Australian Genomics Business Continuity Plan

Submission to the NHMRC and Australian Department of Health (GNT2000001)

Professor Kathryn North AC
Lead, Australian Genomics
Director, Murdoch Children's Research Institute

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Australian Genomics

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1. EXECUTIVE SUMMARY

Australian Genomics is an Australian Government-funded initiative supporting genomic research and its translation into clinical practice. Through broad engagement and a national collaborative approach, Australian Genomics improves the efficiency, reach and timeliness of genomic research projects. It supports the implementation of genomics research outcomes to inform evidence-based policy development and clinical practice.

Australian Genomics has been supported by time-limited research grants via National Health and Medical Research Council (NHMRC) since 2016. Through this valuable partnership with the Australian Government over the past six years, Australian Genomics has strategically built a national, multidisciplinary genomic network with excellent connections to international genomic initiatives.

Current funding expires in 2023, at a time of unprecedented genomic market growth; increasing complexity in the genomic health ecosystem; and ongoing investment by Commonwealth and State/Territory governments in genomic research, policy, and clinical translation. In this context, Australian Genomics plays a vital role in analysing and strategically preparing stakeholders for future directions and priorities, in identifying sources of funding and in providing operations to support the continuation of world leading genomics in Australia.

This document articulates a strategy to secure a long-term funding mechanism, beyond time-limited grants which will be essential for the translational benefits of health genomics to be achieved (e.g. acute care genomics, newborn screening, pharmacogenomics, novel precision therapies). The options available to Australian Genomics are evaluated and a way forward is proposed. This Business Continuity Plan is for submission to NHMRC and the Department of Health for approval under Section 10.5 of the Australian Genomics Grant Program grant guidelines.

Summary of findings

Australian Genomics has been instrumental in progressing the integration of genomics into mainstream healthcare in Australia. Today, Australian Genomics is at a pivotal point of change, looking to transition from its strong foundations toward a sustainable ongoing mechanism of funding and operations. This comes at a time of unprecedented momentum in global health genomics implementation, growing demand for clinical genomic testing globally and rapid acceleration of investment in genomic research in Australia, both at a national and jurisdictional level. Health genomics is projected to transform the way healthcare is delivered throughout the course of an individual's life, enabling rapid diagnosis, prevention, early intervention and targeted therapies. Australian Genomics now has the opportunity to drive excellence in genomics innovation and translation to improve the health and wellbeing of all Australians, at all stages of life.

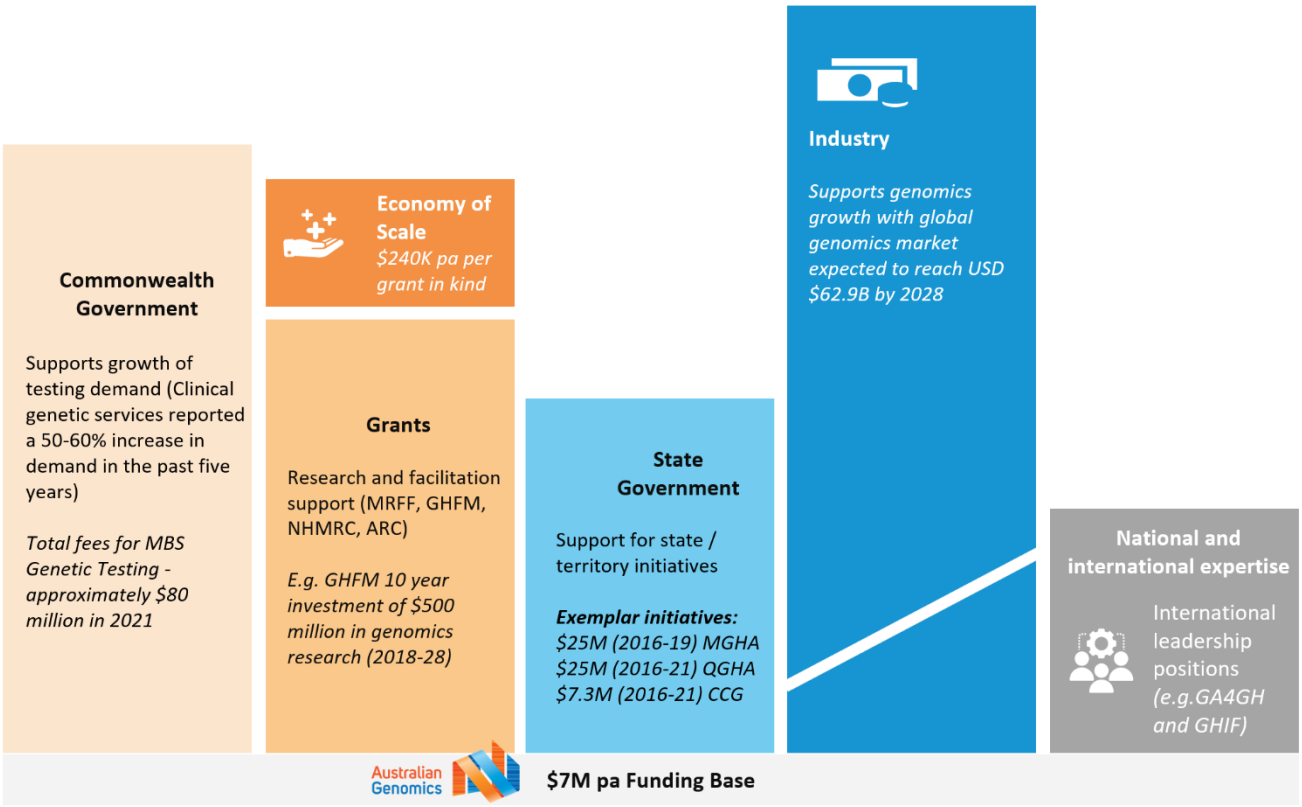
The genomics ecosystem in Australia is complex, delivering a multi-million dollar value chain spanning research innovation and capabilities, health system policy and strategy, through to clinical genomic service provision and diagnostics. Genomics policy and health practice in Australia has evolved rapidly, however, the implementation of genomic medicine as standard of care requires commitment and alignment across this entire ecosystem. Australian Genomics is uniquely placed to drive national genomic initiatives, with a network of collaborators and investigators, across Governments, health services, researchers, diagnosticians, industry and infrastructure providers.

While the existing model has been critical in driving genomics innovation, this Business Continuity Plan has determined a future vision for Australian Genomics to "***foster, support and translate genomics innovation: from research to health outcomes, involving all Australians in sharing the benefits of health genomics***".

The future role determined for Australian Genomics focuses on driving a national approach to improving the impact and efficiency of genomic research and translating this into policy and practice. This positions Australian Genomics as a "***connector***": a central piece of the genomics ecosystem in Australia that convenes expertise to drive key implementation programs. It provides a "unified voice" for key bodies across genomics and mitigates the risk of having scattered pockets of research across Australia.

An assessment of future government and non-government funding sources determined that due to specific dynamics of the nascent genomic research and medicine market in Australia, **the most effective model of funding is a primarily Australian Government funded entity to act in the public interest, with an annual budget of $7 Million,** in line with national genomic initiatives around the globe**.** Australian Genomics will transition to a company limited by guarantee, led by a CEO, with the potential to attract philanthropic funding and partner with industry bodies to develop supplementary revenue streams. Further discussion with Government will be required to determine the optimal governance structure to provide certainty for the future of genomics in Australia.

As Australian Genomics provides a unifying single point of contact across the genomics value chain, its role in amplifying existing investment is critical. As depicted below, investment in Australian Genomics acts as an impact multiplier for investments in genomics from Government, grants, industry and international players (with the significant size of investment reflected in respective heights). Working in collaboration, Australian Genomics will enhance investment to transform the way we deliver Australian healthcare throughout the life-course.

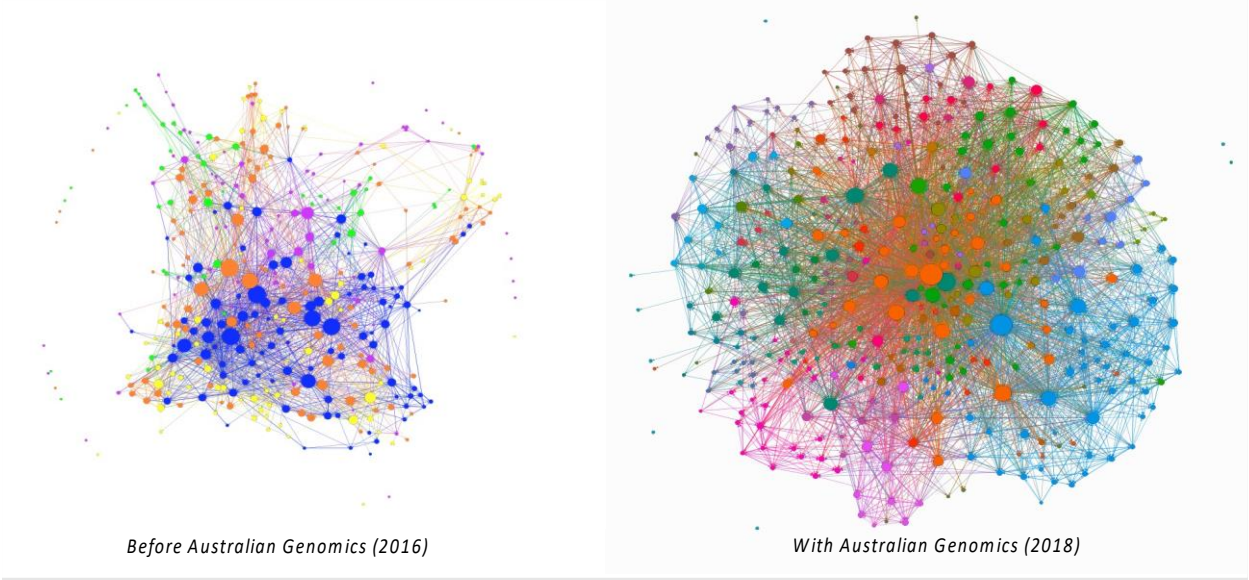


There is ongoing and critical need for a national genomic initiative in Australia as a single and independent point of connectivity across this complex ecosystem: driving health safety, quality and equity of access – and driving health innovation for the benefit of all Australians.

2. AUSTRALIAN GENOMICS TODAY

Australian Genomics was launched with a NHMRC Targeted Call for Research (TCR) 'Preparing Australia for Genomic Medicine' ($25M, 2016 – 2020). This grant evaluated the clinical utility and impact of offering genomic testing to over 5000 Australians with rare disease or cancer, across 18 different clinical 'flagship' pilot studies. In parallel, we developed and piloted the infrastructural elements that are required for the implementation of genomics into the health system: education and upskilling the workforce; development of national approaches to data capture, analysis, governance and federation; and evaluation of the economic, policy, regulatory and ethical implications of genomics in Australian health care. As part of this program of work, Australian Genomics developed an extensive portfolio of genomic research tools and resources, and built a national coordination network, including the clinical genetics devices and diagnostic laboratories in every State and Territory. The effectiveness of these capabilities has been demonstrated through the successful delivery of the TCR, as well as the support of several other government-funded research initiatives including Mackenzie's Mission ($20M), the Cardiac Genetic Disorders Flagship ($6M), Massimo's Mission ($3M) and the Acute Care Genomics study ($5M). Critically, in those five years Australian Genomics has established a large, inclusive, national multidisciplinary genomic network, connected to international genomic initiatives. The engagement of this genomic learning community of over 450 investigators and 100 partner organisations has been recognised as instrumental to the successful implementation of genomics into healthcare, and is a significant legacy of the TCR[1].

Figure: Growth of the Australian Genomic Learning Community[2]



Before Australian Genomics (2016)

With Australian Genomics (2018)

The Australian Genomics Grant Program (AGGP, $15M, 2021 – 2023) builds upon this foundation of research, partnerships, and international networks, to ensure genomic advances continue to be integrated into research, policy and health care practice - and can meet new challenges and opportunities. Australian Genomics is improving the efficiency, reach and timeliness of genomic research, and the implementation of genomic advances into healthcare. Under this model, Australian Genomics has changed focus to reflect the changing needs for progressing genomics in Australia: providing research services, developing genomic infrastructure, and supporting health system translation.

[1] Stark, Z, et al; AJHG 105, 7-14, 2019
[2] Long, JC, et al; BMC Medicine 17(44), 2019

What we have achieved:

Australian Genomics has supported the translation of government-funded research programs that are now ready to be implemented into health care:

- **Mackenzie's Mission** implementing reproductive carrier screening for 750 severe genetic conditions *(a world first)*
- **A national Acute Care Genomics program** for rapid 3-day diagnosis of critically ill children in every state and territory (*a world first*)
- **Rare disease diagnosis** delivering five times the diagnostic rate at a quarter of the cost (*now a Medicare item*)
- **Cancer diagnosis and surveillance** leading to early diagnosis and targeted interventions
- **National collaborative clinical and research networks** to catalyse high quality research and translation
- A national system for **sharing genetic evidence between diagnostic laboratories**, '*Shariant*'
- Partnership with Aboriginal and/or Torres Strait Islander advisers and communities to develop **Indigenous-led genomics programs**
- **A robust governance structure** with jurisdictional membership to consult and drive implementation into clinical practice
- Leadership roles in the **Global Alliance for Genomics and Health** and the **Genomics and Health Implementation Forum,** international consortia focussed on responsible sharing and utilisation of clinical and genomic data

What we are developing:

- A comprehensive suite of **tools, resources and capabilities** to enhance the efficiency, impact and translation of government-funded genomic research in Australia
- National program in **pharmacogenomics** to reduce preventable harm from medicines in the primary care setting (*responsible for $1.4B cost per annum to the health system*[3])
- An evidence-base for genomics to **predict high burden disease risk** (*e.g. diabetes, cardiovascular disease and cancer*)
- National platforms and technologies in **functional genomics** to increase diagnostic yield and underpin precision medicine and cell and gene therapies
- The prototype for a **National Approach to Genomic Information Management,** a national legacy dataset of clinical and genomic data for clinical applications and research.

[3] Pharmaceutical Society of Australia: Medicine Safety, Take Care (2019)

OUR IMPACT

5,200
Patients recruited / genomic testing

100%
Results returned

32
Recruitment sites

450
Collaborators & Investigators

103
Organisational partners

117
Jobs created

55
PhD / Masters research students trained

$99 million
Research supported

21
Cancer & Rare Disease studies

129
Peer-reviewed papers

21
Reports

115
Ethics & site submissions per year

23
Sub-projects in Genomic Health Service Delivery

723
Presentations

54
Workshops & conferences

13
Policy submissions

4
Position statements

10
Advisory Boards

3. THE HEALTH GENOMICS LANDSCAPE - National and International

The momentum of **global health genomics implementation** is extraordinary. The global genomics market is expected to reach USD 62.9billion by 2028 with the Asia Pacific region predicted to exhibit the highest growth internationally (19% compound annual growth rate, 2021 – 2030)[4,5]. There are now 41 countries who have founded national genomic initiatives to drive the implementation of genomic medicine into healthcare, and a further 86 countries have targeted genomic projects[6]. 95% of these initiatives are either public funded, or public-private partnerships, for example: Japan ($126M AUD), Singapore ($210M AUD), USA ($1298M AUD), Switzerland ($100M AUD)[3].

These national genomic initiatives develop clinical and population cohorts; establish research and health system infrastructure including standards and platforms for the collection, storage and sharing of data; and resources for workforce and public genomic education.

The Global Alliance for Genomics and Health (GA4GH) unites 670 member organisations from 90 countries to share approaches, develop guidelines and standards, and through this collaborative approach avoid duplication of effort7. The members of the GA4GH Genomics in Health Implementation Forum (GHIF) represent 32 national initiatives that are progressing government funded programs for the translation of evidence-based policy in genomics into standard of care in their healthcare systems. Australian Genomics holds leadership roles in both of these international genomic consortia.

Investment in **genomic research in Australia** has also been accelerating. The National Health and Medical Research Council and Australian Research Council have together awarded an estimated $338M toward genetic/genomic-related research through competitive grant schemes since 2014 (around $292M and $46M, respectively)[8]. Notably, almost half of this funding was awarded in the past 2.5 years, with $157M allocated since 2019. The Australian Government's Medical Research Future Fund (MRFF) has funded $218M in genetic/genomic projects since 2017, of which $150M was allocated from the $500M Genomics Health Futures Mission. Other Federal Government investments include the Zero Childhood Cancer Program ($20M), and the Australian Cancer Genomic Medicine Program ($50M). There has also been $116M in jurisdictional investment in health genomic research and translation in the past decade, with State/Territory Government genomics alliances launched in Victoria, New South Wales, Queensland, ACT and South Australia.

Despite the substantial Government investment in health genomic research, it is well documented that the path to **translation of clinical research** is long, and inefficient. Approximately 85% of biomedical research investment is wasted: with only 14% innovations entering clinical practice, and 1% to important health impacts[9]. To capitalise on Australia's genomic research investment and expertise, a strategic and sustained platform to support translation into healthcare is required, particularly given the complexities of the Australian health policy and practice landscape.

Genomics policy and health practice in Australia has also rapidly evolved in the past five years. The Australian Government Department of Health's National Health Genomics Policy Framework (NHGPF) was published in 2017[10], and the Project Reference Group on Health Genomics was established to oversee delivery of State and Territory participation in the NHGPF

[4] Next Generation Sequencing Market, Allied Market Research, July 2021
[5] Research and Markets, https://www.researchandmarkets.com/research/7wb4qz/genomics_market?w=5
[6] Kovanda, A. et al; Human Genetics (2021) doi.org/10.1186/s40246-021-00315-6
[7] https://www.ga4gh.org
[8] Publicly available grant outcomes were searched for the terms 'genetics' and 'genomics'. Epigenetics was not included. Projects were considered individually for relevance and inclusion. https://www.nhmrc.gov.au; https://www.arc.gov.au
[9] Chalmers I, Glasziou P. *The Lancet* 2009;374(9683):86-89.
[10] National Health Genomics Policy Framework, 2018; ISBN 978-1-76007-328-2

Implementation Plan[11], however with the dissolution of the AHMAC under the National Cabinet, the PRG was disbanded in 2020, with some functions falling under the remit of the Health Chief Executive's Forum. Most States and Territories have also independently developed their own genomic strategies. As health genomic technologies become mainstreamed, and with the pan-disciplinary nature of the technology, genomic priorities will also be reflected in broader Government strategies and policies, including the National Preventative Health Strategy (2021), and Primary Health Reform (2021), as well as the Zimmerman Report (2021) (*see below*). Further, the reliance on - and generation of - data associated with genomics intersects with the activity of the Australian Digital Health Agency, as reflected in the National Digital Health Strategy and National Healthcare Interoperability Plan (2022).

Beyond policy, the **funding of clinical genomic testing** in Australia is fragmented. Most aspects of clinical genomic service provision are via State/Territory funding, block funding, Activity Based Funding – or paid by the patient themselves. There is a lack of clarity and jurisdictional consistency with respect to funding of genetics services which results in diminished service capacity, inequity of patient access, and insufficient budget for genomic interventions – even though genomics has been demonstrated to drive system-wide efficiencies in both clinical benefit and cost effectiveness[12]. The Medical Services Advisory Committee (MSAC) is receiving an increasing number of applications for genomic services, but not all genomic interventions are appropriate for MBS subsidisation[13].

Concurrently, the **demand for clinical genomic testing** in Australia is growing rapidly. The Stocktake conducted by the Royal College of Pathologists in 2017 reported 1,700 genomic tests (whole exome or genome sequencing) conducted in the 2016/17 financial year[14]. By mid-2019, public accredited pathology services reported a volume of 4,000 genomic tests annually, and growing. Clinical genetic services across the country have also reported a 50-60% increase in demand in the past five years[18].

Industry players in the genomic value chain in Australia span from medtech/device companies, biotech infrastructure or laboratory supply organisations, through to pharmaceutical companies and private pathology providers – many of which are large multinationals, with a local presence. Effective engagement with industry will be critical to health genomic research translation, genomic testing capacity in Australia, and the generation of commercial outcomes – delivering both health, and wealth for the nation.

Genomics in Australia is a complex ecosystem delivering a multi-million dollar value chain spanning research innovation and capabilities, health system policy and strategy, through to clinical genomic service provision and diagnostics. It includes stakeholders from Governments, health services, researchers, diagnosticians, industry, infrastructure providers and an increasingly health-consumer oriented public.

