InGeNA Consolidated Response to the National Health Genomics Policy Framework and Implementation Plan (2026-2030)

Introduction

This submission presents InGeNA's consolidated industry response to the draft National Health Genomics Policy Framework and Implementation Plan (2026–2030). It reflects insights gathered through the Genomics Industry Roundtable and ongoing consultation with our members, partners, and Consumer Advisory Group.

There was broad consensus from roundtable participants that while the draft framework represents a welcome step forward, it lacks critical implementation detail, particularly in relation to timeframes, measurable outcomes, and funding mechanisms. Genomics is no longer experimental. Clear national targets, accountability mechanisms, and strategic investment are required to transition from pilots and research into routine clinical care.

We believe it is essential for the policy to clearly articulate the cost avoidance and economic benefits of genomics, as well as its social impact. Continued partnership with industry will be vital to ensure that policy development is grounded in practical implementation expertise and real-world innovation.

This document is a complement to our submission via the online Department's consultation platform and to outline our practical recommendations and the rationale behind them. It is designed to ensure key collective issues are clearly articulated and that the final framework is strengthened to support timely and scalable implementation of genomics in Australia's health system.

General Overview

The 2025 National Genomics Policy Framework (NGHPF) is a welcome step towards embedding genomics in Australia's health system. It is positive that the Framework has aligned the federal and state health departments in genomics.
There has been significant evolution of genomics over the past decade since the original Policy Framework was introduced to support alignment between State and Commonwealth activities. Genomics is now embedded more widely across the health system, with a growing number of stakeholders, including patients, healthcare professionals, researchers, and industry engaged in delivering or benefiting from genomic-based care. It is critical that the revised Framework responds to this shift by providing content that supports effective planning, expectation management, and collaboration across this broader stakeholder landscape.

The contributions and potential of these stakeholders is not adequately captured as a framework to align and enable the diversity of the ecosystem to drive progress through to 2030.

Our highlighted priorities for the Government's response include:

1. **Establish a National Genomic Test Directory within five years**.
2. **Set measurable national targets** for implementation at scale (e.g., CGP access, data integration benchmarks).
3. **Introduce a strategic funding priority**, encompassing sustainable financing models, public-private co-investment, a Genomics Innovation Fund, and fast-track funding mechanisms.
4. **Embed FAIR principles** into the national genomic data infrastructure.
5. **Formalise consumer and industry partnerships** under Genomics Australia.
6. **Support implementation through Health Technology Assessment (HTA) reform, workforce development, and regulatory alignment**.
7. **Address jurisdictional inconsistencies**, particularly for access and funding in public hospitals.

As the peak industry body feedback from our members is that **it lacks a sense of urgency and ambition**, particularly when compared with global leaders like the UK, which has set clear objectives through Genomics England. Our key concerns include:

- The language remains **conservative and tentative**, lacking strong delivery-oriented verbs.
- Most activities are still **foundational rather than strategic**, not moving us forward. We believe the policy must shift from an exploratory phase to one focused on implementation, as genomics is already in routine use.
- There is **no national stocktake of current genomics activity or capability** to anchor the framework, nor a strategic "gap analysis" to plan where we want to be in 3, 5, and 10 years. This contrasts with recent genomics frameworks like the National Framework for Genomics in Cancer Care.
- The framework **lacks integration with key stakeholder contributions and activities** already in place, including the roles of industry, universities, and clinical bodies.
- **Desired outcomes and national actions frequently do not match**, or their relevance is unclear.
- **Implementation detail**, such as timeframes, responsibilities, and deliverables, is absent, limiting accountability.
- The **integration of funding into Strategic Priority 2 weakens its visibility and actionability**; we believe a dedicated strategic priority for funding and investment should be reinstated.

- The document risks **keeping genomics in a research and translation phase without sufficient focus on implementation**.
- We need a **bolder, globally competitive vision statement**, similar to Genomics England's leadership framework, with measurable goals. The policy framework should clearly articulate what we will have in place and our goals for 2030, aligning with ambitious existing plans like the cancer genomics framework.
- The framework currently focuses too heavily on testing rather than emerging targeted therapies and treatment applications.
- The framework lacks set targets or outline to what will be delivered
- The guiding principles proposed outcomes do not go far enough to meaningfully support these principles.

We acknowledge that a significant overarching challenge is the inherent difficulty in national policy implementation within a federated system, especially concerning funding models and achieving equitable access. We await the public release of the detailed, concrete, and measurable Monitoring and Evaluation Plan.
We are committed to a collaborative partnership with government and all parties in the genomics ecosystem.

Updated Priorities – Naming and Rationale

The 2025 draft National Genomics Policy Framework reflects a shift in the number and naming of strategic priorities compared to the 2019 framework. While we acknowledge the rationale for these shifts – such as expanding the Person-Centred Approach to include family and community, combining workforce, financing, and services into a Dynamic and Sustainable Genomics Ecosystem, broadening the Data priority, and introducing a new Aboriginal and Torres Strait Islander-led Genomics priority – **we strongly advocate for specific changes and additions to strengthen the framework's ambition and implementation focus.**

The Government's Draft Strategic Priorities are:
1. Person-, Family-, and Community-Centred Approaches
2. Dynamic and Sustainable Genomics Ecosystem
3. Genomics Information Management
4. Aboriginal and Torres Strait Islander-led Genomics

We suggest strengthening the language and structure of these priorities, proposing the following updated naming and the addition of a crucial fifth priority to ensure explicit focus on funding for implementation:
1. **Empowering People, Families, and Communities through Genomics**

2. **Building a Scalable and Sustainable National Genomics Ecosystem**
3. **National Genomics Data & Digital Infrastructure**
4. **Indigenous Leadership in Genomic Research & Healthcare**
5. **Funding and investment to implement genomics**

We have raised the critical issue of funding as a distinct strategic priority in discussions with industry and government, despite the current government draft that it no longer needs to be separate. We maintain that the **assessment and funding mechanisms are a significant barrier limiting adoption and access to genomic testing**, making a dedicated funding priority essential for meaningfully achieving equitable access for all Australians.

