Clinical and population genomic data for Aboriginal and Torres Strait Islander peoples is crucial for equitable provision of expanded carrier screening in Australia.

Easteal, Simon1*; Arkell, Ruth, M.2; Balboa, Renzo, F.1; Bellingham, Shayne, A.1; Brown, Alex, D.3,4; Calma, Tom5; Cook, Matthew, C.6; Davis, Megan7; Dawkins, Hugh, J.S.8–12; Dinger, Marcel, E.13; Dobbie, Michael, S.1,2; Farlow, Ashley 1,14; Gwynne, Kylie, G.5,15; Hermes, Azure1; Hoy, Wendy, E.16; Jenkins, Misty, R.17,18; Jiang, Simon, H.6; Kaplan, Warren19; Leslie, Stephen 1,14; Llamas, Bastien20; Mann, Graham, J.2; McMorran, Brendan, J.2; McWhirter, Rebekah, E.21; Meldrum, Cliff, J.22; Nagaraj, Shivashankar, H.23; Newman, Saul, J.24; Nunn, Jack, S.25; Ormond-Parker, Lyndon26; Orr, Neil, J.5; Paliwal, Devashi 1,2; Patel, Hardip, R.1; Pearson, Glenn27; Pratt, Greg, R.28; Rambaldini, Boe5; Russell, Lynette, W.29; Savarirayan, Ravi30; Silcocks, Matthew 1,14; Skinner, John, C.5; Souilimi, Yassine 1,31; Vinuesa, Carola, G.2; The National Centre for Indigenous Genomics1; Baynam, Gareth32–34**;
Affiliations

1 National Centre for Indigenous Genomics, Australian National University, Canberra, Australian Capital Territory 2600, Australia.

2 John Curtin School of Medical Research, Australian National University, Canberra, Australian Capital Territory 2600, Australia.

3 Aboriginal Health Equity, South Australian Health and Medical Research Institute, Adelaide, South Australia 5000, Australia.

4 Faculty of Health and Medical Sciences, University of Adelaide, Adelaide, South Australia 5005, Australia.

5 Poche Centre for Indigenous Health, University of Sydney, Sydney, New South Wales 2006, Australia.

6 Department of Immunology, Canberra Hospital, Canberra, Australian Capital Territory 2606, Australia.

7 UNSW Law, University of New South Wales, Sydney, New South Wales 2052, Australia.

8 HBF Health Limited, Perth, Western Australia 6000, Australia.

9 School of Medicine, The University of Notre Dame Australia, Sydney, New South Wales 2010, Australia.

10 Sir Walter Murdoch School of Policy and International Affairs, Murdoch University, Murdoch Western Australia 6150, Australia.

11 Division of Genetics, School of Biomedical Sciences, University of Western Australia, Nedlands, Western Australia 6008, Australia.
12 Centre for Population Health Research, Curtin University of Technology, Bentley, Western Australia 6102, Australia.

13 School of Biotechnology and Biomolecular Sciences, University of New South Wales, Sydney, New South Wales 2052, Australia.

14 Melbourne Integrative Genomics, University of Melbourne, Melbourne, Victoria 3010, Australia.

15 Faculty of Medicine and Health Sciences, Macquarie University, Sydney, New South Wales 2113, Australia.

16 Faculty of Medicine, University of Queensland, Brisbane, Queensland 4072, Australia.

17 Immunology Division, The Walter and Eliza Hall Institute of Medical Research, Parkville, 3052, Victoria, Australia.

18 La Trobe Institute of Molecular Science, La Trobe University, Bundoora, Victoria 3086, Australia.

19 Informatics, Garvan Institute of Medical Research, Sydney, New South Wales 2010, Australia.

20 Centre of Excellence in Australian Biodiversity and Heritage, School of Biological Sciences, The Environment Institute, University of Adelaide, Adelaide, South Australia 5005, Australia.

21 Centre for Law and Genetics, Faculty of Law, University of Tasmania, Hobart, Tasmania 7001, Australia.

22 NSW Health Pathology, Sydney, New South Wales 2065, Australia.

23 Institute of Health and Biomedical Innovation, Queensland University of Technology, Brisbane, Queensland 4000, Australia.
24 Biological Data Science Institute, Australian National University, Canberra, Australian Capital Territory 2600, Australia.

