Acknowledgments

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InGeNA recognises Aboriginal and Torres Strait Islander peoples as the First Peoples of this nation. We recognise the ongoing traditional and modern cultural practices and connection to Country held by Aboriginal and Torres Strait Islander peoples. We acknowledge Elders past, present and emerging as the Traditional Custodians and Lore Keepers of the world’s oldest living culture.
Table of Contents

**Executive summary** ................................................................................................................................................. 6

**An industry perspective on genomic data management** ........................................................................................................... 7

- Diversity of the genomics industry ........................................................................................................................................ 7
- Previous genomic data investigations ................................................................................................................................. 8
- Agreeing a position for industry on genomic data management and sharing ........................................................................ 8

**The present state of genomic data in Australia** ................................................................................................................ 10

- Public health and genomics ................................................................................................................................................... 10
  - Australian Government ........................................................................................................................................................ 10
  - New South Wales .................................................................................................................................................................. 11
  - Victoria .................................................................................................................................................................................. 12
  - Queensland ............................................................................................................................................................................ 12
  - South Australia ..................................................................................................................................................................... 13
  - Western Australia ................................................................................................................................................................. 13
  - Tasmania ................................................................................................................................................................................ 13
  - Northern Territory ................................................................................................................................................................. 14
  - Australian Capital Territory .................................................................................................................................................... 14
  - Interstate and private sample testing ................................................................................................................................ 14

- Australian genomic alliances .................................................................................................................................................... 15
  - Australian Genomics ............................................................................................................................................................ 15
  - Melbourne Genomics ........................................................................................................................................................... 16
  - Queensland Genomics ............................................................................................................................................................ 16
  - Sydney Genomics Collaborative ........................................................................................................................................... 17

- Current and evolving state of technology ............................................................................................................................... 17
  - Sequencing and other technologies .................................................................................................................................. 17
  - Storage and analysis of genomic data ................................................................................................................................ 18
  - Curation and interpretation of genomic data ........................................................................................................................ 18
  - Emerging technologies and approaches .............................................................................................................................. 18

**Challenges and opportunities** .............................................................................................................................................. 20

- Data for precision medicine ................................................................................................................................................... 20
  - Data classification ................................................................................................................................................................. 20
  - Data discoverability ............................................................................................................................................................... 20
  - Challenges of federating research data ................................................................................................................................. 21
  - Integration of clinical data ..................................................................................................................................................... 21

- Privacy, consent and data sharing .......................................................................................................................................... 22
  - Understanding genomic privacy ........................................................................................................................................... 22
  - Compliance regimes .............................................................................................................................................................. 23
  - The impact on consent ........................................................................................................................................................... 24
  - Who defines consent? .............................................................................................................................................................. 24
  - Emerging models of consent .................................................................................................................................................. 25
  - Private insurance and genomics .......................................................................................................................................... 25

*Genomic data in Australia - An industry perspective on clarity, certainty and standardisation*
Genomic data in Australia - An industry perspective on clarity, certainty and standardisation

Direct-to-consumer applications.................................................................................................................. 26
Data governance .............................................................................................................................................. 27
   A framework for data governance............................................................................................................... 27
Data security .................................................................................................................................................. 27
Promoting data sharing ................................................................................................................................. 28
Sovereignty and data ownership..................................................................................................................... 28
Reuse versus resequencing............................................................................................................................ 29
Interoperability .............................................................................................................................................. 29
Adoption of standards ................................................................................................................................. 30
   Standards maturity in genomics.................................................................................................................. 30
Evolving synoptic reporting standards.......................................................................................................... 30
The cost of collaboration ............................................................................................................................... 31
Considering diversity .................................................................................................................................... 31
Resources ...................................................................................................................................................... 32
   A common language ................................................................................................................................. 32
Education materials ..................................................................................................................................... 32
Catalogue of endorsed standards.................................................................................................................. 33

The role of the genomics industry .................................................................................................................. 34
   InGeNA’s 2021 projects ............................................................................................................................... 34
   Working with the Australian Government ................................................................................................. 34
   Supporting the states and territories .......................................................................................................... 35
   Engaging professional bodies ................................................................................................................... 35
   Working with consumer advocacy and support groups ............................................................................. 36
   Working with research alliances and groups ............................................................................................. 36
   Services to the industry ............................................................................................................................... 38

Case studies .................................................................................................................................................. 39
   Case study 1: Innovation & Genomic Profiling in Australia by OMICO....................................................... 39
   Case study 2: Genomics as a population health response to the COVID-19 pandemic ......................... 40
   Case study 3: Clinical genomics and data sharing in Victoria .................................................................. 40

Summary of opportunities and activities ..................................................................................................... 42
   Identified opportunities ............................................................................................................................... 42
   Recommended activities.............................................................................................................................. 45
   Remaining challenges................................................................................................................................. 46

Appendix A: Overview of methodology ....................................................................................................... 48
   Range of stakeholders ............................................................................................................................... 48
   Stakeholder survey ................................................................................................................................... 48
   Response rates .......................................................................................................................................... 48
   Survey questions ...................................................................................................................................... 48
   Survey analysis ......................................................................................................................................... 50
Executive summary

The collection, management and exchange of genomic data into healthcare delivery is critical to supporting better person-centred care, driving value to the health system and to building a thriving research ecosystem. The delivery of mature genomics information management which supports the integration of genomics into mainstream health care requires a comprehensive and collaborative approach.

The genomics industry has an important role in delivering this maturity and ensuring that value is derived for all stakeholders. With the establishment of the Industry Genomics Network Alliance (InGeNA) in 2020, it is now appropriate to examine industry’s role in leveraging the value of genomic data to support better health outcomes for Australians.

Engagement with stakeholders from industry, healthcare, research and consumers indicated three consistent themes, being a need for clarity in how the sector communicates, certainty about regulatory and compliance matters, and the importance of standards adoption to support the application of genomics to realise the value for people, the healthcare sector and the research community.

As a united voice for the genomics industry, InGeNA has undertaken a review of the Australian genomic data landscape to identify the challenges and opportunities evidence regarding the management, governance, sharing and use of genomic data to support better outcomes for all Australians. This report then considers the role that InGeNA can play in supporting the delivery of this potential.

By establishing shared positions regarding how genomic data is shared, managed and used in Australia, InGeNA can engage with governments at all levels, and with professional bodies, consumer groups, research groups and the broader healthcare sector to support and encourage the appropriate use of genomic data in Australia without the perception of self-interest that any one organisation may attract. Indeed, the very diversity of interests among the InGeNA membership ensures that a balanced and positive position can be found across a range of topics.

This report is intended primarily as an input to planning for InGeNA. However, it should also serve as a useful guide to the value industry can bring to public health, research and the broader sector.
An industry perspective on genomic data management

Australia’s National Health Genomics Policy Framework identified data as one of five strategic priorities to support the integration of genomics into health care [1]. The collection, management and exchange of genomic data into healthcare delivery is critical to supporting better person-centred care, driving value to the health system and to building a thriving research ecosystem. The delivery of mature genomics information management which supports the integration of genomics into mainstream health care requires a comprehensive and collaborative approach. The genomics industry has an important role in delivering this maturity and ensuring that value is derived for all stakeholders.

Diversity of the genomics industry

Genomics is inherently complex and involves the coordination and collaboration of organisations within a diverse ecosystem [2]. As illustrated in Figure 1, there are five broad phases in the delivery of genomics:

- **Sampling**, through which the patient or related part provides a biological sample in readiness for processing. This may include use of a biobank for the sample.
- **Processing** of the sample using the relevant technology to produce the raw genomic data. The type of data produced will vary on the technology used.
- **Analysis** of this data to identify genomic variants or other markers of genetic significance.
- **Interpretation** of the clinical significance of the analysed sample.
- **Application** of this information for clinical diagnosis or treatment options, or as part of a program of research or development.

The data generated through these processes vary depending upon the nature of the technologies engaged. Exome or genome sequencing can produce large volumes of sequencing data requiring bioinformatics pipelines to identify genomic variants, while other technologies may produce comparatively smaller datasets that require less intensive processing.

Solutions to deliver these capabilities are provided by a range of industry participants [2], including:

- organisations providing sequencing and other technology products or services
- bioinformatics solutions, commonly cloud-based, to support processing, analysis and exploration of the data
- specialist organisations developing diagnostic tools or biomarker solutions for use in the interpretation or application phases
- solutions for managing the data in on-premises, cloud or hybrid solutions
- pharmaceutical organisations who rely on genomic data to support research into new pharmaceutical products, therapies or diagnostic tools
- organisations translating research into clinical practice through clinical trials.

Organisations addressing these capabilities range from multinational companies with a broad range of interests to small and medium organisations that are targeting specific sector requirements.

Other participants in the genomics ecosystem include:

- patients, who must be the primary beneficiary of genomics technologies
• research groups, who improve genomic technologies and provide the evidence required to support the wider adoption of clinical genomics
• clinicians within public and private health establishments who rely on genomics to support prevention, diagnosis and treatment decisions
• public and private diagnostic providers who incorporate genomics into the suite of services provided
• clinical colleges and professional groups responsible for setting professional standards for clinicians
• governments who provide funding within policy, legislative and regulatory frameworks.

All these groups must work together to deliver sustainable genomics data capabilities.

Previous genomic data investigations

Following the 2017 Council of Australian Governments Health Council endorsement of the National Genomics Health Policy Framework [1], and the Implementation Plan for the framework developed and released in 2018, projects were initiated to address the five strategic priority areas. The (then) Australian Health Ministers’ Advisory Council (AHMAC) commissioned Queensland Health to develop a National Approach to Genomics Information Management (NAGIM) in 2019 with a project led by Queensland Genomics and its project partner, the Commonwealth Scientific and Industrial Research Organisation (CSIRO). The NAGIM project supported harmonisation of investments in, and linkage between, clinical delivery systems and research endeavour and infrastructure to progress rapid and safe adoption of medical genomics.

The NAGIM project worked with public health agencies across Australia, and research and infrastructure groups and experts in genomics and data management to produce two key outputs.

The first, the Report on a National Approach to Genomics Information Management (the NAGIM Report) [3], defined the framework for the project, and reported on investigations into current clinical genomics activities and related data management within each state and territory in Australia. The NAGIM Report, intended to inform government planning, included opportunities for future activities.

The second output was the Blueprint for a National Approach to Genomics Information Management (the NAGIM Blueprint) [4], intended to support implementers working in jurisdictional health agencies and research groups across Australia. The NAGIM Blueprint set out to represent a future state for genomics information management in Australia but recognised the varying starting points of actors in the Australian healthcare system, and hence positioned horizons of activity and collaborative action.

The NAGIM Blueprint set out principles to guide future implementations, a genomic data categorisation framework to provide a consistent language to describe genomic data, a governance framework, and options for logical architectures for genomic information supporting interoperability in the sector.

While the NAGIM project was the first wide-angle assessment of genomics data in Australia, its primary focus was on public health and research. While some limited engagement occurred with industry participants, full engagement with the genomics industry was outside the scope of the project and at that stage, no formal industry body existed to support such engagement.

Agreeing a position for industry on genomic data management and sharing

With the establishment of the Industry Genomics Network Alliance (InGeNA) in 2020, it is now appropriate to revisit the industry’s role in leveraging the value of genomic data to support better health outcomes for Australians.
This paper covers a range of topics, including:

- articulating the relationships between industry participants and other stakeholders along the genomics value chain
- identifying the role(s) industry can play in delivering the full potential of genomics to enable personalised health care in Australia
- describing the barriers and roadblocks to data sharing and the industry’s role in addressing those changes
- identifying key areas where industry can contribute to robust data management for long-term health prevention, diagnosis and treatment
- the value of an agreed position on genomic data management and sharing to provide certainty to sector participants and coordination of activities by industry participants
- support clear and transparent industry communication and build a balanced industry position for discussion, influence and action
- establishing the value of a collective position to bring consistency and balance to ongoing discussions and development across the genomics industry.
The present state of genomic data in Australia

While the value of genomics in health care is clear [5], widespread adoption of genomics in mainstream clinical care has not yet been achieved in Australia. Significant progress has been made, especially in a research context, with Australian governments at national, state and territory levels already making significant investments in clinical and research genomics.

This section presents a short summary of activities from across the Australian genomics landscape, major areas shown in the figure below.

![Figure 2: Genomics data in Australia](image)

Public health and genomics

Organisations at Australian, state and territory levels are working to develop the sustainable application of genomics in clinical and research settings. The following summarises these activities and directions of the states and territories (listed by population) and the Australian Government at the time of writing.

**Australian Government**

The publication of the *National Health Genomics Policy Framework* (the Policy Framework) [1] and its supporting documents, endorsed by the Council of Australian Governments Health Council illustrated the importance that governments across Australia placed on establishing national agreements on the use of genomics and the management of genomic data in the public health sector. This was further supported by funding for various projects under the *Implementation Plan*,

The *Framework* identified five key strategic priority areas that required increased national efforts:

1. a person-centred approach to integrating genomics into health care
2. building a skilled workforce
3. the need for sustainable and strategic investment to make genomics cost-effective
4. maximising the quality, safety and clinical utility of services
5. the responsible collection, storage, use and management of genomic data.