Vision Statement

> Vision: *All people who access health care in Australia benefit from appropriately embedded genomics within the health system.*

The statement “appropriately” makes this an unambitious and it is unusual to have such conditional language. InGeNA recommends stronger action verbs and explicit commitments throughout the framework to move beyond tentative language
Compared to Genome UK 2020 and Genomics England’s mission, Australia’s draft framework lacks similar ambition or commitment to transforming healthcare.

Previous framework vision was "helping people live longer and better through appropriate access to genomic knowledge and technology to prevent, diagnose, treat and monitor disease."

Suggested Bolder Vision Statements:

1. **Emphasising Global Leadership and Transformation:**
   Vision: Australia will **lead the world** in establishing an **advanced and integrated genomic healthcare system**, ensuring **universal and equitable access** that **transforms health outcomes** for **all people**.

2. **Focusing on Ubiquitous Integration and Health Impact:**
Vision: Genomics is **fully integrated** across Australia's health system, **delivering personalised care** and **optimising health outcomes** for **every individual**.

3. **Concise and Ambitious:**
Vision: **Transforming health** for **all Australians** through a **world-leading, universally accessible genomic healthcare system**.

Mission Statement

> Mission: *To establish and/or maintain processes that support person- and family-centred, culturally safe, efficient, effective, ethical, and equitable implementation of genomics-informed health care and research in Australia.*

This focuses on foundational processes and maintenance level delivery rather than scaling access to care in Australia and using genomics to deliver a shift in the health system towards prevention, prediction and early intervention. Additionally, that genomics underpins considerable reduction in health system adverse events and waste that is inherent in the overburdened and unsustainable with the current dominant approaches to healthcare delivery.
The UK's Genome UK 2020 explicitly aims to "create the most advanced genomic healthcare system in the world" and to "Transfer world-leading advances in genomic research to front line diagnoses and treatments" to "ultimately provide better health outcomes at lower cost". Genomics England's mission is about "Transforming healthcare: Working with the NHS to deliver and continually improve genomic testing". The Australian mission does not convey such a transformative aim.

Suggested Bolder Mission Statements:

1. **Focusing on Proactive Integration and Systemic Shift:**
   Mission: To **proactively integrate and scale genomics across Australia's health system**, fundamentally **transforming healthcare delivery towards predictive, preventive, and personalised care**, thereby **optimising health outcomes and enhancing efficiency for all Australians**

2. **Emphasising Delivery, Equity, and World-Class Standards:**
   Mission: To **accelerate the delivery of world-class, equitable, and person-centred genomic healthcare** across Australia, **ensuring all individuals benefit from genomic insights** for **advanced diagnostics, targeted interventions, and proactive health management**

3. **Concise and Impact-Oriented:**
   Mission: To **drive the widespread adoption of genomics** to **transform health outcomes** and **deliver sustainable, personalised, and preventive care** for **every Australian**

Guiding Principles

The current draft states the Guiding Principles:

1. Equity
2. Empower individuals and communities

3. Evidence-based and socially accepted
4. Prevention-focused
5. Whole-of-system focused

Our recommendation

1. **Transformative Impact & Global Leadership:**

Rationale: This principle captures the aspiration for Australia to be at the forefront of genomic healthcare, driving fundamental shifts rather than merely maintaining existing processes.

2. **Universal, Equitable, and Person-Centred Access:**

Rationale: This principle expands on the existing equity principle to explicitly demand **widespread and fair access for all Australians**, including those in rural areas, indigenous communities, culturally and linguistically diverse (CALD) communities, and other vulnerable groups, ensuring no one is left behind. It also acknowledges the need for patient advocacy and support services to keep pace with public awareness and service access.

3. **Sustainable, Integrated, and Action-Oriented Implementation:**

Rationale: This principle emphasises a commitment to **active and measurable implementation** of genomics across the entire health system, supported by **sustainable funding models** and seamless integration of research into routine care. It moves beyond "processes" to "deliver" and "scale".

4. **Proactive, Predictive, and Personalised Health Outcomes:**

Rationale: This principle ensures that genomics is leveraged for a **fundamental shift towards proactive, tailored healthcare**, aiming for earlier diagnosis, targeted treatments, and prevention of illness, ultimately leading to better health outcomes at lower cost.

5. **Data-Driven Excellence & Interoperable Infrastructure:**

Rationale: The efficient and ethical management of genomic data is repeatedly highlighted as critical for scalability and effective use.

6. **Agile Innovation & Collaborative Advancement:**

Rationale: This principle commits to fostering a **dynamic environment that encourages rapid translation of research into clinical practice**, adopting innovative pathways, and actively partnering with all stakeholders to accelerate the adoption of new genomic technologies

Enablers of Success:

1. Collaborative and culturally appropriate governance, leadership, information-sharing and, where necessary, coordination across Australian governments
2. Flexibility
3. Individual, community and stakeholder engagement and partnerships

4. International engagement and partnerships

Suggested updates

1. **Strategic & Unified National Governance:**

This enabler expands on "Collaborative... governance" to emphasise the **proactive and integrated leadership required to overcome systemic challenges inherent in a federated health system**. It requires **stronger coordination across Australian governments**, ensuring **consistent and sustainable funding models** and **interoperable data sharing across siloed health systems** (both state and clinical/research). The commitment should extend to formalising governance relationships between national bodies and state-level delivery, and incorporating the diverse contributions of **industry, universities, and clinical bodies** beyond just government entities. Implementing genuine co-design at a national scale also requires **dedicated funding, clear power-sharing frameworks, and robust mechanisms** for resolving potential disagreements, which are not currently detailed.