25 Public Health, La Trobe University, Melbourne, Victoria 3086, Australia.

26 Melbourne School of Population and Global Health, University of Melbourne, Melbourne, Victoria 3010, Australia.

27 Aboriginal Health, Telethon Kids Institute, Perth, Western Australia 6009, Australia.

28 Aboriginal and Torres Strait Islander Health, QIMR Berghofer Medical Research Institute, Brisbane, Queensland 4006, Australia.

29 Centre of Excellence in Australian Biodiversity and Heritage, Monash Indigenous Studies Centre, Monash University, Melbourne, Victoria 3800, Australia.

30 Victorian Clinical Genetic Services, Murdoch Children’s Research Institute, and University of Melbourne, Parkville, Victoria 3052, Australia.

31 School of Biological Sciences, The Environment Institute, University of Adelaide, Adelaide, South Australia 5005, Australia.

32 Genetic Services of Western Australia, Department of Health, Government of Western Australia, Perth, Western Australia 6004, Australia.

33 The Western Australian Register of Developmental Anomalies, Department of Health, Government of Western Australia, Perth, Western Australia 6004, Australia.

34 School of Medicine, Division of Paediatrics and Telethon Kids Institute, University of Western Australia, Perth, Western Australia 6009, Australia.
Corresponding authors

*Professor Simon Easteal
Email: simon.easteal@anu.edu.au

**Dr Gareth Baynam
Email: Gareth.Baynam@health.wa.gov.au
Abstract

Expanded carrier screening (ECS) for recessive monogenic diseases requires prior knowledge of genomic variation including DNA variants that cause disease. The composition of pathogenic variants differs greatly among human populations, but historically research about monogenic diseases has focused mainly on people with European ancestry. By comparison, less is known about pathogenic DNA variants in people from other parts of the world. Consequently, inclusion of currently under-represented Indigenous and other minority population groups in genomic research is essential to enable equitable outcomes in ECS and other areas of genomic medicine. Here we discuss this issue in relation to the implementation of ECS in Australia, which is currently being evaluated as part of the national Government’s Genomics Health Futures Mission. We argue that significant effort is required to build an evidence base and genomic reference data so that ECS can bring significant clinical benefit for many Aboriginal and/or Torres Strait Islander Australians. These efforts are essential steps to achieving the Australian Government’s objectives and its commitment “to leveraging the benefits of genomics in the health system for all Australians”. They require culturally safe, community-led research and community involvement embedded within national health and medical genomics programs that ensure that new knowledge is integrated into medicine and health services in ways that address the specific and articulated cultural and health needs of Indigenous people. Until this occurs, people who do not have European ancestry are at risk of being, in relative terms, further disadvantaged.
Introduction

Genomic technologies have enabled major advances in understanding and treating rare monogenic diseases. Greater accessibility to genomic data and the knowledge to interpret it have: improved diagnostic rates for existing conditions; greatly expanded the number of diseases for which diagnostic tests are available; led to greater understanding of biological processes underlying pathology; enabled development of better and targeted therapies; and resulted in improved prenatal and preimplantation testing\(^1\)\(^{-}\)\(^4\). Genomic technologies have also created the possibility of pre-conception expanded carrier screening (ECS), by which prospective parents are simultaneously screened as potential carriers of a range of different recessive diseases\(^5\)\(^{-}\)\(^7\).

Pre-reproductive carrier screening is generally targeted at specific genes and carried out where there is increased risk of a child being born with a specific recessive condition due to ancestry or based on clinical information\(^8\). It has been extremely effective, e.g., in reducing the incidence of Tay-Sachs disease (MIM: 272800) in Ashkenazi Jewish populations around the world\(^9\)\(^,\)\(^10\). ECS is an extension of this approach that involves simultaneous screening for many pathogenic variants responsible for a broad range of diseases in the general population. This broad-scale approach to screening is achieved by sequencing the entire genomes (genome sequencing) or the fraction of the genome that encodes proteins – the exome (exome sequencing) – of prospective parents. Although data are obtained for the whole genome or exome, screening is often targeted at a predetermined subset of genes and/or variants\(^11\)\(^,\)\(^12\).

