Essentially Ours

ASSESSING THE REGULATION OF THE COLLECTION AND USE OF HEALTH-RELATED GENOMIC INFORMATION

Rebekah McWhirter, Lisa Eckstein, Don Chalmers, Jane Kaye, Jane Nielsen, Margaret Otlowski, Megan Prictor, Mark Taylor and Dianne Nicol

Centre for Law and Genetics Occasional Paper No 11

UNIVERSITY of TASMANIA

THE UNIVERSITY OF MELBOURNE

National Library of Australia Cataloguing-in-Publication Data

ISSN 1445-2766

This issue may be cited as

Rebekah McWhirter et al, 'Essentially Ours: Assessing the Regulation of the Collection and Use of Health-related Genomic Information' (Occasional Paper No 11, Centre for Law and Genetics, 2021).

Published by

Centre for Law and Genetics
Faculty of Law
University of Tasmania
Private Bag 89
Hobart Tasmania 7001
Australia

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OCCASIONAL PAPER NO 11

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Preface

The analysis presented in this Occasional Paper was undertaken by members of the Centre for Law and Genetics at the University of Tasmania and members of the Health, Law and Emerging Technologies Programme at the University of Melbourne. Drs Rebekah McWhirter and Lisa Eckstein led the analysis, and Professor Dianne Nicol was the project lead. Otherwise, all authors contributed equally to the analysis, writing and review of this occasional paper. The authors have no conflicts of interest.

This analysis was undertaken pursuant to a contract for consultancy services (reference ID Health/19-20/06077) from the Commonwealth Department of Health. This Occasional Paper is published with the consent of the Department. This analysis was also in part informed and funded by the Australian Research Council through Discovery Grant DP180100269 for the project *Genomic Data Sharing: Issues in Law, Research Ethics and Society* awarded to Centre for Law and Genetics members. We are most grateful to the Commonwealth Department of Health and the Australian Research Council for their funding, but emphasise that this Occasional Paper should not be seen in any way as representing the views of these agencies.

The analysis presented in this Occasional Paper was informed by a scoping review involving a literature review and interviews with key stakeholders. The report on the scoping review is provided in Appendix 1 to this paper. Dr Rebekah McWhirter, Dr Megan Prictor and Dr Lisa Eckstein took primary carriage of this aspect of the project.

We thank officers from the Commonwealth Department of Health together with a range of experts from Commonwealth and state governments, government agencies and academia for reviewing earlier drafts of this Occasional Paper and providing valuable and insightful feedback. We do note, however, that all errors, misinterpretations and omissions are our own.

We thank Madeleine Archer, Rachel Hay and Nikka Milani for research assistance, and thank Kelly Eijdenberg and Jamie Roberts for graphic design.

The analysis for this Occasional Paper was completed on 31 December 2020.

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Executive Summary

Essentially Ours provides an account of the current modes of regulation of health-related genomic samples and data in Australia. This Occasional Paper revisits some of the issues addressed in the 2003 Report, *Essentially Yours*, authored jointly by the Australian Law Reform Commission and Australian Health Ethics Committee pursuant to a reference from the Commonwealth Government ('*Essentially Yours*'). *Essentially Yours* emphasised 'that fundamental human dignity requires that individuals have a high level of control over their own genetic material... and that human genetic information is personal, sensitive, and deserving of a high level of legal protection'.

The information presented in this Occasional Paper differs from *Essentially Yours* in two important ways, largely resulting from the technological and societal changes that have occurred in the intervening years. The first is that the term genetics has been replaced by the term genomics. Genomics refers to the study of the whole genome whereas genetics tends to focus on individual genes. Rapid technological advances mean that genomics is now the most common form of analysis. Secondly, although genomics provides increased clinical and research opportunities, it also raises particular individual and group-member risks. These changes demand reconsideration of the ethical, legal and social implications of and regulatory responses to advances in health-related genomics in Australia.

Although the focus of this Occassional Paper is descriptive—that is, to account for the manner in which current laws apply to genomic samples and data—it necessarily brings to light regulatory gaps and fissures. In particular, traditional regulatory frameworks focus on controls at the level of the individual, either through consent or through efforts to strip genomic information of its identifiers. In the genomic era, these fail to recognise the essential nature of genomic samples and data as inherently identifiable and as shared within families, communities, and populations. This points to the need for a reorientation in the way genomic information is regulated in order to find a balance between 'yours' and 'ours'.

We trust that *Essentially Ours* will provide a rigorous description of the regulatory landscape relevant to genomics in Australia and a tool for future legal analysis and law reform.

Key Findings

CONSENT AND WAIVERS OF CONSENT

- Consent remains a central control on the collection and use of genomic samples and data in both clinical and research settings. There are limits on the extent to which consent from individuals acts as a protection, due to questions about the appropriateness of broad or unspecified consent for future uses, consent for complicated or technical activities, or consent for activities with the potential to affect other people.
- Waivers of consent are available in some circumstances, relying on Human Research Ethics Committees (HRECs) as gatekeepers. Significant concerns remain about the capacity of HRECs to make these discretionary judgments. Additional strategies for oversight, transparency, and support of HREC decision making warrant consideration.

PRIVACY AND ITS LIMITATIONS

- Privacy legislation is a key regulatory influence in this field, but its efficacy is hampered by jurisdictional inconsistencies and by reliance on distinctions that are increasingly inappropriate for application to genomic information, including between clinical and research uses, public and private, and identifiable and non-identifiable information.
- Uncertainty for those working across jurisdictional or organisational lines, combined with uncertainty regarding duties owed under common law, creates a complex national picture.

ABORIGINAL AND TORRES STRAIT ISLANDER IDENTITY AND GENOMICS

- The use of genomics in healthcare and research raises risks for Aboriginal and Torres Strait Islander populations, arising from consultation failures, lack of informed consent, underrepresentation, and inappropriate use of samples and data.
- Caution should be exercised in any discussions regarding a potential role for genetic testing in establishing Aboriginal and Torres Strait Islander identity.
- Current regulatory frameworks inadequately account for group interests. Achieving equity in genomics will be complex and, in relation to Aboriginal and Torres Strait Islander people, will require widespread uptake of culturally appropriate protocols in both clinical and research contexts, supported by Aboriginal and Torres Strait Islander governance and management of genomic resources.

GENETIC DISCRIMINATION

- Although a moratorium on the use of genetic test results for life insurance was introduced in June 2019, genetic discrimination continues to be an issue of public concern, and has been identified as a barrier to the uptake of genomic services.
- The voluntary insurance moratorium extends some protection to consumers; however, its limited terms and self-regulated nature have created uncertainty and complicated the messaging around the effect of the moratorium for consumers.

CONTROVERSIAL SECONDARY USES

- Privacy legislation allows disclosure of genomic information for law enforcement purposes without the knowledge or consent of the individual. Disclosure is permitted for a wider range of offences than those for which primary genetic testing may be undertaken by law enforcement agencies, which is restricted to serious and indictable offences. Higher protections exist for specific contexts, including My Health Record, and could be applied more broadly.
- Secondary use of genomic information for commercial purposes is primarily mediated by consent, but the quality of this consent may be problematic. Both broad and specific consent models demonstrate limitations in ensuring participants have trust in or oversight of the commercial use, nor do they assure public benefit, or necessarily provide a clear insight into benefit sharing.

RETURN OF FINDINGS

- The generation of large amounts of genomic data through clinical and research testing raises questions about obligations on data custodians, including individual researchers, to analyse and reanalyse data and to communicate findings to patients and participants. Guidance comes from privacy legislation, the regulatory regime related to genetic testing and testing laboratories, the research ethics system, and tort law.
- Recent amendments to the *National Statement on Ethical Conduct in Human Research* have clarified the types of matters that researchers need to take into account in making decisions about what to analyse and what to return to individuals, although the practical implications remain to be assessed.

GENETIC TESTS AND GENETIC TESTING

- The most significant legal and regulatory requirements pertaining to genetic tests and genetic testing relate to pre-market assessment of the tests themselves and accreditation of the testing laboratories. The supply of genetic tests with a therapeutic purpose directly to consumers from Australian sources is prohibited, on the basis it falls within a prohibition on self-testing in vitro medical devices.
- Non-therapeutic direct-to-consumer genetic tests available in Australia and all forms of genetic tests and genetic testing performed in other jurisdictions are not covered by these regulatory requirements.

TISSUE SAMPLES AND DATA

- Genomics research involves the movement of tissue samples and associated data from participants and patients to researchers, clinicians and beyond. Mechanisms for the control of samples and data include: ownership rights, ethical frameworks, intellectual property rights, custodianship sovereignty models, and provenance records. People have intuitive assumptions about rights of ownership or control over their samples and data. However, the legal position is more complicated and frequently does not provide the control that may be assumed.

1 Introduction

1.1 Context and Purpose

Genomics is expected to play a transformative role in the Australian healthcare system. In order to leverage the benefits of genomics for health, action is being taken across Australia to address risks and develop infrastructure in a coordinated and collaborative way. The National Health Genomics Policy Framework 2018-2021 was developed by the Commonwealth and States and Territories under guidance from the Australian Health Ministers' Advisory Council ('AHMAC').[1] It was approved by the Council of Australian Governments ('COAG') Health Council in November 2017, and the associated Implementation Plan was approved by the same body a year later. When considering the Framework, Health Ministers agreed that addressing the key genomics-related ethical, legal and social issues of most concern to the community would be a priority, and this was reflected in Actions 1-3 of the Implementation Plan. In developing the Implementation Plan, stakeholders recommended that an evidence-based assessment of the current ethical, legal and social issues for the health genomics-related policy landscape was necessary, particularly with regard to the collection and use of human genomic information.

The ethical, legal and social challenges arising from the collection, use and disclosure of genomic samples and data have long been recognised. Indeed, the Australian Law Reform Commission and Australian Health Ethics Committee undertook a two-year inquiry into these issues, culminating in Report 96, *Essentially Yours: The Protection of Human Genetic Information in Australia ('Essentially Yours')*, published in 2003. Over the subsequent two decades, genomic technologies have advanced in technical capacity and ubiquity, and the inadequacies in some regulatory frameworks have become correspondingly apparent. Many of the tensions identified in *Essentially Yours* – such as the difficulty in reconciling individual and community interests, and the need to foster innovation while maintaining proper ethical scrutiny and legal control – have only become more acute as we have moved from single gene tests to whole genome sequencing.

1.2 Defining the Scope of the Analysis

This document provides a summary and assessment of the relevant legislation and regulations as they relate to current and emerging issues associated with the collection, use and disclosure of health-related genomics information in research and clinical settings. It focuses on human genomics – that is, genomics targeting human material – rather than microbial genomics, which is also an important health-related use of genomic technology. While in many respects the analysis presented in this document may be viewed as an update to *Essentially Yours*, not all of the same issues have been addressed. This analysis is therefore not an exhaustive survey of ethical and legal issues in this area, but rather focuses on those issues of most concern to the community currently, identified through a scoping review undertaken prior to initiating the legal assessment (presented in Appendix 1).

The scoping review included a national and international literature review, combined with the results of a national consultation process, comprising in-depth interviews with a diverse range of stakeholders undertaken in February and March 2020. Participants were recruited for diversity in professional roles (including clinical geneticists, genetic counsellors, health researchers using genomic data, data custodians responsible for genomic data repositories, chairs of genomic data

1 After finalisation of the analysis presented in this Occasional Paper, AHMAC was reviewed as part of the mid 2020 review of the Council of Australian Government and became the Health Chief Executives Forum.

access committees, and representatives of patient groups), gender and geographical location (all States and Territories were represented). Aboriginal and Torres Strait Islander stakeholders were purposively recruited for an enriched sample, given both their unique status within the Australian community and the additional risks presented to Aboriginal and Torres Strait Islander communities by the misuse of genomic samples and information. This sub-cohort included people with expertise in community engagement, genomics research, clinical care (as both health service providers and patients), data sovereignty and health policy. Ethics approval was provided by the Human Research Ethics Committees at the University of Melbourne (ID: 1956048) and the University of Tasmania (ID: H0018640) for this aspect of the project.

These semi-structured interviews were conducted to identify what participants viewed as: key issues in the collection and use of health-related genomic information in clinical and research settings; important legal and regulatory factors; and new issues likely to emerge in the future.

Data were analysed using deductive and inductive approaches, mapping participant responses against expected themes, as well as identifying and grouping responses that generated new themes. All resultant issues, both from the literature and the consultation process, were grouped by theme and then ordered into five broad categories:

1 issues relating to the regulatory environment;

2 issues arising within the healthcare system;

3 issues arising at the overlap between clinical care and research;

4 issues relating to research; and

5 issues arising from other uses of genomic information.

From this list of issues and an associated literature review, a subset of priority issues for legal analysis was agreed on with the Department of Health, taking into consideration current policy issues.

1.3 What is Genomic Information?

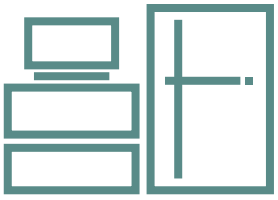
The World Health Organisation defines genetics as the 'study of heredity' and *genomics* as the 'study of genes and their functions, and related techniques'.[2] A distinction is made between the two terms on the basis that genetics focuses on the function and composition of single genes, whereas genomics encompasses all genes (the whole genome) and their interactions. However, the terms are also used interchangeably, to some extent, and it is common for references to genomics to implicitly include genetics. In this report, genomics is used in this way and as the preferred term, except when referring to specific sources where the term genetic is used, or where the standard term is genetic (for example, in relation to genetic discrimination).

Within Australian legislation, the term *genetic information* is used, reflecting the time at which it was drafted and the state of the art at that time. Given the purposes of these various acts (such as privacy and human tissue legislation), and employing established principles of statutory interpretation, it is reasonable to read this term as including genomic information.

The scope of this term in relation to specific pieces of legislation is discussed in more detail in the following chapters, but in general it can be taken as referring to information arising from genetic or genomic testing, and does not include the genomic sample from which the information was derived (see Figure 1).

2 Genomics and World Health, *Report of the Advisory Committee on Health Research*, Geneva, WHO (2002); WHA 57.13: Genomics and World Health, Fifty Seventh World Health Assembly Resolution; 22 May 2004.

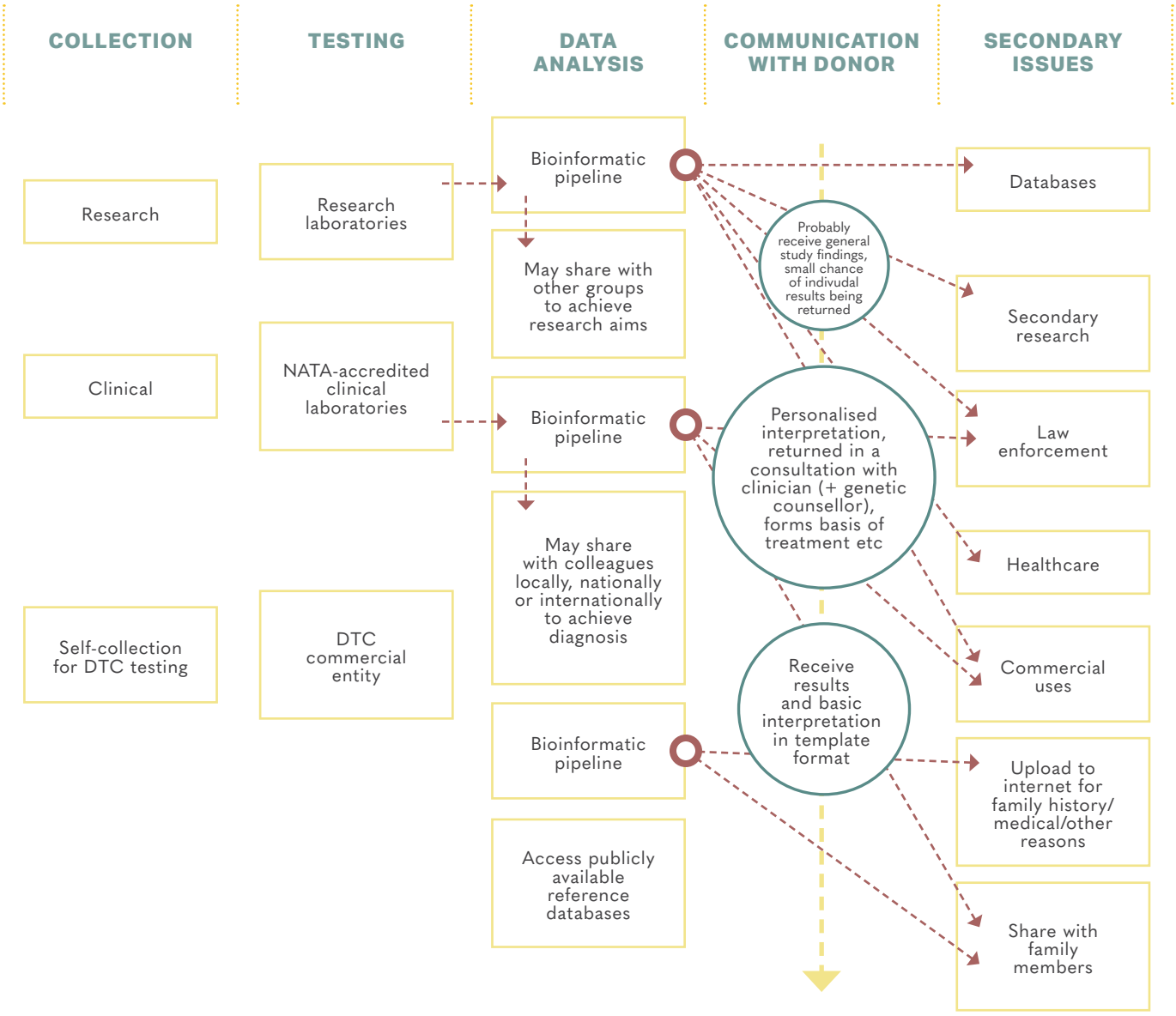
FIGURE 1: SCOPE OF GENOMIC INFORMATION



SAMPLE	TEST	ANALYSIS	COMMUNICATIONS
	GENETIC		
E.g. blood, saliva, buccal, fresh/frozen or formali-fixed paraffin-embedded tissue	Raw data E.g. microsatellites, quantitative polymerase chain reaction or targeted sequencing	Alignment, refer to online databases for interpretation	Result relating to gene of interest may be communicated with patients, participants, research community etc.
	GENOMIC		
	Raw data E.g. unfiltered reads and quality control information	Bioinformatic pipeline to identify variants of interest	Only a subset of all variants identified will be communicated, and will usually be accompanied by information to aid interpretation
IDENTIFIABILITY			
Technically reidentifiable depending on availability of reference data, and arguable as to whether it is 'reasonably identifiable'	Genetic: Unlikely to be reasonably reidentifiable Genomic: Technically reidentifiable, difficulty depending on other available information	Genetic: Unlikely to be reasonably reidentifiable Genomic: Technically reidentifiable, difficulty depending on other available information	Summaries only, unlikely to be reidentifiable once identifiers removed, depending on the rarity of the findings

The flow of genomic information, from the collection through the testing and analysis processes and then to communication and potential secondary uses, can be complex as it includes clinical, research and individual- driven contexts. The most common pathways are summarised in Figure 2.

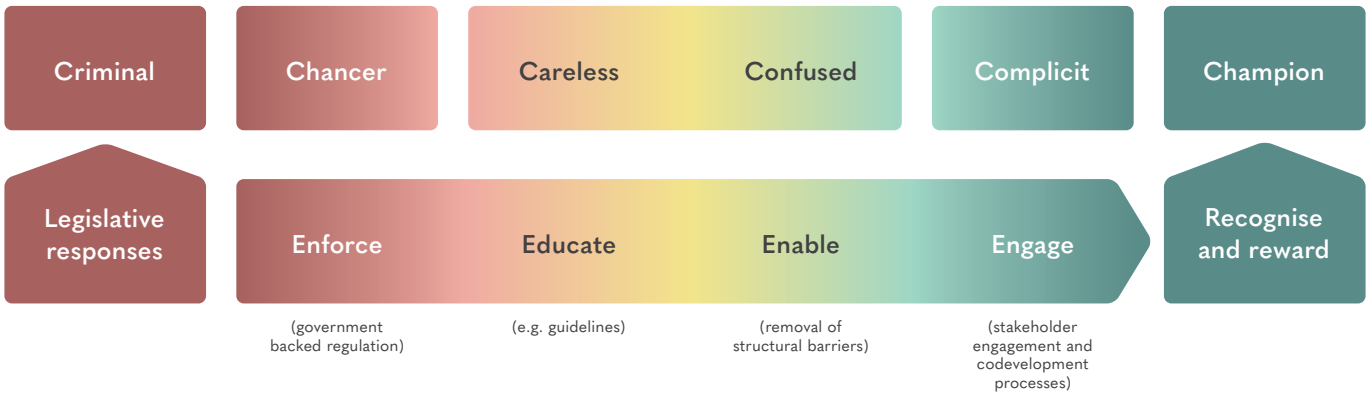
FIGURE 2: FLOW OF GENOMIC INFORMATION



1.4 Regulation of Genomic Information

Regulation of the collection, use and disclosure of genomic information should aim to provide appropriate protections for the individuals to whom that information relates, their families, communities, and wider society. It is important for public trust that the individuals and institutions responsible for genomic information handle that information appropriately. When this trust is undermined, genomic research and healthcare are compromised. Regulation does not inevitably entail legislative responses. Throughout this report, a broad range of regulatory responses are considered. Figure 3 illustrates the regulatory continuum and the options that encompasses, as well as the categories of people targeted by different levels of intervention.

FIGURE 3: REGULATORY CONTINUUM



Adapted from South Australian Environmental Protection Authority, epa.sa.gov.au/business_and_industry/compliance_and_enforcement

2 Consent and Waivers of Consent

2.1 Summary

Consent to medical procedures and for participation in research is a foundational requirement in law and ethics, and provides a key means of ensuring individual control over the way in which their samples are used. Standards for consent to clinical genomic testing will mirror consent requirements for medical procedures more generally: that is, to advise patients of those risks to which a 'reasonable person in the patient's position, if warned of the risk, would be likely to attach significance to it or if the medical practitioner is or should reasonably be aware that a particular patient, if warned of the risk, would be likely to attach significance to it'.[1] In the research context, consent will be governed by the requirements of the *National Statement on Ethical Conduct in Human Research ('National Statement')*,[2] including the need for consent to be 'voluntary' and 'based on ... an adequate understanding of the purpose, methods, demands, risks and potential benefits of the research'.[3]

However, genomics is testing the meaning of consent, particularly as it applies to the storage of genomic samples and data for future unspecified uses. These future uses can be sought for samples and data originally obtained for either clinical or research purposes. In the first instance, the acceptability of such future uses will depend on the scope of the original consent. In circumstances in which the future use is for research, a waiver of consent may be available, based on principles and guidance set out in the *National Statement* as well as Commonwealth, State and Territory privacy laws, and other relevant legislation.

Human Research Ethics Committees ('HRECs') provide a crucial gatekeeper role for decisions about consent and waivers of consent for genomic research. There is no equivalent gatekeeper in the clinical context, with these decisions largely made by individual practitioners. Longstanding concerns about timeliness and variation among HRECs in their decision making have been addressed to some extent through the National Certification Scheme of Institutional Processes Related to the Ethical Review of Multi-centre Research ('National Certification Scheme'), although there is evidence of ongoing delays due to site-specific assessments by Research Governance Officers.[4]

However, individual HRECs are still tasked with making highly discretionary judgments about the acceptability of consent models and waivers of consent for genomic research under the *National Statement* as well as Commonwealth, State and Territory privacy laws.

Additional strategies for oversight, transparency, and support of HREC decision making may warrant consideration. This might include the consideration of publicly established, specialised HRECs for specific kinds of research — for example, to authorise genomic data sharing based on a waiver of consent. This may ameliorate the potential risks of relying on institutional Committees with variable membership, experience, and training to make such decisions.

1 *Rogers v Whitaker* (1992) 175 CLR 479.

2 National Health and Medical Research Council, Australian Research Council and Universities Australia, *National Statement on Ethical Conduct in Human Research* (at 2018).

3 Ibid, [2.2.2].

4 Matilda A Haas et al, 'The Ethics Approval Process for Multisite Research Studies in Australia: Changes Sought by the Australian Genomics Initiative' (2019) 211(10) *Medical Journal of Australia* 440; Elisabeth De Smit et al, 'Heterogeneity of Human Research Ethics Committees and Research Governance Offices across Australia: An Observational Study' (2016) 9(2) *The Australasian Medical Journa*l 33; VM White et al, 'Inconsistencies and Time Delays in Site-Specific Research Approvals Hinder Collaborative Clinical Research in Australia' (2016) 46(9) *Internal Medicine Journal* 1023.

2.2 Consent in Genomics

Consent for genomic medicine is mediated through healthcare practitioners, with oversight provided through tort law and the Australian Health Practitioner Regulation Agency, national health practitioner boards, and relevant State and Territory ombudsmen. The standards applied to such consent will mirror consent standards across medical practice more broadly. As described in *Rogers v Whitaker*, under the tort of negligence, medical practitioners are required to provide patients with information about those risks to which a 'reasonable person in the patient's position, if warned of the risk, would be likely to attach significance to it or if the medical practitioner is or should reasonably be aware that a particular patient, if warned of the risk, would be likely to attach significance to it'.

The onus will be on a patient to bring forward any legal action, which has limitations such as cost and reactivity, given an action can only be made after damage has been sustained. Notifications about a health practitioner's failure to obtain an adequate informed consent also can be lodged with the Australian Health Practitioner Regulation Agency, national health practitioner boards, and relevant ombudsmen. Any liability will depend on the adequacy of the consent process, which goes beyond information provided in a written form. In other words, a written information sheet can provide evidence for the communication of information about risks, but additional forms of communication may be necessary for an adequate consent process.[5]

In the research context, consent forms and processes require approval by an HREC before any research is allowed to commence based on the requirements set out in the *National Statement*. HREC oversight will also apply to the secondary use of genomic samples or data for which participants have provided broad consent, and samples and data for which researchers are seeking a waiver of consent.

In Australia, HRECs are constituted and operate under the *National Health and Medical Research Council Act 1992* (Cth), which requires the NHMRC – through its Principal Committee the Australian Health Ethics Committee – to issue guidelines for the conduct of human research.[6] The *National Statement*, updated 2018 (the *National Statement)* is the most recent version of these guidelines. Over 200 HRECs are currently registered with the NHMRC, including Committees associated with public health institutions like major teaching hospitals, universities, and health departments.[7]

Although review by an HREC is not legally mandated for all Australian research involving human participants (including through tissue samples and/or data), various laws and regulations ensure a requirement for review of the vast majority of such research, including:

1. human research that is funded by, or takes place under the auspices of, the NHMRC, the Australian Research Council, or Universities Australia,[8] or conducted by institutions that receive such funding;
2. clinical trials of unregistered therapeutic goods;[9]
3. research that seeks a waiver of consent for the use of personal information under the *Privacy Act 1988* (Cth) and some State and Territory privacy laws; and
4. research approved under the *Research involving Human Embryos Act 2002* (Cth).[10]

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5 See, eg, *Hassan v Minister for Health* [2005] WADC 182.

6 s 10.

7 National Health and Medical Research Council, *Human Research Ethics Committees* (Web Page) <https://www.nhmrc.gov.au/research-policy/ethics/human-research-ethics-committees>.

8 National Health and Medical Research Council, Australian Research Council and Universities Australia (n 2) 6. The NHMRC funding agreement requires any institution receiving funding to ensure that all of its research complies with the *National Statement*, and as well as the research of partner institutions.

9 *Therapeutic Goods Administration Act 1989* (Cth) s 18; *Therapeutic Goods Regulations 1990* (Cth) s 12AD.

10 *Research involving Human Embryos Act 2002* (Cth) s 21.

The right to withdraw funding has been identified as 'the most important and direct mechanism by which the NHMRC may induce compliance with the *National Statement*'.[11] Evidence of HREC review also may be required for the publication of the results of research,[12] and as a means of demonstrating 'reasonable care' in any litigation arising from a research project.[13]

Additional guidelines apply for research with Aboriginal and Torres Strait Islander Peoples and communities.

The Guidelines clarify the need for researchers to have appropriate cultural competency and articulate six core values for research with Aboriginal and Torres Strait Islander Peoples, being:

- spirit and integrity;
- equity;
- respect;
- reciprocity;
- responsibility; and
- cultural continuity.[14]

The quality of, and inconsistencies between, HREC reviews of research have been raised as issues, including in the consultation for this Project.[15] Over the past decade, extensive policy reforms have been introduced to address these concerns. One such initiative has introduced strategies to support single ethical review of research being conducted at multiple sites. Previously, multi-site research required review and approval by an HREC at each study site. The *National Statement* now makes clear an obligation on HRECs to minimise duplication of ethical review.[16] The National Certification Scheme supports this streamlined approach by allowing research institutions to accept an ethics review conducted by a certified HREC at another institution. To have its HREC certified, an institution must evidence acceptable HREC policies, processes, and procedures.[17] Certification does not assess the quality of an HREC's deliberative review, nor does it review the operation of policies, processes and procedures in practice.

The timeliness of reviews of large-scale genomic research projects, including due to the need for site-specific assessments by research governance officers, has recently been reported as an ongoing challenge.[18]

11 Australian Law Reform Commission and Australian Health Ethics Committee, *Essentially Yours: The Protection of Human Genetic Information in Australia* (ALRC Report 96) (ALRC Report, No 96, May 2003) 14.6 ('*Essentially Yours*').

12 International Committee of Medical Journal Editors, *Preparing a Manuscript for Submission to a Biomedical Journal* (Web Page, 2009) <http://www.icmje.org/recommendations/browse/manuscript-preparation/preparing-for-submission.html>.

13 *Essentially Yours* (n 11) 14.25.

14 Australian Government National Health and Medical Research Council, *Ethical Conduct in Research with Aboriginal and Torres Strait Islander Peoples and Communities* (Report, 2018).

15 See Appendix 1, p135

16 National Health and Medical Research Council, Australian Research Council and Universities Australia, (n 2) 5.3.

17 National Health and Medical Research Council, *National Certification Scheme for the Ethics Review of Multi-Centre Research* (Web Page) <https://www.nhmrc.gov.au/research-policy/ethics/national-certification-scheme-ethics-review-multi-centre-research>.

18 Haas et al, (n 4).

2.3 Overview of Law

2.3.1 STANDARDS FOR CONSENT FOR GENOMIC ANALYSIS

FIGURE 4: INFORMATION FOR CONSENT TO GENOMIC TESTING

Mediated by clinical standards and guidelines

Requires prior HREC approval based on National Statement *requirements*

CONSENT FOR CLINICAL USE

Consent should consider:

- The purpose of testing
- The potential for results of uncertain significance
- The potential for changing knowledge in the future
- The potential for secondary findings
- Insurance implications
- Future information sharing with health practitioners and genetic relatives
- Obtaining consent for future research

National Model of Consent for Clinical Genomic Testing: Clinical Genomic Testing Consent Form (January 2021)

CONSENT FOR RESEARCH

In seeking consent, researchers should consider:

- The scope of the research and the information it will generate
- The plan for communication of findings
- Health implications for participants and their relatives
- Additional implications for participants and their families, such as insurance, employment, social stigma
- Potential for reidentification
- Any future sharing of information, including for research
- Future use of information and specimens, including commercial applications

National Health and Medical Research Council, National Statement on Ethical Conduct in Human Research (Guidelines, 2018), 3.3.10.

At its most basic level, consent for the collection of genomic samples or data must satisfy the requirements of being voluntary, sufficiently informed, and based on adequate information to make an informed decision. These elements will be crucial to the taking of genomic samples for clinical and/or research purposes, as well as any subsequent uses of those samples and the resulting data. In the genomics context, perhaps the key challenge is the question of what constitutes 'adequate information to make an informed decision'. This is especially challenging when it comes to future uses of genomic samples and information, which may be unknown at the time of collection. There is considerable ethical debate about the amount of information required for consent in these circumstances.

2.3.1.1 Standards for consent for clinical genomics

Several templates and guidelines have been put forward to guide the information with which patients should be provided when it comes to clinical genomic testing.

The Human Genetics Society of Australasia ('HGSA') made available genomic testing information sheets and consent forms to its members in 2012. The information sheet explains the testing process as well as possible outcomes, including a causal finding, variations of unknown significance, and incidental findings. The information sheet advises that 'Your DNA sample will not be sent to other laboratories for research studies unless you give prior permission and the study is also approved by the hospital ethics committee'.[19] An option for de-identified data sharing for the purpose of research is included in the associated consent form.

The information and sharing practices set out by the HGSA are broadly reflected in the 2017 National Pathology Accreditation Advisory Council ('NPAAC') *Requirements for Human Medical Genome Testing Utilising Massively Parallel Sequencing Technologies*. This document outlines the minimum best practice requirements for medical pathology laboratories undertaking the performance and implementation of human genetic testing utilising massively parallel sequencing. Most relevantly, S1.4 of the Requirements advises that patients must provide 'additional informed consent' for any research uses of their genomic data. Moreover, 'this consent must be distinct from the consent process for clinical testing'.[20]

In 2019, the Australian Genomics Health Alliance ('AGHA') published a clinical consent form and supporting materials.[21] As with the HGSA form, information is included about the potential outcomes of the test, including the chance of results of unknown significance and incidental findings, the potential for the results to affect patients' ability to obtain some kinds of insurance, and familial implications of the results. However, it diverges from the HGSA and NPAAC when it comes to future uses of samples and data. The AGHA consent provides:

- that de-identified samples and data may be shared 'to advance scientific knowledge'. This sharing is required as a part of the clinical consent process; no option is made available to patients to refuse such sharing.
- a check box option for patients to consent to share their re-identifiable sample and genomic data for research.

The NSW Ministry of Health has developed a national model for clinical consent to genomic testing on behalf of the former Australian Health Ministers' Advisory Council ('AHMAC') Project Reference Group on Health Genomics.[22] The Consent Form includes options to consent to the sharing of test results with other health practitioners and genetic relatives. Rather than seeking consent for the use of samples or data in research, it advises patients that they may be contacted in the future regarding potential participation in research.

These different model consent forms and guidance documents highlight variation in what might be considered the 'blurred line' between clinical and research uses of genomic samples and data. The Consent Form developed by the NSW Ministry of Health maintains the most stringent separation, in permiting only consent for patients to be contacted in the future about potential research uses. The HGSA forms permit some collapsing of the boundaries in allowing patients to consent—at the time of any clinical consent – to future research uses of deidentified data sharing.

19 Human Genetics Society of Australasia, *Genomic Testing* (18 July 2012).

20 National Pathology Accreditation Advisory Council (NPAAC) *Requirements for Human Medical Genome Testing Utilising Massively Parallel Sequencing Technologies* (Web Page) <https://www1.health.gov.au/internet/main/publishing.nsf/Content/npaac-pub-mps>.

21 Australian Genomics Health Alliance, *National Clinical Consent* (Web Page) <https://www.australiangenomics.org.au/tools-and-resources/national-clinical-consent-forms/>.

22 NSW Ministry of Health, *National Model of Consent for Clinical Genomic Testing Final Report* (2021) <https://www.health.nsw.gov.au/services/Pages/genomic-testing-national-model.aspx>

The AGHA collapses the boundaries further by requiring patients to accept research uses of deidentified data and samples, and permitting unspecified consent for future research uses of reidentifiable samples and data.

Evident from the above is significant divergence in standards for consent for the future use of genomic information collected for clinical purposes.

2.3.1.2 Standards for consent for genomic research

Chapter 2.2 of the *National Statement* details the requirements for consent when it comes to research participation, based on the 'guiding principle' that 'a person's decision to participate in research is to be voluntary, and based on sufficient information and adequate understanding of both the proposed research and the implications of participation in it'.[23] The Chapter goes on to address the potential for participants to consent to future uses of their data or tissue in research. Importantly, it states that 'unspecified' consent for the use of data or tissue in future research 'can still be sufficient and adequate for the purpose of consent'.[24]

Further information on consent requirements for genomic research are specified in Chapter 3.3 (Genomic Research) of the *National Statement*, including that:

> Consent specific to the research may not be required or a waiver of the requirement for consent may be considered by an HREC if:
>
> (a) the data or information to be accessed or used was previously collected and either aggregated or had identifiers removed; or
>
> (b) prior consent for the use of the data or information was provided under the scope of a research program that encompasses the proposed research project; or
>
> (c) prior consent for the use of the data or information was provided in the clinical context for research that encompasses the proposed research project; or
>
> (d) unspecified consent has been provided.[25]

The acceptability of various consent models for specific genomic research projects therefore will be at the discretion of reviewing HRECs. Reviewing HRECs will also be responsible for determining the acceptability of future uses of genomic data without going back to participants for additional consent, as well as determining whether conditions for a waiver of consent have been satisfied.

The *National Statement* sets out standards for future uses of genomic samples or data based on unspecified consent. Paragraph 2.2.16 specifies that:

> When unspecified consent is sought, its terms and wide-ranging implications should be clearly explained to potential participants. When such consent is given, its terms should be clearly recorded.[26]

It is unclear whether clinical consent forms that seek permission for future research uses of samples or data satisfy this threshold.

Dynamic consent was developed to address a number of challenges that are raised by genomics research such as informed consent, increased requirements for engagement and management of secondary use of data, as well as the issues associated with biobanks and other medical research infrastructure.[27]

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23 National Health and Medical Research Council, Australian Research Council and Universities Australia, (n 2) 2.2.1.

24 Ibid 2.2.14(c).

25 Ibid 3.3.14.

26 Ibid.

27 Jane Kaye et al, 'Dynamic Consent: A Patient Interface for Twenty-first Century Research Networks' (2015) 23(2) *European Journal of Human Genetics* 141.

Dynamic consent uses internet-based platforms to enable two-way, ongoing communication between researchers and research participants and to support participant-led involvement in research studies. It allows participants to develop greater understanding of the research project; to choose from more granular consent options and to change consent choices over time (including for future use of their data); to indicate preferences for return of results; and to engage in the research process as much as they choose, all according to their own level of commitment and timeframes.[28]

For the users or custodians of data, it provides a way to check the consent preferences attached to data and to re-contact individuals for a new consent. It also enables clinical trial managers, researchers and clinicians to know what type of consent is attached to the use of data they hold and to have an easy way to seek a new consent if the use of the data changes. It is able to support greater accountability and transparency, streamlining consent processes to enable compliance with regulatory requirements. Such ongoing interface may also increase participants' understanding of research and positively impact retention rates.[29] Dynamic consent may be useful in supporting Indigenous Data Sovereignty and supporting culturally-acceptable data governance in health research involving Indigenous people and communities.[30] However, Indigenous scholars have pointed to some of the limitations of a dynamic consent approach, including issues relating to internet accessibility and limited awareness of research activities, both of which have the potential to undermine the informed nature of the consent.[31]

2.3.1.3 Consent requirements for tissue samples

State and Territory human tissue laws impose additional consent requirements for the removal of human tissue from which genomic information may be derived, including the taking of blood samples. Most of these permit the taking of blood samples from adults for medical or scientific reasons contingent on the person's consent.[32] *The Human Tissue Act 1983* (NSW) specifies that such consent must be in writing.[33] With the exception of NSW, the Acts also specify that nothing in the Act applies to the removal of tissue from a living person (adult or child) in the course of medical or dental treatment when carried out by a registered medical practitioner, or the use of the tissue so removed.[34] Therefore, in these States and Territories, tissue removed for clinical purposes can be used for medical research without the need for additional consent, provided a waiver of consent is granted in accordance with the *National Statement* and relevant privacy laws. This removes decision-making from individuals about how their samples should be used.

In comparison, section 21X of the *Human Tissue Act 1983* (NSW) provides that any use of tissue for 'therapeutic, medical or scientific purposes' expressly requires the person from whom the tissue was extracted to give written consent for the use of the tissue for that purpose. This represents an inconsistency in regulatory requirements across Australia, and potentially a regulatory gap that may require consideration.

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28 Matilda A Haas et al, 'CTRL: a Dynamic Consent Platform for Genomic Research' (in press).

29 Isabelle Budin-Ljøsne et al, 'Dynamic Consent: A Potential Solution to Some of the Challenges of Modern Biomedical Research' (2017) 18 *BMC Medical Ethics* 4; Patrick Cheong-Iao Pang et al, 'The Use of Web-Based Technologies in Health Research Participation: Qualitative Study of Consumer and Research Experiences' (2018) 20(10) *Journal of Medical Internet Research* e12094.

30 Megan Prictor et al, 'Australian Aboriginal and Torres Strait Islander Collections of Genetic Heritage: The Legal, Ethical and Practical Considerations of a Dynamic Consent Approach to Decision Making' (2020) 48(1) *The Journal of Law, Medicine & Ethics* 205.

31 Nanibaa' A Garrison et al, 'Entwined Processes: Rescripting Consent and Strengthening Governance in Genomics Research with Indigenous Communities' (2020) 48(1) *The Journal of Law, Medicine & Ethics* 218.

32 *Transplantation and Anatomy Act 1978* (ACT) s 20; *Human Tissue Act 1982* (Vic) s 21; *Transplantation and Anatomy Act 1979* (NT) s 14; *Transplantation and Anatomy Act 1979* (Qld) s 17; *Transplantation and Anatomy Act 1983* (SA) s 18; *Human Tissue Act 1985* (Tas) s 18; *Human Tissue and Transplant Act 1982* (WA) s 18.

33 *Human Tissue Act 1983* (NSW) s 19.

34 *Transplantation and Anatomy Act 1978* (ACT) s 46; *Human Tissue Act 1982* (Vic) s 42(1)(a); *Transplantation and Anatomy Act 1979* (NT) s 26; *Transplantation and Anatomy Act 1979* (Qld) s 47; *Transplantation and Anatomy Act 1983* (SA) s 37; *Human Tissue Act 1985* (Tas) s 28; *Human Tissue and Transplant Act 1982* (WA) s 32.

Notably, an August 2020 article authored by senior officers of the US National Institutes of Health recommends seeking consent for the use of all biospecimens in research, regardless of identifiability. This goes beyond current legal requirements in the US but is argued as being an important foundation for public trust.[35]

2.3.2 HREC ESTABLISHMENT AND COMPOSITION

Under paragraph 5.1.1 of the *National Statement*, institutions must ensure that any human research they conduct or for which they are responsible receives ethical review by an HREC. Negligible risk research and certain kinds of low-risk research can be reviewed by non-HREC levels of ethical review.

However, the *National Statement* clarifies that:

> As a general principle, research including genomics will require review by an HREC; however, if no information that can identify an individual is used and no linkage of data is planned, the research may be determined to carry low risk.[36]

Institutions that establish an HREC must take steps to support the HREC's role of reviewing the ethical acceptability of research, including ensuring that:

- members have relevant experience and/or expertise;
- members undertake an appropriate induction and continuing education;
- HREC review of research proposals is thorough, and processes and procedures are expeditious;
- HREC decisions are transparent, consistent, and promptly communicated; and
- actual and potential conflicts of interest are identified and managed.[37]

The *National Statement* further specifies minimum membership requirements for HRECs, which include:

- a suitably qualified chairperson;
- at least two lay people, one man and one woman;
- at least one person with knowledge of, and current experience in, the professional care, counselling or treatment of people;
- at least one person who performs a pastoral care role in a community;
- at least one lawyer; and
- at least two people with current research experience that is relevant to research proposals to be considered by the HREC.[38]

There is no explicit requirement for HREC members to have expertise in genomics, although this might be captured in the requirement for members with research experience 'relevant to research proposals to be considered by the HREC'.

2.3.3 CONSENT FOR SPECIAL POPULATIONS

Some additional consent requirements apply to genomic testing of embryos, children, and deceased persons.

35 Carrie D Wolinetz and Francis S Collins, 'Recognition of Research Participants' Need for Autonomy: Remembering the Legacy of Henrietta Lacks' [2020] *JAMA*, <https://jamanetwork.com/journals/jama/fullarticle/2769506>.

36 National Health and Medical Research Council, Australian Research Council and Universities Australia (n 2) 3.3 and Introduction.

37 Ibid 5.1.28.

38 Ibid 5.1.30.

2.3.3.1 Embryos and fetuses

Clinical and research activities with gametes and embryos in Australia must comply with the NHMRC *Ethical Guidelines on the Use of Assisted Reproductive Technology in Clinical Practice and Research 2017*. The Guidelines specifically address ethical issues associated with preimplantation genetic testing ('PGT'), and require individuals or couples seeking PGT to undergo counselling before they may consent to the procedure.[39] The Guidelines go on to require that clinics 'provide individuals or couples who seek PGT with sufficient information to facilitate an understanding of the limitations of the technology, including the likelihood of false positive and false negative results and the potential for developmental abnormalities to occur despite PGT'.[40]

Under the *Research involving Human Embryos Act 2002* (Cth), research on human embryos can only occur after a licence is issued by the NHMRC Embryo Research Licensing Committee. A condition of any such licence being issued is a determination by the Committee that appropriate protocols are in place to enable proper consent to be obtained.[41] Assessment and approval by an HREC is also required.

2.3.3.2 Children and young people

The predictive nature of genomic information has led to longstanding limitations on allowing children and young people to consent to certain kinds of genomic testing. There is evidence to suggest more flexible approaches in clinical practice and guidance would be advantageous.[42] The permissibility of consent by, or on behalf, of a minor for predictive genomic testing will be tested through tort law and potential disciplinary actions through professional societies.

Limitations on the taking of blood samples and other human tissue from children for the purpose of research also apply in some State and Territory human tissue laws. These permit parental consent to blood samples being taken from children for the purpose of research, generally preconditioned on this not being prejudicial to the health of the child[43] and/or the child's agreement.[44] Less clear is whether there is authorisation under the Acts to remove other kinds of tissue (eg, tumour samples) from children for scientific purposes, although removal is permissible for medical purposes with subsequent use of the tissue for scientific purposes.

An open question is the need for reconsent for future use of biological samples and data taken from minors after the minor reaches 18 years of age. Under the National Statement and human tissue Acts, authorisation for such extraction and use is provided by parents on the minor's behalf. Some guidelines and policies provide that researchers should obtain consent from the minor themself for studies that occur after the time that they can provide consent on their own behalf.[45] However, this position is contestable,[46] and is not currently established in Australian law or the National Statement.[47]

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39 National Health and Medical Research Council, *Ethical Guidelines on the Use of Assisted Reproductive Technology in Clinical Practice and Research* (at 2017) 8.18.

40 Ibid 8.18.2.

41 *Research involving Human Embryos Act 2002* (Cth) s 21(3)(a)(i).

42 Cara Mand et al, 'Predictive Genetic Testing in Minors for Late- Onset Conditions: A Chronological and Analytical Review of the Ethical Arguments' (2012) 38(9) *Journal of Medical Ethics* 519.

43 See, eg, *Transplantation and Anatomy Act 1978* (ACT) s 21; *Human Tissue Act 1982* (Vic) s 22; *Transplantation and Anatomy Act* (Qld) s 18; *Transplantation and Anatomy Act 1983* (SA) s 19; *Human Tissue Act 1983* (NSW) s 20; *Human Tissue Act 1985* (Tas) s 19; *Human Tissue and Transplant Act 1982* (WA) s 19.

44 See, eg, *Human Tissue Act 1982* (Vic) s 22; *Transplantation and Anatomy Act* (Qld) s 18; *Transplantation and Anatomy Act 1983* (SA) s 19; *Human Tissue Act 1983* (NSW) s 20; *Human Tissue Act 1985* (Tas) s 19; *Human Tissue and Transplant Act 1982* (WA) s 19.

45 See discussion in Benjamin Berkman et al, 'Reconsidering the Need for Reconsent at 18' (2018) 142(2) *Pediatrics* 1.

46 Ibid.

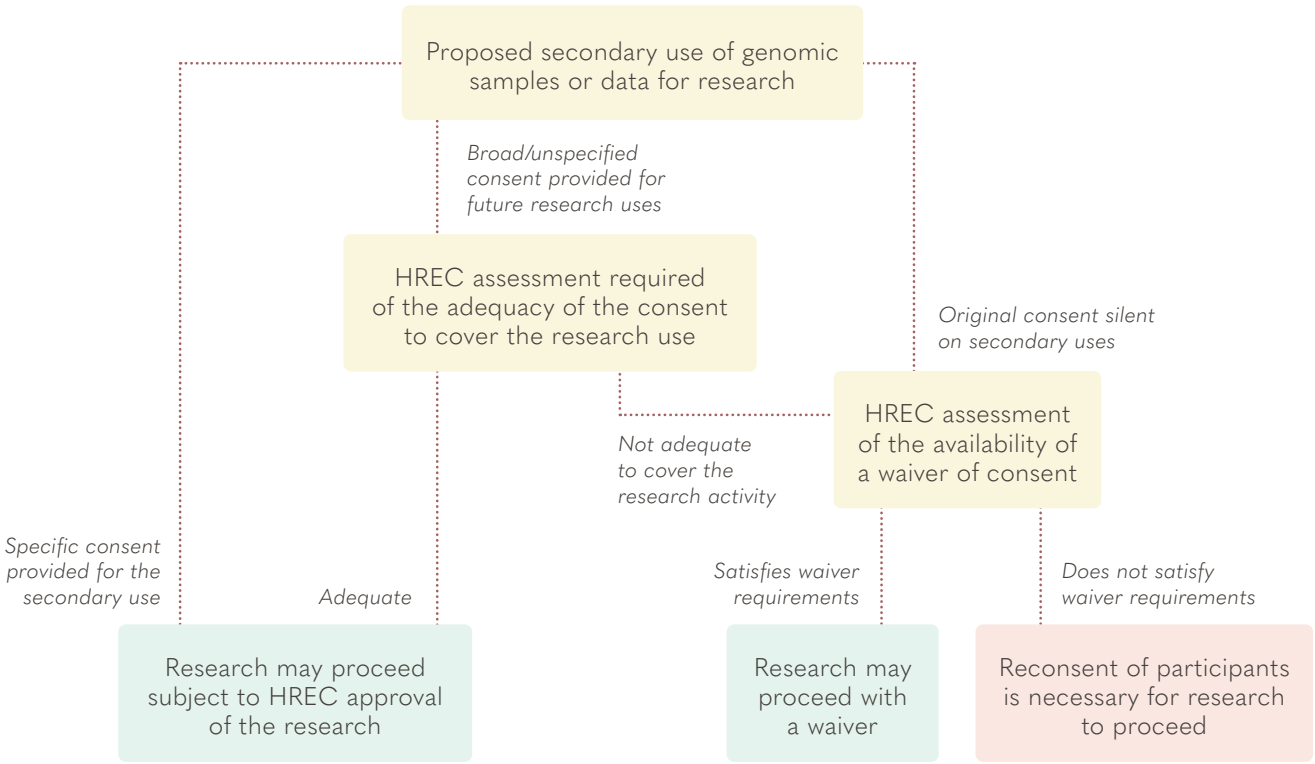
47 Amanda Rush et al, 'Opinions of Young Adults on Re-Consenting for Biobanking' (2015) 167(4) *The Journal of Pediatrics* 925.

2.3.3.3 Deceased persons

The indefinite storage of biospecimens in many biorepositories means that researchers may seek to sequence a sample from a deceased person.[48] Consent usually, but not always, will have been obtained from the person for inclusion of the tissue into the biorepository, with varying degrees of specificity in terms of its future uses. One possibility is to obtain consent from a next of kin for future research activities, but this becomes progressively harder as the sample ages.[49] The death, or likely death, of persons from whom samples are being requested for testing is also a common justification for a waiver of consent, and will be discussed below.

2.3.4 WAIVERS OF CONSENT

FIGURE 5: CONSENT FOR PROPOSED SECONDARY RESEARCH USING GENOMIC INFORMATION AND SAMPLES



The *National Statement* permits waivers of consent for participation in research in certain circumstances. Other privacy exceptions may apply to sharing activities for clinical purposes, as discussed in Chapter 3.

48 Rebekah McWhirter and Margaret Otlowski, 'Regulation of Non-consensual Genetic Testing in Australia: Use of Samples from Deceased Persons' (2016) 24(1) *Journal of Law and Medicine* 150.

49 Ibid.

Paragraph 2.3.9 of the *National Statement* clarifies that only an HREC can grant a waiver of consent for research using personal information in medical research, or personal health information. Non-HREC review bodies established by an institution may grant a waiver of consent for other research. Opt-out consent also is permissible under the *National Statement*; however, it advises that this approach should not be used in genomic research.[50]

Paragraph 2.3.10 provides that, before approving a waiver of consent, an HREC or other review body must be satisfied that:

(a) involvement in the research carries no more than low risk to participants

(b) the benefits from the research justify any risks of harm associated with not seeking consent

(c) it is impracticable to obtain consent (for example, due to the quantity, age or accessibility of records)

(d) there is no known or likely reason for thinking that participants would not have consented if they had been asked

(e) there is sufficient protection of their privacy

(f) there is an adequate plan to protect the confidentiality of data

(g) in case the results have significance for the participants' welfare there is, where practicable, a plan for making information arising from the research available to them (for example, via a disease-specific website or regional news media)

(h) the possibility of commercial exploitation of derivatives of the data or tissue will not deprive the participants of any financial benefits to which they would be entitled

(i) the waiver is not prohibited by State, federal, or international law.[51]

Additional requirements apply to waivers of consent that involve the collection, use, or disclosure of personal information under Commonwealth and State and Territory privacy laws (noting that the *National Statement* advises that the use of identifiable genomic information will not usually meet the criteria of 'low risk' to warrant a waiver of consent).[52] Sections 95 and 95A of the *Privacy Act 1988* (Cth) allow for the NHMRC to issue guidelines for the use of certain kinds of personal information in research. The *Guidelines Approved under Section 95 of the Privacy Act 1988* (the s 95 Guidelines) apply to requests for waivers of consent for the collection, use or disclosure of personal information held by Commonwealth agencies for the purpose of 'medical research'.[53] In comparison, the *Guidelines Approved under Section 95A of the Privacy Act 1988* (the s 95A Guidelines) apply to requests for waivers of consent for the collection, use or disclosure of personal health information held by organisations for the purposes of research, or the compilation or analysis of statistics, relevant to public health or public safety.[54]

The extent of any resulting gap between the permissibility of waiving consent for the use of genomic data held by agencies as compared with organisations will depend on how broadly 'medical research' is interpreted. The Australian Privacy Commissioner finding in *'PA' and Department of Veterans' Affairs (Privacy)*[55] suggests the likelihood of a broad interpretation. At issue was the establishment of an information database by the Department of Veteran Affairs 'to assist in health research projects', including the Mental Health and Wellbeing Transition Study.

50 National Health and Medical Research Council, Australian Research Council and Universities Australia (n 2) 3.3.15.

51 Ibid 2.3.10.

52 Ibid 3.3 and Introduction.

53 National Health and Medical Research Council, *Guidelines Under Section 95 of the Privacy Act 1988* (at November 2014).

54 National Health and Medical Research Council, *Guidelines Approved under Section 95A of the Privacy Act 1988* (at March 2014).

55 [2018] AICmr 50.

Based on principles of statutory interpretation, the Privacy Commissioner adopted a deliberately broad interpretation of medical research as including 'the investigation or study to gain knowledge and understanding of the causes, treatment and prevention of human diseases'.[56] In this case, the proposed uses of the databases were considered to fall within the scope of the exception since their broad purpose was the study of 'the causes, treatment and prevention of human diseases'. This was the case even though the Commissioner noted that at least some of the research for which the database would be used may fall outside the field of medicine.

Although there are some differences in scope, the s 95 Guidelines and the s 95A Guidelines operate in a broadly consistent manner. Both authorise acts that otherwise would breach an Australian Privacy Principle provided an HREC has approved the activity in accordance with the specified criteria. For both, the Guidelines seek to provide a mechanism for ensuring that approval is granted only in circumstances in which the public interest in the research activity substantially outweighs the public interest in maintaining privacy protections. Prerequisites for such an assessment include:

- that seeking consent from the affected individual/s is impracticable;
- that the research activity cannot achieve its purpose through the use of de-identified information.

Decisions of an HREC to approve a waiver of consent under sections 95 or 95A of the *Privacy Act* must be reported to the NHMRC annually.

Most Australian States and Territories also establish research exceptions in their privacy laws, though these differ to some extent in scope. Notably, there is no clear research exception in the ACT privacy framework, and the Commonwealth and South Australian exceptions are limited to health-related data.[57] Consistently with the Commonwealth regime, these primarily precondition a waiver of consent for research that meets a specified public interest test and where obtaining consent is 'impracticable'. Some add requirements for the information to be in de-identified form and/or a reasonable belief that the recipient will not disclose the information.[58]

2.3.5 IMPRACTICABILITY

The 'impracticability' requirement is one of the most challenging aspects of the criteria for approving waivers of consent for the collection, use, or disclosure of personal information in research—both under the *National Statement* and under Commonwealth and some State and Territory privacy laws. This may be argued for research projects seeking to use genomic data or samples donated many years earlier without clear consent for future use. Another situation in which a waiver might be sought is for the release of deidentified data to medical journals in satisfaction of journal data-sharing policies. In the consultation process for the Scoping Review, a researcher expressed discomfort and uncertainty about proceeding with research on the basis of a waiver of consent with notification for samples collected years earlier. The researcher noted, however, the impracticability of the alternative of reconsenting a thousand participants. Aboriginal and Torres Strait Islander stakeholders also reported significant discomfort with the use of waivers of consent in relation to Aboriginal and Torres Strait Islander genomic data.

56 Ibid [52].

57 Ruthie Jeanneret et al, 'Enhancing Early Detection of Cognitive Impairment in the Criminal Justice System: Feasibility of a Proposed Method' (2019) 31(1) *Current Issues in Criminal Justice* 60.

58 Ibid Table 1.

The thresholds for HREC assessment of claims of impracticability as one of the requirements for a waiver of consent are unclear, including when impracticability is based on the cost of obtaining consent.[59] No definition is included in the *National Statement*, other than the explanation that it might apply 'due to the quantity, age or accessibility of records'. This can be compared, for example, with Canada's *Tri-council Policy Statement*, which permits a Research Ethics Board (which are broadly equivalent to Australian HRECs) to approve waivers of consent for research if the Board is satisfied that 'it is impossible or impracticable... to carry out the research and to address the research question properly... if the prior consent of participants is required'. The Canadian Statement clarifies that '"impracticable" refers to undue hardship or onerousness that jeopardises the conduct of the research. It does not refer to mere inconvenience'.[60] This definition appears considerably narrower than the definition of 'impracticable' in the Macquarie dictionary, being something 'that cannot be put into practice with the available means'.

In its review of federal privacy laws, the ALRC also noted situations in which seeking consent might be 'practicable' but would be unreasonable, for example, because of an adverse impact on the integrity and validity of a research program, or where previous unspecified consent for future uses of samples or tissue has been obtained.[61] The ALRC recommended that waivers of consent should be available for the purposes of research where it is 'unreasonable or impracticable' to seek that consent.[62] This recommendation is not reflected in current Australian laws.

2.3.6 INVOLVEMENT IN THE RESEARCH CARRIES NO MORE THAN LOW RISK

Waivers of consent can only be approved under the *National Statement* for research that an HREC assesses as 'low risk'. The *National Statement* clarifies that:

> As a general principle, research including genomics will require review by an HREC; however, if no information that can identify an individual is used and no linkage of data is planned, the research may be determined to carry low risk.[63]

As with other criteria, this leaves individual HRECs to assess the risks associated with the use of deidentified genomic data or samples in research. This may include potential group harms stemming from genomic research, including any stigmatization and dignitary harms to members, particularly of vulnerable groups.[64,65]

59 Lisa Eckstein et al, 'Australia: Regulating Genomic Data Sharing to Promote Public Trust' (2018) 137(8) *Human Genetics* 583.

60 Canadian Institutes of Health Research, Natural Sciences and Engineering Research Council of Canada and Social Sciences and Humanities Research Council of Canada, *Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans* (at 2018) 3.

61 *Essentially Yours* (n 11) 65.

62 Ibid Rec 65-5.

63 National Health and Medical Research Council, Australian Research Council and Universities Australia, (n 2) 3.3 and Introduction.

64 Mark A Rothstein, 'Is Deidentification Sufficient to Protect Health Privacy in Research?' (2010) 10(9) *The American Journal of Bioethics* 3.

65 Elisabeth De Smit et al, 'Heterogeneity of Human Research Ethics Committees and Research Governance Offices across Australia: An Observational Study' (2016) 9(2) *The Australasian Medical Journal* 33.

2.3.7 NO KNOWN OR LIKELY REASON FOR THINKING THAT PARTICIPANTS WOULD NOT HAVE CONSENTED IF THEY HAD BEEN ASKED

A further requirement in the *National Statement* for approving a waiver of consent is that there is no known or likely reason for thinking that participants would not have consented if they had been asked. This has been characterised as 'presumed consent' of participants.[66] Australia is reportedly the only country to include a presumed consent requirement in its research ethics code for waivers of consent.[67] As with impracticability, interpretation of the presumed consent requirement is highly subjective. No guidance is available on the acceptable threshold for presumed consent. Ballantyne and Schaefer note that this could refer to 'a simple majority (1/2), a clear majority (2/3), nearly all (4/5) or all (100 per cent) of the public'.[68] This is challenging to reconcile with empirical research reporting varying rates of decline for genomic sequencing research, including 13 per cent whose reported reasons for declining participation were privacy or discrimination concerns.[69] Depending on a Committee's views on the acceptable threshold and the characteristics of a proposed study, this could equally support or refute an application for a waiver of consent for use of previously collected genomic data or samples.

2.3.8 HRECS AS AUTHORISING BODIES FOR WAIVERS OF CONSENT

Commonwealth and some State and Territory privacy frameworks rely upon reviewing HRECs to make decisions about the acceptability of a waiver of consent for research, including genomic research. This raises the question of the appropriateness of these institutional Committees to assess conformity with legal compliance. In comparison, in England and Wales, any access to personal information without consent, or alternative statutory basis, for the purpose of medical research requires review by the Confidentiality Advisory Group ('CAG') – an independent statutory body established to advise the Health Research Authority. This advice process operates in addition to review of research for ethical acceptability by a Research Ethics Board. Meeting minutes are published online, including detailed information on CAG deliberations.[70]

2.3.9 HREC OVERSIGHT, ACCOUNTABILITY AND GUIDANCE

Scholars assessing HRECs and their international equivalents note that these Committees receive little guidance when it comes to the ethical concerns raised by data-intensive projects such as large-scale genomic research.[71] As such, it is unsurprising that considerable variability has been reported in HREC decision making.[72] Variation in the expertise and views of different HRECs was identified as a problem by several participants in the Scoping Review.

