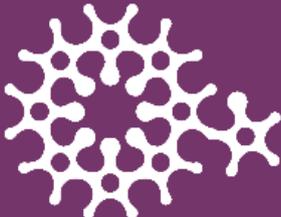


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Deeble Institute for Health Policy Research

## Perspectives Brief

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### Transforming the genomics workforce to sustain high value care

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## Executive Summary

Genomics is the study of the full set of a person's genes, including the interactions of different genes with each other and the person's environment. Genomics is used to diagnose disease, identify future risk of disease, optimise treatment and surveillance plans, and inform whole of life decision-making for affected individuals and their families (such as family planning). As such, genomics is delivering huge benefits for people in Australia and holds the promise for future gains. However, the bulk of the workforce is not prepared for genomics.

Genomics is a complex process requiring new and multidisciplinary ways of working. Teams of highly trained practitioners must work together in ways that are not currently well defined in order to:

- identify when a genomic test is appropriate for a patient,
- interpret the gigabytes of data that each test produces and its relevance to the person's healthcare, and
- explain the often life changing results to patients and their families.

Health professionals are attempting to form these teams in healthcare settings around the country on an *ad hoc* basis, and in the face of enormous need, without dedicated resources or training.

More than 1 million genetic and genomics related tests<sup>1</sup> are performed in Australia annually (RCPA, 2019); demand is increasing, and the workforce is under pressure. General practitioners (GPs), specialists, pathologists, medical scientists, genetic counsellors, clinical geneticists and many others, all play a key role in the process and often contribute hours of unpaid work every week in order to make the system work for patients.

Education in this rapidly evolving field is already lacking, and classroom education alone will never be sufficient to prepare the workforce. Knowledge has to be accompanied by development of hands-on skills such as how to explain and discuss the test with a patient, as well as how to interpret the implications of results for management of the patient. Decisions around genomic testing, as well as implications of findings, deeply impact the individual and their families as well as healthcare providers from primary care, for example GPs, through to tertiary care in hospitals, in public and private care.

Genomics is enormously valuable to society and will only grow in demand. Personal test results can provide both health and psychological benefits, reducing uncertainty and leading to more tailored care, reducing unnecessary screenings and invasive procedures, and leading to high-value care for the individual and their family.

In order to truly realise the benefits, a national strategy is required to address the consequences of a fragmented workforce, the substantial burden of unpaid work, critical gaps in education and training, and lagging funding for research into evidence-based solutions and ongoing evaluations of interventions.

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<sup>1</sup> Including biochemical tests

## Introduction

### Genomics is supporting the delivery of better value care

Genomics, where all a person's genes are examined, as well as the interactions of those genes with each other is proving a powerful tool for medicine. It is providing definitive diagnoses for people with rare diseases, directing optimal treatment choices for people with cancer, allowing informed family planning for couples and, as has been illustrated in the current COVID-19 pandemic, giving crucial information regarding infectious diseases.

Australian Genomics and state-based programs such as Melbourne Genomics Health Alliance are building the evidence base of genomics in Australia. Their translational activities are crucial to link researchers and clinicians, create and support learning communities and move policy into practice. Funding for Genomics Australia, announced March 2022 and commencing January 2024, is a welcome addition.

#### **Box 1: The promise of genomics**

##### ***Rare disease diagnosis***

After months looking for an answer to unexplained seizures in their baby son, Joshua, a genomic test showed he had Dravet syndrome, a rare genetic disease. A diagnosis opened the door to targeted treatment and allowed his parents to avoid triggers in the environment that they knew made the condition worse (Australian Genomics, 2021).

##### ***Stopping unnecessary treatments***

Ryan<sup>2</sup> had drug-resistant epilepsy that resulted in frequent seizures and meant that he could not drive. Invasive surgery to remove the suspected problem area of his brain was being planned when genomic testing revealed he had a subtype of epilepsy in which surgery and the medications he was currently on were making his condition worse (Perruca et al., 2017). Genetic diagnosis led to a change in Ryan's treatment that has greatly improved his quality of life. Ryan was saved from having needless procedures, and the health system avoided unnecessary costs in the order of \$50,000.

