Australian Genomics Health Alliance

Clinical Genomics: Investigating perceptions of unmet need in Australia. An Initial Study

NATIONAL DIAGNOSTIC AND RESEARCH NETWORK PROGRAM, AUSTRALIAN GENOMICS

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Executive Summary

Genetic and genomic testing within the clinical realm is rapidly progressing, but anecdotal reports suggest that uptake across Australia is not uniform. This study posed this question to genetics experts in the field, to establish if there is unmet need and, if so, where attention should be focused in further investigating and understanding the need.

We investigated the views and experiences of unmet needs as perceived by Heads of Genetic Services across Australia. The purpose of this study was to understand the perspectives of these clinicians to address unmet need and prioritise a future research agenda to ensure subsequent policy is directed appropriately. Twenty interviews of genetic service leads, or their delegates, were conducted over a three week period in May 2019. Coding and content analysis were completed using the Framework approach (Ritchie and Lewis, 2003).

Unmet need has been defined as "*the difference between services judged necessary to deal appropriately with health problems and services actually received*" (Carr & Wolfe, 1976). In the context of genomics, we define *unmet need* as *patient groups in whom a clinical geneticist thinks a genomic test is clinically indicated and who want genomic testing but cannot access it*. Findings varied greatly between and within states/territories, with some areas reporting little to no unmet need while others had significant challenges. Almost all clinics had a gatekeeping process for ordering genomic testing, and ordering was subject to multidisciplinary team (MDT) discretion. Categories of unmet need indicated from this study included: populations who are unable to access genomic testing; populations not presenting to genetics services; budgetary constraints resulting in challenges in meeting needs; and workforce implications to ensure clinicians know when to refer, how to refer, and what genetics can offer is considered important. Themes as to where future research should be directed included: education of non-genetic clinicians and the general population; how to increase equity of genomic testing; finding ways to direct outreach services to less served populations; and how to address funding challenges within genomic testing.

Recommendations/Future Direction

Genetic professionals report varied and inconsistent access to genomic testing, with access varying by state, geographic region, with particular challenges for patients in regional areas, of Indigenous background and those with an intellectual disability. Accepting that genomic testing is increasingly being shown to be a powerful tool in improving healthcare in individuals with genetic conditions, this finding is of concern and merits further investigation. In particular, further study is recommended to:

- Understand the observed disparities between states
- Elucidate the reported disparities in genomic service provision for;
    - Indigenous populations
    - Specific patient populations such as individuals with intellectual disability
    - Rural and remote populations – **we recommend future study focuses on this area**

In addition, the patient population who do not know about the possibility of genomic testing represent an important unmet need, but one which we acknowledge is difficult to study or capture, and whose size may be reduced by ongoing efforts in education of non-genetics specialists and the general public.

Contents

Introduction

The role of genomic testing in clinical practice is growing. From 2011 to 2016/17 the volume of genomic tests completed in Australia had grown by 22%, mainly within the molecular sector. In addition, the scope of genomic testing and complexity of interpretation have increased (Royal College of Pathologists of Australia, 2018). There is an increased demand for genomic testing, but anecdotal reports suggest that uptake across different spheres of clinical practice is not uniform.

Despite anecdotal reports of unmet need, there is a lack of research in this area. Other research groups are attempting to understand this question from different perspectives (e.g. the patient perspective). We are aiming to complement this research adding the perspective from genetic professionals, seeking to explore their perceived areas of unmet need and their views on how the future research agenda should be prioritised.

Potential causes of unmet need are wide ranging and vary within and between countries. Previously identified causes of unmet need in healthcare include social and service-based drivers with "*i) population-driven unmet need, individuals are not entering or accessing the healthcare system"* and… *"ii) service-driven unmet need, services are not reaching the target population, or they drop out of the system*" (Academy of Medical Sciences, 2017 p.8).