Given the discretion involved in determining whether a project has satisfied the criteria for ethical acceptability set out in the *National Statement*, some variability in decision making is to be expected. However, the limited avenues for HREC oversight and accountability means there are few avenues for resolving differences among Committees.

66 Angela Ballantyne and G Owen Schaefer, 'Taxonomy of Justifications for Consent Waivers: When and Why Are Public Views Relevant?' (2019) 45(5) *Journal of Medical Ethics* 353.

67 Ibid.

68 Ibid.

69 Laura Amendola et al, 'Why Patients Decline Genomic Sequencing Studies: Experiences from the CSER Consortium (2018) 27(5) *Journal of Genetic Counselling* 1220.

70 NHS Health Research Authority, *Confidentiality Advisory Group* (Web Page) <https://www.hra.nhs.uk/about-us/committees-and-services/confidentiality-advisory-group/#:~:text=The%20Confidentiality%20Advisory%20Group%20(CAG,Health%20for%20non%2Dresearch%20uses>.

71 Edward S Dove et al, 'Ethics Review for International Data-Intensive Research' (2016) 351(6280) *Science* 1399.

72 De Smit et al (n 65).

An extensive search of Australian legal databases did not reveal any actions directly against HRECs regarding their review of research. HREC decision making, including authorisations for a waiver of consent for the use or disclosure of personal information under ss 95 and 95A of the *Privacy Act*, have been characterised as unlikely to be subject to judicial review.[73]

Although not related to genomic information, the case of 'PA' and Department of Veterans Affairs (Privacy) regarded a Commonwealth agency's disclosure of personal information for research purposes based on HREC approval of an opt-out consent process.[74] The Commissioner reviewed the minutes of the HREC meeting approving the research, and determined that the HREC deliberations were conducted consistently with the s 95 Guidelines. Therefore, the agency's disclosure was permissible under the *Privacy Act*. The Commissioner's oversight of the HREC review was procedural rather than substantive in nature: that is, ensuring that the HREC had assessed the relevant criteria for a waiver of consent, rather than considering the quality of the HREC's assessment.

A small number of Freedom of Information applications have been lodged to seek additional information on HREC processes and procedures. Tribunals have varied in their willingness to release deliberative information.[75]

The limited availability of information about Australian HREC deliberative processes and oversight of their decision making can be contrasted with some international frameworks. For example, in the US, the Office for Human Research Protections sits within the Department of Health and Human Services. A part of its role is investigating potential non-compliance with research ethics requirements. This includes the failure of Institutional Review Boards (the US HREC equivalents) to provide an adequate review. Assessments are made publicly available.[76]

In England, the National Research Ethics Service ran multiple cycles of a Shared Ethical Debate exercise. The exercise sought to foster greater dialogue and reflection about Research Ethics Committee ('REC') processes and judgments. It involved the distribution of a single research application to a number of RECs. Each REC was expected to participate as a part of its accreditation. Comments are recorded and feedback provided to Committees to promote self-refection and consistency among Committees.[77]

Australian HRECs have weighty responsibilities for regulating the use of genomic samples and data, however, there are gaps in oversight of their operations. Federal and state governments could help to mitigate this risk by increasing oversight of Committees. One suggestion would be to audit Committee processes and decision making, and/or implementing programs similar to the English Shared Ethical Debate exercise to encourage continued Committee self-reflection. Publicly established, centralised Committees for specific kinds of research applications – for example, to authorise genomic data sharing based on a waiver of consent – also may ameliorate the potential risks of relying on variably constituted and resourced institutional Committees.

73 *Essentially Yours* (n 11) 65.1444.

74 *PA' and Department of Veterans' Affairs (Privacy)* [2018] AICmr 50.

75 *Re Whitely and Curtin University of Technology* [2008] WAICmr 24; *Battin v University of New England* [2013] NSWADT 73; *Raven v The University of Sydney* [2015] NSWCATAD 104.

76 OHRP OHRP Determination Letters and Other Correspondence (Web Page, 23 June 2009) <https://www.hhs.gov/ohrp/compliance-and-reporting/determination-letters/index.html>.

77 Peter Heasman, Alain Gregoire and Hugh Davies, 'Helping Research Ethics Committees Share Their Experience, Learn from Review and Develop Consensus: An Observational Study of the UK Shared Ethical Debate' (2011) 7(1) *Research Ethics* 13.

3 Privacy and its Limitations

3.1 Summary

This chapter considers Commonwealth, State and Territory privacy laws as they relate to genomic information, including the disclosure of genomic information to relatives and the treatment of data relating to the deceased.[1] Privacy law, at the Commonwealth level, reflects international commitments to the International Covenant on Civil and Political Rights and OECD Privacy Guidelines.[2] It serves to promote the protection of the privacy of individuals, responsible handling of personal information, and to facilitate free flow of information across national borders in ways that respect individual privacy. State and Territory privacy law is typically concerned with not only the protection of individual privacy but also the security and integrity of data and the establishment of the conditions for appropriate use and disclosure in pursuit and protection of individual and public interests. This is important in the context of genomic information due to the significance of the information from the perspective of individual and group privacy and the importance of establishing the conditions under which genomic data can flow between organisations and across borders for the purposes of health care and research.

Whether genomic information is within the scope of privacy law will typically turn on whether it is about a reasonably identifiable individual. This can be difficult to assess. There are also jurisdictional differences on if, and why, genomic information is categorised as sensitive information. In some jurisdictions genomic information will be categorised as sensitive information only if genomic information falls within the broader category of 'health information.' However, under the *Privacy Act 1988* (Cth) ('*Privacy Act*'), 'genetic information' is specifically defined as sensitive information whether or not it is health information.

Currently, all Australian jurisdictions regulate the disclosure of genomic information to at-risk relatives.[3] However, the regulatory approaches can differ in scope and substance. While small, these differences may introduce uncertainty when operating across jurisdictional or organisational (public/private) lines.

Protection for data relating to the deceased varies across States and Territories. In particular, the length of time for which data relating to the deceased may be subject to statutory protection varies considerably. Alongside uncertainty with regard to duties owed under common law duties of confidentiality, this creates a complex national picture.

1 Since the analysis for this Occasional Paper was completed, the Commonwealth government released a Discussion Paper pursuant to its review of the *Privacy Act 1988*. The Discussion Paper includes proposals for reform which, if implemented, will have significant ramifications for the ways that the collection and use of genomic information is regulated through privacy law. <https://www.ag.gov.au/integrity/consultations/review-privacy-act-1988>.

2 *International Covenant on Civil and Political Rights*, opened for signature 19 December 1966, 999 UNTS 171 (entered into force 23 March 1976) ('ICCPR'); OECD, *Guidelines on the Protection of Privacy and Transborder Flows of Personal Data* (Guidelines, 11 July 2013) ('OECD Privacy Guidelines').

3 *Health Records (Privacy and Access) Act 1997* (ACT) sch 1; *Health Records and Information Privacy Act 2002* (NSW) sch 1, s 10; *Information Act 2002* (NT) sch 2; *Hospital and Health Boards Act 2011* (Qld); *Health Care Act 2008* (SA) s 93; *Personal Information Protection Act 2004* (Tas) sch 1; *Health Records Act 2001* (Vic) sch; *Health Service Act 2016* (WA), s 220 in conjunction with the *Health Services (Information) Regulations 2017* (WA).

3.2 Issues Considered

Stakeholders identified a number of issues of community concern in relation to the use and disclosure of genomic data for healthcare and research purposes related to Australian privacy laws. These included the material scope of privacy law in Australia and its application to genomic information. For example, it was not considered clear in practice when genomic information falls within scope of statutory protection due to questions about whether it is 'about' an 'identified or identifiable individual.' This gave rise to doubt over whether material scope extends to data in all situations of concern to people. A specific concern was expressed with regards to the impact that privacy legislation has on data sharing for research purposes.

There was a perception that privacy legislation may inhibit sharing and, in particular, data linkage may be discouraged by a concern that linkage may increase the identifiability of a dataset. Waivers of consent for research purposes are addressed in Chapter 2.

Where genomic information does fall within scope of privacy law, then questions arise about appropriate access and disclosure to family members, including without the consent of the person to whom the information relates. Provisions to ensure such access when there is a serious (although not necessarily imminent) threat to a person's life, health or safety was a key recommendation of *Essentially Yours: The Protection of Human Genetic Information in Australia* ('*Essentially Yours*'). This has been enacted in the *Privacy Act*; however, only some States and Territories have enacted equivalent amendments to their privacy legislation.

The chapter further addresses community concerns expressed with regard to uncertainty on the level of protection for genomic data relating to the deceased.

3.3 Overview of Law

3.3.1 MATERIAL SCOPE OF LEGISLATION: WHEN IS GENOMIC INFORMATION COVERED?

Stakeholders raised questions about the material scope of privacy law in Australia and the lack of clarity with regards to a number of definitional issues. Stakeholder 19 said, 'It would be disappointing if people responded by just siloing information because they are unsure about the [privacy] framework and where they sit with the law'.

Privacy and genomic information sharing in Australia is governed by a complex patchwork of Commonwealth, State and Territory legislation. The relevant Commonwealth legislation is the *Privacy Act*. This applies to Commonwealth bodies, as well as to private companies with an annual revenue over $3 million or providing a health service and holding any health information (other than in an employee record).[4] The Commonwealth regime further applies to organisations that have obtained genomic or other personal information through providing health services to individuals. These provisions apply to health professionals, including general practitioners, working in private practice.[5]

4 See *Privacy Act 1988* (Cth) s 6D ('*Privacy Act*'); See also Rebekah McWhirter, Carolyn Johnston and Jo Burke, 'Disclosure of Genetic Results to At-Risk Relatives without Consent: Issues for Health Care Professionals in Australia' (2019) 27(1) *Journal of Law and Medicine* 108, 109; see also Natalia Meggiolaro et al, 'Disclosure to Genetic Relatives without Consent – Australian Genetic Professionals' Awareness of the Health Privacy Law' (2020) 21(1) *BMC Medical Ethics* 13, 14.

5 Margaret Otlowski, 'Disclosing Genetic Information to At-Risk Relatives: New Australian Privacy Principles, but Uniformity Still Elusive' (2015) 202(6) *Medical Journal of Australia* 335, 336.

Generally, clinical genomics services and other medical practices are in the State or Territory public health sector.[6] Accordingly, the collection, sharing and use of medical (including genomic) information from these services will be governed by State and Territory regimes.[7] Each of the legislative regimes incorporate a set of privacy principles, which articulate high-level privacy objectives. The Australian Privacy Principles ('APPs') are set out in Schedule 1 of the *Privacy Act*. Similar but not identical privacy principles are included in State and Territory privacy laws.

3.3.2 IS GENOMIC INFORMATION 'PERSONAL INFORMATION' UNDER PRIVACY LAWS?

Genomic information will typically only fall within scope of privacy laws if it meets the legislative definition of personal information.[8] This is an issue that the community has suggested can be challenging if there is any uncertainty about the identifiability of genomic information, particularly in the context of data sharing and linkage activities.

At the Commonwealth level, personal information is defined as information or opinion 'about an identifiable individual, or an individual who is reasonably identifiable'.[9] Therefore, where information is 'no longer about an identifiable individual or an individual who is reasonably identifiable',[10] this information is no longer subject to the provisions of the *Privacy Act*. However, information that retains a code or other means of reasonably being linked to data from other data sources to re-identify an individual is 'reasonably identifiable'. This means such information would still be considered personal information, and therefore still subject to the protections of the *Privacy Act*.[11]

It can be difficult for those processing genomic information to confidently assess when it is reasonably identifiable; this is particularly the case as data moves between different collections with different permutations of possible (future) data linkage. The risks of re-identification may be mitigated to varying degrees by organisational, technical, and contractual measures. The entity subject to the legal obligations (eg, the relevant APP entity in the case of the *Privacy Act*) is legally responsible for compliance and must determine whether genomic information they collect, use, or disclose falls within scope of privacy law all things considered. In practice, the expertise needed to assess whether definitional thresholds are met is likely to sit with experts who may be distant from organisational responsibility for legal compliance. For example, health researchers in a university may be well placed to assess risks of identifiability in day-to-day practice but may be relatively poorly placed to contribute that understanding or insight to organisational decision-making regarding legal responsibilities in light of broader organizational, technical and contractual arrangements.

Fragmented and siloed expertise relevant to assessment will undermine the quality of decision-making and the opportunities for regulatory compliance. These risks are exacerbated where legal thresholds are themselves unclear on important considerations. For example, the definition of personal information does not indicate the extent to which account should be taken of all objective factors, such as the costs of and the amount of time required for identification, the available technology at the time of the processing, and the possibility of technological developments.[12]

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6 Ibid.

7 At least to the extent that regulation of medical data sharing cannot be shoehorned into legislation made under one of the Commonwealth's existing heads of power (eg, corporations, external affairs, interstate trade and commerce). The *Privacy Act* (n 4) is a good example of this. However, the Act is limited in its scope as it relies predominantly on the external affairs power. As a result, the text of the *Privacy Act* is necessarily limited to legislation deemed appropriate and adapted to implement Australia's international obligations under various treaties (see, eg, ICCPR and the OECD Privacy Guidelines (n 2)). <https://www.oecd.org/sti/ieconomy/oecdguidelinesontheprotectionofprivacyandtransborderflowsofpersonaldata.htm>.

8 It is to be noted that, in some cases, the scope of legislative protection may be determined by the concept of 'confidential information' rather than 'personal information', see for example the *Hospital and Health Boards Act 2011* (Qld).

9 *Privacy Act* (n 4) s 6(1) (definition of 'personal information').

10 Ibid (definition of 'de-identified'); Office of the Australian Information Commissioner, 'De-identification and the Privacy Act' (OAIC, March 2018) 1 and 9.

11 Ibid 8.

12 Cf Regulation (EU) 2016/679 of the European Parliament and the Council on the protection of natural persons with regard to the processing of personal data and on the free movement of such data (General Data Protection Regulation), Recital 26.

That said, at a high level, all State and Territory definitions of 'personal information' are similar. This has meant that considerations for determining whether an individual's identity can be determined through data linkage – and subsequently whether information is classified as 'personal information' and protected under State or Territory privacy legislation – are largely similar. This creates an opportunity for consistent additional guidance to be given on when an individual is to be understood as reasonably identifiable. For example, the NSW Civil and Administrative Tribunal has applied a test of whether 'more than moderate steps [are] necessary to match data from different sources in order to ascertain an individual's identity'.[13] There is a regulatory opportunity to clarify how consistently 'reasonable identifiability' will be interpreted across the relevant Commonwealth and State and Territory legislation.

One of the concerns expressed by stakeholders was that uncertainty might impact upon willingness to share genomic information for research purposes. This was independent of concerns that there may be inconsistent conditions attached to sharing for research purposes across Australia.

3.3.3 IS GENOMIC INFORMATION 'SENSITIVE' OR 'HEALTH INFORMATION' OR BOTH?

Under the *Privacy Act*, genetic information which is 'personal information' is classified also to be sensitive information.[14] This includes both genetic information that is health information[15] and genetic information about an individual that is not otherwise health information. Sensitive information is provided more robust legislative protection than other kinds of personal information under the *Privacy Act*.[16]

Each State and Territory has its own privacy legislation, apart from Western Australia (WA) and South Australia.

Relevant legislation in Queensland currently makes no specific mention of genetic or genomic information. Genomic information would be regulated only so far as it fell within broader categories of personal information[17] or confidential information.[18] New South Wales,[19] the Northern Territory,[20] Tasmania[21] and Victoria[22] all expressly include genetic information within the legislative definition of health information.

In WA, limited privacy protection is provided through Acts such as the *Freedom of Information Act 1992* (WA), the *State Records Act 2000* (WA) and the *Health Services Act 2016* (WA). The *States Records Act 2000* (WA) places controls on access to records containing information about a person's medical condition but does not expressly mention genetic or genomic information, nor does the *Health Services Act 2016* (WA). In South Australia, the Cabinet Administrative Instruction 1/89 sets out Information Privacy Principles that are binding on the public sector. The *Health Care Act 2008* (SA) also deals with confidentiality and disclosure of information. Neither specifically mentions genetic information.

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13 *AIN v Medical Council of New South Wales* [2016] NSWCATAD 5 at [39] – [44]. While this decision was applied in the context of the *Privacy and Personal Information Protection Act 1998* (NSW), this piece of legislation and the NSW Health Records and Information Act 2002 adopt an identical definition of 'personal information'.

14 *Privacy Act* (n 4) s 6(c) (definition of 'sensitive information').

15 Ibid s 6FA(d): "genetic information about an individual in a form that is, or could be, predictive of the health of the individual or a genetic relative of the individual"

16 *Information Privacy Act* (Qld), s 12

17 *Hospital and Health Boards Act 2011* (Qld), s 139

18 See *Privacy Act* (n 4) s 6FA(d). See also McWhirter, Johnston and Burke (n 4) 109.

19 Genetic information is specifically regulated by section 4 and section 6(d) of the *Health Records and Information Privacy Act 2002* (NSW) which together provide a definition of 'genetic information'. Section 5 also includes genetic information explicitly as a form of 'personal information'.

20 Genetic information is specifically mentioned in section 3(d) of the *Information Act 2002* (NT) and is regulated as 'health information'.

21 Genetic information is specifically mentioned in section 3(d) of the *Personal Information Protection Act 2004* (Tas) and is regulated as 'health information'.

22 Genetic information is specifically mentioned in section 3(d) of the *Health Records Act 2001* (Vic) and is regulated as 'health information'.

It is to be noted that even if genetic information is expressly mentioned by privacy legislation, where the material scope of legislation is bounded by the concept of personal information, then genomic information will only be regulated where it also satisfies the definition of personal information. Not all genomic information will be personal information, hence only some may fall within the scope of relevant statutory definitions even if genetic information is specifically mentioned.

TABLE 1: GENETIC INFORMATION IN PRIVACY LEGISLATION

JURISDICTION	ACT/SECTION	SUMMARY
CTH	*Privacy Act 1988* (Cth).[23]	Genetic information isspecifically mentioned in s 6(1) (under 'sensitive information'), s 6FA ('health information'), s 16B(4) ('use and disclosure') and s 95 AA as: whether or not also and health information, and may be subject to guidelines approved under s 95AA.
ACT	*Health Records (Privacy and Access) Act 1997*	***Genetic information is not specifically mentioned.***
NT	*Information Act 2002* (NT). Section 4 (definition of 'health information')	Genetic information is specifically mentioned in s 3(d) of the *Information Act 2002* (NT) and is regulated as 'health information'.
NSW	*Health Records and Information Privacy Act 2002* (NSW).[24] Section 4, Section 5, Section 6	Genetic information is specifically regulated by s 4 and s 6(d) of the *Health Records and Information Privacy Act 2002* (NSW) which together provide a definition of 'genetic information'. Section 5 also specifically notes that personal information includes such things as 'genetic characteristics'.
QLD	*Hospital and Health Boards Act 2011* (Qld)	***Genetic information is not specifically mentioned.***
SA	*Health Care Act 2008* (SA)	***Genetic information is not specifically mentioned.*** Although the power of an incorporated hospital to carry out testing for the purposes of determining parentage or other human genetic relationship is established (s 31(1a)(e))
TAS	*Personal Information Protection Act 2004* (Tas).	Genetic information is specifically mentioned in s 3(d) of the *Personal Information Protection Act 2004* (Tas) and is regulated as 'health information'.
VIC	*Health Records Act 2001* (Vic).	Genetic information is specifically mentioned in s 3(d) of the *Health Records Act 2001* (Vic) and is regulated as 'health information'.
WA	*Health Services Act 2016* (WA)	***Genetic information is not specifically mentioned.***

3.3.4 WHAT ARE THE CONDITIONS OF RESEARCH ACCESS TO GENOMIC DATA HELD IN CLINICAL RECORDS?

Chapter 2 addresses access to genomic information for research purposes, including through waivers of consent approved by Human Research Ethics Committees. As discussed in that chapter, given the structure of Australian privacy laws, the answer to this question will depend on where the practitioner works (eg, a public or private institution) and the jurisdiction in which they operate. It should be

23 *Privacy Act* (n 4) s 6(c) (definition of 'sensitive information'), s 6FA, s 95AA.

24 *Health Records and Information Privacy Act 2002* (NSW) s 4 (definition of 'genetic information') s 6.

noted that disclosure to researchers located outside of Australia will be governed by an added layer of regulation under APP 8, relating to disclosure of personal information outside of Australia.

3.3.5 FAMILIAL ISSUES

In accordance with individual privacy interests, disclosure of personal genomic information without the consent of the individual to whom the information relates should be avoided where possible. However, the familial nature of genomic information means that in the event that a person refuses permission to disclose, health practitioners can be faced with an ethical and legal tension between their duties of confidentiality and disclosure for the benefit of an at-risk relative. Genomic information will often relate to more than one person and its shared nature can give rise to questions regarding whether it is best conceived ethically as data relating to an individual or a family. Where data is considered sufficiently significant to an at-risk relative, but a person serving as the starting point for the genetic study of a family declines consent to disclosure, disclosure may still be legally permitted. However, the lack of a consistent standard for when disclosure without consent is permissible, uniform across all Australian jurisdictions, has been noted to be an issue of concern.

Currently, the disclosure of genomic information to at-risk relatives is governed by either Commonwealth or State or Territory legislation as well as common law obligations of confidentiality. Applicability is contingent on the nature of the entity gathering the information and the jurisdiction in which this entity operates. All Australian jurisdictions regulate the disclosure of genomic information to at-risk relatives.[25] However, the regulatory approaches vary in scope and substance.

Section 16B of the *Privacy Act* applies to private sector organisations (and not government agencies)[26] and permits disclosure of genomic information to at-risk relatives where there is a reasonable belief that disclosure of the information is necessary to lessen or prevent a serious threat to the relative of the patient.[27]

Section 16B(4) specifies that disclosure will be permissible where:

(a) the organisation has obtained the information in the course of providing a health service;

(b) the organisation reasonably believes there is a serious threat to life, health or safety of a genetic relative of the individual to whom the genetic information relates;

(c) the use or disclosure is necessary to lessen or prevent that threat;

(d) in the case of disclosure, the recipient of the information is a genetic relative of the individual to whom the genetic information relates; and

(e) the use or disclosure is conducted in accordance with the NHMRC Guidelines.

Any disclosures made under s 16B must also comply with guidelines issued by the National Health and Medical Research Council ('NHMRC') and approved under section 95AA of the *Privacy Act* (s 95AA Guidelines).[28]

Table 2 lists the s 95AA Guidelines.

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25 *Health Records (Privacy and Access) Act 1997* (ACT) sch 1; *Health Records and Information Privacy Act 2002* (NSW) sch 1, s 10; *Information Act 2002* (NT) sch 2; *Information Privacy Act 2009* (Qld) sch 4; *Health Care Act 2008* (SA) s 93; *Personal Information Protection Act 2004* (Tas) sch 1; *Health Records Act 2001* (Vic) sch 1; *Health Service Act 2016* (WA), s 220 in conjunction with the *Health Services (Information) Regulations 2017* (WA).

26 This is different from comparable provisions in State legislation available to health professionals working in the public setting. For a table providing detailed comparison see Jane Tiller et al, 'Disclosing genetic information to family members without consent: Five Australian case studies' (2020) *European Journal of Medical Genetics* 63, 104035.

27 See *Privacy Act* (n 4) s 16B(4). Note also that s 16A recognizes general situations in which it is permitted for an APP entity to collect, use or disclose personal information. These include where it is unreasonable or impracticable to obtain the individual's consent and the entity reasonably believes that the collection, use or disclosure is reasonably necessary to lessen or prevent a serious threat to the life, health or safety of any individual, or to public health or safety.

28 *Privacy Act* (n 4) s 16B(4)(c). Discussed further below in relation to the question 'when is it legally permissible to disclose clinically actionable genomic results to family members without consent?'.

TABLE 2: SECTION 95AA GUIDELINES

GUIDELINE	SUMMARY
Guideline 1	Use or disclosure of genetic information without consent may proceed only when the authorising medical practitioner has a reasonable belief that this is necessary to lessen or prevent a serious threat to the life, health or safety of a genetic relative.
Guideline 2	Specific ethical considerations must be taken into account when making a decision about whether or not to use or disclose genetic information without consent.
Guideline 3	Reasonable steps must be taken to obtain the consent of the patient or his or her authorised representative to use or disclose genetic information.
Guideline 4	The authorising medical practitioner should have a significant role in the care of the patient and sufficient knowledge of the patient's condition and its genetic basis to take responsibility for decision-making about use or disclosure.
Guideline 5	Prior to any decision concerning use or disclosure, the authorising medical practitioner must discuss the case with other health practitioners with appropriate expertise to assess fully the specific situation.
Guideline 6	Where practicable, the identity of the patient should not be apparent or readily ascertainable in the course of inter-professional communication.
Guideline 7	Disclosure to genetic relatives should be limited to genetic information that is necessary for communicating the increased risk and should avoid identifying the patient or conveying that there was no consent for the disclosure.
Guideline 8	Disclosure of genetic information without consent should generally be limited to relatives no further removed than third-degree relatives.
Guideline 9	All stages of the process must be fully documented, including how the decision to use or disclose without consent was made.

The legislation as currently enacted in States and Territories is similar and provides an overall environment that permits conditional disclosure of genetic information to at-risk relatives. South Australia, Tasmania, Queensland, WA and Victoria all permit genetic information to be disclosed where necessary to lessen or prevent a serious threat to the life, health or safety of a person.[29] There is no requirement that the threat be imminent. NSW has a similar approach, but limits the disclosure to genetic information, which is specifically defined in the NSW legislation.[30] In 2012, NSW also introduced the *Health Legislation Amendment Act 2012* (NSW) which amended the *Health Information Privacy Act 2002* (NSW) to allow the NSW Information and Privacy Commissioner to approve the NSW Health Privacy Guidelines.[31] This brought the NSW approach largely in line with the approach at the Commonwealth level.[32]

29 See *Information Privacy Act* 2009 (Qld) sch 4, NPP2; *Hospital and Health Boards Act 2011* (Qld), s 147; *Health Care Act 2008* (SA) s 93(3); *Personal Information Protection Act 2004* (Tas) sch 1, s 1(d)(i); *Health Records Act 2001* (Vic) sch 1; *Information Act 2002* (NT) sch 2, s 2.1(d)(i); *Health Service Act 2016* (WA), s220 in conjunction with the *Health Services (Information) Regulations 2017* (WA).

30 *Health Records and Information Privacy Act 2002* (NSW) s 4(1) (definition of 'genetic information') and sch 1, s 11(1)(c1).

31 Information and Privacy Commission New South Wales, *NSW Genetic Health Guidelines: Use and disclosure of genetic information to a patient's genetic relatives: Guidelines for organisations in NSW* (Guideline, October 2014) <https://www.ipc.nsw.gov.au/nsw-genetic-health-guidelines-use-and-disclosure-genetic-information-patients-genetic-relatives-guidelines-organisations-nsw>. See Natalia Meggiolaro et al, 'Disclosure to Genetic Relatives without Consent – Australian Genetic Professionals' Awareness of the Health Privacy Law' (2020) 21(1) *BMC Medical Ethics* 13, 14.

32 Ibid 14; Otlowski (n 5) 337.

In Tasmania, the Health Department has endorsed the use of the NHMRC guidelines when interpreting Tasmanian legislation and provided guidance on how to interpret the NHMRC guidelines in the context of the Tasmanian Health Service.[33]

Tasmania, Queensland and Victoria also permit disclosure where there is a threat to a person's welfare.[34] Both the NT and ACT retain a requirement that the relevant threat be both serious *and imminent*.[35] While these differences seem small on their face, it has been suggested that they may introduce significant uncertainty for health care practitioners who move between jurisdictions or between private and public practice. As McWhirter, Johnston and Burke note:

> Small differences between jurisdictions create challenges for [practitioners] moving between jurisdictions... clinical geneticists from a private non-profit organisation in Victoria [may] have serviced regional patients through public hospitals in both Tasmania and NT. This means the geneticist needs to be familiar with Commonwealth, Tasmanian, Victorian and NT legislation, and be clear on when each would apply. There is potentially scope for further confusion, if a proband is seen through one organisation and their at risk relative, living interstate, is seen through a different organisation. This may be particularly challenging if either patient is resident in the NT, where the 'imminent' requirement is retained.[36]

Legislation at both Commonwealth and State and Territory levels is consistent in its approach to permitting disclosure to family members where non-disclosure would present a risk to their safety, health or wellbeing. However, disclosure to family members may be permissible in some States where the information does not present any material risk to the family members' safety, health or wellbeing. For example, in all States other than the NT and WA, disclosure may be made where a family member also acts as a carer for the patient or is otherwise responsible for the patient.[37] Some jurisdictions also permit disclosure for other reasons, such as compassionate grounds,[38] or the death of a family member.[39]

33 Tasmanian Health Service 'Disclosure of genetic information to at-risk relatives without patient consent' (Tasmanian Health Service: Guideline, July 2019. See also Jane Tiller et al, 'Disclosing genetic information to family members without consent: Five Australian case studies' (2020) *European Journal of Medical Genetics* 63, 104035.

34 *Information Privacy Act 2009* (Qld) sch 4, 2(1)(d); *Personal Information Protection Act 2004* (Tas) sch 1, 1(d)(i); *Health Records Act 2001* (Vic) sch 1, 2.2(h)(i).

35 See *Health Records (Privacy and Access) Act 1997* (ACT) sch 1, to 8-3"; Australian Government, *First Stage Response to ALRC Report 108* (Report, October 2009) 28.

36 McWhirter, Johnston and Burke (n 4) 113.

37 *Health Records (Privacy and Access) Act 1997* (ACT) sch 1, 10(4); *Health Care Act 2008* (SA) s 93(3)(c); *Personal Information Protection Act 2004* (Tas) sch 1, s 2(4); *Health Records Act 2001* (Vic) sch 1, s 2.4(a)(i); *Information Privacy Act 2009* (Qld) sch 4, s 4(3).

38 *Health Records Act 2001* (Vic) sch 1, s 2.4(a)(ii); *Personal Information Protection Act 2004* (Tas) sch 1, s 2(4)(b)(ii); *Health Records and Information Privacy Act 2002* (NSW) sch 1, s 11(g).

39 *Health Records Act 2001* (Vic) sch 1, s 2.5.

3.3.6 DECEASED PERSONS

The *Privacy Act* has no jurisdiction over deceased persons.[40] Consequently, the Act is silent on whether consent must be sought for genetic testing of deceased individuals.[41] Further, if consent *should* be sought, the *Privacy Act* provides no indication about from whom. While the ALRC recommended the *Privacy Act* be amended to include protection for deceased individuals,[42] this recommendation was not accepted by Government and no amendment to this effect has been made.[43]

There are indications from English Law that the common law duty of confidentiality survives the death of the patient.[44] This may prevent the disclosure of confidential information by a medical practitioner without a legal basis for disclosure. Further clarity would be desirable on the interaction between laws of privacy and confidentiality to ensure health professionals are aware of their responsibilities to deceased patients.

As with privacy protections in other areas, protections for deceased persons vary between States and Territories. Currently, the ACT, Queensland,[45] the NT, NSW, Victoria and Tasmania have some form of privacy protection for deceased persons (Table 3).

South Australia and WA do not apply statutory protections to data relating to the deceased.

Despite the fact that the privacy legislation in most States and Territories affords deceased individuals with some protection, there is not a clearly consistent national position with regards to the circumstances under which a genomic sample previously obtained from a living person now deceased may be tested, including the process by which any consent should be obtained, and from whom. The removal of tissue from the deceased, for purposes including medical or scientific purposes, is typically regulated consistent with the ALRC Recommendations on Human Tissue Transplants.[46]

Jurisdictions also vary on rights to access previously collected information about deceased persons. Under Commonwealth legislation as well as the privacy regimes of most States and Territories, the relatives of the deceased do not have a clear right under the Access principle to access the data of the deceased. The key exceptions are Victoria and the ACT, where the living relatives of a deceased person are afforded a right to access the data of their deceased relative.[47]

40 See Stephen Argument, 'Do the Deceased have a Right to Privacy?' (2006) 3(2) *Privacy Law Bulletin* 20, 20.

41 National Health and Medical Research Council, 'Use and disclosure of genetic information to a patient's genetic relatives under Section 95AA of the Privacy Act 1988 (Cth) – Guidelines for health practitioners in the private sector' (NHMRC, 2014) 1, 5.

42 Australian Law Reform Commission, *For Your Information: Australian Privacy Law and Practice* (ALRC Report 108) (2008) Recommendation 8–3.

43 'The Government acknowledges that there are arguments both for and against extending privacy protections to personal information about deceased persons where held by organisations. Having taken further advice on this issue, the Government is aware of the significant constitutional limitations on the Commonwealth's power to legislate in this area. The Government therefore does not accept the ALRC's recommendations 8-1' Australian Government, 'Enhancing National Privacy Protection: Australian Government First State Response to the Australian Law Reform Commission Report 108 (October, 2009), 28.

44 *Lewis v Secretary of State for Health* [2008] EWHC 2196; *Bluck v The Information Commissioner* (2007) 98 BMLR 1; for commentary see Taylor MJ 'Confidentiality and Data Protection' in Laing, McHale, Kennedy and Grubb (eds) *Principles of Medical Law* (4th Edition) (OUP, 2017) s 12.76 – 12.79.

45 However, note that the definition of personal information contained within the *Information Privacy Act 2009* (Qld) (s 12) limits it to information about an individual. The definition of 'individual' in the *Acts Interpretation Act 1954* (Qld) is 'a natural person', which does not include a deceased person.

46 Australian Law Reform Commission and Australian Health Ethics Committee, *Essentially Yours: The Protection of Human Genetic Information in Australia* (ALRC Report 96, 28.07.10), Chapter 19 'Human Tissue Collections; Australian Law Reform Commission, Human Tissue Transplants (ALRC Report No 7, November 1977).

47 *Health Records (Privacy and Access) Act 1997* (ACT) s 27(2); *Health Records Act 2001* (Vic) s 95(2).

TABLE 3: PRIVACY PRINCIPLES AND THE DECEASED

ACT	*Health Records (Privacy and Access) Act 1997* (ACT).	Section 27 – Deceased Consumers (1) The privacy principles apply in relation to a deceased consumer, *so far as they are reasonably capable of doing so*, in the same way as they apply in relation to a consumer who is not deceased[48]
NT	*Information Act 2002* (NT).	Section 4 – Definitions 'person' means, subject to s 4B, an individual and includes a deceased individual *within the first 5 years* after death
TAS	*Personal Information Protection Act 2004* (Tas).	Section 3A – Reference to personal information in Part 3A and Schedule 1 Personal information means any information or opinion in any recorded format about an individual ... *'who is alive or has not been dead for more than 25 years'* (s 3)
VIC	*Health Records Act 2001* (Vic).	Section 95 – Deceased individuals (1) This Act applies in relation to a deceased individual who has been dead for *30 years or less*, so far as it is reasonably capable of doing so, in the same way as it applies in relation to an individual who is not deceased
QLD	*Hospital and Health Boards Act 2011* (QLD)	Part 7 – Division 2 Confidentiality Prohibited disclosure of confidential information 'applies even if the person who could be identified is deceased'
NSW	*Health Records and Information Privacy Act 2002* (NSW)	Section 5(3) Personal information does not include any of the following: (a) information about an individual who has been dead for more than 30 years

48 It is relevant to note that it is permissible to delete or destroy health records under privacy principle 4.1 if the consumer is under 18 years old when the information is collected—the day the consumer turns 25 years old; or if the consumer is an adult when the information is collected—7 years after the day a service was last provided to the consumer by the record keeper; *Health Records (Privacy and Access) Act (ACT)* 1997, 1.

Case Study: Data Sharing

A National Repository?

Large-scale genomic data repositories facilitate timely, high-quality research, including a better understanding of population genomic diversity and analysis of phenotype–genotype relationships. There are efficiency benefits associated with centralised repositories, and there is evidence that the Australian public are more likely to trust an Australian repository with their data than an overseas one. Data for a such a repository can be obtained through clinical sequencing as well as genomic research data. The value of genomic data is further enhanced when combined with phenotypic and other data. However, collecting, storing, and sharing these data raise key legal and policy issues. How would an Australian genomic data repository operate under current regulatory conditions?

Implications for Consent

A threshold issue is the requirements for consent for the transfer of data to any repository. To the extent that information is identified or 'reasonably identifiable', consent for such a transfer will be required under Commonwealth, State and Territory privacy laws. Given that the most useful data repositories will include extensive clinical information—as well as the growing ease of reidentifying individuals based on de-identified datasets— this is likely to be the case for many and perhaps most genomic sequencing data collections. Maintaining identifiers also allows for the future return of clinically valuable findings from research using repository data.

At least arguably, a data repository could collect deidentified genomic sequencing data without individual consent—for example, as an automated step following on from an individual undergoing clinical genomic sequencing. Given the information is deidentified (provided the depth of genomic sequencing and/or associated clinical data is not inherently reidentifying), privacy laws are not applicable to the transfer.

Any transfer of data without consent for the purpose of future research would require a waiver of consent by a Human Research Ethics Committee ('HREC').

This will depend on satisfying the requirements of paragraph 2.3.10 of the *National Statement on Ethical* Conduct in Human Research ('*National Statement*').[1]

This requires that, in the determination of a properly constituted HREC:

(a) involvement in the research carries no more than low risk to participants

(b) the benefits from the research justify any risks of harm associated with not seeking consent

(c) it is impracticable to obtain consent (for example, due to the quantity, age or accessibility of records)

(d) there is no known or likely reason for thinking that participants would not have consented if they had been asked

(e) there is sufficient protection of their privacy

(f) there is an adequate plan to protect the confidentiality of data

1 Australian Government National Health and Medical Research Council, *National Statement on Ethical Conduct in Human Research* (2007).

(g) in case the results have significance for the participants' welfare there is, where practicable, a plan for making information arising from the research available to them (for example, via a disease-specific website or regional news media)

(h) the possibility of commercial exploitation of derivatives of the data or tissue will not deprive the participants of any financial benefits to which they would be entitled

(i) the waiver is not prohibited by State, federal, or international law.

At least for prospective genomic sequencing data, it would be challenging for a national data repository to satisfy the threshold of being 'impracticable to obtain consent'.

It is also questionable whether making sequencing data widely available to researchers nationally and internationally would satisfy the criterion of 'no known or likely reason for thinking that participants would not have consented if they had been asked'.

This would be especially problematic if data from the repository is shared with industry, given empirical research reporting commercialisation as a key concern for potential biobank participants.[2]

Requirements for Consent

Accordingly, a clear and informed consent would be needed for genomic sequencing data and any associated clinical information. (It is worth noting that under paragraph 3.3.15 of the *National Statement*, an opt-out approach should not be used in genomic research.) The consent would need to address the criteria set out in paragraph 3.3.10 of the *National Statement*:

(a) what information will be generated by the research;

(b) what may be discovered by the research;

(c) what will be deliberately excluded from the scope of the research;

(d) which, if any, of the findings of the research will be communicated to participants and, if so, how;

(e) what the health implications are of the information for participants and their relatives;

(f) whether there are any other implications for participants and their families of being given this information (e.g. insurance, employment, social stigma);

(g) the potential for the information generated by or used in the research to result in participants being re-identified;

(h) whether information generated by the research will be shared with other research groups; and

(i) potential future use of information and biospecimens, including commercial applications.

2 Dianne Nicol et al, 'Understanding Public Reactions to Commercialization of Biobanks and Use of Biobank Resources' (2016) 162 *Social Science & Medicine* 79

Subsequent Sharing Following Consent

If the above requirements for consent are satisfied, there are unlikely to be barriers to sharing data across jurisdictions or internationally subject to any local legislative requirements.

Sharing data with industry also could be addressed through the consent process; however, given known sensitivities with such sharing, additional oversight strategies are suggested below. These are designed to promote and maintain public trust in use of repository data.

Beyond Consent

Beyond the necessarily limited information conveyed at the time of initial consent, additional oversight and communication about the projects for which data has been released would be important to ensure the repository's ongoing ethical acceptability. Public disquiet stemming from a lack of such community consultation and transparency were well illustrated through the care. data controversy in England.[3]

Strategies could involve, for example, a Data Access Committee with broad-based membership, including expertise in law, ethics, and science, along with community representation on the Committee itself or through a Community Advisory Board.[4] The Confidentiality Advisory Group that operates in England and Wales provides another model for such oversight and communication.[5]

3 Mark Taylor, 'Information Governance as a Force for Good - Lessons to Be Learnt from Care.Data' (2014) 11(1) *SCRIPTed: A Journal of Law, Technology and Society* 1.

4 Christine Grady et al, 'Broad Consent for Research With Biological Samples: Workshop Conclusions' (2015) 15(9) *The American Journal of Bioethics* 34.

5 NHS Health Research Authority, 'Confidentiality Advisory Group', Health Research Authority <https://www.hra.nhs.uk/about-us/committees-and-services/confidentiality-advisory-group/>.

4 Aboriginal and Torres Strait Islander Identity and Genomics

4.1 Summary

The use of genomics in healthcare and research raises risks for indigenous populations, arising from consultation failures, lack of informed consent, underrepresentation, and inappropriate use of samples and data. These issues are relevant to many socially identifiable groups, but are particularly acute for indigenous peoples in settler-colonial states. Awareness of these issues is reflected in the strategic priorities in the *National Health Genomics Policy Framework*.[1] The concerns raised by Aboriginal and Torres Strait Islander people about genomics – relating to identity, group interests and equity[2] – are difficult to address through purely legal means. Nevertheless, these concerns intersect with existing regulatory frameworks in complex ways, and need to be accounted for in any reform efforts.

Three conclusions are clear from an analysis of stakeholder concerns within the relevant regulatory frameworks. First, caution should be exercised in any discussions regarding a potential role for genetic testing in establishing Aboriginal and Torres Strait Islander identity. Second, current regulatory frameworks inadequately account for group interests. Third, achieving equity in genomics will be complex, requiring widespread uptake of culturally appropriate protocols in both clinical and research contexts, supported by Aboriginal and Torres Strait Islander governance and management of genomic resources.

4.2 Genomics in Healthcare and Research with Aboriginal and Torres Strait Islander People

Any significant development in the way healthcare is delivered needs to be implemented in a way that recognises and accounts for its implications for Australia's First Peoples. As a result of a combination of historical and sociological factors, Aboriginal and Torres Strait Islander people experience higher rates of chronic disease, worse health outcomes, and lower life expectancy than non-Indigenous Australians.[3] If Aboriginal and Torres Strait Islander people are excluded from the benefits of genomic medicine, the widespread integration of genomics into clinical care risks exacerbating existing health disparities.[4]

1 Australian Government Department of Health, Supplementary Information to the *National Health Genomics Policy Framework* (at 2017) Strategic Priorities 1.5, 1.8, 2.1, 2.2 and 5.2.

2 For other issues also of relevance to Aboriginal and Torres Strait Islander people, see Chapter 6 Controversial Secondary Uses; section 9.3.1.2 Proprietary rights in genomic data; section 9.3.4 Custodianship and sovereignty; and section 10.2 Identifiable versus Deidentified Data.

3 Australian Government, Closing the Gap 2020 (Report, 2020); Australian Health Ministers' Advisory Council, *Aboriginal and Torres Strait Islander Health Performance Framework 2017* (Report, 30 May 2017).

4 Rebekah McWhirter, Dianne Nicol and Julian Savulescu, 'Genomics in Research and Health Care with Aboriginal and Torres Strait Islander Peoples' (2015) 33(2-3) *Monash Bioethics Review* 203.

Inclusion will depend upon trust in genomics and data governance being established and maintained in both the research and clinical contexts. Achieving this will require overcoming a history of poor research practices, in Australia and internationally, in which researchers used genomic samples with inadequate consent, for inappropriate purposes, and in ways that contributed to damaging stereotypes, exploitation or cultural undermining.[5] It will also require the specific concerns of Aboriginal and Torres Strait Islander people in relation to genomics being addressed.

In addition to the issues arising from the use of genomics in healthcare and research that affect all Australians, there are a number of issues that are of particular concern to Aboriginal and Torres Strait Islander people. In particular, stakeholders interviewed as part of the consultation process for the Scoping Report highlighted issues associated with using genomics to determine identity; issues related to representing group interests in genomic data governance; and issues associated with equity in the use of genomics in healthcare and research.

4.3 Defining Aboriginal and Torres Strait Islander Identity

Defining Aboriginal and/or Torres Strait Islander identity is a sensitive issue with significant implications for individuals and communities. Stakeholders interviewed as part of the consultation identified two key concerns in this respect: the potential for genomics to influence personal identity as an Aboriginal or Torres Strait Islander person; and the potential for genomics to be used to limit rights or access to services.[6]

4.3.1 LEGAL DEFINITION OF ABORIGINAL OR TORRES STRAIT ISLANDER IDENTITY

Historically, Aboriginal or Torres Strait Islander descent was the key consideration in determining identity, and early tests focused on 'quantum of blood'-style definitions to the exclusion of other criteria.[7] In the 1980s, the then Commonwealth Department of Aboriginal Affairs undertook a review of the definition of Aboriginal and Torres Strait Islander, proposing a new tripartite definition:

> An Aboriginal or Torres Strait Islander is a person of Aboriginal or Torres Strait Islander descent who identifies as an Aboriginal or Torres Strait Islander and is accepted as such by the community in which he [or she] lives.[8]

The three criteria of descent, self-identification, and community recognition have been widely adopted. No weight is placed on the proportion attributable to Aboriginal or Torres Strait Islander heritage. Government departments continue to use this approach as their working definition for assessing eligibility for access to programs and services reserved for Aboriginal and Torres Strait Islander people. It has further been applied on a number of occasions by the High Court, including in relation to s 51(xxvi) of the Constitution,[9] in relation to native title,[10] and – most recently – when considering whether Aboriginal Australians are within the reach of the 'aliens' power in s 51(xix) of the Constitution.[11]

5 Michael Dodson and Robert Williamson, 'Indigenous Peoples and the Morality of the Human Genome Diversity Project' (1999) 25 *Journal of Medical Ethics* 204; Nanibaa' Garrison et al, 'Genomic Research Through an Indigenous Lens: Understanding the Expectations' (2019) 20 *Annual Review of Genomics and Human Genetics* 495.

6 Centre for Law and Genetics, and Centre for Health Law and Emerging Technologies, *Assessment of Legislation and Regulations Applying to the Collection and Use of Health-Related Genomic Information: Scoping Review* (2020) 56.

7 Australian Law Reform Commission, *Recognition of Aboriginal Customary Laws* (ALRC Report 31, 1986) 89.

8 Department of Aboriginal Affairs, *Report on a Review of the Administration of the Working Definition of Aboriginal and Torres Strait Islanders* (Report, 1981).

9 *Commonwealth v Tasmania* (1983) 158 CLR 1, 273-4 (Deane J).

10 *Mabo v Queensland* (No 2) (1992) 175 CLR 1, 70 (Brennan J).

11 *Love v Commonwealth* [2020] HCA 3, 25-6 (Bell J) and 165-6 (Edelman J).

The difficulties of establishing Aboriginal descent were explored thoroughly in *Shaw v Wolf*,[12] in which extensive use was made of archival sources and family history materials. The absence or unreliability of written records can make it difficult to establish descent. The problems 'have been exacerbated', as Merkel J noted, 'by the tragic historical fact that actual or perceived racism was such that many Aboriginal persons regarded their Aboriginal identification and public recognition of it with shame and as a distinct disadvantage.'[13] Indeed, in *Gibbs v Capewell*,[14] Drummond J concluded that, given the challenges associated with proving descent, community recognition would sometimes be the best available proof of descent. However, this raises particular challenges for members of the Stolen Generation, who were removed from their families by the state, resulting in dislocation from community and consequent difficulty in establishing evidence of meeting the tripartite test.

The use of genetic testing as a tool to 'prove' Aboriginality has been raised as an issue on several occasions. In 2002, debate over who was eligible to vote in Aboriginal and Torres Strait Islander Commission elections in Tasmania led to some potential voters resorting to genetic testing to provide evidence of descent from colonial-era individuals known to be Aboriginal.[15] More recently, One Nation politician Mark Latham called for self-identification to be replaced by a system requiring genomic testing and a result of at least 25 per cent 'Indigenous' for Aboriginal or Torres Strait Islander identity to be accepted.[16] Both occasions attracted significant controversy, and were heavily criticised on cultural, scientific, and ethical grounds. Notably, in *Essentially Yours*, the ALRC and AHEC expressed scepticism over 'whether there is any proper role for genetic testing in determining Aboriginal identity, which is basically a social and cultural matter.'[17]

4.3.2 SELF-DETERMINATION AND IDENTITY

Article 33 of the *United Nations Declaration on the Rights of Indigenous Peoples* recognises the right of Indigenous peoples 'to determine their own identity or membership in accordance with their customs and traditions.'[18] The tripartite test in current use was not developed by Aboriginal and Torres Strait Islander people, but has nevertheless gained widespread acceptance.

Significantly, this definition has been adopted within Aboriginal and Torres Strait Islander institutions. The National Aboriginal Community Controlled Health Organisation, for example, uses the three-part definition and describes it as 'the only acceptable definition of Aboriginality.'[19] Stakeholders reported being content with the current definition and its application, and expressed concern that if genetic testing were to be used to provide evidence for the 'descent' limb of the test, that this 'scientific' evidence may overwhelm the other elements of identity, and undermine rights to self-determination, essentially causing a return to 'blood quantum' style definitions of Indigeneity. As stakeholder 60 reported:

> The notion of Aboriginality and what does genetics tell us, if anything, about being Aboriginal is a genuine concern that people have expressed on multiple occasions to me. There's a sense there's been enough damage done already by misunderstandings and misuse around the concept of Aboriginality and who belongs.

12 [1998] FCA 389.

13 Ibid.

14 [1995] FCA 25, 585.

15 'Blood lines called into question' *Sydney Morning Herald* (online, 26 August 2002) <https://www.smh.com.au/national/blood-lines-called-into-question-20020826-gdfkpf.html>.

16 Greg Dunlop and Jack Latimore, 'One Nation wants Aboriginal people to 'prove' ancestry with DNA tests' *NITV News* (online, 11 March 2019) <https://www.sbs.com.au/nitv/article/2019/03/11/one-nation-aboriginal-indigenous-dna>.

17 Australian Law Reform Commission and Australian Health Ethics Committee, *Essentially Yours: The Protection of Human Genetic Information in Australia* (ALRC Report 96, 2003) 36.5 ('Essentially Yours').

18 61/295, 61st session, UN Doc A/RES/61/295 (2 October 2007) 33.1.

19 National Aboriginal Community Controlled Health Organisation, *Definitions* (Web Page, May 2020) <https://www.naccho.org.au/acchos>.

Further, stakeholder 41 said:

> I hope the government won't take this [genetic results] into account, and respect the fact Aboriginal people have their own way of determining Aboriginality, which does not come from having a percentage. This is what I am really worried about.

Similarly, stakeholder 51 observed:

> Aboriginal people have to prove their Aboriginality when other people don't have to prove who they are ... The percentage is a white obsession and it has been there right from colonial times ... Aboriginal people do not care, it's about connection and relationships.

Despite the well-recognised right of Indigenous peoples to self-determination, the fact that a number of legislative instruments restrict eligibility for services, programs or applications under legislative schemes such as Native Title means that administrators and courts will inevitably be called upon to determine Aboriginal and Torres Strait Islander status. Although international law is of limited force at the domestic level, here it highlights the importance of facilitating Aboriginal and Torres Strait Islander leadership in determining both the appropriate legal tests for determining Aboriginal and/or Torres Strait Islander identity, and the evidence accepted in establishing that an individual meets the test.

4.3.3 LIMITATIONS OF THE TECHNOLOGY

In addition to ethical and legal reasons, there are technical reasons to limit the weight given to genomic evidence in determining Aboriginal and Torres Strait Islander identity.

Genomic analysis can identify whether two individuals are closely related. For this reason, genomic methods may be of use to individuals who have lost their connection with their biological families, such as through adoption or through the state-sanctioned removal of Aboriginal and Torres Strait Islander children from their families, known as the 'Stolen Generations'. Link Up organisations working to reunite families, however, currently focus on traditional family history methods and archival resources.

Establishing a connection through genomic methods is more challenging if the two individuals being compared are distantly related. The proportion of the genome inherited from particular ancestors varies and, after a handful of generations, it is possible for no evidence of shared DNA to be present. Further, establishing a genomic connection between two contemporary individuals may not provide evidence of Aboriginal or Torres Strait Islander descent if they, like most people, are of admixed heritage. This means that genetic testing is not an infallible means of determining descent. This is particularly relevant in the Australian context, where the proportion of ancestry is not determinative, and the identified ancestor may be several generations prior.

Determining an individual's ethnicity using genetic testing is not as clear-cut or definitive as the results presented by commercial genetic ancestry tests make them seem. An individual can be compared to different populations to ascertain whether their genome resembles the patterns of genetic variation of particular groups of people, but this is not determinative, and the limits of genomic measures of 'ancestry' are well documented.[20] Its utility in this latter context will depend heavily on the nature and quality of the reference dataset to which the individual is being compared. The genomic differences between the many Aboriginal and Torres Strait Islander peoples across the continent are estimated to be as great as those between populations across Europe and Asia combined.[21] A reference dataset that only included Aboriginal individuals from Arnhem Land, for example, would be unlikely to correctly assign ancestry for an Aboriginal person from Victoria.

20 See, for example, Mwenza Blell and M Hunter, 'Direct-to-Consumer Genetic Testing's Red Herring: "Genetic Ancestry" and Personalized Medicine' (2019) 6 *Frontiers in Medicine* 48; Anne Huml et al, 'Consistency of Direct-to-Consumer Genetic Testing Results Among Identical Twins' (2020) 133 *American Journal of Medicine* 143.

21 Simon Easteal et al, 'Clinical and Population Genomic Data for Aboriginal and Torres Strait Islander Peoples is a Pre-requisite for Equitable Provision of Expanded Carrier Screening in Australia' (2020) 107(2) *American Journal of Human Genetics* 175.

This has proved to be a limiting factor for direct-to- consumer ('DTC') ancestry tests. Currently, the majority of DTC genetic ancestry tests do not include an assessment of Aboriginal or Torres Strait Islander ancestry, owing largely to a lack of reference data. One company offered an 'Aboriginality Report' based on genetic analysis, but this was withdrawn from public sale in September 2019 following an investigation by the Australian Competition and Consumer Commission into possible misleading and deceptive conduct under s 18 of the *Australian Consumer Law*.[22] In their public apology, the company outlined the three criteria used by Government agencies to determine Aboriginality and noted that: 'None of these criteria can be determined by DNA testing'.[23]

In summary, genomic evidence may be useful for demonstrating a positive connection between two individuals, or between an individual and a reference dataset, but could not be used to conclusively determine whether an individual was or was not of Aboriginal or Torres Strait Islander descent.

Although increased inclusion of Aboriginal and Torres Strait Islander people in genomic research will improve reference datasets, this will be primarily of clinical, rather than genealogical benefit. This is consistent with the ALRC and AHEC's finding, in *Essentially Yours*, that Aboriginal identity was 'basically a social and cultural matter' rather than a matter to be determined through genetic testing.[24]

4.4 Group Interests

Around the world, the experiences of Indigenous peoples with genomic research highlight the risk of group harms resulting from the misuse of genomic data. For example, genomic samples and data from members of the Havasupai Indian tribe were originally collected for diabetes research, but were later used in secondary research into schizophrenia, inbreeding and population genetics, all of which were potentially stigmatising or undermined Havasupai cultural beliefs.[25] There are currently limited regulatory mechanisms for including group interests in decision-making processes around the collection, use and disclosure of genomic data. The two key mechanisms in use are individual consent and ethics review processes, both of which exhibit significant limitations in this respect.

4.4.1 CONSENT

The obtaining of informed consent is the key mechanism relied upon for the protection of the interests of both research participants and patients. Consent is relevant at a number of points: at the time of the collection of the sample, prior to undertaking any genomic testing on the sample, and in relation to the use or disclosure of genomic data.

In the legal context, consent based on a general description of the process will be sufficient to avert a claim of battery, although more detailed information may be required to satisfy the duty to warn of a material risk, arising from either the collection process or the subsequent use of the sample, in order to prevent a claim in negligence.[26]This gives rise to additional considerations for genetic counsellors working with Aboriginal and Torres Strait Islander people, given the increased risk of their results being uninterpretable in the absence of appropriate reference data.

22 *Competition and Consumer Act 2010* (Cth) sch 2.

23 GTLDNA, *Ancestry Report* (Web Page) <https://www.gtldna.com.au/dna-ancestry-tests/>. Note that this public apology was only required to be posted for 30 days and is no longer available.

24 *Essentially Yours* (n 17), 36.5.

25 Michelle Mello and Leslie Wolf, 'The Havasupai Indian Tribe Case – Lessons for Research Involving Stored Biologic Samples' (2010) 363 *New England Journal of Medicine* 204.

26 *Rogers v Whitaker* (1992) 175 CLR 479, 490.

The focus on individual consent is replicated in privacy and human tissue legislation regulating the collection, use and disclosure of samples and data, as also discussed in Chapters 2 and 3.[27]

An individual's consent is usually taken as determinative, with very little opportunity for the interests of others to be represented. While clinical guidelines recognise that it is often culturally appropriate to include family in planning and decision-making with Aboriginal and Torres Strait Islander patients, there are systemic barriers limiting this in practice.[28]

While an individual's genomic data is uniquely theirs, it also has implications for their relatives and community, as is evident from the group harms experienced by Indigenous peoples. Under the current regulatory frameworks, an individual is free to consent to any use of their genomic data without reference to any other person and without consideration of potential group harms. One possible exception to this was raised in the stakeholder consultation, by a stakeholder who reported that a collection of historic samples, for which they had consent from individuals or next of kin, had been classified as 'objects' under the *Aboriginal Heritage Act 1988* (SA) and that other Aboriginal people or groups were therefore able to make representations objecting to the use of the samples for genomic analysis. It is not yet clear whether this will represent an example of collective interests trumping individual consent, but it gives an indication of one possible model for taking collective interests into account.

That the current dependence on individual consent is unable to account for collective interests or protect against group harms represents a significant gap in the regulation of genomic samples and data, and one of particular significance to Aboriginal and Torres Strait Islander peoples.

4.4.2 ETHICAL GUIDELINES

Individual consent is as central to the protection of participant interests in research settings as it is to patients receiving clinical care. However, research guidelines explicitly recognise that in some circumstances it is appropriate for others to be involved:

> Within some communities, decisions about participation in research may involve not only individuals but also properly interested parties such as formally constituted bodies, institutions, families or community elders. Researchers need to engage with all properly interested parties in planning the research.[29]

Further, the *National Statement* recognises that:

> Genomic research is frequently considered to be greater than low risk, especially in the context of research involving Indigenous peoples. For this reason, relevant on-going community consultation and active agreement on the part of communities and traditional owners is an essential component of the planning and conduct of this research.[30]

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27 See, eg, *Privacy Act 1988* (Cth); *Health Records and Information Privacy Act 2002* (NSW); *Health Records Act 2001* (Vic); *Health Care Act 2008* (SA); Department of the Premier and Cabinet Circular, *Information Privacy Principles Instruction* (Cabinet Administrative Instruction PC012, 6 February 2017) <https://www.dpc.sa.gov.au/resources-and-publications/premier-and-cabinet-circulars/DPC-Circular-Information-Privacy-Principles-IPPS-Instruction.pdf> ('SA Information Privacy Principles'); *Personal Information Protection Act 2004* (Tas); *Health Records (Privacy and Access) Act 1997* (ACT); *Information Act 2002* (NT); *Information Privacy Act 2009* (Qld); *Transplantation and Anatomy Act 1979* (ACT); *Transplantation and Anatomy Act 1979* (NT); *Human Tissue Act 1982* (Vic); *Human Tissue and Transplantation Act 1982* (WA); *Human Tissue Act 1983* (NSW); *Transplantation and Anatomy Act 1983* (Qld); *Transplantation and Anatomy Act 1983* (SA); *Human Tissue Act 1985* (Tas).

28 See, eg, Australian Commission on Safety and Quality in Health Care, *National Safety and Quality Health Service Standards: User Guide for Aboriginal and Torres Strait Islander Health* (at 2017) 5, 33.

29 National Health and Medical Research Council, Australian Research Council and Universities Australia, *National Statement on Ethical Conduct in Human Research* (at 2018) s 2.2.13.

30 Ibid Ch 3.3.

Good genomic research involves proper community consultation as a minimum, community co-design is preferable, and community control through models such as Indigenous Data Sovereignty or community governance is best practice. The extent to which evidence of this sort of engagement is required will depend on the particular Human Research Ethics Committee (HREC) undertaking ethical review (see also Chapter 2). While some HRECs are very familiar with the appropriate methods for engaging with particular communities, this varies considerably across Australia, and no single HREC will be familiar with all communities. Some HRECs only review research relating to Aboriginal and Torres Strait Islander people, such as the Aboriginal Health and Medical Research Council HREC, or have Aboriginal and Torres Strait Islander sub-committees, whose members provide specialist review of research proposing to target Indigenous participants, while others have individual Aboriginal and Torres Strait Islander members, and still others have no Aboriginal and Torres Strait Islander representation at all. As part of the process of review, researchers may be required to demonstrate that they have the support of the community that they intend to include in their research. Research that includes visiting remote Aboriginal and Torres Strait Islander communities may require researchers to gain approvals from the relevant Land Councils and Health Boards, and these bodies may also wish to see evidence of community support for the project.

However, the guidelines lack detail on: what sort of evidence of community support is acceptable; from whom should community consent be sought; or how community input should be handled in urban settings, in cases where an individual is dislocated from their (genetic) community, or for projects that propose to include Aboriginal and Torres Strait Islander participants from across Australia. Further, Hudson and colleagues have criticised the dependence on HRECs on the basis that:

> The use of ethics review committees as sole arbiters of appropriate access to and use of Indigenous samples and/or genomic data is inconsistent with Indigenous rights and interests. Institutional ethics committees largely lack Indigenous representation, do not recognize or promote Indigenous research ethics principles and often fail to support Indigenous governance of research and data. Ethics review processes are limited in their ability to deal with increasing expectations for open access and secondary use.[31]

Neither individual consent nor ethics review are currently able to adequately represent Aboriginal and Torres Strait Islander interests or to protect against group harms arising from the use of genomics in research and healthcare, and this is a significant gap in the regulation of genomic data. In New Zealand, this problem is being addressed through frameworks for the governance and management of genomic resources that is led, co-curated and governed by Maori and Pacific representatives,[32] and similar discussions, founded in Indigenous Data Sovereignty (see Chapter 9) are emerging in Australia.