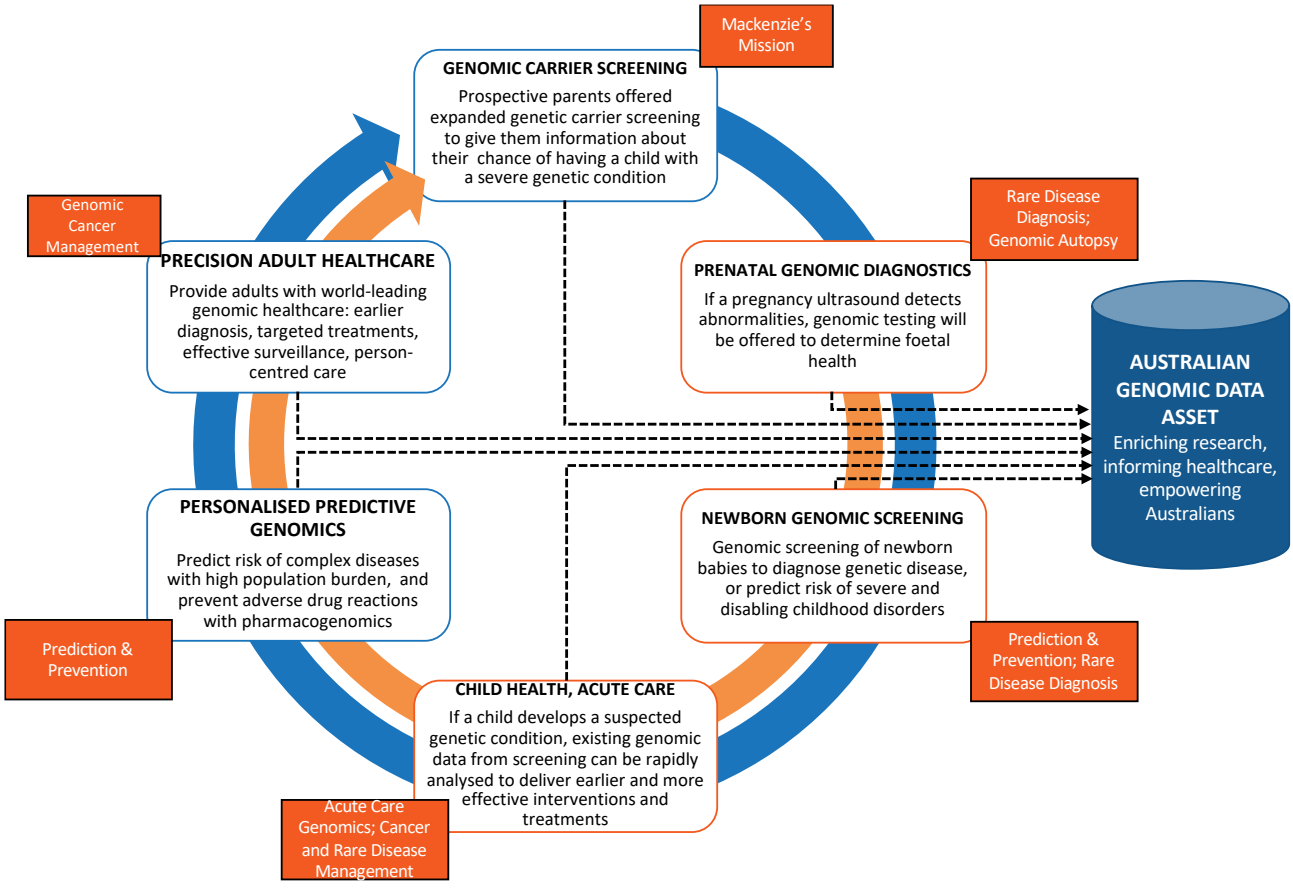
See Appendix 1 for a more comprehensive description of the Health Genomics Landscape Today.

[11] Implementation Plan - National Health Genomics Policy Framework, 2018; ISBN 978-1-76007-363
[12] Australian Genomics response to the Independent Hospital Pricing Authority (IHPA) Consultation Paper on the Pricing Framework for Australian Public Hospital Services 2022-23
[13] Norris, S., et al. J Community Genet (2021)
[14] https://www1.health.gov.au/internet/main/publishing.nsf/Content/national-health-genomics-stocktake

THE FUTURE FOR HEALTH GENOMICS IN AUSTRALIA

Internationally, Australia is celebrated as an early, and effective adopter of health genomic innovation. Considering the substantial research investments by Federal and jurisdictional Governments; the proactive development of genomic health policy; and the eagerness to embrace and implement genomic technologies by researchers, clinicians and health consumers alike; Australia has established the infrastructure and expertise to translate genomic promise, into system benefit.

Genomics is a pan-disciplinary technology that has the potential to bring cost efficiencies and clinical utility beyond tertiary referral centres – genomics will be increasingly mainstreamed into non-genetic specialty services (including nephrology, cardiology, obstetrics, paediatrics), primary care (carrier screening and prediction of disease risk), and even into pharmacies (pharmacogenomics to predict adverse drug events). This pan-disciplinary nature of genomics also speaks to a future where **health genomics will transform the way we deliver healthcare throughout the life course, enabling rapid diagnosis, prevention, early intervention and targeted therapies.**



With genomics informing personalised diagnosis, risk prediction and person-centred care throughout the life course, a nationally coordinated approach becomes critical to drive research strategy and efficiency; to support research outcomes to navigate the path to uptake and implementation; and to ensure Australian health policy and practice in health genomics is evidence-based, equitable, and sustainable. It is critical to drive truly patient-centred genomic healthcare: balancing the need to implement genomics as an appropriate intervention, considering potential harms, health system implications and public trust. Further, with the commercial potential and market growth of the genomic industry, without sustained and strategic development of genomics locally, Australia risks losing its competitive edge in the international genomic stage, and losing market share and expertise to global players.

There is ongoing need for a national genomic initiative in Australia as a single and independent point of connectivity across this complex ecosystem: driving health safety, quality and equity of access – and driving health innovation for the benefit of all Australians.

This is not a question of sustainability of Australian Genomics, but the sustainability of genomics in Australia.

'The Zimmerman Report' The New Frontier – Delivering better health for all Australians

In November 2021, The House of Representatives Standing Committee on Health, Aged Care and Sport published the report of their inquiry into the approval processes for new drugs and novel medical technologies in Australia, with a particular focus on those for the treatment of rare diseases and conditions where there is high and unmet clinical need[15]. Informed by 207 submissions and public hearings, the report makes a suite of recommendations to ensure Australia continues to be well positioned to access and implement novel medical technologies efficiently and effectively, in response to global trends. Australian Genomics applauds the Committee's findings, and notes that many of the recommendations align with the established activities and structures of Australian Genomics - and would be readily transferrable into a future national Australian genomics healthcare initiative:

#	Recommendations of the Zimmerman Report	Australian Genomics Alignment
1	Establish a Centre for Precision Medicine and Rare Diseases. - Advise the Department of Health and the Australian Medical Research Advisory Board on research priorities - Education and training information, support for patients and horizon scanning for novel medical technologies	- Australian Genomics coordinated the GHFM Scientific Strategy Committee to identify strategic research priorities - Genomicsinfo.org.au is a resource for patients and publics on genomics and health - The Australian Genomics 'future diagnostic technologies' working group is establishing a living map of emerging health technologies
2	Establish a National Genomics Testing Program - Building on GHFM, establish a jointly funded national genomics testing program to provide equitable access to genomic testing nationwide; ensuring the provision of genomics counselling for all patients	- Opportunity to build upon Australian Genomics nationally coordinated approach; network of clinical and diagnostic genomic services; and national network of clinical sites - Our work on the capacity and development of the Genetic Counselling workforce can be leveraged, in collaboration with HGSA
3	Establish an Office of Clinical Evaluation - To serve a "living evidence" function to ensure HTA is based on the most up-to-date global health practices	- Opportunity to leverage Australian Genomics' national network of experts and international collaborations
5	Health Economists (labour market and skills strategy)	- Australian Genomics has championed novel approaches to economic assessments, incorporating empirical measures of public and patient preferences to interventions
16	Increase International Collaboration	- Opportunity to leverage Australian Genomics' leadership roles in international health consortia, and active collaborations (inc. NHS England)
21	Improve Newborn Screening (NBS) Program - Standardisation of newborn screening nationally	- Opportunity to build upon Australian Genomics nationally coordinated approach; network of clinical and diagnostic genomic services; and national network of clinical sites in collaboration with public health units

[15] https://www.aph.gov.au/Parliamentary_Business/Committees/House/Health_Aged_Care_and_Sport/Newdrugs/Report

	- Expand NBS based on genomic insights and international best practice	- The application of genomics in NBS is a sensitive and important topic, and requires broad community consultation and evidence-based approaches informed by existing policies[16]
22 to 26	Improve the Clinical Trial System in Australia 'Urgently' harmonise HREC & SSA processes A national clinical trial register - facilitate patient identification, patient recruitment, patient retention and completion	- Australian Genomics has published on the need for harmonization of research HREC/governance processes, and established a national network of 32 recruitment sites nationally - We have developed participant platforms that facilitate engagement, dynamic consent and quality of data capture
27	Research and Development	- Opportunity to build upon Australian Genomics nationally coordinated approach; network of clinical and diagnostic genomic services; and national network of clinical sites
28	The Patient Voice	- Involve Australia is a community-led project in Australian Genomics to develop guidelines to optimize consumer involvement in research

Implementation of the recommendations of the 'Zimmerman Report', and those of the National Approach to Genomic Information Management, will be critical to Australia harnessing the transformative potential of the 'new frontier' of medical innovation. Building upon Australian Government research investments to date, Australian Genomics can promptly address many of the aspirational recommendations of these reports – and accelerate the realisation of gains to the health system, and the Australian public.

[16] Newborn Bloodspot Screening National Policy Framework ISBN: 978-1-76007-364-0; Population Based Screening Framework, ISBN: 978-1-76007-370-1

4. THE BUSINESS CONTINUITY PLAN CONSULTATION PROCESS

Australian Genomics partnered with PwC to support with the development of the Business Continuity Plan. PwC has a depth of experience in delivering work across the Health and Government sectors in Australia and globally. The team brings expertise in business and operating model design and provided independent perspectives to Australian Genomics.

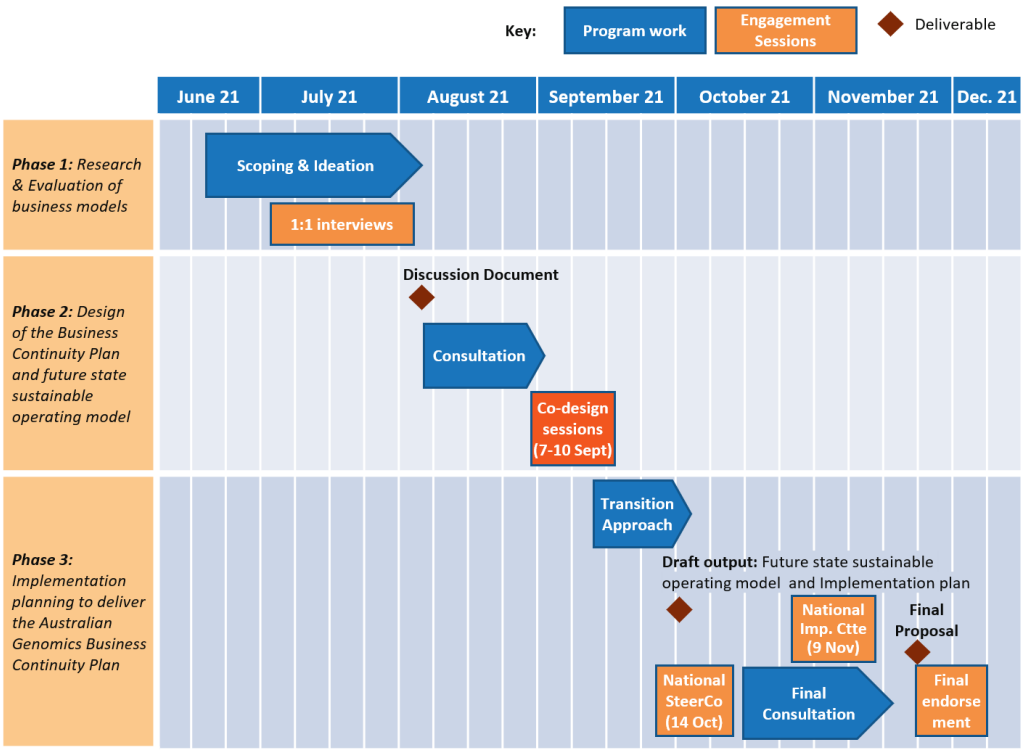
The development of the Business Continuity Plan followed three broad phases: research, design and implementation planning. The approach was designed to ensure broad engagement with stakeholders to identify how Australian Genomics can best continue to drive the implementation of genomics into standard practice.

Phase 1 included research and evaluation of different organisational models, as well as interviews conducted to generate in-depth insights from senior stakeholders with expertise into the future for genomics in health – and what will be needed to drive ongoing momentum and excellence in genomic research, health system innovation, and commercialisation for Australia.

These insights informed the Discussion Document distributed in Phase 2 of the Business Continuity Plan process, which presented an opportunity for a broad stakeholder consultation to inform Australian Genomics' future roles, activities, priorities, funding models and operations.

Following the written consultation period, co-design sessions were held with the Australian Genomics Community Advisory Group and Independent Advisory Board in early September. The sessions provided opportunities to workshop emerging themes and outputs from interviews and consultation, then consider Australian Genomics' vision, to clearly establish its purpose aspirations. The sessions also determined the optimal future role that Australian Genomics should embody to optimise Australia's genomic research efficiency and drive the translation of genomics research into healthcare appropriately, equitably, and sustainably.

The final phase of the process involved amalgamating consultation findings into a final report and conducting implementation planning to deliver the Australian Genomics Business Continuity Plan.



5. EXPLORING FUTURE ROLES FOR AUSTRALIAN GENOMICS

Through the process of Phase 1 research and stakeholder interviews, three broad roles were identified that could support the future optimisation of genomics research and translation in Australia: connector, standards provider, and standards endorser. These roles were described and shared widely with stakeholders for discussion in Phase 2 of the Business Continuity Plan process. Each role has distinct focus areas across each of the four key activities outlined above, and leverages different governance structures, funding mechanisms and capabilities *(see Appendix 2: Consultation Feedback).*

Key takeaway:

The roles consider the scope of current Australian Genomics activity, and depict what roles a national genomics initiative for Australia should encompass into the future. Stakeholders considered the elements of each role against their potential to best position Australian Genomics to continue to drive health genomic implementation in Australia and globally.

The transition to a national Australian genomics healthcare initiative will necessitate a focus on connecting and enabling bodies across the genomics ecosystem to improve the impact and efficiency of genomic research and drive the translation of that research into health policy and practice. The connector focus is critical to Australian Genomics driving the translation of evidence-based policy in genomics into standard of care in the health system to improve the health and wellbeing of all Australians. Its ability to build "critical mass" and work to avoid fragmentation in all domains across the federation is a critical requirement of the genomics ecosystem. This role best positions Australian Genomics to promote competition but avoid duplication across genomic bodies in the future.

Additionally, a focus on facilitating greater cooperation will be necessary to drive uniform infrastructure and data interoperability nationwide, to enable Australia to realise the full potential of genomics healthcare in Australia. Feedback was provided that Australian Genomics should define its role in relation to how it operates and works with different bodies in the genomics landscape in Australia, which has been addressed in Section 6: Proposed Future for Australian Genomics.

For additional detail see Appendix 2: Consultation Feedback

The role of Australian Genomics in progressing a national health genomics initiative

The role that Australian Genomics has defined in the current genomics ecosystem has been instrumental to the progressive integration of genomics into healthcare in Australia. As detailed in Section 2, under the current model, Australian Genomics effectively provides research services, develops genomic infrastructure, and supports health system translation. Throughout the stakeholder consultation process, Australian Genomics was referred to as "a common and central connection point" for genomics healthcare.

While the existing model has been critical to progressing genomics innovation to date, this plan seizes the opportunity to look at the sustainability and future evolution of genomics in Australia and its implications for Australian Genomics' model, funding, governance and capabilities. As genomic medicine will transform the way healthcare is delivered throughout the life course of Australians, it is imperative that this plan explores how the future role of Australian Genomics can best support the progress of the Australian health care system to one of prediction, prevention and personalised treatment. This section explores these options and looks to comparator organisations for best practice.

Chosen Role and Key Activities

Key Activities	Connector (Chosen Role)	Standards Provider	Standards Endorser
Research Enabling Activities			
Facilitate Information Sharing	✓	✓	✓
Uplift Research Efficiency and Reach	✓	✓	✓
Streamline Research Processes (e.g. Ethics)	✗	✓	✓
Provide Accreditation for Processes (e.g. Ethics)	✗	✗	✓
Data Management, Governance & Infrastructure			
Provide a Data Platform	✓[17]	✓	✓
Provide Data Management Leadership	✓	✓	✓
Advise on Data Management Issues	✓	✗	✗
Provide E2E Data Infrastructure	✗	✗	✗
Health System Translation			
Advocate for Health System Translation	✓	✓	✓
Advocate as a Message Amplification Leader	✓	✓	✓
Act as a Peak Advisory Body	✓	✗	✗
Become the key Government Adviser	✗	✗	✗
Key Partnerships / Collaborations			
Engage with Local and National Genomics Bodies	✓	✓	✓
Engage with International Genomics Bodies	✓	✓	✓
Engage with International Healthcare Bodies	✓	✗	✗
Engage with International + Adjacent Industries (e.g. pharma)	✓	✗	✗

The Connector Role

This role focuses on driving a national approach to improving the impact and efficiency of genomic research and translating this into policy and practice. This places emphasis on leveraging Australian Genomics' unique national scope to bring order across the genomics landscape to avoid duplication and allow competition, so that all stakeholders can contribute to a collective whole. Members of the Independent Advisory Board described this role as strengthening Australian Genomics' natural trajectory into being that of a "circuit board" for genomics in Australia; a central piece of the genomics ecosystem in Australia that advances research and its translation into healthcare. The role posits a national genomics healthcare initiative that drives excellence in genomics innovation and translation to improve the health and wellbeing of all Australians, at all stages of life.

[17] *Note: Data Platform provision under Connector Role is the maintenance of the Shariant platform*

The connector role places a continued focus on enabling research, data management and health system translation advisory. A "unified voice" in the health translation space is a critical component as it mitigates the risk of scattered pockets of research across Australia, which would diminish the safety, access and quality of healthcare. The larger scale of Australian Genomics' future role accounts for the increased funding profile for genomics research over the coming years[18], and increasing scope of genomics applications, spanning rare diseases, cancer, infectious diseases, screening, pharmacogenomics, and common disease. By strengthening its role in the growing genomics ecosystem, Australian Genomics is also positioned to equitably disseminate tools and resources, including consent materials, education materials and surveys, to support more efficient research ethics and governance. This enables access for patients across the continuum of care in Australia to the same genomics healthcare and standard clinical processes, ensuring the safety and equity of our healthcare system.

Exemplar Organisations:

A connector-style role has been employed by different organisations internationally within and beyond genomics healthcare. Astronomy Australia Limited (AAL) is a national non-profit organisation, with members from Australian universities and research organisations, that fosters collaboration between academia and industry by building networks and attracting co-investment[19]. It presents a compelling model as the organisation generates "buy in" and generates additional revenue through members' subscriptions. However, all three 'connector' exemplar organisations are supported by substantial, sustained

[18] Medical Research Future Fund (MRFF) 10-year investment plan

[19] https://astronomyaustralia.org.au

DECLAN

Leukodystrophy Flagship

Declan loves rugby union and surfing.

When his legs started to fail him, he and his family went through a difficult journey of hospital visits and countless tests in search of answers.

While at first tests suggested Declan had an unknown leukodystrophy, genomic analysis through our Leukodystrophy Flagship helped piece together that Declan had a very rare genetic condition called VPS13D Spinocerebellar Ataxia. A confirmed diagnosis has given Declan a path forward in managing his condition so he can focus more on doing what he loves.

"The results were good, as they gave my condition a name… it's helped bring peace of mind to me, my family, and my friends," says Declan.

government funding (e.g. $16.6M FY20/21, Astronomy Australia).

Genomics England and Genome British Columbia both work to connect healthcare ecosystems across geographies and are recognised as leading national voices for genomics health system translation in their jurisdictions[8,20]. The pertinence of these exemplars is demonstrated in the similarities in the scope of activities, strategic engagement and global presence: the 'connector' role' effectively positions these organisations as national initiatives driving research innovation and translation.

6. CONSIDERING FUTURE FUNDING MODELS FOR AUSTRALIAN GENOMICS

The funding models that Australian Genomics could adopt have been analysed, with the below section detailing the viability of both government and non-government revenue streams. Given the expanding scope of genomic healthcare globally, the significance of Australian Genomics' role is only expected to increase into the future. Therefore, the sustainability of each future funding model option has been a core assessment criterion. Australian Genomics has been funded entirely by time-limited NHMRC grants to date, however, a combination of funding models in the future has been explored for consideration, as detailed below.

Key findings:

Australian Genomics is currently funded by Australian Government (NHMRC) research grants. An assessment of future government and non-government funding sources identified that continued core government funding will be required. The health genomics landscape in Australia is a nascent market, and implementation into public health practice is not yet mature. Consequently, ongoing government investment will be required to stabilise, and capitalise on, Australia's progress in health genomics to date. Over the coming years, Australian Genomics can look for opportunities to develop compelling propositions to open up new funding streams, some of which have been explored in detail below. However, until the genomics landscape matures, Australian Genomics should focus on government appropriation models that support its future operations, through providing more sustainable income predictability.

Of all the considered government funding models, the most feasible model to enable Australian Genomics to sustain its activities and drive genomics innovation is annual appropriation. With an agreed investment plan based on the costings in this report, this model allows for the efficient allocation of funds to entities based on historic appropriation with adjustments for inflation and potentially efficiency dividends.

Government funding models

Different forms of government appropriation and their viability have been analysed in supporting the sustainable future of Australian Genomics.

One-off Grants & Contributions funding

This funding mechanism utilises grants and contributions from Government bodies, administered for a finite period to research bodies through competitive or targeted research mechanisms. Australian Genomics is currently funded by NHMRC (GNT2000001) to support government-funded genomic research nationally and improve research efficiency, effectiveness and impact. Such funding has enabled Australian Genomics to strategically build

[20] https://www.genomebc.ca

a national, multidisciplinary genomic network, which is recognised as being instrumental in the successful implementation of genomics into healthcare in Australia.

One-off grants provide the potential to receive substantial income with some flexibility. However, as Australian Genomics looks to build a more sustainable operating model, longer-term income predictability is necessary. Moreover, employing a method that provides stronger consistency of funding allows for Australian Genomics employees to reprioritise time spent on grant applications into more outcomes focused activities, and would permit the development and execution of longer-term projects.

There was a consensus from stakeholder and government feedback that one off grant funding will no longer be viable for Australian Genomics in the future. This form of appropriation hinders the assurance provided by Australian Genomics to 'customers' and beneficiaries that its support can still be provided in the years to come.