*Genomic data in Australia - An industry perspective on clarity, certainty and standardisation*
Addressing this last point, the *NAGIM Blueprint* proposed a staged implementation framework, acknowledging that the public health sector includes organisations at different levels of digital maturity and progress towards genomic technology and use. While the public health departments in some jurisdictions are well advanced in using genomics for diagnostic testing, this is not universal. This reflects the reality of differing populations, health priorities and funding capacities across the states and territories.

The Australian Government has a central role to enable to support a unified approach to genomic data in Australia through:

- legislative and regulatory changes that support privacy and consent mechanisms while still encouraging appropriate data sharing
- funding arrangements, including to research into genomic data standards and practices
- leading collaboration between state and territory public health bodies into harmonised approaches to dealing with genomic data
- working with industry to ensure that other activities are pragmatic, implementable and consistent with international practices.

Factors that will influence the development of this approach include:

- the timeframes on the *Policy Framework* and the *Implementation Plan* [1] coming to a close and the potential to refresh these documents
- funding by the Australian Government to support Australian Genomics to undertake implementation projects arising from the *NAGIM Blueprint* [6]
- publication of the *Australia’s Digital Economy 2030 strategy* [7] which seeks to drive digital innovation in Australia
- upcoming publication of the *Commonwealth Digital Health Blueprint* [7], which positions the Department of Health’s vision for digital health in Australia
- the refresh of *Australia’s National Digital Health Strategy* [8] which is due in 2022.

**New South Wales**

New South Wales is well progressed with genomics, and has published the *NSW Health Genomics Strategy* [9] and *NSW Health Genomics Strategy Implementation Plan (2021-2025)* [10]. These documents specifically call out the need for data governance and the considerations required for the “big data” associated with large scale genomic sequencing. Key focus areas include understanding the data sharing landscape in NSW, the development of standardised genomic test results reporting and appropriate data infrastructure.

NSW Health continue to invest in maturing processes around ordering genomic tests through their enterprise systems and have significant investment in software to record family history and pedigree data. The various electronic medical record (EMR) implementations hold the clinical data, but there is limited capability to present this as phenotype data.

New South Wales Health Pathology has implemented cloud-based analysis, interpretation tools and data storage focused on clinical genomics using AWS services. The NSW Health Pathology Sequencing Service has capability for both whole exome (WES) and whole genome (WGS) sequencing. Clinicians are encouraged to use local testing services, but other external sources are used, especially for specialist tests.

NSW Health have worked closely with Melbourne Genomics (see below and on page 16) to share information on approaches, intending to move to data sharing for research as appropriate mechanisms and governance can be put in place. Their technology stack is very similar to that of the Victorian GenoVic system, although the
orchestration element present in GenoVic is absent. NSW have released a policy [11] and strategy [12] that endorse the use of public and private cloud capabilities.

NSW are involved in cross-jurisdictional investigations of communicable diseases, and the Centre for Infectious Diseases and Microbiology (CIDM) [13] are part of network of laboratories undertaking microbial genomics testing.

**Victoria**

Victoria has published their strategy for genomics in the *Genetic and genomic healthcare for Victoria 2021* [14]. Key data-related themes within the strategy included the importance of raising awareness of the use of genomic information in healthcare, including the limitations and benefits, and the establishment of trust to build public confidence in the way genomic information is handled and used.

This strategy for genomics is built upon broader digital health principles and priorities outlined in Victoria’s *Digitising health* strategy [15]. This underscores the importance of state-wide clinical data sharing and the use of deidentified clinical data to support research.

The Victorian Government has provided funding to Melbourne Genomics, focusing on delivering clinical genomics capabilities to Victoria’s public sector hospitals through the GenoVic platform. This uses AWS cloud storage and compute capabilities in line with the *Victorian Government Information Technology Strategy 2016-2020* [16].

A second round of funding announced in 2020 will extend these capabilities, including the agreements required for clinical and research data sharing. More information on Melbourne Genomics can be found on page 16.

In addition to the data held by the GenoVic platform, laboratory information systems (LIMS) hold genomic diagnostic test results. Victorian public health clinicians can order genomic tests through the Victorian Clinical Genomics Service (VCGS), or through interstate or overseas providers. Interstate ordering is most common in cases where a laboratory was known to be a centre of excellence in a specific domain (e.g. the Harry Perkins Institute of Medical Research in Western Australia for neuro-muscular diseases). Where external testing is undertaken, only the diagnostic reporting data was generally available.

**Queensland**

Queensland Health have published the Queensland’s *Statewide Genetic Health Queensland Service Plan 2017-2022* [17] which established programs of work in genomics.

Following on from Queensland Health’s investment in Queensland Genomics [18], work has been undertaken between Queensland Health and CSIRO on understanding the processes and data associated with genomic test ordering and results return to the EMR. Genetic Health Queensland operates a statewide service using the TrakGene software to record family history and pedigrees, however this is not tightly integrated with all other enterprise systems and is not available to general clinicians, limiting access to or capture of this data by groups outside the genetic counselling area. Pathology Queensland’s LIMS holds the results from local and external genomic tests but does not have access to the detailed genomic data, even if it available. Phenotype data used for ordering tests and interpreting results is largely handwritten.

Work is under way to map out the next stage of work for genomics and precision health, with publication of a roadmap for genomics and precision health anticipated soon. This roadmap should support Queensland Health’s policies for sustainable clinical genomics and the state’s approach to supporting genomics research. This will require investments in workforce, services and infrastructure.

In particular, the roadmap is likely to outline Queensland Health’s plans regarding the governance and management of genomic data to encourage the responsible use of this data for research. Pathology Queensland
is exploring cloud based data storage of genomic data for tests they perform, in accordance with the Queensland Government Cloud Computing Strategy [19].

Uniquely, Queensland Health recognises the importance of working with industry to realise the benefits of genomics, through investments in infrastructure, collaboration with researchers and industry leaders, and the use of clinical trials to translate research into clinical practice.

South Australia

Like other states, the South Australian Clinical Genomics Plan 2022 [20] outlines their approach to clinical genomics. One of the 5 priorities identified in the strategy is the collection and analysis of genomic data. Activities called out include strengthening public trust in data systems and mechanisms and collaborating with the Commonwealth and other jurisdictions on national genomics infrastructure to support collection, storage and sharing of genomic data.

In July 2020, the South Australian Genomics Centre was established to support genomics and bioinformatics research in South Australia across all disciplines from environmental, plant and agricultural research to human health. Services will include including RNA sequencing, small RNA sequencing, exome and genome sequencing, epigenomics, metagenomics, single cell genomics.

In addition, the SA Genomics Health Alliance is being formed with Australian Genomics and the South Australian Health and Medical Research Institute (SAMHRI) [22].

While use of cloud services is permitted under South Australia’s Cloud Services Policy [23], long term storage of genomic data in compliance with NATA requirements has typically been on tape.

Western Australia

The Office of Population Health Genomics Strategic Plan [24] supports the translation of genomic research into clinical genomics in Western Australia. A genomics strategy has been under development and publication is anticipated shortly that sets out a 5-year plan for genomics in Western Australia. The draft of this strategy [25] released for public consultation identified the need for meaningful partnerships and collaboration with a range of stakeholders, including private industry and the research community. The draft strategy also calls out the need to better manage genomic data in WA, and to integrate it appropriately in the health system.

WA Health provides a statewide diagnostic and counselling service through Genetic Services of WA (GSWA) [26]. Family history and pedigree data is held in several systems, but they are not integrated with other enterprise systems.

Not all hospitals in Western Australia have an EMR, and genomic reports are typically held in paper medical files, although the roll out of EMRs will eventually change this. Historically, long-term storage of genomic data was based on hard disks, but Western Australia’s recently adopted Cloud Policy [27] may see this change as part of the yet-to-be released genomics strategy.

Tasmania

While the Tasmanian Clinical Genetics Service provides clinical genomics support for the people of Tasmania, a formal strategy has not been developed and most genomic sequencing and analysis is outsourced to mainland laboratories such as VCGS or other external laboratories. No detailed genomic data is held, with only diagnostic test results being recorded.
Northern Territory
The Northern Territory outsources all genomic services to VCGS, including genetic counselling services. This reflects constrained budgets and a focus on health needs specific to the Northern Territory.

Australian Capital Territory
ACT Health’s *Digital Health Strategy* 2019-2029 [28] mentions the use of clinical genomics, although no formal genomics strategy is published.

Instead, ACT Health have funded diagnostic services for genomic testing for rare diseases through Canberra Clinical Genomics (CCG). CCG is accredited to provide this whole exome gene sequencing as a diagnostic service. It is a collaboration between the ACT Health Directorate, Canberra Health Services and the Australian National University [29].

ACT Health is currently rolling out an EMR, and historical data was in paper or PDF form only, although CCG provide an online portal. The new EMR is expected to include only test results not detailed genomic data. CCG leverages the National Computational Infrastructure for analysis and storage.

**Table 1: Summary of government public health genomics**

<table>
<thead>
<tr>
<th>Group</th>
<th>Genomics program</th>
<th>Strategy released</th>
<th>Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Australian Government</td>
<td>NA</td>
<td>Yes, due for refresh</td>
<td></td>
</tr>
<tr>
<td>New South Wales</td>
<td>Yes</td>
<td>Yes</td>
<td>Cloud-based data storage used</td>
</tr>
<tr>
<td>Victoria</td>
<td>Yes</td>
<td>Yes</td>
<td>Cloud-based data storage used</td>
</tr>
<tr>
<td>Queensland</td>
<td>Yes</td>
<td>Due for release soon</td>
<td>Data storage capabilities being developed</td>
</tr>
<tr>
<td>South Australia</td>
<td>Yes</td>
<td>Yes, due for refresh</td>
<td></td>
</tr>
<tr>
<td>Western Australia</td>
<td>Yes</td>
<td>Due for release soon</td>
<td>Data storage capabilities being developed</td>
</tr>
<tr>
<td>Tasmania</td>
<td>Yes</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Northern Territory</td>
<td>No</td>
<td>No</td>
<td>Outsourced services to VCGS</td>
</tr>
<tr>
<td>Australian Capital Territory</td>
<td>Yes</td>
<td>No</td>
<td>Outsourced services to CCG</td>
</tr>
</tbody>
</table>

**Interstate and private sample testing**

Australia has a large private pathology network providing National Association of Testing Authorities (NATA) accredited services, including in many cases genomics [30]. In 2017, out of 73 laboratories offering genetic or genomic investigations, 20 were in the private sector and 12 were in research settings [31, p. 17].

Recent Federal budgets have included funding to reimburse an expanded range of genomics tests. Adding these genetic tests to the Medicare Benefits Schedule resulted from submissions from a range of organisations including pathology laboratories, research groups and the genomics industry [32]-[34].

Besides those jurisdictions who outsource the bulk of their genomic testing to international laboratories, there is a significant level of interstate genomic testing undertaken nationally. A 2018 report by the Royal College of Pathologists of Australasia (RCPA) indicated that interstate transfers of samples for testing the 2016/17 financial...
year almost doubled to 19.8% since the 2011 period [31, p. 6]. International genomic services also represent a significant volume of testing. Between 2011 and 2016/17, international testing increased 31% [31, p. 6]. Such interstate or international transfers are typically done for reasons of specialised testing unavailable locally, or lower cost or turn-around times associated with scale of operations.

Private pathology laboratories have specific data management needs and practices, including:

- The National Pathology Accreditation Advisory Council (NPAAC) mandates minimum requirements for the storage of diagnostic materials and laboratory records [35, p. 12] which applies to the storage of genomic data. For read data and microarray analysis files, this is currently 4 years, with a 10-year retention of the variant call files. Under current levels of testing this is manageable but if genomic data testing volumes increase, especially for whole exomes or whole genomes, this will increase demands on data storage.
- Detailed sequence data is generally not returned as part of the test report. This is partly due to a lack of demand for such services, a lack of demand and possible issues around consent.
- Private testing laboratories do not make data available for other purposes (such as research) as they are not provided with consent to do so.
- Development of national genomic data management and sharing practices that were intended to involve private laboratories would need to understand the practical and commercial realities associated with large data transfers and the associated consent management.

**Australian genomic alliances**

Through a combination of public and private funding, several genomic alliances have been established in Australia to support development, translation and standardisation of genomics in research and clinical settings.

**Australian Genomics**

Established in 2016 with National Health and Medical Research Council (NHMRC) funding for an initial 5 years, in 2021 Australian Genomics was funded for a further 5 years through the Australian Government’s Genomics Health Futures Mission and other philanthropic funding. Through a network of partners and collaborators, such as hospitals, universities, research institutes and centres, laboratories and community organisations, Australian Genomics coordinates, funds and conducts genomic research [36].

By developing a national diagnostic and research network to deliver genomic testing through two flagship studies (rare disease and cancer), Australian Genomics built evidence to inform policy change. These studies also supported development of consistent national approaches for the clinical delivery of genomics, including those for patient consent, pathology reporting, and gene variant classification [37].

As the Australian Driver Project of the Global Alliance for Genomics and Health (GA4GH), Australian Genomics are piloting implementation of GA4GH standards, tools, and frameworks for responsible data sharing [38]. Building upon the GA4GH work, Australian Genomics have undertaken a program of coordinated work to develop national approaches to the generation, processing, curation, storage and sharing of genomic data [39]. This has led to their commissioning by the Australian Government to undertake further implementation of the NAGIM Blueprint [6].