##### ***Tailoring cancer treatments***

Children with aggressive cancer that gives them less than a 30% chance of survival can now enrol in a program based on genomic testing of their tumours and rapid trialling of tailored, new treatments that can be lifesaving. Two-year-old Thomas, diagnosed with a rare brain cancer received a new experimental medication that targeted his tumour's growth based on the genomic results and he made a remarkable recovery (Zero Childhood Cancer, 2020).

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<sup>2</sup> Pseudonym used

### ***Genetic carrier screening***

Mackenzie Casella was 7 months old when she died from a rare genetic disease, spinal muscular atrophy. Neither parent knew they were carriers of the disease. Genetic screening can identify couples that have a higher chance of giving birth to a child with a devastating condition. Genomic testing gives couples information to help them make decisions, be prepared and plan ahead (Mackenzie's Mission, 2020).

### ***Infection control***

Bacteria and viruses also have a genome so useful information can be gained from genomic testing of disease-causing microbes. This has been crucial in the fight against Tuberculosis in some areas, identifying variants and strains that were becoming resistant to standard treatments. This means appropriate tailored treatments can be used. Genome sequencing has also been crucial in the identification of new variants of COVID-19, allowing more accurate contact tracing.

## **Genomics in Australia today**

### *The rapidly evolving genomics workforce*

The genomic workforce can be broadly divided into genetic professionals and non-genetic professionals.

#### *Genetic Professionals*

Genetic professionals have recognised qualifications in genetics in addition to basic medical or other postgraduate or professional training.

In 2017, there were an estimated 637 clinically trained genetic professionals in Australia: 150 clinical geneticists and 480 genetic counsellors, of which 220 were practising in clinical roles (Table 1) (Nisselle et al., 2019). Settings in which genetic professionals work include genetics services and familial cancer services, typically attached to public hospitals. They receive referrals from medical specialists to investigate cases of suspected or known genetic disease, including testing and interpretation of results. The family rather than a single patient is the focus of care so wider family testing and family planning as appropriate are also considered by the service.

There are 50 genetics services in Australia (Centre for Genetics Education, 2022), staffed by clinical geneticists and genetic counsellors. Expert genetics services are typically concentrated in metropolitan areas with regional areas supported by outreach services. This can limit access for those living in rural and remote areas. Genetics services diagnose and manage genetic conditions through investigation, counselling, advice, and medical management as appropriate. Alongside, or sometimes within, these services there are also familial cancer services aiming to reduce risks relating to hereditary cancer. Long waitlists exist for many genetics services, for example in NSW (ACI, 2021).

Table 1: Clinically-based genomic professional workforce description

Health professional	Role	Education and training (registration agency)	Workforce size (2017)(Nisselle et al., 2019)
Clinical geneticist	Medical specialist diagnosing and treating genetic conditions	Bachelor of Medicine and Surgery, basic specialist training, advanced training in clinical genetics under auspices of RACP (AHPRA)	150 Approx. 10 new trainees per year
Genetic counsellor	Allied health professional who assists people with or at risk of a genetic condition to understand and adapt to the implications	Bachelor degree, Master of Genetic Counselling, training in genetic counselling under auspices of HGSA (NASRHP)	477 (220 in clinical practice) Approx. 40 new graduates each year

In addition to these clinically-based genetic professionals, laboratory-based genetic pathologists, medical scientists and data analysts with additional genetic training perform genomic testing and interpretation ('curation') of results. The Royal College of Pathologists Australasia identified 87 laboratories across Australia that performed genetic or genomic testing in their survey of 2017 (RCPA, 2019).

Table 2 Laboratory-based genomics professional workforce details

Health professional	Role	Education and training	Workforce size
Genetic pathologist	Medical specialist who analyses genetic testing data, interprets its clinical significance, and reports test findings	Bachelor of Medicine and Surgery, trained or training in genetic pathology under auspices of RCPA	22.3 FTE*
Medical scientist	Performs genomic testing and curation of results	Bachelor degree in science, training in genetics under auspices of HGSA (MHGSA, FHGSA) or RCPA (FFSc)	~ 169 FTE**
Clinical bioinformatician and IT/computer scientist	Develops and improves methods for acquiring, storing, organising and analysing biological data	Bachelor degree in science, additional relevant qualification (e.g. Masters of Bioinformatics, PhD) may have training under auspices of RCPA (pathology informatics)	39.4 FTE
Other fellows, technicians	Various	Overseas or other fellowship e.g., FAACB, PhD	47.9 FTE

\*An additional 37.7 pathologists with training in other subspecialties, for example haematology, also work in genetic testing laboratories.