Capability based funding

This funding mechanism involves funding based on capabilities which are revisited periodically, typically through an investment plan. Government may commit to a total amount of funding, which enables entities to engage in long-term planning within this funding "ribbon".
An Integrated Investment Program can be used to plan and agree to the cost of acquiring and sustaining capabilities (e.g. ADHA digital capabilities) over a longer time period, with offsets for major projects outside the Integrated Investment Program. For organisations which require large scale infrastructure or equipment investments in complex assets, this approach is most appropriate. It ensures that entities are accountable for meeting agreed, specific strategic goals, and Government is accountable for funding the required capabilities. However, this approach is not flexible as it locks into the development of specified capabilities.

Government cost-recovery

This is a fee-for-service funding model based on full cost recovery of operating expenditure, which is not used commonly except for extenuating circumstances (e.g. emergency funding to mitigate the impact of a pandemic). Whilst this method determines required funds based on organisational expenditure for services, it is seen to be inefficient spend from a Government perspective and used more commonly as a last resort measure.

Activity-based funding model

This funding model is based on level of activity, commonly used in public health whereby entities get paid for the number and mix of health services delivered. The question in this mechanism lies in whether Australian Genomics has a mandate to "clip the ticket" on genomics research and translation activities. This model would tie funding more closely to genomics healthcare, especially as it is mainstreamed, however, the inherent complexity in changing the funding structure of health genomics services renders it unpreferable for Australian Genomics and funding bodies alike.

Annual Appropriation

This form of government funding is a type of funding whereby funds are allocated to entities based on historic appropriation and is adjusted for inflation and potentially efficiency dividends. This traditional distribution of revenue is considered to be simple for Governments to administer. It relies on simple and accurate costings and is considered to provide reasonable value for money for Government if it is underpinned by an agreed investment plan. Potential pitfalls for government lie in the fact that funding may not be adjusted for shifting priorities, however, this can be mitigated if Government agree to additional line-item funding for specific initiatives, determined on a case-by-case basis.

While this model is most commonly employed for funding government departments and agencies, for the case of simplicity and transparency, Australian Genomics will target this as its preferred model. This does not render other government funding models as unviable, however this model provides the most simplicity in approaching government bodies.

Exemplar organisations were investigated, Australian Radiation Protection and Nuclear Safety Agency (ARPANSA) and Cancer Australia all receive annual appropriations. It should also be noted that Government appropriation may be diversified across portfolios based on the focus of genomic activity – for example the Department of Education, Skills and Employment in support of research infrastructure and capabilities (through the National Research Infrastructure Scheme); Department of Industry, Science, Energy and Resources; and the Australian Department of Health – acknowledging the risk of competing priorities, and unclear reporting lines.

Alternative funding models

Fee-for-Service

A Fee for Service Payment model involves the unbundling and separate payments for services or activities. Many stakeholders posited a fee-for-service model as a viable option with careful consideration of risks. Notably, Australian Genomics supports significant genomic research projects to the value of $99 million. It facilitates and supports research projects to reduce duplication of effort across jurisdictions, provide access to international best practice standards, increase exposure for national reach, and streamline any project mobilisation processes. The collective in-kind value of these research capabilities, tools and resources amounts to $240,400 per annum for each project (*for additional detail please see Appendix 3: Australian Genomics support of genomic research projects*). Such support may provide additional funding mechanism opportunities as a proportion of research grant value could be reappropriated from research grants to Australian Genomics. It should be noted that this funding model would likely necessitate facilitation or promotion by grant agencies, asking that research projects include a line item in their budgets to access Australian Genomics services, rather than the current in-kind support. While this 'mandate' to partner with Australian Genomics would be particularly relevant to genomic information management of government funded projects and build a national data asset for future research use, this model has additional layers of complexity in requiring wider government body support and endorsement and would likely not be feasible.

Subscription Model

A subscription model charges members a recurring fee at regular intervals for access to an organisations' network, resources and/or activities. Stakeholders advised careful consideration towards how Australian Genomics might charge an annual "membership fee" with concerns that changing from a "free service" to a membership model particularly for services required in clinical settings may be disruptive and impact upon equitable access to genomic services. Upon considering what subscription fees could be charged, it was determined that PanelApp Australia should remain a free platform as it is currently an open platform used by laboratories globally, clinical and research groups to record and share structured gene-disease validity assessments, with a subscription model potentially reducing crowdsourcing contributions.

Shariant Australia[21] provides real-time national sharing of information about the pathogenicity of genetic variants between Australian laboratories and clinical services. It is currently used by 19 laboratories, and there is interest from private pathology providers, and New Zealand services. The possibility of charging a yearly fee to laboratories for use is being considered, detailed below (*see Vignette 1: Shariant*), which may be feasible if the use of a curated variant sharing

[21] https://shariant.org.au/accounts/login/

database is mandated by laboratory accreditation guidelines, as opposed to "encouraged"[22]. However, without accelerated uptake through a reliance on Shariant, willingness to pay would be a significant inhibiting factor. Based on a current state of 19 laboratories using Shariant today and an assumption of constrained user growth over the next three years, a membership fee of $20,000 would only generate $440,000 in 2024. If the subscription fee was $50,000, the revenue generation opportunity is still only $1,100,000 in 2024 *(See Appendix 5: Australian Genomics Funding Source Modelling for further detail)*. Without significant user uptake, Australian Genomics will struggle to charge subscription fees that promote equity of access whilst generating a substantial level of funding. However, as detailed below, a subscription model remains an option for consideration in future years, depending on future accreditation requirements, and regional uptake.

VIGNETTE 1: Shariant and subscription model

Shariant, an initiative of Australian Genomics in partnership with QIMR Berghofer and SA Pathology, is improving the efficiency, consistency and safety of genomic diagnostics in Australia.

Shariant is a controlled access platform that enables Australian laboratories and clinical services to automate the sharing of detailed and structured scientific evidence about clinically curated variants; communicate in real-time to resolve variant interpretation differences; and access gene and disease-focused expertise.

Variants are changes in a patient's genetic code identified by a genomic test that may help diagnose or treat their condition. Decoding the clinical significance of DNA variants is complex and challenging when clinical laboratories operate in silos. Sharing variant interpretations across laboratories improves accurate diagnoses and the management of patients' health. Importantly, it supports a process to automatically identify and resolve discrepancies in variant interpretation. Shariant also facilitates the sharing of variant interpretations internationally.

As of December 2021, Shariant has more than 14,000 prospective variant interpretations from 11 clinical genetic testing laboratories across four states. Eight laboratories from another two states are in the process of joining. Given the rapid growth in large-scale genomic testing in Australia, Shariant will be instrumental in assisting healthcare professionals deliver better genomics results, thereby improving Australian patient management and treatment.

Philanthropy

Philanthropy is a higher-risk funding mechanism that relies upon philanthropic donations to generate funds. Australian Genomics considered the potential to attract donors and whether appetite exists in the genomics field, looking to exemplar organisations to determine how actively philanthropy was utilised. Notably, Genomics England received a £27 million philanthropic donation from The Wellcome Trust for genome-sequencing hub in 2014. However, whilst organisations may be eligible to receive philanthropic donations, there are considerable drawbacks: attracting philanthropic investment requires considerable organisational investment and effort to obtain; bequests may be received sporadically and without long-term certainty. Nevertheless, Australian Genomics can look to define its legal structure as a company limited by guarantee to attract philanthropic funding as a means of

[22] S1.8, Requirements for Human Medical Genome Testing using MPS technologies – NPAAC

generating additional revenue, particularly in the context of programs addressing specific disease areas.

Conclusion

To maintain Australian Genomics and position it as a national genomic initiative driving health safety, quality and equity of access, government funding will be required, as genomics in Australia is still a nascent market subject to considerable research activity and health translation. Opportunities to access additional revenue through alternative streams will be actively pursued in the future - via interactions with industry, philanthropy, and data providers. Australian Genomics should focus on targeting annual appropriation, due to its economic viability, simplicity for Governments to administer, value for money and predictability when administered through an agreed investment plan. Annual appropriation should also provide opportunities for additional line-item funding to be agreed for specific initiatives on a case-by-case basis.

EVIE

Acute Care Flagship

Meet seven-year-old Evie. "Evie is a vivacious, bubbly, strong-willed, happy-go-lucky little girl," says her Mum, Natasha Wagner.

In 2019, Evie became critically ill with a life-threatening liver condition. From the intensive care unit at The Royal Children's Hospital, Evie was referred to Australian Genomics' Acute Care study for ultra-rapid genome sequencing.

The results showed Evie had a rare recessive genetic condition called Wilson's disease.

The rapid diagnosis set Evie on an extraordinary journey that has seen her circle back to the family's dairy farm in Western Victoria where once more she leads a happy, healthy life.

7. PROPOSED FUTURE FOR AUSTRALIAN GENOMICS

A proposed future for Australian Genomics has been defined based on broad stakeholder consultation and genomic landscape analyses. To determine the future state, this section defines Australian Genomics' vision, its role and activities, governance structure, capabilities and cost base and proposed funding model. Each component sets the foundations to support the sustainability of genomics in Australia and position Australian Genomics to drive the implementation of genomic medicine as standard of care.

Key findings:

Australian Genomics places the beneficiaries of genomics healthcare at its core, defining its future vision to "foster, support and translate genomics innovation: from research to health outcomes, involving all Australians in sharing the benefits of health genomics."

Based on Section 5, Australian Genomics will be focusing on connecting bodies across the genomics landscape, as it is best positioned to leverage its national reach to progress health translation, support data interoperability and avoid duplication across genomic bodies.

Australian Genomics will operate as a company limited by guarantee, independent of Murdoch Children's Research Institute, with its current advisory structure providing strong foundations for coordination of a national, multidisciplinary genomics network in Australia and globally. Any required changes will be factored into this structure upon administrative or legal advice.

New organisational capabilities required under Australian Genomics' future operating model include building business-to-business and potentially business-to-consumer engagement as genomics is mainstreamed. Brand management will become more important for the organisation, and marketing capabilities in services pricing may also be necessitated through alternative funding models in future, to ensure the organisation is responsive to stakeholder preferences and member willingness-to-pay.

Section 6 determined the optimal funding model to be annual appropriation, with detailed modelling of different funding options detailed below. Deeper analysis of the current cost base and future state funding requirements revealed three possible scenarios for Australian Genomics' future funding base in 2024, dependent on available funding:

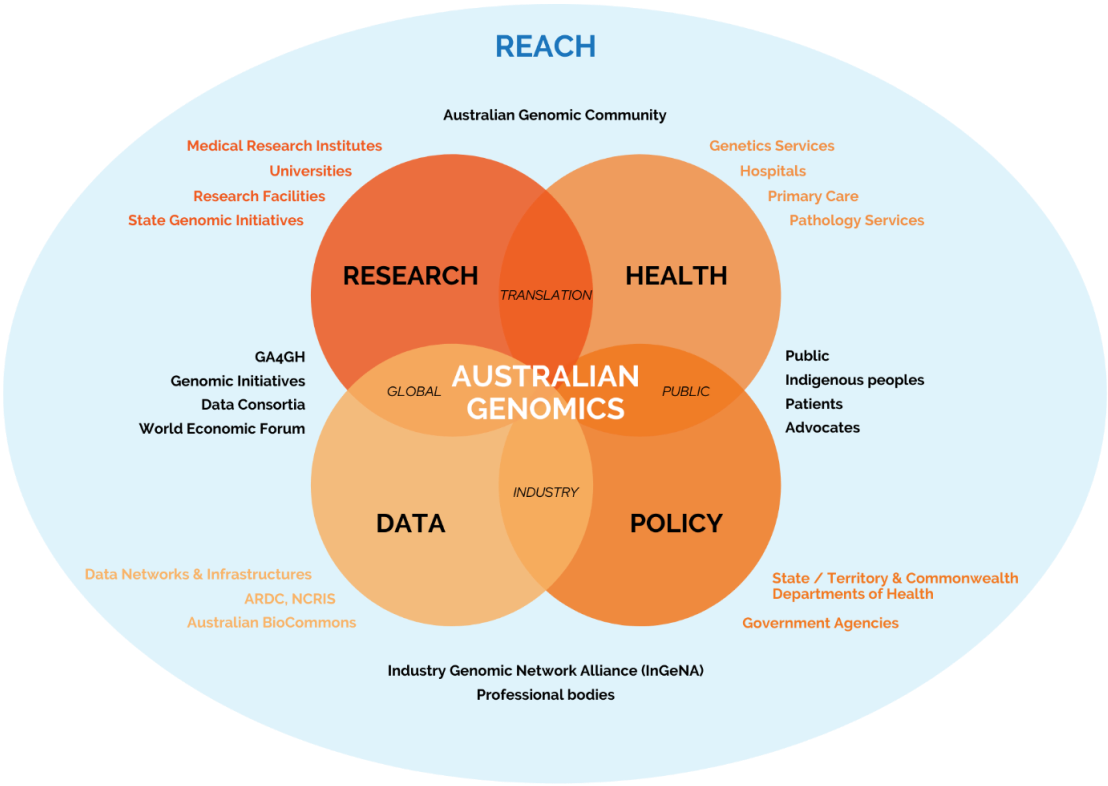
- **Option 1:** $5M funding base would limit the breadth of organisations engaged as a "connector", with policy development, community engagement, international involvement and data management to be jettisoned.
- **Option 2:** $7M funding base would support genomics research and provide capability for Australian Genomics to drive policy development, continue community engagement, international involvement, and also provide core data services.
- **Option 3:** $10M funding base would allow Australian Genomics to drive a national approach to data management and enrich genomics databases, as well as focus on policy and research translation to identify commercial opportunities and support research outputs in industry, and enable stronger community and industry engagement, and international leadership.

Vision

Australian Genomics engaged the Independent Advisory Board and Community Advisory Group to workshop a vision that is substantial and visionary, reflecting the genomic community's idealistic motivations for doing Australian Genomics' work. It underpins the Business Continuity

Plan, and provides a unifying focal point of effort in determining immediate and longer-term aspirations.

Foster, support and translate genomics innovation: from research to health outcomes, involving all Australians in sharing the benefits of health genomics.



Chosen Role and Key Activities

The connector role positions Australian Genomics to allow for competition but avoid duplication and disaggregation across genomic bodies. As outlined in Section 5, the role's focus on connecting bodies with a streamlined national approach will enable the convening of expertise to drive key implementation programs. The "unified voice" also provides opportunities to build future infrastructure at scale, especially with future funding mechanisms looking beyond MSAC, as much genomic innovation is not fit for small scale decisions.
The activities underpinning the role are detailed below, expanding on Section 5, with the reach of each activity determined largely by the future funding base:

Research Enabling Activities: Research Priorities Setting
Support in determining prioritisation across genomics space of most important initiatives – becoming a mechanism to align investment priorities for greater impact - providing direction, framework, and strategic planning in collaboration with key stakeholders
Australian Genomics supports research projects through providing governance, facilitation support, access to international resources, data storage and project mobilisation streamlining (e.g. Research ethics approvals). This enables stronger integration of collaboration, coordination and research tools and resources into the day-to-day operations of genomic research in Australia. Australian Genomics leverages its network to not only enable researchers to maximise their impact through genomic health translation, but to determine where the most important priorities lie within the genomics landscape and provide direction guided by Australian and international experts. The scope of Australian Genomics research activities will depend on its future funding, as detailed in Cost Base Modelling below.

Infrastructure: Data Management Adviser

Engage with the data community to improve the interoperability of datasets and infrastructure - moving from being a "provider" to supporting infrastructure providers to maintain international best practice in data management

Stakeholders emphasised the importance of Australian Genomics' role in improving the interoperability of data, enabling secondary research and efficiency in data discovery and sharing. As numerous entities in the genomics space in Australia have adopted a more active role in providing platforms, tools and resources, Australian Genomics looks to focus less on being an infrastructure "provider" and more towards a "connector" to support infrastructure providers to maintain international best practice, specifically with regards to data management.

Australian Genomics can be a force to mitigate duplication of effort while still allowing for competition so that all organisations can contribute to a collective whole. Hence, stronger data interoperability prevents incorrect diagnoses and lack of access to treatments caused by fragmentation across datasets. The level of funding made available to Australian Genomics (detailed below) will determine if it can leverage its national and international reach to progress and implement research outcomes addressing challenges in clinical and genomic datasets. Australian Genomics has the potential to drive an approach to enriching genomics databases, thus improving equity of access and public benefit derived from genomics in healthcare.

Health System Translation: Peak Advisory Body

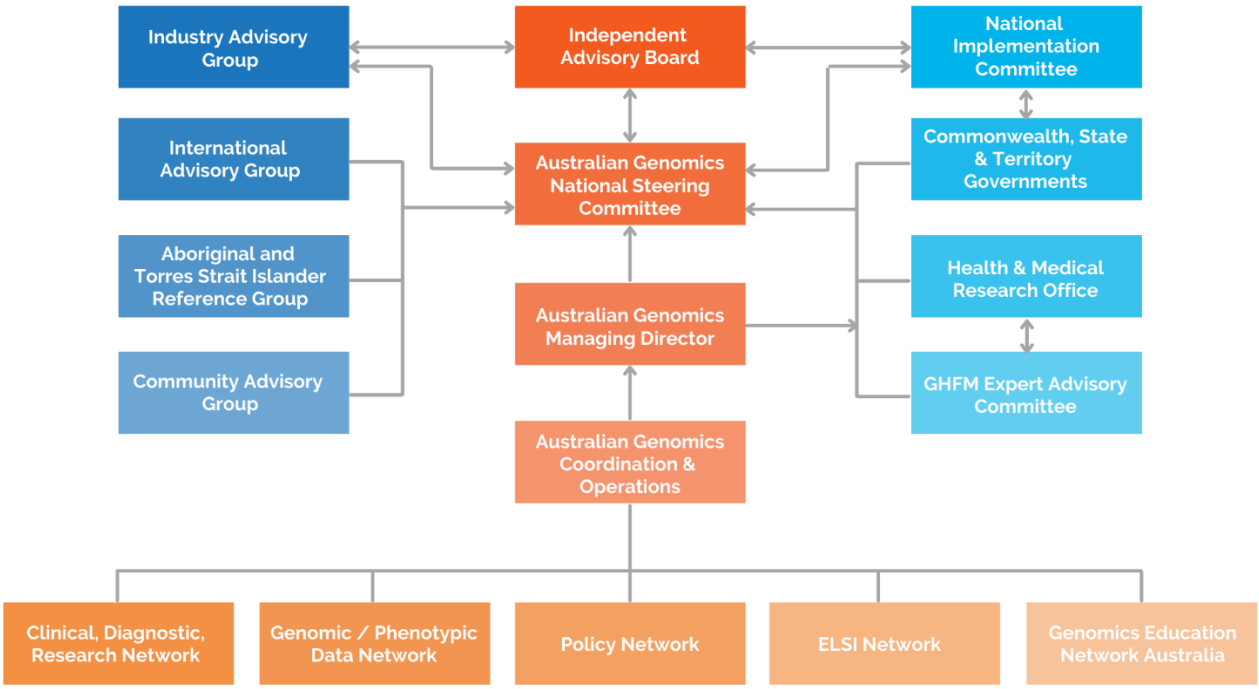
Leverage the network to advocate for and amplify messaging to inform genomics policy and practice and provide a "unified voice" to government, driving optimal policy intervention

Australian Genomics will strive to provide a voice for all states and territories, with engagement to enhance local research implementation and provide investigator support. As genomics is mainstreamed, Australian Genomics' focus on distilling research into policies informed and endorsed by experts can only continue with a higher base level of funding. Additionally, community involvement was highlighted by stakeholders as valuable to improve public awareness and genomic literacy as well as encouraging the number and quality of health-technology assessments for the reimbursement of genomic tests and services.

Key Partnerships / Collaborations: Local and International Genomic Initiatives

A collaborative 'hub' for the genomics community - continue to be global collaborators progressing standards in data interoperability, access and sharing. Consider opportunities to expand to adjacent industry bodies

A focus on external communications will be increasingly important to maximise communication of Government funded genomic activity and research outputs. Industry engagement management should also be an additional focus for Australian Genomics staffing profile as engaging InGeNa and industry bodies will become increasingly critical for better genomic uptake and implementation in healthcare. Retaining the international presence that Australian Genomics has worked to create will be determined by the funding base option available.



Governance and Legal Structure

Australian Genomics should look to transition to a company limited by guarantee, led by a CEO, independent of Murdoch Children's Research Institute. Australian Genomics can therefore attract philanthropic funding and partner with industry bodies. This will position Australian Genomics to transition to a national genomics healthcare initiative to support and drive translation and clinical implementation, policy development, data interoperability, and community, industry and international engagement and education. The organisation is best placed to meet new challenges and opportunities as an independent entity.

Authorisation and support from Federal Government is essential, alongside partnerships across state and territory jurisdictions. Therefore, the organisation should consider a reporting line to the Secretary of the Australian Government Department of Health and the Health CEOs. In order to achieve a level of accountability and scrutiny by Governments, but retain legal and financial separation from the Commonwealth, Australian Genomics could consider government body status as a commonwealth company with possible GBE status. This governing relationship would see Australian Genomics register as a company limited by guarantee under the Corporations Act, with additional requirements to meet appropriate standards of public sector accountability under Chapter 3 of the PGPA Act. Further discussion with Government is required to determine the optimal governance structure to ensure an appropriate balance of independence and funding predictability to support the future of genomics in Australia.

Australian Genomics' current advisory structure provides an exemplar for large research entities. Broad consultation through advisory and reference groups ensure voices from across Australia are elevated and heard. The advisory structure is key in the coordination of a national, multidisciplinary genomics network in Australia and globally. Any required changes will be factored into this structure upon administrative or legal advice.