Australian Genomics has partnered with InGeNA under a Memorandum of Understanding (MOU) to gain industry input into their continuing program of work [40].
Melbourne Genomics

The Melbourne Genomics Health Alliance was established in 2013 with support from the Victorian Government. It is a collaborative of 10 hospitals, research and academic organisations in Victoria, focusing on translational research that demonstrates genomics can contribute to health care that is effective and efficient [41]. The Alliance undertook 16 clinical flagship projects during its initial periods of funding, covering a range of clinical areas to support evaluation of the effectiveness of genomics in health care [42]. This program of work was extended with an additional round of funding from the Victorian Government in 2020 [43].

To support their program of work, Melbourne Genomics developed a cloud-based platform to undertake processing, curation and storage of genomic data taken from laboratory sequencing. This platform, GenoVic, is used to provide clinical genomics in four Alliance hospitals and one laboratory external to the Alliance. As a shared platform, it enables management and use of genomic data by laboratories, clinicians, patients and researchers, while still providing controls to ensure access to genomic data is limited to appropriate users [44].

Through their clinical flagship projects, a data sharing system called DASh, and the GenoVic platform, Melbourne Genomics have supported development of data sharing agreements and approaches that ensure reliable and efficient access to participant genomic data [45], [46].

Melbourne Genomics have delivered the GenoVic cloud-based analysis, interpretation and data storage system. This was delivered through partnership with industry. The system is based on Amazon Web Services (AWS) cloud technology and the Fast Health Interoperable Resources (FHIR) standards. The system supports the use of third-party bioinformatic pipelines (for example those from Illumina) as well as custom platforms developed by specialist groups. A partnership with industry has delivered enhancements to the Alissa Agilent interpretation software that forms part of the GenoVic solution.

The initial focus for GenoVic was to deliver exome and genome sequencing capabilities to support clinical genomics in Victoria’s public sector hospitals. To achieve this, Melbourne Genomics have undertaken work on data governance and clinical data sharing agreements and worked with hospital stakeholders to uplift capabilities in these areas and will be exploring research data sharing.

Queensland Genomics

In 2020, Queensland Genomics completed a 5-year program of work funded by Queensland Health. Three rounds of funding supported clinical projects and capability initiatives within Queensland Health to build:

- a workforce that incorporates genomics into health care
- an evidence base for clinical genomics
- timely and cost-effective diagnostic workflows
- genomic sequence results that are used to benefit patients
- public awareness and understanding
- a system for managing clinical genomic data
- accelerated research translation
- a positive contribution, nationally and internationally.

In addition to the work above, Queensland Genomics worked with CSIRO to deliver the National Approach to Genomic Information Management project which delivered a Report [3] to government and a Blueprint [4] which Australian Genomics will now carry forward into a number of implementation projects.
A key element of the Queensland Genomics project was establishing evidence for the use of clinical genomics to support a sustainable genomics program in Queensland. The ongoing management of genomic data management and capabilities has transitioned to Queensland Pathology [18].

**Sydney Genomics Collaborative**

Founded in 2014 with funding from the New South Wales Government, the Sydney Genomics Collaborative boosted genomic research across New South Wales into inherited diseases and disorders with a genetic component, including cancer [47]. The Collaborative projects included:

- a reference bank containing approximately 4000 whole genome sequences from healthy, aged people for control purposes in disease-specific genomic research
- funding for researchers to undertake whole genome sequencing
- a research program focusing on application of genomics to the understanding, early detection, prevention, and management of cancer.

**Table 2: Summary of data related aspects of genomic alliances in Australia**

<table>
<thead>
<tr>
<th>Group</th>
<th>Currently active</th>
<th>Data specific interests</th>
<th>Relationship</th>
</tr>
</thead>
<tbody>
<tr>
<td>Australian Genomics</td>
<td>Yes</td>
<td>Federated data repository, dynamic consent management, research data sharing agreements, genomic data standards, NAGIM implementation</td>
<td>MOU</td>
</tr>
<tr>
<td>Melbourne Genomics</td>
<td>Yes</td>
<td>Clinical data sharing agreements, cloud-based data repository,</td>
<td>None</td>
</tr>
<tr>
<td>Queensland Genomics</td>
<td>No</td>
<td>NAGIM Blueprint and Report</td>
<td>NA</td>
</tr>
<tr>
<td>Sydney Genomics Collaborative</td>
<td>No</td>
<td>Reference repository</td>
<td>NA</td>
</tr>
</tbody>
</table>

**Current and evolving state of technology**

Technology underpins all parts of the genomics value chain described on page 7 and has seen major improvements over the last 20 years [48].

**Sequencing and other technologies**

While Next Generation Sequencing (NGS) receives much of the attention when discussing genomics, improvements in Sanger sequencing continue to allow it to play an important role in genomic sequencing today [49]. Additional technologies including using nanopores and long read technologies promise further improvements in coming years [49]–[51].

Cytogenetic testing, including fluorescent in situ hybridisation, continues to be a valuable tools for chromosome studies [52]. Recent advances in microarray testing now allow for detection of copy number variants in chromosome not normally discernible using traditional optical techniques, such as microdeletions or microinsertions. The speed with which microarray testing can be undertaken is considered a key advantage [53], [54] and they can be used for population studies, studying RNA, drug sensitivity testing and oncology testing [55].
Storage and analysis of genomic data

As clinical genomics becomes more mainstream, the volume of data stored in clinical settings will increase to meet the National Pathology Accreditation Advisory Council (NPAAAC) requirements of retention of clinical data [35], and this will stress existing data storage capabilities. Cloud storage is one of the available options to address this need [56]–[58].

Bioinformatics pipelines for the analysis of these datasets have traditionally required access to high performance computing facilities. However, application of cloud technologies for both data storage and execution of bioinformatic pipelines has increased in recent years and are now available from a wide range of providers and for both research and clinical applications [59]–[64].

Curation and interpretation of genomic data

While the analysis phase is largely automated to identify variants of interest, the curation and interpretation of these variants has traditionally been significantly more manual. In addition to significant knowledge and experience, curation and interpretation of genomics requires access to contemporary resources (such as academic publications) and datasets to support clinical diagnosis and research interpretation.

However, knowledgebases can vary in content, structure and their use of primary literature, which can limit their use in curation [65]. This has led to the development of meta-knowledgebases that harmonise the disparate data. Another approach is the “crowd sourcing” of curation and interpretation of genomic variants, such as that used by the Shariant platform made available by Australian Genomics. This provides shared scientific evidence about clinically curated variants [66].

Emerging technologies and approaches

To support genomics researchers and variant curators, the application of artificial intelligence (AI) and machine learning (ML) algorithms is increasing being applied. The identification of variants (variant calling) is one area in which algorithms and AI techniques can produce results equal or superior to traditional methods. For instance, the DeepVariant variant caller, relying on deep neural networks, has been demonstrated to outperform other methods [67], [68]. Neural network based techniques are also believed to improve basecalling, the translation of electrical signals from sequencing technology to determine nucleotide sequences [67], [69].

The classification of coding variants is another area in which AI/ML techniques are gaining prominence. Australia’s Australian eHealth Research Centre (part of CSIRO) have developed a cloud-based machine learning platform called VariantSpark to support association analysis of polygenic phenotypes [70], [71]. Similarly, CancerVar [72] is applying deep learning approaches to support clinical interpretation of somatic variations in cancer. Significant investment in AI/ML for genomics is occurring internationally, and this is an area of opportunity of industry participants [73], [74].

AI-based national language processing can also be applied to analysis and data extraction of phenotypic descriptions from clinically relevant information in electronic health records [75] to support identification of candidate pathogenic variants for individuals with rare disease.

Despite the potential for advances in this area, AI and ML are challenged or limited by several factors, including:

- the challenges of achieving regulatory compliance for the underlying algorithms for use in clinical settings
- a lack of transparency in the logic used in so-called “black box” algorithms
- bias called by the data used to train algorithms or other systemic factors
These concerns underly government and academic activities to identify and address the uses of AI/ML in many fields, including clinical applications. Relevant activities to address these factors include a report by the Australian Human Rights Commission and others [76] and the publication of Australia’s AI Ethics Principles by the Department of Industry, Science, Energy and Resources [77].
Challenges and opportunities

While the adoption of genomics in Australian health care is proceeding, the rate of adoption has not been as fast as considered optimal [5]. The governance and management of genomic data is one of the factors that influences adoption. However, with these challenges come opportunities to improve the rate of adoption and the value returned to the health sector through genomics.

In this section, we will explore some of these challenges and opportunities evident in academic literature and through survey and interviews with project participants from both genomics industry and healthcare sectors. The figure below notes the headline areas in the discussion.

![Challenges & opportunities](image)

**Data for precision medicine**

One of the repeated themes in the workshop discussions was data discoverability. While the GA4GH predicts that clinical genomics will not only replace research projects as the majority source of genomics data, it will increase the overall volume of data available [78]. This will lead to several new challenges.

**Data classification**

In the NAGIM Blueprint [4, Sec. 3], “genomic data” depends upon data types traditionally considered part of non-genomic systems, such as clinical and administration data. The richness of this clinical and other data is part of the attractiveness of clinical genomics, as the genomic sequence data can be correlated with phenotype data derived from clinical systems. However, this same data is collected for clinical and funding purposes and may not be sufficiently granular or coded appropriately to support the use of phenotype-genotype correlation.

**Opportunity:** To increase the value of clinical data for genomics, improvements are necessary in the way clinical systems capture and code phenotype data. Australian Genomics have several projects looking at standardisation of phenotypes in the Australian healthcare setting. InGeNA members may be able to support aspects of these projects.

**Data discoverability**

The increase in available data conversely increases the difficulty in locating data unless metadata is available to support discoverability. Survey respondents and workshop participants note that while the provision of genomic data for research was desirable and necessary, making clinical data “research ready” was time consuming, resource intensive and not core business. One of the key challenges is therefore how such data discoverability efforts can be funded to support downstream research use.

The NAGIM Blueprint [4, Sec. 5] proposed a federated approach to national genomic datasets. This would allow state and territory health organisations to manage their own clinical data (including clinical genomics). However,
this also means that any approach to metadata supporting data discoverability needs to be consistent across all jurisdictions to support discovery of genomic data at a national level.

**Opportunity:** A standards-based approach to metadata capture may reduce the burden of making clinical data ready for research and data discoverability. The Australian Government is keen to leverage investments in research and clinical settings and can provide leadership in supporting cross-jurisdictional standardisation efforts. Australian Genomics is planning projects to promote policies and procedures that support secondary use of data. InGeNA can work with both groups to support a national approach to genomic metadata.

**Challenges of federating research data**

Another challenge of federating research data lies in the diversity of research data sets themselves. One lesson learned through the Melbourne Genomics program was the complexity of retrospective data retrieval and transfer processes when collating data from historic research data sets which may include unstructured data or data coded in diverse ways [46]. Differing consent models (see below) can also limit the reusability of data, even if these obstacles can be overcome.

**Opportunity:** A standards-based approach to clinical and genomic data capture may reduce the burden of providing research data for federated use. Implementation of aspects of the NAGIM Blueprint by Australian Genomics including pilot data implementations may offer InGeNA the opportunity to support these pilots to gain understanding of the standardisation process.

An alternative to bringing research from federated data sources into a national research repository is to bring the computational resources to the data. So-called compute-to-data approaches require the data custodian to execute algorithms/pipelines on compute infrastructure (often cloud-based) associated with their data. Several proponents of this approach are looking at the commercial models associated with this, such as data markets. While not without challenges, this approach is gaining interest [79]-[81].

The next step is to use federated analytics to allow for the execution of queries across disparate cloud data storage and compute capabilities is an approach that some believe can reduce costs, increase security and improve privacy over data [82], [83]. Using federated analytics to provide privacy protection is used by several organisations, especially those providing access to real world data for clinical trials [84], [85]. However, these techniques are not applicable to all types of analysis.

**Opportunity:** Exploring the further use of federated analytics and compute-to-data capabilities may allow the industry to inform the approaches needed to support national genomics research while maintaining privacy. Working with industry organisations that currently use capabilities in this area may inform further investigations.

**Integration of clinical data**

The NAGIM Blueprint outlined the importance of combining clinical data with genomic testing [4, p. 44]. With the increasing interest by public health services in the use of genomics to support clinical decision making, the potential for rich, high-quality clinical data and whole exome or genome sequencing data (amongst others), there is a significant data potential if this data can be made available for research and use in commercial product or solution development.
Opportunity: As Australian Genomics has carriage of the work to look at implementations of the NAGIM Blueprint, InGeNA should actively engage with those projects to ensure that the requirements and skills of the industry are applied to implementation planning and activities.

Possibilities for use of this data include development of better algorithms and pipelines to support analysis of genomic data and development of novel software applications to analyse or visualise genomic data.

Opportunity: The establishment of a national capability that included clinical and genomic data provides opportunities for innovation. While this can be leveraged by large industry participants, it also represents an opportunity for InGeNA to encourage start-up participation through mentoring or other incentives to make innovative use of such a data capability. One example might be an incentive program or a national challenge to target a specific issue. Partnering with one or more universities or groups like the Digital Health CRC may be beneficial in this regard.