4.5 Equity

Issues of equity in relation to genomics in healthcare and research are relevant for many groups, and are exemplified by Aboriginal and Torres Strait Islander peoples in the context of colonial dispossession and ongoing marginalisation, and the effects of this context on health outcomes and life expectancy. The *National Health Genomics Policy Framework* identifies equity of access to the benefits of genomics, in both the clinical and research contexts, as a priority area for action. It is equally important that the risks and burdens of genomics do not disproportionately affect Aboriginal and Torres Strait Islander people.

31 Maui Hudson et al, 'Rights, Interests and Expectations: Indigenous Perspectives on Unrestricted Access to Genomic Data' (2020) 21 *Nature Review Genetics* 377, 378.

32 Stephen Roberson et al, 'Genomic Medicine Must Reduce, Not Compound, Health Inequities: The Case for Hauora-Enhancing Genomic Resources for New Zealand' (2018) 131 *New Zealand Medical Journal* 81.

To be effective, genomic medicine requires the sharing of genomic data from large cohorts, so that information about the identity and frequency of genetic variants can be used to facilitate the interpretation of individual results for clinical care. To date, most available data have been collected from people of European ancestry.[33] Although progress has been made in recent years towards improving the representation of Asian and, to a lesser extent, African populations, the representation of Indigenous peoples (including Aboriginal and Torres Strait Islander peoples) has actually decreased. Although many of the findings made in European cohorts will be relevant to all people, there are also many differences between populations that will be clinically relevant. The current lack of evidence will lead to clinical genomics returning more likely-pathogenic variants and variants of unknown significance for Aboriginal and Torres Strait Islander peoples than for non-Indigenous Australians, which has the potential to lead to inappropriate clinical intervention when incorrectly interpreted.[34] Improving representation in research will be critical to equity in clinical care, and projects aimed at developing a reference genome for Aboriginal or Torres Strait Islander people, such as that being undertaken by the National Centre for Indigenous Genomics, are an important step in this process.[35]

Law plays only a minor role in ensuring diversity in genomic research. The *Racial Discrimination Act 1975* (Cth) prohibits:

> any act involving a distinction, exclusion, restriction or preference based on race, colour, descent or national or ethnic origin which has the purpose or effect of nullifying or impairing the recognition, enjoyment or exercise, on an equal footing, of any human right or fundamental freedom in the political, economic, social, cultural or any other field of public life.[36]

While the overall effect of exclusion of Aboriginal and Torres Strait Islander people from genomics is within the spirit of this provision, it will be rare for any individual study to fall within its purview.

The *National Statement* further provides that 'exclusion of some groups may amount to unfair discrimination, and/or exclude individuals and groups from the potential benefits of research'.[37] However, the ethical guidelines also require that research that targets Aboriginal and Torres Strait Islander people meet additional conditions and a higher threshold of ethical review. Researchers, constrained by budget considerations or perceiving research with Aboriginal and Torres Strait Islander people as harder or riskier than with non-Indigenous people, tend to avoid focusing on Aboriginal and Torres Strait Islander cohorts. Further, much research excludes Aboriginal and Torres Strait Islander participants implicitly, through failing to include culturally appropriate protocols for engagement and recruitment.[38] Legal responses are not well suited to addressing systemic exclusion of this nature.

More significant is the role played by cultural norms; that is, what is considered standard and best practice in research and clinical care. Culturally appropriate protocols for research and clinical care are described in the literature, but require systemic changes to become commonplace.[39]

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33 Alice Popejoy and Stephanie Fullerton, 'Genomics is Failing on Diversity' (2016) 538 *Nature* 161.

34 Easteal et al (n 21); Caroline F Wright et al, 'Assessing the Pathogenicity, Penetrance, and Expressivity of Putative Disease-Causing Variants in a Population Setting' (2019) 104 *American Journal of Human Genetics* 275.

35 Easteal et al (n 21); McWhirter, Nicol and Savulescu (n 4).

36 *Racial Discrimination Act 1975* (Cth) s 9(1).

37 National Health and Medical Research Council, Australian Research Council and Universities Australia (n 29) s 3.1.15.

38 McWhirter, Nicol and Savulescu (n 4).

39 See, eg, QIMR Berghofer Medical Research Institute, Genomic Partnerships: Guidelines for *Genomic Research with Aboriginal and Torres Strait Islander Peoples of Queensland* (Guidelines, 2019); Gareth Baynam et al, 'Indigenous Genetics and Rare Diseases: Harmony, Diversity and Equity' in Manuel Posada de la Paz, Domenica Taruscio and Stephen Groft (eds), Rare Diseases Epidemiology: Update and Overview (Springer, 2017) 511; NSW Health, *The National Model of Consent for Clinical Genomic Testing: Guidance for Health Professionals obtaining Consent for Clinical Genomic Testing* (online, 2021) <https://www.health.nsw.gov.au/services/Publications/genomic-testing-consent-guidance.pdf>.

The need for culturally appropriate protocols to ensure inclusion is a potential limitation of a uniform approach to consent to genomic testing, as proposed in the National Model of Consent for Clinical Genomic Testing.[40] This report briefly acknowledges that additional considerations are relevant when consenting Aboriginal and Torres Strait Islander people, with feedback from the consultation process noting that:

- Aboriginal Liaison Officers or other health workers should be made available to assist with consent processes, and when using interpreters, it will be 'important to recognise that in some Aboriginal languages there may not be a specific word or description communicate the process';
- 'Health professionals should receive training on how to support Aboriginal and Torres Strait Islanders';
- The consequences of the lack of genomic reference data for Aboriginal and Torres Strait Islander peoples 'needs to be discussed with participants'; and
- Health practitioners may need to 'seek informed consent, consult with, and/or support from trusted representatives in the community prior to relevant clinical genomic testing.'[41]

The terms employed in this document are suggestive, rather than directive, in nature and will need to be supported by additional measures if meaningful change is to occur.

There is potential for accountability regarding equity through the National Agreement on Closing the Gap 2020.[42] Targets for measuring access to information are intended to assess Aboriginal and Torres Strait Islander people's access to the information and services that can enable participation in informed decision-making about their lives. Commitment in support of this target could consider investment in reference genomes, access to diagnostic screening and availability of genetic counselling.

Hudson and colleagues argue that achieving equity will require engaging with Indigenous critiques of genomic research, revolving around issues of trust, accountability and equity, and establishing frameworks that recognise Indigenous rights and interests in genomic resources.[43] This may include developments in domestic law that give expression to international law obligations, including Article 31 of the UN Declaration on the Rights of Indigenous Peoples, which states that 'Indigenous peoples have the right to maintain, control, protect and develop their cultural heritage, traditional knowledge and traditional cultural expressions … including human and genetic resources'.

40 New South Wales Ministry of Health, 'National Model for Clinical Consent to Genomic Testing' (Final Report, January 2021) 9.

41 Ibid, 9.10.

42 The Coalition of Aboriginal and Torres Strait Islander Peak Organisations and all Australian Governments, *National Agreement on Closing the Gap* (Agreement, July 2020).

43 Hudson (n 31) 377.

5 Genetic Discrimination

5.1 Summary

Genetic discrimination is defined as the differential treatment of asymptomatic individuals on the basis of their actual or presumed genetic characteristics (i.e. of individuals who may be genetically predisposed to a disease due to a genetic mutation).[1] People can experience genetic discrimination within a range of contexts, including in the family and social domains through to public institutional domains. Internationally and in Australia, the focus of debate regarding genetic discrimination has been within the context of insurance, and to a lesser extent, employment.

Although a difficult-to-verify phenomenon, there is clear evidence of genetic discrimination occurring in Australia.[2] Despite the ALRC and AHEC making a number of reform recommendations in *Essentially Yours: The Protection of Human Genetic Information in Australia* ('*Essentially Yours*') to protect against genetic discrimination,[3] these were never implemented and, until recently, insurers have been free to use genomic test information provided they could justify their decisions actuarially. However, following further scrutiny of the life insurance industry by the Parliamentary Joint Committee on Corporations and Financial Services,[4] a voluntary moratorium on the use of genetic test results for life insurance cover up to $500,000 was introduced by the peak insurance body, the Financial Service Council ('FSC'), in June 2019.[5] The moratorium will be re-evaluated in 2022 to determine whether it will be continued beyond the initial five years duration (ending June 2024).[6] It remains to be seen whether consumer interests are sufficiently protected by the moratorium and whether its terms are well understood by consumers and their health care professionals.

5.2 Regulatory Response to Genetic Discrimination

Debate on regulatory responses to genetic discrimination in Australia dates back many years, to Senator Stott Despoja's Private Members Bill, the Genetic Privacy and Non-discrimination Bill 1998, based on draft legislation from the United States.[7] This Bill had been referred to the Senate Legal and Constitutional Legislation Committee which reported, in 1999, significant privacy and ethical issues in relation to access to, and control over, personal medical records containing genetic information.[8] There was no further follow up since then until the ALRC/AHEC Inquiry that resulted in *Essentially Yours*.

1 Paul Billings et al, 'Discrimination as Consequence of Genetic Testing' (1992) 50(3) *American Journal of Human Genetics* 476.

2 Margaret Otlowski et al, 'Investigating Genetic Discrimination in the Australian Life Insurance Sector: Use of Genetic Test Results in Underwriting 1999-2003' (2007) 14(3) *Journal of Law and Medicine* 367; Kristen Barlow-Stewart et al, 'Verification of Consumers' Experience and Perceptions of Genetics Discrimination and its Impact in Utilization of Genetic Testing' (2009) 11 *Genetics in Medicine* 193.

3 Australian Law Reform Commission and Australia Health Ethics Committee, *Essentially Yours: The Protection of Human Genetic Information in Australia* (ALRC Report 96) (ALRC Report, No 96, May 2003) 14.6 ('Essentially Yours').

4 Parliamentary Joint Committee on Corporations and Financial Services, Parliament of Australia, *Life Insurance Industry* (Report, March 2018) 1.

5 Financial Service Council, *Standard No. 11: Moratorium on Genetic Tests in Life Insurance ('Moratorium')* (Standard, 21 June 2019).

6 Ibid s 5.1.

7 *Genetic Privacy and Non-discrimination Bill 1998* (Cth).

8 Senate Legal and Constitutional Legislation Committee, Parliament of Australia, *Genetic Privacy Bill* (Report, 31 March 1999).

Concern about the use of genetic information by the insurance industry was one of the main factors leading to the establishment of this Inquiry.[9] The Inquiry's Terms of Reference specifically included consideration of how best to protect against unfair genetic discrimination.[10] Use of genetic test information for purposes of life insurance underwriting was thoroughly examined by the Inquiry.[11] While supporting the fundamental principle underlying the market in voluntary, mutually rated insurance, namely, equality of information between the applicant and the insurer, the ALRC/AHEC recognised the need for some adjustment in favour of consumer interests.[12] This was seen as necessary to ensure appropriate protection of consumers from inappropriate use of genetic information in insurance underwriting, noting the limited actuarial data available regarding the impact of genetic tests on insurance underwriting.[13]

A number of specific recommendations were made for insurance underwriting involving the use of genetic information. One was for the proposed Human Genetics Commission of Australia ('HGCA')[14] to establish procedures to assess and make recommendations on whether particular genetic tests should be used in underwriting mutually rated insurance. In doing so, the report stipulated that the HGCA must have regard to the genetic tests' scientific reliability, actuarial relevance and reasonableness.[15] The report placed corresponding obligations on the then peak insurance body, the Investment and Financial Services Association ('IFSA'), and the Insurance Council of Australia ('ICA') to develop mandatory policies to ensure member compliance with HGCA recommendations regarding use of a particular genetic test in underwriting.[16] Further, the report recommended that IFSA and ICA should require their members to provide applicants with accurate information in relation to those genetic tests that the HGCA recommended not be used in underwriting.[17] Additional recommendations proposed to strengthen consumer protections included an entitlement to written reasons for an adverse underwriting decision based on genetic information,[18] and the establishment of an independent review mechanism for decisions involving genetic information.[19] Introduction of a compulsory moratorium was one of the options that the ALRC/AHEC Inquiry had canvassed but ultimately rejected, resolving instead to retain the current system but with modifications to restrict which genetic tests can be used in insurance underwriting.[20]

Notwithstanding the references to the role of HGCA in operationalising these proposed reforms, the Government in its Response to *Essentially Yours* concluded that '[t]hese recommendations are directed at IFSA and the ICA' [i.e. the life insurance industry] and the recommendations were never implemented.[21]

In 2009, the Human Genetics Advisory Committee ('HGAC') was established as a Principal Committee of the NHMRC. Genetic discrimination in insurance was on the committee's agenda for a number of years and the committee sought to engage with the life insurance industry on the issue. However, no substantive reforms were achieved before the HGAC was disbanded in 2015.

9 *Essentially Yours* (n 3) 44.

10 Ibid 100.

11 Ibid.

12 Ibid 690-693.

13 Ibid 707-711.

14 Ibid Recommendation 5-1 and 5-2, 211. This was recommended to be established as an independent statutory authority which provided advice to Australian governments about current and emerging issues in human genetics and similar high-level advice on the ethical, legal and social implications arising from these developments.

15 Ibid Recommendation 27-1, 711.

16 Ibid Recommendation 27-2, 711.

17 Ibid Recommendation 27-3, 711.

18 Ibid Recommendation 27-5, 723.

19 Ibid Recommendation 27-9, 733.

20 Ibid 684.

21 Full Australian Government Response to ALRC Report 96, 18 October 2010, <https://www.alrc.gov.au/inquiry/protection-of-human-genetic-information/full-australian-government-response-to-alrc-report-96/>

The issue of genetic discrimination in life insurance once again came under regulatory review through the Parliamentary Joint Committee on Corporations and Financial Services which reported in March 2018.[22] The report highlighted challenges with industry self-regulation and the negative impact on applicants for insurance of genetic discrimination by insurance companies. It also expressed concern that the problem of genetic discrimination is likely to become even more substantial in the near future with continual developments and cost reductions in the area of genetics.[23] Life insurer concerns about adverse selection[24] were found to be overstated and the committee concluded that there is greater benefit to consumers in preventing a duty of disclosure from arising in respect of predictive genetic tests.[25] Significantly, the committee was 'highly concerned about evidence received that individuals are not undertaking potentially life-saving genetic testing due to fears of unfair treatment by life insurers.'[26] The committee also expressed concern that the use or perceived use of genetic information by life insurers has impacted on participation in public health research projects and other forms of research. It recommended the introduction of a moratorium similar to that which exists in the UK, banning use of genetic test information for underwriting in life insurance. Further, in the event of failure by the industry to comply with a moratorium, the Committee recommended legislation be introduced prohibiting insurers' access to genetic test information.

As reflected in both these inquiries, there is a major concern of genetic discrimination in both clinical and research settings. There is clear evidence that genetic discrimination is acting as a deterrent to people undertaking clinically relevant genetic testing, as reported in published studies.[27] Guidance for health practitioners highlights the importance of advising of these insurance implications of genetic testing and a failure to do so would be analogous to the failure to warn of medical risks associated with a procedure.[28] There is also clear data of the negative effect that disclosure of the insurance implications of genetic testing has on participation by potential research participants. A paper by Keogh et al reported that the proportion of participants among those informed of insurance implications who then declined genetic testing, was more than double the proportion of those who did not have this information.[29]

These public health-related concerns have been a driving factor behind the proactivity of the Human Genetics Society of Australasia in urging the Australian Federal Government to take a more active role in regulating the use of genetic information in insurance and calling for a moratorium on the use of genetic test results.[30]

22 Parliamentary Joint Committee on Corporations and Financial Services (n 4).

23 Ibid 154.

24 Adverse selection refers to situations in which an insurance company extends insurance coverage to an applicant who knows their actual risk to be substantially higher than the risk known by the insurance company. The insurance company suffers adverse effects by offering coverage at a cost that does not accurately reflect its actual risk exposure. Implications of adverse selection in the context of genetic testing was outlined in the *Essentially Yours* Report (n 3) 679-680.

25 Parliamentary Joint Committee on Corporations and Financial Services (n 4).

26 Ibid 155.

27 Béatrice Godard et al, 'Factors Associated with an Individual's Decision to Withdraw from Genetic Testing for Breast and Ovarian Cancer Susceptibility' (2007) 11(1) *Genetic Testing* 45; Ilias Goranitis et al, 'The Personal Utility and Uptake of Genomic Sequencing in Pediatric and Adult Conditions: Eliciting Societal Preferences with Three Discrete Choice Experiments' (2020) *Genetics in Medicine* 1.

28 *Rogers v Whitaker* 1992 [HCA] 58 and see the Consent chapter.

29 Louise Keogh et al, 'Is Uptake of Genetic Testing for Colorectal Cancer Influenced by Knowledge of Insurance Implications?' (2009) 199(5) *Medical Journal of Australia* 255. See also Keogh et al, 'Choosing Not to Undergo Predictive Genetic Testing for Hereditary Colorectal Cancer Syndromes: Expanding Our Understanding of Decliners and Declining' (2017) 40(4) *Journal of Behavioral Medicine* 583; Amelia Smit et al, 'A Pilot Randomized Controlled Trial of the Feasibility, Acceptability, and Impact of Giving Information on Personalized Genomic Risk of Melanoma to the Public' (2017) 26(2) *Cancer Epidemiology, Biomarkers and Prevention* 212.

30 Human Genetics Society of Australasia, *Genetic Testing and Personal Insurance Products in Australia* (Position Statement, 28 February 2018).

Another dimension of the problem is the unequal bargaining position of an applicant compared to an insurer. Although the onus is on the insurer to justify any use of genomic test information in underwriting, in practice, it is difficult for an individual applicant to challenge an adverse insurance decision. This is compounded by low levels of awareness of avenues to challenge treatment perceived as unfair and for those wishing to pursue legal redress, costs can be prohibitive.[31] Indeed, the case of 'James' who achieved a successful outcome on his life insurance application only after commencing legal proceedings alleging genetic discrimination against the insurer, stands out as a rare example of a particularly well-informed, resilient and persistent consumer who was able to stand up for his rights.[32]

Against this background, it was not entirely surprising that most of the stakeholders interviewed for the Scoping Review[33] drew attention to community concerns associated with discrimination in the context of insurance.

Viewed in the international context, Australia has been relatively slow to respond and protect consumers from genetic discrimination. In contrast, a number of European countries have legislation to prohibit insurers' use of genetic test information including Belgium, Austria, Denmark, France, Germany, Lithuania, Norway, Portugal and Sweden. The United Kingdom has had a moratorium in place since 1997, negotiated between the UK Government and the Association of British Insurers. This moratorium is periodically reviewed, most recently in 2018, with the endorsement of a *Code on Genetic Testing and Insurance*.[34] This new *Code* has no set end date but will be reviewed every three years.[35] In the US, the *Genetic Information Nondiscrimination Act* (GINA), which took effect in 2009 provides some protection against genetic discrimination in insurance but this is limited to health insurance and notably does not cover life insurance long term care policies. In Canada, the *Genetic Non-Discrimination Act* was passed in 2017 and prohibits the use of any genetic test (including diagnostic genetic tests) in insurance underwriting. Although the Quebec Court of Appeal ruled this legislation to be invalid in 2019,[36] a recent decision of the Supreme Court of Canada has upheld its constitutional validity.[37]

5.3 Overview of Law

The regulation of genetic discrimination in Australia is primarily through anti-discrimination legislation – the *Disability Discrimination Act 1992* (Cth) (*'Disability Discrimination Act'*) at the federal level, and State and Territory anti-discrimination legislation (see Table 4).[38] The focus here is on genetic discrimination which is unlawful. Potentially, this could be as a result of declining a life insurance product outright or because of the imposition of unfavourable terms (e.g., charging higher premiums or imposing exclusions)[39] if it can be shown that the decision was not actuarially justifiable.

31 Anita Goh et al, 'Perception, Experience, and Response to Genetic Discrimination in Huntington's disease: The Australian Results of The International RESPOND-HD Study' (2013) 17(2) *Genetic Testing and Molecular Biomarkers* 115.

32 Louise Keogh and Margaret Otlowski, 'Life Insurance and Genetic Test Results A Mutation Carriers Fight to Achieve Full Cover' (2013) 199(5) *Medical Journal of Australia* 363.

33 Centre for Law and Genetics, and Centre for Health Law and Emerging Technologies, *Assessment of Legislation and Regulations Applying to the Collection and Use of Health-Related Genomic Information: Scoping Review* (2020).

34 *Code on Genetic Testing and Insurance: A voluntary code of practice agreed between HM Government and the Association of British Insurers on the role of genetic testing in insurance* (UK).

35 Ibid 3.

36 *In the matter of the: Reference of the Government of Quebec concerning the constitutionality of the Genetic Non-Discrimination Act enacted by Sections 1 to 7 of the Act to prohibit and prevent genetic discrimination*, 2018 QCCA 2193.

37 *Canadian Coalition for Genetic Fairness v Attorney General of Canada and Attorney General of Quebec* 2020 SCC 17.

38 *Discrimination Act 1991* (ACT); *Anti-Discrimination Act 1992* (NT); *Anti-Discrimination Act 1977* (NSW); *Anti-Discrimination Act 1991* (Qld); *Equal Opportunity Act 1984* (SA); *Anti-Discrimination Act 1998* (Tas); *Equal Opportunity Act 1995* (Vic); *Equal Opportunity Act 1984* (WA).

39 Margaret Otlowski et al (n 2) reported on Australian empirical research which identified a number of cases of unlawful genetic discrimination on the basis of unjustifiable exclusions imposed by the insurer.

TABLE 4: ANTI-DISCRIMINATION LEGISLATION IN AUSTRALIA AND THE EXEMPTION FOR LIFE INSURANCE

	Cth *Disability Discrimination Act 1992*	ACT *Discrimination Act 1991*	NT *Anti-Discrimination Act 1992*	NSW *Anti-Discrimination Act 1977*	QLD *Anti-Discrimination Act 1991*	SA *Equal Opportunity Act 1984*	TAS *Anti-Discrimination Act 1998*	VIC *Equal Opportunity Act 1995*	WA *Equal Opportunity Act 1984*
Disability/impairment									
definition: specifically includes genetic predisposition	*yes 'disability that may exist in the future (including because of a genetic pre-disposition to that disability)'*	*yes 'disability that a person may have in the future, including because of a genetic disposition to the disability'*	**no**	**no**	**no**	**no**	**no**	*yes 'disability that may exist in the future (including because of a genetic predisposition to that disability)'*	**no**
broad definition: future, presumed and/or imputed disability which encompasses genetic status	–	–	**no**	*yes*	*yes (s8 re meaning of discrimination on the basis on an attribute)*	**no**	*yes*	–	*yes*
Life insurance									
Comes within meaning of provision 'goods and services'	*yes*	*yes*	*yes*	*yes*	*yes*	*yes*	*yes*	*yes*	*yes*
Exemption from disability discrimination	*yes*	*yes*	*yes*	*yes*	*yes*	*yes*	*yes*	*yes*	*yes*
[+or other data]	*yes*	*no*	*yes*	*yes*	*yes*	*yes*	*no*	*yes*	*yes*
Prohibition on requests for information which may be used to discriminate (would apply to discrimination on grounds of genetic status)	*yes (uses example of genetic information)*	*yes*	*has a section which prohibits asking for information on which unlawful discrimination might be based but as impairment is defined narrowly doesn't apply to genetic status*	**no**	*yes*	**no**	**no**	*yes*	*yes*

5.3.1 INSURANCE

5.3.1.1 Life insurance

Life insurance products (e.g., death cover, critical illness cover or cover for total permanent disability) are a form of voluntary mutually rated insurance. Approximately 30% of Australian adults have a dedicated life insurance policy. Applicants for life insurance are required to disclose to the insurer all information that is known, or which reasonably ought to be known, to be relevant to the insurer.[40] Failure to disclose information that the applicant had a duty to disclose can result in the insurance contract being found to be void. Alternatively, it may reduce the amount paid to the person insured. For most life insurance products this duty only applies at the time of initial application; once life insurance is secured, it is guaranteed renewable, irrespective of a subsequent change in health status or genetic information becoming available.

Although seemingly well settled, it is in fact open to question which genetic test results an applicant for life insurance must disclose under their duty of disclosure. It is clear from the terms of the former Financial Services Council Genetic Testing Policy as well as the current moratorium document, that from an industry perspective, applicants for life insurance are expected to disclose not only genetic test results obtained in a clinical context but also genetic tests obtained as a result of participation in a research study where the results have been returned to the applicant.[41] It is, however, arguable that not all genetic tests are appropriate for underwriting e.g. research results that have not been confirmed by a National Association of Testing Authorities accredited laboratory or potentially unreliable direct-to-consumer genetic test results. Use of such results could be contested from a relevance or 'materiality' perspective. Establishing relevance or materiality of data to be disclosed is a question of fact and the onus lies on the insurer to do so.[42] It is not sufficient that an insurer is interested in the information; in order for information to be considered material there must be some relationship between that information and the assessment of risk.[43] Even for many clinical genetic test results this could be contested, let alone for unverified research results or direct-to-consumer genetic tests. Notably in the UK, which is one of the few jurisdictions to have reviewed the reliability of genetic test results for life insurance underwriting, the only genetic test approved for use for underwriting applications in excess of the moratorium threshold is the test for Huntington's Disease. Notwithstanding such arguments being raised, this has not been the subject of legal challenge and in academic commentary as well Australian government inquiries into this area, it appears to be widely assumed that all predictive genetic tests fall into the category of information that must be disclosed for the purposes of a life insurance application.[44]

In broad terms, there is protection from disability discrimination in the provision of goods and services[45] under Federal and State and Territory anti-discrimination law.[46] As a result of amendments made in response to the *Essentially Yours* Report, the *Disability Discrimination Act* is explicit in including within the meaning of disability, a disability that may exist in the future because of a genetic predisposition to that disability.[47] In addition, the more general provisions in the

40 *Insurance Contracts Act 1984* (Cth) s 21. After the finalisation of the analysis in this paper, this Act has been amended to include new s20B, applying to all consumer insurance contracts (including life insurance), shifting the onus on the insurer to specifically ask questions about information they regard as relevant. This has changed the duty of the insured from a duty of disclosure to a new duty to take reasonable care not to make a misrepresentation.

41 Financial Services Council, Standard No.11, *Genetic Testing Policy 2016*, cl 10.3.2; FSC, *Moratorium* (n 5) cl 3.2.

42 *Western Australian Insurance Co Ltd v Dayton* (1924) 35 CLR 355; *Visscher Enterprises Pty Ltd v Southern Pacific Insurance Co Ltd* [1981] Qd R 561.

43 *Barclay Holdings (Aust) v British National Insurance Co Ltd* (1987) 8 NSWLR 514.

44 Otlowski et al (n 2); See, eg, coverage in *Essentially Yours* (n 3) ch 25, footnote 5.

45 'Services' is defined to include insurance – see the *Disability Discrimination Act 1992* (Cth) s 4, as well as under State and Territory anti-discrimination legislation, eg, *Anti-Discrimination Act 1998* (Tas); *Equal Opportunity Act 1995* (Vic) s 44.

46 See also *Discrimination Act 1991* ACT s 5AA.

47 s 4 (j).

legislation of most States and Territories would cover genetic discrimination on the basis of genetic status as this would qualify as an imputed or presumed disability or impairment.[48]

While the legislation would certainly cover discrimination by an insurer on the grounds of a person's predisposition to a genetic disease, there is an exemption from liability for disability discrimination conferred on insurers underwriting insurance provided that they can give reasonable justification for their decisions. Under this legislation it is not unlawful for a person to discriminate on the grounds of a person's disability by refusing to offer, or altering the terms and conditions of a life insurance policy or other policy of insurance provided that the discrimination is based on actuarial or statistical data on which it is reasonable for the insurer to rely and that the discrimination is itself reasonable having regard to the matter of the data and other relevant factors.[49] Alternatively, in a case where no such actuarial or statistical data is available and cannot reasonably be obtained, the insurer's conduct will be protected under the exemption if the discrimination is reasonable having regard to any other relevant factors.

Under State and Territory legislation,[50] the language of the insurance exemption varies between jurisdictions, but most provisions contain elements similar to those in s 46 of the *Disability Discrimination Act*. These exemptions acknowledge that insurance inherently involves differentiating between individuals but seeks to prevent discrimination which is not based on justifiable grounds.

There has been no Australian case law alleging disability discrimination against an insurer on the basis of their use of genetic test information but some guidance on the interpretation of the insurance exemption can be found in the Federal Court decision *QBE Travel Insurance v Bassanelli*.[51] This case dealt with alleged discrimination against an applicant for travel insurance on the grounds of her metastatic breast cancer after the insurers, QBE, had declined to issue any policy of travel insurance to her for a short overseas holiday. The case confirmed that the onus of proof is upon the insurer to qualify for the exemption once it is shown that the insurer has engaged in discrimination that would be in contravention of the Act.[52] Further, insurers cannot rely on 'other relevant factors' without first seeking out relevant actuarial and statistical data as required under the provision.[53] If there is actuarial or statistical data available to or reasonably obtainable by the insurer, then 'other relevant factors' cannot be relied upon.[54] The reasonableness of the discrimination will not be judged only by the material known to the insurer; rather, the insurer must consider 'any matter which is rationally capable of bearing upon whether the discrimination is reasonable'.[55]

Although these insurance exemptions for insurers from disability discrimination remain in force, in July 2019 the FSC introduced a voluntary moratorium on the use of genetic test information in life insurance underwriting subject to specified thresholds.[56] The moratorium is in the form of a self-regulatory standard and has limited the use by life insurers of genetic test information to those circumstances where the applicant is applying for insurance above the specified thresholds: $500,000 for lump sum death cover and total permanent disability cover and $200,000 for trauma and critical illness cover. The effect of the moratorium is to limit an applicant's duty to disclose genetic test information to circumstances where the amount of insurance applied for

48 *Anti-Discrimination Act 1977* (NSW) s 49A; *Anti-Discrimination Act 1991* (Qld) s 8; *Anti-Discrimination Act 1998* (Tas) s 3, *Equal Opportunity Act 1995* (Vic) s 4, *Equal Opportunity Act 1984* (WA) s 4.

49 *Disability Discrimination Act 1992* (Cth) s 46; See *Disability Discrimination Act 1992* (Cth) s 12, although the legislation expressly preserves some residual role for State and Territory legislation – see ss 13(2) and (5) which give the complainant a choice of jurisdiction in which to pursue the complaint.

50 *Discrimination Act 1991* (ACT) s 28; *Anti-Discrimination Act 1992* (NT) 49; *Anti-Discrimination Act 1977* (NSW) s 49Q; *Anti-Discrimination Act 1991* (Qld) s 72; *Equal Opportunity Act 1984* (SA) s 85; *Anti- Discrimination Act 1998* (Tas) s 44; *Equal Opportunity Act 1995* (Vic) s 47; *Equal Opportunity Act 1984* (WA) s 66T.

51 [2004] FCA 396.

52 Ibid [37].

53 Ibid [33].

54 Ibid [34].

55 Ibid [53].

56 FSC, *Moratorium* (n 5).

exceeds the specified thresholds. This is reinforced in the moratorium document which makes clear that non-disclosure of genetic test information for applications for life insurance below the specified thresholds does not amount to a breach of duty to disclose.[57]

However, genetic information from other sources, such as family history, must still be disclosed and can be taken into account. The moratorium provides for the continued use of a favourable genetic test result that an applicant chooses to disclose, to offset a relevant family history.[58] Under the moratorium, FSC members who are 'Life Insurance Providers'[59] are bound to comply with its terms, however as the moratorium is a voluntary arrangement introduced by the FSC it is not legally enforceable in the event that an insurer does not comply with its terms.

5.3.1.2 Evaluation of the FSC Moratorium

The introduction of the moratorium on the use of genetic tests is a positive development as it limits disclosure obligations and the detrimental use of genetic test information for the purposes of underwriting life insurance products. As a regulatory measure, a moratorium has some advantages over a fixed legislative approach, particularly its flexibility and amenability to updating, especially in light of rapid advances in the field of genomics and genetic testing.

There are, however, a number of important limitations. As noted earlier, recommendations in *Essentially Yours* that intended to balance the rights and obligations of consumers and insurers in this area were never implemented, leaving consumers vulnerable. Foremost amongst these was the recommendation for an independent committee (HGCA) to be established to vet which genetic tests are suitable for use in underwriting mutually rated insurance based on the tests' scientific reliability, actuarial relevance and reasonableness. In the UK where this approach has been adopted, only the test for Huntington's Disease has been approved by the Genetics and Insurance Committee.

While the moratorium provides some protection to an applicant in terms of what must be disclosed and what information can be used for underwriting purposes, its limited terms both with respect to financial thresholds and duration have raised concerns as reflected in the stakeholder consultations:

> [People are in] a better position than they were in when there was no moratorium... but we see the limits that apply ($500,000) only covers what some people need in terms of proper life insurance cover (Participant 3).

Notably, the terms of the moratorium, specifically the financial thresholds are not as generous as the equivalent insurance moratorium in the UK (£500,000 for Life Insurance, £300,000 for Critical Illness Insurance, and £30,000 for Income Protection Insurance). Applicants seeking insurance above the specified thresholds are still required to disclose all genetic test results. In the UK, the suitability of predictive genetic tests has been heavily scrutinised, resulting in only the test for Huntington's disease qualifying for disclosure. Under the FSC moratorium, all genetic tests must be disclosed for applications above the threshold irrespective of whether obtained in a clinical or research context. As a result, the potential for genetic discrimination remains with all the attendant risks vis a vis deterrence of take up of genetic tests with and participation in genetic research.

57 Ibid s 4.

58 Ibid s 3.4.

59 As defined in the *FSC, Guidance Note 5: Industry Terms and Definitions* (Guidance Note, 21 June 2019).

Furthermore, there is uncertainty for consumers/patients and health care providers as genetic test information still needs to be disclosed in some circumstances. This, in turn, has implications for genetic counselling prior to genetic testing and also in the health research context in terms of the information research participants must be informed about before agreeing to participate in a genetic/genomic research study. This uncertainty and resulting confusion came out clearly in the stakeholder consultations:

> [The moratorium is creating] confusion for clinicians...consumers, [and] patients ... (Participant 3)

> We now have to inform people about the moratorium and therefore it is creating more uncertainty in the people, being reluctant to be involved because we are told we have to bring this up in conversation ... This whole issue has created an unnecessary hesitancy for people to be involved in research that will benefit them more than harm them (Participant 25).

Options for recourse for those who believe they have been subject to unfair genetic discrimination are limited. *Essentially Yours* recommended an accessible independent review process of underwriting decisions involving genetic test information.[60] This recommendation was never implemented. While some legal avenues to challenge decisions do exist, in practice they are usually not taken up – either because people are not aware of their legal rights or do not have the resources to litigate.[61] This is particularly concerning given that the onus is actually on the insurer to justify their decision.

Furthermore, as the moratorium is in the form of a Standard which FSC members have agreed to comply with, it is not *legally* binding and could not be legally enforced if an insurer does not comply.[62]

There are also risks associated with the self-regulated nature of the moratorium which currently lacks any government oversight.[63] The operation of the moratorium will be reviewed by the FSC in consultation with stakeholders to consider, among other things, appropriateness of the amounts of cover, advances in genomics and genetic testing and the impacts of the moratorium on the sustainability of the life insurance industry,[64] but this lacks the rigour of an independent review.

The moratorium is time-limited (stated to apply for five years from 1 July 2019 to 30 June 2024, with potential for extension). The moratorium in the UK has been extended a number of times and now exists in the form of a Code which has no end date but is subject to a three-yearly review. Therefore, while there may be a reasonable expectation of renewal of the FSC moratorium, its time-limited status does create considerable uncertainty. Persons who undertake genetic/genomic testing in the interim years would be obliged to disclose those tests for new life insurance applications in the event that the moratorium is not extended. Given the 'guaranteed renewable status' of life insurance, policies taken out during the moratorium would remain in place provided premiums are regularly paid however ending the moratorium could have significant financial implications for individuals wanting to take out new life insurance cover or increase the amount of cover in being able to access the cover sought. This would also carry with it the attendant risks of genetic discrimination as a result of potentially unjustified decision making. Such adverse financial impact extends beyond the individual applicant to their families, particularly where they have dependents.

60 *Essentially Yours* (n 3) Recommendations 27-9 733.

61 Margaret Otlowski et al, 'The Use of Legal Remedies in Australia for Pursing Allegations of Genetic Discrimination: Findings of an Empirical Study' (2007) 9(1) *International Journal of Discrimination and the Law* 3.

62 It should be noted that, at the time of writing, the FSC has plans to add the Moratorium to its Code of Practice which would enhance its enforceability: Adrian Black, 'Genetic testing moratorium needs more research, tweaking' *Australian Banking Daily* (20 May 2020).

63 For discussion of some of the limitations of an industry self-regulated model for insurance see Ainsley Newson et al, 'Genetic and Insurance in Australia: Concerns Around a Self-Regulated Industry' (2017) 20(4) *Public Health Genomics* 247.

64 FSC, *Moratorium* (n 5) s 5.

It will therefore be important that ongoing evaluation of the impact of the moratorium on Australian consumers occurs. Relevantly, the Genomics Health Future Mission has recently funded a three-year Ethical, Legal and Social Implications Project to monitor the moratorium's impact.[65] It is also imperative to ensure the FSC's proposed review of the moratorium in 2022 takes account of all relevant information. As one interviewee cited in the Scoping Report reflected:

> The moratorium was a clever strategic move on part of the FSC. People particularly in government who were concerned think it is fixed now. As we gather data, I think we need to gather data about the impact of the moratorium and whether it is actually protecting consumers. I think it certainly is an issue (Participant 18).

While the moratorium has its limitations as noted, it does offer some protection to consumers. With the expansion of genomic testing and increase in the scale of genomic information available about an individual, the potential for genetic discrimination will increase if there are no safeguards in place. This is particularly the case in the event of expansion of genomic testing into population screening which potentially includes genomic information of children (even newborns) whose parents provide consent on their behalf. Genetic/genomic testing of children is contentious, especially for late onset disorders.[66] The usual disclosure obligation on an applicant for life insurance includes all genetic test information of which they are aware, so would apply in the case of applicants who became aware of genetic risk as a result of population screening, irrespective of age at the time of testing and whether it had been undertaken with their consent. The impact of adverse life insurance outcomes and potential for unjustified decision making is exacerbated in circumstances where the applicant must disclose genetic test information but had not been given a choice as to whether that testing should be undertaken. This highlights the need to develop stronger and more enduring safeguards to protect consumers.

5.3.1.3 Travel insurance

Travel insurance is part of general insurance and therefore falls outside the scope of the FSC Moratorium on the use of genetic tests in underwriting, which only applies to life insurance products. As a form of general insurance coming within the *Insurance Contracts Act 1984* (Cth), travel insurance is subject to the usual principles about insurance contracts, that contracts must be made with the utmost good faith and that failure to make full disclosure of required information can result in the contract being invalidated.

As travel insurance usually involves individual risk assessment which takes account of health status, the exemption in the *Disability Discrimination Act* allowing insurers to discriminate on the basis of person's disability, applies provided that such discrimination can be justified in accordance with the requirements of the legislation.[67] However, because of the typically short term-nature of such insurance, it would be rare for a positive (adverse) genetic test result to be relevant for underwriting purposes. The legal onus rests with the insurer to demonstrate the relevance of information on which they seek to rely but this is seldom brought to account because in practice, consumers who may have experienced discrimination on the basis of genetic status rarely seek to challenge this conduct.[68]

65 Jane Tiller et al, 'Monitoring the Genetic Testing and Life Insurance Moratorium in Australia: A National Research Project' (2021) 214 *Medical Journal of Australia* 157; Monash University, 'A-Glimmer: Australian Genetics and Life Insurance Moratorium: Monitoring the Effectiveness & Response', *Australian Genetics and Life Insurance Moratorium* (Website, 24 November 2021) <https://www.monash.edu/medicine/a-glimmer/home>.

66 Committee on Bioethics, Committee on Genetics and the American College of Medical Genetics and Genomics Social, Ethical, and Legal Issues Committee, Policy Statement, 'Ethical and Policy Issues in Genetic Testing and Screening of Children' (2013) 131 *Pediatrics* 620.

67 See section 5.3.1.1.

68 Otlowski et al (n 61).

Amendments to the *Insurance Contracts Act 1984* (Cth) made in 2013 regarding the insured's duty of disclosure specified travel insurance contracts as a category of insurance contracts to which particular rules should apply. The effect of this amendment introduced through new section 21A of the Act, is that for this category of general insurance (as well as motor vehicle insurance, building insurance and home context insurance), the responsibility lies with the insurer to ask specific questions that are relevant to the decision of the insurer whether to accept the risk and if so on what terms. If the insurer does not do so, they are taken to have waived compliance with the duty of disclosure in relation to the contract. This is beneficial for consumers (although probably not widely known or understood) and may help to remove some uncertainty about what exactly must be disclosed.[69] To the extent that specific questions are included, it is difficult to see how questions about predictive genetic test results could be relevant in the case of asymptomatic applicants.

Although the literature on this is sparse, there is some evidence of genetic discrimination occurring in relation to travel insurance. In 2001, Barlow-Stewart and Keays documented two cases in Australia where individuals reported a loading of their premiums for travel insurance on the basis of a genetic test result.[70] Neither of these individuals reported symptoms affecting their ability to travel. More recent Australian empirical research has identified instances where predictive genetic tests were treated as a pre-existing condition for the purposes of travel insurance applications which is clearly inappropriate.[71] In summary, the potential for genetic discrimination in travel insurance in Australia is an area that may warrant policy attention, particularly as this is not covered by the moratorium on the use of genetic test information. Similar concerns have been reported in the United Kingdom and Canada.[72]

5.3.1.4 Health insurance

Health insurance has been a significant domain for genetic discrimination in some countries such as the US pre-*GINA*, but this issue has not featured significantly in discussions about genetic discrimination in Australia. This is because Australian legislation regulating health insurance is solidarity-based and mandates a 'community rating' approach whereby premiums are based on the health care experience of the membership as a whole, rather than individual members.[73] This means that everyone is entitled to buy the same health cover, at the same price, regardless of age, gender, race, health status, claims history, perceived risk of requiring future treatment or any other reason.

All health insurers are required to provide health insurance to people with pre-existing conditions. Insurers can apply a maximum 12 month waiting period for pre-existing conditions and obstetric treatment before their members will be covered for hospital treatments. However, cover for psychiatric, rehabilitative or palliative care can be accessed after waiting just two months (whether or not for a pre-existing condition). Waiting periods are designed to ensure equality among insured persons. Without waiting periods, health insurers would be open to the risk of persons taking out short-term cover for the treatment of known conditions and cancelling their membership after the bills have been paid. The costs would then be borne by long-term members.

So, while health status is not normally relevant in accessing health insurance, it can be taken into account in connection with pre-existing conditions. A pre-existing condition is 'any ailment, illness or condition in respect of which the person had signs or symptoms during the six months prior to the person becoming insured under the policy.'[74] The existence of a pre-existing condition

69 Since the analysis in this paper was completed, s21A has been repealed and replaced with new s20B applying to all consumer insurance contracts (see n40 above).

70 Kristine Barlow-Stewart and David Keays, 'Genetic Discrimination in Australia' (2001) 8(3) *Journal of Law and Medicine* 250.

71 Kristine Barlow-Stewart et al, 'Genetic Discrimination Concerns in Travel Insurance—The Pre-existing Medical Condition Rule', 52nd European Society of Human Genetics (ESHG) Conference: Gothenburg, Sweden June 15-18, 2019 European Journal of Human Genetics (2019) 27:1174-1813 <https://doi.org/10.1038/341431-019-0494-2>.

72 Anya Prince, 'Comparative Perspectives: Regulating Insurers Use of Genetic Information' (2019) 27 *European Journal of Human Genetics* 340; Michelle Lane et al, 'Genetics and Personal Insurance: The Perspectives of Canadian Cancer Genetic Counselors' (2015) 24 *Journal of Genetic Counselling* 1022.

73 *Private Health Insurance Act 2007* (Cth) div 5

74 Ibid ss 75-15.

precludes health insurance payments for the duration of the waiting period for claims arising from that condition, irrespective of whether the condition was diagnosed at the time of taking out the policy.

The question arises as to whether a positive (adverse) predictive genetic test may be treated as a pre-existing condition. There had been some early reports of genetic discrimination in health insurance as a result of health insurers classifying a positive (adverse) predictive genetic test a 'pre-existing condition',[75] however this issue has not been tested in the courts. Clearly, if an individual is experiencing physical signs or symptoms of a genetic disease that would come within the meaning of 'pre-existing condition.' It is equally clear that in the absence of symptoms or signs of the condition at the time of the application, an adverse predictive genetic test should not be treated as a 'pre-existing condition'. To hold otherwise would be the equivalent of treating an adverse predictive genetic test as a diagnosis.

Use of genetic information by health insurers was not considered by the ALRC/AHEC Inquiry. The Inquiry took the view that because health insurers are prevented from using health information to assess individual risk due to the principle of community rating, the use of genetic information in relation to health insurance does not raise the same issues as for other personal insurance products.[76] There was no consideration given to the potential for genetic discrimination in connection with the interpretation of the pre-existing condition provision.

Although health insurance has not been prominent in the contemporary debate about genetic discrimination in Australia, in practice there is often confusion among members of the public as to the types of insurance genetic discrimination may impact. There have also been reports of consumer concerns about health insurance discrimination even though this is largely unfounded due to the nature of these products as community risk rated. Concern has also been expressed that the current community rating premise of Australian health insurance may change. This is reflected in the comments of one participant cited in the Scoping Report:

> Life insurance is still an issue. Who knows whether health insurance will change its tune down the track and say we will not insure people who have genetic conditions? The moratorium hasn't gone far enough (Participant 13).

This underscores the importance of retaining community rating for health insurance in Australia to ensure that genetic discrimination does not become a significant issue in this area.

5.3.2 EMPLOYMENT DISCRIMINATION

In comparison to genetic discrimination in insurance, relatively little attention has been given to the issue of genetic discrimination in employment. This was reflected in the stakeholder consultation process, where discrimination in the context of employment was referred to much less than discrimination in the insurance context by stakeholders.

While there may be legitimate use of genetic screening and monitoring in the workplace on occupational health and safety grounds, concerns have been raised about employer use of genomic information to determine whether to employ a person (in order to exclude from the workforce individuals who have been identified as being at risk of developing a genetic condition that may affect their future capacity for work).

Reported instances of genetic discrimination in employment in Australia[77] and overseas[78] have most commonly involved persons with genetic risk of neuro- degenerative disorders such as Huntington's

75 Joseph Alper and Jon Beckwith, 'Distinguishing Genetic and Nongenetic Medical Tests: Some Implications for Antidiscrimination Legislation' (1998) 4 *Science and Engineering Ethics* 142.

76 *Essentially Yours* (n 3) 654.

77 Barlow-Stewart and Keays (n 70).

78 Pauline Kim, 'Regulating the Use of Genetic Information: Perspective from the U.S. Experience' (2009) 31(4) *Comparative Labor Law & Policy Journal* 693.

Disease. However, past research into the practices and attitudes of Australian employers found limited evidence of interest by employers in use of genetic testing.[79]

The use of genetic information in employment was examined by the ALRC/AHEC Inquiry, although in less detail than the area of insurance – reflecting the relative nascent status of the issue at the time. *Essentially Yours* recommended that employers should not collect or use genetic information in relation to job applicants or employees, except in the limited circumstance consistent with privacy, anti-discrimination and occupational health and safety legislation.[80] The expansion of the definition of disability in the *Disability Discrimination Act* to include a genetic predisposition to disability, has been an advancement in this area.[81] That *Act* was also amended to restrict the information that an employer may seek from a prospective employee; an employer may not require a prospective employee to provide genetic information if the employer intends to use that information to unlawfully discriminate against the employee on the ground of a disability of the employee.[82]

5.3.2.1 The Law

Federal and State and Territory anti-discrimination legislation provides protection against discrimination in employment[83] on the grounds of disability/impairment, which under most acts is broadly defined to include a future or imputed disability. It therefore applies to genetic discrimination. Litigated cases, however, are rare.

The case of *Trindall v NSW Commissioner for Police*[84] illustrates the potential for misuse of an employee's genetic information; in this instance, discrimination was established under the *Disability Discrimination Act* against an asymptomatic individual on the basis of a carrier status.

In the United States, *GINA* prohibits employers from discriminating against individuals based on results from genetic testing;[85] this is particularly relevant in that jurisdiction in light of the responsibility of employers to provide health insurance for their employees.

Although there is an existing legislative framework which provides protection against genetic discrimination in employment, there appears to be a lack of awareness about the laws covering this area as reflected in the stakeholder consultations:

> Discrimination in employment can be an issue. The laws do not cover genetic discrimination and it is a fine line, it is hard to prove sometimes, might be that the other candidate is a better candidate for the job. But if you know two people, one with a child with genetic health condition [and the] other doesn't, you'll probably employ the one that doesn't because they won't be as absent from work (Participant 13).

> In Australia do we have any laws that prevent you from discriminating if an employer has your records and sees a high risk of Alzheimer's onset? Would this be allowable? I do not know (Participant 9).

79 Margaret Otlowski et al, 'Practices and Attitudes of Australian Employers in Relation to the Use of Genetic Information: Report on a National Study' (2010) 31 *Comparative Labor Law and Policy Journal* 637.

80 Ibid chs 30-32.

81 *Disability Discrimination Act (Cth)* s 4(j).

82 *Disability Discrimination Act 1992* (Cth) s 30. A number of States and Territories make general provision to prohibit request for information on which unlawful discrimination might be based: *Discrimination Act 1991* (ACT) s23; *Anti-Discrimination Act 1992* (NT) s 26; *Anti- Discrimination Act 1991* (Qld) s 124; *Equal Opportunity Act 1995* (Vic) s 107; *Equal Opportunity Act 1984* (WA) s 66O.

83 *Disability Discrimination Act 1992* (Cth) s 15; *Discrimination Act 1991* (ACT) s 10; *Anti-Discrimination Act 1992* (NT) s 31; *Anti-Discrimination Act 1977* (NSW) s 49D; *Anti-Discrimination Act 1991* (Qld) s 15; *Equal Opportunity Act 1984* (SA) s 67; *Anti-Discrimination Act 1998* (Tas) s 22; *Equal Opportunity Act 1995* (Vic) s 18; *Equal Opportunity Act 1984* (WA) s 66B.

84 [2005] FMCA 2.

85 *Genetic Information Nondiscrimination Act 2008* Title 1, Health Insurance.

6 Some Controversial Secondary Uses

6.1 Summary

Once a sample has been collected and tested, for either clinical or research purposes, the resultant data is a valuable resource that can be re-used by other parties. The focus of this chapter will be reuse for law enforcement and for commercial purposes. The traditional controls of individual informed consent and deidentification (so-called 'ask or anonymise') have significant limitations in these contexts.

The *Privacy Act 1988* (Cth) ('*Privacy Act*') and equivalent State and Territory Acts all have provisions permitting discretionary disclosure by those holding identifiable genetic and genomic information to law enforcement agencies without the knowledge or consent of the individual to whom the information relates. This applies in both clinical and research settings, although in some states the confidential information of hospital patients is not subject to these provisions.[1] Higher privacy protections for genomic information have been set out in specific contexts, such as for information held on the My Health Record system, and arguably this level of protection may be warranted more broadly. Further, the current regime allows secondary disclosure of genomic data for a wider range of offences than those for which law enforcement agencies may undertake primary genetic testing, which is restricted to serious and indictable offences.

Secondary use of genomic information for commercial purposes is primarily mediated by consent, but the quality of this consent may be problematic. Significantly, broad consent fails to ameliorate some risks of commercial re-use of health-related genomic data such as a lack of trust, whether participants will receive a financial benefit, and whether they have sufficient control over the data re-use. Yet even specific consent to each proposed type of re-use may be an inadequate response, as well as being expensive and impractical (see Chapter 2). It does not guarantee that data subjects will have trust in or oversight of the commercial use, nor does it assure public benefit, or necessarily provide a clear insight into benefit sharing.

6.2 Secondary Uses of Genomic Samples and Data

Genomic data, once created, can be reused by many different secondary parties. In many cases, reuse is a desirable outcome: a whole exome sequence undertaken for one purpose may be reanalysed in light of new information; data generated for one research project may be pooled with data from other projects to increase power and provide additional value from the public funds originally expended. However, other potential secondary uses are more troubling. Two controversial secondary uses identified in the stakeholder consultation were: use for law enforcement purposes, and commercialisation.

1 For example, in Queensland and Victoria the more permissive provisions in the health records and information privacy laws are subordinate to the health services laws which preclude discretionary disclosure on this basis.

6.2.1 LAW ENFORCEMENT AGENCY ACCESS

FIGURE 6: COMPARING BASES FOR DISCLOSURE UNDER THE PRIVACY ACT AND THE MY HEALTH RECORDS ACT

Bases for disclosure to law enforcement agencies, of identifiable genomic information held in medical records, research databases, DTC-GT services (covered by Privacy Act)

Entity considers it reasonably necessary for law enforcement purposes	Court order	Individual consent	Consent is impracticable and entity considers it necessary to reduce serious threat to someone's life, health or safety

Bases for disclosure to law enforcement agencies, of identifiable genomic information held in My Health Record

Entity considers it reasonably necessary for law enforcement purposes	Court order	Individual consent	Consent is impracticable and entity considers it necessary to reduce serious threat to someone's life, health or safety

Enforcement agencies, principally police, may have a legitimate interest in obtaining access to the health-related genomic information of Australians in clinical, commercial and research contexts. Such access may assist them in identifying, confirming or eliminating individuals from likely involvement in a serious offence, where DNA has been deposited at a crime scene.

Essentially Yours addressed this issue,[2] particularly in relation to police accessing resources such as newborn bloodspot screening ('Guthrie') cards and other sources of genomic data outside the forensic procedures legislation.[3] At that time the Australian Law Reform Commission and Australian Health Ethics Committee recommended the development of:

> nationally consistent rules governing disclosure of newborn screening cards for law enforcement purposes. These rules should provide for disclosure only: (a) with the consent of the person sampled or a person authorised to consent on his or her behalf; or (b) pursuant to a court order.[4]

2 Australian Law Reform Commission and Australian Health Ethics Committee, *Essentially Yours: The Protection of Human Genetic Information in Australia* (Report No 96, May 2003) ('*Essentially Yours*').

3 Ibid ch 19.

4 Ibid rec 19.100.

This recommendation was not implemented directly, although there is now a Newborn Bloodspot Screening National Policy Framework (Department of Health, 2017) providing some guidance. *Essentially Yours* also recommended that:

> AHMAC[5], in consultation with the HGCA[6], the NHMRC[7] and key professional bodies, should review the need for nationally consistent rules in relation to the collection, storage, use and disclosure of, and access to, other human tissue collections— including collections of pathology samples and banked tissue.[8]

Provisions in the *Crimes Act 1914* (Cth) and equivalent State and Territory legislation address the collection of DNA samples *directly* for forensic purposes, as well as their sharing with other agencies (in Australia and overseas), retention and disposal.[9] They also regulate the State, Territory and national criminal DNA databases, including data sharing.[10] Such legislation does not address police access to and use of DNA samples and information collected for health-related clinical, commercial or research purposes. Such access may be seen as a means for police to circumvent the forensic procedures set out in legislation, which generally constrain sample collection to situations involving either a person's consent, court order, or the authorisation of a senior police officer.

Community concern about police access to health-related genomic information held by pathology laboratories, clinical records and research databanks was expressed during the stakeholder consultation for the Scoping Review, with almost all the Aboriginal and Torres Strait Islander participants flagging the potential for police to access this data as a serious issue.[11]

The traditional reliance on anonymisation or consent as safeguards of privacy in relation to the use of health-related genomic data is unsuited to law enforcement agency access to this data. The data's usefulness lies in its identifiability—so anonymisation is not a helpful safeguard here – and police neither need to invite data subjects to give consent nor inform them about access.

6.2.2 COMMERCIALISATION OF HEALTH-RELATED GENOMIC INFORMATION

Commercial entities such as biotechnology, pharmaceutical and medical device companies have an interest in the primary or secondary use of identifiable genomic information generated in clinical and research settings. They can use this information to conduct research and generate new products.

Stakeholder interviews revealed community apprehension that organisations holding genomic data could proceed to share it with commercial entities. This is supported by academic research showing that, for instance, trust and intention to participate in a biobank was negatively impacted if the biobank was described as being a private rather than public research entity.[12] Nicol and colleagues reported that establishing proper arrangements for benefit sharing in the case of commercial involvement could be very important in ensuring public trust.[13]

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5 AHMAC is the Australian Health Ministers' Advisory Council, which was the former advisory and support body to the Council of Australian Governments Health Council.

6 HGCA was the Human Genetics Commission of Australia that the *Essentially Yours* report recommended be established.

7 NHMRC is the National Health and Medical Research Council.

8 *Essentially Yours* (n 2) rec 19.103.

9 *Crimes Act 1914* (Cth) pt ID ('*Crimes Act*'). See, eg, *Crimes Act 1958* (Vic) sub-div 30A.

10 Ibid pt ID.

11 Esther Han, 'My Health Record Can Store Genomic Information but Critics Say it's Not Ready', *Sydney Morning Herald* (online at 5 August 2018) <https://www.smh.com.au/healthcare/my-health-record-can-store-genomic-data-but-critics-say-it-s-not-ready-20180801-p4zuxz.html>.

12 Christine Critchley, Dianne Nicol and Margaret Otlowski, 'The Impact of Commercialisation and Genetic Data Sharing Arrangements on Public Trust and the Intention to Participate in Biobank Research' (2015) 18(3) *Public Health Genomics* 160 <https://doi.org/10.1159/000375441>.

13 Dianne Nicol, 'Public Trust, Intellectual Property and Human Genetic Databanks: The Need to Take Benefit Sharing Seriously' (2006) 3(3) *Journal of International Biotechnology Law* 89 <https://doi.org/10.1515/JIBL.2006.012>.

Untangling the bases for this trust deficit is less straightforward, but research suggests that they include: concerns about unexpected use of personal information beyond the scope of the original consent; a lack of benefit sharing;[14] compromising the public benefit/altruistic nature of the original data donation; and use of the data in ways that are morally concerning or discriminatory.[15]

The traditional approach of 'ask or anonymise'[16] fails to protect data subjects in the face of these issues. The inherent identifiability of genomic information coupled with the value-add that phenotypic data provides in genomics research limits the usefulness of anonymisation as a privacy-protecting mechanism. Consent therefore remains the principal legal basis for commercial use and reuse of health-related genomic information gathered in the clinical or research setting, as described in the overview of the law given below. However, using consent as a legal basis for reuse is also problematic, for a number of reasons.

If a data subject originally provided broad consent, for example when donating a tissue sample and associated data to a publicly-funded biobank, the scope of this consent may have included future unspecified commercial use. While broad consent might be legally sufficient for the commercial use, it is probably inadequate for providing data subjects with control over this use (especially in relation to morally-challenging uses of the data), for the return of benefits to them, and even in terms of the degree of information provided. Even the more intensive and tailored consent mechanisms (for example: dynamic consent, tiered consent) do not allow for prior consultation with affected individuals and groups, nor governance to support transparency and oversight and address conflicts of interest.

6.3 Overview of Legal Protections

This section reviews the protections that apply to genomic information held in health and health-research records as they apply to secondary use for law enforcement and commercial purposes. See Table 5.

6.3.1 SECONDARY USES FOR LAW ENFORCEMENT

The general discussion of privacy obligations in Chapter 3 of this report is relevant here, because all entities involved in producing and holding identifiable health-related genomic data will be bound by privacy obligations. Genomic information that is purportedly de-identified is unlikely to be useful to enforcement agencies in the investigation and prosecution of criminal offences.

6.3.1.1 How does the Privacy Act protect health-related genomic information in relation to law enforcement?

Identifiable health-related genomic information held by an entity that is subject to the *Privacy Act* — which includes many healthcare providers and research entities as well as direct-to-consumer genetic testing services (DTC-GT) — may be disclosed for the purposes of an 'enforcement related activity' without the data subject's knowledge or consent. This is established at Australian Privacy Principle (APP) 6.2(e),[17] which permits the use or disclosure of a person's identifiable genomic information (and other types of personal information) by an APP entity that 'reasonably believes that the use or disclosure of the information is reasonably necessary for one or more enforcement related activities conducted by, or on behalf of, an enforcement body'.

14 The patenting of genetic information is considered in detail in chapter 9.

15 This could compromise the public benefit/altruistic nature of the original data donation, see Critchley, Nicol and Otlowski (n 12) 170. There could also be concern about use of the data in ways that are morally concerning or discriminatory: Ipsos Mori, Report prepared for the Wellcome Trust, March 2016, 'The One-Way Mirror: Public attitudes to commercial access to health data' pp 57-60 <https://www.ipsos.com/sites/default/files/publication/5200-03/sri-wellcome-trust-commercial-access-to-health-data.pdf>

16 Mark Taylor, *Genetic Data and the Law: A Critical Perspective on Privacy Protection* (Cambridge University Press, 2012) 206.

17 *Privacy Act 1988* (Cth) sch 1 pt 3 ('*Privacy Act*').

'Enforcement related activity' is wide-ranging and is defined as:[18]

(a) the prevention, detection, investigation, prosecution or punishment of:
  (i) criminal offences; or
  (ii) breaches of a law imposing a penalty or sanction; or
(b) the conduct of surveillance activities, intelligence gathering activities or monitoring activities; or
(c) the conduct of protective or custodial activities; or
(d) the enforcement of laws relating to the confiscation of the proceeds of crime; or
(e) the protection of the public revenue; or
(f) the prevention, detection, investigation or remedying of misconduct of a serious nature, or other conduct prescribed by the regulations; or
(g) the preparation for, or conduct of, proceedings before any court or tribunal, or the implementation of court/tribunal orders.

The disclosing entity must make a written note of the use or disclosure (APP 6.5).

It is important to note that the provision is permissive rather than mandatory. Hence, an entity holding identifiable health-related genomic information *may* lawfully disclose it if they have the requisite reasonable belief but is not *required* to do so under APP 6.2(e).

The basis for the 'reasonable belief' is not defined in the Act. In the guidelines issued by the Office of the Australian Information Commissioner ('OAIC'), it is suggested that a signed and dated written request by an enforcement body will be a clear basis for a 'reasonable belief.'[19] In the case of *Jones v Office of the Australian Information Commissioner*,[20] it was indeed held that the existence of a search warrant was sufficient basis for a doctor's reasonable belief.