The Australian government is evaluating the potential benefits and challenges that ECS presents\(^7\)\(^,\)\(^12\)\(^{-}\)\(^17\) with a view to its introduction into the national healthcare system\(^18\). Our focus here is on the significant challenges of achieving inclusion and equitable benefits for
Indigenous Australians from this approach and, by extension, medical genomics generally. While our focus is on Indigenous Australians, many of the points we raise apply to other groups that are under-represented in current genomic reference data.

Ethical, cultural, social and policy considerations are of over-riding importance in genomics. Implementation of ECS in Aboriginal and Torres Strait Islander communities raises questions about: the cultural appropriateness of screening in different communities; how prospective parents should be counselled and appropriately informed about ESC; the means by which consent should be obtained; the potential impact on social and cultural norms; the potential for group, family and/or individual stigmatisation; how screening can be harmonised with cultural practices, lifestyles and traditional concepts; whether the autonomy of patients, families and communities can be preserved; the proportion of the population likely to benefit from this approach; how screening will be administered through community controlled and other local health services; and whether there is the capacity for culturally safe counselling and follow-up clinical care.

Fully articulating these complex issues for health professionals and Indigenous communities is a substantial undertaking that needs adequate resourcing to ensure appropriate support. Consequently, we address only the salient points here. Our main focus is on scientific evidence about genetics and its medical implications for Indigenous Australians, as a foundation to better inform this process.

The core challenge for ECS implementation is lack of knowledge about genomic variants in Indigenous populations and of appropriate clinical and genomic reference data. Carrier screening depends on prior knowledge of pathogenic variants, most of which comes from studies of people of European ancestry, which may have limited or sub-optimal applicability to other populations19–27.
As Australia is a culturally and ancestrally diverse nation, there is a need to recognise how genomic information is interpreted, incorporated and translated meaningfully in the lives, experiences, and healthcare of individuals from diverse cultural and ethnic backgrounds. In particular, there is a national imperative to ensure equitable benefit for Aboriginal and Torres Strait Islander Australians, who collectively experience significant disparity in morbidity and mortality\textsuperscript{28} and access to health services\textsuperscript{28,29} compared with non-Indigenous Australians.

We discuss how the involvement of Indigenous people must be fully embedded within national health genomics initiatives such as ECS to ensure that the needs of Aboriginal and Torres Strait Islander people are met, and to ensure that these initiatives deliver outcomes consistent with the equity principles that underpin Australia’s public healthcare system: universal cover and universal access.

**Medical genomics in Australia**

The national introduction of ECS is being evaluated as part of the Genomics Health Futures Mission (GHFM), a program funded by the Medical Research Future Fund (MRFF). Projects funded through the GHFM operate within the policy settings provided by Australia’s National Health Genomics Policy Framework (NHGPF) developed by the Australian Health Ministers’ Advisory Council (AHMAC) and agreed by the Council of Australian Governments (COAG) Health Ministers in November 2017 (Box 1). The NHGPF recognizes the importance of addressing the requirements for Indigenous inclusion in the implementation of genomic medicine (Box 1).
Box 1. The National Health Genomics Policy Framework, Medical Research Future Fund (MRFF) and Genomic Health Futures Mission (GHRM)

The NHGPF provides the blueprint for embedding genomics in the Australian health system. It “presents a shared commitment to leveraging the benefits of genomics in the health system for all Australians”

The principles underpinning NHGPF priorities are:

- The application of genomic knowledge is ethically, legally and socially responsible and community trust is promoted
- Access and equity are promoted for vulnerable populations
- The application of genomic knowledge to health care is supported and informed by evidence and research.

Recognising the importance of equity and inclusion, particularly in relation to Indigenous Australians, the priority areas of action of the National Health Genomics Policy Framework 2018–2021 include:

- 1.5. exploring the potential for discrimination, and evaluating the delivery of genomic services in terms of being accessible, appropriate and culturally secure and responsive for Aboriginal and Torres Strait Islander peoples.
- 5.2. Promote culturally safe and appropriate genomic and phenotypic data collection and sharing that reflects the ethnic diversity within the Australian population, including for Aboriginal and Torres Strait Islander peoples.