Considering genomics specifically, evidence from the US points to the lack of access to genomic testing by disadvantaged groups (Smith et al 2016) with Amendola et al (2018) reporting on the work of the Clinical Sequencing Evidence-Generating Research (CSER) consortium as it investigates '*the effectiveness of integrating genomic (exome or genome) sequencing into the clinical care of diverse and medically underserved individuals in a variety of healthcare settings and disease states*'. The need for new models of service delivery are identified to overcome some challenges (for example Stoll et al 2018 consider the use of genetic counsellors to overcome geographical access barriers).

Responding to concern that precision medicine could focus services on those already accessing healthcare, reinforcing patterns of former health service provision, Gray et al (2017) highlights the need to learn from healthcare payers, clinicians and patients to maximise the potential of genomics in clinical practice. This study is a first step in identifying and exploring where the unmet need for genomic testing lies in Australia and where the future research agenda needs to be concentrated to ensure future policy is directed appropriately. Related ongoing studies including an audit of current models of care in cardiac clinics and the focus group survey of patients with neurodevelopmental disability will also be important perspectives to consider from non-geneticist healthcare providers.

Policy Background

Reviewed below are Australian genomic policies, activity around unmet need, and demographics essential to the discussion surrounding unmet need.

Unmet Need in Australia

To ensure equity, universal access, and sustainability within genomic testing, collaboration and coordination across Australia is necessary. The Australian Government has recognised the need for research and collective integration of genomics into healthcare, funding the Australian Genomics Health Alliance (Australian Genomics) through the National Health and Medical Research Council (NHMRC) and developing a *National Health Genomics Policy Framework* (Commonwealth of Australia, 2017) and *Implementation Plan* (Commonwealth of Australia, 2018). However, for genomic testing to be accessible and encompass all populations, systems need to be set in place to ensure all goals of the National Health Genomics Policy Framework are met. Key priorities for governing institutions are education campaigns, decision aids, programs to implement genomics into healthcare systems, and empowering less resourced groups (Burns et al, 2019).

Unmet needs encompass those who are unable to access genomic testing. Burns et al (2019) discuss national standards for genomic testing and encourage implementation to ensure equitable and safe access to genomic testing throughout Australia. Outlined are five critical priority areas necessary for sustainable implementation of genomics within the Australian health care system: data, workforce, finances, person centred care, and services. Resource allocation to all five of the priority areas are essential for an effective implementation of genomic testing. This study encourages development of national standards for genomic testing and includes recommendations from a governmental perspective.

Geography presents a particular challenge within Australia due to its vast geographical distance and access to cities. Those living within rural and remote areas lack access to genetic services. Within these regions, telehealth has been utilised to help minimise disparities if genomic testing is offered. Additionally, the availability in some areas of access to genomic testing on a user pay basis means that those who are unable to pay for testing have an inequity of access to genomic testing. There are currently different tests available publicly and privately (e.g. Non-Invasive Prenatal Testing (NIPT), reproductive carrier screening, and Preimplantation Genetic Diagnosis). Private sector genomic testing and

direct to consumer genomic testing have a skewed population group towards higher genomic literacy, education and means to access these venues (Burns et al, 2019).

Indigenous populations in relation to genomic testing are also focused on within the literature. Inequalities for Aboriginal and Torres Strait Islanders occur for a number of reasons including a lack of genomic representation data to make diagnosis, referral bias, and living in remote areas decreasing access to genetics services (Baynam et al, 2017). Other cultural barriers include a distrust of research (Thomas, Bainbridge & Tsey, 2014). Additionally, genetic and genomic research is less effective for this population due to lack of relevant population variant data (Garrison et al, 2019). For equitable genomic health, there is an urgent need to improve models of care for Indigenous populations. Suggestions to bridge this gap include community involvement, the use of aboriginal healthcare workers, development and implementation of Indigenous guidelines, and the reduction of referral bias (lack of referrals to genetics or specialists from general practitioners) to introduce an inclusive approach of Aboriginal and Torres Strait Islander integration into genetics (Baynam et al, 2017; Garrison et al, 2019).

Policy

The overarching genomics health policy that governs Australia is the National Health Genomics Policy Framework 2018-2021 (Commonwealth of Australia, 2017). This policy framework outlines five strategic priorities as person-centred approach, workforce, financing, services, and data. This framework was developed to address a need to allow for a cohesive approach to genomic medicine across Australia (Burns et al, 2019). The implementation plan is currently under development with Australian Health Ministers' Advisory Council (AHMAC) oversight through the Project Reference Group on Health Genomics.

