State and Territory Health Systems Consistency Project

Final Report

July 2024

Acknowledgement of Country

In the spirit of reconciliation Australian Genomics acknowledges the Traditional Custodians of country throughout Australia and their connections to land, sea, and community.

We pay our respect to their elders past and present and extend that respect to all Aboriginal and Torres Strait Islander peoples today.

Artwork by Yorta Yorta artist, Alkina Edwards, for Australian Genomics.

State and Territory Health Systems Consistency Project

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#legacy – June 2021 – January 2022

The document and associated appendices were compiled by project coordinators MJ, MQ, and AS, and subsequently reviewed and approved by project leads.

Executive Summary

Project Overview

Currently, each Australian jurisdiction operates according to their respective legislation, policies, frameworks, funding structures, and geographical constraints when providing and implementing genomic health care. This project therefore strived to determine what jurisdictional (in)consistencies exist across health practices in Australia, and how to facilitate equitable access to appropriate publicly funded clinical and diagnostic genomic services for all Australians.

The overarching aims of this project were firstly to ensure mechanisms were in place for regular exchange and consultation between jurisdictional representatives and their respective clinical genomics services. Secondly, we aimed to promote sharing of experiences across jurisdictions regarding translation of genomics and provide a national framework that is acceptable to each jurisdiction.

Methods

Mapping of state and national genomic plans:

Available jurisdictional genomic plans/strategies for NSW (2017 and Implementation 2021-2025), WA (2022-2032), SA (2022), VIC (2021), QLD (2017-2022) and the National Health Genomics Policy Framework 2018-2021 were reviewed, and the content mapped against key subject areas, such as services; digital infrastructure / IT; education of genomic workforce; Indigenous genomics / regional priorities, and integration of research into standard practice. Side-by-side comparison enabled identification of areas in the plans that were (in)consistent and revealed several gaps in proposed and prioritised strategic delivery of genomic healthcare.

Key Findings

While highly concordant, the high-level review of available genomic plans identified 21 gaps (not addressed by any plan) and/or inconsistencies (addressed by some plans or specified to a varying extent). Of these, 13 were mapped to existing projects already underway by Australian Genomics or other national initiatives (e.g. Australian Genomics priority projects, or Genomics Health Futures Mission (GHFM) funded research grants). The remaining 8 of these were presented to the project leads, of which 3 were prioritised for detailed review and consultation process towards national service and professional guideline development.

The prioritised gaps/inconsistencies due to national need and working group expertise were:

- Professional standards for bioinformaticians (gap)
- Standard genetic referral criteria (inconsistency)
- Testing/referral processes for non-genetic specialists (inconsistency)

Impacts

Identifying and addressing gaps and inconsistencies between jurisdictional strategic priorities and strategies provides an opportunity to improve the equity of access to genomic healthcare across Australia. While states and territories must operate in accordance with their respective legislative and policy requirements, connecting subject matter experts and sharing experiences enhances the application and implementation of genomics, facilitates collaboration, and avoids unnecessary duplication of work.

Recommendations

Feedback genomic plan evaluation to the appropriate government departments to help inform the review and update of the National Health Genomics Policy Framework and Implementation Plan.

Establish pathways to allow open communication and the transfer of knowledge between clinical and diagnostic services to enable ongoing improvement in genomic healthcare delivery within Australia.

In the 2024-2025 Australian Genomics grant opportunity, continue to progress work to achieve greater equity and quality of genetic health service delivery.

Conclusion

In a federated health system model such as in Australia, there is an ongoing need for evaluation of service delivery across geographical areas, to prevent any potential 'postcode lottery'. Further communication is necessary in both the clinical and diagnostic space to ensure consistency in approach. The refining of the National Health Genomics Policy Framework and Implementation Plan offers an opportunity to continue to address the gaps and inconsistencies identified in this project.

It is important to note, that while genomic plans are a useful tool for identifying potential areas of inconsistency, not all jurisdictional plans were developed during the same time period, and not all strategies or action plans specified will progress to completion.

Plain Language Summary

Currently, each Australian state and territory adopts different approaches when implementing genomics into health care. This project aimed to determine what differences existed across Australian genomic health practices, and how to improve equitable access to appropriate clinical and diagnostic genomic services. Through review of jurisdictional genomic strategic plans, we found that there were gaps and inconsistencies in key areas relating to genetic service delivery. We progressed further research in three areas to address these differences, including a review of what referrals are accepted by public genetics services in each state and territory. We also investigated how standard qualifications for clinical bioinformaticians could be assessed. Finally, we produced decision support tools that will aid non-genetic specialists to collect appropriate clinical information and order genetic and genomic tests in the most consistent and efficient manner. In each case, results will be distributed back to respective decision makers and those that are able to progress the implementation of findings.

Background

The State and Territory Health Systems Consistency project is a national collaborative effort that brings together state and territory clinical, diagnostic, and policy service leads, to establish consistent inter-state health practices, facilitating a standard genomic referral and testing framework for all Australians.

Currently, clinical and laboratory workflows and processes are independently governed by local legislation, frameworks and policies. However, if shared, established evidence-based practices at each local jurisdictional health service would allow future streamlining of prospective policy and framework changes.

Therefore, mapping and comparison of the current practices that exist within each jurisdiction, and frequent consultation between genetic service and laboratory leads, will improve consistency and encourage the establishment of harmonious guidelines.

This project aims to implement and maintain a nationwide minimum set of standards in select issues relating to clinical and diagnostics genetic service delivery, ensuring that all Australians can access equitable, quality standard of care testing in the diagnostic genomic space.

Introduction

The Australian health care system is a complex network of public and private providers and settings (Australian Institute of Health and Welfare 2016). Public healthcare is overseen by three levels of government (federal, state/territory, and local). Public hospitals, from which public genomic services are provided, are managed by state/territory governments and funding is shared between the Australian and state/territory governments. Therefore, genomics services in each jurisdiction are guided by nuanced approaches creating challenges in delivering nationally consistent genomic healthcare due to a variety of factors; including different funding models, geographical challenges, and legislative and policy differences.