\*\* A further 702 medical scientists and 185 technicians work in laboratories but do not have formal training in genetics. 47.9FTE have overseas or other qualifications not specific to genomics for example, FAACB.

FTE	Full time Equivalents
RCPA	Royal College of Pathologists of Australasia
FAACB	Fellow of the Australasian Association for Clinical Biochemistry and Laboratory Medicine
FRCPA	Fellow of the Royal College of Pathologists of Australasia
FFSc	Fellow of the Royal College of Pathologists of Australasia Faculty of Science
FHGSA	Fellow of the Human Genetics Society of Australasia



**Box 2. Case study - Dr Charlotte Slade<sup>3</sup>**

Dr Charlotte Slade is a clinical immunologist and clinician scientist— one of an increasing number of specialists now incorporating genomics into their field. Dr Slade co-led an immunology project with the Melbourne Genomics Health Alliance. The project provided genomic sequencing to selected patients to evaluate its usefulness for medical practice.

***What conditions did you investigate and why?***

The immune system is responsible for fighting infection in the body; immune disorders occur when the body mistakenly attacks its own body tissues. This project investigated a number of immune disorders that have a known genetic basis. What is common to these conditions is that they create a severe impact on people’s daily lives and are typically very difficult to diagnose accurately.

***Which patients were involved?***

Adults and children who were patients of the Royal Melbourne Hospital, The Royal Children’s Hospital, Austin Health and Monash Medical Centre. These hospitals draw patients from across Victoria.

***What was your role in the immunology project?***

I was the key coordinating point for all the professionals involved, across the four hospitals. We held regular multidisciplinary meetings, involving clinicians and scientists, to decide which patients would most benefit from a genomic sequencing test. Once patients were selected, I helped advise the genetic counsellors who met with them and their families to discuss the testing and what kinds of results might be expected, so that patients were in the best position to decide whether they wished to have the test.

This work brought together my dual roles as a clinical immunologist at the Royal Melbourne Hospital and immunology researcher with the Walter and Eliza Hall Institute.

***What did you achieve?***

Patients with immune conditions often remain undiagnosed and without appropriate treatment for many years. We call this the “diagnostic odyssey”, and it can result in invasive and unnecessary testing, as well as loss of good health and quality of life. Diagnosing infants is particularly difficult, and children with immunological conditions can grow to adolescence or adulthood without a clear diagnosis.

We saw how genomics can help us achieve more accurate diagnosis more quickly for at least some patients. Diagnosis can make an enormous impact on the management of a patient’s condition, as well as guiding more personalised care. Over time, it is expected that the use of genomics will also result in better understanding of the mechanisms of immune responses, leading to the potential for better, more targeted therapies.

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<sup>3</sup> <https://www.melbournegenomics.org.au/professionals/profiles/dr-charlotte-slade>

### Non-genetic professionals

Non-genetic professionals are the mainstream health professional workforce who are not formally qualified in genetics, but whose practice includes care of people who may benefit from – or have undertaken – genomic testing. This encompasses medical specialists (for example, in cardiology, neurology and general practice), nurses, midwives, and allied health professionals (for example audiologists and dieticians). A large section of the workforce is not yet prepared, with evidence from the Australian medical workforce that they lack knowledge, skills, and confidence to practise genomic medicine (Cusack et al., 2021; Nisselle et al., 2021).

Historically, and across all fields of medicine, experts have effectively shepherded colleagues through the introduction of new technologies, for example early use of MRIs, taking on complex cases themselves but supporting the routine practice of their colleagues. This is also evident over many decades in the introduction of new screening and diagnostic procedures for genetic conditions, from early chromosome analysis to now genomic testing. Genetic experts play a critical role developing the genomic capability of this workforce, through structured activities, for example, multidisciplinary teamwork and delivery of genomic education, and *ad hoc* advice on request (McClaren et al., 2020).

