THE UNIVERSITY OF MELBOURNE

Melbourne School of Population and Global Health

Better Indigenous Genetic (BIG) Health Services project

POLICY BRIEF–August 2020

Better Indigenous Genetic (BIG) Health Services project

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PROJECT AIMS AND PURPOSE

The rapidly developing field of genomics has the potential to revolutionise how health care is delivered. With increasing genomics capability, the opportunities for positive impacts on clinical medicine are immense (1). Yet, how these impacts are best translated to enhanced patient outcomes is not well defined. There is evidence of significant unmet need for clinical genetics and genetic counselling among Aboriginal and Torres Strait Islander Australians (2, 3), and where services are available there are considerable gaps persist in terms of the comprehensiveness of those services and continuity of care. With an increasing trend of integrating genomics into clinical practise, addressing these issues is crucial to improving provision of effective genetic health care services to Aboriginal and Torres Strait Islander Australians (4). This imperative has been recognised in the National Health Genomics Policy Framework 2018–2021 (5) which includes a focus on equity as its first strategic priority.

The Better Indigenous Genetic (BIG) Health Services project was funded by the NHMRC, Lowitja Institute, MJD Foundation, Northern Territory Health Department, Queensland Health and Western Australian Department of Health. The BIG Health Services project worked with clinical genetic health services to develop more effective models for meeting the needs of Aboriginal and Torres Strait Islander Australians. The specific aims of the project were to:

1. Explore the extent to which current models of genetic service provision and follow-up support meet the needs of Aboriginal and Torres Strait Islander patients;
2. Identify barriers and facilitators to effective provision of genetic services and follow-up support for Aboriginal and Torres Strait Islander patients;
3. Develop recommendations to improve the ability of genetic services to better meet the patient and family needs, and to support improved access to follow-up services.

CLINICAL GENETICS SERVICES

Clinical Genetic Services are at the vanguard of the advances brought about by the shift from medical management of patients to genome-level diagnostics and precision medicine. The services they offer encompass a broad range of capabilities relevant to patients throughout the life course, from pre-implantation genetic diagnosis to prenatal, paediatric and adult genetics (1, 4). Even where treatment does not currently exist, the considerable benefits of genetic diagnosis include providing certainty, reproductive confidence, reducing unnecessary investigations, increasing support and access to education, as well as providing a platform for community activism.

Northern Territory Genetics Service (NTGS) operates as a specialist outpatient service which has been contracted by the NT Department of Health to Victorian Clinical Genetics Services (VCGS) since 2014. This report includes data for the period 2014-2018. During this time, all clinical staff were based in Victoria and NTGS was operated on a fly-in fly-out basis with two clinical geneticists and one genetic counsellor traveling to the NT approximately four times annually. Clinics were held in Darwin and Alice Springs. Telehealth appointments are also available where appropriate. Due to the intermittent nature of NTGS appointment availability, all clinics are general. In the period from 2014-2018, NTGS saw 722 patients, 20.4% of whom identified as Aboriginal and/or Torres Strait Islander.

Genetic Health Queensland (GHQ) operates as a state-wide hub and spoke model service. The hub is Brisbane-based and offers general clinics as well as specialised clinics for cardiac genetics, renal genetics, prenatal counselling and paediatric genetics. Visiting clinical genetics services are provided at the Gold Coast, Toowoomba, Nambour, Bundaberg, Rockhampton, Mackay, Townsville and Cairns Hospitals. Telehealth services are also available to patients throughout the state. In the period from 2014-2017, GHQ saw 5443 patients, 4.7% of whom were Aboriginal and/or Torres Strait Islander.

Genetic Services of Western Australia (GSWA) operates as a state-wide hub and spoke model service. The hub is Perth-based and offers general and antenatal clinics as well as specialised clinics for cancer, paediatric, dermatology and eye disease genetics. GSWA conducts outreach clinics in Albany, Bunbury, Geraldton, Joondalup, Kalgoorlie, Port Hedland and Rockingham throughout the year. In the period from 2015-2018, GSWA saw 9350 patients, 3.4% of whom identified as Aboriginal and/or Torres Strait Islander.