There are also many private healthcare providers offering genomic services. These are funded through a combination of out-of-pocket fees and (where eligible) private health insurance or the Medicare Benefits Scheme (MBS), which is funded by the Australian Government. However, there are only a limited number of diagnostic genetic/genomic tests or services that are subsidised by the MBS (Stark et al, 2019) or health insurance. While private providers of genomics tend to have shorter wait times, the out-of-pocket costs make such services inaccessible for many consumers.

The mix of public and private healthcare within the federated system of healthcare in Australia, which is both complex and fragmented, likely amplifies some of the differences in state and territory genomic service provision.

There are both national (Australian Genomics, refer to Stark et al., 2023) and state led collaborative efforts (for example Melbourne Genomics Health Alliance (refer to Genomics in Victoria: What's changed in 10 years: 2013-2023) and Queensland Genomics Health Alliance (refer to Vidgen et al., 2021) that inform evidence-based implementation of genomics, including diagnostic rates of genetic testing, health economic analysis of cost effectiveness, investigations into equity of access to genetic testing and translation from clinical research. However, integration of genomics into healthcare presents unique challenges as the technology around diagnostic testing and techniques is constantly evolving (Burns et al., 2019) and current systems are not designed to cope with such rapidly evolving technologies.

There is a need for a systematic investigation of the gaps and inconsistencies that exist for genetic and genomic service delivery that are contributing to inequity of access Australia wide. This project therefore aimed to undertake a nationwide review of existing genomic healthcare priorities and implementation strategies and identify and facilitate areas for improvement. Available jurisdictional genomic plans and strategies were reviewed alongside the national framework as a first step to identify gaps and inconsistencies. Following working group review, three main areas were prioritised for further progression in an attempt to assist in national equity of access and service delivery.

Aims

Overarching aims:

- Identify and evaluate key genomic healthcare priorities and barriers to equitable translation.
- Ensure mechanisms are in place to allow transfer of knowledge and acquired learnings through regular exchange and consultation between jurisdictional representatives and their genomic healthcare services.
- To develop and promote a nationwide minimum set of recommendations in targeted areas to inform policy development to facilitate equitable and quality standard of care for clinical and diagnostic genomic health care.

Objectives

- Perform a systematic review of available jurisdictional and national genomic plans and collate a list of gaps and inconsistencies in strategic priorities.
- Map against existing projects being conducted through other mechanisms (e.g. Australian Genomics priority projects; GHFM funded projects; other national initiatives)
- Undertake a desktop review and associated consultation with relevant parties to address a subset of chosen gaps and inconsistencies.
- Provide summarised information to relevant stakeholders and partners.

Intent

The genomic healthcare landscape in Australia is continuously evolving, and despite a high level of concordance in the scope of clinical genetics and diagnostic services, these are governed by each jurisdiction. To assist in equity of access and consistent service delivery, the State and Territory Health

Systems Consistency project undertook landscape analyses across 3 areas; jurisdictional genomic plan review, professional standards for bioinformaticians, and standard genetic referral criteria. Primary information sources such as documents, websites, and job advertisements reviewed for each of these sub-projects were assessed at certain point(s) in time, with the intent to identify gaps/inconsistencies, barriers/enablers and minimum criteria required to deliver consistent and equitable genomic healthcare. The purpose of the information gathered is to assist in local, jurisdictional, and national planning, rather than identifying or highlighting service or area specific differences. Similarly, what is documented is a balanced perspective based on evidence at a certain time point, and certain aspects of these assessments may require more granularity, time, or funding to be realised.

Inputs

Project Leads: Clara Gaff, Cliff Meldrum, Julie McGaughran, Kristen Nowak, Hamish Scott.

Project Coordinators: Matilda Jackson, Michael Quinn, Ami Stott, Julia Dobbins (maternity leave cover for Matilda Jackson between June 2021 – January 2022).

Subproject working groups:
Cardiac decision support tool working group: Chirag Patel (QLD, chair); Belinda Gray (NSW); Jodie Ingles (NSW); Ivan Macciocca (VIC); Julie McGaughran (QLD); Jason Davis (QLD); Ami Stott (WA, coordinator).

Endocrine genetics decision support tool working group: Chirag Patel (chair); Chris Richmond (QLD); Sunita De Sousa (SA); Lisa Hayes (QLD); Nicolas Boyer (QLD); Michael Quinn (QLD, coordinator).

External Collaborators:

- Clinical genetic service representatives
- Clinical diagnostic laboratory representatives
- Australian Bioinformatic and Computational Biology Society (ABACBS)
- Commonwealth Department of Health and Aged Care representatives

Engagement with First Nations Communities:

- Consultation on ‘Aboriginal and Torres Strait Islander Genomics Health Guiding Principles’

Stakeholders: Clinical genetic service leads, clinical diagnostic laboratories and staff, Human Genetics Society of Australasia (HGSA), Royal College of Pathologists of Australasia (RCPA)

Other Resources: Decision tool design has been led by Liana Cross (Digital Communications Manager, Australian Genomics) in conjunction with Chirag Patel (Chair, decision support tool working groups), decision support tool coordinators and Queensland Metro North human factors consultants.

Milestones and Timeline

The first meeting with leads and coordinators was held on 2 September 2021.

MILESTONE	TIMELINE	ACTIVITIES
Establish working group	Q3 2021	• Define scope and aims of project • Decide on format of meetings • Identify starting objectives
Establish a key contact list	Q1 2022	• Identify key clinical, laboratory, data and policy contacts in each state and territory
Jurisdictional and national genomic plan document identification and review	Q2 2022	• Identify available national, state and territory genomic and implementation plans • Review plans for detailed content in the following areas: clinical services; digital/IT; education of genomic workforce; priorities for Aboriginal and Torres Strait Islander people; funding models; integration of research into clinical practice
Genomic plan gaps and inconsistency mapping to existing projects	Q3 2022	• Identify gaps or inconsistencies across jurisdictional genomic plans and map to other Australian Genomics or national initiatives (e.g. GHFM funded research)
Selection of gaps or inconsistencies to progress	Q3 2022	• Identification of key differences (that lack consistency) across jurisdictions and prioritisation for alignment directed by national need and working group expertise
Gap - Standards for bioinformaticians	Commenced Q3 2022, complete June 2024	• Mapped 20 vacant positions (July 2021 – Aug 2022) • Compared to previously employed positions within diagnostic laboratories (March 2014 – October 2020) • Mapped information on job roles and requirements; essential qualifications and skills; pay scales and employment stream
Inconsistency - Standard referral processes	Commenced Q3 2022, complete June 2024	• Map referral criteria for 24 public genetic service including: conditions referred (e.g. cardiac, cancer, etc.), additional testing offered (e.g. paternity, reproductive screening, etc.), referral process (e.g. self-referral, family or individual referrals, etc.), and clinical prioritisation categories (e.g. pregnant, cat 1, 2, 3, etc.) • Provided summarised data back to local genetic service asking for confirmation and/or additional comments • Compared publicly available to service confirmed information • Reviewed overall referral processes nationally and across entire jurisdictions (e.g. NSW as statewide referral vs individual service delivery)