Future Capabilities

As Australian Genomics transitions to an independent organisation and possibly adopts alternative funding mechanisms it requires different organisational capabilities for its future operating model. Australian Genomics should continue its focus on international presence and public affairs, and also retain its government relations expertise in grants applications, despite

not being grant funded, but to continue supporting partners in their applications for continued research funding. Australian Genomics currently supports GHFM-funded research programs that are close to completion and ready to be implemented into healthcare today. As it moves into the future, Australian Genomics additionally needs to mobilise its subject expertise to enable research translation to effectively drive the implementation of genomic medicine as standard of care.

Industry engagement and community outreach are current strengths of Australian Genomics, as seen through its network of collaborators and investigators. Moving into the future, Australian Genomics should look to deepen its capabilities in business-to-business engagement and eventually business-to-consumer engagement as genomics is mainstreamed. Moreover, as an independent not-for-profit organisation, Australian Genomics is directly accountable for its brand management. It should continue a focus on building its internal functions, people and culture, finance and risk management. Marketing capabilities in services pricing are also necessitated by alternative funding models, to ensure the organisation is responsive to stakeholder preferences and member willingness-to-pay.

Future State Forecast Australian Genomics Costs

Current State View

To determine a current state cost breakdown, Australian Genomics costs were broken down firstly by which activity they are attributable to across researching enablement, infrastructure, health translation and key partnerships / collaborations. Under the current NHMRC funding (2021 – 2023), Australian Genomics attributes the following costs to each key activity:

ACTIVITY	$ million p.a.
Research enabling activities and capabilities	1.21
Infrastructure and data	1.32
Health system translation	1.42
Management, partnerships, collaborations	1.05
TOTAL	**5.0**

The three cost areas of Australian Genomics are personnel, equipment and other direct costs, with personnel costs attributing to approximately 83% of its cost base.

Future Cost Base Options

The costing of three different scenarios for 2024 was modelled:

- **Option 1: $5M Funding Base** - Scale Back of Current Activities
- **Option 2: $7M Funding Base** – Maintenance of Activities
- **Option 3: $10M Funding Base** - Moderate Expansion of Activities

To determine requirements of each scenario, additional workforce costs were calculated across the four key activities and non-people costs were calculated as the required changes in operating and platform costs as a % increase to current state costs (see Appendix 6: Australian Genomics Funding Base Modelling for further detail).

Option 1: $5M Funding Base - Scale Back of Current Activities

In 2018, the Government's decade-long investment of $500 million in genomics research set a robust foundation for genomic healthcare to deliver better outcomes for Australians. Additionally, the Government has launched two 2021 medical research grant rounds for

genomics and cardiovascular research, worth $90 million. This demonstrates an ever-increasing focus from the Australian Government on accelerating Australian-led genomics. Moreover, alongside a growing quantum of genomics projects funded through GHFM, there is an increasing rate of projects engaging national consortia. As the scope, reach and impact of genomics research projects grow rapidly, the workforce requirements for Australian Genomics expands.

At a $5M funding base in 2024, the breadth of work that Australian Genomics could support in its "connector" role would be limited, with Australian Genomics having to reprioritise its efforts across the organisation purely into research project support with a translational research focus. Currently, Australian Genomics funding supports 18.1FTE in research support and partnerships/collaborations, partnering with 18 research projects with in-kind support totalling $10 million. With Genomics Health Futures Mission (GHFM) investing $500 million over 10 years in genomics research, the GHFM funding profile indicates that there will be at least 10 new projects per annum over the next 7 years, with an increased project lifespan and scope. Australian Genomics' current partnership uptake of 58% successful grants is additionally set to increase, as Australian Genomics has recently been included in GHFM Guidelines for entities to consider partnership. Based on these forecasts, Australian Genomics would require at the minimum an additional $1.7M beyond its current funding base to effectively support 31 concurrent projects in 2024, to enable the delivery of government funded genomic research outcomes into the future. Therefore, under a $5M funding base, activities relating to policy development, community engagement, maintaining an international presence for Australia and providing core data services would require alternative funding sources outside the base funding.

Option 2: $7M Funding Base – Continuation of Activities

At a $7M funding base in 2024, Australian Genomics would be adequately positioned to support genomics research as the GHFM funding profile grows, with additional FTE to enable stronger integration of collaboration, coordination and research tools and resources into the day-to-day operations of genomic research in Australia. The additional $1.7M required to effectively support 31 concurrent projects in 2024 would enable the delivery of government funded genomic research outcomes into the future. Additionally, funding provision extends support that Australian Genomics currently offers to GHFM-funded research programs that are close to completion and ready to be implemented into health care, to additional input and support beyond the life of the research grant are required to achieve this. A $7M funding base enables capability for Australian Genomics to progress research translation policy, providing a "unified voice" for states and territories and reducing complexity for Government.

Option 2 provides opportunities to continue community engagement and involvement activities and supports a more active role for Australian Genomics at the international genomics stage. Australian Genomics would also be positioned to fund core data services, leveraging its partnerships with various institutions to provide and receive advice on information management, support the interoperability of data and promote data sharing across the genomics landscape. Specific initiatives to realise the full potential of genomic healthcare could be funded through line-item funding for specific initiatives could additionally be agreed on a case-by-case basis. For example – to support the national implementation of a prototype for clinical and genomics information management.

Option 3: $10M Funding Base – Moderate Expansion of Activities

Option 3 extends the policy and research translation focus to distil research from across the network into identifying commercial opportunities and supporting research outputs in industry. Stronger engagement with InGeNA and industry bodies will lead to better genomic uptake and implementation, improving the efficiency of healthcare delivery as genomics is mainstreamed.

Alongside enhanced communications and community engagement, Australian Genomics can also solidify its role as an international leader on the global genomics stage. Under this funding base, Australian Genomics can leverage experts across the genomics and data community to support the development of scalable functional genomics platforms and progress and implement research outcomes addressing challenges in clinical and genomic datasets. Australian Genomics will be well-positioned to drive a national approach to enriching genomics databases, thus improving equity of access and public benefit derived from genomics in healthcare, as detailed in the NAGIM case study below.

VIGNETTE 2: NAGIM and data coordination ($10M funding model)

The Blueprint for a National Approach to Genomic Information Management (NAGIM) is a digital genomics blueprint developed for the Commonwealth Government. From evaluating the current jurisdictional, operational, and technical landscape in Australia, the NAGIM Blueprint concluded that a standards-based approach, using a federated model, would be the most appropriate strategy for a national approach to genomic information management in Australia. The Blueprint also included a proposed roadmap of activities, as relevant to infrastructure, genomic medicine, governance, and genomic research.

To understand genomic test results and make new genomic discoveries, researchers and clinical services rely on information generated by the combination of genomic data from many different people and produced at different organisations. For this to happen, data needs to be collected, processed and shared using common and standardised approaches.

Australian Genomics was tasked by the Australian Department of Health to develop recommendations for implementing the Blueprint and progressing the national approach to genomic information management for Australia. We are undertaking pilot projects to develop and encourage adoption of standardised, interoperable and extensible approaches to the collection, storage and use of genomic data in Australia.

National infrastructure stakeholders were invited to participate in prototype construction, addressing these priority areas, with the goal of identifying the best combination of components that can serve as the basis for long-term national infrastructure. The open call for prototype projects resulted in 12 submissions from key genomic infrastructure providers working together to leverage datasets. A community of practice has been established, both formal (monthly meetings with more than 40 attendees per meeting) and informal (Slack channel, with 54 members). Detailed key recommendations will be delivered to Governments in May 2022.

"This is an impressive activity and Australia should be proud to lead in this way and provide downstream benefits to its citizens. I can already say with certainty that the outcomes of NAGIM, including the assessment framework, will be used by organizations (and countries) in the future to guide their own work." Bob Freimuth, Mayo Clinic, US.

Required People Changes in each funding base option (2024)

	Option 1: $5M Funding Base	Option 2: $7M Funding Base	Option 3: $10M Funding Base
Research Enabling Activities	No Change (reprioritisation of effort)	+6 FTE Project Officers / State Coordinators for project support and local research implementation	+10 FTE (2 per state) Project Officers / State Coordinators for local research implementation + Senior Evaluation for Research Alignment
Health Translation	-2 FTE - Policy Project Officer Scaleback (+ reprioritisation of current FTE to Research Enabling Activities)	+1 FTE - Policy Project Officer + Clinical Implementation	+5 FTE Policy Project Officers + + Senior Evaluation - Research to Industry Outputs
Infrastructure	-2 FTE - Data standards officers / Engineer Scaleback (+ reprioritisation of current FTE to Research Enabling Activities)	+1 FTE Data Administrators to support data governance policy development etc.	+4 FTE Data Administrator and Assistants to support data governance interoperability + development
Key Partnership / Collaborations	-1 FTE - Comms Manager Scaleback (+ reprioritisation of current FTE to Research Enabling Activities)	+1 FTE addit. Comms Manager to maximise comms of Govt funded activity + community engagement	+5 FTE - 2 FTE to Comms Manager and Assistant + 2 FTE Industry engagement
Other	No Change	No Change	+2 FTE for Admin Costs of new Governance Model

Proposed Funding Model

An assessment of funding models in Section 6 determined that the optimal funding model for Australian Genomics to pursue is continuous annual appropriation, with additional alternative funding options of fee-for-service, subscription and project-specific funding remaining as options in future years. Revenue streams can be further diversified by pursuing project-specific partnerships to drive genomic research and translation, with industry (e.g. cell and gene therapies; targeted interventions) and philanthropy (consumer and patient-driven research priorities). Deeper analysis of the current cost base and future state funding requirements revealed three scenarios to fund Australian Genomics' future cost base in 2024:

1. Government Appropriation

The preferred model of government appropriation is $7M of core annual funding. Diversification of revenue will be sought with additional line item funding for specific Government initiatives which can be agreed on a case by case basis, and will also be pursued with industry, philanthropy, and other funding partners. Alternative revenue generating streams will be periodically re-assessed as genomics becomes a standard concept in Australian healthcare.

However, until this shift fully materialises, annual appropriation will be necessary to ensure the sustainability of genomics in Australia.

2. Subscription Model for Shariant

As detailed in Vignette 1: Shariant in Section 6, Australian Genomics partners with various institutions to maintain key infrastructure tools and support the maintenance of platforms including the Genomic Data Repository, Shariant and PanelApp. The continued management of Shariant in Australian Genomics' future role provides an opportunity to consider subscription options, however, as detailed in Section 5, its viability is heavily dependent on future actions to potentially mandate Shariant use in laboratories, as well as current willingness to pay from users. Moreover, at a funding base of Option 2: $7M, subscription revenue at $20,000 per annum would account for just 6% of Australian Genomics' 2024 costs:

Year	2024	2025	2026	2027	2028
$ Revenue from Subscription Model	$439,898	$461,892	$484,987	$509,236	$534,698
% of Funding from Subscription Model	6%	6%	6%	6%	6%

3. Fee-for-service for research project support

As detailed in Section 5, the collective in-kind value of research capabilities, tools and resources amounts to projects is $961,634 across a four-year project *(for additional detail please see Appendix 3: Australian Genomics support of genomic research projects)*. This can provide additional funding opportunities through reappropriating a proportion of research grant value from research grants to Australian Genomics. However, caution is advised for the method by which this fee could be charged. Reappropriating a portion of each funding grant presents significant challenges as the average GHFM research project grant size in 2020 was $2.73 million. This would mean that charging an annual in-kind value-based fee per project of $240,409 (annualised in-kind value) to new projects would reapportion approximately 9% of each project grant. Conversely, charging a more nominal fee-for-service (eg. $54,600 – 2% of average 2020 GHFM research grant) would generate less than $1 million based on the projection of 8 new projects funded and supported by Australian Genomics annually. Charging a fee-for-service may prove to be feasible if the fee is charged back to GHFM based on Australian Genomics research support uptake. However, "clipping the ticket" on grants would be prohibitive for the research institutes and universities conducting genomics research.

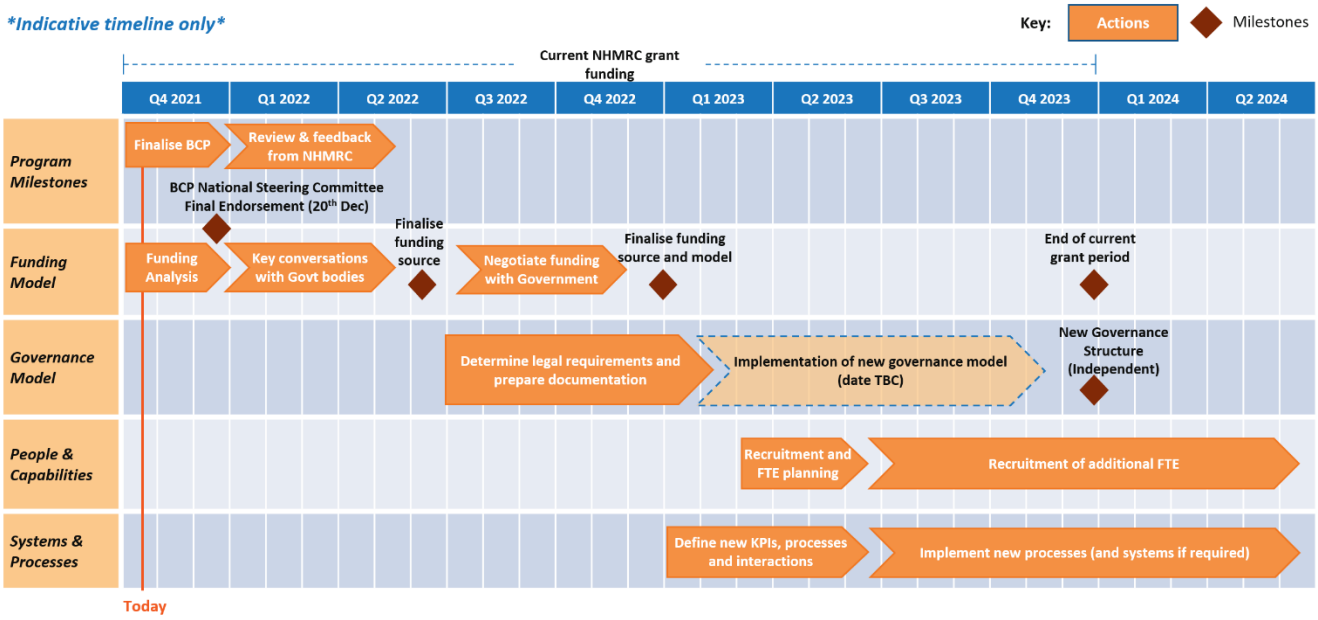
Conclusion

Australian Genomics' defined vision is to "foster, support and translate genomics innovation: from research to health outcomes, involving all Australians in sharing the benefits of health genomics." Its connector focus brings this to life by connecting stakeholders across the genomics ecosystem to support research translation, policy development, data interoperability, and community, industry and international engagement. Australian Genomics should transition to a company limited by guarantee, led by a CEO, continuing to leverage current advisory and reference groups, with specific governance arrangements to be discussed with Government. Additional capabilities required under Australian Genomics' future operating model include building business-to-business and potentially business-to-consumer engagement as genomics is mainstreamed. Deeper analysis of the current cost base and future state funding

requirements revealed three scenarios for Australian Genomics' future funding base in 2024, with the feasible scenario being $7 million of core annual funding.

8. IMPLEMENTATION ROADMAP AND TIMELINE

Implementation Roadmap



As the proposed future model is implemented, there will be required shifts in funding, governance, people and capabilities and systems and processes. The Business Continuity Plan is to be submitted to NHMRC and the Department of Health for approval in January 2022. Feedback will be reviewed and actioned over the first quarter of 2022, setting a foundation to engage in key funding conversations and gain funding and governance approvals from Government. Australian Genomics will aim to finalise its future funding model by 2023, negotiating potential options to transition to the new funding model in part before the NHMRC grant period concludes. Legal counsel will be engaged to identify the legal requirements of transitioning to a company limited by guarantee and determine the appropriate governance structure and government body relationship for Australian Genomics. Depending on whether Australian Genomics is confirmed as a Commonwealth company, legal counsel will need to confirm additional requirements for public sector accountability under Chapter 3 of the PGPA Act. Recruitment and FTE planning will be undertaken following the finalisation and approval of the proposed funding and structure. It should be noted that the process outlined in the above indicative roadmap could be shifted forward if funding negotiations are implemented earlier than anticipated and/or if government funding is approved to supplement Australian Genomics NHMRC grant funding before 2024. New processes and systems should be implemented once requirements of the new structure, staffing profile and funding are finalised.

Implementation Challenges

Effort required to transition to a new operating model may disrupt business and place strain on resources. It is suggested that Australian Genomics set up a working group that focuses on managing the transition whilst maintaining BAU role functions. The funding model provides costings and justifications for key priorities of driving research and data standardisation; supporting governments in translation and implementation; sustaining the multidisciplinary genomic community; and managing the implementation of genomic research into clinical practice. There is a risk that Australian Genomics priorities may change in the future, due to new policy positions or shifting immediate needs in genomics. Australian Genomics can mitigate the risk but drawing back to its vision to "foster, support and translate genomics innovation: from research to health outcomes, involving all Australians in sharing the benefits of health genomics". This places a focus on maximising patient benefit and promoting equity of access at the centre of any future prioritising decisions.

9. BENEFITS OF THE PROPOSED FUTURE FOR AUSTRALIAN GENOMICS

The ability of Australian Genomics to bring consensus to the complex genomics ecosystem and streamline perspectives is a significant competitive advantage at a national level. This will only become more advantageous as Australia's genomics network is mobilised to build the appropriate national infrastructure and ecosystem required to support future models of care. Australian Genomics is uniquely placed to bring together a diverse network of experts to drive excellence in genomics innovation and translation, working to improve the health and wellbeing of all Australians.

Convening an international expert network

Australian Genomics' network nationally and internationally has deep expertise and breadth, involving laboratories and clinical services; functional genomics networks; clinical recruitment sites and international organisations. Australian Genomics has a proven ability to convene expertise to drive implementation programs, which will become increasing critical to achieve key national health policy objectives, as recommended in the Zimmerman Report. Additionally, the international reach of Australian Genomics, exhibited through its leadership roles in international health consortia (GA4GH, Genomics and Health Implementation Forum) and active collaborations (inc. NHS England) will be instrumental in progressing Australian healthcare innovation. Such leadership positions are extremely advantageous for Australia reputationally and provide Australians with access to best practice genomic treatments, enriched through the development of frameworks for genomic data.

Driving future models of care

During this period of rapid innovation, Australian Genomics is becoming increasingly central to supporting future genomic models of care and analysis across care settings. Today, genomics innovation and translation spans across areas of healthcare including rare diseases, cancer, infectious disease, screening, pharmacogenomics and common diseases, with analysis tools including but not limited to bioinformatics, deep phenotyping, multiomics data, bio specimens and longitudinal data. The reach of genomics across the lifecycle continuum should also be noted, from genomic carrier screening, prenatal diagnostics, newborn genomic screening, child health, acute care, additional genomics findings to precision adult healthcare. The impact of genomics in transforming the lives of Australians is growing rapidly, and Australian Genomics is at the centre of this national shift, harnessing its network of over 450 collaborators nationally and internationally.

Ensuring research policy alignment

Australian Genomics' future role in connecting government, industry, researchers and beneficiaries of genomics healthcare will become increasingly critical as the genomics ecosystem is set to grow rapidly in Australia. In retaining the national momentum on genomics innovation, Australian Genomics provides opportunities to export expertise, stimulate investment and create smart jobs for Australia's economy. The Genomics Health Futures Mission (GHFM) is investing $500 million over 10 years in genomics research through MRFF to improve disease testing and diagnosis, innovate treatment options to improve health outcomes, and reduce unnecessary interventions and their associated health costs[23]. As announced in September, GHFM has up to $56 million available over two years from 2021–22 through six streams to support Australian medical research and medical innovation projects[24]. Australian Genomics is a critical enabler to support and uplift the efficiency and reach of research projects, with a central role in ensuring that the $500 million is utilised most effectively to drive genomics innovation into clinical practice. Moreover, Australian Genomics will be positioned to mobilise its national network to determine key research priorities in the genomics space, aligning priorities for greater impact; providing direction, frameworks and strategic planning.

Capitalising on future growth

The global genomics market is expected to reach USD 62.9billion by 2028 (19% compound annual growth rate, 2021 – 2030)[25,26]. As genomics transitions beyond diagnostic and clinical testing, into public health and prevention, industry involvement in the genomic value chain in Australia will increase. Australian Genomics provides a single point of contact for industry to access its national and coordinated network. It is also uniquely placed to leverage its State and Territory Government partnerships to support future service provision. Core investment from the Commonwealth Government will enable Australian Genomics to leverage its networks' capabilities, look to industry for partnerships, and respond to new technological advances and opportunities as they arise. Additionally, as the genomics landscape matures, Australian Genomics will strive to employ alternative revenue-generating opportunities (i.e. fee-for-service, philanthropy, membership fees) to increase its financial independence in a manner that is not prohibitive for organisations in the genomics value chain. By positioning Australian Genomics as a national genomics healthcare initiative, it can drive research translation and clinical implementation, promote and enhance data interoperability and support industry engagement and policy development to drive future models of care. This enables Australia to realise the full potential of genomic healthcare for all Australians on a sustainable basis.

Counterfactual

Without substantial, consistent and ongoing funding, the future of genomics healthcare in Australia is placed at risk. The potential consequences are dire: an absence of collaboration and data interoperability; diminished support for critical GHFM projects; a loss of connectedness between both researchers and Government as well as Australia's international partnerships; decreased translation into clinical care; and diminishment of patient outcomes. Additionally, without a demonstrated commitment to Australian Genomics, access to genomics would remain only through ad hoc research efforts, with reduced equitability for all Australians.