2. **Agile Adaptation & Responsive Innovation:**

Moving beyond "Flexibility," this enabler champions **dynamic, responsive, and accelerated decision-making processes** to keep pace with the rapidly evolving field of genomics. It means **investigating, developing, and implementing expedited approval pathways** for clinically beneficial technologies, drawing on successful international models like the UK Cancer Drugs Fund. This enabler also highlights the need to explore **alternative, flexible service delivery models** to ensure diverse and equitable access options for all Australians.

3. **Genuine Co-design & Strategic Multi-Sector Partnerships:**

This strengthens "Individual, community and stakeholder engagement and partnerships" by emphasising **authentic co-design and empowered, strategic alliances**. It calls for **formalising partnerships** with key stakeholders, including **consumers, patient advocates, Aboriginal and Torres Strait Islander communities**, and critically, **industry and academia**. These partnerships should be foundational to **enhance investment, accelerate clinical adoption**, contribute to service delivery and data platforms, and support workforce development.

4. **Proactive International Alignment & Global Leadership:**

This enabler elevates "International engagement" to a strategic imperative. Australia needs to **actively benchmark against global leaders** like the UK, learning from their ambition and commitment to transforming healthcare. This includes **drawing lessons from successful international efforts** such as the UK's federated data model to "leapfrog a few years of figuring it out", and modelling key initiatives like a **National Genomic Test Directory** on international systems. It also involves ensuring that any national principles and standards **adhere to global best practice** to facilitate the provision of tools and systems from international industry. This positions Australia not just as an engaged partner, but as a nation striving for **global competitiveness and leadership** in genomic healthcare.

Strategic Priority 1: Person-, Family- and Community-Centred Approach

This priority aims to empower informed decision-making and improve public genomics knowledge. We fully support ongoing efforts to raise public awareness and foster social license for genomics, recognising that public trust was critical to the success of Genomics England.

However, this must be coupled with **sufficient access to services**.

A significant barrier for individuals and families is the **lack of genomics literacy and confidence among healthcare professionals**. Many clinicians feel unprepared to integrate genomics into their practice due to knowledge gaps and a lack of confidence. If access to genomics services does not evolve at the same pace as public awareness, it could create unmet demand and hinder clinician adoption.

Our concerns and recommendations include:

- The current activities **do not outline how they will support the patient advocacy and support services** that deliver a lot of the patient and family awareness and other services in the community. These services are at risk of overload if their value and benefit to the community are not adequately supported.
- We advocate for establishing a **formal Consumer Advisory Group and Ethics and Legal Advisory Group** under Genomics Australia, ensuring co-design and genuine partnership rather than tokenistic consultation.
- Concerns were raised that without investment in community and consumer support, including psychosocial services, the uptake of genomic services may be undermined. There is a **critical gap in post-test support infrastructure** for patients, highlighting the need for ongoing assistance based on genomic test outcomes.
- There is a large array of educational materials already developed. Navigation to access these readily needs coordination and we advocate that the focus needs to on this rather than materials creation.
- The outcomes focus on awareness and education rather than patient access to services.
- Family centred services are lacking which are critical for inherited conditions.
- We stress the importance of a **consistent quality experience across the whole patient healthcare journey with a genomically informed health workforce** to ensure equity.
- The role of **clinical training and genetic counselling needs to be explicitly called out** within this priority. We need a more explicit path for professional development that includes interaction with patient and family advocacy and support services.

- The priorities currently speak primarily to public hospital services, but we need to understand how they will address other touchpoints, such as **GPs and specialist services**, to reflect the diverse ways people access healthcare.

Regarding Equity and Priority Populations:

- While Strategic Priority 4 is dedicated to Aboriginal and Torres Strait Islander-led genomics, the policy also states that "other priority populations will need to be considered" and that all activities will "apply an equity lens". This is not reflected in the actions.
- We express strong support for elevating **families with known inherited conditions, rare diseases, culturally and linguistically diverse (CALD) communities, and rural populations as explicit priority groups**, rather than just applying a broad "equity lens".
- Cultural safety for both Indigenous and non-Indigenous CALD communities is a critical factor.
- We call for a commitment to **"leave no one behind,"** particularly as data gaps and inequities persist in access, counselling, and diagnostics. The framework currently has an oversight regarding regional/remote access beyond Indigenous communities and for an expanded definition of disadvantaged populations. There is a need for sustained long-term support infrastructure beyond testing for inherited conditions.

Strategic Priority 2: Dynamic and Sustainable Genomics Ecosystem

This priority seeks to realise the full potential of genomics-informed healthcare by fostering a dynamic and fit-for-purpose genomics ecosystem that supports research, innovation, workforce development, and infrastructure.

Outcome 2.1: Understand and develop specialised workforce capacity, infrastructure, and health service design &

Outcome 2.2: Support the mainstream workforce

We are concerned that the activities proposed in the draft do not adequately link with education providers to sufficiently increase the genomics literacy and confidence of the workforce across various health disciplines for clinical practice. There is an **urgent need for stronger connections with education providers, clinical colleges, and professional bodies to drive comprehensive workforce development**. Industry also plays a crucial role in supporting professional development and ensuring alignment with current state-of-the-art delivery and foundational understanding.