Australian Genomics is a large and diverse research network of more than 80 organisations, building the evidence for a whole-of-system approach to integrating genomics into healthcare. Its aim is to evaluate and accelerate the application of genomic testing in healthcare and to do this with a collaborative approach based on federation principles. Its partners include research institutes, hospitals, sequencing laboratories, universities and community groups across Australia and internationally.

The Australian Genomics research model encompasses clinical projects in rare diseases, cancers and reproductive carrier screening, and interdependent programs for advancing the diagnostic, health informatics, regulatory, ethical, policy, and workforce infrastructure

necessary for the integration of genomics into the Australian health system (Stark et al, 2019).

While national policies provide a framework, public hospitals and state genetics services are funded by the individual states and territories. Ultimately, the state is responsible for providing healthcare to its inhabitants. In addition to the National policy, most individual states are now developing state-specific genomics policies, outlined here:

QLD: Queensland has a State-wide Genetic Health Queensland Service Plan 2017-2022 (State of Queensland, 2017) and the Queensland Genomics Health Alliance (QGHA) is working to develop a plan bringing clinical, pathology and policy work together.

NSW: The NSW Health Genomics Strategy (2017) instituted five sub-committees to assist in implementing a shared vision for NSW health over a three phase, seven-year process. Six recommendations of the policy include establishing a governance committee, enhancing clinical need, utility and validity, developing new delivery models, integrating genomics into healthcare, identifying workforce needs, and working with the general public to gain confidence in genomic medicine.

VIC: Victoria aims to have better health, better access and better care by 2040. Four priorities outlined in the Genetic and Genomic Healthcare for Victoria 2021 document (State of Victoria Department of Health and Human Services, 2017) are to develop and implement a state-wide genetics and genomics plan, establish a genomic health clinical network, undertake community consultations, and reduce superbugs. These four priorities are supported by additional work to strengthen the healthcare system, build trust, raise awareness, and grow knowledge.

SA: The South Australian Clinical Genomics Plan 2022 (Government of South Australia, 2019) outlines a need to implement and support a clinical genomics program. The National Health Genomics Policy Framework 2018-2021 was amended to suit South Australia's individual needs. The SA advisory group will collaborate with all stakeholders to implement a successful genomics program into SA.

WA: The WA Health Department implemented a State Strategic Framework for Rare Diseases 2015-18 (Dept of Health, Western Australia, 2015) a further Rare Disease Strategy and WA Genomic Health Strategy are currently under development.

These state policies illustrate the divergence of state-based priorities in the genomics field.

Demographics

The cultural and regional distribution of populations vary greatly between the states and territories within Australia. Indigenous Australians are more commonly located in less populated areas, living in more remote areas of Australia. Three quarters of non-Indigenous people in Australia live in major cities, in contrast to the one third of Indigenous people who live in major cities. Inner regional, outer regional, remote and very remote Australia have higher percentages of Indigenous people than non-Indigenous inhabitants (Australian Institute for Health and Welfare, 2017) (see figure 1). This suggests that, geographically, a greater proportion of Aboriginal and Torres Strait Islander Australians have less access to all health resources than non-Indigenous people in Australia.

