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The Australian LINEAGE Study: advancing and implementing international guidance on genomic data within local governance frameworks

Tamra Lysaght, Rachel Ankeny, Alex Brown, Rebekah McWhirter, Dianne Nicol, Margaret Otlowski, Bernadette Richards & Ainsley J. Newson

The importance of generating and sharing human genomic data for human health is widely recognised. Whilst several international high-level frameworks provide guidance, local governance challenges and concerns of previously under-represented populations mean that the full potential of genomic data sharing remains unrealised. This commentary introduces the LINEAGE study, an Australian research project that is building an empirically-informed and normatively robust governance framework for the generation and sharing of genomic data.

The potential benefits of generating and sharing genomic data for human health are widely recognised. Yet the activities required to generate, store, use and share genomic data remain subject to many governance challenges, including unresolved ethical, legal and social issues (ELSI). As a result, genomic datasets globally remain poorly representative, under-developed, under-networked and under-utilised[1]. To address this problem, several international frameworks for the governance of genomic data have been released in recent years. Whilst there is general, high-level consistency across these frameworks, they were never intended to be uniformly applicable across all jurisdictions where genomic data may be generated, stored, shared and protected. Rather such frameworks are intended to provide the high-level guiding principles that local jurisdictions can apply according to their specific legal, cultural, ethical and socio-political contexts. Although, how this translation is achieved in practice requires further consideration.

In this commentary, we introduce an Australian research project that aims to bridge the gap between international guiding principles and their local application. The study, *LINEAGE: Law, Sociology and Ethics in Data Governance for Genomics*, is building an empirically-informed governance framework for enabling ethically defensible, legally robust, socially acceptable and culturally safe sharing of genomic data. The ambition is that the LINEAGE framework will apply to genomic data generated in a variety of settings, including in basic and translational research, clinical care and population health. It will address the complexities of the federated (multi-jurisdictional) Australian regulatory system and the impetus to share data with, or facilitate data access for, researchers internationally. It will also incorporate patient-public preferences and cultural considerations, including perspectives from Australian Aboriginal and Torres Strait Islander Peoples. Outcomes will include a road map for other jurisdictions seeking to apply international guiding principles to generate national frameworks for the ethical governance of genomic data and its sharing across the data life cycle in ways that account for local ethical, legal, social and cultural considerations.

Existing international guidance on human genomic data sharing

A number of notable international guidelines for sharing human genomic data are available: the UNESCO *Universal Declaration on the Human Genome and Human Rights*[2], UNESCO *International Declaration on Human Genetic Data*[3], the European 1+*Million Genomes (1 + MG) Framework*[4], the Global Alliance for Genomics and Health (GA4GH) *Framework for Responsible Sharing of Genomic and Health-Related Data*[5], and the World Health Organisation (WHO) *Principles for Human Genome Data Access, Use and Sharing*[6]. As summarised in Fig. 1, there is some uniformity in the principles endorsed across these documents. Respect for individuals, including their rights to privacy and self-determination (autonomy), are shared between frameworks, as are collective interests, such as fairness, equity, solidarity and reciprocity. However, the documents differ in their scope and application.

For example, the two UNESCO declarations set out guiding principles for Member States in the formulation of legislation and policies. The 1997 declaration on the human genome has precedence and thus influences the interpretation of the 2004 guidance on handling genetic data. However, individual jurisdictions are left to interpret and apply these guidelines in relation to their own legal systems, state of technological advancement, resourcing and cultural context.

At a supra-national level, 1+*Million Genomes (1 + MG) Framework* recognises the complexities arising from overlaps, gaps and conflicts between EU and Member State laws and policies. The *Framework* comprises numerous guidelines and recommendations that are intended to be used as a reference point when implementing genomics initiatives at regional, national, and European levels[4]. However, it is also highly complex and does not include comprehensive implementation plans.

In 2024, the WHO released their *Principles for Human Genome Data Access, Use and Sharing*[6]. While these principles provide useful high-level guideposts, they do not aim to account for variations in local contexts, nor do they aim to guide granular implementation in individual jurisdictions. As