It is crucial that both specialised genomics professionals and general healthcare workers, such as General Practitioners (GPs) and nurses, understand and can effectively

utilise genomics to support patient decision-making and consent processes. Our specific concerns and recommendations for workforce development include:

- The **lack of explicit recognition of genetic counsellors and their essential role** was repeatedly highlighted in our consultation. However, we also warn against creating bottlenecks and advocate for **team-based, scalable models for counselling**.
- Current education for the existing mainstream workforce is insufficient, negatively impacting their confidence in genomics. The proposed activities do not adequately foster new levels of capability and confidence for the mainstream workforce to integrate genomics into their practice. There is a need to establish stronger links with education providers and clinical practice-influencing bodies to address this gap.
- We promote a multidisciplinary engagement model, similar to that successful in the cancer community, emphasising **on-the-job training over formal academic training** where appropriate.
- **Accreditation, licensing, and the role of clinical education bodies** (e.g., colleges and universities) should be explicitly linked to implementation planning.
- There is a clear lack of a skilled workforce and insufficient remuneration for skilled services.
- The stepping stones listed in Outcome 2.1 do not reflect enough action to ensure the workforce, infrastructure, and services will be developed and put into action.

Workforce needs need to encompass the end-to-end aspects of genomics including talent for **the whole Genomics-Impacted Workforce including:**

- **Specialists** – geneticists, genetic counsellors, bioinformaticians, molecular scientists.
- **Mainstream Clinicians** – GPs, specialists, nurses, pharmacists.
- **Support Services** – counsellors, psychologists, social workers, navigators.
- **Digital & Data** – IT, informatics, data governance professionals.
- **Education** – universities, clinical colleges, training providers.
- **Regulatory & Policy** – ethics, legal, HTA, funding experts.
- **Research & Innovation** – researchers, clinical trial and pipeline experts.
- **Public Health** – epidemiologists, screening, health planners.
- **First Nations Workforce** – Indigenous clinicians, researchers, community leaders.

Outcome 2.3: Decision making processes, funding pathways and sustainable investment to enhance clinical translation

Funding and Reimbursement Pathways (Major Industry Concern):
We explicitly recommended the addition of a fifth strategic priority titled "Funding and investment to implement genomics," considering it vital to ensure an "explicit focus on

funding for implementation". Our proposals included a long-term, sustainable, and nationally coordinated funding model, equitable funding across states and territories, reimbursement reforms, public-private co-investment, a Genomics Innovation Fund, and a fast-track funding mechanism. We believe that Outcome 2.3, which deals with these issues, needs to be a strategic priority, as it represents a major barrier to equal access for all Australians.

A critical issue is that current state funding for public hospitals often **prevents claiming Medicare Benefits Schedule (MBS) benefits for genomic testing, leading to unequal access for public patients**. We suggested considering complex reforms to shift all genomic testing to the MBS to allow for monitoring of test access and utilisation, which would highlight disparities across locations and jurisdictions. We advocate for embedding funding for genomics-based care into the National Health Reform Agreement (NHRA) to secure sustainable financial support and address disparities.

The 2025 draft integrates funding aspects under Strategic Priority 2, rather than as a standalone priority as it was in the 2019 framework. The draft's language, such as "developing options" and "promoting consideration," is perceived by us as less committal than our proposed specific mechanisms like a "Genomics Innovation Fund" or a "fast-track funding model". It does not explicitly address the fundamental issues of consistent federal and state funding or direct MBS reform for public hospital genomic testing.

Our suggested improvements include:

- **Elevate Funding:** Re-introduce "Financing" as a distinct strategic priority, as it was in the 2019 framework, to provide a more explicit focus on funding for implementation.
- **Stronger Action Verbs:** Change "Promote consideration of genomics as part of broader HTA improvement" to **"Actively integrate genomics into broader HTA improvement and reform processes to establish efficient and timely adoption pathways for genomic applications into clinical standard of care"**.
- **Specify Funding Mechanisms:** Instead of "Develop ethical models for partnerships," we propose to **"Establish and fund specific mechanisms for public-private partnerships and co-investment, including a Genomics Innovation Fund, to accelerate translational research and clinical adoption of validated technologies"**.
- **Commit to NHRA Integration:** Directly state **"Embed funding for genomics-based care into the National Health Reform Agreement (NHRA) to secure sustainable financial support and address disparities in genomic testing access across states and territories"**.
- **Establish alternative reimbursement pathways and funding mechanisms** to enable the efficient and timely adoption of genomics applications into clinical standard of care.
- **Recommend a minimum ratio for clinical implementation funding relative to research funding**, including investment in major infrastructure.
- We identify the **lack of infrastructure within existing pathology networks and minimal to no support for the adoption of new technology** as significant barriers.

- The current funding is minimal with confusing and restrictive conditions, placing a large onus on sponsors to create applications with no guarantee of success. We seek **collaborative and proactive support from government to identify testing per national priorities**.
- The language around supporting alternative investment and funding models at the Federal level is very soft.
- If MBS funding is utilised, we must establish an **alternative assessment pathway that incorporates early value assessment for technology that addresses unmet clinical needs and allows for the generation of real-world evidence**.
- It is likely necessary to explore alternative service delivery models to ensure diverse and flexible access options for all Australians, as well as explore funding options for wrap-around services linked to genomic-led care plans.
- Outcome 2.3 does not recognise the inherent barriers existing today in terms of access to and funding for instrumentation and other necessary infrastructure, the significant delays in funding through existing HTA pathways, the patchy coverage of existing funding across test menu, and the absence of a universally accessible and easy-to-understand test directory.
- We need to more clearly identify the funding pathways, including early value assessment processes, mission-based approaches that "pull" in relevant technology and tools, and outcomes/value-based assessment pathways.
- The current HTA processes must shift from disease-based to technology-based assessment and support real-world data, small-n populations, and personal utility. Australia is seen as lagging, and the adoption of innovative pathways (e.g., NICE's ILAP or UK interim funding) is strongly encouraged.