Inconsistency - Testing processes/guidelines for non-genetic specialists: Cardiac and endocrine genetics	Commenced Q2 2023 – ongoing until Q1 2025	• Formation of specialised working groups (one each for cardiac and endocrine genetics) • Defining scope, audience and outputs • Design of decision support tools • Evaluation

Frequency of meetings / structure of activity:

- Quarterly, virtual meetings were held with project leads and coordinators.
- Coordinators had fortnightly meetings to progress research activities and subprojects.
- *Ad hoc* meetings were conducted as needed, e.g. for review of specific documents, briefing of a lead for a committee presentation.

Working groups for the decision support tool subproject were formed and have been held approximately monthly since April 2023.

Budget, Expenditure and Resourcing

This project relied on input and progression of milestones from Australian Genomics coordinators and in-kind contribution by the project leads.

For the design of decision support tools, a human factors consultant from Metro-North HHS (Qld) has been approached and will be involved in review of the tools. This is anticipated to occur once the decision support tools have been finalised by cardiac and endocrine genetics working groups and will be approximately 8 hours of work at a cost recovery fee of $250 per hour. A formal quote will be generated once decision support tool content and design are finalised and will be funded by Australian Genomics.

Methods

The comparison of jurisdictional and national plans formed the foundational evidence base from which the project was able to progress.

Methodology for Genomic Plan Review:

Process:

- Jurisdictional genomics plans were identified and reviewed for NSW (2017 and Implementation 2021-2025), WA (2022-2032), SA (2022), VIC (2021), and QLD (2017-2022), with no plan available for NT, ACT, TAS at the time of review (Q2 2022), in addition to the National Genomics Health Policy Framework (2018-2021) and accompanying Implementation Plan (AHMAC).
- Key priority statements were summarised from each genomic plan under target areas
    - Services, digital/IT, education of genomic workforce, priorities for Aboriginal and Torres Strait Islander people; funding models; and integration of research into clinical practice

- More detailed information was collated across jurisdictions, and gaps and/or inconsistencies were identified.

Comparisons:

- Gaps and/or inconsistencies were mapped against current national projects already underway (e.g. Australian Genomics priority projects or GHFM funded projects) to understand areas that were not already being addressed.

This activity identified 21 gaps/inconsistencies in total. Of these, 13 were mapped to existing projects already underway by Australian Genomics or other national initiatives. The remaining 8 of these were areas for potential progression under this project.

Proposal:

After review, the working group identified three priority areas for detailed review and alignment directed by national need and working group expertise, outlined below.

Subproject 1: Standards for Bioinformaticians

Currently, there is a lack of any accreditation requirement for bioinformaticians working in clinical diagnostic laboratories, including no standard level of education and no standard level of experience. The aim of this subproject was to determine the transition of clinical bioinformatic job roles as complex genomic testing becomes more widely accessible and commonly referred to diagnostic laboratories.

Process:

- Mapped 20 vacant positions (July 2021 – Aug 2022) and compared against those currently employed within clinical diagnostic laboratories (March 2014 – October 2020)
    - Utilised SEEK for current positions
    - Search terms within Health and Medical sub-category: bioinformatics, bioinformatician, genomics, genomics AND IT, software developer
    - Utilised direct contact and “past jobs” for prior positions
- Mapped information on:
    - Job roles and requirements
    - Essential qualifications and skills
    - Pay scales and employment stream

Comparisons:

- Compare job roles requirements, essential criteria, advertised salaries, employment streams, etc between two different time points (pre-2020 vs post July 2021)

Proposal:

Recommendation of a standardisation of award structures, conditions and minimum entry level requirements for bioinformaticians and consistency across jurisdictions.

Subproject 2: Standard Referral Criteria

Each state and territory have either a specific or partial referral plan, but jurisdictional differences exist between referral processes and pathways. This can often result in jurisdictional differences in genetic service delivery and patient equity of access. The aim of this project was to determine what genetic conditions are referred and accepted at clinical genetic services, and what referral processes are established.

Process:

- Project coordinators data mined publicly available information for 24 public, national genetic services (using clinical service websites) and collated referral criteria information.
- Mapped each genetic service for:
    - Condition(s) referred (e.g. cardiac, cancer, etc.)
    - Additional testing provided (e.g. paternity, reproductive screening, etc.)
    - Referral process (e.g. self-referral, family or individual referrals, etc.)
    - Clinical prioritisation categories (e.g. pregnant, cat 1, 2, 3, etc.)
- Summarised information was shared with respective clinical services seeking confirmation of the accuracy of the data and any additional comments.

Comparisons:

- Publicly available information versus service confirmed information
- Overall referral processes nationally, and across entire jurisdictions where a statewide service was not operational (e.g. NSW statewide referral vs individual local health network service delivery)

Proposal:

Establish a minimum set of accepted conditions nationwide, in additional to providing genetic services and jurisdictional government representatives with a summary document collating the differences, and consistent referrals vs non-referrals nationwide to assist with ongoing service delivery planning and development.

Subproject 3: Testing processes/guidelines for non-genetic specialists

Genomic testing is fast moving towards becoming a first line, standard-of-care approach for many genetic disorders, with recent MBS items providing subsidies for genomic testing in renal, cardiovascular, and other monogenic disorders. Recent success has been achieved in the kidney space with the development of genetic kidney disease decision support tools co-designed by KidGen (Australian Genomics) and Queensland Genomics. The aim of this subproject is to build on this body of work and adapt the decision tool format for other genetic conditions.