Name	Endorsing Authority	Aim and Scope	Guiding Principles
Universal Declaration on the Human Genome and Human Rights	UNESCO,1997	To promote genome research that fully respects human dignity, freedom and human rights, and reject all forms of discrimination based on genetic characteristics.	Respect for human dignity Individual rights to privacy and autonomy Solidarity Benefit sharing Genetic non-discrimination
International Declaration on Human Genetic Data	UNESCO,2004	Ensure respect of human dignity and protection of human rights and fundamental freedoms in the collection, processing, use and storage of human genetic data, in keeping with the requirement of equality, justice and solidarity	Privacy and Confidentiality Individual rights have priority over collective Proportionality Special status of genetic data Transparency Equality Consent
Framework for Responsible Sharing of Genomic and Health Related Data	Global Alliance for Genomics and Health, 2014	The purpose of this Framework is to provide a principled and practical framework for the responsible sharing of genomic and health related data.	Respect Individuals, Families and Communities Advance Research and Scientific Knowledge Promote Health, Wellbeing and the Fair Distribution of Benefits Foster Trust, Integrity and Reciprocity
Principles for human genome data access, use and sharing	WHO, 2024	Sets out globally applicable and inter-connected principles on the collection, access, use and sharing of human genome data, to promote human health and well-being, including responsible medical advances and scientific research	Social Justice Solidarity Equity Collaboration & partnership Transparency Stewardship
1+Million Genomes (1+MG) Framework	European 1+Million Genomes, 2023	Aims to enable secure access to genomics and the corresponding clinical data across Europe for better research, personalised healthcare and health policy making.	Consistency FAIR Principles Quality guidelines Consent Collaboration

Fig. 1 | Summary of international guidance for human genome research.

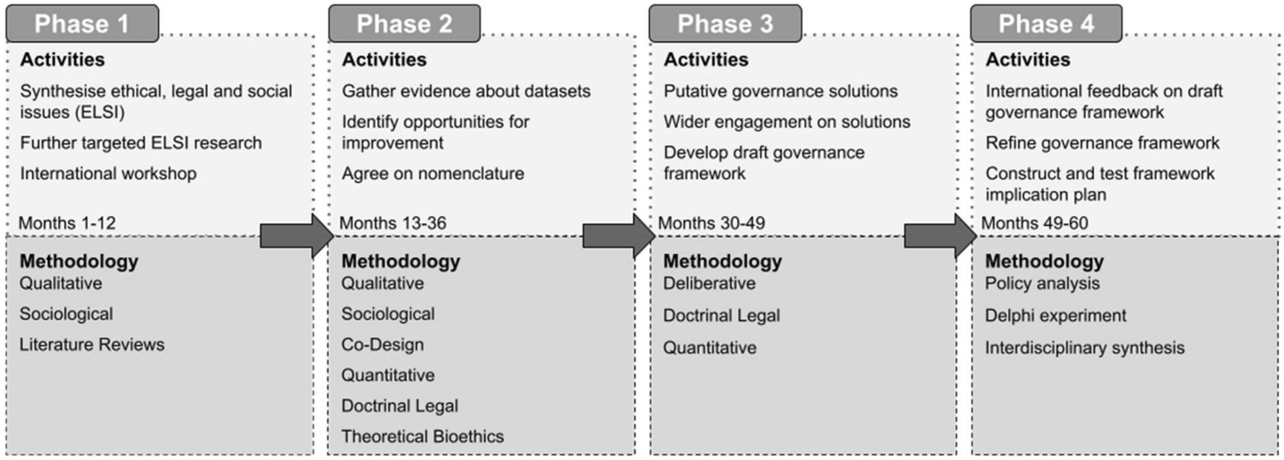

Fig. 2 | Multi-phased research design and methodologies of the LINEAGE Study.

a result, operationalisation of the WHO Principles will require member countries to develop their own genomic governance frameworks that suit cultural, social and logistical needs.

The international not-for-profit coalition, GA4GH, was formed in 2013 to foster global collaboration and establish standards for the sharing and use of genomic and clinical data. Through its eight work streams and other initiatives, GA4GH develops policy tools and technical specifications to promote interoperability, data sharing and ethical standards across diverse jurisdictions. The GA4GH Framework provides the foundational principles that shape the development of all GA4GH tools and specifications[5]. The aim is to provide guidance to organisations in responsible data stewardship, recognising the diverse contexts within which they work. The Framework explicitly states that the application of its principles should account for local cultural norms and practices in different countries.

Bridging the Gap: the LINEAGE study

Alongside these high-level principles, locally established frameworks are needed to provide agile and responsive governance. In Australia, as in other countries[7–10], large-scale research activity and rapidly increasing use of genomic sequencing in clinical care are generating large volumes of genomic data[11]. Yet, while a blueprint and implementation plan for a National Approach to Genomic Information Management (NAGIM) in Australia have been developed[12], data remains extensively siloed and there has been no specific attempt to bring international frameworks into domestic governance.