The intended outcomes of the Medical Research Future Fund (MRFF) are:

1. life changing discoveries such as new treatments, drugs and devices
2. continuous improvement and innovation in the health system that benefits all Australians
3. strengthening domestic research capacity through support, collaboration and the development of expert talent
4. positioning Australia’s health and medical research sector at the forefront of the innovation economy
5. improving Australia’s reputation as a global leader in health and medical research.

The objective of the Genomics Health Futures Mission (GHFM) is to:

1. deliver better diagnostics and targeted treatments
2. avoid unnecessary health costs
3. improve patient experience and outcomes.

The fund supports research projects that aim to:

1. provide the pathways for the development of new diagnostics, medicines and treatments from genomics research
2. expand genomics research effort and reach, allowing researchers and commercial partners to sustain efforts in discovery
3. build evidence for scaling applications, and build new markets
4. ensure that later stage translation and flagship work is not hampered by a lack of investment in early research.
Pathogenic variants are generally rare and population-specific

Most monogenic diseases are caused by many different DNA variants in one or more specific genes, almost all of which are rare. These variants may occur only in people with ancestry from a particular geographic region, in one small community, or even in a single family. Thus, for example, more than 2,000 different known pathogenic variants in the CFTR gene (MIM: 602421) can cause the recessive monogenic disease cystic fibrosis (CF; MIM: 219700). Approximately 1 in 3,000 people are affected by CF in northern Europe. Elsewhere, however, it is much rarer and is usually caused by local, rare variants that are not found in European patients. Thus for example, in China and in the many substantial Chinese communities elsewhere in the world, where CF, although rare, affects an estimated 20,000 people, carrier screening panels designed for ancestrally European populations fail to detect most CF carriers.

The rarity and geographically restricted origins of pathogenic variants have important consequences for Australia’s diverse society.

1. The makeup of pathogenic variants is likely to be unique, reflecting the unique diversity of Indigenous peoples and the ancestral makeup of settlers and immigrants.

2. For the same reasons, it is likely that there are many pathogenic variants that have not been previously characterized. These novel variants may cause already described clinical phenotypes. It is likely, however, that even if they have similar molecular properties to known variants some will cause different phenotypes. These include milder or more serious forms of disease and different treatment responses. Clinical and functional investigation will generally be required to establish their pathogenicity and associated disease phenotypes.
3. For recessive diseases, many novel combinations of pathogenic variants are likely. A recessive disease can be caused by \( \frac{n(n-1)}{2} + n \) combinations of \( n \) pathogenic variants. Only a small fraction of these combinations can occur where the geographic distribution of variants is restricted. However, in a society with many people of mixed ancestry many novel combinations of variants are likely. These novel combinations may cause novel disease phenotypes and have differing effects on treatment responses.

4. Genomic-background, environment and lifestyle are more likely to influence the phenotypic manifestation of recessive diseases caused by pathogenic variants, even potentially causing normally pathogenic variants to become benign\(^{35}\) or normally benign variants to become pathogenic\(^{36,37}\) because:

   I. The environment and lifestyle of many people has rapidly changed due to alterations in economic or social circumstances, changes in diet, or as a result of displacement or migration.

   II. There are many people of mixed ancestry in whom the effect of a variant on disease may have changed after it arrived in a genomic background different to the one in which it had previously existed.

**Pathogenic variants in Aboriginal and Torres Strait Islander communities**

Global prevalence estimates\(^{7,38}\) suggest that, to a first approximation, more than 30,000 Aboriginal and Torres Strait Islander people may be affected by monogenic diseases, and that many more may be carriers of pathogenic variants. Many of these variants will be different from those causing the same diseases in people with ancestry from Europe and other parts of the world. Many may cause either formerly unknown diseases or phenotypic
manifestations of known diseases that have not previously been encountered in a clinical setting.

Some Aboriginal and/or Torres Strait Islander people have pathogenic variants inherited from non-Indigenous ancestors. However, with few exceptions, like Machado-Joseph Disease\textsuperscript{39} and a complex phenotype resulting from an \textit{MTOR} gene variant\textsuperscript{40}, little is known about pathogenic variants originating within Indigenous populations.