As noted earlier, the options to support curation and interpretation of genomic data are anticipated to be enhanced using AI and ML. The development of a national data capability combining genomic and clinical data provides an option to ensure that algorithms are trained using a broad set of data to reduce bias and increase the transparency of algorithms.

Opportunity: As part of any implementation of the NAGIM Blueprint by Australian Genomics, consideration of the use of this data to support AI and ML technologies in line with Australia’s Artificial Intelligence Ethics Framework [77]. InGeNA role should include providing Australian Genomics with advice from industry about requirements for AI and ML in this area.

A national genomic and clinical data capability also provides a basis for the development of clinical evidence at scale to support the health economic evaluations and the adoption of specific genomic testing approaches and technologies within the healthcare sector.

Opportunity: InGeNA’s Data and Technology Innovation Working Group should liaise with other working groups and Australian Genomics to establish a process for leveraging the national capability to deliver evidence to government that demonstrate the economic benefit to the health sector to support improved sustainability.

Privacy, consent and data sharing
While data-rich digital technologies such as genomics offer significant benefits to health care, they also strain traditional data governance concepts such as consent and privacy, and if incorrectly addressed, can erode public trust in such technologies [86, pp. 27–29].

Understanding genomic privacy
The key challenge of privacy in genomics is generating valuable clinical insights without disclosing personal genome data. While data sharing for research is usually based on removing personally identifiable information from the dataset, it can be argued that genomic data cannot be completely anonymised due to its inherent uniqueness to an individual. This is concerning for individuals, such as those with rare diseases, who may be concerned about genomic discrimination [87, Ch. 16 p. 3], [88, p. 2].
The use of clinically derived data for research and commercial applications is an issue that has caused much debate in Australia and elsewhere. The use of de-identified data from Australia’s My Health Record system “solely for commercial and non-health related purposes” has been explicitly excluded [89] following debate during the opt-out period in late 2018 [90]. Despite this, availability of de-identified data for approved uses is still not available [91]. It is anticipated that the storing of genomic data and the potential uses of it will also cause debate [92].

**Opportunity:** InGeNA is uniquely positioned to engage in the debate about the commercial use of genomic data. A position paper that describes the nature of use and safeguards may hold address some of the concerns likely to be raised by the public and media. This would be specifically relevant to those organisations with an interest in clinical trials and research.

Privacy-preserving technologies including the use of federated queries may reduce the risks associated with the re-identification of patients via their genomic data, however, may not be suitable if access to the raw data is required. Organisations such as IQVIA and TriNetX already provide access to networks of clinical and genomic real world data [84], [85].

**Opportunity:** Adopting a standardised approach to structuring and operating federated query systems in some circumstances may allow participating data sources to preserve the privacy of their data while supporting the research outcomes possible from the broader data environment. There may be opportunities for organisations with existing technologies and approaches to engage with (or join) InGeNA to support such investigations.

### Compliance regimes

Australia has a complex set of legislation and regulatory compliance requirements operating at both national and state/territory levels.

At a national level, these include:

- **Privacy Act 1988** (Privacy Act) and Australian Privacy Principles (APPs)
- **National Health Act 1953**
- **National Health and Medical Research Council Act 1992**
- **National Health Reform Act 2011**
- **Therapeutic Goods Act 1989**
- **Healthcare Identifiers Act 2010**
- **My Health Records Act 2012**

Many states and territories also have health and privacy related legislation, such as:

- **Privacy and Personal Information Protection Act 1998** (NSW) and **Health Records and Information Privacy Act 2002** (NSW)
- **Privacy and Data Protection Act 2014** (Vic) and **Health Records Act 2001** (Vic)

The compliance requirements are not static, and recent activity in the Australian legal ecosystem include the Data Availability and Transparency Bill [93], the Australian Competition and Consumer Commission (ACCC) Consumer Data Right project [94], the Australian Data Strategy and the Digital Atlas of Australia announced as part of the **Digital Economy Strategy 2030** [7], and the **Australia’s Artificial Intelligence Ethics Framework** [77].

Groups such as Australian Genomics have undertaken reviews of Australian legislative and regulatory requirements [95].

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**Genomic data in Australia - An industry perspective on clarity, certainty and standardisation**
Besides Australia’s requirements, organisations working with international participant data may also be subject to other controls. The European Union’s General Data Protection Regulation (GDPR) [96] applies to data for European Union citizens. While similar to the Australian Privacy Principles, GDPR includes additional restrictions yet to be tested in Australia [97].

**Opportunity:** While each organisation must ultimately make their own assessments on legal compliance, a contemporary review by InGeNA of the Australian legal and regulatory environment for genomic data would assist InGeNA members and other organisations operating with genomic data. Such a review would also inform any InGeNA position regarding introduction of or changes to data privacy regulation in Australia.

**The impact on consent**

While sharing genomic data for clinical and research purposes can deliver value for the healthcare system through better prevention, diagnosis and treatment [88], the nature of this data and how it is interpreted is challenging existing models of consent because of the reliance on of secondary use of data from larger cohorts to support clinical interpretations of genomic data [87, Ch. 13 p. 2], [98, p. 33].

Research in Queensland [99] suggests that most study participants expected that genomic data collected within the public health system would be made available to researchers as long as consent had been sought at least once. Current Queensland legislation does not match these expectations, and it is likely that similar situations occur throughout Australia. The nature of genomic data also means that while consent from a person for use of their genomic data may be obtained, information about family members of that person may be indirectly determined [88].

Workshop participants noted that existing fragmented patient consent approaches, such as differing consent forms between jurisdictions and between service providers, limited interoperability and data sharing for research purposes.

**Opportunity:** Australian Genomics have planned continued work on consent and their dynamic consent platform (CTRL). Engagement with Australian Genomics in this work would allow InGeNA to bring national and international experience to the table, and support moves to a national approach to managing consent across public, private and research genomic data.

**Who defines consent?**

The subject of consent is broad, sometimes difficult to define and frequently emotional. It also frequently becomes overly technical and burdened with legal jargon to limit obligations.

When asked who should be engaged in processes to define and explain consent, survey respondents included a range of participants, including:

- governments
- consumers
- clinicians
- researchers
- ethicists

While this provided a broad and balanced discussion of the subject, consumer representatives were keen to see any discussion of consent and data sharing stay focused on the importance of data sharing to research and...
diagnosis of genomic diseases. There was a concern expressed by one participant that sometimes data sharing may be constrained by privacy experts remote from the need of patients undertaking diagnostic odysseys.

**Emerging models of consent**

Consumers were also keen to be given the choice about the level of involvement they might wish to have with genomics research undertaken with their data. This might range from nothing at all to being informed of the outcome of specific research programs. They also noted that they reserved the right to alter their position (although noting this would only apply to subsequent use of the data).

This reinforced the need for a consent model that can be adaptable to a range of requirements and support ongoing engagement. Typically referred to as dynamic consent, it can improve trust with some consumers, but also requires technology platforms to allow consumers to engage with the data custodians [100]. The CTRL model being developed by Australian Genomics is an example of a platform that allows consumers to engage (or not) with research using their data [101].

**Opportunity:** There is a strong appetite for further consultation and consensus development with health consumer groups on consent around genomics data. Such work would inform approaches that build fine-grained real-time consent to let patient cohorts calibrate their data sharing preferences more actively. As Australian Genomics have a project to continue development of consent models, the opportunity exists for InGeNA to be engaged in this work.

Building on dynamic consent, a growing area of research and industry interest is the application of blockchain technologies to genomic data to support consent (and monetarisation) of biobanks and genomic data by the consumer or the research organisation providing access to data [79], [100], [102]–[104]. While blockchain is commonly associated with financial markets, the blockchain’s distributed ledger is designed to provide increased security, boost trust and ensure transparency of transactions, which can equally apply to the use and management of data [100], [102].

Some blockchain models are focused on managing access to data, while others, such as Shivom [105], promote data sharing through the incentivisation of data owners to share their genomic data [104]. However, blockchain technologies are not the only method of securing genomic data, and involve a level of overhead and complexity that has come into question [106].

**Opportunity:** An InGeNA sponsored review of blockchain technologies related to genomic data sharing may help inform public and industry debate around the application of such technologies to increase transparency and security of genomic data considering issues associated with blockchain.

**Private insurance and genomics**

While the benefits of genomic testing are established, there remains public concern regarding the impact genomic testing may have on insurance products [107].

In Australia, the *Private Health Insurance Act 2007 (Cwlth)* prohibits discrimination by private health insurers [107]. The existence of a genomic indicator of a potential illness where there are no symptoms does not constitute a pre-existing condition, and hence waiting periods will not apply [108].

However, this is not the case with risk-rated insurance products such as life insurance. In 2019, the *Moratorium on Genetic Tests in Life Insurance* came into effect. The moratorium is in place until at least 30 June 2024 and
constrains life insurance companies from using genomic test results as part of an insurance application up to predefined limits. This is inclusive of genomic testing undertaken as part of research projects [107], [108].

The moratorium does not apply to other forms of risk-rated insurances, including travel insurance, and sickness and personal accident insurances.

What happens after the period of the moratorium is yet to be decided in the lead to June 2024. Concerns remain over the implications the end of the moratorium may have on clinical and research application of genomics despite the value they represent to the health system [107].

**Opportunity:** Supporting an ongoing discussion on the implications of private insurance use of genomics may reduce concerns about the long-term impact of involvement in genomic research. Continued engagement by InGeNA with the Australian Government in this regard is advisable.

**Direct-to-consumer applications**

In response to increasing interest by the public in genomics, there is a growing number of Direct-To-Consumer (DTC) genetic tests available in Australia, frequently via online channels offered by international laboratories. Many are available without a medical referral and hence sit outside the Australian health system [109], [110].

As many of these laboratories are outside the accreditation requirements of the Therapeutic Goods Administration (TGA), they may not be held to the same standards of evidence and process required here in Australia. Concerns voiced in Australia include [109]-[111]:

- the quality control of these laboratories cannot be assessed by regulators which may place the accuracy of results in question
- the level of evidence for tests may not meet Australian standards
- a lack of suitable evidence may set unrealistic community expectations and so undermine confidence in clinically valid genetic tests
- consumers who have received results may present to Australian General Practitioners and genetic services looking for interpretation of the results
- results are frequently provided with little or no pretesting genetic counselling or medical advice
- consumers may engage with DTC testing for so-called recreational purposes but learn of incidental findings in the absence of counselling
- protocols for the retention of samples and associated data vary between DTC laboratories and may not be within the control of the consumer
- the sale of genetic data to third parties, in some case without the knowledge of consumers, has been raised in several instances.

While some Australian DTC laboratories work to the standards required by the TGA and address these issues, and some ensure that the data provided by customers remains private, the lack of control over others and international laboratories remains a concern.

**Opportunity:** An InGeNA sponsored development of a voluntary DTC ‘code of conduct’ would allow these services to indicate their data management practices of consumer data against a set of guiding principles. An information campaign about the code would better inform customers of DTC products in how their data will be used. A voluntary code may be a precursor to a more formal accreditation regime if appropriate.
Data governance

The NAGIM Blueprint [4, Sec. 6] outlined a range of factors that need to be considered regarding governance of genomic data. These included:

- the importance of establishing governance structures over genomic data
- the lifecycle of genomic data, and the impact of data retention requirements
- data sovereignty issues and data ownership versus data custodianship, especially for Aboriginal and Torres Strait Islander peoples
- the aspects of consent across clinical and research domains
- managing the privacy and security of genomic data
- the need for data sharing in clinical and research settings
- understanding the quality aspects of data
- and the importance of metadata to record provenance and support discoverability.

Beyond this, the clinical utility of incorporating genomics and diagnostic data into decision-making requires additional governance and presents unique challenges.

A framework for data governance

While organisations handling genomic data are establishing data governance practices over the assets they manage, there is no national framework for such data governance. This requires organisation to assess their legal and regulatory requirements individually and establish policy and processes despite the likelihood there are several repeatable patterns that could inform organisations and streamline the process.

Survey respondents all agreed there was value in establishing a national framework that could provide consistent guidelines for organisations.

**Opportunity:** While each organisation must ultimately make their own assessments on compliance and data governance, a national framework that outlined guidance across a suite of implementation patterns would encourage more consistent and robust data governance. As Australian Genomics is currently looking at some areas of this, continued engagement with them would be advisable.

Data security

The sensitivity and inherent identifiableness of genomic data requires the maximum protection possible to avoid eroding public trust.

While security standards for health-related data are mandated under the Health Insurance Portability and Accountability Act of 1996 (HIPAA) Security Rule [112], these only apply to organisations operating the USA. The Australian Privacy Principles do not mandate explicit security standards or practices.

Despite this, survey respondents and workshop participants felt that security was manageable under generally accepted standards such as the ISO 27000 series and other related standards. Including standard security constructs in cloud-based data storage and computation was cited by several as the attraction for these, although participants also note that the responsibility still lay with the organisation using the services to ensure controls were in place.
Opportunity: A lightweight suite of tools to self-assess data security, coupled with some pragmatic guidance to support improvements could improve the understanding and use of appropriate cybersecurity for genomics data. This is an area in which the Australian Digital Health Agency (ADHA) has previous experience and InGeNA could partner with ADHA to support the development of such tools.

