NATIONAL APPROACH TO GENOMIC INFORMATION MANAGEMENT (NAGIM)

IMPLEMENTATION RECOMMENDATIONS OVERVIEW

Australian Genomics

EXECUTIVE SUMMARY

To secure our data assets and make the most of Australia's investments – a national genomic data infrastructure is urgently required to better support healthcare and medical research.

Australia has embraced the potential of genomics, with significant investments in genomic research and healthcare systems. The datasets from these endeavors are already substantial, but siloed.

A National Approach to Genomic Information Management (NAGIM) will deliver an integrated **genomic data asset for Australia, using secure digital systems and nationally aligned frameworks.**

This will allow seamless access to data – with appropriate consent and regulatory controls, **to benefit healthcare, patients and medical research**.

A NAGIM for Australia requires a federated approach - to support autonomy of jurisdictions and organisations. This will involve adoption of interoperable systems, standardised approaches to data, cloud-based services and international best practice.

Australian Genomics has developed Recommendations for Implementing NAGIM, informed by architectural prototyping, international review and broad consultation.

Eight high-level recommendations are presented, with corresponding workstreams, priority areas for action, and an overall strategy for commencing NAGIM.

Effective involvement of genomics stakeholders will be critical to the success of the NAGIM strategy, including those of clinical services, Indigenous peoples, the community and industry.

The NAGIM Implementation report presents a vision, a means and a method to progress a national approach to genomic information management for Australia.

INTRODUCTION

The promise of genomic data.

This is a time of unprecedented momentum in global health genomics. There is growing demand for clinical genomic testing and accelerating investment in genomic research.

With the right infrastructure, governance and oversight there is an exciting opportunity for genomic data to be collectively and ethically used to:

✓ Achieve new diagnoses and better treatments;

✓ Improve the efficiency and impact of healthcare systems;

✓ Advance knowledge about health and disease; and

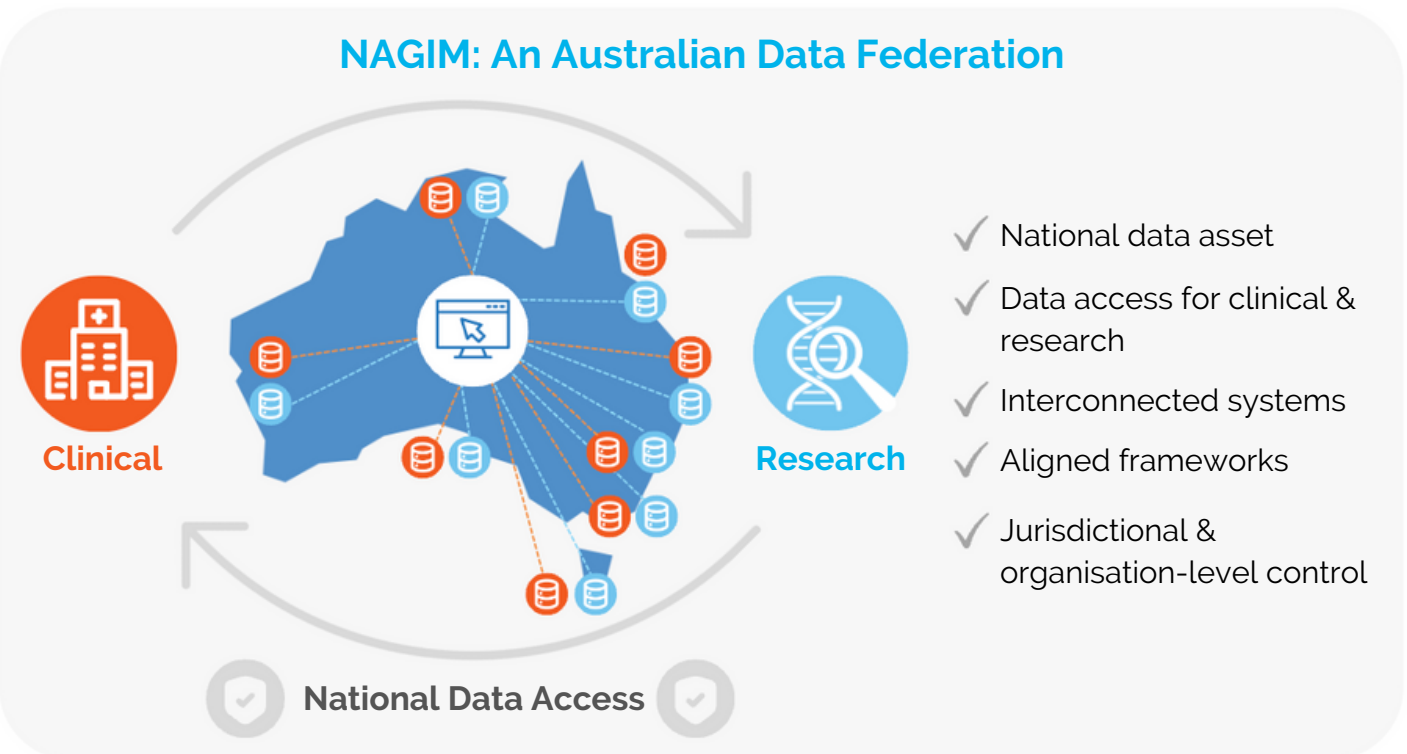✓ Progress equitable and sustainable health genomics for all Australians.