Beyond this, there are further provisions that support disclosure of health-related genomic information to law enforcement agencies. APP 6.2(b) permits disclosure where it 'is required or authorised by or under an Australian law or a court/tribunal order'. The consequences of a failure to act in accordance with a statutory requirement or court order include being found guilty of contempt of court and incurring financial penalties. APP 6.2(c) permits disclosure where a 'permitted general situation' exists. A 'permitted general situation' is defined at section 16A(1), and relevantly includes disclosure without consent where the entity reasonably believes that it is necessary 'to lessen or prevent a serious threat to the life, health or safety of any individual, or to public health or safety'. In this case, there must be 'one or more clear reasons that make it unreasonable or impracticable to obtain an individual's consent'.[21] APP 8 addresses the transfer of data overseas and provides a number of avenues for such transfer including APP 8.2(f) that specifically allows transfer to an enforcement body for enforcement-related activities.

18 Ibid s 6 (definition of 'enforcement related activity').

19 Office of the Australian Information Commissioner, *Australian Privacy Principles Guidelines: Privacy Act 1988* (Guidelines, July 2019) cl 6.60 <https://www.oaic.gov.au/__data/assets/pdf_file/0009/1125/app-guidelines-july-2019.pdf> ('APP Guidelines').

20 [2014] FCA 285.

21 APP Guidelines (n 19) cl C.6.

TABLE 5: AUSTRALIAN LEGISLATIVE PROVISIONS FOR DISCLOSURE OF HEALTH DATA WITHOUT CONSENT

	Cth *Privacy Act 1988*	Cth *My Health Records Act 2012*	ACT *Information Privacy Act 2014*	NT *Information Act 2002*	NSW *Health Records and Information Privacy Act 2002*	QLD *Information Privacy Act 2009*	SA *Department of the Premier and Cabinet Circular PC012 – Information Privacy Principles (IPPS) Instruction*	TAS *Personal Information Protection Act 2004*	VIC *Health Records Act 2001*	WA *Health Services Act 2016 and Health Services (Information) Regulations 2017*
Test for disclosure without consent for law enforcement purposes	*Reasonable belief that disclosure is reasonably necessary*	*Order from judicial officer; disclosure is reasonably necessary; no other way for agency to obtain the information; disclosure would not unreasonably interfere with person's privacy*	*Reasonable belief that disclosure is reasonably necessary*	*Reasonable belief that disclosure is reasonably necessary*	*Disclosure is reasonably necessary … where there are reasonable grounds to believe that an offence may have been, or may be, committed*	*Reasonable belief that disclosure is reasonably necessary*	*Disclosure is reasonably necessary*	*Reasonable belief that disclosure is reasonably necessary*	*Reasonable belief that disclosure is reasonably necessary AND is not a breach of confidence*	*Disclosure is in good faith*
Test for disclosure without consent to lessen or prevent serious* threat to life, health or safety of individual, or public health or safety	*It is unreasonable or impracticable to obtain individual's consent, and there is a reasonable belief that disclosure is necessary*	*It is unreasonable or impracticable to obtain individual's consent, and there is a reasonable belief that disclosure is necessary. (The issue of individual consent does not apply to a serious threat to public health or public safety)*	*It is unreasonable or impracticable to obtain individual's consent, and there is a reasonable belief that disclosure is necessary*	*Reasonable belief that disclosure is necessary*	*Reasonable belief that disclosure is necessary*	*Reasonable belief that disclosure is necessary*	*Belief on reasonable grounds that disclosure is necessary*	*Reasonable belief that disclosure is necessary*	*Reasonable belief that disclosure is necessary, and disclosure is in accordance with the guidelines (if any) issued by the Health Complaints Commissioner. [There are no such guidelines]*	*Disclosure in good faith; disclosure is reasonably necessary*
Test for disclosure without consent in accordance with a law, court or tribunal order	*Disclosure is required or authorised by or under an Australian law or a court or tribunal order, or in court proceedings*	*Disclosure is only required in relation to court or tribunal proceedings relating to the My Health Records Act; unauthorised access to information in the My Health Record system; or the provision of indemnity cover to a healthcare provider*	*Disclosure is required or authorised by or under a law or a court or tribunal order*	*Disclosure is required or authorised by law; reasonable belief that disclosure reasonably necessary for court proceedings*	*Courts, tribunals and Royal Commissions not affected by the Act*	*Disclosure is authorised or required by or under law; reasonable belief that disclosure reasonably necessary for court proceedings*	*Disclosure is required or authorised by or under law*	*Disclosure is required or authorised by or under law; reasonable belief that disclosure reasonably necessary for court proceedings*	*Disclosure is required, authorised or permitted, expressly or impliedly, by or under law, or for a law enforcement function (includes court proceedings)*	*Disclosure is in good faith: under another law; or in court proceedings; or under a court order*

** Test varies between jurisdictions, eg. threat must be 'serious', or 'real and immediate', or 'serious or imminent', or 'serious and imminent' etc.*
***This does not apply to the disclosure of information which could identify a person who is receiving or has received a public sector health service: see Part 7, Hospital and Health Boards Act 2011 (Qld).*
****This does not apply to the disclosure of information by a public or private hospital subject to the Health Services Act 1988 (Vic) s 141.*

There is no case law that directly considers the disclosure under the *Privacy Act* of health-related genomic information for law enforcement purposes. The penalties for interferences with privacy under the *Privacy Act* range up to 2000 penalty units for a serious or repeated interference with privacy.[22]

6.3.1.2 How does the My Health Records Act 2012 protect health-related genomic information in relation to law enforcement?

At present, genetic test reports rather than raw genetic data can be uploaded to the national electronic health record My Health Record. This may limit the Record's current utility for law enforcement purposes. From its 2012 enactment until amendment in 2018, the *My Health Records Act 2012* (Cth) ('*My Health Records Act*') contained provisions, like those of the *Privacy Act*, permitting the disclosure of health information included in a person's My Health Record to police or another enforcement body without external oversight. For disclosure to be permitted, the System Operator (the Australian Digital Health Agency ('ADHA')) only needed to have a reasonable belief in the reasonable necessity of disclosure to an enforcement body for, among other things, 'the prevention, detection, investigation, prosecution or punishment of criminal offences'.[23]

In 2018 this provision was repealed and new sections 69A and 69B were introduced in the underpinning legislation. These require an agency seeking to access a person's My Health Record information in this way to first obtain an order from a judicial officer for its release. The order may be obtained from a State or Territory magistrate or a judge who, in making the order, must be satisfied of certain matters, including that: the particular disclosure is reasonably necessary; there is no other effective means for the agency to obtain the information; and the disclosure of the information would not, on balance, unreasonably interfere with the privacy of the healthcare recipient. The person is not told about the disclosure but ADHA must make a written record of it.

As with the Commonwealth *Privacy Act*, and the State and Territory legislation discussed below, there are other statutory mechanisms for police to obtain any genomic information held in a person's My Health Record.

In particular, section 64 of the *My Health Records Act* permits disclosure by ADHA or a registered healthcare provider if the disclosing entity reasonably believes that the disclosure is 'necessary to lessen or prevent a serious threat to an individual's life, health or safety' and obtaining the person's consent to the disclosure is unreasonable or impracticable.[24] Further, a person may validly consent to the disclosure of their information under section 67.

Penalties for police obtaining information from My Health Record in breach of the above provisions include up to 5 years' imprisonment and 1500 penalty units.[25]

6.3.1.3 How do the State-based health records laws protect health-related genomic information in relation to law enforcement?

The *Privacy Act* does not apply to State and Territory public sector health service providers such as public hospitals. In the ACT, NSW and Victoria, private sector health services are required to comply with both Commonwealth and their State or Territory privacy laws in dealing with health information (see also Chapter 3).

Consistent with the Commonwealth legislation, the State and Territory public sector health authorities are entitled to *not* disclose a person's genetic information for law enforcement purposes in the absence of a legal obligation to disclose it.

22 *Privacy Act* (n 17) s 13G.

23 *My Health Records Act 2012* (Cth) s 70(1)(a) ('*My Health Records Act*').

24 Ibid s 64(1)(a).

25 Ibid ss 59-60.

All States and Territories[26] have legislative provisions that permit the disclosure of health-related genomic information (and other health information) by public sector agencies for law enforcement purposes without the data subject's knowledge or consent.[27] Victoria alone stipulates that the disclosure not be a breach of confidence, hence drawing in a 'public interest' balancing exercise to the decision. In some states, Queensland and Victoria for example, legislation protecting the health information of hospital patients overrides the provisions in other laws that allow discretionary disclosure for law enforcement purposes.[28]

Six of the States and Territories provide for the disclosure of genetic information from health records to a third party such as police or a court, where the disclosure is required or authorised by another law or a court order.[29] In South Australia, the Information Privacy Principles and the *Health Care Act 2008* (SA) permit the disclosure of personal information to a third person as is 'required or authorised by or under law'; a court order is not explicitly mentioned.[30] There is no analogous provision in the NSW legislation.[31]

Disclosure of information from health records to agencies involved in enforcing the criminal law is also possible under provisions in all States and Territories permitting disclosure where the organisation holding the information has a reasonable belief that the disclosure is necessary to lessen or prevent a serious threat to an individual's life, health, safety or welfare, or to public health, safety and welfare. The provisions vary slightly between jurisdictions, but all stipulate the requirement of a reasonable belief that the disclosure will diminish a serious threat or risk to a person or people.[32] Finally, disclosure to interstate and international agencies is possible under state legislative provisions such as the *Health Records Act 2001* (Vic) Health Privacy Principle 9 (Transborder Data Flows).

6.3.1.4 Protections applying to information held by law enforcement agencies

Once health-related genetic information has been provided to Australian law enforcement agencies, they are bound by the applicable privacy and data protection and/or health records legislation in their use and further disclosure of the information. If the agency is an APP entity under the *Privacy Act*, it is subject to APP 5 which requires notification to the individual of the collection of personal information if it is 'reasonable in the circumstances'. Not all jurisdictions provide special protection to genetic information specifically.

26 The *Health Records (Privacy and Access) Act 1997* (ACT) does not contain specific permission to disclose for law enforcement purposes, but public sector health services in the ACT must also comply with the Commonwealth *Privacy Act 1988*, which does contain such permission.

27 Department of the Premier and Cabinet Circular, *Information Privacy Principles Instruction* (Cabinet Administrative Instruction PC012, 6 February 2017) cl 10(e) <https://www.dpc.sa.gov.au/resources-and-publications/premier-and-cabinet-circulars/DPC-Circular-Information-Privacy-Principles-IPPS-Instruction.pdf> ('*SA Information Privacy Principles*'); *Health Services Act 2016* (WA) s 220(1)(g); *Health Records Act 2001* (Vic) sch 1 cl 2.2(j); *Information Act 2002* (NT) sch 2 cl 2.1(g); *Hospital and Health Boards Act 2011* (Qld); *Personal Information Protection Act 2004* (Tas) sch 1 cl 2(1)(g); *Health Records and Information Privacy Act 2002* (NSW) sch 1 cl 11(1)(j) ('*NSW Health Records and Information Privacy Act*').

28 See, eg, *Hospital and Health Boards 2011* (Qld) s 142(1); *Health Services Act 1988* (Vic) s 141(2).

29 *Health Services Act 2016* (WA) s 220(1)(d)-(f); *Health Records Act 2001* (Vic) sch 1 cls 2.2(c),(j) (including the 'no breach of confidence' criterion); *Information Act 2002* (NT) sch 2 cls 2.1(f), (g)(v); *Health Records (Privacy and Access) Act 1997* (ACT) sch 1 cl 10.2(e); *Information Privacy Act 2009* (Qld) sch 4 cls 2(1)(f), (g)(v); *Personal Information Protection Act 2004* (Tas) sch 1 cls 2(1)(f), (g)(v).

30 *SA Information Privacy Principles* (n 27) cl 10(d); *Health Care Act 2008* (SA) s 93(3)(a).

31 *Health Records and Information Privacy Act 2002* (NSW).

32 *SA Information Privacy Principles* (n 27) cl 10(c); *Health Care Act 2008* (SA) s 93(3)(e); *Health Services (Information) Regulations 2017* (WA) regs 5(1)(a)-(b); *Health Records Act 2001* (Vic) sch 1 cl 2.2(h); *Information Act 2002* (NT) sch 2 cl 2.1(d); *Health Records (Privacy and Access) Act 1997* (ACT) sch 1 cl 10.2(d); *Information Privacy Act 2009* (Qld) sch 4 cl 2(1)(d); *Personal Information Protection Act 2004* (Tas) sch 1 cl 2(1)(d); *Health Records and Information Privacy Act 2002* (NSW) sch 1 cl 11(1)(c).

6.3.1.5 Sample overseas provisions

In Europe, the processing of data for law enforcement purposes is regulated not by the *General Data Protection Regulation* but instead by EU Directive (Directive 2016/680).[33] Under national legislation giving effect to the Directive, there may be special protections pertaining to the processing of genetic data as a category of sensitive data. For example, Chapter 2 of Part 3 of the *Data Protection Act 2018* (UK) addresses law enforcement data processing and establishes six data protection principles requiring that:

- processing be lawful and fair;[34]
- purposes of processing be specified, explicit and legitimate;[35]
- personal data be adequate, relevant and not excessive;[36]
- personal data be accurate and kept up to date;[37]
- personal data be kept for no longer than is necessary;[38]
- personal data be processed in a secure manner.[39]

If the data subject has not given consent for the processing of genetic data for law enforcement, then processing can only occur for a stipulated purpose set out in Schedule 8. Further, it must be 'strictly necessary for the law enforcement purpose'.[40]

6.3.2 COMMERCIAL ACCESS

Unlike the situation involving data sharing for law enforcement, the re-use of health- or research-related genomic information for commercial benefit is rarely addressed explicitly in Australian law. The *My Health Records Act* is an exception; it precludes anyone (other than the healthcare recipient) using an individual's personal health information from the My Health Record for insurance or employment purposes.[41] It also prohibits ADHA from providing de-identified data to any insurer.[42]

33 *Council Directive (EU) 2016/680 of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data by competent authorities for the purposes of the prevention, investigation, detection or prosecution of criminal offences or the execution of criminal penalties, and on the free movement of such data, and repealing Council Framework Decision 2008/977/JHA* [2016] OJ L 119/89.

34 *Data Protection Act 2018* (UK) s 35(1).

35 Ibid s 36(1)(a).

36 Ibid s 37.

37 Ibid s 38(1)(a).

38 Ibid s 39(1).

39 Ibid s 40.

40 Ibid s 35(5)(a).

41 *My Health Records Act 2012* (Cth) ss 70A-B, 71A.

42 Ibid s 16.

Further, the secondary use of My Health Record data, for research and public health purposes, including any genomic information held in the My Health Record, is governed by the *Framework to Guide the Secondary Use of My Health Record System Data* (May 2018).[43] This provides for certain protections in regard to the secondary commercial use of data held in the system, for research and public health purposes, including the following:

- a mechanism allowing data subjects to opt-out of all secondary use of their My Health Record data;[44]
- a requirement that the data from the system 'cannot be used solely for commercial ... purposes'[45], although data use by commercial entities that is likely to be in the public interest and/or generate public health benefits is permissible. Direct marketing to consumers and uses of the data for assessing insurance premiums or claims is expressly precluded under the *Framework*;[46]
- a third-party data custodian, being the Australian Institute of Health and Welfare;[47]
- a governing body: the My Health Record Secondary Use of Data Governance Board, with wide representation. This Board will assess applications for access to this data for secondary use;[48]
- a requirement that the data be kept in Australia.[49]

Neither Commonwealth nor State and Territory legislation that applies to the secondary use of health data addresses access for commercialisation purposes specifically. Such access is possible under broader disclosure provisions both with and without the consent of the data subject.

In many or most cases, consent will be an available legal basis for commercial use that is envisaged at the outset, as well as for secondary (commercial) use of health-related genomic information. Generalised consent requirements are addressed consistently in legislation across Australia.[50] The consent must be adequately informed, voluntary, current and specific, and given by a person with legal capacity.[51] Where, as part of the consent process, the information provided about the proposed data use by a commercial entity is inadequate, such use may constitute a breach of the Australian Consumer Law misleading or deceptive conduct provisions (section 18).[52]

43 Australian Government, *Framework to Guide the Secondary Use of My Health Record System Data* (Framework, May 2018) <https://www1.health.gov.au/internet/main/publishing.nsf/Content/eHealth-framework>.

44 Ibid 19.

45 Ibid 7.

46 Ibid 63.

47 Ibid 4.

48 Ibid 4.

49 Ibid 5.

50 *Privacy Act* (n 17) sch 1 cl 6.1(a); *SA Information Privacy Principles* (n 27) cl 10(b); *Health Services Act 2016* (WA) s 220(1)(h); *Health Records Act 2001* (Vic) sch 1 cl 2.2(b); *Information Act 2002* (NT) sch 2 cl 2.1(c); *Health Records (Privacy and Access) Act 1997* (ACT) sch 1 cl 10(2)(c); *Information Privacy Act 2009* (Qld) sch 4 cl 2(1)(b); *Personal Information Protection Act 2004* (Tas) sch 1 cl 2(1)(b); *Health Records and Information Privacy Act 2002* (NSW) sch 1 cl 11(1)(a).

51 *APP Guidelines* (n 19) ch B <https://www.oaic.gov.au/privacy/australian-privacy-principles-guidelines>.

52 See, eg, *ACCC v HealthEngine Pty Ltd*: Australian Competition and Consumer Commission, Concise Statement', Notice of Filing in *ACCC v HealthEngine Pty Ltd*, NSD1255/2019, 7 August 2019. <https://www.accc.gov.au/system/files/ACCC%20v%20HealthEngine%20Pty%20Ltd%20_Concise%20Statement.pdf>.

Under the *Privacy Act* s 16B(3), the disclosure of genomic information without a person's consent, for research relevant to public health or public safety (which might include commercial research[53]) may be permitted. An entity holding identifiable genomic information may choose to disclose it under this provision if:

(a) the use or disclosure is necessary for research, or the compilation or analysis of statistics, relevant to public health or public safety; and

(b) it is impracticable for the organisation to obtain the individual's consent to the use or disclosure; and

(c) the use or disclosure is conducted in accordance with guidelines approved under section 95A for the purposes of this paragraph; and

(d) in the case of disclosure—the organisation reasonably believes that the recipient of the information will not disclose the information, or personal information derived from that information.

The s 95A Guidelines that are referred to in subsection (c) outline the requirement for the proposed research to be approved by a Human Research Ethics Committee (HREC).[54] That the Guidelines do not mention commercial use explicitly is an opportunity for reform, and alignment with the principles for secondary use of My Health Record data. Currently, the Guidelines apply to the research use of genomic information without consent, on the basis of an opt-out approach or a waiver of consent. In both instances only low-risk research can be pursued using these approaches, which may limit their application in the context of using *identifiable* genomic information. Further, in determining whether to grant a waiver of consent, the *National Statement* requires that HRECs be satisfied that 'the possibility of commercial exploitation of derivatives of the data or tissue will not deprive the participants of any financial benefits to which they would be entitled'.[55] Hence both the data steward and a HREC would need to be satisfied that the disclosure is necessary, that consent is impracticable, that the research is low risk, that the commercial use will not deprive the data subjects of a financial return, and that the entity receiving the data will not disclose it further. In practice it is unclear whether or not these criteria offer sufficient protection against the unconsented disclosure of genomic data for research.

The use of deidentified genomic data without the consent of the data subject, for the purposes of research, is also addressed in broadly similar terms in State and Territory legislation and subsidiary guidelines in most jurisdictions (excepting South Australia).[56]

53 The phrase 'relevant to public health or public safety' is not defined. Guidelines issued by the Office of the Australian Information Commissioner contain the following illustrative examples: 'research or the compilation or analysis of statistics relating to communicable diseases, cancer, heart disease, mental health, injury control and prevention, diabetes and the prevention of childhood diseases': APP Guidelines (n 19) cl D.13.

54 National Health and Medical Research Council, *Guidelines Approved under Section 95A of the Privacy Act 1988* (2015) <https://www.nhmrc.gov.au/about-us/publications/guidelines-approved-under-section-95a-privacy-act-1988>.

55 National Health and Medical Research Council, *National Statement on Ethical Conduct in Human Research 2007* (Updated 2018) para 2.3.10(h).

56 *Information Act 2002* (NT) sch 2 cl 2.1(ca); *Health Records Act 2001* (Vic) sch 1 cl 2.2(g); *Health Services Act 2016* (WA) s 216(d); *Health Services (Information) Regulations 2017* (WA) reg 3; *Health Records (Privacy and Access) Act 1997* (ACT) sch 1 cl 10(3); *Information Privacy Act 2009* (Qld) sch 4 cl 2(1)(c); *Personal Information Protection Act 2004* (Tas) sch 1 cl 2(1)(c); *Health Records and Information Privacy Act 2002* (NSW) sch 1 cl 11(1)(f).

7 Return of Findings

7.1 Summary

The generation of large amounts of genomic data by research laboratories and clinics across Australia raises questions as to the extent to which this data should be analysed, and the extent to which the findings from this analysis should be returned to individuals. Advances in our understanding of the significance of genomic variants are further complicating this matter, on the basis that variants once considered to be of unknown significance could potentially become relevant to the individual. This might be because variants become recognized as pathogenic, or because variants previously thought of as pathogenic may be reclassified as benign.

No single regulatory instrument provides complete guidance on the legal obligations that clinicians and researchers need to take into account in deciding which of the findings from their genomic analyses should be returned to individuals. Guidance will come from privacy legislation, the regulatory regime related to genetic testing, accreditation of testing laboratories and best practice standards for genomic analysis and return of findings, the research ethics system, and tort law. The scope of patient/participant consent, discussed in Chapter 2, is also clearly a relevant consideration.

In the context of clinical genomic analysis, the National Pathology Accreditation Advisory Council ('NPAAC') provides detailed best practice standards in a series of documents, the most relevant of which are discussed below. However, the precise scope and content of the obligations of laboratories with regard to analysis and return of clinical findings is not articulated. While the courts may ultimately provide such guidance through actions in tort law, they have not yet had the opportunity to do so.

Recent amendments to the *National Statement on Ethical Conduct in Human Research ('National Statement')* have clarified the types of matters that researchers need to take into account in making decisions about what to analyse and what to return to individuals. It is not yet clear whether these new amendments provide adequate guidance to researchers and Human Research Ethics Committees ('HRECs') in determining the precise scope and content of their obligations with regard to analysis and return of research findings.

7.2 Introduction

The technological capacity to undertake genomic analysis has changed dramatically in the past 30 or so years, since the start of the human genome project. Genomic research and clinical practice are no longer constrained by the limited capacity to compare single genes from an individual with reference genes known to carry mutations connected with particular disease manifestations. There are many and varied ways in which genomic analysis is undertaken, ranging from mapping of single nucleotide polymorphisms through to analysing whole genome or whole exome sequences. Whole genome sequencing is now frequently performed for both research and clinical purposes, although there is still lingering doubt about its efficacy from a health economics perspective.[1]

1 Katharina Schwarze et al, 'Are Whole-exome and Whole-genome Sequencing Approaches Cost-Effective? A Systematic Review of the Literature' (2018) 20 *Genetics in Medicine* 1122.

As a consequence of these technological developments, the amount of genomic data that could exist about a particular individual has increased massively. However, this raw sequence data is of limited value to the individual unless it has been analysed. This highlights the importance of maintaining a clear distinction between data and findings.

In the clinical context, analysis of sequence data may provide findings to the individual about the presence or absence of clinically relevant sequence variants sought out by them (usually through their physician). However, this genomic data analysis may also reveal variants that are of unknown significance ('VUS'). Variants that are currently of unknown significance may become clinically relevant over time, as more information is accumulated.

Genomic analysis may also incidentally reveal the presence of variants that are clinically relevant, but that were not actually sought out by the individual. In the research context, genomic analysis may be directed towards population-wide trends rather than identifying variants specific to an individual. Each of these aspects of genomic analysis raises complex ethical, legal and social questions around the extent of genomic analysis that should be undertaken and the obligation to return the findings from this analysis to the individual. The potential for harm from overdiagnosis is clearly a key consideration in this context.[2]

This chapter considers laws and other forms of regulation that are relevant both to the types of analyses of genomic variants that should be undertaken and to the obligations to return the findings of those analyses. The language of 'return of findings of genomic analysis' has been adopted, because this is the language that is commonly used in the policy and academic literature.[3]

It should be noted that the *National Statement*[4] variously uses the language of 'return of results or findings', 'return of findings or results' and 'return of findings'. These terms are not defined and appear to be used interchangeably, suggesting that it is not necessary to distinguish between return of results and return of findings.

A recent review of international, regional and national laws relating to return of research results and incidental findings across 20 countries revealed a wide divergence in approaches.[5] This illustrates that there is no one jurisdiction to which Australia might turn for guidance. In this chapter, the approach in the USA is highlighted, given the particular direction that jurisdiction has taken with regard to return of incidental findings.

The legal and regulatory environment within which genomic analysis and return of the findings from that analysis are situated is complex, whether the analysis is undertaken for clinical or for research purposes. The first reason for this is the breadth of the options available to clinicians and researchers for genomic analysis and for return of findings. The second reason is because the legal and regulatory requirements relating to genomic analysis and return of findings are highly context dependent. In the case of clinically relevant findings resulting from genomic analyses undertaken for clinical purposes, it seems clear that a clinician has both ethical responsibilities and legally binding duties to their patient.[6]

2 Karen M Meagher and Jonathan S Berg, 'Too Much of a Good Thing? Overdiagnosis, or Overestimating Risk in Preventive Genomic Screening' (2018) 15 *Personalised Medicine* 343.

3 See, eg, Robert C Green et al, 'ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing' (2013) 15(7) *Genetics in Medicine* 565.

4 NHMRC, Australian Research Council and Universities Australia, 'National Statement on Ethical Conduct in Human Research' (Australian Government, 2007, Updated 2018) ('*National Statement*').

5 Adrian Thorogood, Gratien Dalpé and Bartha Maria Knoppers, 'Return of Individual Genomic Research Results: Are Laws and Policies Keeping Step?' (2019) 27 *European Journal of Human Genetics* 535.

6 See, eg, Australian Medical Association, *Code of Ethics*, particularly Chapter 4, Ethics of Genetics & Reproductive Medicine <https://www.ama-assn.org/delivering-care/ethics/code-medical-ethics-genetics-reproductive-medicine>; *Breen v Williams* (1996) 186 CLR 71; *Rogers v Whitaker* (1992) 175 CLR 479; *Rosenberg v Percival* (2001) 205 CLR 434.

The precise scope of these responsibilities and duties remains somewhat unclear – for example, what findings must be returned, who should be responsible for the return of findings, and must return of findings be accompanied by provision of genetic counselling?[7] Although it is hard to imagine a circumstance where there would be an obligation not to return findings of analyses directly related to clinical care, there is a body of commentary suggesting caution about the return of incidental findings, given that this may expose a clinician to legal liability.[8] Clinician liability is, of course, but one consideration. Other complex considerations apply when considering harm to the individual and their family, particularly in the context of return of incidental findings. The third reason for this complexity is that laws and other regulations have traditionally made a clear distinction between research and clinical practice.

FIGURE 7: RETURN OF FINDINGS FROM GENOMIC ANALYSIS

TYPES OF FINDINGS

- Clinically relevant findings requested by clinician
- Incidental clinically relevant findings
- Finding of variants of unknown significance (VUS)
- Clinically relevant findings from reanalysis of VUS
- Individual research findings
- Cumulative research findings
- Raw sequence data

REGULATORY INSTRUMENTS

- Privacy laws
- National and international standards and accreditation requirements
- In vitro diagnostic medical devices regulation
- Human research ethics
- Tort law

As discussed elsewhere in this report, it is becoming increasingly difficult to make this clear distinction between research and practice. As succinctly noted by NPAAC, '[t]he unclear boundary between validated medical testing and medical research is recognised across all pathology testing.'[9]

7 Although now slightly dated the following National Health and Medical Research Council (NHMRC) publication addresses some of these issues: NHMRC, 'Medical Genetic Testing: Information for Health Professionals' (Information Paper, Australian Government, National Health and Medical Research Council, April 2010) <https://www.nhmrc.gov.au/about-us/publications/medical-genetic-testing-information-health-professionals>.

8 See, eg, Ellen Wright Clayton and Amy L McGuire, 'The Legal Risks of Returning Results of Genomics Research' (2012) 14(4) *Genetics in Medicine* 473; Amy L McGuire et al, 'Can I be Sued for That: Liability Risk and the Disclosure of Clinically Significant Genetic Research Findings (2014) 24 *Genome Research* 719.

9 Australian National Pathology Accreditation Advisory Council, *Requirements for Human Medical Genome Testing Utilising Massively Parallel Sequencing Technologies* (2017), 1, <https://www1.health.gov.au/internet/main/publishing.nsf/Content/FB649C2C2A42CACDCA2580A400039643/$File/Reqs%20MPS%20Technologies%202017.pdf>.

Aside from analysis and return of clinically relevant findings specifically requested by the individual, through their clinician, the main points that need to be considered include:

- return of findings from clinical genomic analysis that constitute VUS;
- return of findings from research-related genomic analysis (whether cumulative or individual, whether of unknown significance, or potentially clinically relevant, once verified);
- genomic analysis for and return of incidental findings, in both clinical and research-related contexts (noting that the term 'incidental' is, of itself, loaded with ambiguity – this issue will be addressed in the next section);
- re-analysis and return of findings from genomic analysis when new information comes to light (in either the clinical or research setting); and
- return of raw sequence data from clinical and research-related genomic analysis.

7.3 Stakeholder Views and Key Points from the Literature

Stakeholder interviews undertaken for the scoping review illustrate the problematic nature of these issues. They raise questions such as: what constitutes clinically relevant information; what constitutes meaningful consent in the context of incidental findings; in what circumstances should changing views on clinical significance be reported to individuals; and how should privacy and research ethics be understood in the context of raw sequence data?

There is a large body of literature debating the propriety of return of findings in each of these contexts. The key outcomes from this literature analysis are summarised in the following six points:

1 It appears from the literature that it is appropriate to draw a clear distinction between those findings that must be returned to the individual because they are of clinical relevance and those findings that are of unknown significance, where the existence of an obligation to return is much less certain. As Friedman and colleagues have asked, 'The question is, in what circumstances should uncertain results be fed back to patients and their families given associated burdens on the healthcare system and potential harms from false or uncertain information?'[10]

2 There is uncertainty around whether there is an obligation to undertake analysis to identify the presence or absence of common disease-causing variants in the clinical context. The American College of Medical Genetics and Genomics ('ACMG') has attempted to address this matter, by issuing a set of guidelines with a 'minimum list' of variants that should be assessed by clinicians on the basis that they have a high degree of clinical validity and utility.[11] To date, the ACMG guidelines have not been endorsed in the clinical context in Australia. Rather, organisations including the Human Genetics Society of Australasia[12] and NPAAC have called for more conservative approaches.[13]

3 Although it has been argued that an obligation to undertake analysis to identify common disease-causing variants should also flow to genomic researchers,[14] this appears not to have been accepted in Australia.[15]

10 Jan M Friedman et al, 'Genomic Newborn Screening: Public Health Policy Considerations and Recommendations' (2017) 10(9) *BMC Medical Genomics* 1, 6 <https://doi.org/10.1186/s12920-017-0247-4>.

11 Green et al (n 3); Robert C Green et al, 'Corrigendum: ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing' (2017) 19 *Genetics in Medicine* 6.

12 Human Genetics Society of Australasia, HGSA Commentary on ACMG Recommendations (Web Page, 2018) <https://www.hgsa.org.au/hgsanews/hgsa-commentary-on-acmg-recommendations>.

13 Australian National Pathology Accreditation Advisory Council (n 9). See also, Lisa Eckstein and Margaret Otlowski, 'Strategies to Guide the Return of Genomic Research Findings: An Australian Perspective' (2018) 15 *Journal of Bioethical Inquiry* 403, 405 <https://doi.org/10.1007/s11673-018-9856-7>.

14 Julian J Koplin, Julian Savulescu and Danya F Vears, 'Why Genomics Researchers are Sometimes Morally Required to Hunt for Secondary Findings' (2020) 21(11) *BMC Medical Ethics* 1, <https://doi.org/10.1186/s12910-020-0449-8>.

15 Eckstein and Otlowski (n 13).

4 There is variability in the literature as to the meaning of the term 'incidental findings'[16] and the scope of any obligation to return them.[17] The ACMG uses the term to mean 'the results of a deliberate search for pathogenic or likely pathogenic alterations in genes that are not apparently relevant to a diagnostic indication for which the sequencing test was ordered.'[18] In contrast, the Australian National Health and Medical Research Council (NHMRC) defines incidental findings quite differently, focusing on their 'unexpected' nature. In the *National Statement* the following distinction is drawn between pertinent, secondary and incidental findings:

   i **Pertinent findings:** Also known as primary findings, pertinent findings are those that were the primary objects of the investigation.

   ii **Secondary findings:** Findings that were not the primary target of the investigation, but were either specifically sought or are related to the primary target and anticipated as likely to arise.

   iii **Incidental findings:** Findings of potential clinical significance unexpectedly discovered during the investigation. NB: With respect to full spectrum 'discovery' investigations and direct-to-consumer testing, one is explicitly searching for any and all findings and so no findings can be considered 'unexpected'.[19]

   Given that the *National Statement* refers to secondary and incidental findings of genomic analysis collectively, the term 'incidental findings' is adopted in this chapter to include both findings for which searches are deliberately undertaken (secondary findings using the language of the *National Statement*) and unexpected findings.

5 On reanalysis and recontact in the research context, a 2019 position statement developed by the American Society of Human Genetics[20] and endorsed by the Human Genetics Society of Australasia[21] advises that there will be an obligation to do so in some circumstances.

6 On return of un-analysed raw sequence data, empirical research reveals that potential research participants have considerable interest in receiving such data, including to use it to seek out their own clinical interpretations.[22] However, scholars raise concerns about the accuracy and utility of such data, and the consequences that may flow from its release.[23]

In sum, there is a body of literature discussing each of these issues, but there is a lack of clear consensus on the way forward. These are important questions, yet given the speed of technological advances and the lack of consensus as to the approach to be taken, it is difficult for legal and regulatory responses to keep pace. The Australian Law Reform Commission and Australian Health Ethics Committee report, *Essentially Yours*, makes no mention of terms such as return of findings, return of results or incidental findings.[24] This is hardly surprising, given that whole genome sequencing and whole exome sequencing were not widely available in 2003.

16 Noting that incidental findings can also be referred to as 'unsolicited' or 'secondary' findings. See Koplin, Savulescu and Vears (n 14).

17 Green et al (n 3); Green et al 'Corrigendum' (n 11); Lisa Eckstein, Jeremy R Garrett and Benjamin E Berkman, 'A Framework for Analyzing the Ethics of Disclosing Genetic Research Findings' (2014) 42(2) *Journal of Law, Medicine & Ethics* 19.

18 Green et al (n 3); Green et al 'Corrigendum' (n 11).

19 *National Statement* (n 4) 52.

20 Yvonne Bombard et al, 'The Responsibility to Recontact Research Participants after Reinterpretation of Genetic and Genomic Research Results' (2019) 104(4) *American Journal of Human Genetics* 578, <https://www.sciencedirect.com/science/article/pii/S0002929719300709>.

21 Ibid.

22 Anna Middleton et al, 'Potential Research Participants Support the Return of Raw Sequence Data' (2015) 52(8) *Journal of Medical Genetics* 571 <https://doi.org/10.1136/jmedgenet-2015-103119>.

23 Adrian Thorogood et al, 'APPLaUD: Access for Patients and Participants to Individual Level Uninterpreted Genomic Data' (2018) 12(1) *Human Genomics* 1 <https://doi.org/10.1186/s40246-018-0139-5>.

24 Australian Law Reform Commission and Australian Health Ethics Committee, *Essentially Yours: The Protection of Human Genetic Information in Australia* (Report No 96, May 2003) 355 ('*Essentially Yours*').

Since then, there have been some developments in Australia which attempt to address these issues. For example, the NHMRC has considered the issues associated with return of research findings from genomic analysis in some depth in its rolling review of the *National Statement*. The most significant amendments to the *National Statement* in the context of genomic analysis occurred in 2018, as discussed below. The NHMRC has also produced a set of useful resources relating to genomics for health professionals and consumers, some of which provide guidance on return of findings.[25]

In the clinical context, the 2017 NPAAC guidance document on massively parallel sequencing (*Requirements for Human Medical Genome Testing Utilising Massively Parallel Sequencing Technologies*),[26] together with an earlier document, *Requirements for Medical Testing of Human Nucleic Acids*,[27] provide a set of minimum best practice standards. Two other NPAAC guidance documents, *Requirements for Cytogenetic Testing* (Third Edition 2013) and *The Provision of Direct-to-Consumer Genetic Tests: Guiding Principles for Providers* (Second Edition 2014) are also relevant.

Together with the overarching *Requirement for Medical Pathology Services* (Third Edition 2018),[28] these documents provide guidance on the requirements that providers of such services must adhere to in order to achieve accreditation. The requirement for accreditation of pathology laboratories is specified in the the Health Insurance (Accredited Pathology Laboratories-Approval) Principles (2017), made under the *Health Insurance Act 1973* (Cth). Accreditation is required for a pathology laboratory to be eligible for Medicare reimbursement.

The NPAAC guidance documents also represent minimum best practice standards for good laboratory practice that are harmonised with international best practice (where relevant) and are comprehensive but not prescriptive.

The National Association of Testing Authorities (NATA) is the only assessing body currently endorsed for pathology accreditation in Australia.[29]

The legal and regulatory issues relating to return of clinical and, particularly, research findings, including incidental findings, have received considerably more attention elsewhere, most notably in the US. The US National Academy of Science, Engineering and Medicine has undertaken a major review of return of individual findings to research participants, a comprehensive report of which was published in 2018 ('National Academies Report').[30] Chapter 6 of that report provides a detailed assessment of the legal and regulatory landscape within which return of individual research findings is situated.[31]

The analysis in chapter 6 of the National Academies Report indicates that major federal laws and other regulations in the US could, on the one hand, create barriers to the return of individual research findings to participants, or, on the other hand, create onerous obligations on researchers and clinicians.[32] Perhaps the worst case scenario is for these instruments to be conflicting or otherwise uncertain or ambiguous.

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25 'Genomics Resources for Clinicians and Researchers', *NHMRC* (Web Page) <https://www.nhmrc.gov.au/health-advice/genomics/genomics-resources-clinicians-and-researchers>.

26 Australian National Pathology Accreditation Advisory Council (n 9).

27 NPAAC, *Requirements for Medical Testing of Human Nucleic Acids* (2013).

28 NPAAC, *Requirement for Medical Pathology Services* (Third Edition 2018)

29 Department of Health, National Pathology Accreditation Advisory Council (NPAAC) (Web Page) <https://www1.health.gov.au/internet/main/publishing.nsf/Content/health-npaac-index.htm>.

30 National Academies of Sciences, Engineering, and Medicine, *Returning Individual Research Results to Participants: Guidance for a New Research Paradigm*, ed Autumn S Downey, Emily R Busta, Michelle Mancher and Jeffrey R Botkin (National Academies Press, 2018).

31 Ibid 241-75.

32 Ibid.

The analysis presented in that chapter illustrates the legal and regulatory quagmire facing US-based researchers. The following quote from the National Academies Report sums up the challenges.

> As currently written and implemented, ... the laws and regulations governing access to laboratory results are not harmonized; they afford inconsistent and inequitable access for participants to permitted results, and the regulatory conflicts create dilemmas for laboratories, forcing them to choose which regulation to intentionally violate in order to comply with the other.[33]

The analysis presented in this chapter suggests that we are not presently facing quite the same quagmire in Australia. However, this may be because Australia has not yet addressed some of the questions that have been considered in the US.

The rest of this chapter is organized into three sections. First, obligations imposed by NPAAC and National Association of Testing Authorities ('NATA') accreditation requirements, privacy legislation and the Therapeutic Goods Administration ('TGA') will be considered in the Australian context under the heading 'Regulation in the Clinical Context'. Secondly, a separate and more detailed analysis of the research ethics obligations follows. This is necessary, given the detailed attention to this issue in the *National Statement*. In addition to the legal and regulatory instruments, tort law may also have an important role to play in determining the obligations of clinicians and researchers to return/not to return findings from genomic analysis. The role of Australian tort law will be a focus for analysis in the last section this chapter.

7.4 Regulation in the Clinical Context

As noted above, the major regulatory instruments that are relevant in the context of return of findings from genomic analysis are NATA accreditation (which requires compliance with NPAAC standards), TGA requirements, privacy provisions and research ethics obligations. The first three of these are considered in this section. The detailed requirements in the *National Statement* relating to return of findings are discussed in a separate section, below.

It should be noted at this juncture, however, that although focus of the *National Statement* is on research, it does include clinical findings in its decision tree for the management of findings in genomic research and health care (discussed further below).[34] In the clinical context, the decision tree distinguishes between findings that are pertinent to the indication for testing, where standard clinical practice should be followed, and secondary or incidental findings (as defined) where policy or patient preferences should be followed if the patient has consented, and current best practice or national clinical genomics guidelines should be followed if the patient has not consented. As such, the *National Statement* does not add any further layers of regulation in the clinical context, but merely directs the reader to current regulatory instruments and best practice standards, which are discussed below.

To date, no attempt has been made to coordinate reform of the regulatory environment for return of results. Chapter 6 of the US National Academies Report illustrates the importance of a coordinated approach. It shows that well intentioned but uncoordinated reforms can create regulatory dilemmas. For example, significant amendments were made in 2014 to laboratory accreditation requirements and privacy obligations in the US, in light of new technological developments. Yet it appears that the outcome may have been less than optimal for the research sector. According to the National Academies Report, the *Health Insurance Portability and Accountability Act of 1996* (HIPAA) Privacy Rule ('HIPAA Privacy Rule') applies to all laboratories of 'covered entities' (including universities with teaching hospitals) that hold clinically relevant genomic data. The Rule requires them to comply with requests from individuals to access any of their research findings held in 'designated record sets'. Release of findings that are not yet interpreted raises concerns about undue anxiety or unnecessary intervention.[35]

33 Ibid 245.

34 *National Statement* (n 4) 52.

35 National Academies of Sciences, Engineering, and Medicine (n 30) 251.

The report also suggests that any laboratory that is part of a HIPAA covered entity and holds potentially clinically actionable findings needs to be accredited under the Clinical Laboratory Improvement Amendments of 1988 ('CLIA'). The authors of the National Academies report see the requirement for CLIA accreditation as a 'formidable obstacle to the return of individual results to research participants even though the results may meet other quality standards and a right of access to laboratory results has been gaining credibility in other regulatory policies.'[36]

In the Australian context, as noted in Chapter 8, all medical devices including in vitro diagnostic medical devices ('IVD medical devices') supplied in Australia must be included in the Australian Register of Therapeutic Goods ('ARTG'). The regulatory requirements for medical devices in Australia are broadly consistent with other international jurisdictions. It is not a requirement for 'in-house human genetic tests to be included in the ARTG'.[37] This includes genomic analysis undertaken in a laboratory without the use of commercially supplied kits (i.e., using a laboratory developed test), or using modified commercial kits.

Even so, there is still a requirement that a laboratory that is manufacturing in-house IVD medical devices must comply with the conformity assessment procedure provided in pt 6A sch 3, of the *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth). Clause 6A.2 requires the laboratory manufacturing the in-house IVD medical device to satisfy three requirements. It must first notify the TGA of the in-house IVD medical devices it manufactures. Secondly, it must meet the NPAAC standard, Requirements for the Development and Use of In-house In Vitro Diagnostic Medical Devices. Thirdly, it must be accredited as a testing laboratory by NATA as meeting international standards.

The TGA requirements relating to IVD medical devices only apply to therapeutic goods. As such, they do not apply to research laboratories making research findings (i.e. findings for information purposes only and that are not being returned to a clinician or patient, or being used to diagnose, treat or manage a person). Research laboratories that return clinically relevant findings must either comply with the regulatory requirements for IVD medical devices or apply for an exemption to use an unapproved medical device (e.g., a clinical trial exemption).

The TGA, NATA and NPAAC requirements apply to clinically relevant findings. This means that they apply just as much to clinically relevant incidental and re-analysed findings as to original findings. The NPAAC guidance document on massively parallel sequencing is particularly helpful in this regard.[38] It sets out the frameworks, consent documentation and policies that medical pathology laboratories must have in place to comply with best practice standards, with particular guidance on the context of incidental findings.

This NPAAC document includes a set of standards and a set of commentaries on each of those standards.

Of particular relevance, Standard 1.6 requires that laboratories have a clear policy on the reporting of incidental findings. This policy must be made available to patients and clinicians on request. In Commentary 1.6(ii), linked to this standard, it is noted, inter alia, that 'Data management strategies should consider the masking of information that is outside the scope of testing for a given patient sample.' Standard 3.1 states further that 'The laboratory must define the scope of testing in advance of the provision of any genomic pathology test with a view to ensuring it is able to provide a quality pathology service.' Commentary 3.1(i) adds that targeting 'only genes relevant to the disease being investigated will assist with minimising incidental findings'. Standard 8.7 requires laboratories performing genomic testing to have clear policies for the disclosure of incidental findings, and Commentary 8.7(i) adds that 'Laboratories must limit the reporting of incidental findings to variants that are unequivocally classified as pathogenic or likely pathogenic.'

36 Ibid 248.

37 See Chapter 8.

38 Australian National Pathology Accreditation Advisory Council (n 9).

The NPAAC massively parallel sequencing guidance document thus clearly indicates that any genomic analysis for and return of incidental findings should be undertaken with caution. However, neither this guidance document, nor other NATA and NPAAC documentation, provide assistance in determining the nature of any substantive legal obligations relating to: return of clinically relevant findings from genomic analysis; genomic analysis for and return of incidental findings; and analysis (and re-analysis) for, and return of, findings of unknown or changing significance. Nor do these provisions assist in determining the legal obligations that apply to return of research findings, or to return of raw sequence data.

In the context of Australian privacy laws, individuals have a right of access to personal information (with limitations).[39] As such, provided that there are no relevant limitations, and that all other requirements for access are met, these provisions require clinicians and researchers to make health and medical information about individuals available to them on request. This obligation to provide access to personal information is likely to apply most clearly to return of clinically relevant findings in the situation where a specific request is made for clinical genomic analysis by an individual or their physician.

The obligation might also extend to return of clinically relevant incidental findings, unless arguments could be made that one or more of the limitations on access apply. Examples include: where giving access would pose a serious threat to the life, health or safety of any individual, or to public health or public safety; or where giving access would have an unreasonable impact on the privacy of other individuals, as specified in Australian Privacy Principle 12 (APP 12).[40] However, the question of whether there is an obligation under Australian privacy laws to return non-clinically relevant findings from genomic analysis remains to be determined. In part, this will depend on what forms of genomic information satisfy the definition of 'personal information' in Australian privacy statutes (Chapter 3).

7.5 Research Ethics Obligations

The *National Statement* extends to any research involving humans that is in any way affiliated with an organisation receiving federal research funding. The scope and enforceability of the *National Statement* are discussed in Chapter 2. Here, the primary foci for consideration are the guidelines provided in the *National Statement* with regard to: return of findings from research-related genomic analysis; dealing with research findings of clinical significance; obligations for actively identifying secondary/incidental findings in the research context; reanalysis of research findings for variants of unknown significance; and return of raw sequence data.

The *National Statement* was significantly updated in 2018, adding a new Chapter 3.3 dealing specifically with genomic research and including a 'decision tree' for researchers and clinicians making decisions whether to return a genomic result.[41] As noted above, the arm of the decision tree focusing on clinical practice directs attention to standard clinical practice and best practice or national clinical genomics guidelines. The arm of the decision tree focusing on research provides more detailed guidance on the various steps that need to be considered in determining whether findings should or should not be returned to the research participant.

Paragraph 3.3.1 of the *National Statement* recommends that 'ethical issues that arise from activity outside the intended scope of the research are minimised by, for example, developing a list of genes that are excluded from analysis.'

39 See particularly *Privacy Act 1988* (Cth), Australian Privacy Principle 12. Equivalent provisions in state and territory legislation are also relevant. See also, Chapter 3.

40 Ibid Australian Privacy Principle 12.3(a)-(b).

41 *National Statement* (n 4) 52, noting that this decision tree comes from Principles for the translation of 'omics'.

The *National Statement* emphasises the importance of consent in defining the scope of research and the findings that should be returned. For instance, the list of matters to be included for consideration in assessing the appropriateness and scope of consent to genomic research in paragraph 3.3.10 includes, at point (d), 'which, if any, of the findings of the research will be communicated to participants and, if so, how.' Paragraph 3.3.11 also notes that researchers should advise participants that 'information that they may be given about the likely impact of the genomic information may change over time as new knowledge/insight is gained and how to obtain updated information'. Importantly, paragraph 3.3.17 points out that researchers should not assume participants wish to receive the findings of the research they participate in, but rather, if there is to be mandatory return, this should be made clear at the outset.

The genomics chapter also provides a whole section, Element 5, listing the considerations that researchers should take into account in deciding what research findings should be communicated to participants. In summary, paragraphs 33.3.26 and 3.3.27 make it clear that it is for the researcher to decide how and what to communicate, noting that some findings must be returned, some may be returned, and some should not be returned. It should be noted, however, that these decisions must be included in the researcher's ethically defensible plan (discussed further below) and must be approved by the relevant HREC.

Paragraph 3.3.28 explicitly addresses raw sequence data, noting that, while participants may have a strong interest in their individual findings, 'researchers are not expected to return raw genomic data to participants.' In the event that raw sequence data falls within the definition of 'personal information' in the *Privacy Act 1988* (Cth), this would seem to be at odds with APP 12, which requires access, subject to certain limitations. Paragraph 3.3.29 goes on to address clinical significance, noting that once there is sufficient evidence and agreement, participants should be advised that findings will only be returned via a NATA accredited clinical service. Paragraph 3.3.34 refers to reanalysis, noting the responsibility of researchers to give participants the opportunity to reconsider decisions relating to return of findings in light of substantive change in the understanding of the significance of the research findings over time. However, paragraph 3.3.35 notes that any obligation to reanalyse only extends to the end of the research project.

The next section of the *National Statement* requires researchers to develop an ethically defensible plan for the return of findings and individual research results. As noted in paragraph 3.3.36, the purpose of the plan is to 'manage the disclosure or non-disclosure of genomic information of potential importance for the health of research participants or their relatives'. The plan must address, in detail, each of the aspects of return of findings that is relevant to the research project, including: whether findings will be returned; in the event they are to be returned, how findings will be validated and assessed; how consent to return will be sought and how participants will be notified about the availability of relevant findings. The plan must also outline how participant privacy will be protected. The plan must be submitted to an HREC for approval.

While it seems implicit in this chapter of the *National Statement* that some form of return of research findings is required, there is no specific obligation as to what and how to return, aside from the must/may/should not considerations in paragraph 3.3.27. Paragraph 3.3.40 provides some further guidance on matters that that the researcher should consider and address these in the ethically defensible plan, noting that relevant factors include:

(a) analytic (scientific) and clinical validity;

(b) significance to the health of the participants/relatives; and

(c) clinical utility.

There is no mandatory requirement to return raw sequence data, nor to reanalyse data. Nor is there a prescribed list of variants that must be examined whenever research-related genomic analysis is undertaken. Rather, researchers may wish to draw up a list of genes that will be excluded.

What is mandatory is that the decisions made by the researcher regarding return of research findings must be clearly conveyed to the participant at the time that consent is sought. In particular, participants must know if return of specific findings is mandatory. It is also clear that any return of clinically relevant findings must be validated by a NATA accredited laboratory and returned via a clinician (paragraphs 3.3.29-3.3.32).

The *National Statement* thus provides researchers with detailed guidance as to the requirements they must comply with in order to secure ethical approval for research involving genomic analysis. However, it is arguably the case that the 2018 amendments do not provide much in the way of additional guidance to researchers in actually deciding what analyses should be undertaken and what findings should be returned to research participants.

Whenever findings have the potential to be scientifically and clinically valid, of health significance and of clinical utility (listed as relevant factors in the preparation of the ethically defensible plan), it must surely be the case that the researcher must recommend clinical validation to the participant. As such, this list of factors may provide little practical guidance on the scope of the obligation to return findings.

As with TGA, NATA and NPAAC requirements in the clinical context, the *National Statement* largely leaves to researchers and HRECs decisions about the substantive obligations of researchers relating to return of findings (in all their guises) and to re-analysis of findings.

While there has been little opportunity for analysis of the effectiveness of the 2018 amendments to the *National Statement*, some caution has been expressed in earlier commentary as to the extent to which HRECs have the capacity to provide adequate advice to researchers on their obligations with regard to return of findings.

In a 2014 article examining return of research findings in the context of the Australian Pancreatic Cancer Genome Initiative, it was concluded that return of individual results, including incidental findings, was both feasible and ethically defensible. The importance of a dynamic, ethically defensible plan was emphasised, but it was noted that 'perhaps ethical review boards lack the knowledge or expertise to guide researchers adequately on this topic.'[42] A later article by the same group found that the overall process of managing return of findings was 'a resource-demanding activity... labour-intensive, costly and time-consuming.'[43] Despite this, it was concluded that return of findings 'stands to be of significant value to participants and family members if we can work to steadily remove the clinical and procedural barriers to implementation.'[44]

In the context of the 2018 amendments, some might argue that researchers could avoid some of the extensive obligations regarding return of findings by narrowly presenting the scope of their research, as they are required to do through paragraph 3.3.41(c). By so doing, they could significantly narrow their obligations with regard to returnable findings. On the other hand, if they adopt a more expansive (and possibly more realistic) presentation of the scope of their research, the requirements they face with regard to consent and development of an ethically defensive plan are substantial, with some 50 specific points that need to be addressed.

HRECs are also potentially exposed to significant additional workload to properly assess these ethically defensible plans. This suggests that more work may need to be done to ensure national consistency in approaches to return of research findings by researchers and HRECs. HRECs and researchers may also need further guidance and training on the scope of their ethical and legal obligations with regard to genomic analysis and return of findings, and how best to meet these obligations. Researchers also need more training and education on IVD medical device regulatory requirements and obligations.

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42 Amber L Johns, et al, 'Returning Individual Research Results for Genome Sequences of Pancreatic Cancer' (2014) 6(5) *Genome Medicine* 42.

43 Amber L Johns, et al. 'Lost in Translation: Returning Germline Genetic Results in Genome-scale Cancer Research' (2018) 9(1) *Genome Medicine* 41.

44 Ibid.

Given that researchers would be required to undertake additional analyses should there be an increased expectation that incidental research findings will be returned, it seems prudent to consult with the research community about the feasibility of this approach. A recently published article examined the practices of two community-based genomic studies following the 2018 amendments to the *National Statement*. The 2018 article by Johns et al highlights some of the dilemmas faced by researchers in deciding what findings should be returned to participants, particularly when participants are elderly or in cognitive decline.[45]

FIGURE 8: KEY PROCEDURAL AND SUBSTANTIVE REGULATORY INSTRUMENTS

PROCEDURAL	REGULATORY INSTRUMENTS
Clinical ▪ NPAAC/NATA standards and accreditation requirements ▪ Laboratory policies, procedures Research ▪ *National Statement* guidance	Tort law ▪ Duty of care Privacy law ▪ Right of access

7.6 Tort Law

Although the conversation in the academic literature relating to genomic analysis and return of findings often focuses on the extent of any normative ethical obligation to do so, equally relevant is the question of whether there is some form of legal duty.[46] Whether or not patients or participants might be able to bring a successful action in negligence in such an instance is an open question. This question has been addressed to some extent by scholars in overseas jurisdictions,[47] but less so in Australia. There is no case law dealing with the issue of negligence for non-disclosure of findings. However, the law of negligence itself is surprisingly consistent across international jurisdictions. For an action in negligence to be successful a plaintiff (a party bringing a civil action) will need to prove that (i) they are owed a duty of care by another party (the defendant); (ii) that this duty of care was breached by the defendant; and (iii) that any resulting damage suffered by the plaintiff is linked in a causal sense to the breach.

45 Ibid; see also Jane Tiller, Alison H Trainer, Ian Campbell and Paul A Lacaze, 'Ethical and practical implications of returning genetic research results: two Australian case studies' (2021) 214(6) Medical Journal of Australia, doi: 10.5694/mja2.50842.

46 Note that this chapter does not consider liability arising through breach of contractual obligations that might be present due to agreements between researchers and research subjects.

47 Some of the more prominent academic commentaries include: Susan M Wolf, 'The Role of Law in the Debate over Return of Research Results and Incidental Findings: The Challenge of Developing Law for Translational Science' (2012) 13 *Minnesota Journal of Law Science and Technology* 435; Clayton and McGuire (n 8); Elizabeth R Pike, Karen H Rothenberg and Benjamin E Berkman, 'Finding Fault? Exploring Legal Duties to Return Incidental Findings in Genomic Research' (2014) 102 *Georgetown Law Journal* 795; McGuire et al (n 8).

The question is whether tort law imposes a common law duty of care in respect of return of relevant findings from genomic analyses to individuals, or in respect of negligent disclosure, which may lead to false positives.[48] This is the first hurdle when bringing an action in negligence. In Australia, as in other jurisdictions, clinicians owe their patients a duty of care under the tort of negligence: this duty is in the nature of a fiduciary duty.[49] The situation in relation to researchers differs in that the relationship does not involve the same, high level of trust. In the US, it has been clearly articulated that this relationship is not a fiduciary relationship.[50] In contrast, there is no clear judicial statement in Australia on this point and therefore this question remains an open one. Leaving this aside, there would appear to be no reason in principle why a duty of care would not be found to exist in the right circumstances, and this has certainly been the case in the US where a number of lawsuits by research participants have been brought.[51] For the most part these lawsuits have settled, with the result that we have little resulting case law to guide future courts. The position is murkier where a clinician has a dual clinical/research role, and enrols patients in studies in which he or she is involved. In this case the duty owed in each context would be distinguished. It can be stated with some degree of certainty (although there is no direct authority on point) that a duty of care might be owed by any party involved in the management of genomic information, not just the party who originally provides a request for genomic testing/sequencing to be undertaken.

The scope of any duty of care owed by a clinician or researcher also requires defining. The duty of care owed by a clinician extends to a duty to act in the patient's best interests in treatment, diagnosis and advice. The notion of advice referred to in respect of a clinician's duty of care generally involves a clinician advising a patient of risks that may deter them from undergoing treatment. But a number of US cases have demonstrated that a physician's duty will ordinarily extend to disclosing unexpected findings that may lead to future harm,[52] and could conceivably extend to that patient's relatives.[53] In a recent UK case, Justice Yip of the UK High Court was prepared to find that a hospital owed a patient's relative a duty of care to advise her that their patient (her father) had tested positive to Huntington's Disease.[54] Her father had expressly asked that his diagnosis not be disclosed to his daughter. It is worth noting that the plaintiff was in a relationship with the hospital as a participant in family therapy that also involved her father. This gave rise to a close proximal relationship which taken with relevant policy considerations, gave rise to a duty of care to balance the claimant's interest in being informed of her genetic risk, against her father's interest (and that of the public) in preserving confidentiality. This duty of care was not, however, breached, on the grounds that the balance did not sit in the claimant's favour. Legal development may well take a similar path in Australia, although under the High Court of Australia's formulation for establishing a duty of care in novel factual scenarios, there is significant scope in cases involving medical practice, for normative considerations to preclude the imposition of such a duty.[55]

48 Pike, Rothenberg and Berkman (n 47) 813; McGuire et al (n 8) 720-2.

49 *Breen v Williams* (1996) 186 CLR 71, 83; 108 (per Gaudron and McHugh JJ).

50 Clayton and McGuire (n 8).

51 See, eg, *Grimes v Kennedy Krieger Inst Inc* 782 A.2d 807, 858; L Jansson, 'Researcher Liability for Negligence in Human Subject Research: Informed Consent and Researcher Malpractice Actions' (2003) 78 *Washington Law Review* 229.

52 See, eg *Hahn v Mirda*, 54 Cal Rptr 3d 527 (Cr App 2007). For comprehensive discussion of the case law see Thomas L Hafemeister and Selina Spinos, 'Lean on Me: A Physician's Fiduciary Duty to Disclose an Emergent Medical Risk to the Patient' [2009] 86 *Washington University Law Review* 1167.

53 See, eg, *Pate v Threlkel*, 661 So2d 278, 281-82 (Fla 1995); *Safer v Estate of Pack*, 677 A2d 1188, 1192 (NJ Super 1996). In these cases, doctors were held liable for failing to warn of genetic conditions that could be passed on to children. See also *Schroeder v Perkel* 432 A2d 834 (NJ 1981), where a doctor was held liable to a child's parents for failing to inform them she had cystic fibrosis. For further discussion of these cases see Richard L Furman Jr, 'Genetic Test Results and the Duty to Disclose: Can Medical Researchers Control Liability?' [1999] 23 *Seattle University Law Review* 391, 402-4.

54 *ABC v St Georges Healthcare NHS Trust* [2020] EWHC 455 (QB).

55 See, eg, the 'wrongful life' cases such as *Harriton v Stephens* (2006) 226 CLR 52; 226 ALR 391.

As for researchers, the difficulty in finding a duty in such cases is that researchers have a '... primary obligation to produce generalisable knowledge, even if it requires actions that are not primarily for the benefit of the individual research subject.'[56] Research generally does not aim to better the individual health of the research participant. An analogy has been drawn with 'third-party physicians' who are not primary clinicians but still play some part in a patient's care, such as physicians retained by employers and insurers to conduct health assessments.[57] There is also a series of US cases dealing with radiologic imaging which may be analogous. US courts have been willing to find a duty of care exists in both of these scenarios, where there is some prospect of clinical actionability despite a lack of fiduciary duty.[58]

If a duty of care can be established, an injured party is then required to prove that the duty of care has been breached. The process for determining breach of duty is to consider whether the identified risk is reasonably foreseeable, and then whether the defendant's conduct is consistent with the relevant standard of care. A risk will be foreseeable where it is 'not far-fetched or fanciful'.[59] In reality this is a low threshold, and it may well be foreseeable that a research subject may be at risk of harm if findings that may impact on their future health are not conveyed to them.