Australian regulators have recognised the need for a national governance framework for genomic data. This need was stipulated as a key aim in the 2018 National Health Genomics Policy Framework[13]. Further, the need for research to address emerging ethical, legal, and social issues in genomic data governance was recognised as a priority in the Australian government's 2021 Genomics Health Futures Mission (the Mission)[14]. The LINEAGE Study was subsequently funded in 2022 with funding from the Mission through the Medical Research Futures Fund (MRF2015531, 2022-27)[15]. Integrating numerous established research methods from bioethics, health law and the social sciences[16–19], LINEAGE is developing an empirically-informed, philosophically robust and contextually (locally) appropriate framework for the ethical governance of genomic data.

The over-arching research question to inform LINEAGE is: "How should genomic datasets be governed effectively and responsibly?". This question is being addressed through four phases, see Fig. 2. The first phase establishes the current regulatory landscape and ethical tensions, before the project transitions to more in-depth targeted empirical and ethical analyses of key issues, such as trust, custodianship, benefit sharing and ownership in phase 2. In the third phase, putative elements of a governance framework will be articulated and tested empirically with stakeholders. The framework will be finalised in phase 4 and an implementation plan will be developed to ensure the framework is fit for purpose.

The interdisciplinary LINEAGE project team comprises over 50 experts in law, bioethics, social science and genomics. Several investigators have lived experience of a genetic condition, and some are Aboriginal or Torres Strait Islander. At its conclusion, LINEAGE will have undertaken large-scale, transdisciplinary research to develop an ethically defensible, legally robust, socially acceptable, culturally safe and responsive, and implementation-ready governance framework for genomic datasets in Australia. While the final form of the governance framework cannot yet be articulated (because its final design is contingent upon outcomes from phases 1-3), a possible structure for the framework is presented in Box 1. The framework will account for the different forms of genomic data in Australia, the contexts in which they are generated and the various modes of sharing that are available. As identified in preliminary findings from Phase 1, Box 2 outlines some practical examples of challenges the framework aspires to address. Further, the project will develop a process for contextualising high-level principles that are promulgated in international guidelines to then embed them in governance frameworks designed for local jurisdictions.

Genomic data involving Aboriginal and Torres Strait Islanders

The need to improve diversity in genomic data has long been recognised[20]. Until recently, major challenges in Australia included a lack of diversity and a widening gap in access to the benefits of genomics in terms of datasets, research leadership and impact. These gaps have meant that genomic advances are primarily benefiting well-represented and highly resourced populations, predominantly those of European ancestry. However, equity is not just a matter of inclusion in research. To address historical exploitation of Aboriginal communities, including the harm, trauma and loss of trust this has led to, there needs to be substantial change to the way genomic data are collected and managed[21].

Central to these efforts is the need to operationalise Indigenous Data Sovereignty principles and ensure that Aboriginal and Torres Strait Islander people retain control over their genomic data[22]. Core requirements for

Box 1 | The LINEAGE genomic data governance framework will be oriented around a structure such as the genomic data life cycle

It will commence with an overview section that includes:

- Purpose
- Audience
- Key components of the Framework

The body of the framework is likely to contain elements such as:

- Broad values (both procedural and substantive), e.g. transparency, trustworthiness
- Key principles, e.g. data security, public benefit
- Contexts in which the framework will apply
- The kinds of genomic data to which the framework applies
- Application of values and principles to specific topics or issues, e.g. informed consent, stewardship, custodianship, capacity building
- Alignment with regulatory mechanisms
- Key recommendations (excluding operational aspects)

Worked into the framework will be key opportunities for governance improvement (see Box 2). These will be particular use cases or scenarios which will exemplify current challenges. which are being sourced through key stakeholder interviews and from within the LINEAGE consortium.

Box 2 | Examples of where improvements in governance are needed in Australia

1. Inconsistent capture in clinical consent forms of sharing preferences for genomic data.
2. Conflation of clinical and research governance policies can generate confusion and inefficiency.
3. Inconsistent adoption of data sharing standards, leading to limited data interoperability and sharing.
4. Barriers to research data sharing across different jurisdictions (e.g. due to inconsistent research ethics approvals or different local governance).
5. Challenges in maintaining Indigenous data sovereignty and aligning data sharing practices with community expectations and trust.

involving Indigenous Peoples in data governance processes are encapsulated in the CARE Principles for Indigenous Data Governance (Collective Benefit, Authority to Control, Responsibility and Ethics). These principles were devised in consultation with Indigenous Peoples, scholars, non-profit organisations and governments, and emphasise the need for Indigenous People to make decisions about their own data[23]. The CARE Principles are people- and purpose-oriented and reflect the crucial role of data in advancing innovation, governance and self-determination among Indigenous Peoples.