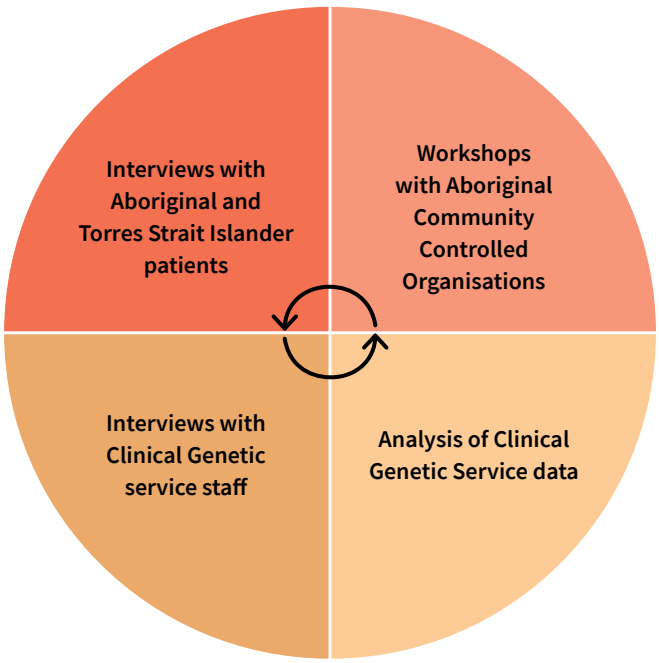
METHODS

Governance of the BIG health services project included a majority-Aboriginal and Torres Strait Islander Project Reference Group and an End-User Group consisting of Aboriginal people affected by or at risk of genetic conditions. These groups met annually to discuss project progress.

A mixed-methods approach was used, including the analysis of service referral and appointment data (n=15,837, 4.5% Aboriginal and Torres Strait Islander) and qualitative data from the following sources:

- Aboriginal and Torres Strait Islander patients/carers attending clinical genetics services (n=73)
- Workshops with Aboriginal Community Controlled Health Services (n=3)
- Clinical genetics/health service staff (n=24).

Information from these data was integrated to inform the development of recommendations included in this report. The aim of these recommendations is to improve the extent to which the services meet the needs of Aboriginal and Torres Strait Islander Australians.



RESULTS

Equity in referral

Table 1 summarises the rates of referral to clinical genetic services in each jurisdiction and in total. Overall, Aboriginal and Torres Strait Islander Australians were 24% less likely than other Australians to be referred to clinical genetic services. This was the case for both paediatric and adult patients in all jurisdictions with the exception of Queensland, where paediatric referrals were higher for Aboriginal and Torres Strait Islanders. Disparities in referral were greatest for older adults. Prenatal and cancer referrals were lower for Aboriginal and Torres Strait Islander Australians compared to other Australians in all jurisdictions. Disparities in referrals for rare and other diseases varied between jurisdictions. With the exception of very remote and inner regional areas in WA, there were no disparities in referrals between Aboriginal and Torres Strait Islander Australians and other Australians due to remoteness.

Table 1: Referrals to clinical genetic services for Aboriginal and Torres Strait Islander (ref) compared to other Australians

	NT (2014-18)	WA (2015-18)	QLD (2014-17)	Total
	RR category			
Adult/Paediatric referral				
Paediatric (0-17 years)	Lower	Lower	Higher	Lower
Adult (18+ years)	Much Lower	Lower	Lower	Lower
Age				
0-9 years	Lower	Lower	Higher	Same
10-19 years	Lower	Lower	Same	Same
20-29 years	Lower	Lower	Same	Lower
30-39 years	Much lower	Lower	Lower	Lower
40-49 years	Much Lower	Much Lower	Lower	Much Lower
50+ years	Much lower	Lower	Same	Lower
Type of referral				
Prenatal referral	Much lower	Much lower	Lower	Much Lower
Rare disease/other	Lower	Same	Higher	Lower
Cancer	Much lower	Lower	Lower	Much Lower
Remoteness				
Major city		Same	Same	Same
Inner regional		Lower	Higher	Same
Outer regional	Same	Same	Higher	Higher
Remote	Same	Same	Same	Same
Very remote	Much Lower	Lower	Same	Much Lower