Fast-Track or Alternative Regulatory Models (Major Industry Concern):
We propose an **"Accelerated Approval Models," such as the UK Cancer Drugs Fund (CDF)**, to simplify regulatory pathways. The UK's CDF model features separate approval pathways for complex technologies, accelerated access through interim funding and usage, and flexible reimbursement models.
The 2025 draft generally mentions "active engagement, cooperation and collaboration with domestic and international stakeholders" and developing "ethical models for partnerships... and co-investment". However, it **does not explicitly address specific regulatory barriers, delays, or the lack of "appetite" for public-private partnerships** that we identify as critical impediments. Concrete actions for "Accelerated Approval Models" or detailed "simplified regulatory pathways" are not outlined as distinct initiatives. We urge the government to adopt an agile, "trial-and-refine" approach to decision-making processes.
Our suggested improvement is to propose to "Investigate, develop, and implement expedited approval pathways for clinically beneficial genomic technologies, drawing on successful international models such as the UK Cancer Drugs Fund, to reduce regulatory delays and accelerate patient access". This shifts the language from merely

considering to proactive investigation and implementation. We also note that HTA review should involve a single pathway agreement for genetic/genomic technology assessment. We have removed the need to demonstrate cost effectiveness a priori when it has not had the opportunity to implement despite clinical effectiveness.

Furthermore, Outcome 2.4, while supporting research, is too focused on the research aspect of appropriate data generation for HTA and fails to recognise that **existing HTA processes are not fit for purpose for genomics**. Any evolution in funding or "financing" needs to consider mechanisms for assessing "cost-effectiveness" to include the full value chain within HTA processes.

Outcome 2.4: Support research and innovation

Less Committal Language on Transitioning Research Programs into Mainstream Clinical Practice (Major Industry Concern):

We explicitly call for the **"transition of successful genomic research programs,"** such as ZERO Childhood Cancer and PrOSPeCT, **into mainstream clinical practice and into the MBS"**. We argue this is essential for broadening the reach of genomic medicine beyond research into everyday healthcare.

The 2025 draft includes an activity to "Develop options for long-term, sustainable, and scalable integration of genomics-informed national pilot initiatives and research-funded programs into clinical practice where they are demonstrated to be clinically beneficial and cost-effective".

We assess that the phrasing **"Develop options for... integration" is less definitive and immediate than our call for "transition," suggesting a more exploratory and less committed approach** to embedding these proven programs into routine care and funding.

Our suggested improvement is to replace "Develop options for" with "Develop and implement structured funding mechanisms and pathways that deliver the long-term, sustainable, and scalable integration of clinically beneficial genomics-enabled national pilot initiatives and research-funded programs into mainstream clinical practice and appropriate funding schemes (e.g., MBS)."

Additionally

Establish a National Pathway and Fund for Therapeutic Development in Ultra-Rare Genetic Diseases

Australia must implement clear pathways to enable the development and delivery of innovative therapies for rare and ultra-rare genetic conditions. Despite having the technology and capability to diagnose and develop personalised treatments, patients are left behind due to the lack of funding, regulatory flexibility, and implementation infrastructure.

For ultra-rare conditions, traditional clinical trials and health technology assessments (HTAs) are unfeasible. A new paradigm is needed—one that enables both the repurposing of existing drugs and the development of bespoke genomic therapeutics.

We recommend the following actions:

1. **Establish a Rare Diseases Therapeutic Innovation Fund**
   A dedicated MRFF-backed fund should support translational and therapeutic development for ultra-rare conditions, enabling equitable access to innovative treatments. This should leverage Australia's existing clinical-grade capabilities in gene editing, organoid models, and manufacturing platforms.
2. **Develop a National Implementation Pathway for Bespoke Therapeutics**
   Regulatory and reimbursement frameworks must accommodate small-n, high-impact personalised therapies. Australia should adopt accelerated and adaptive pathways modelled on international best practice such as the NIH Bespoke Gene Therapy Consortium and UK's ILAP/Cancer Drugs Fund.
3. **Expand the Role of the Australian Functional Genomics Network**
   Its remit should include phenotype validation for genes of known function using patient-derived organoids, enabling rapid, patient-matched compound testing through AI and platform technologies.
4. **Leverage Bioplatforms Australia and NCRIS via a National Voucher Scheme**
   Enable families and clinician-researcher teams to access infrastructure and feasibility testing tools to support therapeutic hypothesis development.
5. **Establish an International Consortium for Genomics and Rare Disease Therapeutics**
   Co-design a multilateral framework with global partners (UK, US, CA, EU, SG, etc.) to share data, trial platforms, regulatory strategies, and public funding initiatives to fast-track global rare disease therapeutic access.

This initiative would build on the success of the MRFF-funded leukodystrophy Massimo's mission and expand its reach, ensuring diversity, equity, and scalability for the rare disease community. Clear policy will enable the diverse industry to be play a role in bring therapies to patients.

Horizon Scanning:

We call for establishing and maintaining a **collaborative horizon-scanning process for emerging health genomics trends, research, and technologies**, actively involving consumers, industry, clinicians, and researchers. This process should inform prioritisation and align with broader national and state-based activities.

While Activity 2.4.4 in the 2025 draft mentions establishing and maintaining targeted horizon scanning for emerging trends to inform prioritisation, **our suggested improvement is that the process should explicitly and actively involve consumers, industry, clinicians, and researchers.**

We believe **industry is best placed to support these activities** due to our 5-10 year product pipeline knowledge. We also believe that horizon scanning activities should be

managed at a Federal level, as the population is too small to focus on State-level capacities.
Furthermore, there is a need to assess existing capabilities in Australia, including local skills, companies, and intellectual property, to leverage them domestically and for export. The framework should also draw lessons from international efforts, such as the UK's federated data model, to potentially "leapfrog a few years of figuring it out".