Process:

Based on the recent MBS subsidies for cardiovascular and endocrine genetic testing, these two areas were a logical choice for which to develop decision tools.

- The project coordinators used a program logic to outline the design, development and evaluation of the decision tools (Niselle et al., 2019)

- With the project leads, two working groups were established, inviting respective subject matter experts. Both groups are led by Chirag Patel who developed the kidney support tools.
- The working groups identified conditions based on best practice guidelines for genetic testing, which formed the basis of the decision tool content. The intended audience for these decision support tools are mainstream endocrinologists/cardiologists (consultants and trainees), clinical geneticists (consultants and trainees), and specialist endocrine/cardiac-genetic multidisciplinary team clinicians, so content was directed to this level of expertise and knowledge.
- Australian Genomics team members are continuing to assist with formatting the content of the decision tools. Human factors consultants from Queensland Health will also be engaged.

Comparisons / Evaluations:

- Following development and formatting, decision support tools will be shared for peer review. Following guidance from Bronwyn Terrill (Education Officer, Australian Genomics) regarding how the tools could be shared and accessed, a member of the Cardiac Society of Australia and New Zealand (CSANZ) has been invited to the cardiac working group and is attending meetings. This is part of the process for the CSANZ to endorse the final product. For endocrine conditions, the aim is for EndoGen (national specialist interest group) to endorse once complete (plus the Endocrine Society of Australia if possible). Endorsement will also be sought from the Human Genetics Society of Australasia for both sets of decision tools and working group members have engaged regarding the process.
- Evaluation of decision tool use may include an online survey of users of the decision tools to assess their utility, effectiveness and breadth of use.

Proposal:

Dissemination of endorsed cardiac and endocrine decision support tools to relevant stakeholders to facilitate standard genetic referrals for these conditions.

Evaluation of decision support tools to determine utility will be undertaken in 2024-2025.

Results

Outputs and outcomes

MILESTONE	OUTPUTS	COMMENTS TOWARDS PROGRESS / COMPLETION DATE
Establish a key contact list	**Appendix 2** - Table of state contacts for policy, lab, clinical and IT **Appendix 3** - SA map; policy, lab, clinical and IT (example of state flow and interconnections)	Q1 2022 *To be updated in 2024-2025 project.*

MILESTONE	OUTPUTS	COMMENTS TOWARDS PROGRESS / COMPLETION DATE
Jurisdictional and national genomic plan document identification and review	**Appendix 4** - Zipped folder of state and national genomic plans and relevant publications utilized for Genomic Plan mapping **Appendix 5** - State and Territory Health Systems Consistency genomics plan comparisons **Appendix 6** - State and Territory Health Systems Consistency_gaps and mapping	Q2 2022
Gap - Standards for Bioinformaticians	**Appendix 7** - Gap – Bioinformatics summary (excel spreadsheet - detailed) **Appendix 8 -** Gap – Bioinformatics summary (PowerPoint - high level)	Commenced Q3 2022, complete June 2024
Inconsistency - Standard referral processes	**Appendix 9** - Inconsistency – referral processes summary (excel spreadsheet - detailed) **Appendix 10 -** Inconsistency - referral processes summary (PowerPoint - high level) *Appendix 9a and 10a provide anonymised summary level information, access to detailed information must be requested and approved by working group representatives.*	Commenced Q3 2022, complete June 2024
Inconsistency - Testing processes/guidelines for non-genetic specialists	**Appendix 11 -** Inconsistency – decision support tool sample (note: not final version) **Appendix 12 -** Inconsistency – decision support tool Program logic for decision aid planning and evaluation **Appendix 13 -** International Clinical Cardiovascular Genetics Conference May 2024. Presentation by JD	Commenced Q2 2023 – ongoing until Q1 2025
Overall project	**Appendix 14 –** HGSA presentation, November 2022. Presentation by MJ **Appendix 15 -** Health systems consistency project summary slide deck **Appendix 16 -** Health Systems consistency 2-slide summary • Various presentations to Australian Genomics National Steering committee (NSC) and National Implementation Committee (NIC)	Completed June 2024

Outcomes from Genomic Plan Mapping

Key findings:

- While highly concordant, high-level mapping of available genomic plans identified 21 gaps and/or inconsistencies across the jurisdictions; 8 of which were found not to be addressed by existing Australian Genomics or other national initiatives.
- Three critical areas were selected for progression, determined by both national need and working group expertise (see below).

Some project members attended an invited meeting with Commonwealth Department of Health and Aged Care representatives following the announcement of Genomics Australia. The Commonwealth were looking to establish a taskforce to determine what strategies and frameworks exist within each jurisdiction and any barriers to implementing a national approach for translating genomics into the health system. The main aim was to determine what requires a local level solution versus a national readjustment. An overview of the state and territory health systems consistency priority project scope was provided to determine where both parties could work collectively, with genomic strategy plan high-level review and mapping data shared post meeting.

Subproject 1: Standards for Bioinformaticians

Employment stream:

- While clinically focussed, three quarters of the job positions were employed under a non-clinical employment stream, with nearly half being in research (likely funded by translational research projects).
- Past jobs - Large number employed within admin stream, most likely due to no dedicated employment stream available at the time of employment.
- Current – Increasing number of job adverts in private sector indicating a shift in funded healthcare delivery.

Qualifications:

- Despite Masters being the highest formal education available for a bioinformatician, large proportion of jobs required a PhD as a qualification requirement. However, there was a shift from PhD to postgraduate requirement in more recent job advertisements.

Job roles:

- Job advertisements are becoming more targeted to a specialised skillset (e.g. core bioinformatics or software development skills), whereas prior jobs (pre-2020) covered a broad range of skills required for undertaking a single role, often including clinical analysis.