Promoting data sharing

The challenges outlined above have a detrimental effect on the ability of organisations to share clinical and genomic data. Healthcare is a conservative field by nature, and where doubt exists about legal, compliance, or consent, the default position is frequently to not share data. This is despite many healthcare organisations recognising the need for and explicitly promoting data sharing.

Organisations including the GA4GH [113], Australian Genomics [39] and Melbourne Genomics [45], [46] have developed a range of data sharing agreements on which to base interorganisational arrangements. However, these need to be examined in local and national legislation.

Workshop participants noted that a national framework for genomic data sharing must support appropriate policy and regulation development.

Opportunity: Co-development with Australian Genomics of a national framework for genomic data sharing will support transparent conversations about the subject to better inform policy and regulation development. This would ensure that an industry lens on data sharing is included in work planned by Australian Genomics.

Sovereignty and data ownership

While legislation applicable to health data constrains the hosting of that data to Australian locations, deidentified data used in research may not be so constrained. Genomics is a science that frequently benefits from international collaborations. When considering data sharing agreements and governance frameworks, it is important that considerations for cross-border data sharing are factored into these documents.

One area of concern related to the rights associated with samples processed internationally, and which fall outside Australian jurisdictional control. To address these issues, contractual arrangements need to be in place with international laboratories.

During the engagement for this project, the role of data ownership and intellectual property appeared to be an important factor. However, survey respondents and workshop attendees almost universally considered that intellectual property was held in the processes for manipulating, analysing and interpreting genomic data, rather than in the data itself.

Opportunity: InGeNA could sponsor or develop a position paper on the limits of intellectual property rights over genomic data may be beneficial to conversations around consent and consumer engagement. This would need to address the issue of IP associated with samples processed internationally and the associated contractual arrangements required. Engagement with the pathology sector including the Royal College of Pathologists of Australasia would be advantageous.

While the workshop participants were less concerned about data ownership, there are increasing moves by some to recognise the ownership of genomic data by consumers, and the monetisation of these ownership rights. Using blockchain technologies is one mechanism suggested to allow consumers to control access to their data [114].
Reuse versus resequencing

Reuse of genomic data is an important way to leverage the value of data for both patient diagnostic purposes and for research.

One advantage of whole genome or whole exome sequencing of germline samples is that once sequenced, the resulting data can be reprocessed or reanalysed. If initial diagnostic testing of specific genomic markers is unsuccessful, additional testing can be undertaken using the same data as no additional cost for sequencing. The data may also be reprocessed as bioinformatic pipelines are improved, or as variants of previously unknown significance are identified, diagnosing a patient where previously this may not have been possible.

Research datasets of genomic and phenotype data also support genome-wide association studies (GWAS), allowing for the identification of markers that may predict a disease. As the samples have been sequenced, there is no additional sequencing cost regardless of how often the data is used.

These factors have been especially important while the cost of sequencing has been high and time consuming. As technology improves, the cost to store and retrieve data may eventually exceed the cost to sequence a patient again. However, the costs of data storage have also been falling. Making this issue of limited concern.

**Opportunity:** Monitoring the evolution of both genomic and storage technologies will be required to ensure the balance between them supports the sustainable application of genomics. Working with the InGeNA working group that is quantifying the benefits of genomics would inform both work programs. Engagement with leading cloud vendors with genomics platforms would ensure currency of this information.

Interoperability

The genomics sector makes use of highly sophisticated data manipulation and analysis tools. The velocity of technical innovation also means that the market is not yet mature, as evident by interoperability challenges that exist.

Survey respondents and workshop participants noted a lack of interoperability between electronic medical record (EMR) systems, laboratory information management systems (LIMS) and systems designed to support genomic analysis and interpretation make the flow of phenotype data from clinical systems and the return of reporting challenging in many instances. These challenges are typical of the broader digital health sector.

In the earlier environment scan, state and territories are already moving to implement genomic data storage and computation capabilities. Historically, the state and territory jurisdictions have developed solutions for clinical systems independently, driven by existing jurisdictional legislative restrictions, operational requirements and funding models for public health systems. Workshop participants noted that as genomic data processing is increasingly moved to cloud compute, different organisations will probably select different compute and storage solutions, which may complicate data sharing.

Different system implementations also increase varying standardisation of coding systems and data standards, which will further compound the challenges of data sharing and establishing national research capabilities.

Workshop participants also expressed concern around the level of interoperability between legacy clinical systems such as laboratory management systems and electronic medical records and systems used to undertake genomics. Clinical information was not standardised and not always digitised in machine-readable formats, and this was hindering data aggregation and linkage to other systems and for research.
Opportunity: The endorsement by InGeNA of key standards around data structures and taxonomies may improve interoperability and provide greater certainty for the genomics sector. Development of a list published on the InGeNA website would increase visibility and provide a common position. This would also provide a platform for further engagement with the Standards community and the Australian Government.

Adoption of standards

Cloud vendors are increasingly addressing the need for interoperability and data sharing by the adoption of standards [61]. Groups such as the GA4GH are working with researchers and clinicians worldwide on developing standards to support genomics. However, as in many areas of health, adoption of standards varies.

Standards maturity in genomics

The rapid pace of changes means that some standards are not yet mature or commonly adopted. This is a problem that the broader digital health sector has faced for many years, with several standards competing for acceptance, including:

- HL7 V2
- HL7 Clinical Data Architecture (CDA)
- HL7 FHIR
- openEHR
- Observational Health Data Sciences and Informatics (OHDSI)

Workshop participants agreed that it was not the role of industry to independently develop new standards, but to engage with the broader standards community supporting developing internationally agreed approaches.

There are already a variety of standards being developed in the genomics space, including by GA4GH, FHIR and Observational Health Data Sciences and Informatics (ODSHI). While some of these standards appear to be converging [115], selecting the correct standard is a challenge for all industry participants.

It was noted however, that in some areas the pace of development was such that some standards were not yet mature enough for endorsement, and this might constrain innovation. However, by communicating these standards, the industry would usefully inform sector participants.

Opportunity: Workshop attendees suggested that rather than developing standards, there was an opportunity for InGeNA to clearly articulate which standards were being collectively endorsed. Such endorsement could be placed on the InGeNA website. Working with the Commonwealth in this regard may also be beneficial. See previous opportunity point as it regards interoperability.

Evolving synoptic reporting standards

Standards in genomics are evolving and have not yet reached maturity in all areas. One such area relates to synoptic reporting.

Synoptic reporting presents pathology data in value pairs instead of (or in addition to) narrative reporting. This approach improves the standardisation of diagnostic testing criteria, the terminology used and the interpretation of data, especially by automated systems [116].

Several survey respondents and workshop participants noted that the lack of standardised synoptic reporting was one impediment to widespread adoption of clinical genomics. Using synoptic reporting standards can [116]:

Genomic data in Australia - An industry perspective on clarity, certainty and standardisation
- reduce the variance in wording selected by individual pathologists
- result in reports largely self-explanatory to clinicians with some familiarity of the field
- facilitates standardised coding systems such as SNOMED-CT and LOINC.

**Opportunity:** InGeNA can engage with leading pathology groups, professional bodies such as the Royal College of Pathologists of Australasia, and the Standards community to support the development of more consistent synoptic reporting capabilities for genomic data to increase interoperability and improve communication and discoverability of results.

### The cost of collaboration

The establishment of InGeNA provides a unique opportunity for the industry body to engage with government, academia and the broader genomics industry. The establishment of key agreements with these groups allows InGeNA to establish itself as a voice for the industry, articulating an agreed position across a variety of topics, as outlined in this report.

However, it should also be recognised that such collaboration comes at a cost. Time and effort are required on the part of the members to establish the agreed positions described above, and investments of either time, money or resources in various projects may be considerable. This last is an important factor, as the health sector has a long history of “pilotitis” [117], as noted by Dr Victoria Wade who described telehealth as having “more pilots than Qantas”[118]. Without a plan for further adoption beyond the initial investment, many pilots or similar investments stall once the initial work is completed.

It is important that all collaborative efforts undertaken by InGeNA consider the costs associated with collaborative efforts.

**Opportunity:** InGeNA should establish a policy around collaborative efforts to ensure that investments made by InGeNA can be assessed for the value and costs associated with them and plans for further adoption considered as part of the collaboration decision.

### Considering diversity

While the term diversity is usually refers to the mix of people in a workforce or social context [119], diversity particularly applies to genomics. Applying genomics requires a standardised reference against which to identify genomic variations that may cause illness in a person [120].

The international reference genome (GRCh38) comprises data from a few people, and 70 per cent is from a single individual. This lack of diversity results in missing data and some data that is not representative of common forms of genes. It does not capture the diversity of a worldwide population [121]–[124].

In an Australian context this results in specific challenges. We have an ethnically diverse population with one in four persons born overseas and 46 per cent having at least one parent born overseas [125]. Further, the long history of human occupation in Australia, large land area and relative isolation have resulted in distinct genomic variation within the Aboriginal and Torres Strait Islander peoples [120].

This diversity means that analysis of genomic data against the international reference genome complicates analysis and interpretation of genomic testing. The National Centre for Indigenous Genomics (NCIG) is embarking on a project to build a reference genome for Aboriginal and Torres Strait Islander peoples to address at least some of this challenge [120].

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**Genomic data in Australia - An industry perspective on clarity, certainty and standardisation**
These efforts are important when considering the use of machine learning and artificial intelligence to support analysing, curating and interpreting genomic data. Unless the data used to train such systems represents human diversity, bias may be introduced into results [67].

**Opportunity:** Continued support for initiatives that improve the diversity of genomic data will support reduced efforts to curate and interpret genomic variants, especially when algorithm processes such as machine learning and artificial intelligence are used. InGeNA might consider approach the NCIG to discuss options for supporting their work in diversity.

**Resources**

Given the range of organisations within the genomics industry, there is a unique opportunity for the industry to provide or co-develop resources useful to the sector. This section outlined some resources identified during this project.

**A common language**

One challenge in communication of highly technical subjects such as genomics is the language used by those engaged in the science. Several challenges were identified by workshop participants, including:

- inconsistency in using genomic terminology between different parts of the sector and internationally
- lack of familiarity of some aspects of genomics terminology that limit the ability to communicate concepts with other clinicians
- genomic terminology can compound the challenges of communicating with patients, carers and the broader community.

While this applies to the broader subject of genomics, it is important when discussing genomic data. The NAGIM Blueprint [4, Sec. 3] has provided a draft data classification framework to support clearer communication. However, this classification framework needs additional validation and refinement, especially in areas outside NGS.

**Opportunity:** Refinement of the genomic data classification framework can support more informed data governance conversations by clearly identifying the type of genomic data under discussion. InGeNA can engage with Australian Genomics on their projects to trial aspects of the NAGIM Blueprint.

**Education materials**

Participants in the InGeNA workshops were very supportive of efforts to leverage existing industry knowledge and experience, often developed within larger industry organisations, to support better communication and education with the broader sector. This was true when considering the consumer groups,

However, further engagement through the workshop series suggested any such work for clinicians should be done in collaboration with and support of the colleges and professional bodies, as these are the most appropriate channels for working with clinicians.

Consume groups, by comparison, are frequently operated by volunteers and poorly funded. They would have difficulty producing educational materials and may be open to the co-development of materials that could be provided to their members and the public.

By educating people on the use and sharing of genomic data, a more structured and productive national conversation might be had around subjects such as consent.
Opportunity: Working with consumer groups, co-develop resources providing consumer-friendly explanations of common terms and types of data used in genomics. Such content could be published on the InGeNA website and made available to individual groups for publication or distribution.

Besides providing educational materials to support consumer groups, it was important to ensure that policymakers had the background with which to develop informed public policy.

Opportunity: Working with clinical and research groups, industry can contribute to ensuring policymakers have the information they require to communicate the need, value and issues associated with genomics in policy setting and public communications.

Catalogue of endorsed standards

Standards underpin system interoperability and facilitate data sharing. There are opportunities for the genomics industry to endorse a suite of mature standards to support systems interoperability and procurement decisions by sector participants.

This is similar to the process undertaken by the Office of the National Coordinator (ONC) for Health Information Technology (HealthIT) in the USA. The Interoperability Standards Advisory (ISA) is the process by which they publish endorsed standards (and those under consideration) [126].

Opportunity: Having endorsed standards, it would be useful to publish a catalogue of these standards to maximise visibility and transparency. Such publication would also include the processes by which the endorsement took place.
The role of the genomics industry

The genomics industry can play a significant role in delivering on the promise of genomic medicine, its integration into health care and realising the potential of genomics to personalise health care. Industry participants bring a skills and experience across a range of organisations, including pharmaceuticals, biotechnologies, diagnostics, informatics, data and technology [127].

Through InGeNA, industry works alongside state and federal governments, consumer groups, academia and professional bodies to enable Australia to be a world leader in the adoption of genomics in health care and accelerate the potential benefits of genomics including improving health outcomes, contributing to the future affordability of health care and strengthening the economy and creating jobs.