Ref=Aboriginal and Torres Strait Islander Rating=Much lower: RR LCI 95% >2 ,Lower: RR LCI 95% >1 and <=2 , Same: RR 95% CI includes 1, Higher: UCI 95% RR<1

Equity in attendance

Table 2 summarises the rates of attendance at clinical genetic services in each jurisdiction and in total. Overall, there was less disparity in attendance between Aboriginal and Torres Strait Islander and other Australians (15% less likely to attend) than disparity in referral. This suggests that lower rate of referral is a greater driver of inequity in service use. Attendance was the same or lower among Aboriginal and Torres Strait Islander patients compared to other patients for both adult and paediatric services, with the exception of people referred for prenatal appointments in Western Australia. The observed differences were driven by lower attendance rates among Aboriginal children in NT and WA and younger Aboriginal adults in NT. Attendance for cancer-related appointments were the same among Aboriginal and Torres Strait Islander patients compared to other patients except in WA where they were lower. Attendance for rare and other disease appointments were the lower among Aboriginal and Torres Strait Islander patients compared to other patients. Attendances were lower among Aboriginal and Torres Strait Islander patients compared to other patients in major cities and outer regional areas in NT and WA.

Table 2: Attendance at clinical genetic services for Aboriginal and Torres Strait Islander (ref) compared to other Australians

	NT	WA	QLD	Total
	RR category			
Adult Paediatric referral				
Paediatric (0-17 years)	Lower	Lower	Same	Lower
Adult (18+ years)	Lower	Lower	Lower	Lower
Age				
0-9 years	Lower	Lower	Same	Lower
10-19 years	Lower	Same	Same	Lower
20-29 years	Lower	Same	Same	Lower
30-39 years	Same	Same	Same	Same
40-49 years	Same	Same	Same	Same
50+ years	Same	Same	Same	Same
Sex				
Female	Lower	Lower	Same	Lower
Male	Lower	Lower	Lower	Lower
Type of referral				
Prenatal referral	Same	Higher	Same	Higher
Rare disease/other	Lower	Lower	Lower	Lower
Cancer	Same	Lower	Same	Lower
Remoteness				
Major city		Lower	Lower	Lower
Inner regional		Same	Same	Same
Outer regional	Lower	Lower	Same	Lower
Remote	Same	Same	Same	Same
Very remote	Same	Same	Same	Same

Ref=Aboriginal and Torres Strait Islander Rating=Much lower: RR LCI 95% >2 ,Lower: RR LCI 95% >1 and <=2 , Higher: UCI 95% RR<1

Overall, disparities between Aboriginal and Torres Strait Islander Australians and other Australians were smaller in Queensland than in other jurisdictions. This is due to higher levels of paediatric referrals among Aboriginal and Torres Strait Islander Australians compared to other Australians in Queensland. There were also fewer disparities in attendance in Queensland with the exception of attendance major cities. In WA and NT referrals and attendance were consistently lower for Aboriginal and Torres Australians compared to other Australians, however this did vary with the type of appointment. In all jurisdictions, attendance was lower for rare and other disease appointments perhaps reflecting a lower referrer and community understanding of these conditions.

Aboriginal and Torres Strait Islander Patient Experiences

Barriers to access and engagement were present at each stage of the patient journey. These included challenges in obtaining a **referral,** long **waiting periods**, limited **genetic literacy,** absence of Aboriginal and Torres Strait Islander **support services**, **communication** challenges and lack of adequate **psychosocial support** and **follow-up** after attendance. Aboriginal and Torres Strait Islander patients reported experiencing significant barriers gaining a referral to clinical genetic services, often due to **racism**. Participants' overall experiences of attending a genetic health service were varied, with positive perceptions tied closely to a diagnosis being achieved. The experience of (and expectation for) recognition of cultural identity and provision of culturally safe care was low among participants. Unaddressed concerns continued to cause significant distress in some people years after their appointment took place.