While medical specialists report an expectation that they will need to become more proficient in all stages of testing, most currently prefer a model whereby they receive support from genetics services (Cusack et al., 2021; Nisselle et al., 2021). The pace of change means there is an urgency for less reliance on genetics services; however, the reality of achieving this is constrained by the shortage of genetic experts and their capacity to do more than meet patient demand. Key skills and knowledge are required for non-genetic trained professionals to be able to:

- recognise patients for which genomics has utility,
- appreciate and discuss with patients and their families the medical, genetic and/or psychosocial implications of these tests,
- appropriately refer a patient to a genetics service or request a test if within scope of practice,
- understand test result reports, and
- discuss results with the patient and act on those results appropriately.

## Genomics is a multidisciplinary and staged process

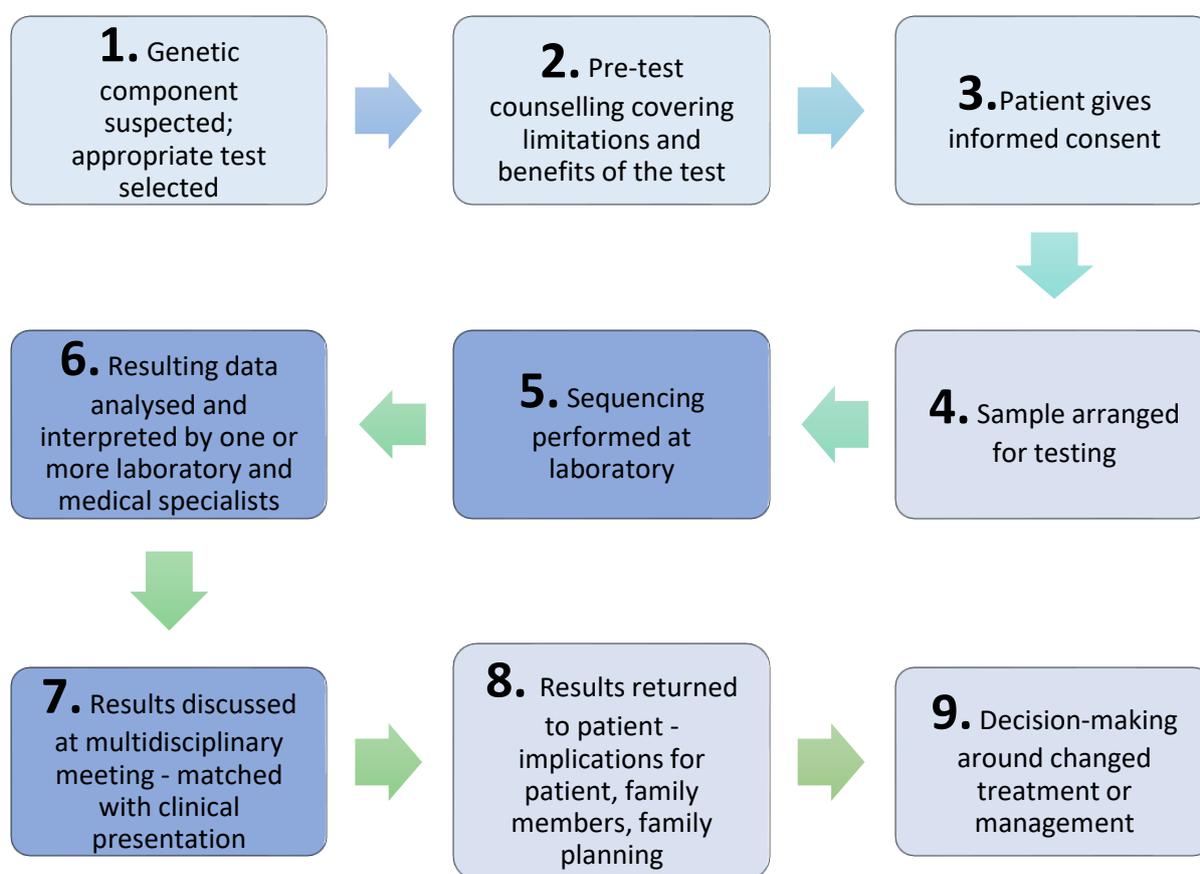
Genomic testing is a complex and staged process involving many different health professionals. It does not just involve a single patient either but the entire family. There is an increased commitment of time involved in the consenting process, testing, interpretation of genomic data, and return of results to the patient and their family.