InGeNA’s 2021 projects

To address the challenge of how industry can support delivering the promise of genomic medicine, InGeNA commissioned projects across all four strategic focus areas:

- **Quantifying the benefits of genomics**: This project undertook a literature review quantifying the benefits of genomics. Supported by case study analysis, it delivered evidence of the broad economic benefits of a thriving genomics industry, the value associated with prevention and early diagnosis of disease using genomic techniques, and the benefits that can be derived from effective treatments and the avoidance of harm from ineffective treatments [128].

- **Access and equity**: After undertaking a literature search, this project worked through solutions to addressing the challenges faced by genomics in addressing Health Technology Assessments (HTAs) in Australia. An Australian genomic test value framework is the key deliverable [129].

- **Workforce**: Development of a validated competency and capability criteria and framework for a genomic workforce is the key deliverable of this project. The framework will cover non-traditional role types including pharmaceuticals, advanced therapies, digital health, diagnostics and medical devices [130].

- **Data and technology innovation**: The final project addresses how industry can support data sharing and the challenges faced in governance of genomic data. This paper is the deliver for this project [131].

Working with the Australian Government

While the Australian Government shares responsibility with state, territory, and local governments for operating the health system, it is the majority funder and provides significant leadership and coordination in terms of policy and legislation [132].

Building a cooperative relationship with parts of the Australian Government will allow InGeNA to contribute valuable knowledge and experience to inform government planning and present a representative view of the genomics industry in such engagements. In discussions with representatives of the Australian Government Department of Health, it was acknowledged that active engagement with the Department would support their work by providing pragmatic industry insights during the development of policy concerning genomics.

Opportunities that exist for engagements include:

- responding to requests for information about genomic data and engagement in discussions with:
  - the Office of the National Data Commissioner [133] on subjects related to genomic data sharing and management [134]-[136]
• the Office of the Australian Information Commissioner [137] on subjects related to genomic data sharing and management as they are affected by the Australian Privacy Principles [138]
• the Digital Health Branch developing the Commonwealth Digital Health Blueprint for the Australian Government Department of Health [7]
• those branches of the Australian Government Department of Health responsible for genomics to provide knowledge and experience to inform government planning around evolving policy positions such as HTAs and the Policy Framework or Implementation Plan [1]
• the Australian Digital Health Agency in the refresh of Australia’s National Digital Health Strategy due in 2022 [8]

• staying aware of and responding to requests for information about genomic data and engagement in discussions regarding:
  • the proposed Data Availability and Transparency Bill [93], [134]
  • projects such as the Health Information Gateway and the modernisation of the national digital infrastructure by the Australian Digital Health Agency [139]
• engaging with other national research and infrastructure programs such as the National Collaborative Research Infrastructure Strategy (NCRIS) [140], the National Computational Infrastructure (NCI) [141], Bioplatforms Australia [142] (with whom InGeNA already has an MOU), and Phenomics Australia [143] - these programs already operate in the genomics and bioinformatics space and are likely to provide opportunities for knowledge exchange and mutual support
• engaging with national funding bodies such as the NHMRC, MRFF and ARC to encourage funding allocations to include requirements for genomic data generated as part of publicly funding research to be made available in standardised formats for including in any national genomic repository for further research, including consideration of the necessary consent required to support this approach.

Supporting the states and territories
There is activity in many states and territories regarding clinical genomics, and generally a strong desire to support data sharing for research.

In collaboration with the Australian Government and its departments and agencies, there are opportunities for InGeNA to support a national approach to sharing genomic data for both clinical purposes and research. These include contributions to areas such as:

• standardisation of exchange formats to reduce confusion and increase interoperability
• supporting pathology professionals and researchers in developing more consistent methods of reporting on findings to increase comprehension of non-genomics clinicians
• agreeing minimum standards for procurement of systems and services to ensure data interoperability is enabled.

Where jurisdictional genomics strategies have been published (or about to be published), there are frequently opportunities for InGeNA to engage with the relevant groups to collaborate. Many jurisdictions are uplifting capabilities and have identified a need to engage with industry. InGeNA has a unique role in that engagement.

Engaging professional bodies
While a large body of knowledge exists within the genomics industry and associated professional bodies, proliferation and understanding of genomics faces challenges for wider adoption in mainstream clinical domains.

Genomic data in Australia - An industry perspective on clarity, certainty and standardisation
While the responsibility for professional education falls to the relevant colleges and professional associations, these groups may have limited capabilities to maintain information and/or the ability to tailor it to their specific cohorts.

There exist opportunities to share the industry’s knowledge and increase broader clinical understanding of genomics through:

- in collaboration with professional bodies, developing an agreed body of knowledge including terms and definitions accessible to the broader clinical community that can be shared and localised by specific groups to meet the needs of their constituents while remaining consistent across all groups
- working with specific professional colleges, developing or enhancing guidelines and advice for clinical specialties that reflects contemporary practice.

By jointly developing this body of knowledge and specifically guiding clinical specialties through their respective colleges and professional associations, the ability for clinicians to clearly communicate with each other on topics related to genomics may be increased. By providing baseline content, InGeNA can provide a valuable service to such groups, reducing the need to “reinvent the wheel” with the associated risk of doing so inconsistently.

Guidelines might include:

- the range of genomic tests available across the sector, nationally and internationally, and the type of data that can result
- guidance about the appropriateness of specific tests
- generic and specialty specific terms to be included in literature, especially regarding the generation and interpretation of genomic data
- information targeted for consumer consumption.

Bodies that InGeNA might partner with include:

- Royal College of Pathologists of Australasia (RCPA)
- Public Pathology Australia
- Australian Pathology
- Human Genetics Society of Australasia (HGSA)
- Royal Australian College of General Practice (RACGP)
- Australian College of Rural and Remote Medicine (ACRRM)
- Australian Medical Association (AMA)
- Clinical Oncology Society of Australia (COSA)
- Cardiac Society of Australia and New Zealand (CSANZ).

Working with consumer advocacy and support groups

InGeNA already has a relationship with several consumer advocacy and support groups. These groups are typically constrained in funding and operated largely by volunteers with a personal interest in their area (for example, rare diseases). Like the professional bodies discussed above, these groups may not have the capacity or capability to invest in information for their constituents. Some of the materials developed to support professional groups may be of value to consumer groups. And if consumer-facing material is co-designed with these consumer groups, it could be made available to professional bodies with the knowledge that consumer shave been involved in its development.

Working with research alliances and groups
On page 15 of this document, there are several genomic alliances in Australia addressing the needs of both clinical genomics and research.

Most relevant of these is Australian Genomics. They are undertaking projects to further implement the NAGIM Blueprint, as well as a range of other projects that involve genomic data. In response they have developed a broad program of projects for the period 2021 to 2023 [40], which covers a range of data related activities including:

- continued development of national approaches to clinical genomic consent
- establishing a multidisciplinary collaborative data group
- progressing the NAGIM Blueprint implementation and developing further recommendations
- supporting the uptake of research capabilities and optimised national standards
- standardising and converging approaches to genomics information management
- progressing the optimal collection, standardisation and sharing of data
- formulating recommendations for data aggregation and quality control
- collaborating on the development and sharing of open-sourced genomic tools
- developing a sustainability model for ongoing data / system management
- incentivising and supporting sharing of research data
- promoting governance policies and procedures supporting secondary research data use
- implementing data access control systems to streamline data sharing
- reviewing national clinical phenotype data collection
- progressing standardised, detailed clinical data collection
- identifying limitations and potential solutions for clinical data sharing
- evaluating Electronic Request Forms for national activity and approaches
- convening Policy Network Meetings to progress genomic discussion and implementation

As InGeNA and Australian Genomics have an MOU to support cooperation, collaboration with Australian Genomics in these projects will be beneficial to InGeNA’s objectives.

Other genomic alliances such as Melbourne Genomics and the newly formed SA Genomics Health Alliance may be interested in engaging with InGeNA and establishing an MOU arrangement with them may be advantageous to represent industry perspectives with these groups.

Finally, there are a range of translation research groups in Australia with programs targeting genomics. Like the genomics alliances, they are actively engaged in genomic research, often in collaboration with the public health sector. Establishment of MOU arrangements to provide industry perspectives on their genomic data research may lend value to both the institutions and InGeNA. Groups that may be considered include:

- Advanced Health Research and Translation Centres (AHRTC) such as Brisbane Diamantina Health Partners (BDHP), Health Translation SA (HTSA) and Sydney Health Partners
- private research institutions such as QIMR Berghofer Medical Research Institute and the Harry Perkins Institute of Medical Research.
Services to the industry

Providing benefits back to the genomics industry itself is also of significant value. Through engagement with respondents, these opportunities have been identified:

- The many regulatory, compliance and other external factors that impact the genomics industry is non-trivial. Examples of external factors include government committees, assessments, programs, parliamentary bills, announcements, funding, public consultations that do or may affect genomic data.

  Specific examples include the *Data Availability and Transparency Bill* [93], the ACCC’s Consumer Data Right project [94], the Australian Data Strategy and the Digital Atlas of Australia announced as part of the *Digital Economy Strategy 2030* [7], the Australian Privacy Principles [138], and the *Australia’s Artificial Intelligence Ethics Framework* [77]. Other government groups that may provide valuable information for members would include the Pharmaceutical Benefits Advisory Committee (PBAC), the Medical Services Advisory Committee (MSAC) and the Health Technology Assessment (HTA) group within the Australian Government Department of Health.

  When asked whether they saw value in InGeNA providing an aggregated information stream gathering information from these various sources, survey respondents uniformly agreed this would be valuable for industry participants. While a considerable body of work that would require updating to maintain currency, the costs could be offset by providing it as a subscription service.

- Respondents also frequently identified the lack of an agreed data governance framework for genomic data as an obstacle that increased risk and reduced certainty. When asked regarding the value of a National Genomics Data Governance Framework that addressed areas such as ethics, privacy, and consent and was aligned with existing NHMRC guides for research related aspects, respondents were also overwhelmingly positive. It is noted this could leverage the work commenced in the *NAGIM Blueprint*, including the principles outlined in that document.
Case studies

Examples of the positive use of genomics data and data sharing at a person or population level can provide useful to understand different aspects of genomic data implementation. The examples selected each demonstrate an issue that is important to the genomics industry and InGeNA in particular.

Case study 1: Innovation & Genomic Profiling in Australia by OMICO

Founded in 2019, Omico is an Australian non-profit, incorporated national cancer network, funded by government, industry and the community, Omico’s aims create a sustainable ecosystem enabling research-led cancer care by providing precision oncology based on genomic medicine. Omico has built a nationwide network of research and treatment centres that facilitate, support and promote clinical trials in genomic cancer medicine [144], [145].

To address the key barriers to the implementation of precision medicine, Omico has:

- created a system for organisations to engage nationally in genomic cancer medicine in a uniform manner across tertiary referral hospitals/centres
- progressively scaled activities to test fundamental concepts such as national engagement of academic centres/research for clinical trials
- delivered a demonstrable ROI for government through commercial engagement.

By screening up to 80,000 Australian cancer patients, Omico aims to enrol an estimated 12,000 of those patients in clinical trials to access novel therapies over the next five years. Eventually, the objective is to establish universal genomic profiling of oncology patients in Australia using a public-private consortium consisting of government, biotechnology and pharmaceutical companies, contract research organisations, investment organisations and data and analytics providers.

As a national network of cancer research and treatment centres, Omico can deliver genomic cancer medicine clinical trials. Omico’s programs seek to understand the genetic causes of cancer, and develop strategies to detect cancers at an early, curable stage for those at increased cancer risk. Omico is currently operating three programs [144]:

- Molecular Screening & Therapeutics (MoST) study: Using genomic technology to characterise molecular changes in a patient’s cancer that may help to identify a targeted therapy.
- Genetic Cancer Risk in the Young (RisC) study: Studying the genetic variants contributing to inherited cancer.
- Surveillance study in Multi-Organ Cancer prone syndromes (SMOC+): Investigating and evaluating the surveillance practices used for people at high risk of multi-organ cancer.

Omico acts as a single point of entry for industry groups seeking participants in clinical trials of new or repurposed therapies, providing access to appropriate patients of biomarker directed studies. InGeNA has a MOU arrangement with Omico and several InGeNA members are industry partners with Omico in their own right.

Omico is establishing a National Genomic Medicine Platform [145] that is hoped to result in improved health benefits for patients, enhancement of the quality and capabilities of the healthcare sector, increased economic activity and the creation of skilled jobs. While targeted at oncology genomics, such a platform would be consistent with the roadmap outlined in the NAGIM Blueprint and Australian Genomics is one of Omico’s partners.

The recent announcement by global clinical research organisation George Clinical of its collaboration with Omico to harness the National Genomic Medicine Platform [146] demonstrates the value placed by leading Australian and international clinical research groups in this platform, and the importance of collaboration between government, research and industry groups.
Case study 2: Genomics as a population health response to the COVID-19 pandemic

Australia, like countries worldwide, has been impacted by the COVID-19 pandemic caused by the SARS-CoV-2 virus. However, a surprising by-product of the pandemic has been increased public visibility of genomic sequencing, with daily media reports describing how public health officials use the technique to trace sources of infection [147].

Underpinning Australia’s response to the pandemic has been a national repository of genomic sequence data for the SARS-CoV-2 virus. The system, called AusTrakka, was developed by the Communicable Diseases Genomics Network (CDGN). AusTrakka allows health authorities around Australia and New Zealand to share, store, analyse and view aggregated COVID-19 genomic data in a single secure, and private repository. Fundamental to the system is a data sharing framework [148].