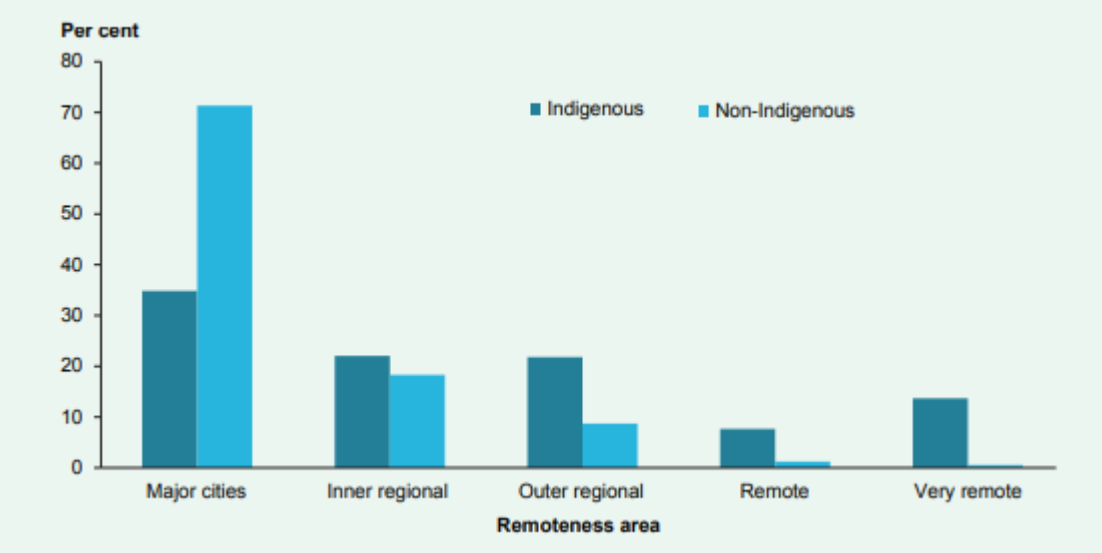

Figure 1: Population distribution by Indigneous/non Indigenous and remoteness area as of June 2011 (data source: *ABS (2013). Estimates of Aboriginal and Torres Strait Islander Australians, June 2011. ABS cat. no. 3238.0.55.001. Canberra: ABS*) AIHW (2017) [1] Reproduced under Creative Commons license.

Each state has varying total populations and population distributions. NSW, VIC and QLD are the most populous states. In WA, VIC, SA, and NSW, approximately 75% of the population lives in major cities. In QLD, there is a lower percentage (62%) living in major cities. In Tasmania 66% of inhabitants live in an inner regional area. Based on ABS data, of the inhabitants of NT, 56% live in outer regional areas, 21% in remote areas, and 23% in

[1] Major cities are "areas where geographic distance imposes minimal restriction upon accessibility to the widest range of goods, services and opportunities for social interaction". Inner regional areas are where geographic distance imposes some restriction, outer regional has moderate restriction and remote/very remote have high and highest levels of restriction imposed based upon geographical location (Australian Bureau of Statistics, 2004).

very remote areas where their geographic distance imposes a moderate to severe restriction upon accessibility to the widest range of goods, services and opportunities for social interaction. In contrast, ACT inhabitants all live within a major city (Australian Bureau of Statistics, 2016a).

Levels of socio-economic advantage also vary between states. Typically, areas within central Australia are less advantaged than coastal areas. Influences on this advantage scale include location, range of opportunities, goods and services to adequately fulfil an individual's needs. The highest proportion of those living in advantaged areas were in ACT, and lowest proportion in Tasmania (Australian Bureau of Statistics, 2016b) (see figure 2).