Internationally, Indigenous groups have begun developing frameworks to support the implementation of indigenous genomic data governance[24], building on a strong foundation of work aimed at promoting ethical genomic research with Indigenous peoples[25–27]. The particular focus on Indigenous genomics at the 2024 GA4GH Plenary[28] shows that there is increasing recognition of the need to be responsive to the concerns of these and other under-represented populations. In Australia, recent targeted funding investments are building capacity in this area, including the establishment of the Australian Alliance for Indigenous Genomics Network (ALIGN). Recognising the critical importance of Indigenous leadership being embedded at every stage, Indigenous perspectives are a cross-cutting research theme of the LINEAGE Study, attracting dedicated resourcing from the project budget and informing all stages of the development of the governance framework.

While the issues affecting the use of Indigenous genomic data are more acute as a result of the history underpinning them, they are largely consistent in type, if not severity, with the concerns of all people regarding the collection and management of their genomic information, including minorities, new migrants and other potentially vulnerable groups. Hence, rather than viewing the strategies for involving Indigenous Australians in the development of genomic data governance as a special case for imposing additional or separate obligations, seeking out processes and solutions from Indigenous Australians may provide benchmarks that could ultimately benefit everyone.

Informing the global dialogue

As outlined above, to apply global frameworks for genomic data governance in any particular locale and to devise appropriate structures for their use, it will be necessary to consider relevant ELSI and other issues that are particularly salient for local populations. Not only are diverse locations likely to have varying levels of existing infrastructures for the collection, storage and use of genomic data but also different legal and regulatory structures (e.g., genetic privacy may or may not be specifically protected) and health systems. Distinct cultural histories, particularly in relation to medicine and genetics require careful consideration: a key example in Australia is its settler-colonial history and the complicity of universities, museums and other research institutions, which historically sidelined Indigenous Peoples as research targets or subjects without appropriate autonomy and consent, or without paying due attention to the potential benefits for them[27]. Even typical ethical approaches may differ place-to-place: for instance, consent can occur at the individual or group level, and be broad or specific depending on a range of factors especially levels of societal trust and the need for cultural safety.

The LINEAGE project is well placed to apply its interdisciplinary expertise at the level of domestic governance, while doing so in a manner consistent with existing global guidelines and priorities. The project will provide a richly detailed process of how broad principles from international frameworks can be adapted to suit local cultural, ethical, legal, social and logistical needs. The processes and the outcomes of this project will demonstrate how internationally recognised needs for the benefits of genomics to human health can be realised by crafting ethically, socially, culturally and legally robust frameworks for local jurisdictions that reflect broadly accepted international principles for genome data governance. We welcome feedback from others working on similar topics and whose local contexts provide opportunities for exploring these urgent issues.

Tamra Lysaght[1,2] ✉, **Rachel Ankeny**[3,4], **Alex Brown**[5,6], **Rebekah McWhirter**[7], **Dianne Nicol**[8], **Margaret Otlowski**[8], **Bernadette Richards**[9] & **Ainsley J. Newson**[1]

[1]Sydney Health Ethics, Sydney School of Public Health, The University of Sydney, Faculty of Medicine and Health, Sydney, NSW, Australia. [2]Centre for Biomedical Ethics, Yong Loo Lin School of Medicine, National University of Singapore, Singapore, Singapore. [3]School of Humanities, Faculty of Arts, Business, Law and Economic, University of Adelaide, Adelaide, SA, Australia. [4]Philosophy Group, Wageningen University, Wageningen, The Netherlands. [5]National Centre for Indigenous Genomics, John Curtin School of Medical Research, Australian National University, Canberra, ACT, Australia. [6]Indigenous Genomics, The Kids Research Institute of Australia, Adelaide, SA, Australia. [7]College of Law, Governance and Policy, Australian National University, Canberra, ACT, Australia. [8]Centre for Law and Genetics, School of Law, College of Arts, Law and Education, University of Tasmania, Tasmania, TAS, Australia. [9]University of Queensland Medicine School, Brisbane, QLD, Australia.
✉e-mail: tamra.lysaght@sydney.edu.au

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Author contributions

T.L. and A.N. as first/senior author conceptualised the paper and led the drafting of the manuscript. R.A., A.B., R.M., D.N., M.O., and B.R. edited and commented on the draft manuscript equally. All authors read and approved the final manuscript.

Competing interest

Dianne Nicol is co-lead of the Regulatory and Ethics Work Stream of the Global Alliance for Genomics and Health. No other co-authors have conflicts of interest to declare.

Additional information

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Correspondence and requests for materials should be addressed to Tamra Lysaght.