Industry Collaboration and Partnerships:

We recommend establishing and implementing strategic partnerships to enhance investment and clinical adoption of genomics, including public-private partnerships. We also suggest developing ethical frameworks for collaboration, engagement, and co-investment with non-government stakeholders that prioritize public benefit and accelerate innovation implementation.
The 2025 draft's approach includes developing "ethical models for partnerships, engagement, and co-investment with non-government stakeholders, including industry and academia" and establishing strategic partnerships.
Our suggested improvement is to **modify the activity to specifically "Establish and implement strategic partnerships to enhance investment and clinical adoption of genomics, including public private partnerships. Develop ethical frameworks for collaboration, engagement, and co-investment with non-government stakeholders, including industry and academia, that prioritise public benefit and accelerate innovation implementation"**.
The roundtable discussions strongly emphasised that **industry must be an integral part of the solution**, contributing to service delivery, data platforms, innovation, and implementation. There were strong calls for a **clear public-private partnership framework**, and for removing barriers to local procurement and adoption of homegrown innovation. We also believe there is currently "soft language around supporting alternative investment and funding models at Federal level", which needs to be strengthened to foster genuine partnerships. Finally, we recommend formalising consumer and industry partnerships under Genomics Australia.

National Target for Universal Access to Comprehensive Genomic Profiling (CGP) for Cancer Patients (Major Industry Concern):

We call for a **commitment to "making Comprehensive Genomic Profiling (CGP) available to all cancer patients within five years"**. We argue this would significantly reduce barriers to biomarker testing, expedite access to precision oncology medicines, and improve lab efficiencies by reducing the need for multiple small panels and minimizing test failures. This aligns with our recommendation to align the framework's vision with existing ambitious plans like the cancer genomics framework.

While the 2025 draft acknowledges "genomics in cancer care involving both germline and somatic variants" and cross-references the "National Framework for Genomics in

Cancer Control," emphasising "shifts to prevention, early intervention, and precision medicine", it **does not explicitly state "universal access to CGP for all cancer patients within five years" as a direct, quantifiable target or a specific strategic activity**. This is a major, actionable goal for InGeNA that is not directly reflected.
Our suggested improvement is to add a new, measurable outcome, such as "Achieve universal access to Comprehensive Genomic Profiling (CGP) for all eligible cancer patients across Australia by [specific target year within the framework's timeframe, e.g., 2030]," and to include activities that directly support this goal. We note that this recommendation does not sit readily in the current structure of the Priorities and Objectives, and therefore recommend that they be updated to ensure that critical areas such as this are readily captured and stand out.

Strategic Priority 3: Genomic Samples, Data and Information Management

This priority focuses on the responsible, secure, and appropriately governed collection, storage, analysis, use, sharing, and management of biological specimens and genomic data.

Outcome 3.1: Safe, secure and trusted data sharing, storage and management

As industry, **we believe we should present a united front to reassure policymakers about end-to-end data and information management, particularly around data security**. This includes not only setting security standards but also implementing mechanisms for regulation and compliance.
We assert that genomic data should adhere to FAIR principles to ensure safe, effective use across clinical and research domains. Given the high value and sensitivity of genomic samples, it is critical to have clear mechanisms to demonstrate adherence to data standards. Concrete measures are needed to secure data, enable interoperability, and ensure proper funding for the entire data management ecosystem, not just producing test results.
Genomic data should be regarded as a national security level of focus and attention.

We also believe that **policy leadership is needed in the legal and regulatory space**, as laws more than 30 years old are inadequate for current genomic data practices. There are four key areas for this priority: building on existing infrastructure, establishing strategic funding, implementing security regulations, and creating a connected national test directory.

Lack of Explicit Reference to FAIR Data Principles (Major Industry Concern):
We strongly advocate for a **national genomic data infrastructure based on the FAIR (Findable, Accessible, Interoperable, Reusable) principles**. There **are no requirements or mechanisms for labs to have FAIR data, which causes a lot of waste and does not**

enable genomics to scale in Australia". We stress that FAIR data supports reuse across research and clinical domains, improves patient outcomes, and protects irreplaceable samples.

While Outcome 3.1 and Activity 3.1.2 in the 2025 draft reference the development of standards-based approaches to information management, including interoperability, security, and privacy, they **do not explicitly commit to the FAIR framework**. We assess that this omission signals a less comprehensive approach to data utility, undermining scalability, reusability, and cross-sector integration. The framework also lacks mechanisms for industry to demonstrate compliance with data standards and does not outline concrete measures for ensuring long-term data value and security.

Our suggested improvement is to integrate FAIR principles directly into both outcomes and activities:

- Outcome 3.1 should read: **"National approaches are developed and implemented that facilitate the FAIR (Findable, Accessible, Interoperable, Reusable) and interoperable sharing of genomic information."**
- Activity 3.1.2 should read: **"Develop culturally informed, future-proof, and sustainable principles- and standards-based national approaches to genomic biological specimen and information management, explicitly based on FAIR principles, for clinical settings and Australian government-funded research."** We also believe it is essential to explicitly state that any principles and standards would adhere to global best practice to avoid inadvertently creating additional barriers when it comes to the provision of tools and systems supplied from industry.

Make Explicit National Genomic Test Directory (Major Industry Concern):

We strongly and unanimously support establishing a **National Genomic Test Directory (NTD)**, modelled on the UK system, to be implemented within five years. The NTD would support "structured and standardised approaches to genomic testing," ensuring equitable access, clinical consistency, and streamlined technology adoption. It would also alleviate pressure on the Medical Services Advisory Committee (MSAC). We believe this directory would streamline access to tests, improve transparency across public/private systems, reduce duplication and delays, and decouple test funding from medicine reimbursement.