Considerable risks would arise for numerous stakeholders if Australian Genomics did not

[23] Medical Research Future Fund - Genomics Health Futures Mission Roadmap
[24] The Hon. Greg Hunt MP, Minister for Health and Aged Care - MEDIA RELEASE - $90 million to help Australians live longer and healthier lives, 28 September 2021
[25] Next Generation Sequencing Market, Allied Market Research, July 2021
[26] Research and Markets, https://www.researchandmarkets.com/research/7wb4qz/genomics_market?w=5

receive sufficient and ongoing funding and was therefore unable to continue its work to date. An inability to effectively translate genomic innovation to improve Australian health and wellbeing would gravely diminish the safety, timeliness, economic viability and reputation of Australia's healthcare system. Without a body to facilitate and streamline the sharing of genomic data, incorrect diagnoses and inequitable access to treatments would place additional strain on the Australian healthcare system. The burden of inefficient and unsafe practices to Government cannot be understated, as a lack of commitment to healthcare innovation would elicit significant backlash from the public.

Additionally, the counterfactual to a future with Australian Genomics impacts patient outcomes, research capability and future data management:

Research capability

Currently, Australian Genomics enables connected research efforts leading to aligned diagnostic research across Australia. Notably, Australian Genomics supports GHFM-funded research programs that are close to completion and ready to be implemented into health care. However, additional input and support beyond the life of the research grant are required to achieve this. Without the continued efforts of Australian Genomics, the clinical outcomes and future patient benefits of today's research projects could be diminished or lost completely. Australian Genomics is uniquely positioned to coordinate a world-class national genomics network. Since 2016, its network has grown to include 103 organisational partners, 450 collaborators and investigators, including over 15,000 research participants engaged in 21 clinical studies and 23 program sub projects. Australian Genomics has continually supported its network, notably completing 80 contracts and 115 ethics and site submissions per year. Without adequate funding Australia would face the loss of a national network with a potential cohort of 25 million people. Hence, research conducted across the country would be fragmented, leading to duplication of diagnostic research efforts.

JOSHUA

Acute Care Flagship

Joshua Stirling was a healthy baby at six months old when he had started having multiple seizures. With no accurate diagnosis, a lumbar puncture was being considered as the next option. After his seizures became longer and more intense, Joshua was transferred to Intensive Care at Monash Children's Hospital. After discussions with his parents, Melissa and Matthew, the head of Paediatric Neurology, A/Prof Michael Fahey referred Joshua to the Acute Care study, led by Australian Genomics - offering rapid genomic testing – providing test results in less than five days for children in neonatal and paediatric intensive care units. A genomic test typically takes three to four months.

It took Joshua's parents just three days to find out their son has Dravet syndrome, a rare genetic epileptic encephalopathy, caused by a change in the SCN1A gene. The implications of rapid screening in ICU are many: a timely diagnosis means doctors can better manage a condition; a diagnosis means no more invasive tests.

For Joshua's parents, finding out their son has Dravet syndrome was life changing, they immediately began looking at targeted treatment options and could avoid certain foods, certain medications, and triggers such as bright light, noisy environments and overstimulation that made his condition worse. They made lifestyle decisions, Melissa gave up part-time work – and accessed support networks. It meant they could stop speculating about the cause, which in Joshua's case turned out to be genetic, and focus instead on managing his condition.

Genomics Data

Australian Genomics has a critical role in supporting national data interoperability for both clinical and laboratory testing. Currently, Australian Genomics is developing the prototype for a National Approach to Genomic Information Management (a national legacy dataset of clinical and genomic data for clinical care and research). Through this work, Australian Genomics envisions a future for an Australian Genomic Data Asset, which will only be possible through sustained government funding. It has also achieved a national system for sharing genetic evidence between diagnostic laboratories, that needs to be upheld and expanded to accommodate for future growth of the genomics ecosystem in Australia and across the world. Australian Genomics' unique position to lead a national response to genomics innovation is central to supporting the Government commitment (through GHFM funding profile) to improve disease testing and diagnosis, innovate treatment options to improve health outcomes, and reduce unnecessary interventions and their associated health costs. Inadequate funding will lead to a loss of data interoperability for both clinical and laboratory testing as well as a loss of data sharing in Australia and internationally.

Patient safety and access to treatments

Today Australian Genomics enables patient access to the latest genomics insights / treatments available from across the globe. It also provides standardised protocols enhancing patient experience and equity of access across Australia and representation for Australia as a key player and leader on the global genomics stage. Without adequate funding, patients risk slower and inadequate access to international genomics insights as well as available treatments. Without collaborative clinical and research networks to catalyse high quality research and translation, Australians would face a lack of standardised protocols diminishing

patient access, experience or worsening clinical outcomes. The implementation of genomic medicine as standard of care depends heavily on the national sharing of genomics research outcomes. Hence, the safety and quality of healthcare is placed at risk, with incorrect diagnoses and lack of access to treatments are a likely reality without a national Australian genomics healthcare initiative. Additionally, Australia would lose its 'leadership position' on the genomics international stage. Comparatively, the fellow 32 members of the Genomics and Health Implementation Forum (including Japan, Singapore, USA, Denmark, Germany, Switzerland) have developed (or are developing) government funded programs for the translation of evidence-based policy in genomics into standard of care. These programs have overarching coordination to drive partnerships across jurisdictions and with industry and are underpinned by national infrastructure for the management of clinical and genomic data. Without a national entity to drive genomic translation, the global genomics leadership positions Australian Genomics holds and the benefits these provide to patients across Australia will be lost.

Conclusion

As Australian Genomics engages community, industry and international partners to support and drive genomics translation and clinical implementation, policy development, data interoperability, the full potential of genomic healthcare can be sustainably realised for all Australians. The importance of Australian Genomics' sustainability lies in its increasingly critical role in ensuring the safety, quality and equity of healthcare access in Australia. Now is the time for the Australian Government to position Australian Genomics as a national Genomics Healthcare Initiative to drive the implementation of genomic medicine as standard of care.

10. APPENDIX

The Appendix includes the following documents:

- Appendix 1: The Health Genomics Landscape
- Appendix 2: Consultation Responses
- Appendix 3: Australian Genomics Support of Government-Funded Genomic Research Projects
- Appendix 4: Exemplar Organisations
- Appendix 5: Australian Genomics Funding Source Modelling
- Appendix 6: Australian Genomics Funding Base Modelling
- Appendix 7: Detailed Analysis of Alternative Roles Proposed for Australian Genomics

10.1 APPENDIX 1: THE HEALTH GENOMICS LANDSCAPE

The International Genomic Landscape

The momentum of health genomics implementation globally is extraordinary. Since the international landscape analysis conducted by Australian Genomics in 2019[27], the number of countries that have founded **national genomic initiatives** to drive the implementation of genomic medicine into healthcare has almost tripled. A recent systematic review has identified 41 active national genomic initiatives globally, and a further 86 countries have genomic projects of a targeted scope[28]. These initiatives progress understanding of normal genomic variation, with 32 active population genomic sequencing initiatives[29]. They are also assembling clinical cohorts to unlock genetic mechanisms of disease: building diagnostic capability in rare disease and cancer has been a strategic priority of 21 of the 24 Global Alliance for Genomics and Health (GA4GH) Driver projects[30], and every European Union country has a rare disease plan[31].

Developing infrastructure is also a key priority of national genomic initiatives, including standards and platforms for the collection, storage and sharing of data; and resources for workforce and public genomic education.

The majority of national genomic initiatives are either public funded, or public-private partnerships, with the scale of funding ranging from <$1M USD (Slovenia) to $9.2B USD (China)[4].

Figure – Exemplar National Genomic Initiatives: Scale, Funding and Priorities

(1) Derived from Kovanda et al (2021)[4]
(2) Australian Genomics data includes directly administered projects
(3) PPP: public-private partnership with pharma, insurers or biotech

Country	WES/WGS	Funding type	$USD M	Population cohorts	Clinical cohorts	Education	Infrastructure	Data Sharing protocols	Genomic data standards	Data Management	Clinical data linkage
Australia (1)	25,000	Public	55	✓	✓	✓	✓	✓	✓	✓	✓
Canada	130,000	PPP (2)	3200	✓	✓	✓	✓	✓	✓	✓	✓
China	100,000,000	PPP	9200	✓	✓	×	✓	✓	×	×	✓
Cyprus	1,000	Public	38	✓	✓	✓	✓	✓	✓	✓	✓
Denmark	1,650	PPP	15	✓	✓	×	✓	✓	×	✓	✓
England	100,000	Public	437	✓	✓	✓	✓	✓	×	✓	✓
Estonia	430,000	Public	6	✓	×	×	✓	✓	✓	✓	✓
Finland	10,000	PPP	59	✓	✓	✓	✓	✓	✓	✓	✓
France	235,000	Public	799	✓	✓	✓	✓	✓	✓	✓	✓
Hong Kong	20,000	Public	1200	✓	✓	×	✓	✓	✓	✓	✓
Japan	250,000	Public	90	✓	✓	✓	✓	✓	×	✓	✓
New Zealand	600	Public	5	✓	✓	✓	✓	✓	×	✓	✓
Poland	5,000	Public	25	✓	×	✓	×	×	✓	×	×
Saudi Arabia	100,000	PPP	80	✓	✓	✓	✓	✓	✓	×	✓
Singapore	10,000	PPP	150	✓	×	×	×	✓	×	✓	✓
South Korea	10,000	Public	15	✓	✓	×	×	✓	×	×	✓
Switzerland	N/A	Public	72	×	×	✓	✓	✓	✓	✓	✓
Thailand	10,000	Public	141	✓	✓	×	✓	✓	✓	✓	✓
USA	1,000,000	PPP	927	✓	✓	×	✓	✓	✓	✓	✓

[27] Stark, Z. et al. AJHG 104(1) 13-20 (2019). https://doi.org/10.1016/j.ajhg.2018.11.014
[28] Kovanda, A. et al; Human Genetics (2021) doi.org/10.1186/s40246-021-00315-6
[29] Frost & Sullivan; Strategic Insights for Global Health Research Data Platforms, July 2019. CSIRO
[30] https://www.ga4gh.org/how-we-work/driver-projects/
[31] Annual Report of the Chief Medical Officer 2017, 'Generation Genome'. www.gov.uk

Genomics England[32] is among the most mature of the national genomic initiatives. Established in 2013 by the Department of Health and Social Care in the UK, it completed the 100,000 Genomes Project underpinning England's national genomics effort. This project established key infrastructure for the delivery of genomics as part of mainstream healthcare through the **Genomics Medicine Pillar of the NHS**, which delivers screening, acute care (ICU) genomics, rare disease diagnosis, pharmacogenomics and cancer diagnosis and surveillance. The Genomics Medicine Pillar aims to deliver patient benefit; enable new scientific insights and discoveries; accelerate the uptake of genomic medicine; stimulate UK industry and investment; and increase public knowledge and support for genomics. This substantive, sustained government investment and support has driven the integration of genomic medicine into the NHS, with the ambition to sequence the genomes of 500,000 patients within healthcare in the next 5 years.

The Global Alliance for Genomics and Health (**GA4GH**) was established in 2013 as an international collaborative of stakeholders in genomics, a community brought together to drive international standard setting through the development of frameworks for genomic data sharing. The GA4GH's mission is to share approaches, develop guidelines and standards, and through this collaborative approach avoid duplication of effort. By 2021, the GA4GH has achieved broad global reach, currently citing 666 member organisations and 3000 members from over 90 countries[33]. The 32 members of the **Genomics in Health Implementation Forum** have developed (or are developing) government funded programs for the translation of evidence-based policy in genomics into standard of care in their healthcare systems. These national programs, which include Japan, Singapore, US, Denmark, Germany, and Switzerland, have overarching coordination to drive partnerships across jurisdictions and with industry and are underpinned by national infrastructure for the management of clinical and genomic data.

Health Genomics Research in Australia

Australia's **National Health and Medical Research Council** (NHMRC) and **Australian Research Council** (ARC) have together awarded an estimated $338M toward genetic / genomic-related research* through competitive grant schemes since 2013 (around $292M and $46M, respectively)[34]. Notably, almost half of this funding was awarded in the past 2.5 years, with $157M allocated since 2019.

The Australian Government's **Medical Research Future Fund** (MRFF) has funded $211M in genetic / genomic projects since 2017, of which $149M was allocated from the $500M **Genomics Health Futures Mission** across identified priorities including rare disease, cancer, carrier screening, acute care genomics, and ethical, legal and social issues (ELSI). Other Federal Government investments include the Zero Childhood Cancer Program ($20M), the Australian Cancer Genomic Medicine Program (ACGMP, $50M) and a $20M Federal investment toward cancer research announced directed to Western Australia.

Over the past decade, several State/Territory genomics research initiatives have also been launched.

The **Sydney Genomics Collaborative** was funded by the NSW Government (2014-2018, $24M), to enable successfully funded clinical flagship projects to access the sequencing services of the Garvan Institute of Medical Research. It also encompasses the Australian Genomic Cancer

[32] https://www.genomicsengland.co.uk/
[33] https://www.ga4gh.org
* Publicly available grant outcomes were searched for theme, keyword, title and summary mentions of the terms 'genetics' and 'genomics'. Epigenetics was not included. Projects were considered individually for relevance and inclusion.
[34] https://www.nhmrc.gov.au; https://www.arc.gov.au

Medicine Program, which is applying genomics to the detection, prevention and management of cancer and has a national reach through Omico[35].

The **Melbourne Genomics Health Alliance** was formed in 2013 with an initial demonstration project (2014-2015), after which a further $25M was allocated by the Victorian State Government (2016-2019) and $10M contributed by 10 organisational partners. This phase conducted 11 clinical projects evaluating the diagnostic and clinical efficacy of genomic testing. Concurrent studies showed strong patient support of genomic testing; upskilled more than 1500 health professionals; and advanced integrated data analytics and management capability through the GenoVic platform. In late 2020, Melbourne Genomics received an additional $35M in Victorian Government funding, with an additional $10M committed by Alliance partners, to progress a further four-year program of work[36].

The **Queensland Genomics Health Alliance** (QGHA) was established in 2016 with $25M funding from the Queensland Government over 5 years. QGHA established a program with translational and implementation focus: clinical demonstration projects evaluating genomic sequencing in the diagnosis and management of specific diseases; and capability projects included workforce development, testing innovation, evaluation and ELSI research. Queensland Genomics concluded in June 2021, with products of the program being embedded in Queensland Health service provision and infrastructure, including the Genomics Institute in Metro North Health[37].

Canberra Clinical Genomics (CCG) was supported by a $7.3M investment from the ACT Government. In 2019, publicly funded, NATA accredited clinical genomic testing became available through CCG, for the approximate 300 patients in Canberra who need it every year to diagnose rare disease.

The **South Australian Genomics Health Alliance** is currently an unfunded network, but operates in an environment where clinically ordered genomic tests have been funded by the State government. The Alliance worked with State government on a Clinical Genomics Plan[38], and the South Australian government committed resources to public health and microbial genomics in 2018.

Despite the substantial Government investment in health genomic research, it is well documented that the path to translation of clinical research is long, and inefficient. Approximately 85% of biomedical research investment is wasted: with only 14% innovations entering clinical practice, and 1% to important health impacts[39]. To capitalise on Australia's genomic research investment, and expertise, a strategic and sustained platform to support translation into healthcare is required, particularly given the complexities of the Australian health policy and practice landscape.

Genomics policy and practice in Australia

The Australian Government Department of Health's **National Health Genomics Policy Framework** (NHGPF) was published in 2018[40], describing six priority areas: a person-centred approach; developing a skilled genomics workforce; service provision; capability, sustainable financing and responsible management of genomic data. The Project Reference Group on Health Genomics (PRG) was established with jurisdictional membership to oversee delivery of State and Territory participation in the NHGPF Implementation Plan[41]. With the dissolution of the

[35] https://sgc.garvan.org.au/initiatives
[36] Gaff et al; Genomic Med (2017) doi:10.1038/s41525-017-0017-4
[37] https://queenslandgenomics.org
[38] South Australian Clinical Genomics Plan 2022; https://www.sahealth.sa.gov.au
[39] Chalmers I, Glasziou P. Avoidable waste in the production and reporting of research evidence. *The Lancet* 2009;374(9683):86-89.
[40] National Health Genomics Policy Framework, 2018; ISBN 978-1-76007-328-2
[41] Implementation Plan - National Health Genomics Policy Framework, 2018; ISBN 978-1-76007-363

AHMAC under the National Cabinet to address the COVID pandemic, and subsequent rationalisation of Government committees, the PRG was disbanded in 2020. As the **States and Territories** are responsible for health service delivery, most have independently developed their own genomic strategies. As health genomic technologies become mainstreamed, genomic priorities will also be reflected in broader Government strategies and policies, including the **National Preventative Health Strategy** (2021), and **Primary Health Reform** (2021). Further, the implementation of genomics in healthcare particularly intersects with the activity of the **Australian Digital Health Agency** (ADHA), which has responsibility for the strategic management and governance of the National Digital Health Strategy and the design, delivery and operations of the national digital healthcare system including My Health Record.

The **funding of clinical genomic testing** in Australia is currently fragmented. Most aspects of clinical genomic service provision are funded by tertiary clinical genetics services, via State funding, block funding, Activity Based Funding – or paid by the patient themselves. There is also a lack of clarity and jurisdictional consistency with respect to funding of genetics services which results in diminished service capacity, inequity of patient access, and insufficient budget for genomic interventions – even though genomics has been demonstrated to drive system-wide efficiencies in both clinical benefit and cost effectiveness[42]. The Medical Services Advisory Committee (MSAC) is receiving an increasing number of applications related to medical services utilising genomic technologies since the consideration of public funding for genetic testing for hereditary mutations predisposing to breast and/or ovarian cancer in 2015. Notably, the first true 'genomic' application recommended for approval by MSAC was the Childhood Syndromes submission progressed by Australian Genomics, which was made available for public funding from May 2020. MSAC applications for Expanded Genetic Carrier Screening (based on Mackenzie's Mission) and Acute Care Genomics are currently in process.

The **demand for clinical genomic testing** in Australia is growing rapidly. The Stocktake conducted by the Royal College of Pathologists in 2017 reported 1,700 genomic tests (whole exome or genome sequencing) conducted in the 2016/17 financial year[43]. By mid 2019, public accredited pathology services reported a volume of 4000 genomic tests annually, and growing. Clinical genetic services across the country have also reported a 50-60% increase in demand in the past five years[18].

Industry players in the genomic value chain in Australia span from medtech/device companies, biotech infrastructure or reagent organisations, through to pharmaceutical companies and private pathology providers – many of which are large multinationals, with a local presence. Until recently, industry involvement with the genomics ecosystem was fragmented, however the Industry Genomic Network Alliance (InGeNA[44]) has brought together 18 companies to date, to progress collaborative priorities and engage with research organisations, policy makers and consumers.

[42] Australian Genomics response to the Independent Hospital Pricing Authority (IHPA) Consultation Paper on the Pricing Framework for Australian Public Hospital Services 2022-23
[43] https://www1.health.gov.au/internet/main/publishing.nsf/Content/national-health-genomics-stocktake
[44] https://ingena.org.au

10.2 APPENDIX 2: CONSULTATION RESPONSES

Responses were received by individuals employed by the following:

- Medical Research Institutes (32%),
- Universities (21%),
- Government (21%),
- Professional societies (16%), and
- Clinical providers (10%).

Stakeholders were invited to complete a survey to answer the following questions:

- What is the core value proposition that you see Australian Genomics delivering in the future to enable Australia to be a global genomics leader?
- Considering the current scope of activities (enabling research; data management, governance and infrastructure; health system translation; and facilitating key partnerships and collaborations), what priorities should Australian Genomics focus on moving forward?
- What exemplar organisation features can provide fresh thinking and perspectives to the review and future direction of Australian Genomics?
- What components of one or more of the three roles do you see as critical to ensure Australian Genomics' long-term value and sustainability?
- What funding mechanisms are feasible and appropriate to enable Australian Genomics to thrive? (e.g. membership model, fee for service, govt appropriation, or some combination).

Current State of Australian Genomics - Core Value Proposition

Stakeholders emphasised Australian Genomics' proven track record of coordinating a strong network and bringing together viewpoints to advise as a core voice on priorities, hurdles and standards. Australian Genomics was referred to as "a common and central connection point" for genomics healthcare. It can provide capacity and capabilities to assist the coordinated implementation of genomics as the future of healthcare looks to research driven clinical care. Stakeholders reflected upon the power of merging efforts of bodies such as "NPAAC, NATA, RCPA and HGSA...to all be talking the same language with respect to genomics implementation". Additionally, providing a single point of contact for international linkages to disseminate information to appropriate stakeholders was deemed to be of great value.

There was wide acknowledgement of the downstream benefits to patients and healthcare that are realised through Australian Genomics' work to promote greater and more equitable access to genomic medicine. The coordination of an equitable approach to genomics in healthcare was highlighted as critical due to the disparity between states and territories in access to clinical and pathology services, as well as policy approaches. Australian Genomics' unique national approach can be employed to engage with all jurisdictions and provide a voice for all states and territories (for example, leveraging learnings from Queensland's standardised genomics sequencing and single e-health medical record to push for a national approach). Engagement with the public in a broader advocacy role was highlighted as valuable to improve public awareness and genomic literacy as well as encouraging the number and quality of health-technology assessments for the reimbursement of genomic tests and services.

Possible Roles and Exemplar Organisations

Priority Areas

Stakeholders stressed that the value proposition of Australian Genomics needs to be more clearly defined to the broader genomic community – and potentially the interdisciplinary communities that increasingly leverage genomics and genomics expertise. The key priorities

posited across the respondents were government advisory, data management and continuing to foster partnerships and collaboration across the network to drive more efficient and effective health system translation. Moreover, the focus on research implementation into healthcare was flagged as essential to then inform and drive other activities, with a focus on research performed specifically for translation into the clinical space (eg. to generate MSAC applications). An emphasis on greater health system translation necessitates regular connection and meaningful engagement across sectors with experts and partners, including clinical trial groups, research, public and private health service providers, industry and other organisations. The prioritisation of supporting the collection, analysis and communication of clinical and research data can enable progress to be effectively measured and unmet needs to be realised.