The problem with Australia's data infrastructure.

Despite our rapidly expanding genomic datasets, Australia lacks nationally coordinated governance and infrastructures. This has resulted in pervasive data silos across Australia. Much needed genomic and health data is often inaccessible for advancing healthcare and medical research.

Australia has fallen at least five years behind other genomic initiatives globally. Without critical national infrastructure, **we are now facing an urgent predicament in genomic information storage, analysis and sharing.**
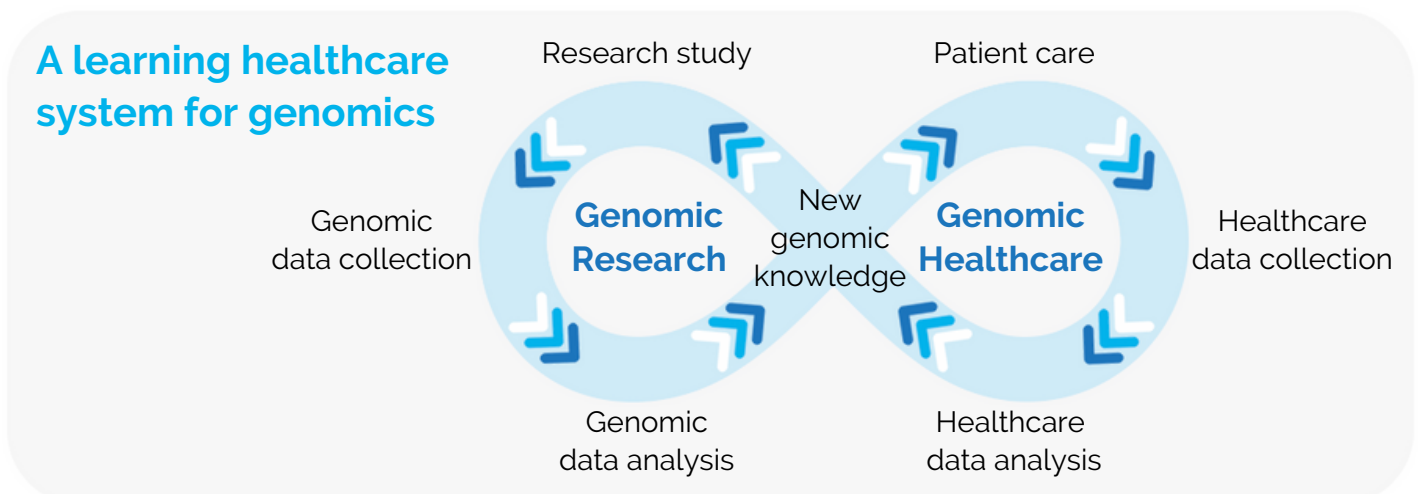
THE NAGIM VISION

A NAGIM for Australia represents **a data asset for the nation**, supported by secure digital genomics systems. This will provide seamless access to genomic and health data, using nationally aligned policies and regulatory frameworks.

NAGIM: An Australian Data Federation

Clinical

Research

✓ National data asset

✓ Data access for clinical & research

✓ Interconnected systems

✓ Aligned frameworks

✓ Jurisdictional & organisation-level control

National Data Access

An Australian NAGIM will ensure data from clinical genomic testing is available for future clinical care and broad medical research, with the appropriate consent and permissions. This will need deep consideration of public and patient perspectives, transparency and community engagement.

With this, a future NAGIM for Australia can ultimately support a learning healthcare system that benefits healthcare, research and patients.

A learning healthcare system for genomics

Research study

Patient care

Genomic data collection

Genomic Research

New genomic knowledge

Genomic Healthcare

Healthcare data collection

Genomic data analysis

Healthcare data analysis

THE NAGIM BLUEPRINT

The **Blueprint for a National Approach to Genomic Information Management** is a digital genomics blueprint that serves as a national framework for managing genomic data. It was developed in 2020, by Queensland Health for the Commonwealth Department of Health, and outlines connected systems and aligned processes, bound by sound data management practices and ethical, legal and social principles.