The 2025 draft references "nationally consistent genomic test result reporting" and "best practice guidelines," but **does not include an NTD**. This is a notable gap; a test directory is a practical and impactful tool that we see as essential for harmonising clinical adoption and funding pathways nationally.

Our suggested improvement is to include a new activity under Strategic Priority 3: "Evaluate the feasibility and establish a plan for the implementation of a National Genomic Test Directory, drawing on international models like the UK, to standardise testing protocols, streamline access, and ensure clinical consistency across

Australia". We also highlight the need for electronic ordering and reporting systems that go beyond a proposed test directory.

Data Sovereignty, Security and System-Wide Scalability:

Industry stakeholders call for **genomic data security and sovereignty to be elevated to a national interest**. Genomic samples and data are often irreplaceable, and their mishandling can cause harm or risk to individuals. There is a need for nationally consistent, enforceable data standards, with mechanisms to demonstrate compliance across public and private sectors. We believe policy leadership is needed in the legal and regulatory space, as current laws (30+ years old) are inadequate for genomic data practices.

While the 2025 draft discusses interoperability, privacy, and consent, it **does not fully articulate mechanisms for end-to-end data security, oversight, or industry co-designed standards**. Without enforceable standards, clear governance mechanisms, and cross-sector accountability, genomic data infrastructure will remain fragmented and inefficient, and will not meet the needs of a future-proof precision health system. We also note the lack of interoperability of data across siloed health systems, both between states and between clinical and research contexts.

Our suggested improvements include:

- Add a new outcome under 3.1 focused on data security and sovereignty: **"Australia's genomic data infrastructure is governed through enforceable standards and best practices that support data sovereignty, transparency, and accountability, enabling secure and trusted use of genomic information across clinical and research contexts."**
- Add a supporting activity: **"Co-develop with industry and other stakeholders a national data sovereignty and security framework that includes accreditation mechanisms for compliance with FAIR-aligned data standards."**
- We strongly support integrating clinical and research data platforms, especially to facilitate real-world evidence generation and reduce duplication.
- National coordination is needed to repatriate genomic data currently sent overseas, ensuring clinician and patient access and control.
- The framework needs a clinical utility focus with a maturity model from research through clinical implementation, requiring population-scale comparison capability.
- The framework narrowly focuses on evaluating specific platforms like Shariant and PanelApp rather than a broader digital genomics infrastructure assessment.

Scalability and Infrastructure Funding (Critical for Implementation):

Realising genomics at scale requires secure, connected, and scalable infrastructure. Stakeholders highlighted the importance of **funding mechanisms that support the safe**

management of genomic data—not just individual tests. We believe that necessary infrastructure is doing a lot of heavy lifting in the current draft, and more explicit outcomes directed to this point are needed to meaningfully achieve goals.
The framework does not identify funding for the data infrastructure layer or mechanisms to support digital platforms that serve the whole system (beyond tools like PanelApp or Share). **We believe this is a critical gap for implementation.**
Our suggested improvements include:

- Add language under Outcome 3.1 recognising that **infrastructure investment must support the entire data lifecycle**.
- Recommend inclusion of a national infrastructure investment activity such as: **"Identify and fund scalable, secure genomic information platforms that support interoperable, whole-of-system data use, including patient-facing communications, real-world evidence, and integration with national digital health records."**
- We need to explicitly identify the mechanisms related to critical data infrastructure in terms of funding, management, and ongoing maintenance and evolution.
- We also note the lack of infrastructure within existing pathology networks and minimal to no support for the adoption of new technology.

Strategic Priority 4: Aboriginal and Torres Strait Islander-led Genomics

This priority emphasises working in partnership with Aboriginal and Torres Strait Islander peoples to build trust, ensure cultural safety, and improve equity of access and outcomes in genomics.

Operationalisation of "Co-Design" and "Partnership" without Specific Mechanisms or Dedicated Funding:

Strategic Priority 4 heavily emphasises "co-design," "leadership," and "partnership," stating that all activities under this priority will be progressed in co-design with relevant Aboriginal and Torres Strait Islander stakeholder groups, ensuring wide representation of Indigenous voices.
However, we recognise a **potential issue that implementing genuine co-design at a national scale can be resource-intensive, complex, and slow without dedicated funding, clear power-sharing frameworks, and robust mechanisms for resolving potential disagreements**, which are not detailed in the draft beyond a general commitment. There is a concern that a collective approach could potentially delay individual access for Indigenous people. We also note the highlighted **structural challenges in proving benefits for Aboriginal and Torres Strait Islander populations in global clinical trials**, and the need for cultural safety requirements and specific reference genomes, emphasising a true partnership approach.

Addressing "Equity" for Other Priority Populations:

Strategic Priority 4 is dedicated to Aboriginal and Torres Strait Islander-led genomics, the policy also states that "other priority populations will need to be considered" and that all activities will "apply an equity lens". However, **specific, detailed mechanisms or dedicated activities for addressing the unique needs and barriers faced by other vulnerable groups** (e.g., those in remote areas, with low socio-economic status, or non-Indigenous culturally and linguistically diverse communities) **are not as prominently articulated**. This could lead to these groups remaining underserved if not proactively addressed through concrete action plans.

We express strong support for **elevating families with known inherited conditions, rare diseases, CALD communities, and rural populations as explicit priority groups**, not just applying a broad "equity lens". We believe there is an oversight in the framework regarding regional/remote access beyond Indigenous communities and for an expanded definition of disadvantaged populations, including CALD communities and paediatric patients.