This framework allows authorised public health laboratories to upload genomic sequences to AusTrakka. A phylogenetic tree is constructed from the data each night that shows how specific sequences are related, support public health officials to trace the outbreak [147].

Specific data privacy rules are embodied in the framework, allowing public health laboratories to access their own information and aggregated data from other jurisdictions. The framework also outlines the governance model for the repository and provides procedures for research data requests within Australia and internationally [148], [149].

While the AusTrakka system was developed by public health bodies rather than industry participants, it demonstrates several factors that are important when InGeNA is engaging with government groups or consumers in general. Using a single system that transcends traditional state-based restrictions on data sharing has been fundamental to the pandemic response in Australia. It has also lifted the public awareness of genomics as a disease surveillance technique and reflected positively on the science [147]. It also demonstrates that data sharing arrangements can be achieved if an appropriate value proposition is identified.

Case study 3: Clinical genomics and data sharing in Victoria

When Melbourne Genomics was established in 2016, one of the key objectives of the program was to establish a common system for clinical genomics across several public hospitals in Melbourne. Because of the independent nature of the hospital system in Victoria, the solution required the ability to keep data separate for each hospital within a common cloud storage and analysis platform [44], [150].

The system, called GenoVic, now supports genomic testing at five organisations:

- Victorian Clinical Genetics Services
- The Royal Melbourne Hospital
- Monash Health
- Australian Genome Research Facility
- The Alfred Hospital

Melbourne Genomics developed the platform using Amazon Web Services (AWS) secure cloud services hosted within Australia [62], and partnered with Agilent, the commercial developers of curation application Alissa Interpret [151], [152], to deliver a standards-based orchestration service using FHIR APIs to manage genomic and clinical data,

While the initial focus has been on delivery of clinical testing, the program has also integrated key applications to support better clinical outcomes. Real-time sharing of detailed scientific evidence about clinically interpreted
variants is available using Shariant, a service delivered by Australian Genomics [153]. A knowledgebase of genes related to human disorders developed by Genomics England called PanelApp has been integrated and allows users to synchronise gene lists using a custom portal [154].

Melbourne Genomics have developed and deployed data access and sharing policies, procedures and agreements to support their research programs and are applying these learnings to clinical data sharing efforts [45].

This project demonstrated that a combination of commercial applications and a standards-based approach to solutions could deliver a functional clinical genomics platform. Further, due to the selection of commonly available standards, the platform can work with other commercial products that support these standards. However, without establishing data sharing agreements and underlying data governance, the technical solution may not have been as successful. Commercial partnering and common data governance arrangements are critical for InGeNA and industry participants.
Summary of opportunities and activities

This report is intended primarily as an input to planning for the InGeNA membership. However, it should also serve as a useful guide to the value industry can bring to public health, research and the broader sector. What follows is a summary of the opportunities, suggested activities and the remaining challenges for these various groups to consider.

Identified opportunities

This report has outlined a series of opportunities for InGeNA and its members to consider.

1. **Standardising phenotypes** – To increase the value of clinical data for genomics, improvements are necessary in the way clinical systems capture and code phenotype data. Australian Genomics have several projects looking at standardisation of phenotypes in the Australian healthcare setting. InGeNA members may be able to support aspects of these projects.

2. **Genomic metadata** – A standards-based approach to metadata capture may reduce the burden of making clinical data ready for research and data discoverability. The Australian Government is keen to leverage investments in research and clinical settings and can provide leadership in supporting cross-jurisdictional standardisation efforts. Australian Genomics is planning projects to promote policies and procedures that support secondary use of data. InGeNA can work with both groups to support a national approach to genomic metadata.

3. **Enabling federation** – A standards-based approach to clinical and genomic data capture may reduce the burden of providing research data for federated use. Implementation of aspects of the NAGIM Blueprint by Australian Genomics including pilot data implementations may offer InGeNA the opportunity to support these pilots to gain understanding of the standardisation process.

4. **Analytics and compute-to-data** – Exploring the further use of federated analytics and compute-to-data capabilities may allow the industry to inform the approaches needed to support national genomics research while maintaining privacy. Working with industry organisations that currently use capabilities in this area may inform further investigations.

5. **NAGIM Implementation** – As Australian Genomics has carriage of the work to look at implementations of the NAGIM Blueprint, InGeNA should actively engage with those projects to ensure that the requirements and skills of the industry are applied to implementation planning and activities.
6. **Start-up innovation** – The establishment of a national capability that included clinical and genomic data provides opportunities for innovation. While this can be leveraged by large industry participants, it also represents an opportunity for InGeNA to encourage start-up participation through mentoring or other incentives to make innovative use of such a data capability. One example might be an incentive program or a national challenge to target a specific issue. Partnering with one or more universities or groups like the Digital Health CRC may be beneficial in this regard.

7. **Guidance on AI/ML** – As part of any implementation of the NAGIM Blueprint by Australian Genomics, consideration of the use of this data to support AI and ML technologies in line with Australia’s Artificial Intelligence Ethics Framework [77]. InGeNA role should include providing Australian Genomics with advice from industry about requirements for AI and ML in this area.

8. **Benefits Reporting** – InGeNA’s Data and Innovation Working Group should liaise with other working groups and Australian Genomics to establish a process for leveraging the national capability to deliver evidence to government that demonstrate the economic benefit to the health sector to support improved sustainability.

9. **Future data use** – InGeNA is uniquely positioned to engage in the debate about the commercial use of genomic data. A position paper that describes the nature of use and safeguards may held address some of the concerns likely to be raised by the public and media. This would be specifically relevant to those organisations with an interest in clinical trials and research.

10. **Federated query** – Adopting a standardised approach to structuring and operating federated query systems in some circumstances may allow participating data sources to preserve the privacy of their data while supporting the research outcomes possible from the broader data environment. There may be opportunities for organisations with existing technologies and approaches to engage with (or join) InGeNA to support such investigations.

11. **Legal and regulatory** – While each organisation must ultimately make their own assessments on legal compliance, a contemporary review of the Australian legal and regulatory environment by InGeNA would assist InGeNA members and other organisations operating with genomic data. Such a review would also inform any InGeNA position regarding introduction of or changes to data privacy regulation in Australia.

12. **Consent management** – Australian Genomics have planned continued work on consent and their dynamic consent platform (CTRL). Engagement with Australian Genomics in this work would allow InGeNA to bring national and international experience to the table, and support moves to a national approach to managing consent across public, private and research genomic data.

13. **Consumer consent** – There is a strong appetite for further consultation and consensus development with health consumer groups on consent around genomics data. Such work would inform approaches that build fine-grained real-time consent to let patient cohorts calibrate their data sharing preferences more actively. As Australian Genomics have a project to continue development of consent models, the opportunity exists for InGeNA to be engaged in this work.

14. **Blockchain** – An InGeNA sponsored review of blockchain technologies related to genomic data sharing may help inform public and industry debate around the application of such technologies to increase transparency and security of genomic data considering issues associated with blockchain.

15. **Implications for insurance** – Supporting an ongoing discussion on the implications of private insurance use of genomics may reduce concerns about the long-term impact of involvement in genomic research. Continued engagement by InGeNA with the Australian Government in this regard is advisable.

16. **Data management code** – An InGeNA sponsored development of a voluntary DTC ‘code of conduct’ would allow these services to indicate their data management practices of consumer data against a set of guiding principles. An information campaign about the code would better inform customers of DTC products in how their data will be used. A voluntary code may be a precursor to a more formal accreditation regime if appropriate.
17. **Data governance patterns** – While each organisation must ultimately make their own assessments on compliance and data governance, a national framework that outlined guidance across a suite of implementation patterns would encourage more consistent and robust data governance. As Australian Genomics is currently looking at some areas of this, continued engagement with them would be advisable.

18. **Data security** – A lightweight suite of tools to self-assess data security, coupled with some pragmatic guidance to support improvements could improve the understanding and use of appropriate cybersecurity for genomics data. This is an area in which the Australian Digital Health Agency (ADHA) has some historic experience and InGeNA could partner with ADHA to support the development of such tools.

19. **Data sharing framework** – Co-development with Australian Genomics of a national framework for genomic data sharing will support transparent conversations about the subject to better inform policy and regulation development. This would ensure that an industry lens on data sharing is included in work planned by Australian Genomics.

20. **IP considerations** – InGeNA could sponsor or develop a position paper on the limits of intellectual property rights over genomic data may be beneficial to conversations around consent and consumer engagement. This would need to address the issue of IP associated with samples processed internationally and the associated contractual arrangements required. Engagement with the pathology sector including the Royal College of Pathologists of Australasia would be advantageous.

21. **Data storage** – Monitoring the evolution of both genomic and storage technologies will be required to ensure the balance between them supports the sustainable application of genomics. Working with the InGeNA working group that is quantifying the benefits of genomics would inform both work programs. Engagement with leading cloud vendors with genomics platforms would ensure currency of this information.

22. **Data structures** – The endorsement by InGeNA of key standards around data structures and taxonomies may improve interoperability and provide greater certainty for the genomics sector. Development of a list published on the InGeNA website would increase visibility and provide a common position. This would also provide a platform for further engagement with the Standards community and the Australian Government.

23. **Standards endorsement** – Workshop attendees suggested that rather that developing standards, there was an opportunity for InGeNA to clearly articulate which standards were being collectively endorsed. Such endorsement could be placed on the InGeNA website. Working with the Commonwealth in this regard may also be beneficial. See previous opportunity point as it regards interoperability.

24. **Synoptic reporting** – InGeNA can engage with leading pathology groups, professional bodies such as the Royal College of Pathologists of Australasia, and the Standards community to support the development of more consistent synoptic reporting capabilities for genomic data to increase interoperability and improve communication and discoverability of results.

25. **Planning collaboration** – InGeNA should establish a policy around collaborative efforts to ensure that investments made by InGeNA can be assessed for the value and costs associated with them and plans for further adoption considered as part of the collaboration decision.

26. **Data diversity** – Continued support for initiatives that improve the diversity of genomic data will support reduced efforts to curate and interpret genomic variants, especially when algorithm processes such as machine learning and artificial intelligence are used. InGeNA might consider approach the NCIG to discuss options for supporting their work in diversity.

27. **Data classification** – Refinement of the genomic data classification framework can support more informed data governance conversations by clearly identifying the type of genomic data under discussion. InGeNA can engage with Australian Genomics on their projects to trial aspects of the NAGIM Blueprint.

28. **Genomics glossary** – Working with consumer groups, co-develop resources providing consumer-friendly explanations of common terms and types of data used in genomics. Such content could be published on the InGeNA website and made available to individual groups for publication or distribution.
29. **Information sharing** – Working with clinical and research groups, industry can contribute to ensuring policymakers have the information they require to communicate the need, value and issues associated with genomics in policy setting and public communications.

30. **Standards catalogue** – Having endorsed standards, it would be useful to publish a catalogue of these standards to maximise visibility and transparency. Such publication would also include the processes by which the endorsement took place.

**Recommended activities**

In addition, the following recommended activities have been identified.

![Activities](image)

1. **Responding to information requests** – responding to requests for information about genomic data and engagement in discussions with:
   a. the Office of the National Data Commissioner [133] on subjects related to genomic data sharing and management [134]–[136]
   b. the Office of the Australian Information Commissioner [137] on subjects related to genomic data sharing and management as they are affected by the *Australian Privacy Principles* [138]
   c. the Digital Health Branch developing the *Commonwealth Digital Health Blueprint* for the Australian Government Department of Health [7]
   d. those branches of the Australian Government Department of Health responsible for genomics to provide knowledge and experience to inform government planning around evolving policy positions such as HTAs and the *Policy Framework or Implementation Plan* [1]
   e. the Australian Digital Health Agency in the refresh of *Australia’s National Digital Health Strategy* due in 2022 [8]
   f. the proposed *Data Availability and Transparency Bill* [93], [134]
   g. projects such as the Health Information Gateway and the modernisation of the national digital infrastructure by the Australian Digital Health Agency [139]

2. **Research programme engagement** – engaging with other national research and infrastructure programs such as the National Collaborative Research Infrastructure Strategy (NCRIS) [140], the National Computational Infrastructure (NCI) [141], Bioplatforms Australia [142] (with whom InGeNA already has an MOU), and Phenomics Australia [143] - these programs already operate in the genomics and bioinformatics space and are likely to provide opportunities for knowledge exchange and mutual support.