Clinical genetic service providers

Among practitioners, there was generally good understanding of logistical barriers that may prevent Aboriginal and Torres Strait Islander patients from attending the service or achieving acceptable levels of satisfaction with their attendance. There was also awareness of the importance of addressing systemic barriers like improving Aboriginal and Torres Strait Islander identification in referral and clinical genetic data. Overall, practitioners' responses suggested hesitancy and lack of confidence in integrating social and cultural considerations that might be relevant to Aboriginal and Torres Strait Islander patients into their practice. There was a high level of motivation to improve the cultural safety of services and support for developing policies and pathways to improve the benefit of clinical genetic services for Aboriginal and Torres Strait Islander patients.

RECOMMENDATIONS

The findings of the BIG Health Services project highlight the need for a systematic approach to increasing the benefits of clinical genetic services for Aboriginal and Torres Strait Islander people. This should encompass an overarching framework to guide system change, including improvements to data systems, referral processes, clinical genetic consultations and support and follow-up. The recommendations that emerged from the project are presented below with suggestions of practical action to aid the implementation of each recommendation.

Recommendation	Practical action to realise recommendation
Overarching Service Responsibilities	
Develop a national plan to guide coordinated approaches to increasing the benefit of clinical genetic services for Aboriginal and Torres Strait Islander patients and their families	In consultation with services and jurisdictions, a national plan should be developed for improving the benefit of clinical genetic services for Aboriginal and Torres Strait Islander patients and their families. This should include the establishment of Aboriginal governance structures to support the equitable development of clinical genetic services. Clinical genetics service staff and referring practitioners should receive cultural safety training with specific relevance to clinical genetics. Measures to improve data management are required for the purposes of referral management, continuity of care, patient and family follow-up, service monitoring. Resourcing is required for pre- and post-appointment support and Aboriginal genetic workforce development.
Enhance the capacity of clinical genetic services to provide an integrated service to Aboriginal and Torres Strait Islander patients and their families	There is a need to expand focus from the delivery of clinical genetic services to strengthening the support provided to patients and their families to ensure that optimal benefits from these services are delivered. Developing mechanisms for appropriate patient support during and after engagement with clinical genetic services would have direct benefits for patients in addition to workforce capacity building.
Improve equity in referrals	Equity in clinical genetic services overall would be improved by improving equity in referrals. Lower rates of referral for Aboriginal and Torres Strait Islander patients and their families is an issue in all jurisdictions, particularly for adults with cancer and for prenatal reasons. Provided that there is appropriate capacity, working with referring services to increase referral rates for Aboriginal and Torres Strait Islander Australians will be an important step in improving equity.
Support the development of networks to increase information sharing and professional learning	The BIG Health Services project provided the opportunity for clinical genetics providers from different jurisdictions to engage in knowledge sharing and ongoing learning with each other and with Aboriginal end-users. This was highly valued and provoked immediate, beneficial changes to practice. Support for sustainable, collaborative professional development will facilitate continued service improvement.
Build relationships with Aboriginal and Torres Strait Islander services/staff and the community-controlled health sector	The referral processes and experience of Aboriginal and Torres Strait Islander patients and their families would be improved by greater engagement with Aboriginal and Torres Strait Islander services and staff in all phases of the clinical process. This could be facilitated by stronger links between genetic services and hospital Aboriginal and Torres Strait Islander liaison services and developing stronger links with Aboriginal Community Controlled Health Services and other service providers for Aboriginal and Torres Strait Islander people.
Strengthen the evidence base in relation to increasing participation for what works in the Australian context	BIG health services project was the first project of its kind. The project highlighted both the need for development of an evidence-based approach to improving equity in clinical genetics services and the benefit this would provide to Aboriginal patients. Research is required to further examine and address the key drivers of lower rates of referral and attendance at clinical genetic services among Aboriginal people.