Stakeholders stressed the value in leveraging Australian Genomics' positioning to garner support from State Health Departments to create clinical and molecular workforce capacity. Particular stakeholders prioritised optimising synergies between discovery and clinical genomics, given the risk to clinical implementation arising from gaps between jurisdictional and national funding for different aspects of a national genomics strategy. It was stressed that "disjunction between research funding and governance of clinical services is a perpetual threat to the overall genomics effort". It was also advised that Australian Genomics clearly define any "peak advisory" role to avoid duplicating health departments and agency responsibilities or creating conflicts of interest. The advisory role was suggested to focus on supporting health departments in communicating and translating genomic research and outcomes and informing policy development and implementation as reflected in the aims of the National Implementation Committee. Data management and interoperability across all jurisdictions was widely acknowledged as a critical priority, supporting "one way to store and share data". Stakeholders emphasised the need to provide support towards developing models that reduce clinical/research boundaries but maintain appropriate controls including dynamic patient consent.

Australian Genomics Roles

A majority of stakeholders articulated how the "connector" would be optimal to drive a genuine national approach with optimal policy intervention, placing a continued focus on enabling research, data management and health system translation advisory. Stakeholders acknowledged the profound impact of having a "unified voice" in the health translation space, highlighting its criticality as "there is too much risk of scattered pockets of research that Australian Genomics can bring together". This positioning also emphasised the value in facilitating greater cooperation to drive uniform infrastructure and data interoperability nationwide. By strengthening its "connector" role across the genomics space, stakeholders flagged how Australian Genomics can also equitably disseminate tools and resources, including consent materials, education materials and surveys to support more efficient research ethics and governance. Australian Genomics was instructed to consider how its government advisory role would work with and align to existing advisory bodies such as HGSA, RCPA and NPAAC, as well as allow space for partners to provide their own perspectives on policy development and implementation. Caution was advised to 'relating solely to genomics', suggesting that roles look to the effective mainstreaming of genomics into routine healthcare delivery and how alignment with existing bodies can progress this.

Stakeholders emphasised the value and expertise of Australian Genomics with components of the standards provider and endorser role to drive data interoperability and consistency across areas including ethics and consent. However, the standards endorser role was critiqued as an additional burden for clinical diagnostic labs if not done alongside NATA accreditation. Particular stakeholders flagged that Australian Genomics would be better placed to bring together existing bodies to act as a peak advisory body with a unified voice, rather than "acting as another standards endorser". However, stakeholders prioritised data management in

supporting and driving ongoing efforts to arrive at standards for data management, variant calling, data storage, and data access. Stakeholders flagged that this could be achieved "through modification of existing accreditation procedures for sequencing laboratories" once standards have been agreed. Overall, there was also consistent messaging for Australian Genomics to continue upholding local and global collaboration initiatives and work to connect healthcare ecosystems nationally and globally, regardless of its future role.

Exemplar Organisations:
Respondents looked to exemplar organisations utilising the 'connector' role - including Genomics England and Astronomy Australia. Genomics England's integrated system with NHS, state pathology service and consistent Health Electronic record system was flagged as achieving strong connection of healthcare ecosystems, an aspiration for Australia's federated healthcare system. Genomics England and Genome British Columbia reflect NAGIM's goal to move as many aspects of genomics standards around data sharing and secondary research for clinical benefit to the Federal level for better equity of access and implementation. Additionally, Astronomy Australia Limited's model of co-investment was flagged as an appealing way to build a sustainable funding model. Stakeholders also looked to building closer partnerships with other prominent organisations, with specific recommendations to align and develop formal lines of communication/consultation with HGSA. Specific feedback to highlight includes a suggestion to focus on transferrable elements when designing and building infrastructure such as big data capability which could incorporate proteomic data as easily as genomic data. One respondent suggested focusing on exemplar organisation features such as peer networking and member benefits through voluntary membership by research institutes and health and medical researchers. Another respondent recommended looking at aspects of how these organisations work internally, and how they make their decisions, including values, diversity, workplace culture, and the impact these factors have on their success and sustainability.

Funding Mechanisms

There was general consensus that grant funding is not viable, and sustainable government funding is vital for current infrastructure to continue, with some stakeholders referencing both Federal funding and state representation. It was noted that the "connector" role specifically lends itself to government funding. Federal activity-based funding linked to clinical services was posited. One stakeholder highlighted national funding support for national sequencing strategy, biobanking effort, MRFF funding for discovery and jurisdictional funding of medical record integration with commonwealth support for establishment of standards. Respondents promoted the exploration of a combination of funding models (that still protect and maintain individuals rights over data). A funding combination was additionally suggested to be protective against accusations of bias, and can improve stakeholder engagement.

Concerns were expressed with particular commercial funding models, articulating that change from a "free service" to a membership model particularly for services required in clinical settings (eg. Shariant) may be disruptive and impact upon equitable access to genomic treatments. An industry type model whereby a fee is charged for access to resources or to remain a member/partner of an alliance may be prohibitive for many organisations. Other precautions included monetising research ethics advice, how to appropriate "tier" memberships, and the maintenance of public trust with any level of industry involvement. Particular stakeholders also called out the government bodies who already act as accrediting bodies, standards development and standards accreditation and flagged that additional licences / fees may discourage industry engagement and investment in both R&D and clinical settings in Australia. Nevertheless, numerous respondents posited a fee-for-service model as being viable with careful consideration of risks. It was suggested that a fee-for-service be paid by government, with a translational / development fund for more research focused genomics that could have

multiple funding sources. The timing of such funding arrangements was considered, with a particular stakeholder noting that “it feels too early to expect anything other than government funding”. Another respondent flagged that while it is not on the horizon currently, it may be appropriate in the long term that the government appoint a national genomics agency with associated funding, and relevant strategy and performance framework. Overall, all respondents emphasised the value that Australian Genomics provides and the criticality of its sustainability. Acknowledging the expanding scope of genomic healthcare, respondents saw the significance of Australian Genomics’ role only increasing into the future, as genomics transitions beyond diagnostic and clinical testing, into public health and prevention.

SALLY

Acute Care Flagship

When Sally* arrived, my husband and I were overjoyed. We had educated ourselves about breastfeeding. but it didn't happen. The midwife hand expressed the colostrum for me and we fed it to Sally in a syringe - then she turned blue. This was the first sign there was something seriously wrong - Sally was transferred to the Royal Children's Hospital NICU. The whirlwind began. Test after test after test. The many medical teams she saw could not work out what was wrong. A big concern was the fact that she was floppy.

The doctor told us they were looking for neurological conditions. I was floored, grief stricken, and panicked. They wanted to do some genetic testing, which would take up to six weeks to come back. We were lucky to be offered more extensive genetic testing, called exome sequencing, through a research project being run by Dr Zornitza Stark and Sebastian Lunke from the genetics team at Australian Genomics and the Victorian Clinical Genetics Services. The results came very quickly – within three days.

At five-weeks-old Sally was diagnosed with a genetic condition, a myopathy, which meant her muscles had not formed correctly. This explained her floppiness and poor feeding. It was with both relief and sadness that we learnt of her diagnosis. . Since we now know her condition is inherited – we will need to have IVF and pre-implantation genetic diagnosis (PGD) to avoid having another baby with the same condition. Having exome sequencing has been a life-changing decision. What the diagnosis offered access to early intervention since she was a newborn and is doing remarkably well.

* Name has been changed. Image by Pexels

10.3 APPENDIX 3: AUSTRALIAN GENOMICS SUPPORT OF GOVERNMENT-FUNDED GENOMICS RESEARCH PROJECTS (2021)

Australian Genomics is a national research collaboration launched in 2016 to build the evidence and inform policy for the integration of genomics into Australian healthcare. Building upon the core research capabilities and infrastructure developed through the first phase of Australian Genomics (2016-2020), the network has been funded by the Australian Government through the NHMRC (GNT2000001) to support government-funded genomic research nationally, and so improve research efficiency, effectiveness and impact.

Australian Genomics also progresses government priorities by informing policy development and health system implementation: bringing together clinicians, researchers, diagnosticians, bioinformaticians, policy makers, and consumers who are united in the aim of equitable, appropriate application of genomics in healthcare.

Australian Genomics can offer the following capabilities in support of projects:

- Support of the Australian Genomics national coordination network of project officers, genetic counsellors and data administrators nationally to facilitate research implementation and activity in each state and territory;
- Guidance and support in the development and submission of human research ethics applications for the project and attainment of site-specific governance;
- Study outreach and advocacy through the activities of the coordination network and the national multidisciplinary collaboration, to improve project awareness and uptake through clinical and research institutions;
- Support the project in adaptation of a suite of established participant materials and resources for research consent, education, information and research evaluation;
- Advise on the standardisation and optimisation of research data capture and the development of study databases, including access to technicians to customise REDCap and alternate data capture instruments;
- Access to the online research consent and engagement platform CTRL ('control') for adaptation and application to the project;
- Guidance on genomic data management and standardisation, and data governance policies and procedures, to support secondary research and efficiency in data discovery and sharing in alignment with international standards and best practice;
- Advise on national genomic research operations, and specialist evaluation (health economics, implementation science), through our expert operational personnel and academics;
- Opportunities for input into research strategies and co-design of materials from partner patient support and advocacy groups, via the Community Advisory Group;
- Australian Genomics will also support the project in the translation of project outcomes into policy and practice.

The collective in-kind value of these research capabilities, tools and resources amounts to $240,410 per annum, per project.

10.4 APPENDIX 4: AUSTRALIAN GENOMICS ANALYSIS - EXEMPLAR ORGANISATIONS

Australian Commission on Safety and Quality in Healthcare

Entity Type: Corporate Commonwealth Entity and part of the Health portfolio of the Australian Government

Established: 2006 by Council of Australian Governments (COAG)

Annual Revenue: $29 million

Registered Location: Level 5, 255 Elizabeth Street, Sydney NSW 2000

Resources: 94 employees

Summary

The Australian Commission on Safety and Quality in Health Care leads and coordinates national improvements in the safety and quality of health care. Its mission is to provide safe and high-quality health care for every person, everywhere, every time. The Commission was established in 2006 to lead and coordinate national improvements in the safety and quality of health care, its permanent status was confirmed with the passage of the National Health and Hospitals Network Act 2011, while its role was codified in the National Health Reform Act 2011.

Revenue Source

Jointly funded by State and Federal Governments

- $29,055,000 Income
- $11,655,000 Federal Government funding
- $8,093,000 State and Territory contributions
- $28,852,000 - Total Expenses
- $13,486,000 - Employee expenses
- $13,258,000 - Supplier expenses
- $203,000 operating surplus

Activities

- Formulating standards, guidelines and indicators relating to healthcare safety and quality matters
- Advising Health Ministers on national clinical standards
- Promoting, supporting and encouraging the implementation of these standards and related guidelines and indicators
- Monitoring the implementation and impact of the standards
- Promoting, supporting and encouraging the implementation of programs and initiatives relating to healthcare safety and quality matters
- Formulating model national schemes that provide for the accreditation of organisations that provide healthcare services and relate to healthcare safety and quality matters
- Publishing reports and papers relating to healthcare safety and quality matters

Governance

- 8 Board Members
- Chief Executive Officer: Adjunct Professor Debora Picone AO - Chief Executive Officer
- Chief Medical Officer and Clinical Directors
- Committees
- The Australian Government Minister for Health appoints the Commission's Board, in consultation with all state and territory health ministers. The Board is established and governed by the provisions of the National Health Reform Act and the PGPA Act.

Customers / Stakeholders

- Independent Hospital Pricing Authority
- NHMRC
- Australian Institute of Health and Welfare
- Australian Digital Health Agency
- Australian Health Practitioner Regulation Agency
- National Blood Authority.

The Commission also works closely with consumer groups, state and territory health departments, clinical colleges and other professional clinical organisations, complaints commissioners, and universities

Key Findings and relevance to Australian Genomics

- Trajectory in 2006 similar to Australian Genomics, possible next steps

Astronomy Australia

Astronomy Australia Ltd (AAL)

Entity Type:
Australian Public Company
Non-profit organisation

Annual Revenue:
$11.38 million
(Total gross income)

Registered Location:
Hawthorn, Victoria (Swinburne University)

15 funded staff
(6.9 FTE)
30 Volunteers
$1.21 million salaries

Registered:
April 2007

Summary

Astronomy Australia Limited (AAL) is a national non-profit organisation, whose members are Australian universities and research organisations with a significant astronomical research capability. AAL work with national observatories, relevant infrastructure providers, astronomers at universities, and the Australian Government to advance the infrastructure goals in the Australian Astronomy Decadal Plan 2016–2025, Australia in the era of global astronomy.

AAL foster collaboration between academia and industry by building networks and attracting co-investment. Since 2007, AAL has coordinated the Australian astronomy response to, and managed the funding for, a number of national schemes and projects, including the Australian Government's investments in astronomy infrastructure through the National Collaborative Research Infrastructure Strategy (NCRIS).

Revenue Source

- $11,383,140 gross income (2019-20)
  94% government funded ($10,702,540 in 2019-20)
  $175,000 AAT Consortium income
- $262,800 Members' subscriptions
- $182,800 investment revenue

Australian Government contribution to date:

- $45.531 million from NCRIS
- $10.0 million under the Super Science Initiative
- $3.474 million under the Collaborative Research Infrastructure Scheme
- $12.185 million from the NCRIS 2013

Activities

- Grant and funding distribution (govt investment in astronomy infrastructure)
- Optical Astronomy
- SKA Pathfinders
- Data and Computing
- International Engagement
- High Energy Astrophysics

Outputs

- $12,582,912 Grants distributed nationally (2019-20)
- Providing key elements of national radio and optical astronomy infrastructure.
- developing eResearch infrastructure to manage astronomical and astrophysical data
- Industry Engagement

Governance

- In 2019/20 there were 15 institutional members of AAL
- Each member organisation has a nominated representative who attends the AGM to elect Board Directors
- 7 independent, skills-based Board Directors with expertise in astronomy, management and finance.
- Member representatives are also consulted throughout the year on key astronomy infrastructure and investment decisions.

Customers / Stakeholders

AAL's members include all Australian universities and research organisations with a significant astronomical research capability:

- Australian Astronomical Observatory (AAO)
- Australian National University
- Commonwealth Scientific and Industrial Research Organisation (CSIRO)
- Curtin University
- Macquarie University
- Monash University
- Swinburne University of Technology
- University of Adelaide
- University of Melbourne
- University of New South Wales
- University of Queensland
- University of Sydney
- University of Tasmania
- University of Western Australia

Australian Digital Health Agency (ADHA)

Australian Government
Australian Digital Health Agency

Australian Digital Health Agency (ADHA)

Entity Type:
Statutory authority in the form of a corporate Commonwealth entity

Annual Revenue:
$223.2 million
(Total operating revenue)

Registered Location:
Sydney, NSW Australia

Resources:
267 employees

Summary

The Australian Digital Health Agency (the Agency) has responsibility for the strategic management and governance of the National Digital Health Strategy and the design, delivery and operations of the national digital healthcare system, including the My Health Record system. ADHA strives to enable better health through digital health services and technologies that provide innovative tools for patients and providers. The Australian Digital Health Agency is jointly funded by the Australian Government and all state and territory governments.

Revenue Source

The Agency is jointly funded by the Commonwealth ($199.0 million) and the states and territories ($34.3 million) in 2019–20 under an Intergovernmental Agreement between Aust Government, States & Territories

- Total operating revenue of $223.2 million
- Total expenses of $249.9 million
- Operating loss of $26.7 million

Activities

- Designing and calling tenders for the next generation of Australia's national health infrastructure managed or operated by the Agency
- Advancing the interoperability of clinical systems across multiple fronts
- Supporting the widespread implementation of electronic prescribing across general practices and community pharmacies
- Establishing multiple Communities of Excellence that showcase practical implementations of community-wide integrated digital health solutions
- Developing and implementing the National Digital Health Workforce and Education Roadmap
- Enhancing support for registration and meaningful use of My Health Record in the specialist sector and residential aged care facilities
- Expanding use of the Service Registration Assistant to streamline practice administration
- Helping ensure that more complete patient health information is available at the point of care

Outputs

- MHR – drive the registration, viewing and uploading of content to the MHR by emergency departments, Residential Aged Care Facilities, community pharmacies, pathology labs, and practitioners. Increase public view / use of MHR.
- Secure Messaging Program for healthcare providers
- Develop and enhance interoperability and data quality across digital health infrastructure providers
- Electronic prescribing scheme to improve medicines safety
- Applying the above infrastructure to support enhanced models of care system-wide, supported by strategic investment in workforce and education

Governance

The Board delegates responsibility for operational management to the Chief Executive Officer, who, with the support of an executive team. The Agency is a corporate Commonwealth entity under the Public Governance, Performance and Accountability Act 2013.

Customers / Stakeholders

- Governments (Australian, State/Territory)
- Other Government entities / Agencies (e.g. AIHW)
- Digital infrastructure providers
- Healthcare providers (pathology providers, pharmacies, practitioners
- Public

Relevance to Australian Genomics

The ADHA exemplar aligns more with the infrastructure, standards and strategy activity of AG. It is envisaged that the ADHA will be:

A. a key partner with AG moving forward and

B. important to align the content, sequencing and complementarity of recommendations / standards across genomics and digital/eHealth moving forward

Australian Genomics

Australian Radiation Protection and Nuclear Safety Agency (ARPANSA)

Entity Type:
Non-corporate Commonwealth Entity

Annual Revenue:
$25.8 million
(Total gross income)

Registered Location:
Victoria
619 Lower Plenty Road, Yallambie VIC 3085
New South Wales
PO Box 655,
Miranda NSW 1490

Resources:
123 (ongoing), 11 (non-ongoing) staff (as on 30/6/20)

Established
February 1999

Summary

ARPANSA is the Australian Government's primary authority on radiation protection and nuclear safety. ARPANSA is a non-corporate Commonwealth entity established under the ARPANS Act 1998 that provides both services to research organisations (radiation monitoring, badges, training) as well as infrastructure for health systems and commercial activity. Its purpose is to protect the Australian people and the environment from the harmful effects of radiation, through understanding risks, best practice regulation, research, policy, services, partnerships and engaging with the community.

Revenue Source

The Agency manages non-financial assets totalling $40.126 million and its asset management strategy emphasises whole-of-life asset management.
Total operating revenue for the year was $25.833 million and consisted of:

- Government appropriation of $12.757 million
- Regulatory license fees and charges of $5.081 million
- Sale of goods and provision of services and other revenue of $7.995 million.

Activities

- Identify, assess and communicate health, safety and environmental risks from radiation
- Promote radiological and nuclear safety and security, and emergency preparedness
- Promote the safe and effective use of ionising radiation in medicine
- Ensure risk-informed and effective regulation
- Enhance engagement with stakeholders
- Enhance organisational innovation, capability and resilience

Outputs

- Publication of the Medical Exposure Code (Radiation Protection Series C-5), developed in collaboration with state and territory regulators and other members of the Radiation Health Committee
- A new program for electromagnetic energy (EME) research for public information
- A national action plan to implement the findings of the Mission Report from the Integrated Regulatory Review Service mission, coordinated by the International Atomic Energy Agency (IAEA)
- Strong performance against indicators of Australian Government's Regulator Performance Framework
- Finalisation of the integration of the monitoring stations in Fiji and Kiribati into the ARPANSA-operated radionuclide monitoring stations of the International Monitoring System established under the Comprehensive Nuclear-Test-Ban Treaty (CTBT)
- Deployment of the Australian Radiation Monitoring System (ARMS) to provide a new early warning system in the event of any radiation releases during nuclear-powered warship visits or at the Australian Nuclear Science and Technology Organisation (ANSTO) Lucas Heights site.

Governance

ARPANSA sits within the Department of Health portfolio. The CEO, Dr Carl-Magnus Larsson, is the accountable authority of ARPANSA. The ARPANS Act establishes several bodies that advise the CEO of ARPANSA. These bodies include the Radiation Health and Safety Advisory Council, the Radiation Health Committee and the Nuclear Safety Committee. Established by the Australian Radiation Protection and Nuclear Safety Act 1998 (ARPANS Act) ARPANSA replaced the Nuclear Safety Bureau and Australian Radiation Laboratory

Customers / Stakeholders

- Australian Government and associated agencies (e.g. EPA)
- Nuclear installation operators
- Professional bodies / institutions / organisations involved in application of radiation – industry (e.g. mining), healthcare, research

Relevance to Australian Genomics

ARPANSA offers a suite of services associated with the safe, appropriate application and use of radiation:
A. including but not limited to development of regulation, and assessment of compliance
B. service provision including maintaining personal radiation monitoring infrastructure and on-site monitoring
C. interventions and exposure control and
D. negotiation with international regulators / policy makers to contribute to and implement new standards/practices. This suite of activities have equivalents in the genomics arena for AG.

Cancer Australia

Entity Type:
Non-corporate Commonwealth Entity
Registered under the Charitable Fundraising Act 1991 (NSW) and has deductible gift recipient status to receive donations.

Annual Revenue:
$21.2 million
(Total gross income)

Registered Location:
Strawberry Hills, NSW 2012

Resources:
Till 30 June 2020, 76 employees (62 ongoing)

Established 2006 by the Australian Government

Summary

Cancer Australia is a specialist agency providing national leadership in cancer control across all cancers for all Australians. Cancer Australia is an agency within the Australian Government's Health portfolio, that aims to reduce the impact of cancer, address disparities and improve outcomes for people affected by cancer by leading and coordinating national, evidence-based interventions across the continuum of care.

Revenue Source

Australian Government (Refer to Appropriation Bills (No.1 and No.2) 2019-20 and Health Budget Statements 2019-20

- Total departmental resourcing $17.506M (appropriation of $13.248M)
- Total administered resourcing $20.162M (appropriation of $19.946M)
- Total resourcing for Cancer Australia 2019 – 2020: $37.668M
- Estimated expenses for Cancer Australia 2019 – 2020: $33.523M

Total appropriation in 2020-21 estimated to be $29.58M & four years of this plan (2020-21 to 2023-24), total appropriation anticipated to remain relatively stable.