Unpublished data compiled by the National Centre for Indigenous Genomics (NCIG) for 160 people from four Aboriginal communities show that:

1. Approximately 25\% of all DNA variants in the genome of an Aboriginal person, disregarding variants inherited from non-Aboriginal ancestors, are unknown in people from outside Australia. Among the large number of Aboriginal and Torres Strait Islander-specific variants there will be some that are pathogenic. These will not be represented in international or Australian clinical databases or in current screening panels. These databases and panels may, therefore, currently be of limited value for screening in Aboriginal and Torres Strait Islander communities.

2. Of these Aboriginal-specific variants, \textasciitilde 40\% are likely to be found in a single region or community. Overall, based on $F_{ST}$ distances\textsuperscript{41} and comparison with data from the Simons Genome Diversity Project\textsuperscript{42}, genomic differences among Aboriginal communities across Australia are as great as those between populations across Europe and Asia combined. Thus, for example, using information about people from the Northern Territory as a basis for treating people in South Western Australia, would be equivalent to treating people with British ancestry on the basis of information about people from Cambodia.
These data can be accessed and used for specific purposes, as determined by the NCIG Indigenous-majority Board, in accordance with the CARE data sovereignty principles and the National Centre for Indigenous Genomics Statute, 2016 (Cth).

Current lack of evidence means that for many people with Aboriginal and/or Torres Strait Islander ancestry, ECS will produce greater uncertainty, revealing more ‘likely-pathogenic variants’ (LPVs) and ‘variants of unknown significance’ (VUSs) than for those with ancestry from Europe and other parts of the world where the causes of monogenic diseases are better understood. This uncertainty could potentially lead to inappropriate clinical intervention if benign variants are incorrectly reported as pathogenic, as has occurred elsewhere.

The risk of variants being falsely reported as pathogenic can be avoided by increasing the threshold of evidence required to assign pathogenicity. This approach, which reduces the risk of false positive reports, also tends to cause under-reporting of pathogenic variants because some do not meet the higher threshold of evidence. The same evidence-based criteria will have a differential effect when applied to populations for which there are different levels of available evidence. Pathogenic variants will tend to be under-reported to a greater extent in populations where there is a relative lack of evidence, as there is for people with Indigenous ancestry.

The result is greater “residual risk”, i.e. more couples with a risk of having an affected child that is not identified by ECS. High residual risk is equivalent to low sensitivity, i.e., high rates of false negative findings. Thus, when residual risk is high, negative findings have little predictive value. For the great majority of prospective parents who test negative, testing provides little information. Thus, participation in testing may raise awareness of potential risk but leave most participants with high levels of residual uncertainty about their own risk.
In addition, increasing the threshold of evidence for pathogenicity reduces the “yield”, i.e. the number of couples identified as being at risk. The result is that, overall, fewer people benefit from screening. If the expected yield for the general population is 1–2%, the lower expected yield for couples with Indigenous ancestry means that many hundreds of couples may be screened without any of them receiving a report that they are at risk of giving birth to a child with a monogenic disease.

Lack of knowledge about variant pathogenicity adds to the challenges of counselling prospective Aboriginal and Torres Strait Islander parents and of supplying the accurate information they need in order to make informed decisions about undergoing ECS.

Novel variants identified through ECS can be functionally and clinically investigated. These investigations are unlikely, however, to provide useful information to prospective parents because of the amount of time required to carry them out. They may, nevertheless, give rise to new evidence that improves the quality of screening for future patients.

These indirect benefits might provide ethical justification for ECS as a medical intervention if it were not possible to obtain them in other ways, even when there is little potential benefit and considerable risk for patients. Novel pathogenic variants can, however, be more effectively identified and their phenotypic effects better characterized at greatly reduced risk through direct clinical investigation of affected patients and their families. This more direct approach is greatly enhanced by characterization of genomic variation in patient communities, which can be critically important for variant discovery and for correct assignment of pathogenicity.
How to address the current disparity?