There is an imperative in genomics to work with multidisciplinary teams. Genomics requires input from a range of disciplines as no one discipline holds enough individual knowledge. This means

multidisciplinary teams are required to create integration of clinical presentation and laboratory data; genetic and non-genetic professionals, medical scientists, and counsellors (Figure 1).

While roles and processes of these teams are still being negotiated and defined, teams have been successfully developed through vocational extension training of junior consultants, building a bridge between genetic and non-genetic specialities (Martyn et al., 2021).

Figure 1: Process map of the stages required for genomic testing which typically involves numerous teams and organisations.



## A landscape view of genomics in Australia

In 2022, Australia is just one of 41 countries that has a national program toward implementing genomics in medicine, a further 86 have smaller projects (Kovanda et al., 2021). This effort is supported by an estimated global investment of US\$14B, international collaborative networks, pooling of data and sharing of lessons learnt around sustainable practice (Stark et al., 2019).

Within Australia, there is a complex context for genomic medicine with a diverse range of stakeholders, organisations, and regulatory, advocacy and governance processes involved.

Additionally, there is strong interdependency between these where the actions and understandings acquired in one sector. For example, increasing real-world use of genomics and industry involved in manufacture and supply of sequencing equipment directly impact the context within which the others operate (see Appendix 1).

### Funding

There is a mosaic of funding for genomic testing in Australia from a variety of sources that clinicians must negotiate to fund a test. These include Medicare Benefits Schedule (MBS) items, state Health Department or health district initiatives, research projects, and philanthropic funding (Long et al., 2021). Individuals may also need to cover the costs themselves to access testing. Most funding sources require specific criteria to be met regarding the suspected condition, age or what diagnostic tests have already been performed. For instance, to be eligible for the genomic testing for childhood syndromes rebate, just one of multiple criteria is that the person must be under 10 years of age.

Funding for testing does not include costs associated with genetic counselling as part of the return of results. The Human Genetics Society of Australasia is in discussion with the MBS Review Advisory Committee regarding mechanisms to address the lack of funding for genetic counsellors through the MBS (2022). Australian Genomics is also in discussion with the Independent Hospital Pricing Authority (IHPA) to reclassify the Activity Based Funding (ABF) codes for genetic counsellors.

Unintended consequences for the genomic workforce from this mosaic of funding have been identified, for example the burden on health professionals and ensuing delays from the need to search for or consult others to find the right scheme to fund their patient's test.

### Pathways into testing

Pathways into testing and transition strategies to move from current practice to incorporation of genomic testing have been worked out for some but not all applications of genomics, mostly through implementation science streams of translational research efforts (Best et al., 2021; Best et al., 2022).

Work is progressing to develop culturally safe and appropriate genomic pathways for Aboriginal and Torres Strait Islander people and others from culturally diverse backgrounds, but more work is needed to translate this into usual care (Bernardes et al., 2014; Dalach et al., 2021; Wild et al., 2013). Some exemplary programs already exist, for example the *Summer Internship for Indigenous Peoples in Genomics* program (SING Australia Supported by Centre of Excellence for Australian Biodiversity and Heritage, and Australian Genomics), and the recently funded National Indigenous Genomics Consortium, at the South Australian Health and Medical Research Institute (SAHMRI, 2021).

**Box 3: Summary of key workforce issues for genomic medicine**

- Genomic testing is a complex, multistage process that requires both knowledge and acquisition of skills.
- There is a demonstrated lack of knowledge, skills and confidence in the wider medical workforce, and the same is assumed for nursing and allied health staff.
- There are long waitlists for expert genetics services due to an increase in demand.
- Genomic workforce shortages are due in part to constraints on numbers of professionals being trained, which in turn are linked to limited workplace rotations available.
- The genetics evidence base is constantly evolving, providing challenges for health professionals to stay up to date and for education courses to retain currency.
- Pathways into testing, and the roles and responsibilities of genetic and non-genetic professionals tailored for each speciality (for example cancer, neonatal, cardiac, renal) are still under development and sometimes lack rigorous evaluation.
- Pathways for Aboriginal and Torres Strait Islander people, those with CALD backgrounds, and other priority populations to receive culturally safe and appropriate care are still being developed.
- Professional roles within genomics are still evolving and being negotiated (for example, medical science and biostatistician roles).
- The mosaic of tightly constrained funding schemes for genomic testing is adding to the burden of having to match the criteria of funding to the patient for many genetic professionals.