For a connected digital genomics ecosystem in Australia, the NAGIM Blueprint states that a **federated approach** or a **hybrid approach** (mix of federated, distributed and centralised) is most appropriate, based on the Australian clinical and research landscape.
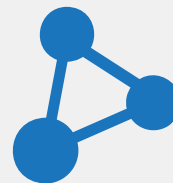
Data Infrastructure Types

Centralised genomic databases	Data commons	Distributed	Federated
Central platform providing information from datasets that have been combined	Separate datasets housed in a single, controlled access repository	Linkage of disparate datasets across different systems without shared components	Independently governed datasets on separate infrastructures aligned to common data elements and rules

However, a **flexible approach** to federating data in Australia will be required.

AN IMPLEMENTATION FRAMEWORK

A NAGIM Implementation Framework has been developed to progress the NAGIM vision. The framework depicts foundational elements to guide future implementations.

Vision

Australia benefits from a national genomic data asset that enriches research; informs clinical care; and improves health, ethically and equitably, for every Australian.

Architectural Enablers of an Australian NAGIM

- Federated frameworks
- Interoperability of systems
- Standards based approaches
- Cloud-based or hybrid solutions
- Alignment with international best practice

Critical Infrastructure Pillars

Interoperability	Scalability	Extensibility	Security	Sustainability	Usability

Fundamental Principles

- Trust, Transparency, Accountability
- Equity of Access
- Indigenous Data Sovereignty
- Alignment to Australian regulatory, legal and cultural context
- Best practice Data Governance and Ethical, Legal and Social considerations

Interoperability: able to interact with other systems to seamlessly exchange and make use of information
Scalability: able to handle growth and large-scale datasets
Extensibility: able to accommodate new additions and expand functionality
Usability: ease of use and ability to meet the needs of all end-users in clinical and research settings
Security: protection of data, infrastructure, and applications from harm or misuse
Sustainability: long term operational and financial maintenance of systems, resources and programs that ensures continuity.

NAGIM IMPLEMENTATION RECOMMENDATIONS

Recommendation 1: Governance, Coordination and Implementation of NAGIM

Appoint a coordinating body, governance structures and working groups to manage, monitor, evaluate and report across all parallel workstreams, clinical and research, to progress NAGIM.

Recommendation 2: Architectural Foundations for Integrated Research Infrastructure

Commence building architectural foundations, and core genomic data management infrastructure, for a federated NAGIM research ecosystem.

Recommendation 3: Clinical Information Systems, Priorities, and Intersections

Establish alignment and intersections of NAGIM research ecosystems with Australian jurisdictional clinical systems, and progress clinical priorities.

Recommendation 4: Data Governance and Ethical, Legal and Social Considerations

Identify and address data governance and legal requirements, and ethical and social considerations, associated with a federated NAGIM.

Recommendation 5: Key National Services for NAGIM

Establish the key national services required to operate a NAGIM in Australia, including services for national digital identities, security, infrastructure users and workforce.

Recommendation 6: Data Standards and Harmonisation

Achieve sector agreement, and support adoption of data standards, harmonisation, and data ingestion, for sector alignment, interoperability and data quality.

Recommendation 7: Production Operations

Establish the strategies, tenders, partnerships and requirements for launching successful production platforms operating as federated NAGIM ecosystems.

Recommendation 8: Innovation in NAGIM

Invest in a sustained strategy of innovation, for new technologies and data types to apply to the NAGIM ecosystem.

WORKSTREAMS & PRIORITY AREAS

Each Recommendation above should be progressed with independent but interrelated **Workstreams** responsible for several **Priority Areas for Action**. Participation of broad stakeholders and robust co-ordination across intersecting workstreams will be necessary.