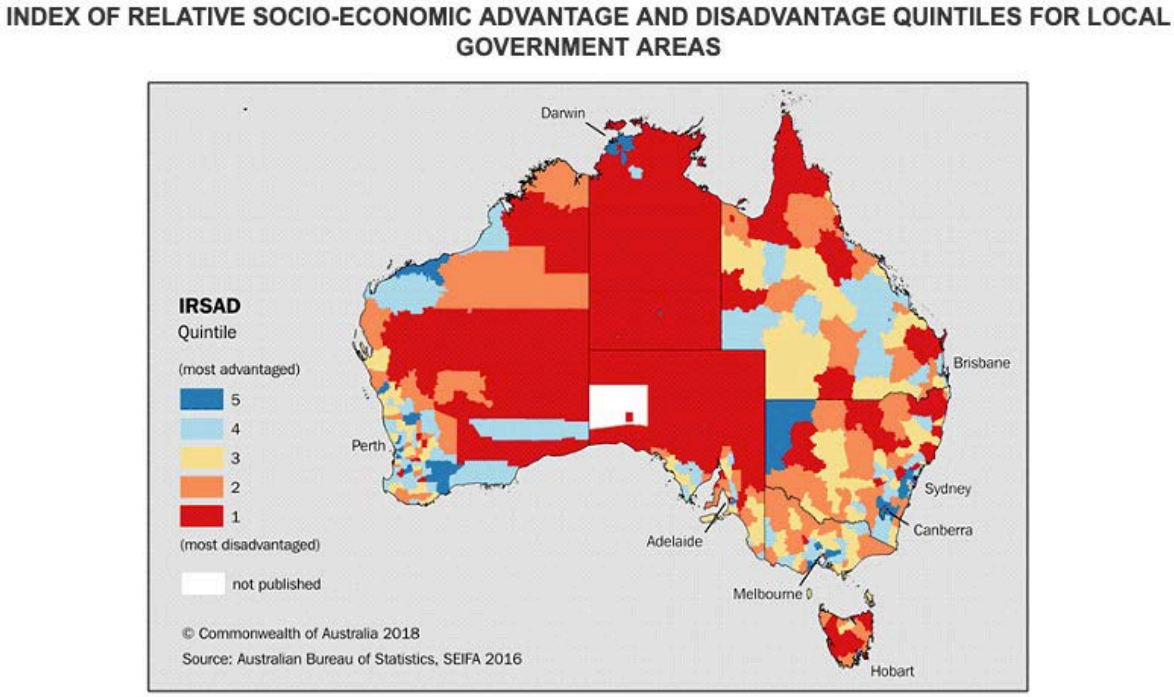

Figure 2: Disadvantage quintiles of Australia. Quintiles are calculated using Socio-Economic Indexes for Areas (SEIFA), and the Index of Relative Socio-Economic Advantage and Disadvantage (IRSAD). The SEIFA IRSAD uses variables which include income, education, employment, occupation and housing characteristics (Australian Bureau of Statistics, 2016b). Reproduced under Creative Commons license. Retrieved from ABS and available here.

Genetic Services by state

There is currently no national coordinated approach for genetics services across Australia: each is funded by State/Territory Departments of Health. These services tend to be centralised, servicing metropolitan, regional and rural areas with outreach clinics (Barlow-Stewart et al, 2007). Western Australia, Tasmania, and Queensland have coordinated services throughout each state and in South Australia genetic services are provided separately for children and adults. Both Victoria and New South Wales each have multiple

genetics services (Victoria 4, NSW 7). WA and Queensland Genetic Services serve the largest geographic areas, 32.9 and 22.5% of Australia respectively. Interestingly, in this initial study, the perception of unmet need correlated better with geographic limitations rather than the presence of a unified state service. This could be explored further.

Familial cancer services are typically distinct (Barlow-Stewart et al, 2007) in most states (though not in WA) and were not covered by this study. Additionally, there are both private and public services throughout Australia and this study focused on public sector provision.

Methods

Rapid structured interviews were conducted with 20 key stakeholders for genetics services across Australia. Questions explored perceptions of unmet need in the area in which the clinical genetic service operates. Stakeholders were Heads of Department for each service (approximately 15 services) or their delegate and followed the inclusion criteria below. Contact details for participants were gathered from publicly available websites and interviewees were invited to identify other key stakeholders.

Inclusion criteria:

Participants must be:

- Senior genetic clinicians
- Maximum of two representatives per service

Data collection

Interviews: Due to the rapidly changing nature of genomics, interviews spanned the course of three weeks (majority of interviews from 14/5/19 - 30/05/19; one interview occurring the first week of July) to gain contemporaneous perspectives. Participants were interviewed via telephone. The interviews lasted approximately 15-20 minutes.

Consent and recording: Participant information was distributed prior to interviews and verbal consent was sought at the outset of the interview. The interviews were recorded using a digital recorder, then transcribed. All participants were assigned a code and interviews were solely identified via codes.

Data Analysis

Iterative data analysis was undertaken continuously throughout the interview process looking to identify common themes in the data. Interviews were transcribed and then coded in Excel using the Framework approach (Ritchie and Lewis, 2003).