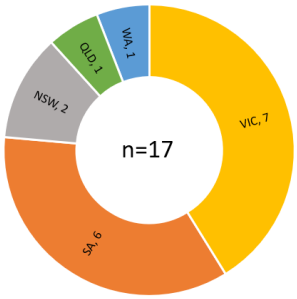
Salary:

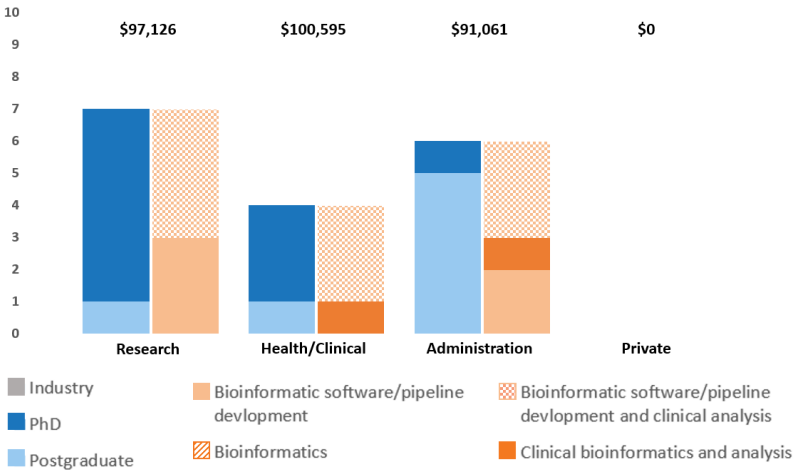
- Those employed in health/medical stream are paid more than those in research or admin streams, however, the private sector paid more than public sector.

Consistent with the evolving nature of genomics healthcare, there is an apparent evolution in job roles and requirements for clinical bioinformaticians. More recent adverts indicate highly specialised role descriptions, with some preference given for experience over qualifications. Despite these developments in the field of bioinformatics, there still exists discrepancies in role and qualification requirements that don’t align with current classifications and training. We propose a recommendation

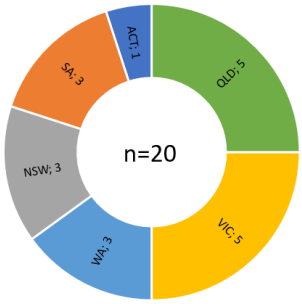
of a standardisation of award structures, conditions and minimum entry level requirements for bioinformaticians and consistency across jurisdictions.

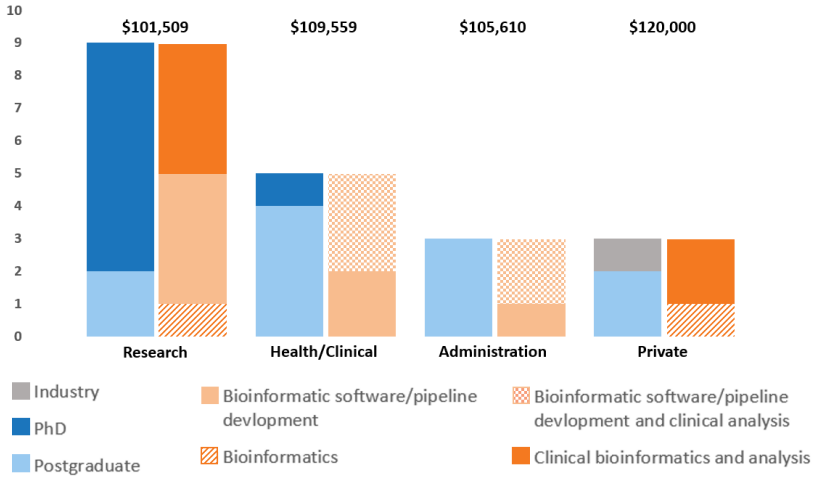
Prior positions - March 2014 – October 2020





Vacant adverts - July 2021 – August 2022





Subproject 2: Standard Referral Criteria

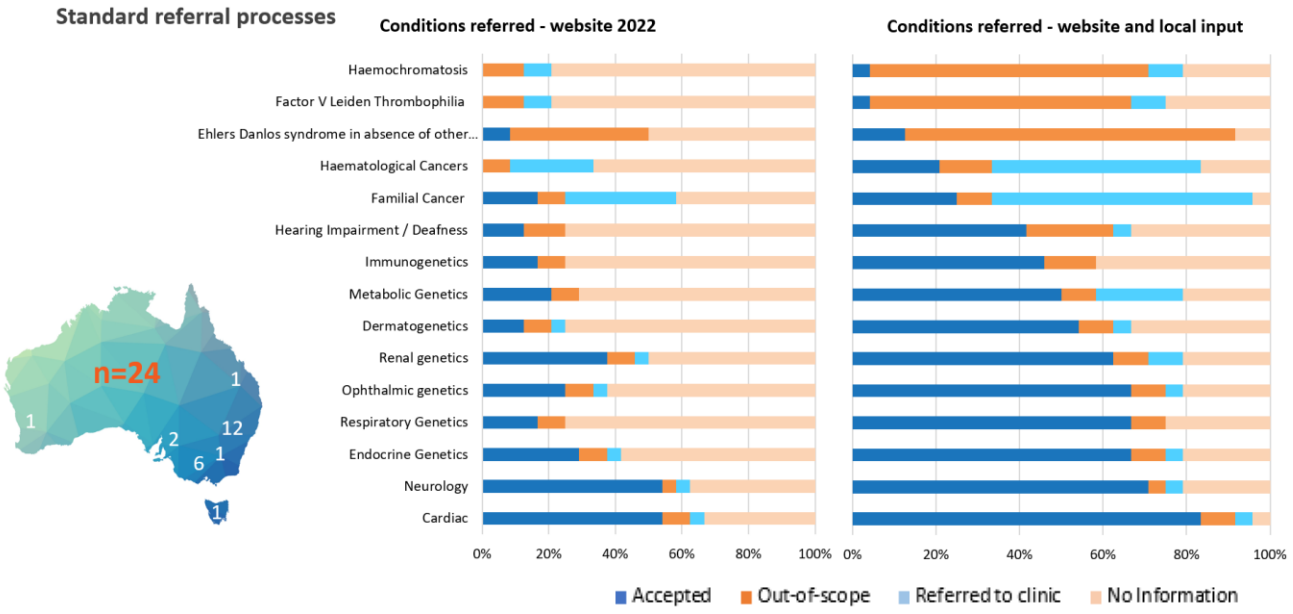
Key findings:

- There is a large disparity of information relating to accepted referrals between information available on public facing genetic service websites and information provided by a genetics service representative (e.g. genetic counsellor, head of service, or executive).
- Nine services updated their service websites between our review period (2022 vs 2024), mostly to update referrals to specialised clinics (e.g. cancer).