For professionals, the relevant standard of care is statutorily prescribed in each Australian jurisdiction.[60] This standard will apply in determining the standard in any cause of action brought by a patient against a clinician. The defendant's conduct will be measured against peer professional opinion to determine whether it aligns with the conduct of peers, or practice that is 'widely accepted in Australia by peer professional opinion as competent professional practice.' The relevant provisions go on to state that peer professional opinion will not be accepted if the court considers it is irrational, but that peer professional opinion can be viewed as 'widely accepted', despite being one of a number of differing opinions, or not universally accepted. Essentially, the question of what constitutes widely accepted peer professional opinion will depend on expert evidence. Guidelines such as those issued under s 95AA of the *Privacy Act 1988* (Cth) would likely be adduced as evidence of the relevant standard (see Chapter 3). The issue will be one of fact and degree, and will depend on the clinical specialty of the practitioner involved, the import attaching to the findings in question, and the perceived importance to a patient (and perhaps their family) of returning those findings.

It is likely that some degree of certainty would need to attach to the findings for a court to conclude that standard practice would demand their communication to patients: where a VUS has been discovered, it is difficult to conceive that a majority of clinicians would currently consider its communication necessary. In reality, it is rare for a finding that a body of opinion is 'irrational'.

Whether researchers are subject to this special standard of care that applies to professionals is unclear. 'Profession' is not defined under the legislation. There is no case law on whether medical researchers are engaged in 'practising a profession' pursuant to the civil liability legislation. *Zhang v Hardas (No 2)*[61] is a NSW case which dealt with whether chiropractors can be said to be practising a profession. In this case, the Justice of Appeal (Leeming JA) found that they are, given that chiropractors are accredited and regulated under specific legislation.[62] This legislation operates in a similar fashion to legislation regulating the practice of medicine.[63] He characterised the practice of chiropractic as a 'licensed monopoly', which admits a limited

56 McGuire et al (n 8) 720-1.

57 Pike, Rothenberg and Berkman (n 47) 818-23.

58 Ellen Wright Clayton et al, 'Managing Incidental Genomic Findings: Legal Obligations of Clinicians' (2013) 15(8) *Genetics in Medicine* 624.

59 *Wyong Shire Council v Shirt* (1980) 146 CLR 40; *New South Wales v Fahy* (2007) 232 CLR 486.

60 *Civil Liability Act 2002* (NSW), s 50; *Wrongs Act 1958* (Vic), s 59; *Civil Liability Act 2003* (Qld), s 22(1); *Civil Liability Act 1936* (SA), s 41; *Civil Liability Act 2002* (Tas), s 22; *Civil Liability Act 2002* (WA), s 5PB (which applies only to health care professionals).

61 *Zhang v Hardas* (No 2) [2018] NSWSC 432. 57.

62 Ibid [145]-[170].

63 Ibid [169]-[170].

number of members based on educational qualifications.[64] Applying these principles, it seems that legislative and ethical requirements govern entry into and the practice of medical research, hence it may constitute a 'licensed monopoly'. The answer to this question is far from clear. Notably however, what constitutes a profession may change over time.[65]

Even if this special standard of care does not apply to medical researchers, the more fundamental approach for determining the standard of care will apply. The standard of care a reasonable person in the defendant's position would have adhered to will be determined by considering relevant factors, including the likelihood of the risk of injury and the seriousness of any injury, the burden of taking precautions to avoid the risk of injury and any social benefit inherent in the defendant's risk-taking conduct.[66] Other factors might come into play, including, importantly, custom and professional practice in the relevant industry or area in which the conduct takes place,[67] and the state of knowledge at the time the cause of action arose.[68]

Questions that arise here relate to the adequacy of study and validation protocols; what constitutes standard practice in relation to identifying variants; and the practicality of identifying incidental findings given the state of science at any given time and the limited purpose for which many research studies are conducted.[69] For example, genomics researchers have a duty to thoroughly investigate and produce replicable results: would they breach this duty if they failed to investigate individual-level VUS they discover incidentally? And what role do ethical obligations to return findings play? As noted earlier in this chapter, the *National Statement* provides guidance but no mandatory directives on return of findings, which would inform the issue of what is standard practice. If the practice of returning incidental findings becomes more entrenched and consistent, or more in the nature of standard practice, researchers who fail to do so may be found to have fallen short of the expected standard.[70]

The final element in establishing a cause of action in negligence involves considering whether any damage suffered by the plaintiff was causally linked to the breach of duty.[71] These requirements will be difficult to make out. A *predisposition* to disease caused by a genetic variant that has not yet eventuated will *not* equate to damage. Any degenerative symptoms a plaintiff suffers by developing that condition will have been caused by the genetic variant, rather than the nondisclosure by the clinician/researcher. The failure to communicate findings is unlikely to be a necessary element of the plaintiff's damage and the requisite statutory provisions are unlikely to be satisfied.

A team of US commentators has raised the possibility of application of the 'loss of chance' doctrine, which allows a plaintiff to argue that the defendant's negligence deprived them of the chance of a better outcome.[72] If not told of an underlying predisposition to develop a genetic disorder, a plaintiff might argue they were denied the opportunity to make lifestyle choices that could delay or prevent its onset. There is some scope under US law to apply this doctrine. The High Court of Australia has rejected the application of the 'loss of chance' doctrine in Australia because it requires a court to consider whether the chance of avoiding the damage which actually occurred, was lost due to the defendant's negligence.

64 Ibid [170].

65 Ibid [142], [145].

66 *Civil Law (Wrongs) Act 2002* (ACT) s 43(2); *Civil Liability Act 2002* (NSW) s 5B(2); *Civil Liability Act 1936* (SA) s 32(2); *Civil Liability Act 2002* (Tas) s 11(2); *Wrongs Act 1958* (Vic) s 48(2); *Civil Liability Act 2002* (WA) s 5B(2).

67 *Woods v Multi-Sport Holdings Pty Ltd* (2002) 208 CLR 460.

68 *H v Royal Alexandra Hospital for Children* (1990) Aust Torts Reports 81-000.

69 See also, McGuire et al (n 8) 722.

70 Pike, Rothenberg and Berkman (n 47) 821-2.

71 *Civil Law (Wrongs) Act 2002* (ACT) s 45(1); *Civil Liability Act 2002* (NSW) s 5D(1); *Civil Liability Act 2003* (Qld) s 11(1); *Civil Liability Act 1936* (SA) s 34(1); *Civil Liability Act 2002* (Tas) s 13(1); *Wrongs Act 1958* (Vic) s 51(1); *Civil Liability Act 2002* (WA) 5C(1).

72 Pike, Rothenberg and Berkman (n 47) 825-7.

In doing so, it lowers the standard of proof for damage from the required standard (on the balance of *probabilities*) to a lower standard: it effectively means a court must ask whether there was a *possibility* that such damage as did occur might not have eventuated had the plaintiff not been denied the chance.[73] The High Court refused to take this step.

The final aspect of the element is that the damage suffered by the plaintiff must be within the scope of the defendant's liability.[74] There are many factors that may take the plaintiff's damage outside the scope of what a defendant should be liable for. A plaintiff's damage might far outweigh a defendant's blameworthiness. One aspect of this is that the damage itself must be a foreseeable consequence of the defendant's breach. In some cases, this might be clear-cut; an incidental finding might predict a specific harm that a defendant ought to have foreseen. In other cases, particularly where a VUS is not communicated to a plaintiff, it will be far more difficult to establish that the harm was foreseeable, where the future significance of the variant is not known at the time of the breach.[75] The requirements in establishing damage under Australian law are complex and their applicability in this scenario warrants more detailed attention.

In sum, there are real questions about whether non-disclosure of findings in both clinical and research settings would give rise to fault-based liability, at least on the basis of current Australian law. In the absence of concrete legal rules, it is unlikely at present that tort law represents a valid option for those alleging they have not been notified of relevant findings, to seek compensation. A key issue worthy of consideration here would be what form of disclosure would likely be sufficient to discharge any obligation to disclose under the requirements of negligence.

73 *Tabet v Gett* [2010] HCA 12. See, in particular, Justice Kiefel's judgment at [152].

74 *Civil Law (Wrongs) Act 2002* (ACT) s 45(1); *Civil Liability Act 2002* (NSW) s 5D(1); *Civil Liability Act 2003* (Qld) s 11(1); *Civil Liability Act 1936* (SA) s 34(1); *Civil Liability Act 2002* (Tas) s 13(1); *Wrongs Act 1958* (Vic) s 51(1); *Civil Liability Act 2002* (WA) 5C(1).

75 Pike, Rothenberg and Berkman (n 47) 827-8.

8 Genetic Tests and Genetic Testing

8.1 Summary

The most significant legal and regulatory requirements pertaining to the provision of genetic tests and genetic testing relate to pre-market assessment of the tests themselves, accreditation of the testing laboratories and compliance with national and international genetic testing standards.

Genetic tests for therapeutic purposes are classified as in vitro diagnostic medical devices ('IVD medical devices') and are regulated under the medical devices regulatory scheme included in the *Therapeutic Goods Act 1989* (Cth), administered by the Therapeutic Goods Administration ('TGA'). In addition, the National Association of Testing Authorities ('NATA') administers the scheme for accreditation of testing laboratories. The National Pathology Accreditation Advisory Council ('NPAAC') standards and international standards provide best practice guidance on the provision of genetic testing. Collectively, these instruments and agencies provides a comprehensive regulatory regime to ensure genetic tests and genetic testing are safe and clinically effective.

The general supply of genetic tests directly to consumers in Australia is prohibited through the IVD medical devices regulatory regime, on the basis it falls within a prohibition on 'self-testing IVDs'. This prohibition has recently been affirmed in a review by the TGA, as reflected in the *Therapeutic Goods (Medical Devices – Excluded Purposes) Specification 2020* (Cth).[1] However, consumers can directly order and import a genetic test for personal use from an overseas providers under an exemption for personal importation and use. Genetic tests provided directly to consumers for ancestry and paternity purposes are also outside the remit of the TGA IVD medical devices regime, as these tests are not claiming therapeutic benefits. Genetic tests carried out for therapeutic purposes on Australian samples by laboratories located in other countries are also beyond the jurisdiction of Australian regulatory agencies.

Additional regulatory regimes that intersect with genetic tests and genetic testing include: privacy, product liability, advertising and consumer protection, and remuneration.

8.2 Introduction

The terms 'genetic tests' and 'genetic testing', as used in this chapter, are intended to embrace both genetic and genomic analyses undertaken for 'health-related' or '*therapeutic purposes*'. As such, this chapter is not intended to cover research-related genetic and genomic analyses, unless the findings of those analyses need to be clinically validated (see chapter 7). It is only at this point that the regulatory regime for genetic tests and genetic testing becomes relevant. This chapter also considers the extent to which the genetic reports provided directly to consumers ('DTC') by the direct-to-consumer genetic testing ('DTC-GT') industry fall within this regulatory regime. This chapter is restricted in its consideration to human-based genetic tests and human genetic testing. As such, it is beyond the scope of this chapter to provide a detailed description of the regulatory environment relating to genetic tests and genetic testing for pathogens.

1 Note that this Specification was amended on 1 October 2021 to allow for the supply of COVID-19 rapid antigen self-tests in Australia. See: *Therapeutic Goods (Medical Devices—Excluded Purposes) Amendment (COVID-19 Rapid Antigen IVD Medical Devices for Self-Testing) Specification 2021.*

8.3 The Australian Genetic Testing Landscape

The Royal College of Pathologists of Australia ('RCPA') has undertaken a series of three comprehensive studies examining the genetic testing landscape in Australia.[2] The most recent study, reported in May 2019, involved a survey of providers offering genetic and genomic tests with medical utility during the 2016-2017 financial year.

The RCPA provides a helpful definition of the distinction between genetic and genomic testing:

> **Genetic [tests]:** Genetic testing seeks to identify changes in chromosomes, genes, or proteins. Identified genetic changes (variants) may confirm the diagnosis of a suspected disorder, predict the likelihood of developing or passing on a genetic condition, or predict response to medication.
>
> Several methods can be used for genetic testing:
>
> - **Gene tests (or molecular tests):** study short lengths of DNA, a single gene, or multiple genes to identify variations or mutations that lead to a genetic disorder.
> - **Chromosomal tests:** analyse whole chromosomes or long lengths of DNA to assess for large genomic changes, such as an extra copy of a chromosome, causative of a genetic condition.
> - **Biochemical tests:** study the amount or activity level of proteins; abnormalities in either can indicate changes in genes that result in a genetic disorder.
> - **Genomic tests:** The study of multiple genes, which may include analysis of all genes across the human genome, involves the use of massively parallel sequencing technology and microarray technology.[3]

For the purpose of this chapter, no further distinction is made between genetic and genomic analyses. Rather, the generic terms 'genetic tests' and 'genetic testing' are used to embrace both forms of tests/testing.

A total of 87 laboratories were identified and invited to participate in the latest RCPA survey.[4] The overwhelming majority of laboratories (90 per cent of those answering accreditation questions) were accredited by NATA.[5] A key finding from the survey, when compared with the last survey, undertaken in 2011, was a substantial increase in the number of accredited providers (from 39 in 2011 to 72 in 2016-7).[6]

Although only 30 per cent of survey participants were in the private sector, they delivered more than 50 per cent of the almost 1.2 million outcomes that were reported. The range of tests on offer was also shown to be increasing, and molecular testing was becoming more common than cytogenic (or chromosomal) testing. Transfers to interstate and overseas laboratories were also becoming more common, which is an important finding given that genetic tests performed by overseas laboratories are not within the jurisdictional oversight of the TGA. It is beyond the scope of this report to examine the regulatory requirements for genetic tests performed in other jurisdictions. Medicare funding applied to around 49 per cent of all tests, compared with 35 per cent in 2011. Most test referrals were ordered by general practitioners or obstetricians, or for cancer related reasons.[7]

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2 Royal College of Pathologists of Australasia, *Australian Health Genetics/Genomics Survey 2017 Report of Key Findings to: Department of Health* (Final Report, May 2019) <https://www.rcpa.edu.au/Library/Practising-Pathology/RCPA-Genetic-Testing/Docs/RCPA-Genetic-Testing-Survey-Report.aspx> ('*2017 Health Genomics Survey Report*'); Royal College of Pathologists of Australasia, *Report of the RCPA Genetic Testing Survey 2011* (2012); Royal College of Pathologists of Australasia, *Report of the RCPA Genetic Testing Survey 2006* (2009).

3 2017 *Health Genomics Survey Report* (n 2) 8.

4 Ibid 4.

5 Ibid 5.

6 Ibid.

7 Ibid.

In addition to these health professional-mediated tests, consumers have the option of ordering genetic reports online from companies located around the world, without a requirement for any direct involvement from the healthcare profession. The rise of this so-called DTC-GT service has been accompanied by a significant body of literature voicing concerns about the potential weaknesses it exposes in regulatory frameworks in Australia and other countries.[8]

The Australian Law Reform Commission and Australian Health Ethics Committee concluded in *Essentially Yours* that there are 'strong arguments for regulating the supply, directly to the public, of products used in some forms of genetic testing'.[9] *Essentially Yours* also canvassed the difficulties associated with regulating foreign companies offering DTC genetic testing via the Internet, acknowledging that 'ensuring accreditation of all genetic testing performed in Australia will not necessarily overcome concerns about access by individuals to non-accredited genetic testing conducted overseas'.[10]

According to *Essentially Yours*, there were no Australian companies offering DTC-GT in 2003.[11] In a study by Nicol et al published in 2014, at least 15 companies were identified as having offices in Australia and making DTC- GT available directly to the Australian public.[12] Since that time, it appears that four of those companies are no longer active, and two have merged. The ten remaining entities offer a mixture of ancestry, parentage, relationship, immigration and other forms of non-health related testing. Six also offer health and wellbeing related testing, and of those, three claim to offer some form of clinical testing. One of these companies states that its clinically-related interactions with consumers are health practitioner-mediated. All but two companies state that they have some form of accreditation, based either on international or national requirements, although for most of them, this form of accreditation is likely to be more directly related to parentage services than for health purposes.[13]

Aside from these Australian-based services, consumers are also freely able to engage with overseas providers, and many do so.[14] As noted in the Scoping Report, the availability of DTC-GT, whether offered by overseas or Australian providers, is of concern both in the literature and for stakeholders. Among others, Matthews, Hall and Carter call for a regulatory framework that better protects consumers.[15]

Two main aspects of the legal and regulatory environment for genetic tests and genetic testing in Australia are considered in detail below: the IVD medical devices regime administered by the TGA; and accreditation and standards for genetic testing through NATA, NPAAC and International Standards Organisation ('ISO') standards. The interaction of genetic testing with privacy law and consumer law and the role of remuneration in the regulation of tests and testing are also briefly described.

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8 See, eg, Jan Charbonneau et al, 'Public Reactions to Direct-to- Consumer Genetic Health Tests: A Comparison across the US, UK, Japan and Australia' (2020) 28 *European Journal of Human Genetics* 339 <https://doi.org/10.1038/s41431-019-0529-8>; Rebecca Mathews, Wayne Hall and Adrian Carter, 'Direct-to-Consumer Genetic Testing for Addiction Susceptibility: A Premature Commercialisation of Doubtful Validity and Value' (2012) 107(12) Addiction 2069 <https://doi.org/10.1111/j.1360-0443.2012.03836. x>; Amy L McGuire and Wylie Burke, 'Health System Implications of Direct-to-Consumer Personal Genome Testing' (2011) 14(1) *Public Health Genomics* 53 <https://doi.org/10.1159/000321962>; Dianne Nicol et al, 'Time To Get Serious about Privacy Policies: The Special Case of Genetic Privacy' (2014) 42 *Federal Law Review* 149; Jacqueline Savard et al, 'From Expectations to Experiences: Consumer Autonomy and Choice in Personal Genomic Testing' (2020) 11(1) *AJOB Empirical Bioethics* 63 <https://doi.org/10.1080/23294515.2019.1701583>.

9 Australian Law Reform Commission and Australian Health Ethics Committee, *Essentially Yours: The Protection of Human Genetic Information in Australia* (Report No 96, May 2003) 355 <http://www.alrc.gov.au/publications/report-96> ('*Essentially Yours*').

10 Ibid 356.

11 Ibid 347.

12 Dianne Nicol, Meredith Hagger, Nola Ries and John Liddicoat, 'Time To Get Serious about Privacy Policies: the Special Case of Genetic Privacy' (2014) 42 *Federal Law Review* 149.

13 For the list of companies see Ibid, Table 1, 167-169.

14 Sylvia A Metcalfe et al, 'Australians' Views on Personal Genomic Testing: Focus Group Findings from the Genioz Study' (2018) 26 *European Journal of Human Genetics* 1101.

15 Matthews, Hall and Carter (n 8).

8.4 Regulation of Therapeutic Goods

In Australia, the TGA has responsibility for oversight of the safety and efficacy of therapeutic goods. The TGA regulatory framework is based on a risk management approach designed to ensure quality and safety by 'reducing the impact of risk to a manageable level'.[16] Until 2010, the TGA regulatory framework did not specifically apply to genetic tests. However, in 2010, a major addition brought these tests squarely within its ambit. One of the drivers for this change was to ensure international alignment in regulatory approaches, as endorsed by the Global Harmonization Task Force for IVDs.[17] This complex regulatory environment for genetic and genomic tests is briefly summarised in this section, which starts with an overview of the broader regulatory environment for therapeutic goods.

8.4.1 THE AUSTRALIAN REGISTER OF THERAPEUTIC GOODS

Any product for which therapeutic claims are made must be entered in the Australian Register of Therapeutic Goods ('ARTG') before the product can be supplied in Australia, unless it is exempt from registration requirements. The ARTG is a database of therapeutic goods for human use approved for supply in, or exported from, Australia. The *Therapeutic Goods Act 1989, Therapeutic Goods Regulations 1990* (Cth), Orders and Specifications set out the requirements for inclusion of therapeutic goods in the ARTG, including advertising, labelling, product appearance and appeal guidelines. Some provisions, such as the scheduling of substances and the safe storage of therapeutic goods, are covered by relevant state or territory legislation.

'Therapeutic goods' are defined in the *Therapeutic Goods Act 1989* at s 3 as, inter alia:

> goods
>
> (a) that are represented in any way to be, or that are, whether because of the way in which the goods are presented or for any other reason, likely to be taken to be:
>
> (i) for therapeutic use ...

'Therapeutic use' is defined in s 3 as use in or in connection with:

> (a) preventing, diagnosing, curing or alleviating a disease, ailment, defect or injury;
>
> (b) influencing inhibiting or modifying a physiological process;
>
> (c) testing the susceptibility of persons to a disease or ailment;
>
> (d) influencing, controlling or preventing conception;
>
> (e) testing for pregnancy; or
>
> (f) replacement or modification of parts of the anatomy.

A key aspect of the regulation of therapeutic goods is pre-market assessment. Products assessed as having a high level of risk (prescription medicines, some over the counter medicines and medical devices) are evaluated for quality, safety and efficacy prior to inclusion on the ARTG and approval for marketing. Products assessed as being lower risk (many over the counter medicines, most complementary medicines and low risk medical devices) are assessed for quality and safety only. All medicines must be listed or registered on the ARTG unless exempt.

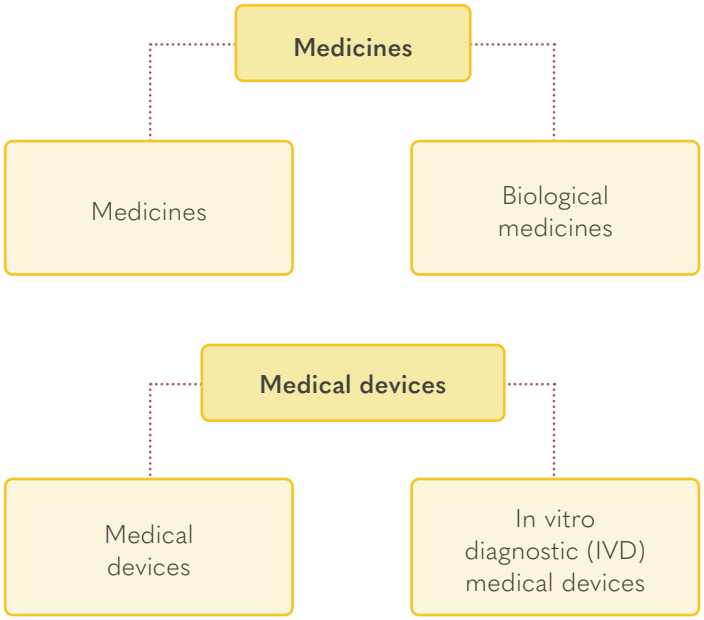
16 Therapeutic Goods Administration, *The TGA Regulatory Framework* (May 2012) 6.

17 Therapeutic Goods Administration, *Overview of the New Regulatory Framework for In Vitro Diagnostic Medical Devices (IVDs)* (July 2011), 5 ('*TGA New Regulatory Framework*'). It should be noted that the Global Harmonization Task Force has since been replaced by the International Medical Device Regulators Forum: <http://www.imdrf.org/index.asp>.

A major amendment to the Therapeutic Goods Act 1989 relating to the regulation of medical devices was made in 2002, through the *Therapeutic Goods Amendment (Medical Devices) Act 2002* (Cth). Prior to this amendment, the regulation of medical devices was much less prescriptive than for medicines. For the first time, the *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth) provided detailed procedural requirements for this new regulatory regime for medical devices.

There are four classes of devices, ranging from class I (for such devices as surgical retractors and tongue depressors) through to higher classes (for heart valves, hip replacements, defibrillators and the like). A conformity assessment procedure ensures that devices conform to requirements for that class. Higher classes require a higher level of conformity.

FIGURE 9: THERAPEUTIC GOODS ACT 1989 REGULATORY SCHEME



Notes:

- *Administered by the Therapeutic Goods Administration*
- *All goods for therapeutic use in Australia must be included on the Australian Register of Therapeutic Goods (ARTG) unless exempt*
- *Requires pre-market assessment commensurate with level of risk (based on classification of the device)*

Of particular relevance in the context of this chapter, medical devices are defined in s 41BD of the *Therapeutic Goods Act 1989* as *inter alia*,

> (1) (a) any instrument, apparatus, appliance, software, implant, reagent, material or other article (whether used alone or in combination, and including the software necessary for its proper application) intended, by the person under whose name it is or is to be supplied, to be used for human beings for the purpose of one or more of the following:
>
> …
>
> (v) in vitro examination of a specimen derived from the human body for a specific medical purpose; and that does not achieve its principal intended action in or on the human body by pharmacological, immunological or metabolic means,

The regulation of medical devices has recently undergone further reform. In particular, amendments to the *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth) were introduced in December 2019 through the *Therapeutic Goods Legislation Amendment (2019 Measures No.1) Regulations 2019* (Cth). These amendments included making specific provision for software, personalised medical devices and IVD companion diagnostics.[18]

8.4.2 THERAPEUTIC GOODS LEGISLATION AND IN VITRO DIAGNOSTIC MEDICAL DEVICES

In 2010 a new regime for regulating IVD medical devices in Australia was established through amendments to the *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth) by the *Therapeutic Goods (Medical Devices) Amendment Regulations 2010 (No 1)* (Cth). As noted by the TGA, IVD medical devices are 'in general, pathology tests and related instrumentation used to carry out testing on human samples, where the results are intended to assist in clinical diagnosis or in making decisions.'[19] The TGA further notes that, prior to the creation of this new regime, regulatory scrutiny of IVD medical devices was very limited, with most being excluded from pre-market assessment.[20] The two notable exceptions were IVDs for human immunodeficiency virus (HIV) and hepatitis C virus, which were subject to extensive pre-market review.[21]

The new framework was designed to ensure all IVD medical devices supplied in Australia, with a few limited exceptions and exclusions, are subject to regulation under the *Therapeutic Goods Act 1989*.

IVD medical devices are defined broadly in the Dictionary to the *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth) to include any medical device intended to be used in vitro to examine a 'specimen derived from the human body' for therapeutic purposes.

IVD medical devicess are separated into two categories: 'IVD medical devices' and 'in-house IVD medical devices'. For the purpose of this chapter, 'IVD medical devices for self-testing' are considered separately, although the TGA does not consider them to be a distinct category. In-house IVD medical devices are limited to those IVD medical devices developed or modified by a laboratory for use specifically within the laboratory (or laboratory network), with no intention to supply them outside of the laboratory (or laboratory network). Commercially supplied IVD medical devices must be included on the ARTG. In contrast, in-house IVD medical devices that fall within classes 1 to 3 are exempt from inclusion in the ARTG, subject to conditions. However, inclusion in the ARTG remains a requirement for class 4 in-house IVD medical devices.

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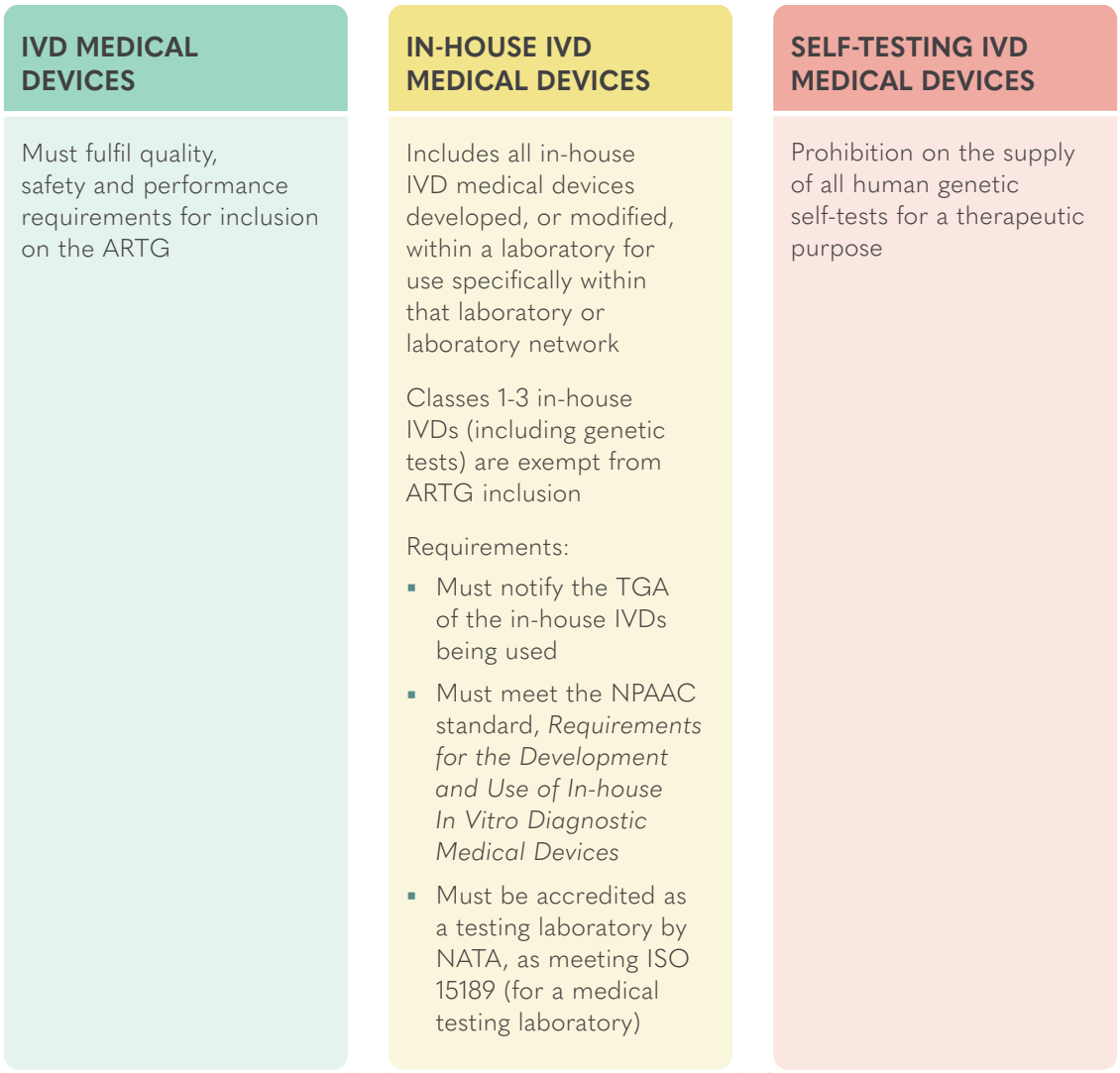
18 *Therapeutic Goods Legislation Amendment (2019 Measures No 1) Regulations 2019* (Cth); 'Therapeutic Goods Legislation Amendment (2019 Measures No 1) Regulations 2019', *Therapeutic Goods Administration* (Web Page, 10 January 2020) <https://www.tga.gov.au/therapeutic-goods-legislation-amendment-2019-measures-no1-regulations-2019>.

19 *TGA New Regulatory Framework* (n 16) 5.

20 *Ibid.*

21 *Ibid.*

FIGURE 10: IVD MEDICAL DEVICES REGULATORY SCHEME



Laboratories performing genetic tests (which include whole genome sequencing and whole exome sequencing analyses) may include use of commercial IVD medical devices included on the ARTG or the laboratory may use their own in-house IVD medical devices. All genetic tests for a therapeutic purpose (commercial or in-house IVD medical devices) must involve the return of results via a health professional who has formal training in a medical field to which the testing relates.

Schedule 3 of the Regulations provides detailed information on the requirements imposed on Class 1-3 in-house IVD medical devices, such as the tests used for health-related genetic testing. In particular, laboratories are required to: notify the TGA of the in-house IVDs it is using; meet the NPAAC standard, *Requirements for the Development and Use of In-house In Vitro Diagnostic Medical Devices*; and be accredited by NATA to ISO 15189 (for a medical testing laboratory). The laboratory must establish a post-market system and report any adverse events to the TGA.

Because DTC-GT are provided directly to consumers, rather than being mediated by a health professional, they cannot be considered as in-house IVD medical devices. Instead, they would be defined as self-testing IVD medical devices, being those intended to be used:

(g) in the home or similar environment by a lay person; or

(h) in the collection of a sample by a lay person and, if that sample is tested by another person, the results are returned directly to the person from whom the sample was taken without the direct supervision of a health professional who has formal training in a medical field or discipline to which the self-testing relates.[22]

This categorisation of DTC-GT IVD medical devices as self-testing IVD medical devices brings them under a different and more proscriptive category of regulation when compared with in-house IVD medical devices.

8.4.3 PROHIBITION ON SELF-TESTING IN VITRO DEVICES

In parallel with the introduction of the IVD medical devices regime, the TGA also recognised that there was a need to respond to the emergence of an unregulated environment for home use of IVD medical devices, including to test for pathogens, to diagnose notifiable infectious diseases, to determine genetic traits and to test for serious disorders including cancers and myocardial infarctions.[23] As a result, the *Therapeutic Goods (Medical Devices) Amendment Regulations 2010 (No 1)* (Cth) were accompanied by the *Therapeutic Goods (Excluded Purposes) Specification 2010* (Cth) which created a new prohibition on certain self-testing IVD medical devices. The *Therapeutic Goods (Excluded Purposes) Specification 2010* (which sunsetted on 1 October 2020) prohibited the supply of self-testing IVD medical devices when used solely for the purpose of: testing for pathogens or transmissible agents; genetic testing for the presence of or susceptibility to serious diseases; diagnosing or assisting in diagnosing serious disorders; and testing for markers for serious disorders. Section 8.4.5, below, provides detail on the *Therapeutic Goods (Medical Devices - Excluded Purpose) Specification 2020* (Cth) which has replaced the 2010 Specification.

8.4.4 PROHIBITION ON IMPORT AND EXPORT OF SELF-TESTING IVDS

Under s 41MI of the *Therapeutic Goods Act 1989,* it is a criminal offence to import into, supply in, or export from Australia an IVD medical device not included in the ARTG (provided none of the exemptions under the *Therapeutic Goods Act 1989* apply). This provision may make consumers of foreign DTC-GT services liable for import and export offences on the basis they are directly involved in the receipt and dispatch of specimen collection kits sourced from outside Australia.

However, under the *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth), a medical device imported into Australia is an 'exempt device' where it is 'for use in the in vitro examination of a specimen obtained from the importer or a member of the importer's immediate family', and the other specified conditions are satisfied. Similarly, the Regulations state that a medical device exported from Australia is exempt provided, among other reasons, 'it is not intended for commercial supply' or 'for use for experimental purposes on humans.' These provisions would seem to protect Australian consumers of DTC-GT originating overseas from criminal liability. In France, by contrast, consumers face criminal liability for requesting genetic tests 'outside the conditions laid by the law'.[24]

22 *Therapeutic Goods (Medical Devices) Regulations 2002* (Cth) dictionary.

23 *Therapeutic Goods Administration, Overview of the New Regulatory Framework for In Vitro Diagnostic Medical Devices* (2010) 8.

24 Pascal Borry et al, 'Legislation on Direct-to-Consumer Genetic Testing in Seven European Countries' (2012) 20(7) *European Journal of Human Genetics* 715, 717. See also, L Kalokairinou et al, 'Legislation of Direct-to-Consumer Genetic Testing in Europe: A Fragmented Regulatory Landscape' (2018) 9(2) *Journal of Community Genetics* 117.

The issue of how to regulate foreign providers of DTC-GT remains unresolved. One option canvassed in *Essentially Yours* was the enactment of Federal legislation, similar to that in place for offensive material and interactive gambling, to regulate advertising of DTC-GT on the Internet. However, it was ultimately concluded it would be premature to implement a similar regime at that stage.[25] Whether or not the situation has changed in the 17 years since *Essentially Yours* was completed remains to be determined. Scholars such as Burdon suggest that the DTC-GT industry is burgeoning,[26] although the study by Charbonneau and colleagues indicates that Australian consumers display a lower level of intention to purchase such tests than consumers in the United States.[27]

FIGURE 11: EXCLUSIONS FROM THE IVD MEDICAL DEVICES REGULATORY SCHEME

Research-related genetic/ genomic analysis

Query: *when does research related analysis become clinically relevant, triggering IVD medical devices regulation? (see return of results chapter)*

DTC genetic/genomic analysis for paternity

Note: *the Family Law Act has relevant regulatory requirements*

DTC genetic/genomic analysis for ancestry

DTC genetic/genomic analysis for other non-therapeutic purposes

Query: *when is analysis for curiosity/ therapy/enhancement?*

Laboratory-based genetic/genomic analysis undertaken outside Australia on Australian samples

Query: *what is the adequacy of the regulatory requirements in the country in which the genetic/genomic analysis is undertaken?*

Note: *protection of privacy, consent and other requirements will apply to the tissue sample collected in Australia*

DTC genetic/genomic analysis undertaken outside Australia

25 *Essentially Yours* (n 8) 357.

26 Kathryn P Burdon, 'Role of Direct-to-Consumer Genetic Testing for Complex Disease in Diagnostics and Research' (2015) 43(6) *Clinical & Experimental Ophthalmology* 503.

27 Charbonneau et al (n 7).

8.4.5 REVIEW OF THE OPERATION OF THE THERAPEUTIC GOODS (EXCLUDED PURPOSES) SPECIFICATION 2010

In 2019, the TGA undertook a review of the operation of the *Therapeutic Goods (Excluded Purposes) Specifications 2010* in the regulation of self-testing IVDs in Australia (including DTC-GT).[28] The TGA noted the dilemma created by the lack of jurisdictional competence to regulate overseas DTC-GT providers:

> DTC-GT intended for determining the presence of, or susceptibility to, a disease or condition are currently prohibited under Therapeutic Goods (Excluded Purposes) Specifications 2010 and cannot be legally supplied in Australia. However, DTC-GT that are advertised and supplied from overseas are currently outside the reach of Australia's legislation. Some of these tests may have limited clinical evidence to support their use.[29]

In March 2020, the TGA published on their website 26 submissions received in response to an Issues Paper released in 2019. Submissions were received from a range of stakeholders including consumer groups, health professionals, government agencies, researchers and IVD manufacturers and industry bodies. In the context of DTC-GT, the TGA noted the majority of submissions favoured continued restriction, based on the high potential for harm and the often-doubtful benefits. Four points were highlighted by the TGA in its summary published with the submissions: lack of regulation of overseas providers; the trend for information collected for non-health purposes to reveal health information; adverse consequences of difficulties in interpretation; and added burden on health care professionals.

Recognising consumer demand for DTC-GT and availability of such services overseas, some submissions favoured the option of allowing certain health-related DTC-GTs to be supplied within a well-regulated environment in Australia. The European Union regime was suggested as a model for regulation of IVDs.

Taking into consideration stakeholder feedback the *Therapeutic Goods (Medical Devices - Excluded Purposes) Specification 2020* was re-made and came into effect from 1 October 2020. The re-made *Therapeutic Goods (Medical Devices - Excluded Purposes) Specification 2020* continues to prohibit self-tests for most pathogenic organisms, transmissible agents, or markers of other serious dieases and conditions, including self-tests for cancer and genetic self-tests, such as DTC-GT. It was considered that the risks associated with DTC-GT could not be safely mitigated to reduce potential harms to an acceptable level.

In addition, the re-made *Therapeutic Goods (Medical Devices - Excluded Purposes) Specification 2020* allows for the supply of self-tests for certain serious infectious diseases including influenza (seasonal) virus, some sexually transmitted infections, hepatitis B and C viruses, HIV and markers for some non-infectious diseases, such as kidney disease, diabetes and cardiovascular disease. It should be noted that the *Excluded Purposes Specification 2020* was further amended on 1 October 2021 to allow for the supply of rapid antigen self-tests for COVID-19 in Australia from 1 November 2021.

8.4.6 THE REGULATION OF IVDS IN EUROPE

Although some submissions to the TGA's review of the operation of *Therapeutic Goods (Excluded Purposes) Specifications 2010* appeared to view the European IVD regime favourably, the European regulatory landscape for health-related genetic testing is not without its own challenges.

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28 Therapeutic Goods Administration, *Review of the Regulation of Certain Self-testing In Vitro Diagnostic Medical Devices (IVDs) in Australia* (Consultation Paper, September 2019) <https://www.tga.gov.au/consultation/consultation-review-regulation-certain-self- testing-ivds-australia> ('TGA Review').

29 Ibid 9.

A recent review of legislation relating to DTC-GT across Europe revealed a 'fragmented regulatory landscape'.[30] The legislation in 26 European countries was analysed in this study, revealing widespread divergence from specific prohibition through to regulation through general laws.

There is a general European Union instrument, Regulation (EU) 2017/746 on In Vitro Diagnostic Medical Devices,[31] which regulates genetic tests as IVD medical devices, in much the same way as the Australian IVD medical devices regime. As pointed out in the TGA review, one major difference between the two regimes is that the European Union regime regulates the entire provision of DTC-GT services over the internet.[32] It is this aspect of the regulation of health-related genetic testing that may hold some interest in Australia.

8.4.7 STANDARDS AND ACCREDITATION OF TESTING LABORATORIES

As noted above, the TGA IVD regime requires laboratories providing in-house tests to notify the TGA of the in-house IVDs it is using; meet the NPAAC standard, *Requirements for the Development and Use of In-house In Vitro Diagnostic Medical Devices*; and be accredited by NATA to ISO 15189 (for a medical testing laboratory).

The fourth edition of the NPAAC standard, *Requirements for the Development and Use of In-house In Vitro Diagnostic Medical Devices* was published in 2018.[33] This document is described by the NPAAC as a 'tier 3B' document, providing 'technical and specific detailed requirements for good medical practice in all pathology services'.[34] It provides a detailed list of 11 general and particular requirements to guide laboratories in the provision of genetic testing services.

The NPAAC also published a tier 4 document (technical publications for specific areas of pathology) *Requirements for Medical Testing of Human Nucleic Acids* in 2013, providing specific standards for genomic analysis. Standard 1.1 in this document provides that 'the Laboratory must provide medical nucleic acid testing only in the context of a clinical service provided by a medical practitioner'. Standard 1.2 distinguishes between level 1 (standard tests) and level 2 (tests with potential complex issues – which include certain tests for heritable variants[35]), with level 2 tests requiring additional procedures. Given the developments in whole genome sequencing and whole exome sequencing since the standards were published in 2013, it seems likely that this document will require significant revision in the near future.

Other relevant tier 4 NPAAC documents include Requirements for Cytogenetic Testing (Third Edition 2013) (currently under review) and *Requirements for Human Medical Genome Testing Utilising Massively Parallel Sequencing Technologies* (2017).

In addition to these technical documents, the NPAAC has also published another document, aimed at providing specific guidance to providers of DTC-GT services, *Provision of Direct-to-Consumer Genetic Tests Guiding Principles for Providers*.[36] The stated purpose of the document is to outline 'what is considered by NPAAC to be the guiding principles that should be followed by DTC-GT providers in order to ensure the safety of their clients who seek this type of testing.'

30 Kalokairinou et al (n 27) 119.

31 Regulation (EU) 2017/746 of the European Parliament and of the Council of 5 April 2017 on in Vitro Diagnostic Medical Devices and Repealing Directive 98/79/EC.

32 *TGA Review* (n 31) 14.

33 National Pathology Accreditation Advisory Council, *Requirements for the Development and Use of In-House In Vitro Diagnostic Medical Devices (IVDs) Fourth Edition 2018* (NPAAC Tier 3B Document) <https://www1.health.gov.au/internet/main/publishing.nsf/Content/health-npaac-dhaivd-2018>.

34 'NPAAC Document Hierarchy', *Department of Health* (Web Page, 26 September 2019) <https://www1.health.gov.au/internet/main/publishing.nsf/Content/health-npaac-publication.htm>.

35 NPAAC, *Requirements for Medical Testing of Human Nucleic Acids* (2013) 19-20.

36 NPAAC, *The Provision of Direct to Consumer Genetic Tests: Guiding Principles for Providers* (2015) <https://www1.health.gov.au/internet/main/publishing.nsf/Content/F57C68E5E41DC946CA257EEC000396F2/$File/Genetic%20DTC%20Guiding%20Principles%20-%20May%202014.pdf>.

The document makes a series of recommendations, including that:

- the DTC-GT provider should be accredited as meeting international standards;
- advice should be sought from the TGA as to whether the service is subject to the IVD regime;
- DTC-GT services should not be provided for high penetrance genotypes associated with serious disorders, prenatal testing, preconception carrier screening, or carrier testing in children;
- limitations should be imposed on certain other forms of testing; and
- various measures should be put in place for consumer protection and in order to recognise ethical, legal and social concerns.

It would seem that strict compliance with these recommendations on the part of DTC-GT providers would address many of the concerns raised by stakeholders in the scoping exercise undertaken as part of this project. However, the recommendations are for guidance only and no study has examined compliance on the part of Australian DTC-GT providers.

NATA is the national accreditation body for providers of testing services in Australia. NATA requirements for accreditation include on-site assessment of the laboratory's resources, procedures and documentation. The objective of the assessment is to establish whether the laboratory can competently perform the tests or examinations for which accreditation is sought. The assessment involves a thorough evaluation of all the elements of a laboratory that contribute to the production of accurate and reliable test data.

Internationally, the International Standards Organisation (ISO) has published an extensive set of standards for clinical diagnostic testing and in vitro diagnostic testing systems.[37]

Collectively, NATA accreditation requirements and NPAAC and ISO standards provide a comprehensive set of instructions to providers of genetic testing services. As noted in chapter 7, accreditation of pathology laboratories is required under the *Health Insurance (Accredited Pathology Laboratories-Approval) Principles* (2017), made under the *Health Insurance Act 1973* (Cth) for them to be eligible for Medicare reimbursement. Given that the most recent RCPA survey reveals that 49 per cent of tests are funded by Medicare, accreditation is becoming a core requirement for genetic testing laboratories. Further, as already mentioned in this chapter, compliance with the NPAAC *Requirements for the Development and Use of In-house In Vitro Diagnostic Medical Devices* and accreditation are two of the TGA requirements for manufacturers of in-house genetic tests. Finally, again as noted in chapter 7, the NPAAC guidance documents also represent minimum best practice standards for good laboratory practice that are harmonised with international best practice (where relevant).

8.5 Other Legal and Regulatory Areas

8.5.1 PRIVACY

The general discussion of privacy obligations in Chapter 3 of this report is relevant here, because all entities involved in health-related genetic testing will be bound by privacy obligations. State and territory-based public hospitals and public pathology laboratories will be bound by privacy obligations within their specific jurisdictions. Most companies offering genetic testing services will satisfy the criteria for APP entities under the federal *Privacy Act 1988*. Although some may be able to rely on the small business exemption under s 6D of the *Privacy Act*, this exemption will not be applicable if they collect health or other sensitive information.

37 STANDARDS BY ISO/TC 212, Clinical laboratory testing and in vitro diagnostic test systems,<https://www.iso.org/committee/54916/x/ catalogue/>.

Those DTC-GT companies offering health and wellness related services could argue that they do not provide a 'health service' under s 6FA(a)(iii) of the Act, as defined in s 6FB(1), if they can establish that they are not performing it:

(a) to assess, maintain or improve the individual's health; or

(b) where the individual's health cannot be maintained or improved—to manage the individual's health; or

(c) to diagnose the individual's illness, disability or injury; or

(d) to treat the individual's illness, disability or injury or suspected illness, disability or injury; or

(e) to record the individual's health for the purposes of assessing, maintaining, improving or managing the individual's health.

Studies on public trust in genetic testing clearly indicate that privacy is one of the key consumer concerns potentially deterring engagement in DTC- GT in Australia.[38] On this basis, voluntary compliance with *Privacy Act* obligations may be encouraged, even in the absence of a binding obligation. Despite this, a review of the privacy policies of 15 Australia-based DTC-GT companies by Nicol and colleagues in 2014 shows that compliance with *Privacy Act* requirements for many of them at that time was low.[39] One of the recommendations in that study was the development of a specific privacy code tailored to the DTC-GT industry.

8.5.2 PRODUCT LIABILITY

The *Competition and Consumer Act 2010* (Cth) may apply if a person suffers loss or damage as a result of a defective product. The product liability provisions create a right to compensation to persons injured or whose property is damaged by a defective product.[40]

There are a number of available defences. The state-of- the-art defence (that the state of scientific or technical knowledge at the time when the goods were supplied by their manufacturer was not such as to enable that safety defect to be discovered)[41] is likely to be the most relevant in terms of supply of genetic products. This enables a manufacturer to raise as a defence that the state of scientific or technical knowledge at the time of supply of the product was not such as to enable the defect to be discovered. In addition, a consumer must bring an action within ten years after supply of the goods.[42] Since defects in genetic products may not yet be part of the state of the art, and may not emerge for many years after supply, the adequacy of product liability protections should be carefully scrutinised.

There are other options for legal redress, including contract and tort, particularly negligence. However, commencing a common law action is an onerous task for consumers, particularly for negligence, given that fault on the part of the manufacturer (that is, a breach of the required standard of care) must be proved by the consumer. Issues relating to negligence, particularly relating to return of findings from genomic analyses, are discussed more fully in Chapter 7. Finally, an action may also be brought against a health practitioner whose conduct falls below professional standards under the *Health Practitioner Regulation National Law Act 2009* (Qld). However, as yet there has been no detailed analysis as to how this might apply in the context of genetic testing.

38 Christine Critchley et al, 'Public Reaction to Direct-to-Consumer Online Genetic Tests: Comparing Attitudes, Trust and Intentions across Commercial and Conventional Providers' (2015) 24(6) *Public Understanding of Science* 731.

39 Nicol et al (n 7).

40 *Competition and Consumer Act 2010* (Cth) sch 2 pt 3-5.

41 Ibid sch 2 pt 3-5 s 142.

42 Ibid s 143.

8.5.3 ADVERTISING AND CONSUMER PROTECTION

Advertising restrictions under the TGA apply to advertising through a range of avenues, including on Australian Internet sites. The *Therapeutic Goods Act 1989* and the *Therapeutic Goods Regulations 1990* include provisions regulating the advertising of therapeutic goods. Advertisements for all therapeutic goods must comply with Part 2 of the Regulations and the *Therapeutic Goods Advertising Code (No.2) 2018*, and there are sanctions available under the Regulations for non-compliance. For example, it is an offence to advertise a genetic IVD on the internet unless the device is listed, and the advertisement is directed exclusively to health professionals (see regulations 4 and 5). But these regulations only apply where the Internet service provider is based in Australia.

Misleading statements would also be covered by the misleading and deceptive conduct and other provisions in Schedule 2, *Australian Consumer Law* of the *Competition and Consumer Act 2010* (Cth), administered by the Australian Competition and Consumer Commission ('ACCC'). The ACCC is also a member of the International Consumer Protection and Enforcement Network ('ICPEN') of consumer protection agencies from 24 countries. As part of this, the ACCC and state/territory fair trading agencies examined more than 3000 sites in Australia for misleading representations or other breaches of the consumer protection law. Yet despite this and other international collaborative efforts, difficulties with enforcement remain.

8.5.4 REMUNERATION

Although only a small number of genetic tests have, in the past, been listed on the Commonwealth Medicare Benefits Scheme, there have been some recent additions, most notably for certain BRCA tests[43] and whole genome and whole exome sequencing for certain childhood syndromes.[44] The bulk of funding for genetic testing is provided through state government healthcare budgets on a 'block-funding' model. Where public providers outsource testing to private laboratories, they are likely to be reimbursed through the public budget.

In some instances, patients will pay private providers for the test directly. Non-invasive prenatal testing, for example, is currently only available via private providers and is not reimbursed through public funds. Aside from this example, however, it would appear that there are few restrictions on remuneration for genetic testing when recommended by a health professional.

43 'Medicare Benefits Schedule – Item 73295', *MBS Online: Medicare Benefits Schedule* (Web Page) <http://www9.health.gov.au/mbs/fullDisplay.cfm?type=item&qt=ItemID&q=73295>.

44 'Changes to the MBS May 2020', *MBS Online: Medicare Benefits Schedule* (Web Page, 19 May 2020) <http://www.mbsonline.gov.au/internet/mbsonline/publishing.nsf/ Content/20200501-News>.

9 Tissue Samples and Data

9.1 Summary

Genomics research involves the movement of tissue samples and associated data from participants and patients ('donors') to researchers and clinicians and beyond. Human tissue samples and data collected for both clinical and research purposes are generally comprised in collections housed in various locations, and conditions surrounding their storage, transfer and disposal are governed by a complex web of overlapping legislation, guidelines and policies. Recent recommendations to clarify the arrangements surrounding the regulation of repositories of human tissue and data have been partially addressed.

Tissue samples collected for clinical purposes are frequently repurposed for research use, which can in some circumstances be achieved with a waiver of consent.[1] Tissue samples collected solely for research purposes are often obtained with broad consent to facilitate onsharing and use, including commercial use. While tissue samples are a scarce and finite resource, there is (theoretically) an infinite capacity to share data, limited mainly by technical capacity for storage.

The chapter highlights the current uncertainty surrounding control and ownership of human tissue samples and genomic data. People have intuitive assumptions about rights of ownership or control over their samples and data. However, the legal position with regard to ownership is complicated and involves intersecting laws (including, for example, the human tissue Acts and intellectual property legislation) and the common law. Ownership rights frequently do not provide the control they may be assumed to provide. As a result, alternative ways of conceptualising possession of tissue and data have developed, including custodianship and sovereignty models. Ethical frameworks (most notably the *National Statement on Ethical Conduct in Human Research* — The *National Statement*)[2] play a very significant role in delineating rights of control over tissue samples and data by donors, and the responsibilities of those who seek access.

9.2 The Concept of Control over Tissue and Data

At the centre of debates around ownership of tissue samples and related health data, lie issues of autonomy and control over use and commercial exploitation. The issues dealt with in this chapter arise from the use and sharing of tissue samples and related health data, and concern the question of who (if anyone) has ownership and/or control of tissue samples and the genomic data derived from these samples. Control is a critical matter for many stakeholders in the genomics sector; for many donors, maintaining control over what use is made of their tissue/data and by whom is their chief concern. This sense of having control over use is often viewed as imperative when decisions are made as to whether to donate tissue. At the same time, clinicians require a degree of control in order to be able to provide effective clinical care, and researchers rely on control over samples and data in order to be able to freely conduct research. To further complicate matters, permitting control over collections of tissue and data (by, for example, those coordinating repositories and biobanks) helps to facilitate strong data protection and at the same time, socially valuable sharing practices. To businesses involved in using tissue samples and data to produce commercial products, the capacity to exercise control over their use of samples and data is viewed as necessary to realise a return on investment.

1 See parts 2.3.1.3 and 2.3.4 of this report.

2 National Health and Medical Research Council, Australian Research Council and Universities Australia, *National Statement on Ethical Conduct in Human Research* (Guideline, 2018).

Not surprisingly given the range of actors involved in genomic medicine and research, control in this context may take a variety of forms. These forms of control may be interrelated but conflicting, in the sense that how we define control in a legal sense differs from what control means to a lay person who might donate tissue. The discussion in this chapter is arranged around various 'modes' of control:

- *Control through ownership and other proprietary rights*. In a legal sense, ownership encompasses the concept of property rights, and has a narrow meaning. Ownership confers certain rights which are elaborated below.
- *Control via ethical frameworks*. Frameworks established through the *National Statement* establish stringent governance structures for the collection, storage and use of tissue samples and data, which in turn play an important role in the management and control of tissue and data in clinical practice and research.
- *Control through intellectual property rights*. Intellectual property rights are intangible rights that may be claimed and exercised by researchers over certain outputs of research. They take a number of forms and confer differing degrees of control to holders of those rights.
- *Custodianship and sovereignty*. Custodianship refers to responsibility for the storage, maintenance and authorised use of genetic or genomic data.[3] Custodianship is the aspect of data governance which concerns its management; data stewardship confers responsibility for what is collected or stored. Data sovereignty is the concept that data governance will be subject to the laws of the country in which the data is stored, an issue that becomes more prominent where there are more than one set of applicable laws (such as statutory versus traditional laws).
- *Controlling provenance*. The provenance of a tissue sample or dataset refers to its source or origin, and ensuring accurate records of sources and transfers of tissue and data helps maintain a strong chain of custody over that sample or data.

9.3 Overview of Law

9.3.1 TISSUE AND DATA STORAGE, USE AND DISPOSAL

Collections of human tissue samples may consist of samples gathered in either clinical or research settings. The Australian Law Reform Commission ('ALRC') and Australian Health Ethics Committee ('AHEC') in their report, *Essentially Yours*, identified three main repositories for tissue samples collected in the clinical context: archived pathology samples; newborn screening cards; and tissue banks.[4] Issues relating to the collection, storage, quality, security and disposal of tissue samples stored in these repositories are dealt with via information and health legislation, the state and territory human tissue laws, guidelines governing pathology laboratories, ethical guidelines and policy statements, and government health authority policies. This complex set of requirements applies equally to tissue sourced for genomic and other analysis. The ALRC and AHEC reviewed these requirements and concluded that they gave rise to some concerns, particularly in relation to the unconsented storage and secondary use of samples for research. The ALRC and AHEC also highlighted the lack of national consistency in policies and practices governing the storage of tissue samples, and recommended that the need for nationally consistent rules be considered.[5]

3 Ibid cl 3.1.55.

4 Australian Law Reform Commission and Australian Health Ethics Committee, *Essentially Yours: The Protection of Human Genetic Information in Australia* (Report No 96, May 2003) 355 <http://www.alrc.gov.au/publications/report-96> ('*Essentially Yours*'). ch 18.

5 Ibid Recommendation, 19.1, 19.2.

These recommendations were accepted by the Australian Government,[6] and national standards which impact on the storage of newborn screening cards subsequently adopted.[7] Development of specific legislation governing collections of other genetic information has not eventuated, and the Government declined to accept recommendations by the ALRC and AHEC[8] relating to using the *Privacy Act 1988* (Cth) *('Privacy Act')* to provide protections to those whose genetic samples were collected, stored, and handled on the grounds that the *Privacy Act* deals with information rather than tangible samples. Instead, the government stated that amendments to the human tissue Acts would be more appropriate. Again, however, amendment to this effect has not occurred. Amendments to the *National Statement on the Ethical Conduct of Research involving Humans* (2007, as amended in 2018) ('*National Statement*') have gone some way towards addressing the ALRC and AHEC's concerns in the research context, although because the National Statement does not apply to clinical practice, it is not applicable in respect of samples collected for clinical purposes and retained in collections that are not subsequently put to research use.

Tissue samples collected primarily for research purposes are generally stored in collections at hospitals, individual research organisations or pathology laboratories. These collections may be held by both public or private/commercial entities. Additionally, many researchers avail themselves of the use of collections derived from clinical collection of human tissue, as described in the preceding paragraph.[9] The ALRC and AHEC broadly defined *research* collections of human tissue and/or human genetic information as human genetic research databases, and observed that the regulation of these databases is via legislation, State and Territory human tissue laws, the *National Statement*, and other guidelines. The ALRC and AHEC pointed to a number of deficiencies in the regulation of human genetic research databases, and recommended review of the *National Statement* to address consent to future uses, and the establishment of a mandatory public register of databases.[10] The NHMRC subsequently revised the *National Statement* to incorporate a number of the safeguards recommended in Essentially Yours, including more stringent conditions on the establishment and operation of databases and the samples and information contained therein.

The legal frameworks existing around tissue management are complex and to an extent, in a state of flux. Recent reviews conducted in respect of tissue donation (including organ donation, retrieval and transplantation, eye and ocular tissue, and haemopoietic progenitor cells),[11] were responded to through the drafting of frameworks which will guide future operations and management of these sectors.[12]

9.3.2 CONTROL THROUGH OWNERSHIP AND OTHER PROPRIETARY RIGHTS

Many people feel strongly that they 'own' tissue samples provided for either clinical or research use, and any associated genetic/genomic data. Legal ownership has a very specific meaning, and refers to the existence of a legally recognisable property right in a good. Property is '…a description of a legal relationship with a thing' and the right to exercise power over that good.[13] In the legal sense, property is commonly referred to as a 'bundle of rights'.[14]

6 Australian Government, *Protection of human genetic information* (Web Page, 2003) <https://www.alrc.gov.au/inquiry/protection-of-human-genetic-information/>.

7 Australian Government Department of Health, *Newborn Bloodspot Screening – National Policy Framework* (Policy, 2018).

8 *Essentially Yours* (n 4) Recommendation 8-1.

9 Ibid 470.

10 *Essentially Yours* (n 4) Recommendations 18.1 and 18.2.

11 See Australian Government Department of Health, *What we're doing about organ and tissue donation* (Web Page, 2020) <https://www.health.gov.au/health-topics/organ-and-tissue-donation/what-were-doing-about-organ-and-tissue-donation>.

12 Review of the organ donation, retrieval and transplantation system, Final Report (Commonwealth of Australia, 2018) and All Governments' Statement, at https://www.health.gov.au/resources/publications/review-of-the-organ-donation-retrieval-and-transplantation-system-final-report.

13 *Yanner v Eaton* (1999) 201 CLR 351, 365-6.

14 See, eg, Wendy Bonython and Bruce Baer Arnold, 'Privacy, Personhood and Property in the Age of Genomics' (2015) 4 *Laws* 377, 386.

Proponents of ownership in the health arena assert that the concept of ownership is necessary because it brings with it certain legally recognised property rights, including (but not limited to) the right to exclude, the right to access or possess and the right to destroy.[15]

The issue of ownership is important, because it is only once questions of ownership have been clarified that the legality of dealings in tissue and data may be examined.[16] Although we frequently think of property rights in terms of donors of genetic material, the issue of property in tissue has also been explored in the legal literature in the context of test kit providers, researchers, clinicians, biobanks, other family members, beneficiaries where donors have died, or commercial entities where patients have used direct-to-consumer testing services or third-party intermediaries.

9.3.2.1 Proprietary rights in human tissue

Whether it is possible to 'own' human tissue has never been definitively resolved in Australia. There is no legislation that deals specifically with property interests in human tissue. The human tissue laws in each Australian State and Territory deal with the donation of tissue,[17] but their scope is limited. The statutes were principally enacted to deal with organ donation from living and deceased donors, and as such, the following is excluded from their operation: tissue removed for purposes other than donation, post-mortem or anatomical study; reproductive material; and tissue removed during consensual medical treatment,[18] or treatment that was necessary for the preservation of life. Given the broad exceptions for tissue and blood samples removed for medical treatment, the transfer of possession of the majority of human tissue that finds its way into research uses is not regulated by the laws. Instead, the most pertinent requirements come through ethical guidelines and policies, in particular the *National Statement*. In any case, the human tissue laws do not deal explicitly with questions of ownership.