Findings

There were varying degrees of unmet need reported throughout Australia. While processes were similar around test ordering and availability of types of genomic tests, perceptions of unmet need had a more unique profile. Suggestions around how to direct further research typically revolved around education of clinicians and the general population.

The initial questions were asked to understand what genomic tests are available how they are ordered within each state/territory. There are similar processes amongst all states/territories regarding which tests are available and who is able to order these tests as perceived by clinical genetics services. All states had clinical geneticists and specialists involved with the ordering process. Panel and exome testing proved to be the most common tests ordered, with limited genome testing utilised clinically. While who could order genomic tests varied, it typically followed a gatekeeping model led by a Clinical Geneticist and/or Multi-disciplinary Team. Typically, specialists need clinical geneticist approval to order genomic testing, with the exception of specific personnel and tests which were 'rubber stamped'. Multi-disciplinary teams were involved in the ordering process when more difficult or expensive decisions were made.

The remaining questions aimed to establish if the clinician felt there was unmet need and understand where it lay. There was agreement that there is an inequity of access to genomic testing. The four categories discussed most often were; i) populations who are unable to access genomic testing, ii) populations not presenting to genetics services, iii) budgetary constraints, and iv) workforce implications.

i) Groups not presenting to genetic services included specific patient populations such as people with intellectual disability, Indigenous populations, rural populations, and those who simply are not being not referred (e.g. due to lack of caregiver or clinician knowledge).

ii) A lack of family history of disease, individuals of all ages with intellectual disability, and 'non-urgent' patients were noted as barriers for access to testing. Wait times for genetics clinics are long and these patients are not prioritised.

iii) A common theme was budgetary constraints. Genetic service leads expressed that there are insufficient resources to accommodate all facets of genomic testing including laboratory staff, interpretation costs, and clinician support. The funding model for genomic medicine across Australia is not sustainable for increased demands on the system.

iv) Workforce constraints were noted as relevant to unmet need. Education of clinicians to ensure they know when to refer, how to refer, and what genetics can offer is considered

important. Additionally, appropriate personnel are essential to ensure genetics services are provided equitably and to the highest standard of care.

There were two main schools of thought within the states and territories. More populated states reported on funding and the lack of, or non-sustainable nature of, the funding systems currently in place. Less populated states focused on the lack of referrals and not seeing representative populations in genetics clinics. This did not correlate with presence / lack of a unified state wide clinical or laboratory service. While these were the main themes, almost all areas mentioned funding and referral challenges. There were exceptions to this finding, one interviewee focused heavily on both funding and the absence of populations within genetics and another did not perceive significant challenges in either category.

The interview concluded with the question "What do you see as the highest priority for future research into unmet need for genomic testing?". Responses ranged from suggesting further research into clinical utility for particular conditions and education around integrating genomics into fields that were not previously thought to have a genetic origin are especially important. The most common response was education of non-genetic clinicians and how to empower them to help compliment the genetics department or service. Additionally, equity of access for those with geographical barriers and research into how services may provide outreach to these individuals was suggested. Reflecting the findings in previous questions several participants felt future work should include understanding the unseen unmet need ("*patients we do not know about*") – though the mechanism to achieve this was not clear – and, how to efficiently provide outreach to under-served communities. The disparity of genomic service provision by state was also highlighted with a preference for increasing consistency throughout states and Australia as a whole.

Limitations

These findings should be considered within the limitations of this study. First, only the perspective of heads of genetics services (or their delegate) was captured, which was predominantly Clinical Geneticists (14 out of 20). Many interviewees suggested we talk to specialists, lab personnel, and others involved within the genomic testing process to gain a more comprehensive perspective. These could be considerations for a future study. Second, the roles of participants varied from clinical geneticist, genetic counsellor, specialist, etc. Thus, attitudes perceptions may vary depending on the clinical role of the individual. Finally, those interviewed were only within the public sector, leaving the private sector as a gap in this study.

Conclusion

Translating genomic medicine to benefit every population within Australia will require substantial stakeholder engagement and recognition of the geographic, political and cultural challenges this goal presents. The policies and guidelines in place act as directions, however ongoing collaboration between community, governmental, and healthcare stakeholders are essential to the ongoing support of equitable implementation of genomics into clinical care.