Activities

- Provide national leadership in cancer control
- Guide scientific improvements to cancer prevention, treatment and care
- Coordinate and liaise between the wide range of groups and health care providers with an interest in cancer
- To make recommendations to the Commonwealth Government about cancer policy and priorities
- Oversee a dedicated budget for research into cancer
- Assist with the implementation of Commonwealth Government policies and programs in cancer control
- Provide financial assistance, out of money appropriated by the Parliament, for research
- The implementation of policies and program
- Any functions that the Minister directs Cancer Australia to perform.

Outputs

- **30 research grants awarded $10.71 million**
- **Knowledge Management:** includes research, the Priority-driven Collaborative Cancer Research Scheme, clinical trials support, data and monitoring and evidence review
- **Service Development and Clinical Practice:** includes CanNET, health professional education, evidence-based guidance materials, models of care and consumer programs
- **Cancer Care:** includes breast, gynaecological cancers; and Indigenous and rural
- **Health Promotion and Communication:** includes health promotion, website, e-communications, corporate communications, media, production coordination
- **Corporate Affairs:** includes human resources, contracts, partnerships, IT, clearinghouse
- **Policy, Strategy and Public Reporting**
- **Finance and Compliance**

Governance

Cancer Australia is led by the CEO, Professor Dorothy Keefe PSM MD. Professor Keefe is supported by the Deputy CEO, Dr Cleola Anderiesz who also has responsibility for Policy, Strategy and Public Reporting. Cancer Australia is established under the Cancer Australia Act 2006. The agency is subject to the Auditor-General Act 1997 and its staff are employees of the Australian Public Service

Customers / Stakeholders

- Governments
- Researchers, research institutions
- Clinical services
- Consumers, public.

Relevance to Australian Genomics

Cancer Australia engages across the key identified domains of Australian Genomics through:
A. research support (and funding)
B. recommendations to Government to inform policy
C. apply research evidence to improve practice and
D. engage with a broad network of stakeholders

Wine Australia

Wine Australia

Entity Type:
Commonwealth Government Statutory Authority
Deductible gift recipient (DGR) status: not entitled to receive donations

Annual Revenue:
$73.1 million
(Total gross income)

Registered Location:
Adelaide, South Australia 5000

Resources:
94 employees (as on 30/6/20)
$12.8M employee expenses

Summary

Wine Australia is funded by grapegrowers and winemakers through levies and user-pays charges, and by the Australian Government, which provides matching funding for research, development and adoption (RD&A) investments. Wine Australia's powers and responsibilities come from the Act which sets out role to coordinate or fund grape and wine R&D, and facilitate the dissemination, adoption and commercialisation of results, control the export of wine from Australia, & promote the sale and consumption of wine, in Australia and overseas.

Revenue Source

Four primary sources of funding are:

- **Research, development and adoption (RD&A) funding:** The grape research levy (grapegrowers pay $2 per tonne of winegrapes crushed) and the R&D component of the wine grape levy (wine producers pay $5 per tonne of winegrapes crushed) are matched dollar-for-dollar by the Australian Government.
- **Marketing funding:** Wineries pay the promotion component of the wine grape levy in a stepped amount per tonne. The promotion component is payable on grapes delivered to a winery once the threshold of 10 tonnes has been reached. Wine exporters pay the wine export charge on wine produced in and exported from Australia. The amount of levy payable is based on the free-on-board (FOB) sales value of wine for the levy year.
- **Regulatory funding:** Regulatory activities are funded on a cost-recovery basis through activity-based fees.
- **User-pays activities:** Wine businesses, regional associations and state governments pay voluntary contributions to participate in marketing activities.

Activities

- 38,193,921 expenditure on research and development contracts and other grants

Outputs

- The Statutory Funding Agreement 2020–30 with the Australian Government ensures that the funding received from levies and matching Australian Government funds for eligible RD&A is expended prudently and in line with levy payers' and the Australian Government's expectations
- The Department of Agriculture, Water and the Environment manages the funding agreement on behalf of the Australian Government

Governance

- Board is established and governed under the Wine Australia Act 2013 and its actions are guided by its Board Charter.
- It has a Chair and between five and seven directors

Customers / Stakeholders

- grapegrowers through the grape research levy
- wine producers through the wine grape levy
- wine exporters through the wine export charge,
- Australian Government through its matching funding for investments in research, development and adoption (RD&A) and the Export and Regional Wine Support Package ($50m Package).
- Other stakeholders
  - those employed by grape and wine businesses
  - national, state and regional representative organisations;
  - researchers;
  - state governments;
  - wine industry suppliers;
  - media and Wine Australia staff.

Relevance to Australian Genomics

Wine Australia as a model was recommended by one of our key interviewees. With the emphasis on research and development, and translation / commercialisation of the outputs of that research there are clear parallels with the activities of AG. Further, Wine Australia supports and grows the industry locally and internationally, and also plays a standards / regulatory role in wine production. The funding model of Wine Australia is particularly intriguing in the context of AG's business continuity planning: a combination of Australian Government funding (including matched research funding) and 25% revenue from the industry via a grapegrowers levy.

Australian Genomics

Bioplatforms Australia

Entity Type:
Australian Public Company, Non-profit organisation

Annual Revenue:
$21.2 million
(Total gross income)

Registered Location:
Macquarie University, Macquarie Park NSW

Resources:
269 funded staff
(226.7 Full time employees)
FTE: 6 Charity / Board
(from ACNC register)

Registered:
June 2007

Summary

Bioplatforms Australia is a non-profit organisation that supports Australian Life science research by investing in state-of-the-art infrastructure and expertise in genomics, proteomics, metabolomics and bioinformatics. Mission: Enhance Australian genomics, proteomics, metabolomics and bioinformatics research capabilities, to support innovation and help transform scientific outcomes into tangible benefits for Australians.

Revenue Source

- 95% government funded - Investment funding is provided by the Commonwealth Govt NCRIS
- Federal Government funded commitment of $111 million over five years (2018).
- Revenue from providing goods or services: $315,562
- Revenue from government including grants: $20,052,541
- Revenue from investments: $799,665

Activities

- Manages Commonwealth and State Government grants for the creation and operation of genomics, proteomics, metabolomics and bioinformatics scientific capability
- Supporting an accessible national network of facilities that provide DNA, RNA, protein and metabolite analysis
- Developing scientific initiatives & collaborations of nat. significance across medicine, agriculture & environmental science
- Lead the Australian BioCommons, an ambitious national bioinformatics infrastructure responsive to the ever-increasing digitisation of biology and evolving analytical requirements.
- Support strategic national investments in scientific research infrastructure and personnel;
- Enhance Accessibility of the national infrastructure to enable Australian research;
- Maximise Quality of research via international best practice;
- Identify and support Impact to ensure socio-economic return;
- Maximise Collaboration in delivery and usage of the national infrastructure; and
- Acting as a Catalyst for research collaborations

Outputs

- 15 world-class facilities and 12 active initiatives
- $18.7 million invested in technology and expertise and 14,194 research contracts
- 874 research papers published in peer-reviewed journals with 109 papers published in top 10% journals
- 150 ARC and NHMRC grants Chief Investigator, partner investigator/organisation
- 17 Bioinformatics training (BioCommons, face-to-face and webinar)
- 5464 training attendees (18% live 82% YouTube channel)
- 2886 clients accessed Bioplatforms capabilities in 2020
- 4% of the clients are based overseas (including NZ)
- Client access by sector: Universities 58%, Medical research institutes (MRI) 18%, Publicly funded research agencies (PFRA) 5%, Industry/commercial organisations 11%, Government departments 8% (incl. local government)

Governance

- An independent Board of Directors and Executive Management Committee

Customers / Stakeholders

- NCRIS
- ACT, NSW, QLD, SA & WA Governments
- Macquarie University, University of Melbourne, University of NSW Sydney, Monash University, University of WA, Australian National University, University of South Australia, University of Queensland, The Australian Wine Research Institute, South Australian Health and Medical Research Institute, Australian Genome Research Facility, Garvan Institute of Medical Research.

Relevance to Australian Genomics

Bioplatforms Australia is relevant to Australian Genomics because it:
A. enhances the research quality and impact by making available state-of-the-art 'omic infrastructure, as well as training services
B. is a significant enabler of life sciences research, engaging with universities, medical research institutes and governments and
C. parallels with AG's research enabling capabilities and data infrastructure services, however BPA generally engages with researchers on a fee-for-service capacity rather than partnering with funded grants.

Australian Academic and Research Network (AARNET)

Entity Type:
Australian Public Company, Non-profit organisation
Deductible gift recipient (DGR) status: not entitled to receive donations

Annual Revenue:
$89.8 million
(Total gross income)

Registered Location:
Chatswood NSW 2067 Australia

Resources:
FTE: 131.40

Established 1989

Summary

AARNet is a network run by AARNet Pty Ltd, a not-for-profit company owned by 38 Australian universities and CSIRO. Established in 1989, AARNet is a national resource, a national research and education network which is widely regarded as the founder of the Internet in Australia and renowned as the architect, builder and operator of world-class high-speed low-latency network infrastructure and an expanding portfolio of products and services, all designed to meet the unique needs of the research and education sector.

Revenue Source

Bulk of AARNet's revenue comes equally from membership subscriptions (~$50M) & revenue from services provision. Supported by Commonwealth Investments

- Revenue from providing goods or services: $85,885,120
- Revenue from government including grants: $454,333
- Revenue from investments: $1,501,934

Activities

- Employee expenses: $25,975,063
- Total expenses: $92,630,817 (All other expenses: $66,655,754)
- Net surplus/(deficit): $-2,808,008

Outputs

- $219.9 million in net assets
- $55.6 million invested during 2020
- 99.96% network availability
- 430+ locations in Australia
- 100+ countries worldwide

Governance

- The Board of Directors is responsible for the overall direction of AARNet and for providing benefits to the shareholders as required under the AARNet Constitution.
- The Shareholders are 38 Australian universities and the Commonwealth Scientific and Industrial Research Organisation (CSIRO).
- The Chief Executive Officer (CEO) is charged with the efficient and cost-effective operation of the company.
- AARNet Advisory Committee (AAC) provides technical and policy advice to the CEO. It is representative of the shareholders and other customers mainly on a regional basis.

Customers / Stakeholders

- AARNet's customers include 38 shareholder universities and CSIRO
- Most of the publicly funded research agencies, such as Australian Nuclear Science and Technology Organisation, Geosciences Australia and Australian Institute of Marine Science
- Several state government agencies, hundreds of schools, many TAFEs and hospitals, and most state and federal galleries, libraries, archives and museums.
- AARNet infrastructure interconnects over two million users—researchers, faculty, staff and students—at institutions across Australia with each other and research and education institutions worldwide, the public internet, and resources such as scientific instruments, data storage and high-performance computing facilities.
- AARNet also interconnects content and service providers and organisations that collaborate with the research and education community.

National Computational Infrastructure (NCI)

National Computational Infrastructure (NCI)

Entity Type:
Organisation unit of the Australian National University

Annual Revenue:
$27.3 million
(Total gross income)

Registered Location:
ANU, 143 Ward Road ACTON ACT 2601

Resources:
$8.16 million in salaries
$16.5 million in expenditure
$16,587,479 total expenditure
$10.6 million surplus

NCI was funded by the Australian Government in June 2007.

Summary

The National Computational Infrastructure (NCI) is Australia's leading high-performance data, storage and computing organisation, providing expert services to benefit all domains of science, government and industry.
NCI's integrated hardware, services and expertise empowers industry, universities and government agencies across multiple research domains. NCI was funded by the Australian Government in June 2007 as the leading high-performance data, storage and computing organisation, providing expert services to benefit all domains of science, government & industry. NCI operates as an unincorporated collaborative venture (the Collaboration) of a number of leading national research organisations, bringing Government and research sector together.

Revenue Source

- The NCI Collaboration Agreement enables many of Australia's leading research intensive universities and science agencies to collectively fund a capability beyond the capacity of any single institution.
- Together, these institutions fund a significant proportion of NCI's operating costs.
- A small but growing proportion of NCI Collaboration income comes from the commercial sector.
- NCI operations are also supported by income received under NCRIS.

Activities

- NCI administers a number of grants and contracts outside of the NCI Collaboration and NCRIS accounts. These special purpose arrangements fund clearly defined projects, infrastructure and services that provide synergistic benefits to the NCI Collaboration.
- Capital funding provided by NCRIS – for NCI's new supercomputer, Gadi

Outputs

- 1,125 Projects
- 6,000+ Users
- 756 million hours - computing time

Governance

NCI is governed by the Australian National University on the advice of the NCI Advisory Board, to within the limits of the Statutes and policies of the University.
The Advisory Board comprises:

- An independent Chair
- Director, NCI
- One nominee board member appointed by each of the Major Collaborators (presently ANU, CSIRO, Bureau of Meteorology and Geosciences Australia)
- Additional independent board members appointed for two-year terms by the NCI Advisory Board.

Customers / Stakeholders

- **Supported By:** NCRIS, Australian Government Dept of Education
- **Major Collaborators:** Australian National University, CSIRO, Bureau of Meteorology, Geoscience Australia
- **Collaborators:** ACE, Deakin University, Garvan Institute, QCIF, RMIT University, UTS, University of Wollongong.
- **Other Contracts:** Australian Research Council, INTERSECT, Macquarie University, Monash University, NSW Planning, Industry and Environment, University of Adelaide, University of Melbourne, UNSW Sydney, University of Queensland, University of Sydney, Victor Change Cardiac Research Institute

Australian Research Data Commons (ARDC)

Entity Type:
Australian Public Company

Annual Revenue:
$61.3 million
(Total gross income)

Registered Location:
900 Dandenong Road Caulfield East VIC 3145

Resources:
48 employees
$844,482 staff costs

Established:
May 2019

Summary

ARDC facilitates partnerships to develop a coherent research environment that enables researchers to find, access, contribute to and effectively use services to maximise research quality and impact. ARDC enables the Australian research community and industry access to nationally significant, data intensive digital research infrastructure, platforms, skills and collections of high-quality data.

In July 2018, the ARDC was formed under the Federal Government's National Collaborative Research Infrastructure Strategy (NCRIS) through the merger of three existing digital research infrastructure capabilities:

- the Australian National Data Service (ANDS)
- National eResearch Collaboration Tools and Resources (Nectar)
- and Research Data Services (RDS).

ARDC has 20 members that enable the Australian research community and industry access to nationally significant, data intensive digital research infrastructure, platforms, skills and collections of high-quality data.

Revenue Source

- 99% government funded
- Revenue from government including grants: $61,045,942
- Revenue from investments: $230,028

Activities

- Employee expenses: $844,482
- Total expenses: $4,973,069 (All other expenses: $4,128,587)
- Net surplus/(deficit): $56,303,584.00

Outputs

- Over the past year the ARDC has led a $66.4 million investment in data-related projects and programs to ensure Australia continues producing cutting-edge research.
- ARDC is also leading the way in promoting collaborative activities and FAIR data principles (Findable, Accessible, Interoperable, Reusable), which are increasingly important to our global research environment.

Governance

- 6 Board Directors
- Executive Team
- ARDC Research and Technology Advisory Committee is to provide independent advice to the CEO relating to longer term national eResearch infrastructure requirements and the efficacy of current ARDC program

Customers / Stakeholders

- Australian National University
- CSIRO
- Curtain University
- Edith Cowan University
- Federation University
- Flinders University
- Griffith University
- Macquarie University
- Monash University
- Queensland University of Technology
- Swinburne University
- University of Adelaide
- University of Melbourne
- University of QLD
- University of Sydney
- University of WA
- University of NSW Sydney
- University of TAS
- University of Technology Sydney
- University of Wollongong

Phenomics Australia

Entity Type:
Research Infrastructure Provider

Established 2007

Annual Revenue:
$TBC million
(Total gross income)

Registered Location:
9 locations across Australia

Resources:
unable to locate

Summary

Phenomics Australia provides research institutes and universities with infrastructure and research models for biomedical research. **For nearly 15 years, Phenomics Australia, formerly the Australian Phenomics Network (APN), has been a forward-thinking research infrastructure provider enabling research discovery & high-impact healthcare outcomes in precision medicine. It partners with health initiatives & infrastructure providers to establish integrated development pipelines for research discovery & translation.**

Revenue Source

Phenomics Australia is an integral part of the National Collaborative Research Infrastructure Strategy (NCRIS) funded by the Australian Government. NCRIS supports Australia's research capability by investing in research infrastructure and making it accessible to researchers across the nation.

Activities

- Offers specialised infrastructure, research services and technical expertise dedicated to advancing our fundamental understanding of health and disease
- Helps in enabling next-generation innovations in healthcare and therapeutic development to benefit all Australians.

Outputs

- 9 locations
- 38+ Research Services
- 50+ Technical experts
- 400+ Projects/year
- $100 million + investment

Governance

- 5 Members: Phenomics Australia Board
- 7 members: Scientific Leadership Committee
- 3 members: Executive Team

Customers / Stakeholders

- NCRIS
- Australian National University
- Monash University
- Harry Perkins Institute of Medical Research
- Peter MacCallum Cancer Centre
- South Australian Health & Medical Research Institute
- University of Melbourne
- Walter and Eliza Hall Institute

Victorian Comprehensive Cancer Centre (VCCC)

Victorian Comprehensive Cancer Centre (VCCC)

Entity Type:
Australian Public Company, Non-profit organisation

Annual Revenue:
$11.3 million
(Total gross income)

Registered Location:
Grattan St, Melbourne VIC 3000 Australia

Resources:
Full-time: 20
Part-time: 14
Casual: 2
FTE: 29.60

Summary

The VCCC is a multidisciplinary, multi-site alliance of 10 leading research, academic and clinical institutions working together to accelerate and amplify cancer research, knowledge and expertise to benefit the Victorian community. The VCCC is an alliance that is majority funded by government, but also generates yearly income from member entities.

Revenue Source

The revenue sources are:
- 78% government funding
- $1.5million yearly income from Member Entities ($151k p.a x 10 members)
- $8.7 million Grants from DHHS
- $143k investments

Annual revenue is consisted of:
- Revenue from providing goods or services: $520,331
- Revenue from government including grants: $8,795,290
- Revenue from investments: $143,404
- All other revenue: $1,848,641

Activities

- Employee expenses: $5,017,482
- Total expenses: $16,375,190 (All other expenses: $11,357,708)
- Net surplus/(deficit): $-5,067,524.00

Outputs

- 400+ committee and working group contributors
- 11 new Health Services research projects
- 28,000 education participants
- 11 new investigator-initiated clinical trials
- 8000+ database subscribers
- 140 nurses join Nurse Led Research Hub
- >$15 million leveraged funds
- 6000 Consumer Toolkit website views
- 3,800 Massive Open Online Course Enrolments from 83 countries

Governance

Board Directors are CEOs of the 10 alliance members

Relevance to Australian Genomics

The collaborative translational research alliance of the VCCC could be an exemplar structure for AG moving forward, with a small contribution to revenues from member organisations, and the bulk of funding from Governments. The size of VCCC is similar to AG currently (~$16M annual turnover; 400 contributors) and the range of activities also has parallels.

Customers / Stakeholders

VCCC alliance members:
- Peter MacCallum Cancer Centre
- Melbourne Health (including The Royal Melbourne Hospital)
- The University of Melbourne
- the Walter and Eliza Hall Institute of Medical Research
- the Royal Women's Hospital
- The Royal Children's Hospital
- Western Health
- St Vincent's Hospital Melbourne (including St Vincent's Institute)
- Austin Health (Olivia Newton-John Cancer Research Institute/Austin Lifesciences)
- Murdoch Children's Research Institute

Omico
Australian Genomic Cancer Medicine Centre (AGCMP)

Omico / Australian Genomic Cancer Medicine Centre (AGCMP)

Entity Type:
Australian Public Company, Non-profit organisation
Deductible gift recipient (DGR) status: DGR1 eligible to receive donations

Annual Revenue:
$10.9 million
(Total gross income)

Registered Location:
370 Victoria St, Darlinghurst NSW 2010 Australia

Resources:
FTE: 1.80
Volunteers: 10

Summary

Omico is a network of Australia's leading cancer research institutions and hospitals. It aims to improve outcomes for Australians with cancer by accelerating the use of precision oncology as a research-led model of care, growing the clinical trials industry and modernising the Australian healthcare system. Omico brings together Australia's cancer centres, leading research institutes, federal and state governments, industry partners and patients.

Omico aims to:

- Improve outcomes for cancer patients
- Using genomics to identify therapeutic options for patients with advanced solid or blood cancer of any type and to manage cancer risk.
- Fuel cancer research across the Australian cancer ecosystem
- Accelerate collaborations with local and global stakeholders
- Serve as trusted advisors and scientific experts in precision oncology
- Become a leader for precision oncology in the APAC region.

Revenue Source

92% government funded but also eligible to receive philanthropic funding,
Annual revenue is consisted of:

- Revenue from providing goods or services: $705,326
- Revenue from government including grants: $10,000,000
- Revenue from investments: $158,440

Activities

- Nationally operating charity – precision oncology network with an innovative model for research-led cancer care
- Partners closely with pharma in delivery of Clinical Trials
- Facilitating the delivery of genomic cancer medicine clinical trials to thousands of Australians suffering from advanced and incurable cancers
- Employee expenses: $198,920
- Total expenses: $9,191,154 (All other expenses: $8,992,234)
- Net surplus/(deficit): $1,672,612.00

Outputs

- Molecular screening and therapeutics
- Health system reform
- Personalised risk management
- Patient support and advocacy

Governance

- 12 Board Members (Directors)
- 8 Executive Leadership members
- 7 Committees

Customers / Stakeholders

Members:

- Royal Hobart Hospital, Govt of WA, PathWest WA, Linear, Peter Mac, SA Pathology, Govt of SA, Pathology Queensland, Metro South Health QLD Govt, NT Govt, University of Sydney, Garvan Institute, NHMRC Clinical Trials Centre.