WORKSTREAM	PRIORITY AREAS FOR ACTION
NAGIM Governance & Coordination (WS1)	• Program governance and oversight • Economic assessments, evaluations and sustainability modelling • Workstream coordination and stakeholder engagement • Reporting, auditing and process evaluation
Architectural Foundations (WS2)	• National data infrastructure planning and strategies for integration • NAGIM research infrastructure implementation pilots • Security and privacy for research systems • Research-clinical infrastructure intersections
Clinical Information Systems & Priorities (WS3)	• Cross-jurisdictional engagement and evaluation • National regulatory frameworks and governance for clinical data • Data standards, minimum requirements and access • Security and privacy for integrated clinical systems • Clinical data pilots and integrations
Data Governance & Ethical, Legal & Social Considerations (WS4)	• National frameworks, policies and agreements for data governance • Ethical, legal and social evaluations • Community engagement and communications • Indigenous data sovereignty • Consent and privacy
Key National Services (WS5)	• Digital identity, access and cybersecurity services • User support services and training • National strategies for use of NAGIM infrastructure • Workforce education, development and engagement
Data Standards & Harmonisation (WS6)	• Consensus on standards and minimum data models • Support technical adoption and implementation of standards • Harmonise and support data ingestion • Clinical and phenotype data harmonisation and use
Production Operations (WS7)	• Tendering and implementation for national operations • Partnerships for operating NAGIM • National security strategies and audits • Long-term sustainability and business continuity planning
Innovation (WS8)	• Development of an independent funding strategy • Tools and innovations for future digital genomics use cases • Tools and innovations for operational management of NAGIM • Tools and innovations for international best practice

OVERALL IMPLEMENTATION STRATEGY

Full national implementation of NAGIM will be a considerable enterprise, both in scale and complexity.

> "Think Big, Start Small, Act Fast."
>
> **NAGIM International Reviewer**

To succeed, the recommended strategy is **an iterative approach delivered with progressive stages**. This would involve targeted initial outcomes or minimum viable products, delivered with iterative development, progressive expansion, and evaluations informed by stakeholders, community and parallel workstreams.

	Delivered	**Short-term**		**Medium-term**			**Long-term & ongoing**		
Year	0	1	2	3	4	5	6	7	→
Primary Delivery Phase	Prototype Phase	**Technical Pilot** WS1, WS2, WS3		**Pre-Production Development** WS1, WS2, WS3			**NAGIM Production: Systems & Services** WS7, WS8 →		
Parallel Workstreams		**Workstreams & Engagement** WS4, WS5, WS6		**Workstreams & Engagement** WS4, WS5, WS6 WS7, WS8			**Production Systems Support** WS3, WS4, WS5 →		
Data Infrastructure		Pilot Data Infrastructure In Use		Pre-Production Data Infrastructure In Use			Prioritised Production Data Infrastructure In Use		

WS1 NAGIM Governance and Coordination
WS2 Architectural Foundations
WS3 Clinical Information Systems and Priorities

WS4 Data Governance and Ethical, Legal and Social Considerations
WS5 Key National Services
WS6 Data Standards and Harmonisation

WS7 Production Operations
WS8 NAGIM Innovation

COMMENCING NAGIM WORKSTREAMS

Progressing implementation of NAGIM should commence with:

A Coordinating Entity

Appoint a coordinating entity to establish overall Program governance, strategy and operations. This entity should be advised by representatives from government, ethics and legal, clinical services, consumer and community groups, industry and data infrastructure providers.

Foundational NAGIM Architecture

Commence a funded NAGIM implementation pilot for research infrastructure, targeting core data management components and key intersections with clinical systems. Cross-jurisdictional engagement on clinical priorities should also commence.

Parallel Workstreams

Assemble key working groups and finalise stakeholders, deliverables and resourcing requirements for workstreams to support preliminary NAGIM infrastructure. These should include those for data governance; ethical, legal and social considerations; national services; and data standards.

Research Data Warehousing

Address the need for a nationally supported research data warehouse. This should reduce current use of overseas data stores and local data silos.

NEXT STEPS

The final scope of works and detailed costings for clinical and research infrastructures will need to be determined to progress the implementation strategy for NAGIM.

Consultation and engagement will be critical to garner the support of Commonwealth, State and Territory Governments: a national genomic data asset will need broad commitment, sustained investment and consensus on governance from all jurisdictions.

Research and clinical organisations are already basing their genomic data management investments and infrastructures on the NAGIM implementation recommendations. We need to move quickly to ensure a nationally coordinated, systematic approach.

A national approach requires partnership with stakeholders across sectors and communities. Key stakeholder groups integral to progressing NAGIM include **clinical services, Indigenous peoples, the community and patients, research institutions and industry**.

With appropriate involvement across stakeholders, the nation can benefit from a national genomic data asset, improving health system efficiency, clinical effectiveness, and enriching research to benefit all Australians.

The lessons learned from other countries will ensure Australia aligns with and benefits from growing international genomics capabilities.

Read the full report on our website:

- Report
- Supplementary Information

"The foresight and resources that Australia and Australian Genomics has dedicated to the development of a national genomics data strategy and implementation plan needs to be commended … this is not easy.

However, it is absolutely essential in order to be competitive in the emerging bio-economy and allow Australians to equitably benefit and participate in the transformative field of genomic medicine.

Go Australia!"

NAGIM International Reviewer

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