Work in this field continues to progress. Future studies should ensure ongoing research projects within existing Australian Genomics work (such as, Implementation Science and Health Economics work around Discrete Choice Experiments for different patient populations, or the exploration of current models of care in specialist clinics (cardiac flagship) are taken into consideration where appropriate. Provision of sufficient access to relevant laboratory services is also relevant and may be addressed by ongoing national genetic laboratory services reviews commissioned by the Department of Health (RCPA 2018) as part of the National Health Genomics Policy Framework (Commonwealth of Australia, 2017).

Further research is essential to determine the extent of unmet need and mechanisms to address unmet need within the clinical health system. Below we outline further directions research may take to inform further policy development.

Recommendations and Future Direction

This study to investigate perceptions of clinical genomic unmet need in Australia confirm previous anecdotal reports and lays the baseline for future research. Ongoing efforts need to be focused on gaining an understanding of the observed disparities between states to elucidate the reported differences in genomic service provision for:

- *Indigenous populations*: much work is already underway exploring the needs of indigenous people. See for example the work of POCHE Centre for Indigenous Health and the National Centre for Indigenous Genomics; the work (est 2011) of the WA Health Department, the Aboriginal Health Council of WA and WA Aboriginal Communities, ACTG – Aboriginal Community-led Translation of Genomics; and the work of Genetic Health Queensland and Queensland Aboriginal Controlled Health Services; and the Better Indigenous Genetics Project for improving genetic health care service delivery in Australia. This list is not exhaustive. We recommend following the activities of these institutions to ensure this unmet need is assessed.
- *The unknown population* who do not know about the potential for genomic testing: this group present a feasibility challenge in developing future study. While this should not discourage future research there is a need for some consideration and careful planning in pursuing this avenue of exploration.
- *Specific patient populations* such as individuals with intellectual disability: this group may be impacted upon to some degree by the expected Medicare item number for funding genomic sequencing in children with intellectual disability. Further studies into adults with intellectual disability should be considered.
- *Rural and remote populations*: the unmet need of rural and remote populations presents as an important area for investigation. Around 7 million or 29% of the Australian population live in rural or remote communities (AIHW, 2018) and we would strongly recommend further analysis of the unmet need in these areas. Study options would include:
    - A post-code and provider number audit of genomic tests in NATA-accredited Australian genomic laboratories, over a specified time period, and compare this with population distribution data to investigate the proportion of metropolitan and regional patients receiving genomic tests compared with what would be expected in terms of demographics. The inclusion of provider numbers may enable analysis of whether regional patients had to travel to metropolitan centres to access their genomic tests, or whether the test was provided in a regional centre. This study would of course require co-operation of Australian laboratories, and may require some administrative support for

data collection. It may be suitable for a student research project and could generate valuable data to inform genomics practice. **This would be our recommendation for future study.**

- A study documenting genetic clinics in metropolitan versus regional centres compared with per head population residing there.
- Survey/audit of rural physicians/paediatricians regarding their perception of genomic test demand and supply.
- Survey of people living in rural/regional areas who would like a genomic test, compared with metropolitan areas. Potential to use social media to deliver this.

References

Academy of Medical Sciences (2017) Unmet need in healthcare Summary of a roundtable held at the Academy of Medical Sciences on 31 July 2017, held with support from the British Academy and NHS England. Available here.

Amendola LM, Berg JS, Horowitz CR, Angelo F, Bensen JT, Biesecker BB, et al. The Clinical Sequencing Evidence-Generating Research Consortium: Integrating Genomic Sequencing in Diverse and Medically Underserved Populations. Am J Hum Genet [Internet]. 2018;103(3):319–27. Available here.

Australian Bureau of Statistics. (2016a) Estimates of Aboriginal and Torres Strait Islander Australians 3238.0.55.001. Australian Bureau of Statistics, Canberra. Available here.

Australian Bureau of Statistics. (2016b) Socio-economic advantage and disadvantage. 3238.0.55.001 Australian Bureau of Statistics, Canberra. Available at here.