Condition(s) referred:

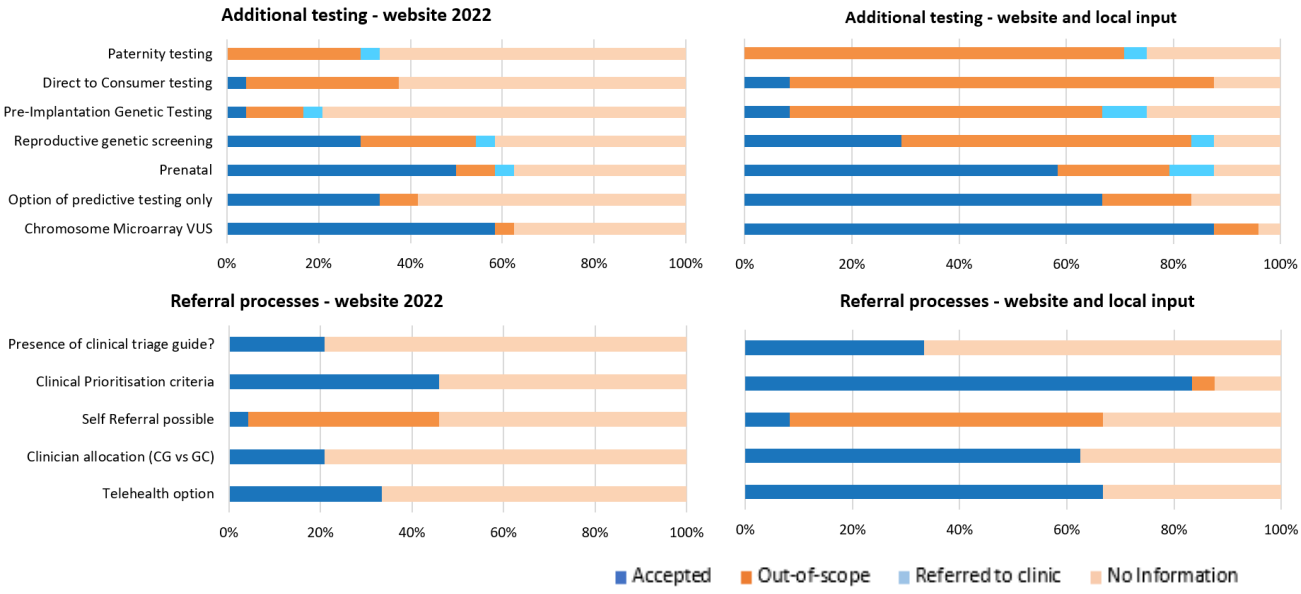
- Haemochromatosis, Factor V Leiden Thrombophilia and Ehlers Danlos syndrome in the absence of other life-threatening complications are the least accepted referrals nationwide.
- Cancers, both haematological and familial, are more likely to be referred to a specialised clinic than accepted by general genetics services.

- While cardiac and neurology are the most accepted referrals nationwide, ophthalmic, respiratory and endocrine are frequently accepted referrals, but are not generally specified as accepted conditions on genetic service websites.

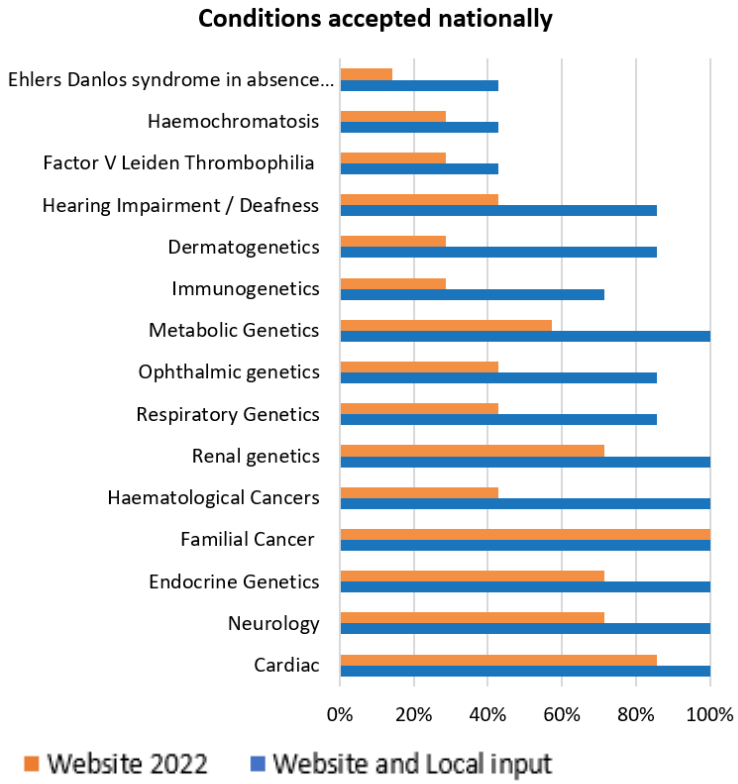


Additional testing and referral processes:

- Paternity testing, direct to consumer testing and pre-implantation genetic testing are the least accepted genetic tests nationwide, chromosomal microarray variants of uncertain significance (VUS) is the most frequently accepted test referral.
- Majority of services have a set clinical prioritisation criteria, but only approximately one-third of services have an established clinical triage guide.
- Most likely due to adaptations established in response to COVID-19, telehealth as a first appointment is accepted at over two-thirds of clinics nationwide.



- Cardiac, neurology, endocrine genetics, familial and haematological cancers, renal genetics and metabolic genetics referrals are an accepted condition in at least one service in each jurisdiction when considering VIC and NSW as a "statewide" service.



Providing relevant, accessible, and up-to-date information in the constantly evolving field of clinical genetic service delivery is often difficult, compounded by emerging or updated guidelines, MBS item numbers, and diagnostic test delivery that potentially shift referrals to in- or out- of scope. There is also the added complexity of workforce capacity and jurisdictional differences in funding models, that again can dictate or determine which referrals are accepted at a point-in-time. Despite this, there was a high level of accepted referrals nationwide across a range of genetic conditions, but this information was not always clearly presented in a public facing forum. We propose a minimum set of accepted referrals nationwide, and distribution of information back to local and jurisdictional representatives to assist in future service delivery and planning.