## Ensuring genomics workforce sustainability

Genomics has an important role to play in the future of Australia's health system. Improving health outcomes of Australians by answering uncertainties for individuals and families with genetic conditions, alerting people in the general population that they are at increased risk, aiding clinicians to tailor patient management plans, and reducing unnecessary procedures. The benefits are immense. Yet, despite the incredible advances in science that make genomics so valuable, the health system will struggle to meet demand until workforce issues are addressed.

A coordinated national strategy is needed to ensure all Australians benefit from genomics and the workforce is sustainable. The establishment of Genomics Australia from 1 January 2024 is a welcome step.

### Develop agile, multidisciplinary teams around forecasted genomic demand

- Defining and building a robust model for genomic multidisciplinary teams, for example, through funded partnership of existing teams with health service researchers, that can be replicated and adapted to different health contexts will be essential.
- Investment in secure digital platforms will be required to facilitate linkage of isolated health professionals to multidisciplinary teams and to build a community of practice, particularly in rural and regional areas.
- The expansion of time-limited data collection is needed to enable forecasting of future workforce issues with some granularity. Specifically, trends in demand for testing, specialist genomic workforce capacity, and non-genetics workforce practice.

### Build the genomics skill base for a systematic, national approach to professional education

- Core capabilities in genetics and genomics for medical students have been developed by the Human Genetics Society of Australasia. These capabilities should be adopted by medical programs around Australia.
- A genetic competency framework for non-genetic health professionals should be developed that can guide professional colleges and educators in the provision of genomics education. This will be essential to meeting future workforce needs and ensuring the capability of the workforce currently in practice.
- Continuing professional development should be supported at a national level. This will ensure the ongoing currency of, and access to, genomics education developed with national research funding.
- The development of “genomic champions” should be supported in non-genetic specialties, through recognition and funding of vocational extension training for health professionals in non-genetic specialties. This could be modelled on the small number of existing programs.
- Culturally safe and appropriate genomic care for Aboriginal and Torres Strait Islander people, those with CALD backgrounds, and other priority populations must be considered in the genomic education of health professionals. This has begun in some areas and can be progressed by the use of co-design frameworks and attracting people from diverse backgrounds to a career in genomics.

### Ensure specialist genomic workforce capacity and capability

- There are only five Commonwealth supported places for Master of Genetic Counselling students (2022). To permit a gradual increase in student numbers and improve access for students from diverse backgrounds, this number should be increased.
- More funded training positions for medical scientists and genetic pathologists with the Royal College of Pathologists Australasia should be established.
- Recognised career paths and Commonwealth support for postgraduate qualifications for emerging roles in genomics should be developed.

### Reform for funding models that can meet contemporary demands for genomic testing

- Genomics service constraints arising from substantial unpaid work evident across Australia must be addressed. This will require reforming current funding models that support the delivery of genetic services overseen by IHPA, and Local Health ABF and staffing models; and subsequent adjustments to staffing levels, role descriptions, workloads and organisational changes.
- The time needed by individual genomic experts to mentor, supervise, and train non-genetic professionals must be acknowledged and appropriately funded.
- HGSA submission to the MBS Review Advisory Committee *The Provision of Services by FHGSA Registered Clinical Genetic Counsellors* must be supported.

### Facilitate embedding genomics into usual practice must be supported

- In order to leverage existing programs and embed genomics into usual care, the National Health and Medical Research Council, Medical Research Future Fund and state Medical Research Councils must continue to support translational research funding in genomics.
- Funding should also include implementation science support for defining patient pathways into genomic testing and models of shared genomic care.

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Appendix 1

Rich picture of the landscape of implementation of genomics in Australia (Long et al., 2021)



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*AHHA acknowledge the Aboriginal and Torres Strait Islander peoples as Australia's First Nation Peoples and the Traditional Custodians of this land. We respect their continued connection to land and sea, country, kin, and community. AHHA also pays our respect to their Elders past, present, and emerging as the custodians of knowledge and lore.*