Australian Institute of Health and Welfare. (2004). Rural, regional and remote health: a guide to remoteness classifications. AIHW cat. no. PHE 53. Canberra: AIHW. Retrieved from and available here.

Australian Institute for Health and Welfare (2017) Australia's welfare. Community factors and Indigenous wellbeing. Available here.

Australian Institute for Health and Welfare (2018) Rural and remote populations. Health of population groups. Available here.

Barlow-Stewart K, Emery J, Metcalfe S, Bylstra Y, Forrest L, Morphett L, Reid G, Ross D. (2007). Genetics in Family Medicine: The Australian Handbook for General Practitioners. Canberra, Biotechnology Australia. Available here.

Baynam, G., Molster, C., Bauskis, A., Kowal, E., Savarirayan, R., Kelaher, M., … Dawkins, H. J. S. (2017). Indigenous Genetics and Rare Diseases: Harmony, Diversity and Equity. Advances In Experimental Medicine And Biology, 1031, 511–520. Available here.

Burns BL, Bilkey GA, Coles EP, Bowman FL, Beilby JP, Pachter NS, Baynam G, Dawkins HJS, Weeramanthri TS and Nowak KJ (2019) Healthcare System Priorities for Successful Integration of Genomics: An Australian Focus. Front. Public Health 7:41. Available here.

Carr W., Wolfe S. (1976) Unmet needs as sociomedical indicator. Int. J. Health Serv.6:417–430. doi: 10.2190/MCG0-UH8D-0AG8-VFNU. Available here.

Commonwealth of Australia. (2018) National Health Genomics Implementation Plan. Available here.

Commonwealth of Australia, Department of Health (2017) National Health Genomics Policy Framework 2018– 2021. Canberra, ACT Available here.

Department of Health, Western Australia. WA Rare Diseases Strategic Framework 2015–2018 (2015) Perth: Office of Population Health Genomics, Public Health Division. Available here.

Garrison, N. A., Hudson, M., Ballantyne, L. L., Garba, I., Martinez, A., Taualii, M., … Carroll Rainie, S. (2019). Genomic Research Through an Indigenous Lens: Understanding the Expectations. Annual Review Of Genomics And Human Genetics. Available here.

Government of South Australia, Department of Health and Wellbeing. (2019). South Australian Health Genomics Policy 2022. Available here.

Gray M, Lagerberg T, Dombrádi V. Equity and Value in 'Precision Medicine.' New Bioeth [Internet]. 2017;23(1):87–94. Available here.

NSW Ministry of Health. (2017). NSW Health Genomics Strategy. Online. Available here.

Ritchie, J & Lewis, J. (2003). *Qualitative Research Practice: A guide for Social Science Students and Researchers.* London: Sage Publications.

Royal College of Pathologists of Australia (RCPA) (2018). Australian Health Genetics/Genomics Survey 2017 – Report Key Findings to: Department of Health. The Department of Health. Retrieved from and available here.

Smith CE, Fullerton SM, Dookeran KA, Hampel H, Tin A, Maruthur NM, et al. Using genetic technologies to reduce, rather than widen, health disparities. Health Aff. 2016;35(8):1367–1373. Available here.

South Australian Clinical Genetics Services. Available here.

Stark, Z., Boughtwood, T., Phillips, P., Christodoulou, J., Hansen, D.P., Braithwaite, J., et al. (2019). Australian Genomics: A Federated Model for Integrating Genomics into Healthcare. Am J Hum Genet, 105(1), 7-14. Available here.

State of Queensland, (2017) State-wide Genetic Health Queensland Service Plan 2017-2022 Available here.

State of Victoria Department of Health and Human Services. (2017). Genetic and Genomic Healthcare for Victoria 2021. Available here.

Stoll K, Kubendran S, Cohen SA. The past, present and future of service delivery in genetic counseling: Keeping up in the era of precision medicine. Am J Med Genet Part C Semin Med Genet. 2018;178(1):24–37. Available here.

Thomas D, Bainbridge R, Tsey K (2014) Changing discourses in aboriginal and Torres Strait Islander health research, 1914–2014. Med J Aust 201(1):S1–S4. Available here.

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