The validity of ECS depends on population reference data and a preexisting evidence-base linking specific DNA variants with disease phenotypes, which has been painstakingly built up through decades of careful direct clinical investigation of affected patients and relevant family members\(^{50, 34, 51}\) mainly in people of European ancestry. Equitable inclusion of Indigenous Australians in the benefits of ECS, and medical genomics more generally, requires a similar level of evidence.

The critical importance of ancestry in the many other areas of health care where genomics now plays an important role\(^ {19-26}\) has led to programs aimed at achieving diversity in genomics, e.g., India\(^ {52}\), Asia\(^ {53}\); Africa\(^ {54}\); Aotearoa/New Zealand\(^ {55}\), USA\(^ {56}\).

An equitable approach in Australia would require prioritization of research involving people of Aboriginal and/or Torres Strait Islander descent, as well as other under-represented groups, as an integral part of national medical genomics programs. National programs should include: 1. Detailed characterization of genomic variation in Aboriginal and Torres Strait Islander peoples; and 2. Careful study, with community involvement and leadership, of pathogenicity and the general clinical, cultural and social consequences of diseases.

Programs must be designed and sufficiently resourced to include Indigenous community leadership to ensure appropriate research conduct at a time when community acceptance of genomics is critically important\(^ {57}\). As in other areas of healthcare\(^ {59, 58, 59}\), extending approaches developed for the general population or retrofitting systems that were not designed to meet the specific needs of Indigenous people will not be effective and may do more harm than good. Hence, there is a need, at all levels and stages, for Indigenous co-
design and development and incorporation of Indigenous data governance and custodianship as the foundations of national medical genomics programs.

Finally, it is essential to account for the significant genomic differences as well as the significant socio-cultural differences among the many Indigenous communities across the Australian continent.

**Conclusion**

ECS is one of many medical applications of genomics that, collectively, can transform the healthcare system for the better. For these developments to contribute usefully to the health and wellbeing of Australians with Indigenous ancestry the current dearth of evidence and lack of reference data must be addressed. To ensure their culturally safe conduct, national genomic medicine programs must ensure that Indigenous communities are empowered by incorporating Indigenous leadership, co-conceptualization and co-design and implementing the principles of Indigenous sovereignty over genomic data.

Australia has an opportunity to embrace the challenges presented by the cultural and ancestral diversity of its people to deliver research and clinical outcomes with significant global impact. New discoveries leading to therapeutic innovation are more likely from clinical investigation of people whose health and disease have previously been neglected, and of illnesses, which, until now, have been ignored, than from focussing on better understood problems in well-studied populations.

In addition, addressing the specific requirements of Australians with Indigenous ancestry and other under-represented groups would directly support the Australian Government’s commitment to equity and inclusion. It would redress past inequities and provide a model for better healthcare practice in Australia and internationally.
Australia has a unique opportunity for medical genomics innovation leading to improved prediction, prevention, treatment and cure of disease that is based on the distinctive characteristics of genomic diversity and its relationship to disease in Indigenous people, a reflection of their continuing ancient presence on the Australian continent\textsuperscript{60}. This comparative advantage derives from Australia’s ancient history and geographical isolation. In realising it, the central role and importance of Aboriginal and Torres Strait Islander peoples must be recognised, they must be at the forefront of national programs, and they must stand to gain an equitable share of the resulting benefits.
Declaration of interests

None of the authors has a conflict of interest.

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Author/s:
Easteal, S; Arkell, RM; Balboa, RF; Bellingham, SA; Brown, AD; Calma, T; Cook, MC; Davis, M; Dawkins, HJS; Dinger, ME; Dobbie, MS; Farlow, A; Gwynne, KG; Hermes, A; Hoy, WE; Jenkins, MR; Jiang, SH; Kaplan, W; Leslie, S; Llamas, B; Mann, GJ; McMorran, BJ; McWhirter, RE; Meldrum, CJ; Nagaraj, SH; Newman, SJ; Nunn, JS; Ormond-Parker, L; Orr, NJ; Paliwal, D; Patel, HR; Pearson, G; Pratt, GR; Rambaldini, B; Russell, LW; Savarirayan, R; Silcocks, M; Skinner, JC; Souilmi, Y; Vinuesa, CG; Baynam, G

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