Based on a growing body of Australian and overseas case law,[19] individuals will only have proprietary rights in human tissue in limited circumstances. It is not clear whether these interests will amount to ownership, and in some cases more than one party may have simultaneous proprietary interests (see the discussion below on the cases concerning storage of sperm). A proprietary or possessory interest is a right to exercise control over property that may amount to a lesser interest than ownership. A tenant with a long-term lease, for example, has a proprietary interest despite not owning the property in question. A proprietary interest may give a right to exercise control, but not include the right to immediate control that ownership gives.

It is well established under Australian law that a body part can constitute property capable of possession where there has been an application of work and skill (such as excision, dissection or preservation).[20] On the basis of this, and case law from other jurisdictions, this principle seemingly applies to preserved tissue,[21] body parts,[22] body tissue removed during surgery,[23] and cell lines derived from tissue samples.[24]

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15 See, for example, Jessica L Roberts 'Progressive Genetic Ownership' (2018) 93(3) *Notre Dame Law Review* 1106, 1128, 1130.

16 Jane Nielsen, Dianne Nicol, Tess Whitton and Don Chalmers, *My Way or the MTA: The Use of Material Transfer Agreements in Publicly Funded Research in Australia* (Occasional Paper No 10, 2018).

17 *Human Tissue Act 1985* (Tas); *Human Tissue Act 1983* (NSW); *Transplantation and Anatomy Act 1979* (Qld); *Transplantation and Anatomy Act 1983* (SA); *Human Tissue Act 1982* (Vic); *Human Tissue and Transplantation Act 1982* (WA); *Transplantation and Anatomy Act 1978* (ACT); *Human Tissue Transplant Act 1979* (NT).

18 Tissue removed during medical treatment is not excluded under the *Human Tissue Act 1983* (NSW).

19 Note that although only Australian case law must be followed by Australian courts, the case law of other jurisdictions can be used to guide judicial reasoning. The case law of England, in particular, has been particularly influential, as has Canadian case law. Where there is very little case law, these cases can be usefully considered.

20 *Doodeward v Spence* (1908) 8 CLR 406.

21 Ibid.

22 *R v Kelly* [1999] 2 WLR 384; *AB and Others v Leeds Teaching Hospital NHS Trust* [2005] 2 WLR 358.

23 *Roche v Douglas* [2000] WASC 146.

24 *Moore v Regents of the University of California*, 249 Cal 494 (Cal Ct App, 1988); *Greenberg v Miami Childrens' Hospital Research Institute Inc*, 264 F Supp 2d 1064 (Fla, 2003).

The real question is in whom this interest resides. It is widely accepted in law that a person does not legally own their tissue while it resides in their body. They do, of course, have legal rights to control what happens to their body, but this does not equate with legal ownership of the constituent parts.[25] Some excision from the body and subsequent application of work and skill is necessary, and at this point, the party who undertakes the relevant procedure (or their institution) generally acquires a proprietary interest.[26] In most cases this will be a clinician or researcher rather than the tissue donor. Under current authority a tissue donor may have no proprietary interest once tissue has been donated, especially where it has been donated for research purposes.[27] The same might apply to tissue removed during surgery, unless agreed otherwise.

Despite this, there has been relatively recent English and Australian authority that men and their beneficiaries have a possessory interest in sperm stored for reproductive purposes after cancer treatment,[28] or after their death.[29] The claims in these cases rested on the fact that the men had a legal right to claim against the laboratories who held their sperm, and were entitled to demand its return. The precise scope of these decisions is not yet clear, but they do suggest that broader proprietary interests in human tissue may be recognised at common law. Importantly, it is not clear from these cases that an *ownership* interest existed: the interests of the donors were not framed in this way, and the interests of the laboratories and the donors existed concurrently.[30] Although a proprietary interest existed, it amounted to less than a comprehensive ownership right.

It is currently unclear what application these cases might have for other forms of human tissue. A primary difference between the 'sperm cases' and instances where tissue is excised for medical purposes and/or donated for research, is that there was an obvious intention on the part of the donors or their beneficiaries to use their sperm. This will not usually be the case with tissue donated for research.[31]

It is also unclear whether tissue stored for other purposes (for example, biomaterials stored in biobanks or blood banks) would be treated similarly.[32] Another case that has been touted as potentially ushering in proprietary rights in tissue samples, is *Roche v Douglas*, a decision of the Supreme Court of Western Australia. Susan Roche sought to access stored tissue which had been removed from the body of the late Edward Rowan during surgery, to enable her to use the sample to determine whether the deceased was her father.[33] Ms Roche needed to establish that the stored sample was property, in order for the court to make the requisite access order. Master Sanderson held that in this particular case it was possible to say the sample was proprietary, and while this was enough to trigger the court's power to make an order in favour of Ms Roche it was not necessary to determine *who* held a proprietary interest.

25 Ibid.

26 *Pecar v National Australia Trustees Ltd* (The Estate of Ivan Urlich deceased) [1996] NSWSC 4; *R v Kelly* [1999] 2 WLR 384.

27 *Moore v Regents of the University of California*, 249 Cal 494 (Cal Ct App, 1988); *Greenberg v Miami Childrens' Hospital Research Institute Inc*, 264 F Supp 2d 1064 (Fla, 2003).

28 See, eg, *Yearworth v North Bristol NHS Trust* [2009] 2 All ER 986; *Roblin v Public Trustee* (ACT) [2015] ACTSC 100.

29 See, eg, Re *Estate of Edwards* (2011) 81 NSWLR 198; Re H, AE (No 2) [2012] SASC 177; *Cresswell v AG for the State of Queensland* [2018] QSC 142.

30 Jane Kaye et al, 'Trends and Challenges in Biobanking' in Ian Freckleton and Kerry Peterson (eds), *Tensions and Traumas in Health Law* (The Federation Press, 2017) 415, 429.

31 Dianne Nicol et al, 'Impressions on the Body, Property and Research' in Imogen Goold et al (eds) *Persons, Parts and Property: How Should We Regulate Human Tissue in the 21st Century?* (Hart Publishing, 2014) 9.

32 Loane Skene, 'Proprietary Interests in Human Bodily Material: Yearworth, Recent Australian Cases on Stored Semen and their Implications' (2012) 20(2) *Medical Law Review* 227, 239.

33 [2000] WASC.

The upshot of this emerging body of case law is that in some circumstances, proprietary rights in tissue samples exist. Any proprietary right that does arise is unlikely to amount to full ownership. There is no clear answer to the question of whether donors have an unequivocal proprietary interest in their own tissue. The problems that would arise for genetic research should such an interest be recognised have been considered.[34] A limitation is that these cases were largely decided on their particular facts, casting doubt on their value as precedent.

What does seem to be more clearly established is that a researcher who works or exercises skill on human tissue has a potentially stronger proprietary interest than the donor. Arguably this interest arises because the donor (assuming they have some proprietary interest in an excised sample) transfers the sample to the researcher (or more accurately the institution employing the researcher)[35] through a gift or some other legal mechanism.[36] In the meantime, there is no impediment on a researcher being able to transfer that sample to other researchers or even commercial entities, provided the researcher adheres to other legal and ethical obligations.[37]

What is unclear is whether there are any more nuanced arguments which may be advanced to enable proprietary interests to vest in a donor of tissue. For example, whether it may be possible for a cancer patient to argue that the provision of access to a stored tissue sample provides the only mechanism for that patient to procure access to a tumour sample. Another possibility is an argument that the tissue sample would not exist 'but-for' the donor donating the sample, although arguments to this effect are untested.

9.3.2.2 Proprietary rights in genomic data

The question of who might have proprietary rights in data is even more problematic than that of proprietary rights in tissue. Comments by consultation stakeholders illustrate the strong views around this issue, with a number making comments about possession of genomic data that used language ordinarily associated with 'ownership': 'what is one's right over their own data?'; 'is [data] considered an asset … and … is the law saying because it is manipulated and not in original form it is not part of you (and you don't own it)?' (Stakeholder 16). These issues were particularly acute for Aboriginal and Torres Strait Islander stakeholders who stressed the importance of sovereignty over their data.

Increasingly, we are hearing calls for the propertisation of health data,[38] which have followed the broader movement towards ownership of digital data.[39] This often takes the form of allowing individuals to 'own' their genomic data and other health information. Many individuals believe their health data belongs to them. This view has been echoed in statements by public officials (including former President Barack Obama).[40] The language of ownership of health information is certainly becoming more mainstream. Services offered by third-party genetic interpretive services offering 'literature searching functions'[41] are often premised on the notion that donors 'own' their genetic data.[42]

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34 See, eg, Justine Pila, 'Property in Human Genetic Material: An Old Legal Question for a New Technological Age' in TK Hervey & D Orentlicher (eds), *The Oxford Handbook of Comparative Health Law* (OUP, 2019); Australian Law Reform Commission, Protection of Human Genetic Information (Report, 2002) ch 17.

35 *Washington University v Catalona* 437 F Supp 2d 985 (ED Mo, 2006).

36 Nielsen et al (n 16) 184-5.

37 Cameron Stewart, Jennifer Fleming and Ian Kerridge, 'The Law of Gifts, Conditional Donation and Biobanking' (2013) 21 *Journal of Law and Medicine* 351, 236-7.

38 There is substantial literature on this point. See, eg, Jessica Roberts, 'Progressive Genetic Ownership' (2018) 93(3) *Notre Dame Law Review* 1105-1172.

39 Paul Schwartz, 'Property, Privacy, and Personal Data' (2004) 117 *Harvard Law Review* 2056.

40 Julie Hirschfeld Davis, 'President Weighs in on Data from Genes' *New York Times* (online 25 February 2016) <https://www.nytimes. com/2016/02/26/us/politics/president-obama-weighs-in-on-data- from-genes.html>.

41 Lauren Badalato, Louiza Kalokairinou and Pascal Borry, 'Third Party Interpretation of Raw Genetic Data: An Ethical Exploration' (2017) 25 *European Journal of Human Genetics* 1189.

42 See, eg, Codegen 'Welcome to Codegen' (Web Page, 2020) <https://codegen.eu/>

An emerging business model in the health sphere is data management intermediaries, which pay patients for their health data and provide connections to researchers and commercial entities.[43] The assumption behind these businesses is that an individual's data is theirs to sell. An argument has even been mounted that the right to own one's data is a fundamental human right.[44] Critically, the focus of these ownership discussions is often not so much about having legally recognised ownership rights in goods, but the ability to exert control over them.[45]

The central issue here is whether it is possible to own genomic data, and if so, in whom proprietary rights might vest. Intuitively, the idea of ownership over one's genomic data is very appealing.[46] To many, it seems 'right' that individuals should be entitled to determine how their data is used, a result which would be brought about if they are considered to own their data.

When calls for ownership of genomic data are made, it is not immediately clear what 'data' means in this context. There is some conflation of a number of terms, including 'data', 'facts' and 'information'.[47] Often these terms are used interchangeably and inconsistently. Data, broadly categorised can be:[48]

- representative in that they involve some kind of measurement;
- implied where they are read into an absence, typically involving inferences; or
- derived, or produced from other forms of data.

Health data may fall into any of these categories. This includes genomic data, depending on the level to which it is annotated.

Facts are in the nature of representative data and can be characterised as being objective realities. Information is 'contextualised facts': '[w]hile facts are raw, information is processed.'[49] The distinction between representative data, facts and information is murky, but what is generally accepted is that facts are considered to be in the public domain.

Why has there been an increasing interest in 'protecting' the rights of data donors through data ownership? At the heart of calls for ownership of personal health information, reside privacy concerns and a desire to provide individuals and vulnerable groups with the means to protect and control genomic information.[50] There are two main grounds for insistence on ownership rights, both of which are donor-centric. The first is that it would safeguard privacy, which is important in the context of genomic data. Ownership, it is argued, would give the highest form of protection for individuals.[51] The second is that it would facilitate self-determination and benefit sharing, particularly among Indigenous populations.[52]

43 See, eg, LunaDNA, 'LunaDNA' (Web Page, 2020) <https://www.lunadna.com/>.

44 See, eg, Hu-manity.co, (Web Page, 2020) <https://hu-manity.co/>; see also, Kara Ching, 'Indigenous Self-Determination in an Age of Genetic Patenting: Recognizing an Emerging Human Rights Norm' (1997) 66 *Fordham Law Review* 687, 707.

45 Amy McGuire et al, 'Who Owns the Data in a Medical Information Commons?' (2019) 47 *The Journal of Law, Medicine and Ethics* 62, 62.

46 The British Academy, The Royal Society and techUK, *Data ownership, rights and controls: Reaching a common understanding* (Seminar Report, 3 October 2018) 5.

47 Teresa Scassa, *Data Ownership*, CIGI Papers No 187 (Centre for International Governance Innovation, 2018) 3-4.

48 Rob Kitchen, *The Data Revolution: Big Data, Open Data, Data Infrastructure & their Consequences* (London: Sage, 2014), 1.

49 Scassa (n 47) 4.

50 See, eg, Richard Spinello, 'Property Rights in Genetic Information' (2004) 6 *Ethics and Information Technology* 29, 30.

51 Ibid 33.

52 Ibid 34-5.

Even if arguments for ownership/proprietary rights are accepted, there is a lack of consensus on *who* should own health data, closely tied to ideas about the desired benefits of an ownership regime. Many scholars argue (on top of the justifications outlined above) that vesting proprietary rights in individuals would advance research, because it would provide them with an incentive to sell access to their data.[53] Others argue for public ownership because it would enhance access to genetic/genomic data by researchers.[54] Complicating the discussion, there is a real issue around whether proprietary rights might vest in *compilers* of data, where genomic data is collated in databases or biobanks.

Some legal developments in the US and the European Union (EU) reflect these apprehensions. Several American states have passed legislation purporting to vest property in genetic data in individuals.[55] However others argue that even without statutory rights, a common law right to property in genetic information is emerging.[56] In two separate cases, US courts have held that donors have a sufficient property interest in genetic data to found a common law action in conversion.[57] Conversion allows a person to claim that an item of personal property has been wrongfully taken and used. Further, Contreras argues that the requirement for informed consent mimics a property regime for genetic data, and that genetic information has now taken on many characteristics of personal property.[58]

Can it be said that genetic/genomic information bears characteristics of property that would give donors rights to control its use? In the EU, the General Data Protection Regulation[59] (GDPR) has given individuals unprecedented levels of control over their personal data, which has been argued are akin to property rights.[60] There have also been several proposals within Europe and Japan that may lead to policy changes that would characterise digital data as property;[61] conceivably this could extend to genetic data. Although some debate as to the need for a standalone data ownership right has been evident in the EU, this push has gained limited traction.[62] One Canadian Supreme Court case held that although a patient retained an ongoing interest in information divulged to a medical practitioner, this did not amount to an *ownership* interest in the information contained in his/her physical medical records.[63]

The position in Australia is even less clear. It is clear that any proprietary interest in the information contained in medical records resides in the creator of those records.[64] There is no Australian authority dealing directly with the issue of whether genomic data is property. It might be assumed, however, that similar conclusions would be reached. As to whether digital data in a general sense might be considered property, there is a similar lack of authority aside from ownership arising in the context of copyright, which is considered below.

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53 See, eg, Mark Hall, *Property, Privacy, and the Pursuit of Interconnected Electronic Medical Records* (2010) 95 *Iowa Law Review* 631, 651.

54 See, eg, Marc Rodwin, 'The Case for Public Ownership of Patient Data' (2009) 302 *Journal of the American Medical Association* 86, 87-8.

55 For discussion see, eg, *Anya Prince, Comprehensive Protection of Genetic Information: One Size Privacy or Property Models May Not Fit All* (2013) 79 *Brooklyn Law Review* 175, 195–8; McGuire et al (n 41) 66; Statutory statements that genetic data is the property of the donor exist via legislation in Alaska, Colorado, Georgia, Louisiana, and Florida.

56 Jorge Contreras, 'Genetic Property' (2016) 105 *The Georgetown Law Journal* 1, 6.

57 *Peerenboom v Perlmutter* No. 2013-CA015257 (Fla. Cir. Ct., 2017); *Cole v Gene by Gene Ltd* No. 1:14-cv-00004-SLG, 2 (U.S. Dist., 2017). Discussed in Roberts (n 4) 1109-10.

58 Contreras (n 56) 20-37.

59 European Parliament and the Council, *Protection of Natural Persons with Regard to the Processing of Personal Data and on the Free Movement of Such Data, and Repealing Directive 95/46/EC* 27 April 2016.

60 Nadezhda Purtova, 'Do Property Rights in Personal Data Make Sense After the Big Data Turn?' (2017) 10 *Journal of Law and Economic Regulation* 208, 214.

61 Jeffrey Ritter and Anna Mayer, 'Regulating Data as Property: A New Construct for Moving Forward' (2018) *Duke Law & Technology Review* 16(1) 220, 226-40. The Organisation for Economic Cooperation and Development has made similar noises in respect of digital data.

62 Scassa (n 43) 15.

63 *Mclerney v McDonald* [1992] 2 SCR 138 [22]; discussed in Scassa (n 37) 13.

64 *Breen v Williams* (1996) 186 CLR 71, 11] – [12]

Those who oppose ownership of data generally do so on the basis that giving donors the right to exclude information from use by third parties could limit the amount of data distributed for sharing in research.[65] The existence of such a right would be antithetical to the development of a biomedical research commons,[66] and has the potential to cause an anticommons.[67] Anticommons was a term initially coined by Heller and Eisenberg to explain the concerns where a field of research is cluttered by patents and subject to underuse where no single party has an effective privilege of use.[68] In the context of seeking genomic data for research, it could arise if the process of negotiating rights to access data from individuals is (or is perceived to be) too onerous. In any case, data pertaining to individuals are more useful in their aggregated form, because it is the aggregation of data that provides a reference base for clinical or research use. Arguments for property more appropriately rest on the assertion that genomic data should be viewed as a form of common property (if indeed any proprietary interest subsists), rather than individual property.[69]

65 See, eg, Spinello et al (n 46).

66 Patricia Deverka et al, 'Creating a Data Resource: What Will it Take to Build a Medical Information Commons?' (2017) *Genome Medicine* 9.

67 Spinello (n 46) 35.

68 Michael Heller and Rebecca S Eisenberg, 'Can Patents Deter Innovation? The Anticommons in Biomedical Research' (1998) 280 *Science* 698, 698.

69 Jonathon Montgomery, 'Data Sharing and the Idea of Ownership' *The New Bioethics* 23 (2017) 81-86.

A Broad Overview of the Application of the GDPR to Ownership and Control of Data

Key Definitions under Article 4 of the GDPR

- Data controllers are natural or legal persons, public authorities, agencies or other bodies which alone or jointly determine the purposes and means of the processing of personal data.
- Data processors are natural or legal persons, public authorities, agencies or other bodies which process personal data on behalf of a controller. Processing means any operation or set of operations performed on personal data or sets of personal data and includes collection, storage, dissemination and erasure.
- A data subject is an identified or identifiable natural subject. Data subjects have a strong degree of control over their personal data, including rights to access, erasure and restriction of processing in certain circumstances.
- Personal data is broadly defined as any information relating to an identified or identifiable person, and includes genetic information and health information. These categories of data are afforded added layers of protection under the GDPR.

An Example of the Operation of the GDPR

- Genomics England was established by the NHS to manage genomics data collected by the National Health Service (NHS) and to run the 100,000 Genomes Project to research cancers and rare diseases. That project is now transitioning to the NHS Genomic Medicine Service (NHS GMS).
- Genomics England's Privacy Policy states that it is a data controller for processing undertaken in a limited number of areas. Pursuant to a contract with the NHS, it is a data processor for all other data it processes, including data derived from tests through the NHS GMS. NHS England is data controller for this information.
- Genomics England does not possess an unfettered right to process and transfer data controlled by the NHS.
- The GDPR (Recital 81, Article 28(3)) provides that a data processor must process personal data only with documented instructions from a data controller, which includes transfers to other countries or international organisations.
- Genomics England's ability to transfer data internationally would therefore be contingent on permissions granted in the contract between the NHS GMS and Genomics England.

Implications

- Under the GDPR, notions of 'ownership' of data by data subjects are strengthened.
- Stringent conditions are placed on processing of data, in respect of which responsibility ultimately lies with data controllers. Heavy penalties may be imposed on those in breach of these conditions.
- Contractual arrangements may operate to provide a data processor with approval to process data in a manner not authorised by Article 28(3) of the GDPR. They may, for example, allow a party such as Genomics England to transfer data internationally.

9.3.3 ETHICAL FRAMEWORKS IMPACTING ON CONTROL

Issues associated with consent and privacy are canvassed elsewhere in this report (Chapters 2 and 3), but these questions are closely related to questions of ownership/property.

The underlying question is whether individual rights of control through privacy and informed consent provide sufficiently strong protection to individuals. Are these rights to control so robust that additional 'ownership' rights would add little?[70] Although individuals do not have proprietary rights in their medical data, they do have rights of access. In Australia, the High Court ruled in *Breen v Williams*[71] in 1996 that the duty owed by doctors to their patients does not extend to providing patients with an unrestricted right to access their medical records, with the High Court observing that any change in the law must be implemented through legislation. The principle has been legislatively overridden in certain Australian jurisdictions.[72]

Subsequently, My Health Record has given Australian health consumers unprecedented levels of access to their medical records. In addition, Australian privacy laws[73] and freedom of information laws give patients rights to request access to their medical information.[74] This is different, of course, to saying that a patient has a proprietary interest in his/her medical records, although even the High Court in *Breen v Williams* was willing to accept that a patient is more likely to be able to successfully argue that medical records obtained on behalf of a patient and paid for by that patient (for example x-rays or pathology reports), give rise to a proprietary interest on the part of the patient.[75] This has implications for genomic test results where those tests have been obtained at the patient's expense. The matter is not so clear cut where tests are obtained under the public system. Nor is there a clear answer where health care is delivered by teams of health care providers, with patient records comprising varying combinations of test results and medical notes produced via intellectual effort by multidisciplinary medical providers.

The situation is comparable in other jurisdictions. Individuals also have a right to access and retain copies of their health data under the HIPAA Privacy Rule in the US.[76] A similar situation exists in Canada and in European jurisdictions (courtesy of the GDPR).[77] These rights are fairly powerful, and access to data is likely to be increasingly easy to facilitate as medical data becomes digitised. Arguably they provide rights that are almost equivalent to proprietary rights. Proprietary rights are not absolute. They rarely lead to unfettered rights to control.[78] Even if a proprietary interest is found to exist, an individual may not successfully obtain a satisfactory remedy in the circumstances. Courts may be unwilling to order, for example the right to return of property, or the right to have property destroyed. Consequently, there is no substantial variance between the proprietary rights in information and the rights attributed by privacy legislation.

70 See, eg, Contreras (n 56). Contreras goes further and would restrict the informed consent regime so that consent does not need to be obtained ex post.

71 (1996) 186 CLR 71.

72 See, eg, the *Health Records and Information Privacy Act 2002* (NSW), which provides patients with the right and an associated process to access health information; see also the *Health Records Act 2001* (Vic).

73 *Privacy Act 1988* (Cth) pt 5, APP 12.

74 Office of the Information Commissioner, 'Freedom of Information' (Web Page, 2020) <http://www.oaic.gov.au/freedom-of-information/>.

75 (1996) 186 CLR 71, [13] (Dawson and Toohey JJ).

76 Deverka et al (n 61).

77 Scassa (n 47) 15; Ritter and Mayer (n 61).

78 The British Academy (n 46) 11-2.

It has also been suggested that another feasible approach in the case of personal data is to shift the debate to consent, rather than focusing on proprietary rights.[79] Giving donors the right to make informed decisions about the use of their genomic data gives them some control over prospective uses (and aligns with privacy laws). However, although this would imbue donors with a sense of control over use of their data, practically it gives rise to many of the same issues that would arise with proprietary rights. It places a heavy burden on individuals to understand and consider the implications of giving informed consent to various uses of their data prior to relinquishing a degree of control over their data.[80] Depending on how broadly it is drafted, a valid informed consent (for example, in the research context) has the potential to remove a patient's right to restrict access to their personal data by that researcher, particularly once data has been incorporated into study results. The limitations of consent as an instrument to facilitate control are canvassed elsewhere in this report.

Despite these potential limitations, a broader focus on ethical research use rather than proprietary rights may be a useful compromise[81] and potentially more feasible given the prevalence of large datasets.[82]

9.3.4 CUSTODIANSHIP AND SOVEREIGNTY

Another way of framing the question of control is in terms of 'custodianship'. In data governance terms, data custodians have particular rights and responsibilities in respect of the management of data, including compliance with relevant legislative requirements. Hence, custodianship incorporates accountability on the part of the custodian of the data. These protections are broadly consistent across jurisdictions. Access to information produced about individuals or held by Australian organisations or government agencies is premised on adherence to strict data custodianship requirements, set out in ethical guidelines,[83] various statutes and other instruments. The Australian Government is currently developing legislation that creates a scheme for sharing of public sector data with accredited users. The legislation requires custodians of public sector data to ensure they have authority to share data and that appropriate safeguards are in place before sharing can take place.[84]

This legislation will sit alongside existing safeguards such as the privacy legislation. The Australian Government Public Data Policy Statement outlines the government's position on open data, which covers non-sensitive public data. This may not include health-related genomic data.[85]

Outside specific statutory requirements, good custodianship practices can help to drive safe data management practice, and should be exercised by all those with access to data, including, for example, researchers, commercial parties and biobanks. Custodianship obligations on the part of funding bodies will also arise in some circumstances. Where clinical genomic testing is conducted pursuant to public funding the public funding body will assume the statutory and ethical responsibilities outlined in the preceding paragraph. Where research is publicly funded, accountability for the management and use of data will reside in those with access to the data (generally researchers and their institutions). This is usually cemented through a contractual relationship between the funding body and the researcher, which will additionally address questions of intellectual property ownership (see further below on intellectual property). In Europe, custodianship obligations that accompany release of personal data have been further cemented by the GDPR which rigidly prescribes the conduct of (among other parties) data custodians, which as we have seen is broadly defined.

79 Ibid 6.

80 Ibid.

81 Ibid 8.

82 Pamela Andanda, 'Towards a Paradigm Shift in Governing Data Access and Related Intellectual Property Rights in Big Data and Health-Related Research' (2019) 50 IIC 1052, 1062.

83 National Health and Medical Research Council (n 2) 3.1.44, 3.1.55- 3.1.57.

84 Office of the National Data Commissioner, *New Legislation* (Web Page, 2020) <https://www.datacommissioner.gov.au/data-legislation>.

85 Australian Government, *Australian Government Public Data Policy Statement* (Policy Statement, 10 December 2020).

Custodianship has also been used to mean a model of data governance in which data is held on behalf of donors with their interests as a guiding principle.[86] This is most clearly evident in relation to marginalised or disadvantaged groups. A related development has been the rise of the Indigenous Data Sovereignty movement, which asserts the collective rights of Indigenous peoples to control 'over the collection, governance, ownership, and application of data about their peoples, territories, lifeways and natural resources.'[87] Although not limited to genomic samples and data, this movement finds particular application in this field, given the group harms that have occurred as a result of misuse by non-Indigenous researchers (Chapter 4).

Arising in response to detrimental experiences with official data and administrative statistics, Indigenous Data Sovereignty is defined as 'the management of information in a way that aligns with the laws, practices and customs' of the group to which the information relates. While this approach includes all data collected in relation to indigenous peoples, it has found particular application in genomics, given the historical misuse of the genomic samples and data of indigenous peoples. Advocates for Indigenous Data Sovereignty in genomics argue that their position is an implication of the United Nations Declaration on the Rights of Indigenous Peoples.[88] Specifically, attention is drawn to article 3, articulating a right to self-determination, and article 31, referring to the right to control and protect human and genetic resources.

Models of genomic data governance in research that promote Indigenous Data Sovereignty include Indigenous-led research, community partnerships, co-design, and participatory governance. Such models are consistent with, and encouraged by, Australian ethical guidelines, including the *National Statement* and *Ethical Conduct in Research with Aboriginal and Torres Strait Islander Peoples and Communities*.[89] While use of Indigenous governance models is increasing in research settings,[90] there remain substantial barriers to widespread uptake, including increased costs and time, research culture inertia, and a currently small pool of Aboriginal and Torres Strait Islander individuals trained in genomics. Funding levers are likely to be the most efficient mechanism for building Aboriginal and Torres Strait Islander capacity in this area and promoting change in research culture.

In the clinical sphere, any move towards Indigenous data governance would need to be part of broader changes to the governance of administrative data. In 2017, the National Advisory Group on Aboriginal and Torres Strait Islander Health Information and Data was disbanded, removing the key mechanism facilitating Aboriginal and Torres Strait Islander voices in health data discussions and leaving a significant gap in any national strategy for data governance.[91]

9.3.5 INTELLECTUAL PROPERTY

Proprietary rights may also provide a right to share in the profits generated by the use of tissue samples or data. It is well established that proprietary rights may reside in intangible assets. The most relevant examples for our purposes are intellectual property rights (IPRs) over creative and inventive works. IP gives a form of 'ownership' right if a party has invested intellectual effort in developing tangible assets or deriving genetic data from these tangible assets.[92] Relevant regimes include patents, trade secrecy or confidential information, and copyright. Among other things, IPRs can be used to exclude others from using works, and can be transferred by gift, sale and bequest.

86 Laura Arbour and Doris Cook, 'DNA on Loan: Issues to Consider When Carrying Out Genetic Research with Aboriginal Families and Communities' (2006) 9(3) *Community Genetics* 153 ('*DNA on Loan*').

87 Ray Lovett et al, 'Good Data Practices for Indigenous Data Sovereignty and Governance' in Angela Daly, S. Kate Devitt and Monique Mann (eds) *Good Data (Institute of Network Cultures, 2019)*.

88 See Tahu Kukutai and John Taylor (eds), *Indigenous Data Sovereignty: Toward an Agenda* (ANU Press, 2016); Lovett et al (n 87); Maggie Walter and Michele Suina, 'Indigenous Data, Indigenous Methodologies and Indigenous Data Sovereignty' (2019) 22 *International Journal of Social Research Methodology* 233.

89 National Health and Medical Research Council, *Ethical Conduct In Research with Aboriginal and Torres Strait Islander Peoples and Communities: Guidelines for Researchers and Stakeholders* (August 2018).

90 See, eg, the National Centre for Indigenous Genomics, *Home* (Web Page, 2020) <ncig.anu.edu.au>.

91 Lovett et al (n 87) 28.

92 McGuire et al (n 44) 65.

Intellectual property issues have arisen most prominently in case law around the patenting of gene sequences. Genomic information is a lucrative commodity, particularly when comprised in large datasets. And yet the commercial interests of developers of genomic technologies may be at odds with donors of genomic information.[93] Equally important, though, is the question of copyright and whether existing regimes provide adequate protection to genomic data, or whether some specific form of protection is necessary.

Patents are granted to reward innovation, and a patent may be granted for an invention that satisfies the statutory requirements for patentability.[94] Critically, a patent gives the patent holder a negative right, in the sense that it allows the patent holder to exclude others from exploiting the patented invention. The most well-known recent example concerning ownership of genetic data is the litigation brought to combat Myriad Genetics' enforcement of its patents over the BRCA DNA sequences. The Australian litigation, ultimately heard by the High Court of Australia, resulted in a pronouncement that the DNA sequence in question was in the nature of 'information' and therefore not patentable.[95] The meaning of 'information' in this context was clear, as the High Court stated that the information was 'discerned' rather than made by human action, and it remained the same regardless of whether it had been isolated or not.[96] The court's reasoning extended to human-made sequences containing the same informational content (cDNA).

What this means is that genomic information, without more, is unlikely to be patentable under Australian law.[97] It is possible that it will be protected as confidential information, or a trade secret. In line with international legal requirements, the equitable action of breach of confidence protects information that 'has the necessary quality of confidence about it', is 'imparted in circumstances that impose an obligation of confidence', and is used in an unauthorised manner.[98] Genetic or genomic data may fit these prescribed conditions where it has been obtained with consent to use in research, and a donor claims their data has been wrongfully released. Sharing of deidentified data is common in research settings. The risk of an action for breach of confidence highlights the importance of de-identifying and anonymising donor data.

Moreover, databases of patient data stand to be very commercially lucrative. For example, Myriad's extensive collection of patient sequence data (including mutations) collected during the period within which it had testing exclusivity for persons seeking genetic testing related to breast cancer susceptibility, is being protected as a highly confidential resource by Myriad.[99] Patient lists themselves have been held to be trade secrets under US case law.[100] Provided the requisite confidentiality is maintained, protection under breach of confidence should be available for the dataset as a whole. A number of individual patients requested access to their genetic sequence information contained in Myriad's database under the Health Insurance Portability and Accountability Act (HIPAA) Privacy Rule, for their own benefit but also to pass onto other researchers. When Myriad refused to grant access, those patients sought a determination from the US Department of Health and Human Services that the HIPAA had been violated.[101]

93 Bonython and Baer Arnold (n 14) 381.

94 *Patents Act 1990* (Cth).

95 *D'Arcy v Myriad* (2015) 258 CLR 334.

96 Ibid [6], [93]-[94].

97 The situation is similar in the US but differs markedly in the EU where DNA sequences are inherently patentable. The situation is similar in the US but differs markedly in the EU where DNA sequences appear to be inherently patentable; see Receptor Tyrosine Kinase, X ZR 141/13 (Bundesgerichtshhof 2016); Illumina, Inc v Premaitha Health Plc, EWHC 2930 (2017).

98 *Coco v AN Clark (Engineers) Ltd* [1969] RPC 41, 47.

99 See, eg, Jane Nielsen and Dianne Nicol, 'The Legal Vacuum Surrounding Access to Gene-based Research Materials and Data' (2016) 24 *Journal of Law and Medicine* 72.

100 Discussed in Sara Ghantous, 'Making the List: What Does it Take to Make a Patient List a Trade Secret?' (2018) *The John Marshall Review of Intellectual Property Law* 83.

101 Jasper Boverberg and Mara Almeida, 'Patents v Myriad or the GDPR Access Right v the EU Database Right' (2019) 27 *European Journal of Human Genetics* 211.

It is not clear whether these actions are yet to be resolved or have been privately settled,[102] but it does highlight the tensions between privacy obligations and the law of confidentiality.

Compilations of data may also be protected by copyright. Copyright is available for original literary and artistic works. While facts are not protected because they form the building blocks of data, in some cases data will be. Compilations of data where original expression has been exercised are one such instance. As part of its inquiry into gene patents, ALRC considered the IP protection of databases. The ALRC concluded that it did not see the need for specific database protection rights under Australian law, on the basis that copyright protects genetic databases.[103] However, since the ALRC completed its gene patent inquiry, there have been some significant judicial decisions that raise questions about the scope of any such copyright protections. This case law has cast doubt on the applicability of copyright to some computer-generated data compilations in Australia where intellectual input by a human author is minimal or non-existent.[104] As a consequence of these decisions, in Australia copyright is unlikely to subsist in collections of data that require no creative effort to compile.[105] The situation is similar in the US[106] but compilations seem to be capable of copyright protection in Canada in some circumstances.[107] Intriguingly, a recent Canadian case found that copyright may also subsist in individual level data where there is sufficient human authorship.[108] This is likely to be restricted to implied and derived data.[109] It has been suggested that a useful step may be to clarify the applicability of copyright to data at an international level.[110] Copyright may be present in some types of medical records (*Primary Health Care Ltd v Commissioner of Taxation* [2010] FCA 419), although not in simple test results. An amendment to the *Copyright Act 1968* (Cth) expressly excludes the collection, use or disclosure of health information in My Health Record from infringing copyright.

There has been no political appetite to enact a specific regime to protect databases in Australia. This contrasts with the situation in Europe where a *sui generis* database regime provides protection to creators of databases that lack the requisite degree of creativity required to attract copyright protection.[111] Having said this, the development of databases in other jurisdictions would not appear to have been unduly hindered due to a lack of a specific regime granting ownership rights over them. One of the major challenges in determining whether to protect compilations of data is in defining what level of human authorship is required in that a significant amount of data generation and arrangement is now automated. It is also not clear how the parameters of protection would be drawn; as we have seen there are difficulties inherent in defining 'data' and determining the rules around which such a regime would operate.[112] The introduction of any new regime protecting data or databases brings with it the risk that it will '... reduce flexibility and increase complexity.'[113] A perhaps more compelling case can be made for increasing emphasis on the custodianship and accompanying ethical obligations that accompany possession of data, rather than focusing on proprietary rights.[114]

102 Ibid.

103 Australian Law Reform Commission, *Genes and Ingenuity: Gene patenting and human health* (ALRC Report 99, 2004) 653-5.

104 *Ice TV Pty Ltd v Nine Network Australia Pty Ltd* (2009) 239 CLR 458; *Telstra Corporation Ltd v Phone Directories Pty Ltd* (2010) 194 FCR 142.

105 Ibid [33].

106 *Feist Publications Inc v Rural Telephone Service Co* 499 US 340, 111 S (Ct 1282, 1991) 348.

107 *Geophysical Service Inc v Encana Corp* [2016] ABQB 230. Leave to appeal denied [2017] CanLII 80435 (SCC) [115].

108 Ibid.

109 Scassa (n 47) 10.

110 Ibid 17.

111 *Directive 96/9/EC of the European Parliament and the Council of 11 March 1996 on the Legal Protection of Databases* [1996] OJ L 77/20, Art 11(3).

112 Scassa (n 47) 16-7.

113 Ibid 17.

114 Andanda (n 82) 1073-5.

9.3.6 MAINTAINING RECORDS OF PROVENANCE AND CHAINS OF CUSTODY

As part of considering questions of ownership, important factors in any transfer of tissue or data include those of provenance. The final issue this chapter touches on is whether proprietary rights are necessary to ensure the maintenance of provenance and recording a clear chain of custody for tissue samples and data.

Even if proprietary rights are denied to donors, researchers and/or compilers of data, ethical obligations inherent in custodianship paradigms demand that efforts are made to act within the boundaries of consent, reduce risk to donors, and ensure benefit sharing is undertaken to the best of researchers' abilities.[115] Acknowledgement of the work of researchers and database administrators in interpreting and compiling data is also an important factor.

An important consideration in achieving research aims is maintaining records of the movement and use of biological materials and genomic data. Material transfer agreements are now in common usage in the research landscape; data transfer agreements are becoming increasingly commonplace.[116] It is only once accurate records of transfers are kept that custodianship obligations to donors can be consistently and adequately met by researchers.

115 See generally, Andanda (n 82).

116 Nielsen et al (n 16)

10 Summary of Findings and Challenges for the Future

10.1 Summary of Findings

This chapter begins with a summary of the key findings for each of the themes presented in chapters 2 to 8 of this document. Each summary follows a consistent format, addressing key aspects of current regulation, risks of the current approach and opportunities for strengthening current arrangements.

10.1.1 CONSENT

How is consent for genomic testing regulated?

- Consent to clinical genomic testing is regulated through the tort law of negligence and professional disciplinary standards administered through the Australian Health Practitioner Regulation Agency, national health practitioner boards, and relevant state and territory ombudsmen.
- In the research context, an additional layer of oversight is provided through requirements for a Human Research Ethics Committee ('HREC') to approve the consent process and forms. HRECs also may authorise the secondary use of genomic samples and data collected with broad consent, and—in some circumstances—waive the requirement for prior consent altogether.

What are the risks of the current approach?

- Key risks relate to the secondary use of data or samples for research based on broad consent, or a waiver of consent. The individuals to whom those samples and data relate may be harmed if secondary uses go beyond the individual's reasonable expectations or, for other reasons, are morally troubling to those individuals.
- The link between deidentification of data and unspecified consent and waivers of consent is likely to be tested by the growing capacity to re-identify genomic information.
- Consent arrangements are predominantly once-off and individual, which may not reflect uses of genomic information over time or the familial and communal nature of genomic information.
- The ethical standards for clinical genomic practice lack the specificity and oversight that apply in genomic research, raising risks for patients and practitioners.

What opportunities are there for strengthening arrangements?

- The Commonwealth Government may consider reviewing the governance, accountability, and expertise of HRECs, including consideration of establishing specialised HRECs to review genomic related research applications, including authorization of waivers of consent. HRECs provide the key mechanism for authorising secondary and unconsented uses of genomic information. HRECs are predominantly established by individual institutions with limited centralised oversight or accountability. There is no specific requirement for HRECs to include members with expertise in genomics.
- The Commonwealth Government may consider reviewing the requirements for a waiver of consent for genomic research to ensure clarity and consistency with public expectations.
- The Commonwealth Government may consider reviewing the protections and oversight processes that apply in the clinical genomics context to ensure their adequacy to uphold ethical standards.

10.1.2 PRIVACY

How is privacy regulated?

- Privacy and data sharing in Australia are regulated through a complex patchwork of law, at Commonwealth, State and Territory level, with different laws applying depending on the type of agency or organisation and whether personal information is health information. The table below lists some key legislation relevant to genomic information but it is not a comprehensive list.[1]

Jurisdiction	Use or Disclosure of Health Data
Cth	*Privacy Act 1988* (Cth)
NSW	*Health Records and Information Privacy Act 2002* (NSW)
VIC	*Health Records Act 2001* (Vic)
QLD	*Information Privacy Act 2009* (Qld); *Hospital and Health Boards Act 2011* (Qld)
WA	*State Records Act 2000* (WA); *Health Services Act 2016* (WA)
SA	*Health Care Act 2008* (SA); Government of South Australia PC012- *Information Privacy Principles* (IPPS) *Instruction* (6 Feb 2017)
TAS	*Personal Information Protection Act 2004* (Tas)
ACT	*Health Records (Privacy and Access) Act 1997* (ACT)
NT	*Information Act 2002* (NT)

- Genomic information is within scope of privacy laws (generally, with some exceptions) if the information is "about" an individual who is 'reasonably identifiable'. Genomic information will typically be categorised as sensitive information and thus subject to additional controls.

What are the risks of the current approach?

- Risks relate to inconsistent understanding of what it means for data to be 'about' an individual and for an individual to be 'reasonably identifiable'.
- Rules permitting disclosure to third parties operate with different thresholds and this may result in confusion and inhibit appropriate disclosure to at-risk family members. Complexity is extended by the fact that some families live across multiple states.
- The significance of genomic data relating to groups is insufficiently appreciated by current legal approaches to privacy protection more generally.

What are the opportunities for strengthening arrangements?

- Governments may wish to consider ways to increase clarity and consistency in all jurisdictions in relation to:
  1. when different types of genomic information (and samples) are 'personal information': importantly, how 'reasonably identifiable' is to be understood in practice. For example, the extent to which account should be taken of all objective factors, such as the costs of and the amount of time required for identification, the available technology at the time of the processing, and the possibility of technological developments
  2. the status of genomic information as sensitive personal information
  3. rules relating to familial disclosure, including to at-risk relatives, and in relation to the deceased
  4. the relationship between common law duties of confidentiality and privacy legislation.
- Governments may further wish to explore the extent to which protections extend to individuals as members of groups and beyond a narrow view of individual privacy.

1 For example, particular confidentiality provisions may apply if data is gathered by a private health facility or through public health powers. These were not specifically considered as part of this report.

10.1.3 ABORIGINAL AND TORRES STRAIT ISLANDER IDENTITY AND GENOMICS

How are Aboriginal and Torres Strait Islander genomic samples and data regulated?

- **Clinical:** regulated in the same way as for non-Indigenous samples and data; ie primarily mediated by consent, with additional protections through the various privacy regimes and human tissue Acts. Local policies regarding the delivery of health services to Aboriginal and Torres Strait Islander people may also apply.
- **Research:** largely the same as for non-Indigenous participants, with key protections derived from consent and privacy legislation, with a pivotal role played by HRECs in approving research protocols that comply with the *National Statement*. There are additional requirements from guidelines specifically relating to research with Aboriginal and Torres Strait Islander people, but application of these depends upon the composition and expertise of the HREC reviewing the application.

What are the risks with the current approach?

- Current regulatory frameworks are ill-equipped to address concerns relating to group interests or equity, and risk further entrenching existing disparities.
- Failing to address the concerns of Aboriginal and Torres Strait Islander people is likely to lead to reduced participation in research and clinical activities, and may contribute to harms from inappropriate practices.
- Heavy reliance on a fragmented and under-resourced HREC system leads to duplicative review, inconsistencies and significant delays. Few HRECs possess expertise in the issues associated with Aboriginal and Torres Strait Islander genomics, leading to ineffective review.

What are the opportunities for strengthening arrangements?

- Commonwealth, State and Territory governments may wish to consider national efforts to improve Aboriginal and Torres Strait Islander representation in genomic datasets, to merge clinical and research genomics, or to create a national repository, will require prioritisation of Aboriginal and Torres Strait Islander leadership and governance for the collection, use and disclosure of Aboriginal and Torres Strait Islander samples and data.
- A review of the ethical review system in Australia may be beneficial, including assessing a role for a centralised review body, comprising diverse Aboriginal and Torres Strait Islander representation and with genomics expertise, particularly for multisite or complex projects.
- Capacity building represents a significant supportive strategy, and consideration could be given to strategies for strengthening Aboriginal and Torres Strait Islander involvement in genomics, bioinformatics, and STEM more generally. For example, creation of an Aboriginal and Torres Strait Islander scientists' association may provide capacity building, networking and advocacy opportunities.

10.1.4 GENETIC DISCRIMINATION

What regulation is in place to prevent genetic discrimination?

- There is general protection under Commonwealth and States and Territory anti-discrimination legislation against disability discrimination in employment which in most cases [not NT and SA] provides protection from genetic discrimination. While more explicit regulation would be beneficial, particularly for educative reasons, this area is not a concern currently.
- The main contentious area for genetic discrimination is life insurance which is currently regulated by antidiscrimination legislation which provides an exemption for life insurers. While insurers retain the right to use genetic test results in underwriting life insurance, the peak body, the Financial Services Council, has introduced a moratorium on the use of genetic test results for life insurance products up to specified financial limits.

What are the risks with the current approach?

- The moratorium applying to life insurers is self-regulated by the industry and has relatively low financial limits (compared internationally). Individuals remain at risk of genetic discrimination for applications for insurance above those financial thresholds and there is uncertainty about whether the moratorium will be extended beyond 2024.
- Public concern about genetic discrimination carries with it the risk of deterrence for uptake of genetic/ genomic testing and participation in genomic research which presents an obstacle to the mainstreaming of genomics.

What are the opportunities for strengthening arrangements?

- One option to protect against risk of and concerns about genetic discrimination is to strengthen the current insurance moratorium on the use of genetic test results by removal of the financial thresholds. It could also be made legally binding rather than self-regulated.
- Further, following the UK model, there could be government partnership in and/or oversight of the moratorium to ensure accountability and enhance public trust in the arrangements.
- An alternative more protective, but also more interventionist, option might be the introduction of legislation banning the use of genetic tests in life insurance underwriting. This would address concerns about genetic discrimination in life insurance and the deterrent effect that genetic discrimination has on uptake of recommended genetic testing and participation in genomic research. It is acknowledged that legislation is a fairly inflexible response but this could be addressed with the inclusion of a sunset clause to allow for review after a specified period of time (e.g. 5 years).

10.1.5 SOME CONTROVERSIAL SECONDARY USES – LAW ENFORCEMENT

How is access by law enforcement regulated?

- Genomic information, like other health information, can be disclosed to law enforcement agencies without the knowledge or consent of the individual, if the entity that holds the data (whether public or private) considers it reasonably necessary for law enforcement activity. No court order is needed.
- Information in My Health Record is an exception to this; a higher standard applies.
- Other bases for disclosure are individual consent, court order, or disclosure to reduce a risk to life, health, safety.

What are the risks with the current approach?

- The possibility of disclosure to law enforcement agencies at the discretion of the entity that holds the data, and without notification to or consent by the individual affected, presents a risk to individual and community confidence in the privacy of their genomic information.
- There may be insufficient protection of data and transparency around disclosure to law enforcement agencies.

What are the opportunities for strengthening arrangements?

- The amendments to the *My Health Records Act 2012* (Cth) in 2018 raised the bar for the protection of personal health information against law enforcement access, removing the possibility of disclosure where the entity that holds the data considered it reasonably necessary for law enforcement purposes.
- Extending this approach through amendments to the *Privacy Act 1988* (Cth) and relevant state-based laws would help to achieve a consistent, high level of protection of personal health information limiting unconsented disclosure to enforcement agencies.

10.1.6 SOME CONTROVERSIAL SECONDARY USES – COMMERCIAL USE

How is access for commercial use regulated?

- Genomic information can be used by commercial entities with the individual's consent. This consent might relate to specific commercial activities or be given broadly for unknown future use by commercial entities.
- Use of genomic information arising from consent that was based on inadequate information (i.e. not properly informed consent) may be a breach of the Australian Consumer Law misleading or deceptive conduct provisions.
- The secondary use of genomic information in My Health Record is an exception to this; a higher standard applies and use for solely commercial purposes is prohibited. Use for partly commercial purposes is not excluded.
- Disclosure of identifiable genomic information for research purposes (including commercial use) may occur without consent where consent is impracticable, the research is low risk and an HREC has approved it.

What are the risks with the current approach?

- Broad consent for unknown future research undermines transparency and individuals' control over their genetic data.
- The risks must be weighed against the administrative burden of specific re-consent for commercial uses if that burden inappropriately impedes beneficial commercial developments.
- Concern about inadequately informed consent (or a lack of consent) for commercial use could lead to a public backlash against certain companies.

What are the opportunities for strengthening arrangements?

- The existing misleading or deceptive conduct provisions in the Australian Consumer Law appear to provide some protection against inappropriate commercial use.
- A dynamic consent or similar approach could mitigate the risk of inadequate transparency and provide additional control and decisional autonomy to individual data subjects.

10.1.7 RETURN OF FINDINGS

How is return of findings regulated?

- Clinically, genomic analysis and return of findings are primarily regulated through the National Pathology Accreditation Advisory Council best practice standards and National Association of Testing Authorities accreditation.
- In research, the *National Statement* is the key source of regulatory requirements.
- Privacy laws require that individuals are provided with access to their personal information, which would include identifiable genomic information.
- The extent to which tort law may provide a legal duty to analyse genomic data and return findings, in both the clinical and research contexts, is not yet clear.

What are the risks with the current approach?

- Individual clinical geneticists and researchers bear the primary burden of deciding whether to undertake genomic analysis for incidental findings and re-analysis of findings of current unknown significance and whether to return findings to individuals. There is some oversight from HRECs in the research context.
- This is likely to result in inconsistency in approaches to return of findings nationally.
- Lack of clarity about the extent of the obligation to return findings carries with it the dual risks of underdiagnosis and overdiagnosis.

What opportunities are there to strengthen arrangements?

- National discussions could clarify the extent of the legal obligation to undertake genomic analysis (including analysis of incidental findings and re-analysis of findings of unknown significance) and return findings to individuals, in both clinical and research contexts.
- Strategies could be put in place for streamlining processes for return of clinical and research findings.
- There could be greater consistency in approaches to return of research findings by researchers and HRECs nationally. Further guidance and training may assist HRECs and researchers in understanding the scope of their ethical and legal obligations with regard to genomic analysis and return of findings, and how best to meet these obligations.
- A regulatory requirement for all Australian laboratories providing genetic testing services to be NATA accredited/comply with NPAAC standards could be considered.

10.1.8 GENETIC TESTS AND GENETIC TESTING

How are genetic tests and genetic testing regulated?

- Genetic tests with a therapeutic purpose are regulated primarily through the in vitro diagnostics medical devices pathway of the Theraueptic Goods Administration regulatory scheme ('TGA IVD medical devices scheme'). Tests that attract a Medicare benefit have additional requirements of NATA accreditation and compliance with NPAAC standards.
- The National Pathology Accreditaiton Advisory Council and National Association of Testing Authorities provide best practice guidance on the requirements for Australian clinical genetic testing laboratories.
- The TGA IVD medical devices scheme appears appropriate for regulating commercial kits and laboratory-based clinical genetic tests for therapeutic purposes.

What are the risks with the current approach?

- Provision of direct-to-consumer ('DTC') genetic testing for therapeutic purposes within Australia remains prohibited under the *Therapeutic Goods (Medical Devices-Excluded Purposes) Specification 2020*. However, provision of non-therapeutic DTC genetic tests is not regulated through the TGA IVD medical devices scheme.
- Likewise, provision of DTC genetic tests for therapeutic and non-therapeutic purposes outside Australia is not regulated through the TGA IVD medical devices scheme. However, it may be regulated by the national regulatory scheme in the jurisdiction where the tests are performed.
- The consequence of unregulated provision of genetic tests and DTC genetic tests, whether for therapeutic or for non-therapeutic purposes, are unknown.

What are the opportunities to strengthen arrangements?

- The TGA IVD medical devices scheme and the prohibition on the provision of DTC genetic tests for therapeutic purposes are not currently in need of strengthening.
- Given the unknown risks associated with unregulated provision of genetic tests for non-therapeutic purposes, and DTC genetic tests for any purposes outside Australia, community awareness of the benefits and risks of DTC genetic tests needs to be raised.
- Compliance of DTC genetic testing with *Australian Consumer Law* could be examined by the ACCC.

10.1.9 TISSUE SAMPLES AND DATA

How are human tissue samples and data regulated?

- The storage, security, transfer and disposal of human tissue samples and data are governed by a complex, overlapping and sometimes inconsistent mix of legislation, guidelines and policies. In particular, storage of tissue and data is covered to varying degrees by the human tissue Acts, information and privacy legislation, accreditation standards and guidelines covering pathology laboratories and health authorities, and ethical guidelines and policy statements.
- Ownership of tissue samples that are provided for clinical or research use is not clearly defined. The existence of ownership interests in data generated from tissue samples is even less clearly articulated. It can be said that a party who has exercised work and skill on a tissue sample may have a stronger ownership interest than any other party (including a donor), as may a creator of data records. Although people make intuitive assumptions about rights of ownership and control over their samples and data, the legal position is complicated and doesn't necessarily provide the control that an individual may assume exists.

What are the risks with the current approach?

- The problems previously identified in relation to storage, security, transfer and disposal of tissue samples and data will continue.
- Uncertainty around ownership and possession are likely to persist in the absence of any action to help clarify. Competing possessory interests are likely to be claimed by multiple parties, and donors of tissue samples and data are likely to continue to feel some degree of dissatisfaction and exclusion. It is likely that no single party will have an exclusive possessory interest.

What are the opportunities for strengthening arrangements?

- In relation to storage, security, transfer and disposal of tissue samples and data, the recommendations made in *Essentially Yours* could usefully be revisited in order to examine the consistency of current legislative and policy regimes.
- Alternative methods of vesting control over access to tissue samples and data in certain parties exist. In some ways these are as effective as rights of ownership. The alternative control mechanisms canvassed here are potentially effective in ensuring donors retain some degree of control: shifting the debate to control rather than ownership and clarifying how control may be exercised is therefore an important educative exercise.

10.2 Challenges for the Future

A recurrent theme present throughout the preceding analysis is that the current regulatory environment is predicated on dichotomies that are increasingly unsupportable: consent versus non-consent, identifiable versus deidentified data, public versus private, and clinical versus research contexts. The law provides some of the frameworks for the use of genomic information but as issues have emerged, more soft law mechanisms have been used, such as the moratorium for insurance (Chapter 5) and the HRECs that oversee research (Chapter 2).

10.2.1 CONSENT VERSUS NON-CONSENT

Our liberal democracies rely on the principle of autonomy and the individual's right to make decisions and to consent to certain activities. Since the publication of the *Nuremberg Code*, consent has been viewed as the lynchpin for protection of patients and participants. As the activities for which consent is sought increase in complexity and scope, the ability for consent to act as a protection diminishes. A number of chapters of this report highlight the complexity of current genomic activity. Once DNA is sequenced from a sample and transformed into a digital form of AGTC base pairs, the potential scope of its use increases. Once derived, genomic information has a wide range of uses, from research to clinical care, to crime detection and insurance uses. How can and should people exercise control over these future uses of their samples and data, particularly given the social benefits of many of these uses?

The traditional focus of the law on individuals obscures the relatedness of people and the mutual interests they may have through their shared DNA. Under the current regulatory frameworks for research, an individual is able to consent to the use of their DNA without seeking the approval of others or the consideration of potential group harms, such as discrimination. Decision-making processes around the collection, use and disclosure of genomic data in research rely on the two key mechanisms of individual consent and ethics review processes, which do not easily accommodate group interests. Genetic privacy protection has relied on consent and anonymisation as a way to alleviate privacy concerns. This does not address the concerns raised by genomic testing in all the areas mentioned above.

Secondary uses of genomic data raise further questions about the limits of consent. As one example, crime prevention and detection are given public interest status in most societies. The role of the state in most societies is considered to be to protect its citizens and so consent is not always a requirement when a DNA sample is taken and analysed. As there is significant interference with civil liberties, these powers are provided for under legislation in Australia. The *Privacy Act 1988* (Cth) and equivalent state and territory acts all have provisions allowing disclosure of information to law enforcement agencies without the knowledge or consent of the individual concerned.

We think of consent as a protection but, in practice, it operates as a means of respect for autonomy, with protections operating via other governance mechanisms. This shows the need for integrated and holistic approaches to reform.

10.2.2 IDENTIFIABLE VERSUS DEIDENTIFIED DATA

Part of the social contract in research is keeping information confidential as a way of respecting individual privacy. Best practice has involved the use of information technology mechanisms such as firewalls, encryption, passwords, and security compliance as well as the restriction of access to raw data that could directly identify an individual. Current research practice requires protections for samples and data such as anonymisation, deidentification of samples and data through the removal of personal identifiers or aggregation techniques. These approaches ignore the inherently identifiable nature of genomic information.[2]

The uniquely identifiable nature of human genetic information means that it is very difficult to guarantee non-identifiability for individuals or their genetically related family members. Exome sequencing reveals rare alleles, including those that are of clinical or personal utility, which increases the risk of identity disclosure as well as breaches of confidentiality or privacy. In 2008, Homer et al demonstrated that it is possible to re- identify individuals who had been genotyped and even those in pooled mixtures of DNA, provided a reference sample is available.[3] Once a person is re-identified, there is the potential for further personal information to be revealed about the formerly anonymous source.

2 Melissa Gymrek et al, 'Identifying Personal Genomes by Surname Inference' (2013) 339(6117) *Science* 321.

3 Nils Homer et al, 'Resolving Individuals Contributing Trace Amounts of DNA to Highly Complex Mixtures Using High-Density SNP Genotyping Microarrays' (2008) 4(8) *PLoS Genetics* 1.

The standard protections, such as encryption, that have been used to anonymise or deidentify sequence information are challenged with new sequencing technology that produces richer and more detailed information on individuals. This information is increasingly powerful for a range of uses when it is linked to clinical, ancestral, public and population-level information, but this raises a number of ethical and legal issues.

As well as being an indicator of disease, DNA also can show an individual's relation to other family members and be an indicator of ancestry. The familial nature of genomic information raises ethical issues of disclosure to other at-risk family members and how to protect individual privacy. It also raises concerns for research in indigenous communities and the need for culturally appropriate protocols that can address individual concerns but also collective interests around the use of samples and information.

Many of the regulatory protections that have been discussed in preceding chapters – especially those in Chapter 3 (Privacy Law) – rely on a distinction between identified and deidentified that is increasingly divorced from genomic reality. Researchers are exploring new techniques for minimising the potential for reidentification of genomic information. If these strategies are not able to keep pace with developments in reidentification, then it may be necessary to consider treating genomic information as personal information at all times for the purposes of privacy law.

10.2.3 CLINICAL VERSUS RESEARCH CONTEXT

The genesis of the development and clarification of the distinction between human research and clinical investigation and uses can be traced back to the ten principles for the ethical conduct of human research stated in the post-war *Nuremberg Code*. These principles were considered by the World Medical Association and reformulated as the ethical and regulatory principles for the conduct of human research in the *Declaration of Helsinki* 1965. As a response to the *Declaration of Helsinki*, in Australia, the National Health and Medical Research Council (NHMRC) issued the *Statement on Human Experimentation* in 1966, which expressly drew on the Declaration.

Conceptually, in Australia, a clear line appears to be drawn between two mutually exclusive sets of activities, human research and clinical interventions, with the two clearly delineated. This is particularly evident through the requirement for deliberation and approval by a Human Research Ethics Committee for research, whereas clinical interventions require no such formal oversight.

It is increasingly difficult to draw such a clear research-practice delineation, with particular challenges in the genomics space. This is especially true for the use of genomics for diagnosis and treatment at the edge of our knowledge, which is simultaneously clinical and research. This is likely to become increasingly apparent as precision medicine expands to become commonplace.

How, then, should we approach regulation, given that current pathways require a clear choice between clinical and research when a given activity may fairly encompass both? The two regulatory pathways apply different standards, and so the consequences of the choice for patient-participants may be significant. Specific groups, including Aboriginal and Torres Strait Islander people, may be at particular risk under this unclear and unsupported approach.

10.2.4 STRENGTHENING CURRENT ARRANGEMENTS

Genomic samples and data have been dealt with through patchworks of existing regulatory frameworks. As the technologies have advanced, the inability of these frameworks to keep pace has become increasingly obvious, leading to the inappropriate dichotomies described here. These features fail to recognise core attributes of genomic information: that it is familial in nature, that it is inherently identifiable, and that genomic technologies are dissolving traditional boundaries between clinical and research contexts. The consequence of this is that the genomics regulatory environment is increasingly unfit for purpose.

Developing a system for the effective regulation of health-related genomic information into the future is therefore a key challenge for achieving the benefits of genomic medicine.

APPENDIX ONE

Assessment of legislation and regulations applying to the collection and use of health-related genomic information

SCOPING REVIEW

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13 MARCH 2020

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Introduction

This scoping review aims to identify current and emerging ethical, legal and social issues of concern to the community in the context of health genomics information. Issues have been identified through a national and international literature review, combined with the results of a national consultation process, involving in-depth interviews with a diverse range of stakeholders. These issues are presented here for the Customer to select those that will form the basis of our assessment of the relevant legislation and regulations.

Genomics is a rapidly advancing field, and applications are being developed for use in many sectors of society. Human genomic information can be uniquely identifying and indicative of our relationships with others at all levels: families, communities and populations. In this way, genomic information presents challenges to our traditional safeguards. The collection, use and disclosure of personal and sensitive information has long been regulated through an approach characterised by 'ask or anonymise'; that is, by asking permission through informed consent processes or, when consent was not feasible, by anonymising (or 'de-identifying') data to prevent harm. Yet, genomic information, in common with other datasets with large numbers of data points per individual, cannot be effectively anonymised, the data and its significance can be complex and require technical understanding, future uses can be uncertain, and the familial nature of genomics means that it is not always clear whose consent should be sought.