3. **Funding design engagement** – engaging with national funding bodies such as the NHMRC, MRFF and ARC to encourage funding allocations to include requirements for genomic data generated as part of publicly funding research to be made available in standardised formats for including in any national genomic repository for further research, including consideration of the necessary consent required to support this approach.
4. **National approach** – support a national approach to sharing genomic data for both clinical purposes and research in areas such as:
   a. standardisation of exchange formats to reduce confusion and increase interoperability
   b. supporting pathology professionals and researchers in developing more consistent methods of reporting on findings to increase comprehension of non-genomics clinicians
   c. agreeing minimum standards for procurement of systems and services to ensure data interoperability is enabled

5. **Jurisdictional capacity** – where jurisdictional genomics strategies have been published, support jurisdictional capability uplift through engagement activities to identify areas of support that can be addressed

6. **Body of knowledge** – in collaboration with professional bodies, develop an agreed body of knowledge including terms and definitions accessible to the broader clinical community that can be shared and localised by specific groups to meet the needs of their constituents while remaining consistent across all groups

7. **Contemporary guidelines** – work with specific professional colleges, developing or enhancing guidelines and advice for clinical specialties that reflects contemporary practice

8. **Consumer content** – leverage work with professional groups to provide content that may be of value to consumer advocacy groups

9. **Industry content** – engage with the various genomic alliances, most notably Australian Genomics, to support existing programs of work by contributing an industry perspective on planned activities

10. **Information stream** – investigate the development of an aggregated information stream gathering information from various public sources for use by industry participants

11. **Data governance** – support development of a National Genomics Data Governance Framework that addressed areas such as ethics, privacy, and consent and was aligned with existing NHMRC guides for research related aspects.

**Remaining challenges**

The opportunities and activities outlined in the last two sections provide a roadmap for future work that will allow the genomics industry to play an active role in supporting the delivery of genomics to the healthcare and research sectors and realising the benefits for all.

Although there is significant potential to deliver benefits from genomics through these activities, significant challenges do remain. While the activities outlined above will help to address these challenges, they are unlikely to be resolved through the efforts of any one group or industry. These challenges include:

- There is a need for Australia to adopt a national approach to managing genomic data to benefit both clinical genomics and genomics research. While these two groups overlap in requirements, workforce and aspirations, they are not identical. Finding a balance that meets the needs for all will require coordination across Commonwealth and state and territory governments, research funders and institutions, commercial partners and private healthcare organisations. There is also a need to recognise that genomics has worldwide application and Australia cannot act independently without jeopardising data use and sharing that can support better clinical outcomes for people who suffer from rare diseases that can be identified only through international collaboration.

- The identification, development and adoption of data standards has been a challenge for the broader digital health sector. A lack of adoption of agreed standards by the software and healthcare sectors has limited interoperability and data use and sharing. Genomics is still “new” enough to avoid some of the data standards...
pitfalls that digital health more generally has encountered but resolving this challenge will require the involvement of clinicians, researchers, governments and the software industry.

- Finally, an informed, national conversation around consent, privacy and data use for public health, research and commercial activities needs to happen to provide certainty for those involved and to establish trust on the part of the Australian public. Such conversations are challenging to arrange, cannot be rushed, and can be easily derailed. Examples in recent times of this challenge include agreements on the secondary uses of the data in the My Health Record and the change from opt-in to opt-out arrangements for the My Health Record system. Such a conversation will need to involve the public, governments, clinicians and researchers, and the commercial sector.

While these challenges are significant, they are achievable if all parties commit to open, collaborative engagement to benefit consumers. This report and the opportunities and the activities it presents are a first step in rising to meet these challenges.
Appendix A: Overview of methodology

This project undertook a two-stage engagement process to gather information from both InGeNA members and those in the broader genomics sector.

Range of stakeholders
Stakeholders were grouped into four broad categories:

- members of InGeNA (Group A, 17 stakeholders)
- stakeholders representing state, territory, or Australian Government organisation, including pathology groups (Group B, 5 stakeholders)
- members of the genomics industry which were not InGeNA members, including MOU holders, clinical research organisations and service providers (Group C, 13 stakeholders)
- professional bodies or colleges and consumer representatives (Group D, 13 stakeholders)

Stakeholder survey
After the engagement list was agreed, stakeholders were sent a survey instrument to gather information about their role in genomics and how the industry could support this work.

Response rates
The following response rates were achieved:

- Group A: 13 respondents (76%)
- Group B: 2 respondents (40%)
- Group C: 4 respondents (31%)
- Group D: 5 respondents (38%)

Survey questions
A range of questions, cleared by InGeNA were included in the survey, as follows:

A little bit about you and your organisation
- In which of the following categories would your organisation identify?

Barriers and roadblocks
- What do you see as barriers or roadblocks to genomic data being shared and the integration of genomics into mainstream health care?
- What steps is your organisation taking to address these barriers?
- What steps could other industry organisations take to address these barriers?

Role of the genomics industry
- What role(s) do you think the genomics industry can play in increasing value and/or benefits through genomic information management?
- Are there any other roles you think the genomics industry can play in increasing value and/or benefits through genomic information management?

The genomics value chain
• Think of the organisations that provide your organisation with genomic/clinical data (i.e., the ones that you rely on to provide information you need to do your role). How would you describe these organisations?
• Still thinking of the organisations that provide your organisation with genomic/clinical data, are there any improvements you can think of regarding the way you obtain this information from them?
• Now think about organisations to which you provide genomic/clinical information. How would you describe these organisations?
• Still thinking about organisations to which you provide genomic/clinical information, are there any improvements you can think of regarding the way you provide this information to them?
• Thinking about the way data moves along the whole genomics value chain (from start to finish), how would you describe the processes involved?
• Still thinking about the way data moves along the whole genomics value chain, are there any parts of it you think could be improved?

Adding value for long-term health prevention and treatment
• Describe the ways your organisation could improve long-term health prevention and treatment options for patients by contributing to genomic information management.
• Describe the ways other organisations in the genomics industry could improve long-term health prevention and treatment options for patients by contributing to genomic information management.

Consumers and genomics
• How does your organisation support consumers to access and/or understand the genomic data about themselves?
• Does your organisation receive requests from consumers for access to or understanding of genomic data?
• Describe the nature of such consumer requests?
• Describe any additional activities that your organisation could do to support consumers?
• Describe any additional activities that the genomics industry as a whole could do to support consumers?

Data ownership, custodianship and access
• Does your organisation take genomic and related data from external sources, and create new/enhanced information from it?
• If so, do you retain any intellectual property rights in this new data that would limit access to it by others?
• If so, how do you address requests for access to this data by either the consumer or researchers?

Use of genomic data in research
• Does your organisation provide access to genomic data for research purposes?
• How does your organisation provide access to genomic data for research purposes?
• Does your organisation use genomic data for research purposes?
• How does your organisation obtain genomic data for research purposes?
• Does your organisation support genomic research through provision of technology or data?
• Describe the way your organisation does this?
• Describe any challenges or opportunities associated with your support for the research community.
• Describe any additional activities that your organisation could do to support genomic research?
• Describe any additional activities that the genomics industry as a whole could do to support genomic research?

Clinical outcomes
• Describe what genomic data should support clinical diagnosis or treatment.
• Describe any challenges or opportunities that clinicians experience when making clinical decisions using clinical data.

Regulation and compliance
• Which organisations/groups do you believe should be involved in the regulation and/or compliance associated with clinical data?
• Describe any changes to regulation/compliance that you think would help the genomics industry manage genomic data.

Genomic data holdings
• Does your organisation hold genomic data?
• More specifically, what types of data are you holding?
• How is your genomic data stored?
• What regulatory or standards are you complying with for the genomics data you are holding?
• How do address the privacy concerns of this genomic data?
• How do you protect the deidentified genomic data from re-identification?
• What requirements do you have for the retention of genomic data?
• Do you hold genomic data for periods longer than the minimum requirements?
• In broad terms, what is the size of your organisation's genomic data holdings?
• In broad terms, what does it cost to store this data per year?
• Who bears the costs of holding genomic data in your organisation?
• Who in your organisation is responsible for protecting genomic data?
• What cybersecurity practices and/or standards do you follow to protect genomic data?
• Please describe any insurance coverage related to genomic data that your organisation has in place in terms of value, scope and limitations.
• Can you describe your organisation's approach to dealing with the risk of ransomware attacks and the like?

Looking forward
• What are the top three things that you think that InGeNA should do with regard to genomic data?
• In the area of genomics, there are many regulatory, compliance and other external factors that impact organisations. What are your thoughts about the value of InGeNA providing an aggregated information stream gathering information from a number of sources (see below) in a subscription service?
• What do you think about the value of a National Genomics Data Control Framework which might include areas such as ethics, privacy and consent and is aligned with NHMRC guides for the research components?
• Do you have any further contributions with regard to data that should be considered by InGeNA when establishing an industry paper on data?

Survey analysis
The responses were analysed to identify consistent qualitative themes. These themes included:
• priority factors influencing genomic data use and sharing
• addressing the security and privacy concerns of genomic data storage and sharing
• contributions that industry partners can make to the advancement of genomic data use and sharing
• resources that would be valuable to organisations to influence their strategies, plans and implementations
the role of data standardisation

Further analysis was undertaken of those questions providing quantitative data.

**Interview workshops**

The survey instruments invited respondents to participate in group workshops. Two workshops were scheduled for each stakeholder group.

**Participation rates**

The following participations rates were achieved:

- Group A: 12 participants from 12 organisations
- Group B: 3 participants from 2 organisations
- Group C: 5 participants from 3 organisations
- Group D: 3 participants from 3 organisations

**Workshop approach**

Workshops were conducted as videoconferences with a prepared set of slides to prompt discussion based upon the six thematic areas identified in the survey stage. For the smaller groups, the slides were used as a guide for the facilitators but not displayed.
## Glossary

The following terms and acronyms appear in this document.

<table>
<thead>
<tr>
<th>Term/Acronyms</th>
<th>Description</th>
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<tbody>
<tr>
<td>ACCC</td>
<td>Australian Competition and Consumer Commission</td>
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<tr>
<td>ACRRM</td>
<td>Australian College of Rural and Remote Medicine</td>
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<tr>
<td>ADHA</td>
<td>Australian Digital Health Agency</td>
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<td>AHMAC</td>
<td>Australian Health Ministers' Advisory Council</td>
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<tr>
<td>APP</td>
<td>Australian Privacy Principles</td>
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<tr>
<td>AWS</td>
<td>Amazon Web Services</td>
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<tr>
<td>Bioinformatics</td>
<td>Bioinformatics is a subdiscipline of biology and computer science concerned with the acquisition, storage, analysis, and dissemination of biological data, most often DNA and amino acid sequences [155]</td>
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<tr>
<td>CDA</td>
<td>Clinical Data Architecture</td>
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<tr>
<td>CDGN</td>
<td>Communicable Diseases Genomics Network</td>
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<tr>
<td>CSIRO</td>
<td>Commonwealth Scientific and Industrial Research Organisation</td>
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<tr>
<td>DTC</td>
<td>Direct-to-Consumer</td>
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<tr>
<td>EMR</td>
<td>Electronic Medical Record</td>
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<tr>
<td>FHIR</td>
<td>Fast Healthcare Interoperability Resources</td>
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<td>GA4GH</td>
<td>Global Alliance for Genomics and Health</td>
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<tr>
<td>GDPR</td>
<td>General Data Protection Regulation</td>
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<tr>
<td>Genomics</td>
<td>Refer to both the study of single genes (genetics) and the study of an individual’s entire genetic makeup (genome) and how it interacts with environmental or non-genetic factors [1, p. ii]</td>
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<tr>
<td>GWAS</td>
<td>Genome-Wide Association Studies</td>
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<td>HGSA</td>
<td>Human Genetics Society of Australasia</td>
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<tr>
<td>HIPAA</td>
<td>Health Insurance Portability and Accountability Act</td>
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<td>HL7</td>
<td>Health Level Seven, Inc.</td>
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<td>InGeNA</td>
<td>Industry Genomics Network Alliance</td>
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<tr>
<td>ISA</td>
<td>Interoperability Standards Advisory</td>
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<tr>
<td>LIIMS</td>
<td>Laboratory Information Management System</td>
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<tr>
<td>LOINC</td>
<td>Logical Observation Identifiers Names and Codes</td>
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<tr>
<td>MOU</td>
<td>Memorandum of Understanding</td>
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<tr>
<td>Term/Acronym</td>
<td>Description</td>
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<tr>
<td>NAGIM</td>
<td>National Approach to Genomics Information Management</td>
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<td>NATA</td>
<td>National Association of Testing Authorities</td>
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<td>NCIG</td>
<td>National Centre for Indigenous Genomics</td>
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<tr>
<td>NHMRC</td>
<td>National Health and Medical Research Council</td>
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<td>NPAAC</td>
<td>National Pathology Accreditation Advisory Council</td>
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<tr>
<td>ODHSI</td>
<td>Observational Health Data Sciences and Informatics</td>
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<tr>
<td>ONC</td>
<td>Office of the National Coordinator</td>
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<tr>
<td>Phenotype</td>
<td>The term &quot;phenotype&quot; refers to the observable physical properties of an organism; these include the organism's appearance, development, and behaviour [156]</td>
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<tr>
<td>RCPA</td>
<td>Royal College of Pathologists of Australasia</td>
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<tr>
<td>Sequencing</td>
<td>Sequencing DNA means determining the order of the four chemical building blocks - called &quot;bases&quot; - that make up the DNA molecule [50]</td>
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<tr>
<td>SNOMED-CT</td>
<td>A systematically organised and computer processable collection of clinical terms providing codes, terms, synonyms and definitions used in clinical documentation and reporting</td>
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<tr>
<td>TGA</td>
<td>Therapeutic Goods Administration</td>
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<td>VCGS</td>
<td>Victorian Clinical Genomics Service</td>
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References


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