Subproject 3: Testing processes/guidelines for non-genetic specialists
Cardiovascular Decision Support Tool:

- The cardiac group has developed content for seven disease-specific decision support tools:
  - catecholaminergic polymorphic ventricular tachycardia (CPVT)
  - long QT syndrome (LQTS)
  - sudden cardiac death
  - hypertrophic cardiomyopathy (HCM)
  - Brugada
  - arrhythmogenic cardiomyopathy (ACM)
  - familial hypercholesterolaemia (FH)
  - aortopathies

- The group plans to develop content for one other condition, dilated cardiomyopathy (DCM).

Endocrine Decision Support Tool:

- The endocrine group has also developed content for a variety of endocrine conditions including:
  - maturity-onset diabetes of the young (MODY)
  - familial pituitary syndrome
  - osteogenesis imperfecta
  - monogenic/familial osteoporosis and hypercalcaemia
  - monogenic diabetes
- The group plans to progress development of content for:
  - Thyroid disorders
  - Primary ovarian insufficiency

A summary page, outlining the conditions covered in each suite will also be developed, as well as supporting pages that address how to order a test, interpret potential results and implications for both conditions. It is anticipated that the development and formatting of the decision support tools will be complete in Q1 2025. The output for both cardiac and endocrine decision support tools will be a PDF suite of guides for non-genetic specialists.

Other project outputs/outcomes:

A state contact list in relevant disciplines was created at the start of this project (**Appendix 2**) with an accompanying representative map (**Appendix 3**). These will be updated throughout the continuing Australian Genomics 2024-2025 project.

Presentations:

- Presentation to Australian Genomics National Steering Committee, 10 February 2022. Presentation by Kristen Nowak titled *"Australian Genomics State and Territory Health Systems Consistency Project".*
- Presentation to Australian Genomics National Implementation Committee, 23 February 2022. Presentation by Kristen Nowak titled *"Australian Genomics consistency of service project and mapping State and Territory genomics plans".*
- Presentation to Australian Genomics National Steering Committee, 12 August 2022. Presentation by Cliff Meldrum titled *"State Consistency Project".*
- Presentation to Australian Genomics Clinical, Diagnostic, Research Network, 15 September 2022. Presentation by Kristen Nowak titled *"Australian Genomics State and Territory Health Systems Consistency Project".*
- HGSA 45th Annual Scientific Meeting presentation, November 2022. Presentation by Matilda Jackson titled *"Australian Genomics State and Territory Health Systems Consistency Project Identifies 21 Gaps and/or Inconsistencies in National Genomics Health Plans".* Authors: Tiffany Boughtwood, Julia Dobbins, Clara Gaff, Matilda R Jackson, Julie McGaughran, Cliff Meldrum, Kristen Nowak, Michael C Quinn, Hamish Scott.

- Presentation to Australian Genomics National Implementation Committee, 23 November 2023. Presentation by Tiff Boughtwood titled "*Australian Genomics State and Territory Health Systems Consistency Project".*
- Presentation at International Clinical Cardiovascular Genetics Conference, May 2024. Presentation by Jason Davis titled *"Development of cardiac decision support tools to support cardiac genetic testing".*

Discussion

Impacts

Significance of the project:
As the application and uptake of genomic technology increases, it is vital to ensure equitable access across the nation. In a complex system providing healthcare to a dispersed population, bringing together specialised genomic resources and expertise encourages efficiency and collaboration. Mapping national genomic healthcare practices highlights the disparity of service delivery and workforce composition in each jurisdiction, warranting systematic service and professional guideline development, and introduction of decision support tools, where relevant. Re-review of local progress against documented jurisdictional priorities remains indispensable for evaluating translation.

Key impact(s):

- Mapping jurisdictional genomic plans and strategies provides an excellent benchmark from which to understand which areas of genomic healthcare are being adequately addressed or require attention. Detailed review of jurisdictional and national genomic plans and strategic priorities can aid in focussing resources without duplicating efforts with other established initiatives.
- Bioinformaticians are an increasingly valuable team members as healthcare becomes more digitalised. Working towards professional standards will aid in attraction and retention of bioinformaticians, while maintaining a high quality of work due to standardised education and training, contributing to improved outcomes across health systems.
- Capturing referral criteria and processes provides a single source of information outlining which genetic conditions are accepted at clinical services across Australia. This information is available online and accessed by clinicians/consumers seeking a referral, therefore ensuring accuracy and equity is critical and serves to improve access to genomic healthcare. This sub-project also aligns with National Health Genomics Policy Framework Priority area for action 1.2 "*Encourage appropriate referrals of genomic testing, that put the welfare and needs of the individual first, thereby avoiding unnecessary testing*."
- The development of decision support tools to guide non-genetic specialists ordering cardiac or endocrine genomic testing will improve the access to clinical genomic testing either directly, or by improving the referral information to clinical services for such testing. Overtime, these tools will ideally work to upskill interested clinicians.

A large amount of high-level and detailed information has been collated over the duration of this project. The generation of summary documents and slide decks that can be disseminated and reviewed by relevant parties and stakeholders are an invaluable resource with the potential to positively impact the delivery of genomic healthcare.

Limitations

Evolution of state genomic plans and review:

- The genomic plan mapping represents a point-in-time comparison.
- Several states have since updated their genomic and implementation plans during the span of the project, which has not been captured in our analysis.
- Mapping was also limited by the unavailability of genomic plans for ACT, NT and TAS.

There is likely disparity between what is documented as a priority and what is progressed and achieved at a jurisdictional and national level. It is understood that this will be addressed by a Department of Health and Aged Care review in preparation for the update national framework and implementation plan.

Closed bioinformatic job adverts:

- On review of closed bioinformatic jobs (advertised pre-2020), there were limitations in jobs which could be included due to broken links or job descriptions no longer being available. Personal communication with jurisdictional diagnostic laboratories did provide some required documents that were no longer available online.