The collection, use and disclosure of genomic information is necessary to realise the substantial benefits promised by genomic medicine to individuals and society. However, these activities raise a range of community concerns, including the use of information for insurance purposes and commercial activities generated through genomic data. Many of these issues are interrelated and overlapping, and categorising them is, in many ways, reliant on value judgments. The categorisation of concerns in this report has been driven by the thematic analysis of the stakeholder interviews, in an effort to reflect the way genomics is understood by the community.

Aim

In consultation with the Department of Health, we agreed on the following research question:

> *What are the current and emerging issues in relation to the collection and use of genetic and genomic information in the clinical and research settings?*

Methods

RAPID REVIEW METHOD

In February and March 2020, a search was conducted in HeinOnline, JSTOR and Pubmed with the following search terms:

1. (genetic or genomic) and (law or ethics or legal) and (data or information) and Australia
2. genetic testing and (ethics or law) and Australia
3. Australia and (genetic* or genomic*) and (use* or disc*) and (law* or legal or governance or ethic*)

One hundred and eight articles were selected as relevant to the agreed research question and satisfying the criteria of high-quality and/or frequently cited. We selected articles for inclusion in this report based on the following priorities:

- Australian authors (including when part of collaborations) and papers that directly assessed the Australian experience;
- Peer reviewed journal articles rather than comments;
- Articles with a legal or ethical focus;
- More recent articles (post-2015).

These articles were downloaded into Zotero and relevant information imported into the literature review template. Additional, more specific searches were conducted for issues raised in stakeholder consultations, but not adequately addressed by the original list of sources.

The search results were supplemented by researchers' own knowledge of important articles across the relevant themes. Our goal was to pragmatically identify an adequate sample of literature—in particular, research published within the preceding five years—to ascertain prevalent issues in relation to the collection and use of genetic and genomic information in clinical and research settings.

STAKEHOLDER CONSULTATION METHOD

Interviews with key stakeholders across Australia were conducted by telephone, internet-based conferencing (eg. Zoom) or face to face, between 7 February and 10 March 2020. Potential interview participants were all adults and were approached due to having an interest in the collection and use of health-related genomics information, e.g. clinical geneticists, genetic counsellors, health researchers using genomic data, data custodians responsible for genomic data repositories, chairs of genomic data access committees, representatives of patient groups. An important sub-cohort was Aboriginal and Torres Strait Islander stakeholders with expertise relating to aspects of the research question. This sub-cohort included people with expertise in community engagement, genomics research, clinical care (as both health service providers and patients), data sovereignty and health policy.

Participants were recruited based on the existing connections held by the project researchers, as a starting point. Potential participants were approached via a standard email. We also invited them to suggest additional potential interviewees based on their awareness of others holding relevant roles. We approached these new potential interviewees directly using contact details available in the public domain.

Participant characteristics are summarised in Table 1. We aimed to recruit for diversity in roles, gender and geographical location. Participant roles are not included in the table, as resulting small cell counts made individuals potentially identifiable. Aboriginal and Torres Strait Islander stakeholders were purposively recruited for an enriched sample, given both their unique status within the Australian community and the additional risks presented to Indigenous communities by the misuse of genomic samples and information. In addition to the eight Aboriginal and/or Torres Strait Islander participants, three non-Indigenous stakeholders with particular experience in working with Aboriginal and/or Torres Strait Islander patients, participants and communities were also interviewed.

TABLE 1: GENDER AND JURISDICTION OF STAKEHOLDERS FOR INTERVIEW

PARTICIPANT CHARACTERISTICS	NON-INDIGENOUS	ABORIGINAL AND/OR TORRES STRAIT ISLANDER
Invited	38	18
Accepted	25	8
Female/Male	15/10	3/5
ACT	1	1
NSW	4	0
NT	1	2
QLD	1	2
SA	2	1
TAS	1	1
VIC	12	↔
WA	3	↨

We conducted semi-structured interviews to identify what participants saw as: key issues in the collection and use of health-related genomic information in clinical and research settings; important legal and regulatory factors; and new issues likely to emerge in the future. All interviews were recorded and contemporaneous notes prepared on structured forms. Notes were then analysed using both a deductive and inductive approach, mapping participant responses against expected themes, as well as identifying and grouping responses that generated new themes. Representative quotations for each of the identified issues were extracted for the report. All resultant issues, both from the literature and the consultation process, were grouped by theme and then ordered into five broad categories:

1 issues relating to the regulatory environment;

2 issues arising within the healthcare system;

3 issues arising at the overlap between clinical care and research;

4 issues relating to research; and

5 issues arising from other uses of genomic information.

These broad categories form the basis of this report.

1 Regulatory environment

1.1 Individual

1.1.1 CONSENT

Consent provides the legal basis for genomic testing in clinical and research contexts. However, its inherent complexities and uncertainties challenge traditional regulatory constructs. Commentators raise questions about the extent to which patients (Meiser et al, 2015) and parents of critically ill children (Wilkinson et al, 2016) undergoing genomic testing understand the information with which they have been presented. Model consent clauses have been developed in some contexts to help to meet patients' informational needs. (Nguyen et al, 2019)

Consent to predictive genomic testing has traditionally been limited in certain respects, such as literature and guidelines cautioning against testing minors for adult-onset genetic conditions. There is ongoing uncertainty in this domain, but evidence to suggest adoption of more flexible approaches in clinical practice and guidance. (Mand et al, 2012) This is supported by empirical assessment of adolescent experiences of predictive-genetic testing for adult-onset conditions, which reported a range of benefits and a relative absence of harms. (Mand et al, 2013)

While initial testing will always require consent, there is an extensive literature on the applicability of consent paradigms to research involving large-scale, long-term biobanks and biorepositories. Many commentators now accept that obtaining specific consent from each participant for each associated project is not feasible. (Otlowski, 2012) Models of broad (Grady et al, 2015) and dynamic consent (Kaye et al, 2014) have been developed in efforts to balance participant autonomy with modern research landscapes. There is concern however that dynamic consent may not be fit for all purposes and contexts. (Prictor et al, 2020)

There remains no criminal offence for non-consensual genetic testing in Australia, despite calls for such an offence due to the sensitive and personal nature of genetic information, and the potential for harm stemming from non-consensual genetic testing. (Otlowski, 2013)

Stakeholder consultation

Most stakeholders commented on aspects of consent for the collection and use of health-related genomic information.

Consent is important

Consent was foregrounded as a crucial but challenging process.

> *"It is not paternalistic [to have] requirements [for] how you let people know exactly what will happen to their data"* [Participant 3].

> *"[In our organisation] a person comes into a clinic and has a trained person go through a consent process to understand what information will come out of these [tests] and potential risks of [the return of genomic results] so I am pretty happy with our processes, we do our best to cover everything"* [Participant 37].

One participant noted that consent was an enabler to data sharing for research purposes:

> *"The importance of informed consent can't be emphasised enough because particularly in the context of research, if you do and have recorded [the] consent discussion then the jurisdictional impediments to sharing data [are somewhat overridden] in the context of the individual consent"* [Participant 18].

Consent is complicated

> *"[Consent] is really complicated and doesn't make it easy so a lot of research groups don't actually venture into this space because it can break you very easily and very quickly and you get discouraged"* [Participant 5].

> *"Consent here [in NSW] is so bound up in law that it is not practical"* [Participant 9].

Aspects of consent processes that were noted as challenging included:

- diverse consent forms and processes in line with the requirements of different ethics committees at different institutions [Participant 25];
- finding a balance between informing people thoroughly, and overburdening them with information: *"The more complex [consent] gets, the longer the form is, the less likely people will read and be informed"* [Participant 34];
- actioning consent withdrawal when samples or data have been shared with third parties [Participant 19];
- understanding the scope of broad consent and when notification and re-consent for further uses is needed: *"if you then go to use [the data] for a different purpose, do you have to notify people?...How does that fit in the law?"* [Participant 19];
- understanding the scope of consent and re-consent requirements when the data subject has died: *"How is consent handled for people who are no longer around?"* [Participant 12].

Is consent truly informed?

Several participants questioned whether data subjects were truly informed, even when they were presented with extensive information about the use of their genomic information.

Giving additional information was seen as impeding participant understanding:

> *"I find it frustrating with patients where I am trying to give the potted summary and I am saying 'now the 1 page form I have given has turned into the 10 page form' and this is becoming like the credit card [terms and conditions] situation"* [Participant 25].

> *"This idea that people agree and it is informed consent even though it is buried in 40 pages of small print is not good enough"* [Participant 3].

> *"If you give a person a 12 page consent form, they don't read it"* [Participant 9].

Meaningful informed consent was noted by some stakeholders to be a particular challenge for people who engaged in 'direct-to-consumer' genetic testing [Participant 37], and who were culturally and linguistically diverse or economically/educationally disadvantaged: *"we still expect everyone to be able to read and write which is wrong"* [Participant 38].

Many of the Indigenous stakeholders recognised the limitations of the traditional model of consent in meeting the needs of Aboriginal and Torres Strait Islander people, particularly those in remote settings. For research participants for whom English was not a primary language, or where efforts to communicate genomic information in culturally appropriate ways had not been made, stakeholders felt that informed consent was an insufficient safeguard, and one that relied on the integrity of the person enrolling participants.

Participant 43 noted that *"If I felt that a lot of people didn't understand then I would ask them to say no. We erred on the side of caution."* Further, Participant 44 reiterated the concern that consent forms contained too much information: *"Each time we do a consent form it has gotten longer. Are we taking it too far? Do people even care with some of it?"*

Participants 42 and 51 both noted that consent for research was undermined when it was attached to clinical care, as participants might *"sign anything to get the care they need"* or were likely to be preoccupied with the illness that had led them to seek healthcare.

Problems with paper-based consent

Paper-based consent forms were noted by Participant 11 as no longer fit for purpose, because the information on the forms is not machine-readable: *"a lot of laboratories scan the paper and put it into their laboratory system...then what happens is you then don't have that consent information in what we call an 'atomic form'... it is not computer-readable."* Progress in this arena could enable the semi-automated authorisation and authentication of researchers and systems to access genomic data.

Dynamic consent

Dynamic consent is a model for ongoing engagement with participants using an interactive IT interface to facilitate greater participant engagement in clinical and research activities, enabling 'participants to consent to new projects or to alter their consent choices in real time as their circumstances change' (Kaye et al, 2015). Several participants referred positively to a dynamic consent approach that provides participants with control over the uses of their genomic information over an extended period of time.

> *"You build a relationship, not 'give me your DNA and we forget about you"*, it is *"transparent, there is engagement, [participants] can ask questions"* [Participant 5].

> *"[With] greater volumes of genomic information, we need a much better and central way as to how to manage and re-use that information in the future that involves the consent of the people who gave it in the first place"* [Participant 9].

A number of participants referred positively to the CTRL dynamic consent tool developed by Australian Genomics Health Alliance (see https://www.australiangenomics.org.au/resources/for-patients/your-personal-platform/), or expressed that they were using similar tools.

> *"We are developing an app where people can go in and change their consent preferences"* [Participant 9].

> *"We have developed a lot of tools around dynamic consent...it is not a one-off decision when talking about your personal information"* [Participant 40].

However not everyone thought that dynamic consent was a useful development. Participant 11 commented that: *"Consent is hard enough at the time of testing... do we have time to go back and do all of that? What is the value in it?"* [Participant 11].

Indigenous stakeholders recognised the difficulties associated with secondary uses of research or clinical data, indicating that broad consent was generally not acceptable, and waivers were inappropriate given the lack of trust, but that dynamic consent was regarded with some suspicion. For instance: *"Yeah there is lots of talk about dynamic consent, but it is not something people I have spoken to are interested in"* [Participant 41].

Participant 51 commented that *"The idea of dynamic consent is a deep concern"* for Aboriginal and Torres Strait Islander participants, particularly when undertaken using apps or other remote technologies, because it assumes a level of genomic literacy that may not exist.

1.1.2 PRIVACY AND CONFIDENTIALITY

The privacy challenges associated with genomic information are two-fold: it can be uniquely identifiable and has an inherently familial nature.

Genomic information can be uniquely identifiable, limiting the potential for perfect anonymisation. This is especially challenging when information is stored for future research, especially given the aforementioned challenges for traditional consent paradigms in this context. Members of the Australian public are more willing to release personal information without consent for the purpose of research if it is de-identified. However, there appears to be limited understanding that removing names and other direct identifiers does not guarantee full privacy protection. (King et al, 2012)

A related privacy trade-off is the availability of personal and scientific benefit from genomic information and the information's identifiability. Personal benefit can come through the return of genetic findings from secondary research. Scientific benefit can come through the potential for data linkage activities and more complete phenotypic information. This has particular pertinence in the context of patients with rare diseases, who often expect that data are shared for scientific advances. Yet patients are also concerned about being identified, which is a risk that is heightened in the rare disease context. (Nguyen et al, 2019)

The familial nature of genomic information raises privacy challenges; in particular, in scenarios in which an affected family member seeks to prevent disclosure of genetic information to other, potentially at-risk, family members. In accordance with *Essentially Yours*, the *Privacy Act 1988* (Cth) has been amended to permit the disclosure of genetic information to an at-risk relative when there is a serious (although not necessarily imminent) threat to that person's life, health, or safety. However, these amendments apply only to doctors and other health professionals working in the private sector. (Otlowski, 2007) Some but not all states have enacted equivalent amendments to their privacy legislation.

Duties of confidence will also apply in some circumstances, although the application of such duties may be challenged by new models of healthcare characterised by increasingly complex dataflows. (Taylor and Wilson, 2019)

Stakeholder consultation

Patients and participants are sometimes concerned and confused about the protection of privacy in the use of their genomic information [Participants 15 and 25]. Genomic information was considered to be special: *"...People worry that the DNA is going to reveal something about them... I guess somehow DNA is the last secret [we have and] don't want to show otherwise we are totally naked"* [Participant 5]. Government access to and use of genomic information was raised as a concern [Participants 15 and 26], even when it was noted that this concern was unwarranted. Anonymisation of the information was not seen as a complete solution: *"...even by being anonymised it is pretty specific data, it can be found again down the line ... by current standards it might be not technically possible to identify me but who knows what will happen in the future?"* [Participant 6].

Aboriginal and Torres Strait Islander participants pointed to past experiences as a driver of concerns about privacy. Participant 44 said *"I know there has been some dodgy stuff done"* and that this has led to a *"certain amount of paranoia"* and a belief that privacy law has been misused to allow inappropriate access to and use of Aboriginal data.

However several stakeholders we interviewed indicated that privacy issues were not an important concern for a cohort of people who were seeking a diagnosis. For instance, Participant 38 said *"If you are sick you don't care [about privacy issues] and if you are not sick you care"* [Participant 38].

The relevance of genomic information to family members was also a challenge to individual privacy. The idea that research participation just involves the individual *"can no longer apply... the [genomic] information is of concern and interest to a wider group"* [Participant 2]. Participant 3 expressed that because of this characteristic, there should be a limit on the exercise of individual privacy: *"although [the patient] should have a right to privacy to an extent, [they] shouldn't be able to control how someone else's health is being looked after once information is known"* [Participant 3].

For researchers, the protection of data subject privacy was seen as complex and as an impediment to research progress. Data linkage raised the risk of re-identification, for instance [Participant 19]. Laws protecting privacy were noted by several researchers as unduly restrictive and complex, particularly around data sharing: *"It would be better if there were clear guidelines and people were feeling confident that they had done everything they need to do to protect the privacy of individuals and that then they could start to collaborate or use the information for other purposes that we do not yet know"* [Participant 19]. Participant 26 indicated that the privacy legislation should address genomic information specifically.

Trust emerged strongly as an important factor in reducing privacy concerns: *"I think the vast majority are happy to share their information as long as they know who has it and what is being done with it"* [Participant 26]. *"If we really want progress we need lots of people to be comfortable with sharing genomic data"* [Participant 30]. The provision in privacy legislation permitting disclosure for research in the public benefit was seen as important for researchers weighing up when to share data with other research groups. *"It gets tricky because we don't want to release data to another third party if ...we don't know what people are using it for"* [Participant 19].

Some stakeholders expressed concern that there were wider societal impacts when privacy fears impeded data sharing which in turn halted progress in both research and clinical care. *"In an ideal world... all these clinical genomic testing results [are available in] one accessible, protected resource which will be accessible by clinical service providers and researchers...This [allows us to] triple the value because my disease can be solved by my neighbour's genome"* [Participant 5]. *"It would be disappointing if people responded by just siloing information because they are unsure about the [privacy] framework and where they sit with the law"* [Participant 19]. Participant 40 stated that there *"needs to be a social responsibility in determining how we use that information"* and that if a person wants to reap the benefits of genomics they *"also have a responsibility to share that information, along the lines of vaccination, which is a good example"*. Two participants highlighted the field of cancer genomics as a good example of how widespread sharing of genomic information can have significant benefits for individual health care and wide-ranging research [Participants 5 and 40].

1.1.3 DATA SUBJECT UNDERSTANDING

There is evidence to suggest that patients and participants can have difficulty understanding some genomic test results, especially when results are indeterminate. (Grover et al, 2009) Uncertain test results also raise challenges for healthcare practitioners, which will have flow on effects for patient understanding. (Turbitt et al, 2013) Empirical research has also found some evidence of 'therapeutic misconception' in genomic research studies, despite clear information in Participant Information Statements explaining that the research might not produce useful or meaningful information. (Gillam et al, 2006)

Stakeholder consultation

Lack of understanding of genomic information and its implications

Participants expressed a range of concerns associated with lack of understanding of the implications of a genomic test. These included lack of understanding of the clinical relevance, societal implications, and fears associated with people turning to misleading commercial products to fill a knowledge gap.

Concerns around clinical relevance were sometimes related to a fear that people may overinterpret the significance of a result: *"I think sometimes the idea of genetic information gets demonised and made to be scary when it is not much different to a lot of other information"* [Participant 40].

An example was given of people who decide to undergo interventions which may be unnecessary e.g. women deciding on mastectomies based on a mutation: *"we could develop a level of paranoia that is not helpful... As a population we need to be more sophisticated than we are"* and take more informed steps [Participant 38]. Another commented that: *"I think the challenge [is in finding] variants that people [incorrectly] latch onto as if they are the cause"* [Participant 30].

A naive genetic determinist view may be compounded by an unduly optimistic opinion regarding the state of current scientific understanding. This can raise expectations unrealistically: Patients in hospital who are living with a condition may expect a diagnosis but the *"realistic expectation around that is also that we mightn't find a solution"* [Participant 30].

> *"I think the concern will still remain that we need to be very clear about realistic expectations and the complexity of understanding the results which will still be a concern in hospitals... Understanding of the results and correct interpretation is something we constantly need to be mindful of"* [Participant 30].

Lack of understanding societal implications extended to an individual's prospects for employment and insurance: the *"broader community have a real lack of understanding about what it means for the individual, whether they are legally obliged to disclose to insurance, jobs, and things like that"* [Participant 29].

In the face of this lack of public understanding a concern expressed was that people might turn inappropriately to commercial products: *"What I worry about is people's ignorance and wanting to be informed can [result in being] led down a path to buying a wristband"* (referring to DNA wristband which recommends products to consumers based on DNA) [Participant 29].

Public understanding of the uncertainties inherent to the analysis of a genomic test may not be helped by marketing of direct-to-consumer genetic tests. One participant referred to having seen genetic testing kits for sale for $50 and their worries about that:

> *"genetic testing is thrown around [by the public without an understanding of what it entails/outcomes/effects]...The public is sufficiently confused that they can't necessarily determine the difference between sending... DNA to Ancestry.com to work out whether you have Viking blood or sending your DNA to someone to have a whole genomic sequence right across with incredible sophistication and depth"* [Participant 29].

The presence on the market of these tests may also contribute to the raised expectations already mentioned:

> *"What is new and emerging is the dichotomy between public expectations driven by things like the direct-to-consumer market versus the in situ capacity of the clinical system and knowledge in physicians to be able to respond"* [Participant 26].

This lack of a sophisticated understanding of the limits and uncertainties associated with genomic test results can extend to some health professionals. It was suggested that: *"[Genetic counsellors and geneticists] are ... sensitive to the technical nature of that information and that a whole bunch of clinicians do not understand this stuff let alone members of the general public"* [Participant 26].

Public (and professional) education requirement

It was suggested that the investment in progressing the science of genomics needs to be matched with an investment in public (and professional) understanding.

> *"I think there is really a need for something, particularly when the government has allocated so much public money, there is a need for the general public to be able to access information in a very easy to understand way...the information] needs to be basic [to inform people of the upsides and downsides of sharing genomic info]...The Genomic Health Futures Mission... have a responsibility to provide some form of easy to understand education, because it is something you just about need a PhD to begin to understand what everyone is talking about"* [Participant 29].

One participant noted that new developments (specific to cancer risk genes) bring with them *"a completely different way of looking at cancer"* and this required *"an important education initiative"* to ensure that participants are informed about the new technology and what it might mean for them [Participant 36].

This may become increasingly urgent as genomic testing becomes more widespread and moves beyond diagnoses and genomic information toward preventive use, and ultimately to inform and develop curative treatments.

> *"The philosophy that there are individuals with normal DNA and there are individuals with bad DNA is totally wrong, we are all vulnerable to errors...We do need to appreciate that [in the future] we are not only going to be diagnosing these diseases but we will be predicting the trajectory of these individuals based on their DNA and [as a society] we need to be prepared"* [Participant 5].

> *"It is illogical, the government [has spent $50 million] on the AGHA, and we do not have a framework in which to even look at translating some of this science into the community and that will become more interesting when you start to look down the track at things like CRISPR and in vivo CAR-T [which are] essentially one stop shop curative cancer treatments"* [Participant 38].

Education initiatives are something that have been done successfully in relation to specific populations and so could be scaled. If communication is effective, then public understanding is possible. One participant noted that that they are involved in a program that was initially focussed on the BRCA1/2 genes, but has extended over time to analyse additional cancer-associated genes - this change *"has not been a problem for participants to get their head around"* and participants who did not have the BRCA1/2 genes *"were really thrilled"* that additional genes were being analysed.

This has extended to the relatively complex issue of incidental findings. In 2010 with the advent of genome sequencing and exome sequencing, a research institute was concerned that incidental findings would be an issue. The project sent out communications to participants about the emerging technology and what to expect and participants received it well with only one out of thousands of participants raising questions - *"[the issue of incidental findings] has not been the nightmare we thought it might be back in 2010...These days we run a 300 cancer gene panel, people love it"*. The view of breast cancer patients and participants is *"anything you can do to stop this is good...[they] are pleased that the technology has developed"* [Participant 36].

Indigenous genomic literacy

Indigenous stakeholders reported that communicating genomic concepts sometimes required sensitivity to language and cultural considerations. Participant 44 noted that:

> *"We have just done work... educating in remote communities around genomics. I think we are dealing with fairly complicated concepts which you do need almost language translation for – because English language is a difficult language – translating this into Indigenous terminology is difficult but usually illustrated stuff is better. Whilst there has been educational materials and resources around Indigenous communities there has not been very much, and the level of literacy is even lower than in the general population."*

However, even in remote areas where language barriers and cultural differences might be hypothesised to influence understandings of genomic information, stakeholders reported very good understanding of genetics and heredity:

> *"People felt comfortable with genetics...they're comfortable because they know where they're from, because I'm going out to their community, and they also know that all their skin grouping stuff is around making sure people marry the right way so you don't have a wrong way baby...so you don't have children with issues...this had been introduced into their family story line for generations... Lots of people watch CSI and all the shows on TV that deal with DNA"* [Participant 43].

Several participants reported that synergies between western understanding of genomic information and Indigenous understanding of kinship systems, inheritance and connections between groups and country facilitated discussions about the implications of genomics.

1.2 Group and public interests in the use of genomic information

1.2.1 GROUP INTERESTS

Genomic information commonly is linked with participants' racial and ethnic categories, raising the potential for group interests associated with the use of such data. (Eckstein, 2011) Australia has only limited regulatory strategies for taking such interests into account when it comes to the collection and use of genomic data. A number of Australian researchers have implemented and published on strategies for respecting group interests in genomic research in the context of Aboriginal communities, noting, for example, that 'genetic research can be both ethically and successfully conducted with Indigenous communities by respecting the authority of the community, involving community members, and including regular community review throughout the research process'. (McWhirter et al, 2012) One such adaptation is the use, for some projects, of saliva samples rather than blood samples for DNA extraction. (Kowal et al, 2015)

However, work remains to be done in this space, including when it comes to the secondary use of genomic data. In the context of data sharing, Jane Kaye and her colleagues advise that 'While in many regions of the world such as sub-Saharan Africa, mechanisms have been developed to engage communities and obtain their consent before research implementation, such mechanisms are not broadly adopted in western countries or other countries with advanced biomedical research'. (Kaye et al, 2018)

Stakeholder consultation

In this theme, participants identified underrepresentation of certain population groups in genomic data sets and the implication of this for health research and care [Participant 18], and issues with communicating to culturally and linguistically diverse populations, the information needed for effective informed consent [Participant 38]. The need for communal as well as individual consent was identified by Participant 51: *"If you've got someone's DNA from a certain community or family*

that flows on to everyone else in that family". This presented a particular challenge in situations where people had been dislocated from their community.

Indigenous stakeholders, on the whole, did not feel the interests of Indigenous peoples as a group were currently well represented in the governance of genomic information. Participant 41 said *"it still feels like Indigenous people are still an afterthought"*, and that:

> *"if people are serious about this, they need to invest a huge chunk of money into Indigenous genomics – they need to invest in proper governance, infrastructure, community engagement. ... so long as there is representation for Indigenous communities and people are being looked after. ... In an ideal world, if I had millions of dollars, I would love Indigenous people to have their own set up with a board governed by good Indigenous people offering scholarships and programs for students, able to care for samples - a biorepository - so that they are cared for properly."*

Another stakeholder warned that Indigenous governance by itself may not be enough to ensure communities' needs were met: *"Ultimately communities still want to have the final say. If it's an Aboriginal person that they don't know [holding the sample or data] they might not necessarily trust them either"* [Participant 43].

Participants emphasised the importance of community consent in protecting group interests, but recognised the practical challenges in obtaining this:

> *"There is a need to think about not just dynamic consent which is great but also individual versus collective consent. The idea that as a person you can consent to your sample being collected, analysed and reported against however the question comes when... making reference to a broader group, a collective. You have consent from the one to be able to collect that sample... but have you got broader consent from the community whom that individual represents to actually undertake that analysis. ...Whilst I think it's ok to get individual consent, if we are making reference to a broader group we probably need to think about the implications for that broader group and then we need to seek endorsement and consent from that broader group to do that work... [but] there is no national body that represents all of the groups that can speak for all of the groups and endorse or consent"* [Participant 42].

> *"If a person is consenting in front of you they are consenting as a private individual – they have a right to do that... If you are working in a community... we need to make sure that community is engaged with what we are doing and what we are potentially going to publish. So you need community support. In that case if you are going to use that group of samples again you would go back to the community."* [Participant 53].

In particular, there was recognition that it is difficult to reconcile conflicts between individual autonomy and community interests in instances when an individual is recruited in a hospital setting or away from community. Participant 53 thought that, if it transpired that the community has said no, but the individual wants to participate, *"it should still be recorded somewhere that the community as a whole has concerns about why it wouldn't want the research."* In practical terms, this might mean ensuring that findings are not reported in a way that associates the research with that community. Participant 51 argued that community input is critical because that is the level at which the harm is caused: *"There needs to be alternate methods where the individual is dislocated from the community."*

Further, the limitations that a requirement for community consent placed on multisite research projects was observed by several participants, with Participant 51 noting that it is difficult to engage with Aboriginal and Torres Strait Islander groups at a national level, because of the number and diversity of groups across the country. Participant 51 linked this difficulty to efforts by successive governments to undermine national Indigenous bodies, such as ATSIC and the proposal for a Voice to Parliament.

1.2.2 RACIAL STEREOTYPING/STIGMATISING

A recognised harm that can result from genomic research involving racial and ethnic groups is stereotyping and stigmatisation, and international examples of this form of group harm are often cited. One prominent example is the use of genomic samples and data collected from members of the Havasupai Indian tribe, originally collected for diabetes research, for secondary research into schizophrenia, inbreeding and population genetics, all of which were potentially stigmatising or undermined cultural beliefs. (Mello & Wolf, 2010) Announcements of the identification of a 'warrior gene' in the New Zealand Maori population without contextualisation of the social factors that could be leading to the purported causation between a gene variant and antisocial behaviours further illustrates this potential for harm. (Wensley and King, 2006) In the context of Aboriginal and Torres Strait Islander peoples, concerns have been raised about potential unauthorised uses of genomic information such as for defining Indigeneity, or for forensic or paternity investigations. (Kowal et al, 2015)

Stakeholder consultation

One participant raised the issue of whether individuals from particular cultures *"will look kindly on whether they are different or not genomically, because it could be a source of bias or racism"* [Participant 2].

Indigenous participants were keenly aware of international and national examples of poor research practice leading to racial stereotyping, and several expressed concern that emergent genomic knowledge would be interpreted in ways that would perpetuate negative narratives about Aboriginal and Torres Strait Islander people. Participant 53 asked *"Are you going to get judged on your genome?"* This participant highlighted the potential for stigmatising genetic associations, such as predisposition to alcoholism or cigarette addiction, being attributed to a particular group leading to inequitable access to healthcare, or employment discrimination.

Participant 51 saw emergent technologies such as epigenetics as dangerous for Aboriginal and Torres Strait Islander peoples, because of the risk of interpreting the biological effects of intergenerational trauma in stigmatising ways:

> *"We know that from the low life span of Indigenous people that we carry the trauma of our mothers with us. The high rates of diabetes and all of these other things may be linked. There's a risk when you start doing that stuff - I don't think we'll be blaming it on the trauma that's been inflicted on our peoples – it'll be blamed on us as genetically weak."*

1.2.3 DATA SOVEREIGNTY

Indigenous control over samples and data is frequently highlighted as the key to rebuilding trust in the collection, use and disclosure of genomic data for clinical and research purposes. One influential way in which control has been conceptualised is the idea of Indigenous data sovereignty, which derives the right to govern the collection, use and interpretation of data from the United Nations Declaration on the Rights of Indigenous Peoples (UNDRIP).

In Aotearoa/New Zealand, Indigenous data sovereignty is finding expression through the development of *Te Mata Ira: Guidelines for Genomic Research with Maori* and the Genomics Aotearoa programme of work. (Hudson et al, 2016; Kennedy, 2018) In Australia, the goals of Indigenous data sovereignty are being pursued by bodies including the Indigenous Data Network and Maiam nayri Wingara. (Maiam nayri Wingara, 2018)

Stakeholder consultation

Indigenous data sovereignty was conceptualised by stakeholders as a legal justification and practical mechanism for effecting Indigenous control over how samples and data collected from Indigenous participants and patients are accessed and used, by whom, and for what purposes, as the following quotations illustrate:

> *"To me, data sovereignty means Aboriginal people being in control of their data; regardless of how much it is watered down, it is still my data. It is still my data and my right to say what you can and cannot use that for. I would see a repository or centre for Indigenous data as taking on that data sovereignty: 'we will hold these samples, but they will always belong to the participant or family or community' and it will always be their decision as to what is useful: disposal or kept?"* [Participant 41].

> *"Control was more around the idea of data sovereignty from an indigenous data sovereignty point of view, i.e., we control who accesses the samples and we control where the samples are stored and we control the duration for which they are stored and we control that after a set and agreed period of time they are returned."* [Participant 42].

> *"...in terms of the aspirational goals this is an opportunity and the first time to really empower Aboriginal people with their national Indigenous stories and to be able to do it by mob, language, region, however Aboriginal people decide they would like to do it and to give them power and control. ... in a larger Aboriginal-controlled dataset, you will really be empowering the individual, the family, the mob and the entire Indigenous population to have their data safely and securely stored, being managed in a culturally safe manner and to be able to have those longer discussions and different discussions about how Aboriginal people want that data to be used for their benefit."* [Participant 66].

Participant 53 believed that Indigenous-led repositories had the potential to be more trusted by Aboriginal and Torres Strait Islander communities, as this model was thought to be more likely to *"give more control back to the people whose samples they are."*

Participant 51 argued that Indigenous data sovereignty was a governance solution that would address the concerns and distrust felt by Aboriginal and Torres Strait Islander people, and that it was founded on an assertion of Indigenous people's rights to data, including but not limited to genomic data, derived from international law:

> *"At the moment we're excluded from all governance and we're expected to stump up and give samples because it's apparently going to be beneficial...well we've been hearing that for 200 years. ...I think this is the only way you will get a genetic reference group - if there's Indigenous people reassuring people. Sovereignty is at the level of the individual and the community, but it's more at the community level because it's at the community level that the harm is done."*

1.2.4 PUBLIC UNDERSTANDING, TRUST

Goodwill from the public is essential for the ongoing viability of biobanks and other research dependent on long-term storage and sharing of genomic data. (Critchley et al, 2015) Once diminished, trust is hard to rebuild. (McGuire et al, 2019) In the Australian Aboriginal context, trust has been recognised as a key explanation for attitudes to the use of blood samples for research, with higher levels of trust associated with a greater willingness among participants for researchers to store and use blood samples, despite their cultural concerns. (Kowal et al, 2015) The recently completed global *Your DNA Your Say* survey addressed the issue of public trust. Results revealed that participants were most likely to trust their medical doctor and that company researchers were least likely to be trusted. (Milne et al 2019)

Stakeholder consultation

One participant held up the Estonian biobank's success as being based on a clear legal framework that protected genomic data from use by employers and insurance companies, hence engendering the necessary trust for this population-wide initiative: *"People have confidence and many have got on board"* [Participant 3]. Participant 11 concurred: *"there needs to be just as much effort in how we manage the public's perception of what we do with their data as well as how to get the technology to work as well as we need it to"*. Untrustworthy behaviour by organisations sharing genomic data with commercial entities could risk damaging trust however [Participant 11].

(Dis)trust among Aboriginal and Torres Strait Islander peoples

Trust, and lack of it, was a dominant theme running throughout the interviews with Indigenous stakeholders. Given the potential risks of harm to individuals, their families and their communities that can arise from genomic information, a high degree of trust is required to be vested in the individual seeking to collect, use or disclose that information.

Generalised distrust took two key forms: distrust of authorities, particularly the government and the criminal justice system; and distrust of researchers. Both forms are founded on a long history of negative past experiences.

Participant 41 observed: *"First and foremost, people talk a lot about relationships with past researchers and the work that people have done previously and have either not followed up with the participant or communities or they have disappeared, or they have gone in and built trust and then have broken that trust."*

Participant 51 viewed the impact of past experiences as critical to understanding Indigenous perspectives on genomics:

> *"For Aboriginal and Torres Strait Islander people - with the very poor record of the research community both in Australia and elsewhere - ...we are deeply suspicious, and we do not think that those doing this research have actually thought it through from an Aboriginal and Torres Strait Islander perspective. ...They [the researchers] have rose-coloured glasses about the benefits of genomic research without any real understanding of the risks involved."*

Participant 66 believed that the impact of past experiences with research were so damaging that genomics with Aboriginal and Torres Strait Islander peoples could only move forward if the discussion was restricted, in the short term, to clinical purposes and aims raised by communities themselves:

> *"Where it gets sticky, is as soon as you raise the word research, you alienate that entire community because again it will be viewed as ... "Why couldn't it be framed purely in the form of health benefits? Now if in the future [there was an] Aboriginal controlled data repository or reference dataset [and people] believed there was some research they would like done, they could call that research but [until then] we need to take research out of the conversation. It should be about capturing and using the old knowledge with the new knowledge for the benefit of the Aboriginal people. ...Once empowered the discussions around research will change. There'll be a difference in competence, there'll be an ability to be able to create a trusting relationship. With the control they're empowered so they feel empowered in the discussion so they're not trying to claw back things. People forget Aboriginal people and culture is set up around reciprocity. We take, take, take and never give. If we were to change that paradigm just once and give and trust their cultural intent would be to...reciprocity. We keep missing a huge opportunity."* [Participant 66].

Trust in specific individuals, groups or institutions was commonly identified, and participants reported that trust could be established through proper consultation processes and investing the time and resources into building relationships with Indigenous communities. In cases where trust was established between the community and the research group, stakeholders reported very little pushback against the use of genomics in health research. However, that trust was unlikely to extend to others who might wish to access the information for secondary purposes, whether that was clinical, research or other purposes. Participant 42 reported their experience of consenting for health research: *"It is consent to store and then analyse but there was a serious concern about who accesses it. Because the longevity of the sample means more potential for somebody to access it who shouldn't access it."*

Lack of trust was so pervasive that even very limited examples of reuse of genomic samples and information were viewed with suspicion. Participant 43 noted that *"We asked if the data could be used on other projects related to [specific disease] and the related illnesses. Only about 50% said yes to future use. That one a lot of people get quite stuck on."*

Distrust was not associated with poor understanding of genomics, and a number of participants reported that genetics tended to reflect Aboriginal understandings of kinship and connection, such that genomic concepts were readily grasped by community members.

1.3 Data 'ownership', control, custodianship

Legal and ethical claims to 'ownership' over samples and tissues remains a vexed issue and any meaningful answers will depend on analysing specific forms of entitlement that might apply. (Roberts, 2018) This may include a limited right to exclude others from accessing genetic data. (Roberts, 2018) Patients and participants will also have rights to access identified genomic data, based on the Access Principles of Commonwealth, state and territory privacy acts. Notably, however, this is inconsistent with paragraph 3.3.28 of the Australian *National Statement on Ethical Conduct in Human Research* (2007), which states that 'While participants may have a strong interest in their own information, researchers are not expected to return raw genomic data to participants'. This apparent inconsistency requires attention.

An associated concern is the need to ensure ongoing participant confidence in the management of their genomic information, including opportunities to serve participants whose data sharing preferences change over time. (Delaney et al, 2016)

Stakeholder consultation

Several participants spoke about the difficult question of the 'ownership' of genomic information; notably this emerged even when a participant acknowledged that information cannot be 'owned' in a legal sense. Participant 16 queried whether the 'owner' was the patient, the test kit vendor or the researcher. Participant 18 reflected:

> *"I get the sense that certainly the health consumer, the patient, the relationship of the patient with their own healthcare has evolved so much in the last few decades and I think this will continue to and will accelerate where it is much more n=1 healthcare and the consumer demands much more control over their clinical management. I think we will get more requests for access to their own data, management of their own data, tools to interrogate. This will have interesting implications for a legal context – what is one's right over their own data?"*

This question of ownership also affected consideration of the inheritance of genomic data, with at least two participants pondering what rights, if any, the family would have after an individual has died: *"is this considered an asset or once it leaves [one database] and is being analysed by another group is the law saying because it is manipulated and not in original form it is not part of you (and you don't own it?)"* [Participant 16].

The need for clarity around questions of ownership, control and responsibility for data as well as samples emerged strongly in the interviews with both researchers and patient advocates. Participant 12 pointed out that as routine sequencing becomes much more widespread in the future, it becomes more pressing that these questions are resolved.

For Aboriginal and Torres Strait Islander stakeholders, the issue of control over genomic information is inextricably linked to trust. As trust decreases, the desire for tighter control increases, and vice versa. The limitations of consent as a mechanism for control meant that the justifications and mechanisms for control included approaches such as: 'ownership' of samples and data, 'custodianship', and 'data sovereignty'. Participant discussions highlighted the need for clarity over who has the right to make decisions about stored data and samples in the longer term. While many participants had trust in the research group or clinical care team they interacted with, they were concerned about subsequent uses. For example, Participant 41 reported community members' concerns about the long term security of their samples:

> *"There were questions asked 'what are you going to do with these samples, who makes decisions for those, are you going to keep them in [location away from community].', and there were concerns about in 20 years' time the laws might change and you might be able to keep the samples for whatever reason if they are a certain age and this did not stick well with the community."* [Participant 41].

There was evidence that framing the discussion in terms of 'custodianship' rather than 'ownership' implied a responsibility to do the right thing in taking care of the data or sample:

> *"My thoughts are about infrastructure allowing participants and the community to control what's done with their data. Who can refute and answer the important questions? There is a real need for that going into the future...I use the term 'data custodian' a lot as it means we have a right over it, or we have been given a right to look after that information and it's up to us to make sure something happens to it rather than just being researchers."* [Participant 53].

2 Genomic information in healthcare contexts

2.1 Workforce

2.1.1 CAPABILITY, TRAINING AND ACCREDITATION: UNDERSTANDING OF GENOMICS AND BIG DATA

Literature review

With the transformation of genomic sequencing from the realm of research to clinical practice, commentators stress the importance of supporting clinicians to integrate genomics into medical practice. (Stark et al, 2019; Delaney et al, 2019) Australian Genomics includes workforce preparedness as a part of its mission, and has developed tools to support evidence-based genomic education and evaluation. (Stark et al, 2019) Others suggest the provision of pre- and post-testing checklists. (Delaney et al, 2016)

Particular challenges arise at the interface of commercial genomic testing and clinical practice. Consumers will often seek help with understanding their test results with a health professional, which places additional pressure on the health system. (Savard et al, 2019) This issue may be especially acute for test results with inconclusive scientific validity and where support is sought for non-health related test results. (Metcalfe et al, 2019)

Stakeholder consultation

Stakeholders reflected on two issues: an insufficient specialist workforce and a lack of training and accreditation of non-specialist clinicians. Participant 26 stated *"It is all well and good for the geneticist to be able to describe in some specialist letter back to the neurologist... what is going on, but primary care has no idea what all this means"*. It was suggested there are risks attached to the use of genetic/genomic information by clinicians who lack appropriate skills. Additionally, there is a wariness by some clinicians of making use of such information or of delving into the field at all, because they lack understanding of it. The use of genomic information in hospitals was identified by Participant 30 as potentially problematic: *"the complexity of understanding the results [..] will still be a concern in hospitals...correct interpretation is something we constantly need to be mindful of."* However the same participant indicated that substantial effort had gone into the translation of genomics into public hospital settings and this had been successful.

Indigenous stakeholders noted two key issues related to workforce considerations. The first was a lack of accommodation of culturally relevant factors, such as the role of family in decision-making or how holding discussions behind closed doors might be perceived [Participant 49], and the impact that culturally inappropriate care had on the delivery of health services. The second was a more general issue that particularly affected regional and remote Indigenous communities: that health services providers in regional and remote areas were poorly equipped to offer, or to support patients in accessing, genomic medicine [Participant 44]. As Participant 66 noted:

> *"...the big hurdle is the fact that most of the interfaces with Aboriginal people are not culturally safe or culturally secure so we still have this Western front door into the service. Once they are into the service Aboriginal people get very good attention... but they are underrepresented in genomic testing by about 80% or so of the number of Aboriginal people you might expect attending, to [actually] attending genomic clinics based on population, it's 80% lower than you'd expect."*

2.1.2 UNDERSTANDING OF LAW BY NON-SPECIALISTS

Literature review

The legal issues arising from genetic testing can require complex assessments by health professionals. Some empirical research suggests a limited understanding among health professionals about how to implement their obligations in these contexts. For example, in the context of non-consensual disclosure of genetic information to at-risk relatives, many genetic health professionals in NSW were found to lack understanding of the scope and clinical application of relevant guidelines. (Meggiolaro et al, 2010) Clinician understanding of the legal issues is further challenged by ongoing legal uncertainties, as exemplified in the recent English case of *ABC v St George's Healthcare NHS Trust & Ors* [2020] EWHC 455 (QB) (28 February 2020).

Stakeholder consultation

Several stakeholders suggested that clinicians lack good knowledge of what the law requires in terms of genomic information use; this is increasingly a problem as genomics moves into mainstream clinical practice: *"it won't just be a genetics professional sitting there who knows what to do"* [Participant 3]. Clear guidelines are required, for instance around the disclosure of clinically-actionable results with family members in the absence of patient consent. Researchers also lack a good understanding of the law, for instance around the sharing of genomic information.

2.2 Patients

2.2.1 EQUITY OF ACCESS (TESTING, TREATMENT, ADVICE RE: TEST RESULTS)

Literature review

State governments fund clinical genetics services, with only a limited number of genetic tests funded federally through the Medicare Benefits Schedule. Private health insurance does not currently cover the costs of genetic testing. Some but not all states have made significant investments in clinical genomics. (Stark et al, 2019)

Stakeholder consultation

Participants commented on significant variation in the availability of access to genetic/genomic testing in different parts of Australia and different clinical contexts, the lack of a Medicare rebate for most testing, and consequent ongoing concern about fairness and equity. Some patients circumvented access challenges by obtaining genomic testing for clinical diagnostic purposes via research studies. A patient advocate, in the rare disease arena [Participant 6], commented *"We hear of some patients whose clinician is tapped into a certain research program that might have access to certain testing, the [patient's sample] may be sent overseas. It is a toss of the coin as to who actually gets access to that"*. Patients with conditions that were particularly complex or not well understood faced higher barriers to access.

Indigenous stakeholders reported some evidence of reluctance on the part of non-Indigenous researchers to do genomic research with Indigenous groups, affecting access to participation in research:

> *"In talking with others in our institute, there was a reluctance to do genomics research with Indigenous people. In workshops it was asked why the reluctance for Indigenous people to engage with genomics? They came back to the Human Genome Diversity Project and [there is] generally some consensus about this being a no-go space and that Aboriginal people were not appreciative of this work and this work did not align with their preferences with respect to health research."* [Participant 42].

Equity was also seen as being adversely affected by the inability of clinical services to provide culturally appropriate care, particularly with respect to Aboriginal and Torres Strait Islander patients. One example concerned consultations being conducted behind closed doors, with the intention of providing privacy. However, this was viewed by some communities as indicative of having something to hide and was linked to increases in the 'shame' associated with genetic disease. Another example was a common failure to take family decision-making and role of family into account when providing genomic health services to Indigenous patients [Participant 49].

Those working in regional areas raised particular barriers to access for lower socioeconomic groups and those living in regional areas. As Participant 44 noted:

> *"Generally there is new technology and it's expensive, and the poorer populations miss out on this, so the other part of genomics is making sure that Indigenous health is at the forefront of genomics and not left behind due to poor technical ability or capacity. The other aspect of that is how we ramp up primary healthcare to better understand genomics and the practical applications of that. If the GPs don't know enough to ask for the test then lots of times our GPs operate in isolated practice so you aren't always keeping up with the latest, certainly genomics. So being across this is knowing of all these broader tests etc. There is a big capacity gap there."*

2.2.2 UNDERREPRESENTED AND VULNERABLE GROUPS

Literature review

The vast majority of participants in genomic research have been people of European descent. Although improvements have been made in recent years in the inclusion of participants of Asian ancestry, the extent to which people of African and Latin American ancestry, Hispanic people and Indigenous peoples across the world are included in genomic research remains disproportionately small. (Popejoy and Fullerton, 2016) This lack of diversity in research has significant implications for understanding disease aetiologies. This has flow on effects for the ability to provide effective care in clinical settings utilising genomic medicine, leading to concerns that "those with the greatest need are the least to benefit from these advances." (Sirugo et al, 2019) In Australia, this issue raises difficulties in relation to a range of minority populations, but is particularly acute for Aboriginal and Torres Strait Islander people. (McWhirter et al, 2015)

Stakeholder consultation

> *"Questions around the efficacy of genomic technologies for underrepresented populations is a big one that we are not addressing well."* [Participant 18].

Several participants noted that underrepresentation of certain groups in genomic databases perpetuates inequality and restricts progress in research and clinical care. Failure to rectify this problem will result in certain populations being *"left behind"* [Participant 18]. The lack of a reference genome was considered to be a particular issue for Indigenous people [Participant 41], with particular effects in emergent screening programs:

> *"An area where it's really unfair because of the lack of an Aboriginal reference is the application of genetic testing and the movement of genetic tests into non-invasive prenatal testing and preconception carrier screening. Assuming both of those are going to at some point come into play, because we lack genomic information and because they are predictive, you cannot apply the same degree of certainty to a predictive outcome to Aboriginal and Torres Strait Islanders as some of the Caucasian, North American and Western European descendants. We do not know what's a pathological mutation and what is not. There the federal government is running ahead with these programs on preconception carrier screening…if an Aboriginal person presents they'll take their blood and they won't do anything with it because they can't do anything with it. We're creating a false hope. We're not being transparent. That to my mind creates a scenario where you could start to erode trust."* [Participant 66].

Indigenous stakeholders were aware of the problems for clinical care raised by the lack of information on genomic variation in Aboriginal and Torres Strait Islander populations, but this was counterbalanced by the perceived risks. Most participants felt that it was important to be included in genomic research, but that it was critical that it was done to the highest ethical standards, and that Indigenous leadership would be a significant factor in successful initiatives. As Participant 51 said: *"I know that genetics is the way of the future so we have to be in there…we have to be part of it, but we have to be at the decision-making table, not as advisors or consultants."*

A number of participants expressed aspirations about the potential role for Aboriginal and Torres Strait Islander people in genomics. For example, Participant 66 stated that the alignment of genomics with traditional understandings of kinship, of songlines, and of connection to country gave them a unique opportunity in this field:

> *"Aboriginal people don't want to be left behind, they want to count and not be counted, and aspirationally they believe that genomics, and this is an aspirational goal they have come to articulate after many years of us in discussion with them about genetics, they believe that Aboriginal people have an opportunity to go forward with genomics and to reap benefits from genetic testing and genomics ahead of the Caucasian population."*

2.2.3 FAMILY INTERESTS

Literature review

The familial nature of genomic information can raise challenging communication issues when family members have different wishes. A common challenge is active and passive non-disclosure by a person who has received genomic information. In this situation, a health professional's obligation to prevent harm to at-risk relatives will conflict with duties of privacy and confidentiality. Amendments to privacy laws, at the Commonwealth level and in some States and Territories, ameliorate this conflict, however, implementation challenges remain. (Meggiolaro et al, 2020) There is considerable variation among genetic health professionals when it comes to writing letters for at-risk family members or other assistance for family communication. (Forrest et al, 2010)

The England and Wales High Court (Queen's Bench Division) case *ABC v St George's Healthcare NHS Trust & Ors* [2020] EWHC 455 found that treating clinicians had a limited duty of care, in some circumstances, to family members, including a possible duty to disclose genetic information against a patient's wishes. It is unclear whether this limited duty to disclose would apply in the context of Australian negligence law, especially given the highly specific factual circumstances at issue.

Conflicting family wishes can require complex informational solutions. One case study reported on a request for predictive genetic testing for an adult-onset neurodegenerative condition for the purposes of reproductive planning. The requestor's at-risk parent advised that, if they found out they had the mutation, they would commit suicide. This raised questions of how to balance duties of the treating team to the person presenting for testing with possible duties to non-presenting family members. The eventual decision relied on the requestor entering into a written agreement not to disclose any results of genomic testing to anyone other than his partner to minimise the chance of unwanted disclosure. (Stark et al, 2016)

Stakeholder consultation

A number of participants identified that the implications of genomic test results for family members presents a challenge throughout the testing process.

At the start of the process there is concern about individual versus familial interests in making a decision about whether to participate in testing. Participant 53, referring to Aboriginal people in the Top End, stated: *"People ask: 'how will this relate to my family if you find something?' … I think that family disagreeing with an individual's decision to participate would impact more on an Indigenous individual than a non-indigenous individual."*

Several participants identified concerns around disclosure to family members in the absence of patient consent in the clinical context. One stakeholder from a national disease-based biobank described their well-considered practices in feeding back results to clinics for return to family members in certain circumstances [Participant 36]. Although they had effective policies and processes in place, the practice of feeding back results remained challenging due to diverse family expectations, views, and family dynamics.

Participant 3 identified that patient privacy obligations are a perceived impediment to disclosure of clinically-actionable results by clinicians to family members; that despite the Commonwealth Privacy Act and all state and territory jurisdictions including provisions that make it possible for clinicians to disclose information to family members, only the Commonwealth and NSW have guidelines on this practice. It was suggested that nationally-consistent guidance for clinicians across both public and private healthcare contexts would enhance practice in this area. Participant 3 also thought *"that people shouldn't be allowed to choose who their data is used to benefit in a family member sense."*

2.2.4 MY HEALTH RECORD

Literature review

In 2018-19 during the switch to an 'opt-out' approach to the national electronic medical record My Health Record in Australia, concerns about the inclusion of genomic test results in the system loomed large in the public debate. Literature on the integration of genomic test results in electronic records derives mainly from the U.S. (For example, Kannry, 2013)

Scheuner and colleagues conducted interviews with healthcare providers in the U.S. in 2006-7 and found a lack of readiness of electronic records for genomic medicine, with one interviewee noting that 'Barriers to integration were mostly related to problems with family history data collection, documentation, and organization'. Also missing were 'standards for data elements, terminology, structure, interoperability, and clinical decision support rules'. (Scheuner et al, 2009) Privacy, confidentiality and security of genomic information in electronic records is seen as crucial. (McGuire, 2008; Hazin, 2013) However researchers have also posited the likely benefits for the genomic literacy of patients and providers of including this information in electronic health records. (Hazin, 2013) One review found that better technical tools for the standardisation, validation, storage and workflow management around clinical genomic data would need to be accompanied by improved consent mechanisms (see 1.1.1) and patient education. (Shoenbill, 2014)

Stakeholder consultation

In relation to genomic information, My Health Record was considered by several participants to be a lost opportunity, stymied by misinformation in the public sphere and lack of trust in government. Participant 26 reflected on the *"ignorant experts"* whose commentary had inappropriately swayed public opinion. This participant reflected on the need for government to support accurate representation of these issues in the media, noting that the large majority of concerns that were expressed about the inclusion of genomic information in My Health Record are already well met in the existing legislation.

3 Dissolving boundary between healthcare and research: the Learning Healthcare system

3.1 Gaps and links between clinic and research

Literature review

The literature evidenced a clear trend towards the breaking down of boundaries between clinical and research activities when it came to genomics. With the advent of increasingly efficient sequencing technology, gaps have become common between the 'discovery of therapeutic benefits in research and actual adoption of the new technology into clinical practice'. (Berkman et al, 2014)

The blurred boundary between clinical and research activities is well illustrated by programs for the diagnosis of cancer and rare diseases, such as the 100 000 Genome Project in the UK and the U.S. National Institutes of Health Undiagnosed Diseases Program. In the latter, participants receive extensive genomic testing, including gene panels, single nucleotide polymorphism analysis, and exome sequencing. The program was established as a research study and has led to extensive research outputs. Yet, it has also provided diagnoses and, in some cases, treatment opportunities, for individuals and families who had been experiencing long-standing diagnostic odysseys in relation to rare diseases. (Gahl et al, 2016)

The dissolving boundary between healthcare and research extends beyond *outcomes* to also encompass the processes and practices employed for genomic testing. Nguyen and her colleagues explain that, in the rare diseases context, the scarcity of patients and the need to share information internationally to find similar cases makes data sharing and 'matchmaking' imperative for clinical outcomes. Machine learning procedures are also being employed for data phenotyping, further blurring research and clinical practice. (Nguyen et al, 2019) The generation of large data repositories from clinically generated sequences for clinical practice, research, and public health will further test these traditional divides. (Capps et al, 2019)

Stakeholder consultation

The traditional distinction between clinical care and health research is seen by some as increasingly anachronistic and problematic. *"We are in this interesting through-the-looking-glass period"* where, in 2018, 20% of all human genome sequences were in health, with the rest in research, but by 2022 it is forecasted to be the opposite, with 80% sequenced in health. [Participant 26]. *"I think we shouldn't be separating so dramatically [as] the boundaries are really disappearing [between the clinic and research settings]"* [Participant 5].

The current distinction drawn between clinical and research contexts is a concern for some due to lost opportunities: *"the loss of opportunity when clinical health information is quarantined just for health purposes and not made available for research"* [Participant 18].

"From a genomic research perspective, data linkage (i.e. accessing administrative data sets, hospital outpatient records etc.) is broken...the system is broken" [Participant 18].

"I know many institutions have been struggling with [cases where] patients on whom they do a lot of genomic testing are not being consented for that data to be used by the research community and that is opportunity missed" [Participant 5].

This may deny patients the opportunity to contribute to research: *"I think we need to take care to allow the option to be retained because many patients particularly with rare diseases are keen to contribute to research and I think we need to keep the option available to patients in clinical settings."* [Participant 18].

It was recognised that closer integration of research into clinical care may be resisted by some health care professionals: there is a *"subset of clinicians who are quite opposed to including an option for research in clinical consent materials"*. [Participant 18].

Potential reasons behind lack of clinical info being shared with research include lack of time for clinicians and non-genetic health professionals being *"unwilling to discuss what genomic research might look like"* [Participant 18]. There is also the issue of differing standards between the clinic and research - in the clinic the pathologist requires patient details (name, DOB etc to ensure samples are identifiable) - but then when the data is used for research: *"if you retain certain [identifying] components how does it affect the researchers who are doing their best to retain distance and not have personal details?"* [Participant 16].

However, it was noted by some that closer integration of clinical and research teams might provide necessary capacity and capability:

> *"I worry that we are all running and doing these tests sometimes five times on a diagnostic basis but they don't progress to a research phase because the services are too busy, separation of clinic and research, but there is huge opportunity in that gap ... I would love to set up a way research can add value because the [clinical genetic] services are not necessarily going to have the time or effort [to bridge the gap and make information available] ...this is where research can be really transformative"* [Participant 5].

It may also have clinical advantages for patients and clinical understanding of variant significance: *"Our data is held by us, and there are a lot of clinicians who have connections to research groups that are looking into potential new variants of significance"* [Participant 37].

"The beauty of an exome is that the data is always there." If new developments in research are made and the clinician notifies the group and asks them to look over an exome again in light of the new information, it can be done - *"data can be surveyed as many times as required based on what the clinician [wants us to look at and if patient consents]."* [Participant 37].

The dissolving boundary between clinical care and research is seen to raise challenges by some stakeholders. They are concerned to achieve common understanding of appropriate data flows between clinical team, research team, and service user.

For Aboriginal and Torres Strait Islander people, this dissolving boundary may pose a particular barrier to care, given the lack of trust extended to secondary use of data for any purpose, including research. One participant went so far as to suggest that it would be necessary to remove research from the conversation entirely, until trust and appropriate governance could be established:

> *"Research is the biggest barrier for the utilisation and enjoying the benefits coming from the new knowledge of genomics... you can't get one step along without someone saying "yeah but how do researchers get access to this without killing the whole debate?" I believe the whole thing has to be set up to be about healthcare and health management and we have to sideline the research agenda."* [Participant 66].

3.2 Data flows and participants

3.2.1 CLINICAL RELEVANCE AND RETURN OF RESULTS

Literature review

Interpretation of results and thresholds for clinical relevance receive considerable attention in the literature, with recognition of a growing amount of results with uncertain significance, uncertain prognostic indicators, or meanings that change over time. (Newson et al, 2016) Challenges in assessing the clinical relevance of a given genomic variant are heightened by the fact that many variants have been identified through research projects, (Cook-Deegan et al, 2017) or are very rare or novel. (Friedman et al, 2017) The question is, in what circumstances should uncertain results be fed back to patients and their families given associated burdens on the healthcare system and potential harms from false or uncertain information? (Friedman et al, 2017)

An ongoing tort action in the U.S. illustrates the issues that can arise with the reporting of results with false or uncertain clinical relevance. In *Williams v Quest Diagnostics* 353 F.Supp.3d 432, a mother brought claims for her son's wrongful death, among other actions in tort, based on the pathology laboratory's allegedly negligent diagnostic testing. The sequencing report indicated a mutation which the laboratory classified as a 'variant of uncertain significance' (VUS), however, the mutation was known at the time to be pathogenic. Allegedly as a result of this misclassification, the child received inappropriate treatment and died. It is unclear whether the misclassification resulted from an administrative error, a failure to update the reference database based on emerging knowledge or corruption of the reference database.

The reporting of a VUS is often presumed, in the literature, to be problematic, with the American College of Medical Genetics and American and Association for Molecular Pathology (ACMG-AMP), for example, recommending that 'VUS should not be used in clinical decision making'. (Richards et al, 2015) An emerging question is whether uncertainty can be explicitly incorporated into genomic practice, with uncertainty directly acknowledged and appraised. (Newson et al, 2016)

Stakeholder consultation

Stakeholders noted that the clinical significance of genetic variation can be unclear. The nature and degree of uncertainty may not be fully appreciated and can result in false expectations or incorrect interpretation of results: *"Understanding of the results and correct interpretation is something we constantly need to be mindful of"* [Participant 30].

The correct approach to defining clinical relevance was unclear to some stakeholders: *"When does an association between a disease and a gene become sufficiently robust to need to be reported?"* [Participant 2].

Also unclear was the responsibility of researchers when results *are* perceived to be clinically relevant: *"How do you define [clinically relevant information] and as a research group, how do you [return clinically relevant information to participants] adequately? Where do you get the funding to do that? How do you follow people up?"* [Participant 34].

In particular, there was perceived to be a lack of consistency with regard to the *authorisation* of return of results: *"[Although there are guidelines that] go back to the treating clinician but it is not clear or straightforward so you have to go through all your [ethics] committees and I am not sure you'd get the same answer across the board"*. [Participant 19].

Concern was expressed in relation to the burden on clinical care teams if results were returned via clinicians, particularly in relation to large studies:[1] *"We are dealing with situations where we have done [a large international study on thousands of children] with neurodevelopmental disabilities … how are these services going to absorb 260 meetings, consultations, testing [for the hypothetical 260 kids who have results].?"* [Participant 5].

The same participant commented: *"I don't like when research data is not returned to participants…[Clinical genetic] services are overworked so we really have to be very creative [with regard to] how to give research data back"* [Participant 5].

Another noting that *"our models [of returning clinically-relevant genetic results] are evolving"*. It is a *"challenging field"* that requires investment to ensure researchers are doing it properly [Participant 34].

A specific concern was expressed in relation to the return of prenatal testing results. One stakeholder suggested that critics of return of genetic results argue that prenatal testing results in the *"termination of children that you do not like being born."* However, the participant fervently disagreed and stated instead prenatal testing has given mothers the *"major outcome [of] reproductive confidence to have healthy children"*. They suggested that the aim of such testing and return of results is to *"empower people, to remove a lot of worries from that space to reassure them that they can have healthy families"* [Participant 5].