Industry Partners:

- AstraZeneca, Roche, Foundation Medicine, Bayer, Pfizer Oncology, Medicines Australia, Sun Pharma, Seagen, Eisai, InGeNA

Collaborators:

- Rare Cancers Australia, BioGrid, NSW Govt, Macquarie University, MRFF, Leukaemia Foundation, Minderoo Foundation, Australian Genomics, Canteen, Cooper Rice-Brading Foundation, QIRM Berghofer, Douglas Hanly Moir Pathology, NSW Health, Australian Government Department of Health, Thoracic Oncology Group Australasia.

American Society of Human Genetics (ASHG)

Entity Type:
Non-profit scientific membership-based organisation

Annual Revenue:
$10.8 million AUD
(Total gross income)

Registered Location:
Rockville, MARYLAND, USA 20852

Resources:
Less than 50 employees

Summary

ASHG is the primary professional membership organisation for human genetics specialists worldwide. ASHG also works to increase the availability of reliable information about genetics and genomics to the public so that people everywhere realize the benefits of human genetics and genomics research. Members include researchers, clinicians, genetic counsellors & others who have a special interest in the field of human genetics and genomics research. ASHG strives to advance the field of human genetics and genomics through excellence in research, education, and advocacy.

Revenue Source

The revenue sources are:
- Annual Meeting ($4.56 million USD)
- Royalty Income ($1.47 million USD)
- Advertising ($362,267 USD)
- Membership ($936,299 USD)
- Government Grants ($156,435 USD)
- Investments ($425,000 USD)

Annual revenue is consisted of:
- $8 million USD in revenue ($10.8 million AUD)
- Net assets over $500,000 USD

Activities

ASHG serves research scientists, health professionals, and the public by providing forums to:
- Share research results at annual meetings and in The American Journal of Human Genetics
- Advance genetic research by advocating for research support
- Enhance genetics education by preparing future professionals and informing the public
- Promote genetic services and support responsible social and scientific policies
- Expenses: $7.84 million USD in expenses ($10.6 million AUD)
- 32% Office and administration costs
- 6% Journal
- 34% Annual Meeting
- 27% Programs: Education, Policy, Workforce, Membership
- 7,575 members from 83 Countries (72% in USA) Members include researchers, academicians, clinicians, laboratory practice professionals, genetic counsellors, nurses
- 8,409 attendees from 80 Countries – Annual Meeting
- Receives philanthropic support

Governance

- ASHG is a professional society, led by volunteers.
- Nominated and elected to serve on the Board consistent with the ASHG Bylaws, members guide the Society's direction and resources

Customers / Stakeholders

Collaborators
- Ad Hoc Group for Medical Research
- American Association for the Advancement of Science
- American Board of Medical Genetics and Genomics
- American College of Medical Genetics and Genomics
- Annual Biomedical Research Conference for Minority Students
- Consortium on Sexual Harassment in STEMM
- European Society of Human Genetics
- Federation of American Societies of Experimental Biology
- International Federation of Human Genetics Societies
- National Human Genetics Research Institute
- National Science Teachers Association
- National Society of Genetic Counselors
- Research!America
- Society for Advancement of Chicanos/Hispanics and Native Americans in Science
- U.S. National Academies of Science, Engineering and Medicine

Relevance to Australian Genomics

Potentially low applicability since its an individual membership model, and it seems most funds are directed toward putting on the annual meeting.

Association of Australian Medical Research Institutes (AAMRI)

Association of Australian Medical Research Institutes (AAMRI)

Entity Type:
Australian Public Company
Deductible gift recipient status: not entitled to receive donations

Annual Revenue:
$745K
(Total gross income)

Registered Location:
Flemington 3052 Victoria, Australia

Resources:
FTE: 3.3 Charity / Board (ACNC register)
100 Volunteers
19,763 members

Summary

AAMRI's vision is to achieve positive outcomes for health and medical research in Australia. AAMRI is the peak body for medical research institutes in Australia. 57 member organisations (medical research institutes, university-based MRIs or a hospital/university alliances) undertake world-class medical research to improve health outcomes. They also work on a broad spectrum of human health issues such as preventive health, chronic disease, mental health, immunology and Indigenous health.

Revenue Source

The revenue sources are:
- 91.7% income - membership fees ($683,441)
- 5.9% income - sponsorship fees ($44,039)
- 2.2% income – customer contracts ($17,000)
- 0.2% income – investments ($944)

Annual revenue is consisted of:
- Revenue from providing goods or services: $744,480.00
- Revenue from government including grants: $0.00
- Revenue from investments: $944.00

Activities

- Promote the benefits of health and medical research
- Advocate for high level policy reform in health and medical research
- Provided authoritative advice and information on health and medical research
- Build the positive profile and strong voice of our members medical research institute organisations
- Represent, engage and connect our members

Outputs

- $611k in operating expenses ($434k salaries)
- 57 member organisations (medical research institutes, university-based MRIs or a hospital/university alliances
- 19,763 members

Governance

- 8 Board Directors

Customers / Stakeholders

- 57 national member organisations (medical research institutes, university-based MRIs or a hospital/university alliances)

Genomics England

Genomics England

Entity Type:
Company

Annual Revenue:
AUD $49.1 million

Registered Location:
England and Wales (Charterhouse Square, London EC1M 6BQ)

Resources:
172 Employees

Registered:
April 2013
Registered number
08493132

Summary

Genomics England is a company set up and owned by the Department of Health and Social Care in the UK, creating a lasting legacy for patients, the NHS and the UK economy, initially through the flagship project sequencing 100,000 genomes from NHS patients with a rare disease and their families, and patients with cancer. It partners closely with NHS England, Health Ed England and Public Health England – as well as 85 NHS trusts / hospitals. Genomics England has been tasked with delivering genomic medicine in partnership with NHS.

Revenue Source

The revenue sources are:

- National Institute for Health Research and NHS England.
- The Wellcome Trust (Philanthropic) - £27 million donation for genome-sequencing hub in 2014
- Cancer Research UK (Charity)
- Illumina - £78 million in services for whole genome sequencing
- Medical Research Council funded research and infrastructure.

Annual revenue consists of:

- Cash GBP £26.5 million (AUD $49.1 million)
- Net Worth GBP £32 million (AUD $59.3 million)
- Current Assets GBP £33.5 million (AUD $62.0 million

Activities

- Research – delivered the 100,000 Genomes Project
- Infrastructure – developed the sequencing capabilities and bioinformatic infrastructure to deliver the Project, which transitioned to supporting the NHS Genomic Medicine Service
- Enabling capabilities – workforce development and education; system adaptation to support genomic medicine (e.g. changes to pathology practices)
- Public involvement and engagement – Genome England has an extensive catalogue of public-directed education, consultation and engagement
- Advocacy – GeL is active in the GA4GH, and international collaborations to share resources and practices.

Outputs

- Flagship project sequenced 100,000 whole genomes from NHS patients with rare diseases, and their families, and patients with common cancers
- In October 2018, the program expanded with a new goal of sequencing 5 million genomes over the next five years
- From 2019, the NHS offer whole genome sequencing (WGS) to all children suspected of having a rare genetic disease or with cancer
- In July 2019 Data Release 7 included the 100,000th whole genome available to researchers

Relevance to Australian Genomics

As a national genomic initiative, Genomics England has a range of activities that are similar in scope to Australian Genomics
A. research support
B. health system translation
C. capabilities development and
D. infrastructure provision and engagement

Governance

- The Secretary of State is the only shareholder
- Governed by a Board who oversees all activities, ratifies all major decisions and sets organisation strategy
- 16 Directors, led by Executive Chairman
- Independent advisory committees that report to the board: Ethics Advisory Committee, Science Advisory Committee, Data Advisory Committee, Access Review Committee, GeCIP Board and the Audit Committee

Customers / Stakeholders

- NHS England
- Health Education England
- Public Health England
- 85 NHS Trusts and hospitals across England

Genome British Colombia

Genome British Colombia (GBC)

Entity Type:
Non-profit organisation

Annual Revenue:
$36.7 million
(Total gross income)

Registered Location:
Vancouver, BC V5Z 0C4 Canada

Summary

Genome British Colombia supports in world-class genomics research and innovation to grow globally competitive life sciences sectors. The organization's initiatives are improving the lives of British Columbians by advancing health care in addition to addressing environmental and natural resource challenges. GBC has helped foster collaboration between academia and industry by building networks and attracting co-investment. GBC has driven genomics innovation on Canada's West Coast and facilitated the integration of genomics into society.

Revenue Source

The revenue sources are:

- Province of British Columbia
- Government of Canada through Genome Canada
- Western Economic Diversification Canada
- Project co-funders

Activities

- Health: 219 projects, $525 Million (AUD $566m)
- Tech/Dev Platforms: 63 projects $183 Million
- Forestry: 32 projects $108 Million
- Fisheries/Aquaculture: 28 projects, $68 million
- Agrifood: 38 projects, $72 million
- Energy/Mining: 16 projects, $38 million
- Environment: 29 projects, $21 million
- Total: 425 projects, $1.018 billion

Outputs

- $1.12 Billion total investment ($1.29 billion AUD)
- 425 genomics research projects and science and technology platforms
- The effort has helped advance 102 companies to date, supported BC job growth and contributed to international recognition
- 32,400 jobs created
- 1,026 partnerships
- 3,404 Scientific Publications
- 699 Patent Applications
- $857 Million Direct Co-Investment attracted

Customers / Stakeholders

- Academia and industry
- Province of British Columbia
- Government of Canada through Genome Canada
- Western Economic Diversification Canada

National Collaborative Research Infrastructure Strategy (NCRIS)

National Collaborative Research Infrastructure Strategy (NCRIS)

Entity Type:
Other organisations

Summary

NCRIS is a national network of world-class research infrastructure projects that support high-quality research that will drive greater innovation in the Australian research sector and the economy more broadly. Users relying on NCRIS range from early career researchers and small businesses who would otherwise struggle to access world-class NRI, to global research leaders tapping into the unique facilities that NCRIS provides.

Revenue Source

As of the 2018 NRI Census, each $1 Government invested in NCRIS saw $1.29 in co-investment from universities, research agencies, state and territory governments and industry.

Annual revenue includes the following:

- The Commonwealth Government will invest $4 billion in NRI from 2018 to 2029
- $251 million will be spent in 2020-21 supporting Australia's national research infrastructure through NCRIS

Activities

- NCRIS currently supports 22 funded projects, plus pilot projects and an international membership
- Currently undertaking a review of funding/priorities
- The projects form a network involving over 200 delivery partnerships, and employing over 1900 highly skilled technical experts, researchers and facility managers

Outputs

- High co-investment rates - each $1 Government invested in NCRIS saw $1.29 in co-investment (2018 NRI Census)
- Projects provide merit-based access for all Australian researchers to their infrastructure
- As of the 2018 NRI Census around 65,000 Australian and 12,000 international users are supported every year
- In 2018-19, the projects overall had a 94% user satisfaction rate

Customers / Stakeholders

- The projects are led by organisations including universities, publicly funded research organisations and private companies.

10.5 APPENDIX 5: AUSTRALIAN GENOMICS FUNDING SOURCE MODELLING

Shariant revenue modelling

Membership Charge to Shariant Users	*Revenue Generation (based on projected Shariant user growth)*				
Membership Fee	**2024**	**2025**	**2026**	**2027**	**2028**
$ 20,000	$ 439,898	$ 461,892	$ 484,987	$ 509,236	$ 534,698
$ 30,000	$ 659,846	$ 692,839	$ 727,480	$ 763,855	$ 802,047
$ 50,000	$ 1,099,744	$ 1,154,731	$ 1,212,467	$ 1,273,091	$ 1,336,745
$ 100,000	$ 2,199,488	$ 2,309,462	$ 2,424,935	$ 2,546,182	$ 2,673,491

10.6 APPENDIX 6: AUSTRALIAN GENOMICS FUNDING BASE MODELLING

OPTION 1 - Australian Genomics Future Cost Base

SCALE BACK OF CURRENT ACTIVITIES

	Current State (2023)	Future State (2024 Onwards)				
	2023	2024	2025	2026	2027	2028
Net New Hires per Year (FTE)		-5	-1	0	0	0
Future People Costs						
Research enabling activities and capabilities	$ 1,228,069	$ 1,266,139	$ 1,184,955	$ 1,221,689	$ 1,259,561	$ 1,298,608
Health system translation	$ 1,245,988	$ 972,572	$ 1,002,721	$ 1,033,806	$ 1,065,854	$ 1,098,895
Key partnerships / collaborations	$ 1,239,908	$ 1,143,379	$ 1,178,824	$ 1,215,367	$ 1,253,044	$ 1,291,888
Infrastructure	$ 1,399,976	$ 1,173,442	$ 1,209,819	$ 1,247,323	$ 1,285,990	$ 1,325,856
Total People Costs	**$ 5,113,941**	**$ 4,555,527**	**$ 4,576,318**	**$ 4,718,185**	**$ 4,864,449**	**$ 5,015,247**
Future Non-People Costs						
Operating Costs	$ 180,000.00	$ 186,272.28	$ 192,763.12	$ 199,480.15	$ 206,431.23	$ 213,624.54
Platform Costs	$ 700,000.00	$ 717,220.00	$ 734,863.61	$ 752,941.26	$ 771,463.61	$ 790,441.62
TOTAL COSTS	$ 5,993,941	$ 5,459,019	$ 5,503,945	$ 5,670,607	$ 5,842,344	$ 6,019,313

OPTION 2 - Australian Genomics Future Cost Base

CONTINUATION OF ACTIVITIES

	Current State (2023)	Future State (2024 Onwards)				
	2023	2024	2025	2026	2027	2028
Net New Hires per Year (FTE)		9	2	2	2	2
Future People Costs						
Research enabling activities and capabilities	$ 1,228,069	$ 1,967,015	$ 2,167,143	$ 2,234,325	$ 2,303,589	$ 2,375,000
Health system translation	$ 1,245,988	$ 1,401,426	$ 1,646,152	$ 1,697,182	$ 1,749,795	$ 1,804,039
Key partnerships / collaborations	$ 1,239,908	$ 1,413,312	$ 1,457,125	$ 1,502,296	$ 1,824,795	$ 1,881,364
Infrastructure	$ 1,399,976	$ 1,560,188	$ 1,608,554	$ 1,926,051	$ 1,985,758	$ 2,331,799
Other	$ -	$ -	$ -	$ -	$ -	$ -
Total People Costs	$ 5,113,941	$ 6,341,942	$ 6,878,974	$ 7,359,853	$ 7,863,937	$ 8,392,201
Future Non-People Costs						
Operating Costs	$ 180,000.00	$ 193,649.40	$ 202,381.44	$ 211,507.22	$ 221,044.51	$ 231,011.85
Platform Costs	$ 700,000.00	$ 753,081.00	$ 787,038.93	$ 822,528.09	$ 859,617.52	$ 898,379.40
TOTAL COSTS	$ 5,993,941	$ 7,288,672	$ 7,868,394	$ 8,393,889	$ 8,944,599	$ 9,521,592

OPTION 3 - Australian Genomics Future Cost Base

MODERATE EXPANSION OF ACTIVITIES

	Current State (2023)	Future State (2024 Onwards)				
	2023	2024	2025	2026	2027	2028
Net New Hires per Year (FTE)		26	3	3	3	3
Future People Costs						
Research enabling activities and capabilities	$ 1,228,069	$ 2,676,067	$ 2,759,025	$ 2,844,555	$ 2,932,736	$ 3,023,651
Health system translation	$ 1,245,988	$ 2,079,971	$ 2,283,601	$ 2,497,857	$ 2,723,202	$ 2,960,118
Key partnerships / collaborations	$ 1,239,908	$ 1,800,058	$ 2,115,444	$ 2,448,654	$ 2,800,491	$ 3,171,788
Infrastructure	$ 1,399,976	$ 2,081,900	$ 2,146,439	$ 2,212,979	$ 2,281,581	$ 2,352,310
Other	$ -	$ 269,933	$ 278,301	$ 286,928	$ 295,823	$ 304,994
Total People Costs	$ 5,113,941	$ 8,907,930	$ 9,582,810	$ 10,290,973	$ 11,033,833	$ 11,812,861
Future Non-People Costs						
0	$ 180,000.00	$ 204,715.08	$ 213,946.09	$ 223,593.35	$ 233,675.62	$ 244,212.52
0	$ 700,000.00	$ 753,081.00	$ 787,038.93	$ 822,528.09	$ 859,617.52	$ 898,379.40
TOTAL COSTS	$ 5,993,941	$ 9,865,726	$ 10,583,795	$ 11,337,095	$ 12,127,126	$ 12,955,453

10.7 APPENDIX 7: DETAILED ANALYSIS OF ALTERNATIVE ROLES PROPOSED FOR AUSTRALIAN GENOMICS

Please see section 7: Proposed Future for Australian Genomics for detailed description of the chosen 'Connector' role for Australian Genomics.

2. Standards Provider

This role would focus on strengthening quality and consistency across health genomics by developing and disseminating a set of standards – addressing the current fragmentation in information management, and ensuring an efficient approach to health genomic data and project mobilisation. By focusing on creating consistency and data interoperability, Australian Genomics can partner with the States and Territories to maintain a more consistent approach in line with global best practice.

- **Research Enabling Activities:** Streamlining Research Processes
  Assist in optimising ethics procedures and creating consistent processes to allow for more efficient research project mobilisation and improved research participant experience and engagement - become an implementable tool provided by Australian Genomics
- **Infrastructure:** Data Management Leader
  Lead the national approach for information management "standards" to ensure the interoperability of datasets and best practice information management
- **Health System Translation:** Message Amplification Leader
  Utilise network to amplify messaging to inform genomics policy and address challenges in developing a cohesive national approach
- **Key Partnerships / Collaborations:** Local and International Genomics Bodies
  Continue to be national collaborators with global alliances for standards in data interoperability, access and sharing, and with international collaborative entities, sharing tools and resources for enhanced credibility

The Standards Provider role achieves efficiencies with current processes while creating resources of value to stakeholders. This can provide new funding mechanism opportunities in monetising standards and processes to academic organisations, industry and health providers. However, it may be difficult to articulate the value to stakeholders of tools and resources that have previously been 'free'. The most appropriate governance structure for this role is that of a company limited by guarantee as independence from government would be key in providing standards that are consistent across Federal and State/Territory jurisdictions.

Exemplar Organisations

Both American Society of Human Genetics (ASHG) and Australian Research Data Commons (ARDC) are public entities with member organisations. ASHG generates 87% of its revenue through annual meetings, royalty income and membership fees, whereas ARDC is government funded ($16M NCRIS 2020). Both exemplar organisations provide standards for organisations but also take on other roles: for example, ASHG organises conferences and forums, and ARDC provides common platforms45. Hence, without providing an "accreditation" element or playing additional roles, it may be harder for Australian Genomics to capitalise on the full value of standards provision, leading to lessened credibility in the genomics community.

[45] https://ardc.edu.au

3. Standards Endorser

This role focuses on becoming a standards and accreditation body that develops research project mobilisation and information management standards into implementable tools to endorse stakeholders for best practice genomics activities. It should be noted that interviewees recognised a gap in the accreditation and standards space specifically for genomic data management and project mobilisation processes. Any accreditation scheme would not focus on areas where accreditation is already established, including overseeing safe clinical testing, providing education to the sector or test classifications. The role would leverage entities adjacent to Australian Genomics to enable control over a niche in the genomics value chain, thus opening up new funding opportunities in standards accreditation. The accreditation role solidifies 'Trusted Advisor' positioning of Australian Genomics when collaborating with different bodies.

- **Research Enabling Activities:** Process Improvement Endorser
  Assist in determining ethics procedures and creating accreditation tools to inform standards for project endorsement - becomes an implementable tool provided by Australian Genomics
- **Infrastructure:** Data Management Leader
  Build credibility by providing data management accreditation through enforcing a set of standards for genomics data that supports the interoperability of data
- **Health System Translation:** Message Amplification Leader
  Leverage network to amplify messaging to inform genomics policy and develop accreditation schemes that address challenges in developing a cohesive national approach
- **Key Partnerships / Collaborations:** Local and International Genomic Bodies
  Continue to be national collaborators with global alliance for standards in data interoperability, access and sharing, seek opportunities to share accreditation tools with international networks for enhanced credibility

Exemplar Organisations

Australian Commission on Safety and Quality in HealthCare (ACSQHC), Wine Australia and Australian Radiation Protection and Nuclear Safety Agency (ARPANSA) each embody a "standards endorser" role. They are all government bodies, positioning them to provide credible endorsement for stakeholders, but potentially lessens the level of perceived independence from government agendas, which is a consideration for Australian Genomics. While ACSQHC is solely government funded, Australian Radiation Protection and Nuclear Safety Agency (ARPANSA) and Wine Australia both generate revenue through regulatory license fees and charges and government appropriation46. Wine Australia specifically generates revenue through government grants, and co-funding of wine/grape research (at 50% of Wine Australia's contribution). The models present compelling opportunities for Australian Genomics, however, may detract from its current core value proposition as a key advocacy body.

Conclusion

Each of the three options adopt different perspectives into the role that Australian Genomics could play in the future. The "connector" focus will best place Australian Genomics to improve the impact and efficiency of genomic research, drive the translation of that research into health policy and practice. The role leverages Australian Genomics' expanding network of collaborators to provide a "unified voice" for transforming health genomic implementation in Australia and globally. By avoiding fragmentation across the federation, Australian Genomics' role will be instrumental in driving the implementation of genomic medicine as standard of care.

[46] proportion of government revenue FY2019/2020 50% ARPANSA, 59% Wine Australia