Improving pre-consultation support	
Develop referral templates that include Aboriginal and Torres Strait Islander identification, referral information and preferred communication	Developing a standardised referral template would enhance the ability of clinical genetics staff to triage, communicate and identify appropriate supports for Aboriginal and Torres Strait Islander patients. Optimally, this would be digitally-integrated or interoperable with clinical systems.
Facilitate patients' access to support services	Standard processes should be implemented to determine patients' eligibility for and access to financial assistance (e.g. PATS). This process should also ensure that Aboriginal and Torres Strait Islander patients are offered the services available to them at the hospital (translation services, Aboriginal and Torres Strait Islander Liaison Officer).
Improve patients' understanding of the reasons for attending clinical genetic services and the processes involved	Improving patients' understanding of the reasons for their referral to clinical genetic services and the potential benefits of this referral is important to improving cultural safety. Prior to their appointment, patients should be provided with clear information that is tailored to them. The information should outline the appointment process so that patients are adequately prepared and can assess their own need for support. Information should include: • Introduction of the practitioner, including their gender • Indication of whether a physical examination will be required • Indication of what results, if any, will be available on the day. Clarification that genetic testing is not always indicated and can be a slow process. • Clarify need for other family members to attend (i.e. could other children have samples collected at home or at the local doctor?) • Recommendation to bring a trusted adult support person as a 'second set of ears'
Improving support during consultations	
Ensure appointment setting are culturally appropriate and able to meet patients' needs	Appointment spaces should be welcoming and comfortably accommodate large families or people with specific needs (e.g. wheelchair access) to improve cultural safety and patient experience. Flexible appointment options (e.g. telehealth) would further enhance the appropriateness of appointments, for example by supporting the engagement of other family members or removing difficulties with travel.
Support genetic health literacy through ongoing communication	Genetic consultations can be stressful and confusing for patients. The challenge of effective communication in clinical genetics is exacerbated by the often complex nature of the information and the fact that outcomes have far reaching implications for families and communities, beyond those for the individual. Patients' genetic literacy should be supported throughout the consultation by establishing mutual understanding of the reason for the appointment and the possible outcomes. In communicating outcomes, it is important that patients are able to re-contact the service and access further consultations by phone or in person in order to clarify information received and resolve any concerns.

Improving support post-consultation	
Ensure reporting supports optimal benefit for Aboriginal and Torres Strait Islander patients	The clinical engagement between practitioners and patients is often short in the context of clinical genetics services. In order for clinical genetics services to deliver benefits to patients, it is critical that detailed, relevant information is provided to patients and referring providers in appropriate language. Reports should not only summarise the outcomes of the consultation but also direct patients/practitioners to appropriate resources for information and psychosocial and medical support. To achieve this, reports should include: • Contact details for clinical genetic services and encouragement to re-contact the service at any time if required. • Clear statement of the "next steps" discussed in the appointment (e.g. talk with family, another appointment to receive results, specific time frame for returning to service - noting that this is the patient's responsibility, the service will not contact them) • Reliable sources of medical information online, specific to the patient's condition or mode of testing • Places to access online (or other) peer or professional support (e.g. disease specific Facebook groups, rare disease organisations, cancer registries) & options for coordinated follow up that is culturally appropriate (e.g. Mappa, mapping care closer to home).
Include primary care providers in reporting	Referrals to clinical genetic services most often come from specialists. While reports are always provided to the referring doctor, in some cases this information is not also communicated to the patient's primary care provider. Ensuring that there is direct reporting to primary care will facilitate improved support to patients and continuity of their clinical care.

CONCLUSION

These recommendations are informed by substantial evidence compiled by the BIG Health Services Project and have been developed in consultation with key stakeholders and Aboriginal end-users. Our recommendations are intended to inform the development of a strategic approach to improving equity of access to clinical genetics services and ensure that that Aboriginal and Torres Strait Islander patients and their families receive the available benefits of engaging with clinical genetics services.

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