Indigenous stakeholders noted that this issue was particularly fraught for Aboriginal and Torres Strait Islander patients, for whom the lack of reference data meant that it was difficult to determine which variants were pathogenic, and that there was a consequent increase in VUSs. As Participant 66 observed, *"you cannot apply the same degree of certainty to a predictive outcome to Aboriginal and Torres Strait Islanders as some of the Caucasian, North American and Western European descendants. We do not know what is a pathological mutation and what is not."*

3.2.2 INCIDENTAL OR ADDITIONAL FINDINGS

Literature review

Genomic sequencing through clinical practice or research can reveal 'incidental' findings that are of potential health or reproductive significance to patients and participants. Most agree that clinicians and researchers have some obligation to disclose at least some genetic findings, provided there is consent for such return. The precise scope of any obligation, however, remains unclear. (Eckstein et al, 2014)

In the clinical context, the American College of Medical Genetics and Genomics issued guidelines, in 2015, recommending that laboratories performing clinical sequencing seek and report on a 'minimum list' of variants assessed as having a high degree of clinical validity and utility. (Green et al, 2015) At least some scholars also argue in favour of such an obligation on genomic researchers. (Koplin et al, 2020) However, in Australia and internationally, there is by no means a consensus on this approach to return of results. Other organisations including the Australian National Pathology Accreditation Advisory Council call for more conservative approaches. (Eckstein and Otlowski, 2018)

The NHMRC *National Statement on Ethical Conduct in Human Research* (2007) has been substantially updated to include a specific 'decision tree' for researchers and clinicians making decisions whether to return a genomic result. The *National Statement* specifies that there generally is no obligation on researchers to look at or assess findings outside of the scope of the research.

1 Referring to the overburdened clinical genetic services - in SA it takes up to 2 years to get an appointment with a clinical geneticist unless the case is urgent [Participant 5]

Stakeholder consultation

Stakeholder comments suggest that concerns raised in relation to clinical relevance and return of results generally may be particularly acute in case of additional or incidental findings. The lack of a national approach to return of additional findings was suggested to be *"a big challenge"* [Participant 18].: *"Australia does not handle the issue of incidental findings well - this will become an economical as well as ethico-legal problem - what percentage risk of developing a disease do we deem high enough to warrant additional screening (and therefore additional funding?)"*. [Participant 12].

Challenges were raised in particular in relation to the ability to obtain a meaningful consent. In a healthcare context participants are likely to have a *"singular focus on diagnosis"* rendering other issues/concerns *"secondary"* to diagnosis and therefore not a big concern for them [Participant 6].

Also, the lapse of time between original consent and identifying additional findings, can be an issue. One stakeholder suggested people may tick at the time of signing a consent form that they want to know incidental findings, but this *"can be terribly confronting later"* [Participant 13].

Presumably, this concern will be particularly pressing if clinical significance is only understood some time later. It was noted that the validity of consent regarding incidental findings in a research context needed to be established at two time points: at the time of joining the research, and in the event of clinically significant findings. The passage of time allowed participants to *"change [their] mind"* [Participant 36].

It was noted that between the clear extremes, referred to as the "black and white" scenarios where either (a) the link is well established, and the finding should be reported, or (b) *"you are researching a gene to find out whether it is associated [with a disease] it is probably not worth reporting back"* [Participant 2], there is a significant grey area. As well as the challenge of determining what results to return because of uncertainties regarding clinical significance and validity of ongoing consent, there were concerns raised in relation to re-analysis and re-contact.

3.2.3 RE-ANALYSIS AND RE-CONTACT

Literature review

Rapidly changing understandings of genomic information raise questions about the duties of clinicians and researchers to provide patients and participants with updated information, for example, to alert them to a variant previously reported as a VUS that is now known to be pathogenic. An obligation to update genetic databases to reflect new understanding was one of the issues in the U.S. case of *Williams v Quest Diagnostics* 353 F.Supp.3d 432,referred to in 3.1.1.

A 2019 position statement developed by the American Society of Human Genetics and endorsed by the Human Genetics Society of Australasia advises that reinterpretation and recontact will be required in some instances of genomics research. The obligation will be strongest when the research project is active and ongoing; the potential for recontact has been raised in the informed consent; the new interpretation has a high degree of certainty; and the reinterpretation would be relevant to the condition under study or would change medical management. (Bombard et al, 2019)

Stakeholder consultation

Concerns are associated with the fact that understanding of clinical significance can change over time. One stakeholder noted that there are issues associated with missing key information at the time of testing (which is found out later) and subsequent disclosure of that information to patients when available. This was likened to the need to trace and contact patients when a previously untreatable disease becomes treatable [Participant 25].

The fact that *"[Genomics] is a very dynamically changing space ... is another complexity in addition to the questions of consent, disclosure of information, return of results"* [Participant 5]. Some research will "discover novel genetic determinants every day". This makes it possible to test for things that *"patients could not have been tested for [and/or] some existing genes that would have been missed by previous technology"* [Participant 5].

One stakeholder noted that there are situations where we find nothing and report to a family, but then something is published which highlights a notable result upon re-analysis and we need to notify families again - some families deal with these complexities well and others don't [Participant 5].

3.2.4 RELEASE OF RAW SEQUENCE DATA TO PARTICIPANTS

Views differ on the ethical and legal issues associated with releasing raw genomic sequence data to patients and participants. As noted previously, the *National Statement* expressly guards against any duty to return raw data to participants. However, to the extent that raw data satisfies definitions of 'personal information', this may conflict with Access principles under Australian privacy acts.

Potential research participants have expressed considerable interest in receiving raw sequence data, including to use it to seek out their own clinical interpretations. (Middleton et al, 2015) Participants also appear to attach intrinsic value to, and 'ownership' of, their genomic data. On the other hand, scholars raise concerns about the accuracy and utility of raw sequence data, and associated consequences. (Thorogood, 2018) Only a limited number of genomic research projects currently provide, or plan to provide, participants with raw data. (Thorogood, 2018) However, given rights to access personal health information under privacy laws in Australian and elsewhere, scholars argue that:

> *"it is likely that clinical laboratories have, or will soon have, a legal obligation to provide individuals their raw genomic data upon request. While it is less likely that a legal right applies in research contexts, we propose that projects should still consider providing a default right of participants to access their own individual-level genomic data upon request."* (Thorogood, 2018)

Stakeholder consultation

Concerns were expressed for privacy and ethical issues to be understood in relation to release of primary data to patients and research participants. One stakeholder noted that the law department at their institute deals with a lot of genomic information transfer cases. In the relevant institute, the practice is for genomic information to be transferred to custodians of the information (but not parents of children). Transfer requires payment and an understanding of privacy and ethical issues outlined by the information sheet [Participant 9].

3.2.5 DATA OF DECEASED PEOPLE AND DISCLOSURE TO FAMILY MEMBERS

Literature review

Samples suitable for genomic testing may be procured from deceased individuals, including biospecimens originally sourced from a living donor but retained after death. (McWhirter & Otlowski, 2016) These samples raise questions about whose consent, if any, should be sought for testing and the consequences of a failure to seek consent. McWhirter and Otlowski note, for example, that a senior next-of-kin might share no genetic information with a deceased person. Should their decision for testing be in preference to a blood relation for whom a decision to test may present potential risks? Approaching relatives of recently deceased persons for permission to test can also cause distress, meaning that researchers commonly seek waivers of consent in such situations. (McWhirter & Otlowski, 2016) Consent to genomic testing and the disclosure of genomic information is further complicated by the lack of privacy protection for deceased persons under some (but not all) Australian privacy acts. (McWhirter & Otlowski, 2016)

A related issue is the disclosure of genomic information to a next-of-kin. This may occur when a deceased person has been a participant in a research study that is reporting results. In this situation, a next-of-kin might not be aware that the deceased person has participated in genomic research or otherwise sought genetic testing. Nina Hallowell and her colleagues report on the experience of disclosing genetic test results to the next of kin for participants in the Australian Ovarian Cancer study. Some relatives experienced distress when they first learned of results, saying 'that they were not sure that they wanted to know this information about themselves'. The researchers suggest obtaining consent or notifying next-of-kin about genomic studies in which results might be generated in the future and/or participants may die before their release. (Halloway et al, 2013) In returning results to a next-of-kin from a cancer research study, Amber Johns and colleagues noted the applicability of the provision in the *Privacy Act 1988* (Cth), which allows healthcare professionals to communicate with family members, without consent of a person under their care, if a medical practitioner considers there is a risk of harm to the family member.

A person's relatives may also request access to genomic information after that person's death, raising questions about any right to information in this scenario. In its 2010 review of Australian privacy laws, the Australian Law Reform Commission recommended that the *Privacy Act 1988* (Cth) should be amended to require organisations to provide third parties with access to the personal information of deceased individuals, except to the extent that providing such access would have an unreasonable impact on the privacy of others, including the deceased individual. Some state privacy acts, for example the *Health Records Act 2001* (Vic), specifically pass on a right of access to a deceased person's relatives. Ethically, disclosure may have benefits for a deceased person's relatives. This must be balanced against any known wishes of the deceased for non-disclosure and the burden of disclosure on a research team. (Chan et al, 2012)

Stakeholder consultation

An issue of concern for a few participants related to the rights of family members to access genomic information relating to the deceased [Participant 16]. One commented that an issue of concern to them was the *"[r]ights people have to access their ancestral genetic info and phenotypic data"* [Participant 25]. They suggested that this was a *"challenge area that will become more apparent"* [Participant 25].

Another put this challenge in terms of ownership and management of personal genomic information and queried whether ownership passed to heirs, siblings and other relatives: *"how is consent handled for people who are no longer around?"* [Participant 16].

3.3 Legal

3.3.1 LEGISLATION NOT FIT FOR GENOMICS

Literature review

The Australian Law Reform Commission, *Essentially Yours* recommended addressing issues raised by genetic testing through changes to existing legal frameworks, rather than through the development of new regulatory frameworks specifically for the protection of genetic information. This differs from the approach taken, for example, in the United States, which enacted the *Genetic Information Nondiscrimination Act* (GINA) (2008) to specifically protect against genetic discrimination. It is notable, however, that GINA provides a level of protection against discrimination in the areas of health insurance and employment (and not life insurance).

Stakeholder consultation

Divergent views were heard in relation to the adequacy of existing law to address the use of genomic data in care and research settings. It was a concern expressed by at least one stakeholder that *"Australia doesn't recognise the importance of a consistent bespoke genetics scheme"* [Participant 3]. In particular, their concern was in relation to lack of *"a coherent, cohesive genetic privacy or non-discrimination legislative scheme"* [Participant 3]. One participant from the insurance sector saw the value of a specific legislative response (although it should be noted that they mistakenly attributed life insurance protection to GINA):

> *"Australia should look to Europe or the US. The US has the GINA policy. This clearly states that you can't use this genetic information. That's life insurance. I think that's important and we're missing something in not having that because currently we just have a moratorium. People with genomic and genetic diseases, the information that we get and the small numbers of people would not have a massive effect on the cost of life insurance."*

The advantage of bespoke legislation was seen to be, at least in part, the clear guidance this may provide to clinicians. For example, clearer legal requirements regarding sharing results with family members will benefit clinicians who may lack knowledge. [Participant 3]. This was considered important as genetic information becomes more mainstream: more clinicians will be encountering it, and *"it won't just be a genetics professional sitting there who knows what to do"* [Participant 3]. On the other hand, others expressed a contrary view, suggesting that there are already *"a lot of [legal] safety nets in place"* [Participant 26] and that *"99% of all issues are actually handled by the legislative frameworks that we have in place today"* [Participant 26]. It was said that though they *"are crude and slow us down ... in general those protections by and large are there in the system"* [Participant 26].

Rather than it being a problem with law, it was suggested to be a problem of coordination and consistent understanding: *"Having better coordination between the ethics community, legal community, and practitioners is a larger hurdle than being able to share a few laws to be able to share data"* [Participant 12].

For example, it was suggested that dynamic consent is needed, and implementing it will be an operational rather than legal problem, with financial implications [Participant 12]: *"At the end of the day I do not think there are laws and regulations that hinder us, it is a case of a lot of groups trying to have to come together to work on things like a standard consent form or consent process and to ensure people have the ability to understand this but to also change their mind (dynamic consent)"* [Participant 12].

3.3.2 RISK AVERSE PRACTICES DUE TO LEGAL COMPLEXITY

Literature review

Unclear and inaccessible legal standards can leave research participants with inadequate protections and can inhibit the progress of research. In England and Wales, the laws surrounding genomic databases have been described as 'highly complex, confusing, uncoordinated, and inadequate'. (Gibbons et al, 2007) A similar complexity is likely to apply in Australia, both for genomic databases and other sources of genomic information.

There is no literature available assessing the prevalence of risk-averse practices in Australia when it comes to sharing genomic information, or the link between such practices and the available legal framework. However, Australia's complex privacy framework has been posited as contributing towards unnecessary limitations on information sharing and a risk-averse culture in the child protection context. (Adams & Lee-Jones, 2017)

Stakeholder consultation

Stakeholders attributed the rise of risk-averse practices across states to legal complexity around genomic data. Participant 16 highlighted that there is a *"general sense of uncertainty around what the law requires"*, and other stakeholders suggested that the uncertainty is evidenced by varying interpretations of laws across data custodians: *"One of the bigger problems is some data custodians' interpretation of the law"* [Participant 34].

> *"I think it is the Commonwealth giving a directive that the states can then get involved, and there is a lack of consistency across the nation in how it is administered"* [Participant 9].

Participants expressed disappointment when describing cases where genetic information is *"silo[ed]"* and that entities become *"defensive with [genomic] information"* due to uncertainty [Participant 19]. One stakeholder expressed that *"the government is making it more difficult to access other data sets... because it is their interpretation and conservative approach to ticking boxes"*, which is *"restrictive"* to research progress [Participant 34]. Comments suggested that the effects of such risk-averse practices are being experienced within research and clinical communities:

> *"In the past 12 months, post-Cambridge Analytica, quite a few researchers including myself have noticed a trend towards medical institutions and other groups also becoming quite conservative in the way they are managing flow of data."* [Participant 16].

As well as adverse media coverage, this may be related to changes in European data protection law and the General Data Protection Regulation (GDPR) (discussed below). One participant commented that:

> *"We are living it at the moment where we have a block of data where we are doing crazy things like sending the Americans to work at the university in the Netherlands to do the analysis there so the American data can come across but the European data does not leave the EU."* [Participant 25].

One stakeholder reflected on the wasteful impacts of such practices, where data collected from research is not made available due to ethics committees precluding access. *"[These situations are] concerning because the information and knowledge wasn't around perhaps at that time when some of these studies were being developed, and does that mean we are wasting large cohorts of datasets?"* [Participant 16].

In addition to hampering research progress, one stakeholder identified the potential for such risk-averse practices to pose a risk to patient welfare: *"You are protecting [patient] identity but [potentially] putting them at risk of inappropriate treatment because [of the] covering up of data, [and introducing potential risk of being unsure of data-patient matches when needing to identify patients]."* [Participant 16].

3.3.3 ALIGNMENT WITH OVERSEAS LAWS

Literature review

The globalised nature of genomic testing and information sharing can raise questions about the alignment between Australian and overseas laws and regulations. The extent to which non-alignment is posing challenges in practice is unclear. In one international survey of cancer clinical sample sequencing initiatives, privacy/ethics issues and international legislation generally were not considered significant barriers to sharing. Some concerns were raised, however, about legislative barriers in specific regions, such as Europe. (Vis et al, 2017)

One option that has been suggested to ameliorate barriers posed by nonalignment of laws is federated data sharing approaches, in which data is mined *in situ*, rather than being transferred to a different jurisdiction. (Vis et al, 2017)

Consent is a common justification for the sharing of personal genomic information under Australian privacy laws. While this remains the case under Australian laws, the introduction of the European Union GDPR suggests a move away from consent as the primary paradigm for disclosure to satisfaction of a public interest test. (Taylor et al, 2018)

Stakeholder consultation

Participants presented varying viewpoints regarding international approaches to the management and sharing of genomic information. One participant pointed to overseas examples from which Australia should take heed, such as the *"more proactive"* approach of European nations: *"In terms of genetics a lot of European countries for their size and weight have overtaken [Australia]... because they are putting money into this...they pass laws"* [Participant 3]. They also referenced the Estonian biobank, and commended the willingness of government *"to put money into"* the *"huge scale effort"*, as well as the Biobank Regulatory Scheme which provides the public with *"confidence"* that *"employers and insurers can't [access their data]."* [Participant 3]. Another suggested that Australia "needs to ride the wave with the Americans" [Participant 34].

In their discussion of the European Union GDPR, stakeholders identified existing inadequacies in Australian law, and the potential for the GDPR to have both beneficial and detrimental impacts for Australia. One stakeholder believed the stricter nature of the GDPR has *"raised the bar"* and provoked thought in Australia, however *"we just haven't mobilised"* [Participant 19]. Another participant highlighted that Australia's approach to data protection is *"vastly inadequate"* as it stands and indicated support for the Therapeutic Goods Administration (TGA) moving towards a European approach to data protection [Participant 3]. One stakeholder emphasized the importance of *"guidance"* for Australia in the wake of the GDPR, as there are *"still a lot of unknowns given that the European requirements are narrower and stricter than what is currently in Australia"* [Participant 16]. A participant viewed such guidance as essential for researchers to be able to *"move data internationally... further down the track"* [Participant 16]. This included guidance on how to meet responsibilities in relation to an individual's right to erasure and data deletion across different sites.

One participant indicated restrictions to international research collaboration as a consequence of *"the way some countries have interpreted [the GDPR]."*. The stakeholder expressed that these responses to the GDPR are creating *"an impasse between the U.S. and Europe at the moment, meaning new data agreements aren't being signed...it is stifling research"* [Participant 34].

One stakeholder provided the example of Australia looking to the U.S. for guidance regarding non-discrimination laws. They mentioned that Australia already has such laws, and following in the footsteps of the U.S. may not be an easy or relevant solution for Australia. The stakeholder expressed concerns regarding the propensity for some nations to follow international legal reforms without an understanding of the context of the developments and their applicability to Australia [Participant 17].

3.3.4 INCONSISTENCY BETWEEN LAWS OF THE STATES AND THE COMMONWEALTH

Literature review

Interjurisdictional issues can also arise in cases of sharing genomic information between Australian States and Territories. Australia's patchwork of federal and state and territory privacy laws can lead to areas of over- and under-regulation along with potential inconsistencies. (Eckstein et al, 2018) For example, the *Privacy Act 1988* (Cth) permits the sharing of personal information for the purposes of research if an HREC determines that the use or disclosure is 'necessary' for the research, the research activity would have an impact on or provide information about public health or public safety, and the research activity could not be achieved through the sharing of de-identified information. Although most states and territory privacy acts include similar provisions, no research exception is on the books in the South Australian administrative instruction or the Australian Capital Territory Act (Eckstein et al, 2018). Challenges raised by different legal provisions may be exacerbated by differing interpretations by HRECs responsible for authorising disclosure decisions.

In accordance with *Essentially Yours*, the *Privacy Act 1988* (Cth) has been amended to remove the requirement for a health threat to be imminent in order for disclosure to be permitted to lessen or prevent a serious and imminent threat to an individual's life health and safety. However, some state and territory privacy laws retain the imminence requirements, for example, the *Health Records Act 2001* (Vic).

Stakeholder consultation

Stakeholders highlighted that the federated model in Australia has resulted in inconsistent laws across states, with adverse consequences for genomic research. One stakeholder referred to directives from the Commonwealth which can result in *"inconsistency because of the states"* [Participant 9]. Another stakeholder commented on difficulties with moving data between states: *"if we get information from other states we have to negotiate jurisdictional boundaries"* [Participant 19]. One participant remarked that *"the government is making it more difficult to access other data sets"* and this is evidenced by the actions of *"some state jurisdictions around access to cancer data"* [Participant 34]. With regard to participant follow-up on cancer status information, the stakeholder noted that *"each state and territory data custodian has to consent... nationally and it varies by state"* [Participant 34].

Participant 37 advocated for the development of *"national guidelines and a national system where everyone is sharing at the same time in the same place"*. They emphasised the need for *"an overarching system for the whole nation... [because] at the moment everything is in pieces"* [Participant 37]. However, another participant pointed to potential difficulties linked to the development of a national model. They identified problems with creating generalised guidelines and regulations in relation to research and clinical care for Aboriginal and Torres Strait Islander peoples, given the differences across Australia:

> *"If you actually want to see something happen at a local level or regional level you need to be sensitive to the politics and the service environment that is at that level. It is great to come up with national frameworks, policies and guidelines and the like but in order to operationalise them at a local or jurisdictional or state level they actually have to have sensitivity to whatever the policies are for the public and hospital and health providers in the space and also the patient services and things like access and whether there are any peak bodies operating in the space."* [Participant 42].

3.3.5 REGULATION OF TESTING AND IN VITRO DEVICES BY THE NATIONAL ASSOCIATION OF TESTING AUTHORITIES AND THE THERAPEUTIC GOODS ADMINISTRATION (TGA)

Literature review

The regulatory environment for genetic testing in Australia has undergone significant change since the release of *Essentially Yours*. The TGA's in vitro devices (IVD) regime was extended to in-house testing through a 2010 amendment to the *Therapeutic Goods Act 1989* (Cth). However, the regulatory implications for direct-to-consumer genetic testing remain uncertain, particularly with regard to overseas suppliers. (Nicol and Hagger, 2015)

There is a significant impact of having diverse national frameworks governing genetic laboratories, such as U.S. legislation requiring that all laboratories that analyse samples from U.S. citizens must be certified under the Clinical Laboratory Improvement Amendments (CLIA) by the U.S. Food and Drug Administration. In Australia, only laboratories accredited under the *Health Insurance Act 1973* (Cth) may seek reimbursement of their expenses through Medicare. The result of different national standards is interference with uniformity and the internationalisation of genetic services. (Tasse et al, 2009)

Stakeholder consultation

Stakeholders made little reference to the accreditation and registration requirements for genetic testing. Specifically, the main requirements are accreditation of laboratories through the National Association of Testing Authorities (NATA) and registration of genetic tests through the in vitro devices regime under the *Therapeutic Goods Act 1989* (Cth), administered by the TGA. However, one stakeholder did express knowledge of the TGA regime in general, and the current review of Therapeutic Goods Regulations relating to direct-to-consumer genetic testing in particular, stating:

> *"The government is reviewing the regulations around the Therapeutic Goods Administration (TGA) DTC testing ... genetic testing is shoved [sic] into the IVD, a genetic test as it stands is a "device" which becomes very problematic when you try and tease out the lab based procedure and all of the interpretation and reporting that comes out of that because they are not separately regulated. The test itself is regulated as one item. Very tricky."* [Participant 3].

4 Research

4.1 Standards and terminology

4.1.1 STANDARD FOR CONSENT/WAIVER OF CONSENT/OPT-OUT

Literature review

The use or disclosure of genomic information that satisfies the definition of 'personal information' under privacy acts usually requires the consent of the person to whom the information relates. There are ongoing debates in the literature about the legal and ethical validity of various consent models, particularly when it comes to the future use of biospecimens or genomic data, as explained in 1.1.1.

Further questions arise where information or specimens have been obtained on the basis of opt-out consent, or where no consent has been provided that covers sharing activities. In empirical research, community members in the U.S. did not consider opt-in consent to be adequate for inclusion in large data repositories (in this research, defined as a 'medical information commons'). They instead favoured a 'dynamic or more granular consent' that involved 'both opting-in to participation and maintaining some individual control over data uses, coupled with knowing how their data are being used and governance structures that ensure fair and meaningful representation'. (McGuire et al, 2019)

Under s 95A of the *Privacy Act 1988* (Cth) and the *National Statement on Ethical Conduct in Human Research* (2007), an HREC may approve a waiver of consent for the use of personal information in research relevant to public health or public safety where there is deemed to be a sufficient public interest in the research, it is impracticable to gain consent and there is no known or likely reason that participants would not have consented if they had been asked. However, further work is needed to assess the circumstances in which a waiver of consent should be granted, including thresholds for 'impracticability' of gaining consent. (Eckstein et al, 2018)

Stakeholder consultation

One researcher we interviewed reported on discomfort about proceeding with the re-use and gathering together of samples that had been collected years earlier on the basis of opt-out consent (with a public notice placed in the newspaper) but felt the alternative of reconsenting a thousand participants would be impractical [Participant 25]. Different ethics committees across Australia that were responsible for different arms of the research took diverse views as to the appropriateness of a waiver of consent for this research.

Indigenous stakeholders reported significant discomfort with any use of waivers of consent and broad consent models in relation to Indigenous genomic data, given the lack of trust among Aboriginal and Torres Strait Islander communities (see sections 1.1.1, 1.2.1 and 1.2.4).

4.1.2 HUMAN RESEARCH ETHICS COMMITTEES AND NEW FORMS OF GOVERNANCE

Literature review

Evident from the above discussion is the pivotal role of Human Research Ethics Committees (HRECs) in Australia when it comes to authorising the use and disclosure of genomic information. The question is whether HRECs are equipped and resourced to perform this role? Scholars assessing HRECs and their international equivalents note that these Committees receive little guidance when it comes to the ethical concerns raised by data-intensive projects. (Dove et al, 2016) Moreover, they stress that reviews of data-sharing research adds to "a system already under fire for subjecting multisite research to replicate ethics reviews". (Dove et al, 2016) The replication of HREC review in multisite genomics research can result in "piece-meal decision-making [that] may not be in accordance with what individuals, groups, and communities want in terms of international data sharing". (Kaye et al, 2018) Recognition of these challenges for HRECs internationally has led to calls for streamlining the ethics review process, including through mutual recognition of ethical review from another jurisdiction. (Townend et al, 2016)

More recently, additional forms of governance have been introduced to sit alongside HRECs, such as Data Access Committees, which many research platforms have established to provide approval for access to genomic data. (Kaye et al, 2018)

Stakeholder consultation

Variation in both the expertise and views of different ethics committees was identified as a problem by several participants. It *"doesn't make sense that five different ethics committees have five different opinions"* [Participant 5]. Participant 19 agreed, noting that not every ethics committee would have the capacity to understand the large data sets that the particular research organisation was dealing with, and fearing that when advice was sought from different committees on whether and how to communicate clinically-relevant findings to research participants, *"I am not sure you'd get the same answer across the board"*. National harmonisation, for instance of consent materials, was not considered to be a suitable solution to this problem because committees would still require amendment to fit local circumstances. Participant 25, a researcher, noted that *"With genetics, most families live around the country so I would have a patient we recruited through [two different hospitals in the same state] - different ethics committees, demanding different things, families needing to read different info from different institutions."* Another researcher, Participant 5, reported on the significant expense of needing to employ someone to manage the ethics applications, and the lengthy delays that can precede ethics approvals.

Aboriginal and Torres Strait Islander stakeholders similarly noted both the time and variation inherent in multisite approval processes: *"The big thing is ethics – there are so many different ethics groups to go through."* [Participant 41]. Several stakeholders expressed concern that HRECs with Indigenous sub-committees were being used as proxies for Indigenous community approval, and noted that it was challenging for HRECs to be representative of the diversity of communities in any given area. *"People use the ethics committees now as a representative of the communities. They are representative of you doing ethical research but not of the community"* [Participant 53].

4.1.3 RE-IDENTIFICATION AND UNDERSTANDING OF TERMINOLOGY (DE-IDENTIFY, ANONYMISE ETC)

Literature review

Much genomic research has proceeded based on assurances of de-identification, with a view that this was the most appropriate strategy for protecting participants. Technological advances mean that de-identification may not secure anonymisation, nor be ethically optimal. It is now possible to infer the identity of some individuals based on their whole-genome sequences along with other publicly available information (Gymrek et al., 2013) This raises concerns that purported de-identification or coding of personal data may not provide sufficient privacy protections for research participants. (Hansson et al, 2016)

There are also growing benefits of maintaining the identifiability of genomic samples. From a scientific perspective, it allows additional data linkage opportunities. For individual participants, it allows the return of results of genomic research, including results with clinical value. (Hansson et al, 2016)

For these reasons, scholars suggest a move away from deidentification towards 'the adoption of governance frameworks, security measures, and standards (i.e., data management/access policies, Privacy Preserving Record Linkage or unique identifying systems)'. (Nguyen et al, 2019)

Stakeholder consultation

The proliferation of data in different locations that are subject to varying de-identification techniques is perceived as complex and as presenting risks to data subjects' privacy: *"if someone wants to find [a person's] data they will find a way"* [Participant 13]. Participant 6 viewed anonymisation as insufficient to protect people: *"[Data can be] anonymised but even by being anonymised it is pretty specific data, it can be found again down the line...by current standards it might be not technically possible to identify me but who knows what will happen in the future?"* [Participant 6].

One stakeholder also spoke directly about the confusion in the use and definitions of key terms, noting the:

> *"lack of understanding of a lot of terminologies in relation to data sharing for genomics, in particular [terms] like identified, re-identifiable, de-identified, anonymised... what do these terms actually mean?...I think they have different implied meanings in different parts of the laws and different policies... I find [trying to interpret and understand these terms and laws] incredibly challenging"* [Participant 11].

4.1.4 OPEN PUBLICATION OF DATA AS A REQUIREMENT OF A PUBLISHER OR FUNDING BODY

Literature review

Data sharing has become a requirement for publishing genomic research as well as for some research funding programs. This is well illustrated by the U.S. National Center for Biotechnology Information's Database of Genotypes and Phenotypes (dbGap), which was established in 2006. Following modifications of the NIH policy for data access in 2014, data sharing was required for research conducted or funded by all NIH institutes. The policy further stipulated that researchers should seek broad consent from participants for such data sharing. (Cook-Deegan et al, 2017)

Stakeholder consultation

Participant 34, who leads a research division, noted that participant privacy is at odds with the requirements of funding bodies and publishers that data are made publicly available. For instance the US National Institutes for Health has a blanket requirement for data release as a condition of funding. Publisher requirements for data release may impede the publication of valuable research findings when participants have not consented for the release, or data cannot be suitably de-identified. The complexity of determining whether a particular data set can or cannot be released, particularly once it has been shared to third parties, is problematic and resource intensive to manage.

This concern was echoed by Indigenous stakeholders, who viewed this requirement as a barrier to publication in some journals. Participant 41 noted that, with respect to data from Aboriginal and Torres Strait Islander participants: *"Another issue I was going to say was you know when you go to publish, and you publish in Nature or whatever and you have to make the data open? That is where we are stuck at the moment. We aren't entirely sure what to do with that - [is it enough] so long as it's deposited somewhere?"* There was a strong feeling that providing open access to data would destroy trust from participating communities.

4.1.5 DATA PARITY, INTEROPERABILITY, QUALITY

Literature review

A key impediment to data sharing comes from the quality and interoperability of genomic data. One international survey of cancer clinical sample sequencing initiatives reported that data harmonisation and bioinformatics concerns, such as a lack of interoperability, were the largest perceived barriers to international data sharing. (Vis et al, 2017) Others have noted that much of the data available for sharing is of such low quality that the data cannot be trusted. (McGuire et al, 2019)

Stakeholder consultation

First, it was noted that data quality may of itself be poor: *"...it is not the case that all data that goes in is fit to be shared...maybe some data going through is cell lines for quality purposes, maybe some isn't actually patient data"* [Participant 11].

This participant, who has systems expertise, noted that standards for storage, sharing and discoverability were also lacking: *"There is still no actual standard for the structure of storing genomic data...[we] don't have standards for APIs [Application Programming Interfaces] so how will we share genomic data between states? How are we going to know what is discoverable? There is a whole lot of work to be done in this space"* [Participant 11].

The need to have high-quality data to support the genomic data itself, such as metadata and clinical data, was discussed by several participants. Participant 11 noted the importance of having meta-data such as the type of sample, the reason for the test, the test location, as well as audit data such as access logs - areas in which international standards are lacking. Participant 19 noted that genomic information alone is not very useful and that associated clinical information is needed, but queried *"Where is the repository that keeps all that info, how do people access that info?...There will be a lot of data linking, big data sets all over the place, who decides what people can link and who controls it; how do you control the quality?"*

4.1.6 PLATFORMS FOR SHARING

Literature review

The design and development of sharing platforms has generally come through 'academia, pharmaceutical companies, clinical health IT systems, funders, insurers, and policy-makers'. (Kaye et al, 2018) Some initiatives for data generation and sharing have also come through patient organisations, especially in the context of rare diseases. (Kaye et al, 2018)

This raises the question of what are the required features of a sharing platform, with key attributes recognised as a participant-centred approach and trustworthiness. (McGuire et al, 2019) In addition, sharing platforms may have obligations to supplement available legal frameworks, for example, by establishing sanctions for unauthorised re-identification of data. (McGuire et al, 2019)

Stakeholder consultation

The consultation identified current technological challenges in sharing platforms, including in data quality (see 4.1.5) and metadata. *"We are lacking national identifiers and family identifiers and a whole lot of basic digital health infrastructure technology that needs to be there as an enabler for streamlined genomic data sharing, particularly for clinical use"* [Participant 11]. Questions also arose about whether particular data can legally be shared. Participant 18 said: *"maybe a patient's data is going through but they are subject to some criminal investigation and despite the patient's consent for research use the lab isn't in a legal position to even share that data"*.

Sharing data across state borders was identified as problematic where it needs to be seamless, with several participants commenting on diverse state laws for sharing and publication. Participant 11 said *"we are going to have challenges in that someone might come along and say, 'great I'm going to take the 5000 people with deafness from across all of Australia and do a big study on it... well what if none of that data can be taken out of any of those jurisdictions?"* . It was also noted that the lack of a national cancer registry was *"seriously clunky and a big problem"* [Participant 19]. Going beyond this, international sharing was foreshadowed: *"if further down the track there are plans to share data, it is quite important that groups around the world pool their information to get the best research out, in those cases you are likely to be sharing that data even in common disease types where there is desire to pool data from hundreds of thousands of patients around the globe"*. [Participant 16].

Tracking data flows from shared platforms in a way that would accord with, for example, the requirements of the GDPR in Europe, was also a technological challenge. [Participant 34]. Sometimes it was easier to share whole datasets instead of *"slicing and dicing"* the data on a few specific genes, meaning that more data is shared than is strictly necessary *"and that is not optimal either"*. [Participant 34]. *"Models like the variant sharing platform [are needed] that allow labs, if they get a variant in a disease they have not seen before, to query other databases to see if other labs have seen it"*. [Participant 18].

Participant 9 advocates for a potential *"national storehouse"* of genomic information, broken down into healthy genomes, and specific disease genomes: *"[With] greater volumes of genomic information, we need a much better and central way as to how to manage and re-use that information in the future..."* [Participant 9]. Annotation of genomic data with clinical information was also needed [Participant 19], and the storage of and access to that data would require further consideration.

Finally, governance of platforms for genomic data sharing was noted by several participants as being critical. As Participant 19 stated:

> *"It would be nice to make sure other researchers could access the information, that there is some governance around it that makes that data findable, accessible, interoperable...Where is the governance?... Who is going to be overseeing access to these datasets?...Currently it is largely ethics committees, but...all the intricacies of these different data sets [are] a lot to expect every ethics committee to know about"*

4.1.7 SECURITY AND HACKING

Literature review

Genomic research participants have raised concerns about what legal recourse may be available if their data is misused. (McGuire et al, 2019) Robust data security measures are a necessary component of data sharing platforms and genomic research activities. However, increasingly common and necessary technologies for such activities, such as cloud computing, raise challenging legal and ethical questions. Cloud computing may lead to data being stored in multiple jurisdictions, and the transfer of data from one jurisdiction to another happening without researcher knowledge. In these circumstances, can and should researchers be held legally responsible for the storage of such data? (Charlebois, 2016)

Stakeholder consultation

A substantial number of interviewed stakeholders commented on the significance of data security risks in terms of causing individual harm and affecting public confidence in genomic information use. One patient advocate described it as their *"number one"* issue [Participant 13] while a genetics counsellor and researcher said that *"Data breaches are a really big deal in relation to genetic data"* [Participant 3].

Some stakeholders considered data breaches via hacking were inevitable: *"if someone wants to find data they will find a way"* [Participant 13]. Recent cybersecurity breaches involving Australian hospitals were cited [eg. Participant 19]. Breaches of genomic data were considered to present a significant risk to individuals because a person's genome is permanent and inherently identifiable: *"Genetic data has a long lifespan in terms of how bad a data breach can be"* [Participant 3]. Even if the data are anonymised *"it is pretty specific data, it can be found again down the line so I think all those standard concerns about where your data will end up, who will access it, how secure can we ensure it is, how by current standards it might be not technically possible to identify me but who knows what will happen in the future?"* [Participant 6]. However one participant with international expertise noted that although the risk exists, there is a *"global effort in terms of designing to minimise data breach risk"* [Participant 40].

The move to cloud-based storage of genomic information was noted as a trend: *"is that sufficient for those looking at privacy issues and cybersecurity?"* [Participant 16]. The same stakeholder also said that researchers need access to technical expertise to ensure the platforms they use to store genomic data are secure. Ultimately cybersecurity breaches would affect public confidence in genomic information use both in research and clinical settings: *"if people don't feel like their data is going to be safe then they won't get on board"* [Participant 3].

4.1.8 EQUITY AND PROVENANCE

Literature review

Genomic research and testing can raise questions of equity and access, particularly given the blurred line between treatment and research. Jane Kaye and colleagues raise the tendency for genomic data sharing systems to 'perpetuate inequalities by obscuring the contributions of different stakeholders along the data stream'. This may result in 'a proportionately greater disadvantage for people in resource-poor countries to contribute to, be recognized for, and benefit from science'. (Kaye et al, 2018)

Stakeholder consultation

"Broadly, equity and access to testing services is an issue" [Participant 6].

People can pay for genome sequencing which *"raises questions around equity of access to that service"*, and many patients access testing under the guise of a research program.

Different situations across patients can make it harder/more expensive to access testing - the stakeholder compared a patient with a family history of mitochondrial disease, which requires a test that looks for one genetic change in mitochondrial DNA which the stakeholder deems is a *"cheap test"*, whereas those with no family history may herald *"a series of complex systems [where] we suspect a mitochondrial disease but do not know which type it is"* which is *"much harder"* [Participant 6].

> *"It is our understanding that there is no Medicare rebate for genetic testing, or for mitochondrial diseases broadly speaking...therefore depending on what clinical service a patient is connected with and what research programs they have available to them, [due to factors such as testing availability and cost], it is not uncommon for patients of mitochondrial disease to go through a pre-targeted test first [in terms of which genes are targeted]. and then broadened out...We hear of some patients whose clinician is tapped into a certain research program that might have access to certain testing, [and as a result the]. patient may be sent overseas [for testing], it is a toss of the coin as to who actually gets access to that"* [Participant 6].

> *"Genomics offers such an incredible future but what the future might look like in terms of access and equity comes up in my mind all the time"* [Participant 15].

> *"You always worry [emerging genomic technologies] won't be equitable across all communities"* [Participant 30].

Provenance emerged as an issue with particular ramifications for those working with older samples from Aboriginal or Torres Strait Islander people. One participant reported that a collection of samples at their institution had initially been determined to not come within the scope of the *Heritage Act 1993* (SA), and were later reassessed as falling under that statute's auspices. This required additional consultation with particular groups, with the power to veto the use of the samples, despite the researchers having consent from the original donor: *"We had individual consent from people and were waiting on results but it was possible that someone else could challenge and say they didn't like an aspect of the study and then it would stop."* [Participant 60].

This is an interesting example of the tension between community and individual interests being resolved in a starkly different way to mainstream consent.

5 Other uses of genomic information

5.1 Private

5.1.1 INSURANCE DISCRIMINATION

Literature review

Discrimination in insurance has been a prevailing ethical and legal issue since the genesis of genetic and genomic testing. Although research has identified lower levels of discrimination than might have been anticipated, overall worry about the risk of discrimination remains prevalent. (Erwin et al, 2010) Fear of genomic discrimination has been linked to limiting uptake of genetic testing in both the research and clinical contexts." (Prince, 2018) As Margaret Otlowski notes, 'While cases of unlawful discrimination are clearly concerning, the very existence of such cases and the potential for such discrimination has created insurance fears that are deterring the uptake of genetic testing and participation in medical research'. (Otlowski et al, 2019)

There are considerable differences internationally in the extent to which insurance companies can use genomic information in decision-making. A strong stance against such use is common in European jurisdictions. The U.S. also has prohibited the use of genomic information by health insurers. (Prince, 2018) In Australia, the primary policies have been anti-discrimination laws along with self-regulation by the Financial Services Council (FSC). Until recently under this model, insurers could use existing genomic test results in underwriting decisions provided there was actuarial justification. (Prince, 2018)

Evidence published on genomic discrimination by Australian insurance companies indicates the occurrence of both legal and illegal discrimination. (Tiller et al, 2020; Barlow Stewart et al, 2018) Although the insurer is responsible for justifying any such use of genomic information, 'in reality it is difficult for an individual applicant to challenge an adverse insurance decision'. (Otlowski et al, 2019) Researchers also have pointed to low knowledge of avenues to challenge treatment perceived as unfair. (Goh et al, 2013)

In March 2018, the Parliamentary Joint Committee on Corporations and Financial Services released a report on the life insurance industry. The report recommended that Australia should have a moratorium on the use of genomic test information in insurance decisions. (Prince, 2018) This was consistent with a 2018 position statement by the Human Genetics Society of Australasia recommending a moratorium on use of genetic test results. (Newson et al, 2018) In July 2019, the FSC commenced a voluntary moratorium on the use of genomic test results for life insurance cover up to $500,000 (lump sum death cover and total permanent disability cover).

Stakeholder consultation

Not unexpectedly, most of the stakeholders drew attention, in one way or another, to community concerns associated with discrimination in the context of insurance. The following two quotes are illustrative of stakeholder perceptions as to these concerns.

> *"I really think the insurance issue is a massive issue that is not spoken about unless it is specifically raised with people."* [Participant 29].

> *"Insurance, when you ask consumers in the general public the first thing they say is insurance"* [Participant 30].

More specifically, a number of stakeholders drew attention to the voluntary moratorium introduced by the Financial Services Council (FSC) in 2019. Some scepticism was expressed by stakeholders as to the effectiveness of the moratorium in assuaging community concerns about the risk of losing or being refused insurance cover based on genetic results, and the confusion that is flowing as a result. The following quotes pick out some of this confusion.

> *"[People are in] a better position than they were in when there was no moratorium... but we see the limits that apply ($500,000) only covers what some people need in terms of proper life insurance cover. ... [The moratorium is creating] confusion for clinicians...consumers, [and] patients ..."* [Participant 3].

> *"We now have to inform people about the moratorium and therefore it is creating more uncertainty in the people, being reluctant to be involved because we are told we have to bring this up in conversation. ... This whole issue has created an unnecessary hesitancy for people to be involved in research that will benefit them more than harm them."* [Participant 25].

Other stakeholders expressed further concern as to the extent to which the moratorium has actually addressed consumer concerns, as illustrated in the following two quotes.

> *"Life insurance is still an issue. Who knows whether health insurance will change its tune down the track and say we will not insure people who have genetic conditions. The moratorium hasn't gone far enough."* [Participant 13].

> *"The moratorium was a clever strategic move on part of the FSC. People particularly in government who were concerned think it is fixed now. As we gather data, I think we need to gather data about the impact of the moratorium and whether it is actually protecting consumers. I think it certainly is an issue."* [Participant 18].

In contrast, one stakeholder expressed stronger support for the moratorium.

> *"I think the moratorium is really helpful. Still there is still a lot of misinformation out there. As a clinician if I was seeing someone that had cancer and they said I don't want a test because it might affect my ability to get insurance you have to nicely say to them you won't get insurance because you have cancer not because you are having a genetic test. There are a lot of myths out there about insurance."* [Participant 9].

It should be noted that other stakeholders were more sanguine about the issue of discrimination in the context of genomics, as encapsulated in the following quote.

> *"People are already being discriminated against for so many things - education, salary. Genomics is relatively minor in the big scheme of things re: discrimination."* [Participant 12].

For the Aboriginal stakeholders, discrimination in the context of insurance was recognised as a potential risk but was, in general, considered one of the less significant discrimination issues that they face. Several participants noted a regional dimension to this issue, with Participant 44 observing that *"Having said that, insurance isn't the biggest thing in Aboriginal communities either."*

5.1.2 EMPLOYMENT DISCRIMINATION

Literature review

Relatively little attention has been given to the issue of genetic discrimination in employment in contrast to the subject of genetic discrimination in insurance. A complicating factor is that there is potential for legitimate use of genetic screening and monitoring in the workplace where this can be justified on occupational health and safety grounds and would generally be in the interests of the workers. The concerns that have been identified in the literature relate to circumstances where genetic information is used by an employer to determine whether to employ a person or to continue to employ a person, in order to exclude from the workforce individuals who have been identified as being at risk of developing a genetic condition that may affect their future capacity for work.

Essentially Yours recommended that employers should not collect or use genetic information in relation to job applicants or employees, except in the limited circumstance where is consistent with privacy, anti-discrimination and occupational health and safety legislation and a number of specific recommendations were made, some of which have been implemented. One key change has been the amendment to the definition of disability in the *Disability Discrimination Act 1992* (Cth) to expressly include a genetic predisposition to a disability.

There have been reported instances of genetic discrimination in employment in Australia (Barlow-Stewart and Keays, 2001) and overseas (Kim, 2010, Otlowski et al, 2012) most commonly involving persons with genetic risk of neuro-degenerative disorder such as Huntington's Disease. There have also been cases before the courts involving misuse of an employee's genetic information as illustrated by the case *Trindall v NSW Commissioner for Police* [2005] FMCA which involved as successful complaint under the *Disability Discrimination Act 1992* (Cth). The complainant, who was a policeman of mixed Aboriginal and black African race, alleged discrimination in his employment after being placed on restricted duties to his sickle cell trait. Persons with sickle cell trait are genetic carriers for sickle cell disease but they do not have the disease and have a normal life expectancy. The condition therefore would not be expected to have any impact on the complainant's capacity to work. The complainant brought an action in the then Human Rights and Equal Opportunity Commission leading to an action in the, then Federal Magistrates Court which found disability discrimination to be established.

Stakeholder consultation

Discrimination in the context of employment was referred to much less commonly by stakeholders than in the insurance context. Nevertheless, a number of stakeholders did make some relevant comments. For example, one stakeholder expressed a concern that this should be a human rights issue rather than an issue that is dealt with through the disability framework [Participant 3]. Others mentioned lack of clarity in the law, and lack of knowledge of relevant laws.

> *"Discrimination in employment can be an issue. The laws do not cover genetic discrimination and it is a fine line, it is hard to prove sometimes, might be that the other candidate is a better candidate for the job. But if you know two people, one with a child with genetic health condition [and the] other doesn't, you'll probably employ the one that doesn't because they won't be as absent from work."* [Participant 13].

> *"In Australia do we have any laws that prevent you from discriminating if an employer has your records and sees a high risk of Alzheimer's onset? Would this be allowable? I do not know. But then I am not a clinician or a lawyer. Laws that protect against discrimination could be repealed at a moment's notice, people are not protected forever."* [Participant 9].

5.1.3 COMMERCIAL USE

Literature review

There is a growing body of academic literature that explores the consequences of the increased push towards the commercialisation of genomic resources and data. Biobanking has been a particular focus for debate. (Caulfield et al, 2014)

In the biobanking context, Critchley et al (2015) point to empirical evidence which shows that "trust and intention to participate in biobank research was significantly eroded when the biobank was described as being run by a private company instead of a public research organization." However, other research from the same group involving a community consultation exercise reveals that "it is possible to counter the 'natural prejudice' that many people have against commercialization through independent governance of biobank resources and transparency with regard to commercial involvement." (Nicol et al, 2016) If there is to be commercial involvement, establishing appropriate benefit sharing arrangements may be crucial to maintaining public trust. (Nicol, 2006)

Stakeholder consultation

Although a number of stakeholders alluded to commercialisation concerns, there were few pertinent references where stakeholders teased out in more detail what it was about commercialisation that was concerning. However, Participant 16 nicely encapsulated what it is that seems to be of greatest concern.

> *"Vast quantities [of data] are generated not just from genomics but from all -omics analyses. When it comes to its application commercially there are implications. You've got any commercial products arising out of it, IP arises out of it, a lot of consent forms will say you won't have any access to any commercial benefits from samples so on and I am wondering whether that is still acceptable for the genomic data coming out of it."* [Participant 16].

Interestingly, one of the stakeholders from the Aboriginal and Torres Strait Islander community put forward a quite different viewpoint.

> *"Not so much concerned about medical or health research nor selling information to big pharma-companies. People don't think about genetics in that sense but rather it is about past researchers and native title and Aboriginality."* [Participant 41].

5.1.4 DIRECT-TO-CONSUMER (DTC) GENETIC TESTING

Literature review

The demand for DTC genetic testing has increased significantly since *Essentially Yours* was released in 2003. 23andme, the dominant player in the global market, surged ahead of its competitors in the provision of health-related and ancestry-related genetic services until the U.S. FDA demanded greater regulatory control (Curnutte, 2017). 23andme has since secured FDA approval for a number of tests that communicate information about individual disease risk (Curnutte, 2017). Scholars suggest the likelihood that DTG testing will extend to include tests based on exome and genome sequencing technologies. (Burdon, 2015) Burdon cautions that, 'In this way, it is highly likely that in the near future anyone will be able to get a genetic test for any disease without the input of medical professionals familiar with the patient's history'. (Burdon, 2015)

Although some direct-to-consumer genetic testing services do exist in Australia, supply is predominantly through online testing companies not located within Australia. (Metcalfe et al, 2018) Concerns have been expressed in the literature in Australia and elsewhere about the rise of the DTC genetic testing industry, including: inadequacies in privacy protection (Nicol et al, 2014); increased burden on the public healthcare system (McGuire and Burke, 2011); commercialisation (Metcalfe et al, 2018); consumer comprehension of information provided (Savard et al, 2020); and

the types of actions that consumers might take in response to test results (Charbonneau et al, 2020).

Concerns associated with public mistrust of commercial involvement are particularly pronounced in the context of direct-to-consumer genetic testing. Research comparing GP-mediated versus direct-to-consumer genetic testing reveals a trust deficit in the commercial context. (Critchley et al, 2015) Matthews et al (2012) raise ethical, as well as social concerns about the commercialisation of direct-to-consumer genetic testing, particularly if the tests don't give the consumer clinically actionable information. They call for a regulatory framework to better protect consumers. (Matthews et al, 2012)

Stakeholder consultation

A small number of stakeholders provided specific feedback on the direct-to-consumer genetic testing industry. Concerns ranged from increased burden on clinical genetic services, through to lack of rigour and accountability and the risks to consumers associated with lack of understanding of the broader implications of engaging with such services, as illustrated in the following four quotes.

> *"If a company wants to move into Australian territory and begin to provide these tests in a health sense, they should be providing support and that should be in their business model so that the commercial benefits are balanced by not just offsetting commercial benefit for that company and dumping it on our health system" ... "What happens with data given to DTC companies is also really important ... this idea that people agree and it is informed consent even though it is buried in 40 pages of small print is not good enough."* [Participant 3].

> *"I was shocked when seeing DNA tests on the market. What I worry about is people's ignorance and wanting to be informed can lead down a path to buying a wristband [re. DNA wristband which recommends products to consumers based on DNA]."* [Participant 29].

> *"[Contrasting with clinical genetic services] buying a $25 box that is sent to the US and they will do your genome and give you all this data back and they share it and are holding your genomic data and there is no governance of that....I am happy with the loops and hoops we [clinicians] have to jump because I feel confident that what we are doing and the results we are providing is much more rigorous than what [direct-to-consumer testing companies] are doing."* [Participant 37].

> *"There is even less control in the corporate space, and issues with Ancestry as we don't know what else they are looking at."* [Participant 53].

For Indigenous stakeholders, commercialisation was part of a wider concern about exploitation of Aboriginal and Torres Strait Islander people and knowledge: *"Aboriginal people have been taken advantage of for so long why wouldn't this be the case? Non-Indigenous people have made money of Aboriginal people in so many ways why not our DNA?"* [Participant 41].

5.1.5 PATENTING

Literature review

The patent landscape has changed significantly since *Essentially Yours* was released in 2003, and since the Australian Law Reform Commission completed their follow-on inquiry into the impact of gene patents on the provision of healthcare in 2004 (*Genes and Ingenuity*, ALRC Report 96). In both Australia and the U.S., the highest courts have decided that patents claiming nucleotide sequences as they are found in nature do not satisfy the requirements for patentable subject matter (Rimmer, 2016). In the specific Australian context, the 2015 High Court of Australia ruling in the case of *D'Arcy v Myriad Genetics* rejected the patentability of Myriad Genetics' claims to BRCA nucleotide sequences. (Bartlett, (2015/2016))

Prior to these decisions, a large body of academic commentary, media reports and policy statements expressed concern about gene patenting because of the power such patents gave to their owners in research, translation and clinical contexts. (Nicol, 2011) Various groups globally recommended the adoption of more collaborative approaches to patent use. (Van Overwalle, 2010)

Since the nucleotide sequence patent decisions were handed down by the courts in Australia and the U.S., patent offices, lower courts and academic commentators have been attempting to define more precisely the boundaries of patentable subject matter in the context of genomics. A marked divergence in approaches has been observed between jurisdictions. (Dreyfuss et al, 2018; Nicol et al, 2019)

Stakeholder consultation

One stakeholder referred to the specific issues associated with gene patents.

"I was against gene patenting when working with XXX. The competition and so on of developing things from genes which were not inventions, they were discoveries. They block everyone else out from investigating them. The High Court aligned with our view." [Participant 2].

5.2 Public

5.2.1 ACCESS BY POLICE AND OTHER ENFORCEMENT AGENCIES

Literature review

In surveys with the public in several countries, including Australia, participants were asked to rate theoretical harms based on what concerned them most. The most frequently identified was 'My DNA being copied and then planted at the scene of a crime'. (Middleton et al, 2019) Concerns about law enforcement access to genetic information came to the fore with the 2018 arrest of a suspect in California's Golden State Killer cases, in large part through a 'partial match' with the killer's DNA via the genealogy website GEDmatch. This raised questions about the privacy expectations of individuals who contribute to genetic genealogy databases. (Guerrini et al, 2018)

Stakeholder consultation

Almost all of the Indigenous participants raised the potential for access to genomic data for law enforcement purposes as a serious concern, exacerbated by Indigenous overrepresentation at every stage of criminal justice. Most considered this issue to warrant legislative protections. Participant 60 attributed this to *"a general distrust of authorities because of past experiences - they don't trust the government... at all."*

5.2.2 IDENTITY INCLUDING INDIGENEITY

Literature review

The extent to which genomic information can inform identity is contextual and dependent on definitions of identity. Race is recognised as a social construct that aligns with genomic information only insofar as it reflects population pressures, and it has long been recognised that genomics as a discipline raises tensions between social and biological paradigms. (Foster and Sharp, 2002) Debates continue to reflect a concern that genome science will lead to an overreliance on biological determinations of identity, undermining pre-existing conceptions of identity, particularly among Indigenous peoples. (TallBear, 2013) The increasing accessibility of genomic testing has resulted in a focus in recent years on what, if anything, genomic information might mean for the definition of Indigeneity among Aboriginal and Torres Strait Islander people. (Kowal and Watt, 2018; Watt and Kowal, 2019)

Stakeholder consultation

The interaction between genomic information and identity was a key theme identified by Aboriginal and Torres Strait Islander stakeholders. This took two main forms: the potential for genomics to influence personal identity as an Australian Indigenous person; and the potential for genomics to be used to curtail rights or access to services.

There was general agreement that people in remote areas were less concerned about genomics as a threat to personal identity than those in urban areas, where mixed heritage or less knowledge about ancestry was common:

> *"Concerns are around a lot more communities who are in urban areas and not so much remote communities in the NT and the Cape ... a lot of urban communities who might have some mixed ancestry or who know they don't have indigenous parentage down the line ... who may not be so comfortable with their identify or who are always questioned about their identify because they are fairer than others."* [Participant 41].

> *"What lots of people hear is not community voices, its other voices speaking on behalf of community people. If you go and speak to community people, they know their story line and they're comfortable with that. If they're brown, then they know that they're Aboriginal and someone else. Territory mob, they know where they're from, even stolen generation. They go back and connect with country if they can"* [Participant 43].

There was, however, an acknowledgement that discovery of misattributed parentage could complicate an otherwise robust sense of identity, including implications for kinship.

> *"People are pretty confident in their family lines. Where it gets sticky is when someone who thinks their father was their father who wasn't, and they have married into the wrong family group. But I don't think people will use genetics as who their family is, they have already worked it out."* [Participant 41].

A number of participants raised concerns about the potential for genomics to be used to further erode rights to self-determination, through changes to the legal definition of Aboriginality. Participant 41 said *"I hope the government won't take this into account and respect the fact Aboriginal people have their own way of determining Aboriginality which does not come from having a percentage. This is what I am really worried about."* Participant 42 viewed this concern as part of wider concerns about the flow-on effects for communities, if the government *"establish[ed] a system that could be used to interrogate people's identity which would have a negative consequence on themselves and the community itself."*

Further, one participant reported:

> *"The notion of aboriginality and what does genetics tell us if anything about being aboriginal is a genuine concern that people have expressed on multiple occasions to me. There's a sense there's been enough damage done already by misunderstandings and misuse around the concept of aboriginality and who belong."* [Participant 60].

Participant 51 linked these concerns to recent political events:

> *"You have people like Pauline Hanson who wanted to create a register of Aboriginal people and wants to genetically test people. I have some concerns that it might be used as a way to oppress people. Aboriginal people have to prove their Aboriginality when other people don't have to prove who they are. ...The percentage is a white obsession and it has been right there from colonial times. ...Aboriginal people do not care, it's about connection and relationships"* [Participant 51].

Participant 44 specifically raised the issue of identity in relation to traditional adoption practices in the Torres Strait, noting that there was a need for clinicians and researchers to take care when handling such cases.

Participant views regarding the potential impact of genomics on members of the Stolen Generation were more mixed. Some saw it as a possible tool for reconnection, while others worried that such a purpose could be co-opted for misuse, based on previous negative experiences (see also s 1.2.4). Participant 53 believed that people would be interested in the use of genomics for the reconnection of dislocated individuals, if it was undertaken with appropriate Indigenous governance:

> *"A centre for this would be great - I would be more inclined to do it if there was an Indigenous centre to look at my samples in the context of where I might belong. And you get to choose what happens to it and say I don't want my sample stored any more"* [Participant 53].

Many participants reported concerns about the use of genomics to determine identity for the purposes of restricting access to services or to Native Title.

> *"Are researchers trying to prove Aboriginality and percent Aboriginality and are the government going to use this information to determine services?"* [Participant 41].

> *"In terms of genetics, lots of things people bring up are things centred around native title and how it will affect them in that process"* [Participant 41].

> In relation to land ownership and Native Title, as well as access to services: *"Generally we are smart enough to be paranoid about some of those things because if it can be used against you, it will be in Indigenous perspectives"* [Participant 44].

> *"The other issue is with Native Title. There's a sense that you can use this genetic information to say that you belong to an area during a period of time needed to prove Native Title. But the most vocal comments we've had during meetings have been about exclusion; some people have said it's an opportunity to say this mob are not from here, they're not from this region originally"* [Participant 60].

Priority Issues for Further Analysis

For the next stage of the project, we will be undertaking an assessment of the legislation and regulations relating to specific issues, selected by the Department of Health from the issues identified in this Scoping Review.

All of the issues identified through the literature review and stakeholder consultation process will be important to address in the implementation of the National Health Genomics Policy Framework. However, not all will be best addressed through primarily legal mechanisms. In prioritising issues for legal analysis, the following considerations provide a useful guide:

- Which issues are amenable to primarily legal analysis?
- Where is there overlap between issues, and which should be considered together?
- Which issues have been the subject of recent reform at the national level?
- For which issues do the findings of the Scoping Review suggest the existence of innovative legal solutions, or solutions that could readily be implemented into policy?
- To what extent do these issues get at the key themes identified in the Scoping Review (1. Reliance on – and limits of – ask or anonymise; 2. Limitations in understanding of genomics and regulatory environment; 3. Dissolving boundaries between public and private, healthcare and research, individuals and communities)?

On this basis, we suggest that the following issues be prioritised for inclusion in the Final Report:

- Genetic discrimination, including insurance and employment;
- Regulation of genetic testing, in vitro devices and DTC testing;
- HRECs, standards and waivers of consent;
- Limitations of ask or anonymise for troubling uses, including access for commercial, insurance or law enforcement purposes;
- Indigeneity and genomics, including issues relating to identification, group interests and equity;
- Privacy and its limitations, including rights of the deceased and family members, and data sharing;
- Recontact and return of raw data and findings (including incidental findings); and
- Chains of custody, control and ownership of data and tissue, including sovereignty, custodianship and intellectual property.

The legal analysis of these issues would be followed by a section that draws together the cross-cutting themes to address two key questions:

1. What are the regulatory distinctions between research and clinical uses of genomic data, and to what extent can and should the distinction between care and research be retained in regulatory frameworks?
2. To what extent can the traditional reliance on 'ask or anonymise' be retained in regulatory frameworks?

A number of issues identified in the Scoping Review are representative of broader themes, and will be addressed as part of the legal analysis of the above issues. These include:

- Legislation not fit for genomics
- Alignment with overseas laws
- Inconsistency between laws of the States and the Commonwealth
- Consent
- Influence of My Health Record

We consider the following issues to be of lower priority for the next stage, because of an unclear legal nexus, because potential solutions are more likely to come from primarily non-legal sources, or because they have been the subject of recent reform:

- Data subject understanding
- Public understanding
- Workforce capability, training and accreditation
- Understanding of law by non-specialists
- Risk averse practices due to legal complexity
- Open publication of data as a requirement of a publisher or funding body
- Data parity, interoperability, quality
- Platforms for sharing
- Security and hacking

Conclusion

Ethical, legal and social issues arising from the collection, use and disclosure of health-related genomic information are numerous, complex and interrelated. Addressing these issues is a daunting and increasingly pressing challenge facing health systems. Here, we have identified the concerns that emerged most strongly from the literature and our community consultations. From this overview, three themes are evident:

1 The traditional reliance in the regulatory environment on 'ask or anonymise' to provide protection for patients and participants is increasingly not fit for purpose;

2 There are limitations in public and professional understanding of genomic data and the regulatory environment, along with gaps, fragmentation, and inconsistency in both regulation and understanding. These are resulting in lost opportunities to realise research/ care synergies and to promote trustworthy governance;

3 Traditional boundaries between public and private, between healthcare and research, and between individuals and their communities, distinctions which have formed the basis of regulatory approaches, are dissolving and highlighting the inadequacies of current regulatory frameworks.

The issues identified in this review are linked to aspects of each of the strategic priority areas identified in the National Health Genomics Policy Framework and will require attention - and in many cases resolution - if the outcomes identified under those priority areas are to be realised. An assessment of the legislation and regulations governing the collection, use and disclosure of health-related genomic information as they pertain to each of these issues will form an important component of the work needed to achieve the outcomes required by the Framework.

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