Accessing referral guidelines:

- As with mapping genomic plans, comparison of online referral criteria information reflects a point in time analysis only.
- It was often challenging to find appropriate information online, without local jurisdictional access (e.g. service log-in).
- There was a large disparity between public facing information and service confirmed information. This is important to consider, especially for non-genetics specialists who may review online information when deciding whether to refer a patient to a genetics service for consultation.
- There were some difficulties in receiving confirmation of publicly available information by a genetic service representative, with two services not able to provide a response.

Decision support tools:

- Subject matter expert time was provided in-kind, and meetings were sometimes challenging to coordinate due to conflicting work schedules and multiple time zones.

Overall: While the project team can undertake the data mining, mapping and initial consultations, the implementation of these findings/recommendations is not within our capacity. We have therefore tried to engage and interact with relevant groups/societies to assist in continued progression. For example, engagement with the CSANZ regarding the testing processes for non-genetic referrals, and engagement with ABACBS for dissemination of bioinformatician data. To aid in appropriate

engagement with other groups/societies the networks of the project leads, National Steering Committee and broader Australian Genomics members can be utilised.

Recommendations and Future Directions

- This project will progress with the Australian Genomics 2024-2025 grant opportunity. An updated project plan has been provided as **Appendix 17**.
- The coordination team will help to facilitate regular interaction with other project working groups during the 2024-2025 grant (including but not limited to: Clinical NAGIM, Indigenous Genomics, Equity, Policy, Translation and Implementation, Support of the National Health Genomics Policy Framework and Implementation Plan Review, Workforce Education).
- Facilitate re-review of respective genomic plans through the dissemination of our mapping data and summary, including involvement in consultation phase of the new framework and implementation guidelines.
- Facilitate and encourage further progression in the areas of standards of bioinformaticians and standard referral criteria through dissemination of our findings back to key stakeholders and interested parties (e.g. clinical and diagnostic genetic services, ABACBS sub-committee, representatives from the RCPA and HGSA, etc).
- Evaluate other international organisations/structures to determine common elements, or key barriers and enablers to the translation of genomics healthcare, e.g. Canadian federal and provincial governance and Pan-Canadian Genomics strategy announced in 2021.

Conclusion

The delivery of clinical genetics in the Australian public healthcare system is complex due to a variety of geographical, logistical, governance and regulatory factors.

This project used a review of jurisdictional genomic plans and strategies as a mechanism to map and prioritise gaps and inconsistencies across the Australian healthcare system, in the context of diagnostic and clinical genomic service delivery.

Three gaps/inconsistencies were chosen for advancement to make a meaningful difference. Information on standardisation of bioinformaticians has been passed on to relevant professional bodies for consideration. A broad review of the scope and acceptance criteria of referral into clinical genetics services will be reflexed back to clinical services with the aim of reducing inequities and fragmentation. Finally, in ongoing work, a suites of decision support tools are being developed to guide cardiac and endocrine specialists and trainees with genomic test requests across Australia.

Although this priority project has achieved many of the planned endpoints, there is still further work to be done – particularly communications between diagnostic and clinical leads across the country; harmonisation of the new health genomics framework and a continued focus to assess genomic health access inequities across groups including Aboriginal and Torres Strait Islander peoples.

References

Genomic Plans as source material for Genomic Planning mapping component:

State

South Australian Clinical Genomics Plan 2022, March 2019
NSW Health Genomics Strategy June 2017
NSW Health Genomics Strategy Implementation Plan 2021-2025
Genetic and Genomic healthcare for Victoria 2021: Improving the health and wellbeing of Victorians
Statewide Genetic Health Queensland Service Plan 2017-2022
WA Health Genomics Strategy 2021
WA Genomics Strategy 2021-2031: Towards Precision Medicine and Precision Public Health
(Note that there were no plans for the Northern Territory or Australian Capital Territory at the time of data collection).

National

Health Genomics Policy Framework 2018-2021
Implementation Plan: National Health Genomics Policy Framework. Driving National Action 2018-2021.

Other References

Australian Institute of Health and Welfare 2016. Australia's health 2016. Australia's health series no. 15. Cat. no. AUS 199. Canberra: AIHW.

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https://www.melbournegenomics.org.au/themes/custom/melbourne_genomics/assets/Genomics-in-Victoria-Whats-changed-in-10-years.pdf

Pan-Canadian Genomics Strategy https://ised-isde.canada.ca/site/genomics/sites/default/files/attachments/2023/pancanadian_genomics_strategy_wwh-en.pdf

Appendices

Summary of Appendices:

1. State and Territory Health Systems Consistency - program logic

2. Table of state contacts for policy, lab, clinical and IT

3. SA map; policy, lab, clinical and IT (example of state flow and interconnections)

4. Zipped folder of state and national genomic plans and relevant publications utilized for Genomic Plan mapping

5. State and Territory Health Systems Consistency genomics plan comparisons

6. State and Territory Health Systems Consistency_gaps and mapping

7. Gap – Bioinformatics summary (excel spreadsheet - detailed)

8. Gap – Bioinformatics summary (powerpoint - high level)

9. Inconsistency – referral processes summary (excel spreadsheet – detailed, password protected)
   *9a – anonymised version *local and jurisdictional information removed, national level presented.*

10. Inconsistency – referral processes summary (powerpoint - high level, password protected)
    *10a - anonymised version *local and jurisdictional information removed, national level presented.*

11. Inconsistency – decision support tool sample (note: not final version)

12. Inconsistency – decision support tool program logic for decision aid planning and evaluation (DRAFT)

13. International Clinical Cardiovascular Genetics Conference May 2024. Presentation by J Davis. Development of cardiac decision support tools to support cardiac genetic testing.

14. HGSA presentation, November 2022. Presentation by M Jackson *"Australian Genomics State and Territory Health Systems Consistency Project Identifies 21 Gaps and/or Inconsistencies in National Genomics Health Plans".* Authors: Tiffany Boughtwood, Julia Dobbins, Clara Gaff, Matilda R Jackson, Julie McGaughran, Cliff Meldrum, Kristen Nowak, Michael C Quinn, Hamish Scott.

15. Health systems consistency project summary slide deck

16. Health Systems consistency 2 slide summary

17. AG 2024-2025 State and Territory Health Systems Consistency project plan