We call for a commitment to **"leave no one behind,"** particularly as data gaps and inequities persist in access, counselling, and diagnostics. There is a need for sustained long-term support infrastructure beyond testing for inherited conditions for these groups. We also note the absence of explicit outcomes to support representative databases and universal accessibility across other populations.

Closing Comments

There was broad consensus from our roundtable participants that the draft framework **lacks crucial implementation detail, timeframes, and measurable outcomes**. We believe that the cost avoidance benefits and social impact importance in genomics implementation should be clearly highlighted. We advocate for continued industry engagement as the policy work develops.

Specifically, we asked for:

- **National targets** (e.g., universal CGP access by 2030, data integration benchmarks).
- **Cost-avoidance modelling and economic indicators**.
- A **clear figure or schema showing "what good looks like"** in genomic service delivery.

Our highlighted priorities for the Government's response include:

1. **Establish a National Genomic Test Directory within five years**.
2. **Set measurable national targets** for implementation at scale (e.g., CGP access, data integration benchmarks).
3. **Introduce a strategic funding priority**, encompassing sustainable financing models, public-private co-investment, a Genomics Innovation Fund, and fast-track funding mechanisms.
4. **Embed FAIR principles** into the national genomic data infrastructure.

5. **Formalise consumer and industry partnerships** under Genomics Australia.
6. **Support implementation through Health Technology Assessment (HTA) reform, workforce development, and regulatory alignment.**
7. **Address jurisdictional inconsistencies**, particularly for access and funding in public hospitals.

This submission was developed in consultation with our members, partners and our consumer advisory group.
We are committed to ongoing engagement with Genomics Australia and the Department of Health to shape a more ambitious, equitable, and implementable national genomics policy.

Appendix

Summary Enablers from Industry

We advocate for deep, and proactive collaboration with industry. This is considered critical for the success and ambition of the National Health Genomics Policy Framework (NGHPF). The current draft is criticised for its "lack of integration" of industry contributions and activities, which stakeholders believe are essential to deliver outcomes into the future.
Key ways the NGHPF needs to work with industry:

- **Elevate Industry as a Core Partner in Strategic Planning and Governance:** The framework currently "lacks integration to other key stakeholder contributions and activities... This includes the role of industry, Universities, clinical bodies. Industry roundtable participants emphasises that **industry must be part of the solution, contributing to service delivery, data platforms, innovation, and implementation**. There's a strong call to **formalise consumer and industry partnerships under Genomics Australia**, moving beyond "tokenistic consultation" towards "co-design and genuine partnership". Industry also highlights the need for clarity on the governance relationship between national body and state-level delivery, impacting their ability to contribute effectively.
- **Prioritise and Reform Funding & Investment Mechanisms:** This is a major industry concern.
- Industry explicitly recommends reinstating **Funding and investment to implement genomics as a distinct strategic priority**, arguing the draft's current approach under Strategic Priority 2 is less committal.
- We advocate for a **long-term, sustainable, and nationally coordinated funding model**, including **public-private co-investment**, a **Genomics Innovation Fund**, and **fast-track funding mechanisms**.
- Industry seeks **proactive government support** in identifying testing based on national priorities, rather than the current system which places a large onus on sponsors to create application with no guarantee of success.

- We urge for **embedding funding for genomics-based care into the National Health Reform Agreement (NHRA)** to ensure sustainable financial support and address disparities across states and territories.

Accelerate Approval Pathways and Reform Health Technology Assessment (HTA):

- Industry urges the government to **fast-track approval and funding pathways** by investigating and implementing expedited approval pathways for clinically beneficial and cost-effective genomic technologies.
- We suggest drawing on successful international models like the **UK Cancer Drugs Fund (CDF)** and adapting HTA processes to support real-world data, small patient populations, and personal utility, rather than solely disease-based assessment.
- This includes the need for an alternative assessment pathway that incorporates early value assessment for technology that addresses unmet clinical needs.
- **Support Workforce Development and Capacity Building:** Industry plays a crucial role in supporting professional development and ensuring it aligns with current state of the art delivery. This involves stronger connections with education providers and clinical bodies to drive comprehensive workforce development and address genomics literacy gaps.

Co-Develop Robust Data Infrastructure and Standards:

- Industry stresses the need for a **federated genomic data infrastructure, with secure, interoperable, and FAIR (Findable, Accessible, Interoperable, Reusable) principles embedded**. We note that the current lack of FAIR data requirements "causes a lot of waste and does not enable genomics to scale in Australia".
- We call for **co-developing a national data sovereignty and security framework with industry and other stakeholders** that includes accreditation mechanisms for compliance.
- Industry is crucial in ensuring that **any national principles and standards adhere to global best practice** to avoid creating additional barriers when it comes to provision of tools and systems supplied from industry.
- **Implement a National Genomic Test Directory (NGTD):** There is strong, unanimous support from industry for a **funded NGTD, modelled on the UK system**, to streamline access, improve transparency, reduce duplication, and decouple test funding from medicine reimbursement. This is considered a major gap in the current draft.

Transition Research Programs into Routine Clinical Practice:

- Industry advocates for a stronger commitment to **"implement and scale"** successful research programs like ZERO Childhood Cancer and PrOSPeCT into mainstream clinical practice and funding mechanisms like the MBS.

Leverage Industry for Horizon Scanning:

Industry has 5-10 year product pipeline knowledge for horizon scanning and is best placed to support these activities. We call for a collaborative horizon-scanning process that explicitly involves industry, consumers, clinicians, and researchers.

In summary, the feedback from industry emphasises a shift from a conservative and tentative framework to one that embraces **stronger action**, **explicit commitments**, and **formalised mechanisms for collaboration and co-investment**, recognising industry not just as a stakeholder, but as an **integral driver of implementation, innovation, and sustainability**