POLICY POSITION DOCUMENT

National Rare Disease Registry for Australia

Medicines Australia

Background and Context

Rare diseases affect an estimated two million Australians, with approximately 80% of conditions of genetic origin. There are more than 7,000 rare diseases that are life threatening or chronically debilitating. Despite this burden, Australia lacks coordinated national data collection. Hospital systems code only a small fraction of rare diseases by international standards, congenital anomaly data are incomplete, and jurisdictional fragmentation obscures patient pathways and outcomes. Existing disease-specific registries are unlinked and ad hoc, entrenching diagnostic delay, inequitable access, and limited research capacity.

The McKell Institute's 2025 report, A Rare Kind of Care, concludes that Australians living with rare disease face a 'postcode lottery' – with long diagnostic journeys, poor treatment access, and missed opportunities for clinical trials varying significantly by jurisdiction. The World Economic Forum (WEF)'s 2026 white paper, Making Rare Diseases Count: How Better Data Can Unlock a Multi-Trillion Dollar Opportunity, reinforces that around 95% of rare diseases have no authorised treatment, and identifies better data as the single most powerful lever for accelerating progress across health, economic, and scientific dimensions.

No state or territory has a comprehensive strategy or action plan for rare disease which provides an overarching framework for delivering health equity for rare disease patients.[1]

The National Strategic Action Plan for Rare Diseases identifies comprehensive data collection and linkage as a critical enabler. The New Frontier report calls for strengthened patient voice, real-world evidence, and an HTA pathway fit for ultra-rare conditions – all dependent on robust patient-level data. A standardised (interoperability) national registry (hereafter referred to as **Registry**), paired with improved rare disease ICD coding across hospitals and the NDIS, would provide the cumulative knowledge needed to inform decision-makers, underpin research, and support accountable, person-centred care.

Preamble

This policy position aligns with:

1. The National Strategic Action Plan for Rare Diseases (Priority 3.1),
2. HTA Review recommendations on real-world evidence,
3. Recommendations for a national approach to rare disease data by Rare Voices Australia and Monash University,

[1] McKell Institute, A Rare Kind of Care. (2025, August). 'The role of states and territories in rare disease', page 24

4. The New Frontier recommendations on precision medicine access, patient involvement, and transparency,
5. Medical Services Advisory Committee's advice regarding implementation of new conditions in newborn screening programmes, states that a publicly funded registry should be developed and commenced simultaneously with newborn screening, to enable long-term follow-up to establish incidence of each disease subtype and their genotype–phenotype relationships and clinical outcomes data,
6. The McKell Institute's 2025 report, A Rare Kind of Care, which maps Australia's rare disease policy landscape and identifies coordinated data collection as a critical unmet need, and
7. The World Economic Forum's 2026 white paper, Making Rare Diseases Count, which provides an international evidence base and five-strategy roadmap for strengthening rare disease data systems worldwide.

Introduction

The National Rare Disease Registry is intended to operate as a federated national data network. It will link, harmonise and enable analysis across existing disease-specific registries and administrative datasets, rather than replace or centralise them into a single monolithic database. Data will remain with existing custodians, while common standards, governance and interoperability arrangements enable national-level insight.

This approach builds directly on Australia's existing registry infrastructure, including national registries such as the Australian Neuromuscular Disease Registry and MSBase, which demonstrate the feasibility of longitudinal rare disease data collection at scale.

A Registry would systematically capture high-quality, longitudinal, person-centred data across the rare disease spectrum, linking clinical, genomic, hospital coding (ICD), NDIS and social care datasets. This may reduce uncertainty in diagnosis and care, illuminate unmet need, strengthen HTA and funding decisions, accelerate research and innovation, and support service planning. A Registry would systematically capture high-quality, longitudinal, person-centred data across the rare disease spectrum, linking clinical, genomic, hospital coding (ICD), NDIS and social care datasets. This may reduce uncertainty in diagnosis and care, illuminate unmet need, strengthen HTA and funding decisions, accelerate research and innovation, and support service planning.

In line with the National Strategic Action Plan for Rare Diseases (Priority 3.1) and recommendations from the New Frontier report and HTA review agenda, a Registry, fuelled by a comprehensive health data linkage would provide the evidence base would provide the evidence base needed to realise timely access to effective therapies, coordinate care nationally, and enable international data collaboration for very small populations.

Alignment with the National Strategic Action Plan for Rare Diseases: Priority 3.1

Priority 3.1 of the National Strategic Action Plan for Rare Diseases calls to 'enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning'.

A national rare disease Registry is the most direct and meaningful mechanism for fulfilling this priority.

> **What Priority 3.1 requires:**
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> *Coordinated and collaborative data collection that facilitates monitoring and cumulative knowledge of rare diseases, improved care management, support for research, and health system planning. The McKell Institute's 2025 assessment of each Australian jurisdiction against this priority found that no state or territory has taken a systematic approach sufficient to meet it. Western Australia's suite of data programmes – including the Western Australian Register of Developmental Anomalies with its implementation of rare disease coding (OPRHAcodes) – represents the closest to full implementation of this objective, but national coordination remains absent.*

The McKell Institute's 2025 assessment of each Australian jurisdiction against Priority 3.1 confirmed the following:

- No jurisdiction has achieved full implementation of Priority 3.1.
- Congenital anomaly data collections represent the most consistent activity across jurisdictions, but these are narrow in scope and are not interlinked nationally.
- Victoria's CareSync Exchange and Western Australia's suite of data linkage programmes represent notable progress, but these remain state-level and do not constitute a national coordinated system.
- The Northern Territory, Australian Capital Territory, and Queensland show no discernible qualifying activity against this priority.
- The absence of a nationally endorsed implementation plan, backed by funding and governance mechanisms, means there is no clear pathway for ensuring progress, accountability, or sustainability.

The WEF's Making Rare Diseases Count report provides an international frame of reference. It finds that many countries lack basic data on rare diseases, that this lack of alignment creates blind spots around a significant cost in the healthcare system, and that an internationally harmonised minimum dataset would provide a common language for understanding rare disease impact and opportunities. The report recommends that countries define and track a minimum dataset aligned to international classification systems – notably ORPHAcodes and ICD-11 rare disease extension codes – as a practical starting point.

Australia is well placed to move. Many countries – starting with France and Germany, extending across Europe and now reaching Australia and Canada – have already embedded ORPHAcodes into public health systems, as the WEF report notes. Western Australia's Register of Developmental Anomalies has implemented ORPHAcodes and serves as a domestic proof of concept. A national Registry would build on these foundations.

The Case for a Coordinated National Data Registry

The evidence base supporting a national rare disease Registry draws from three complementary sources: the direct experience of Australians living with rare disease, the findings of the McKell Institute's 2025 national policy audit, and the WEF's global analysis of data system investments. Taken together, these sources make a compelling, evidence-grounded case.

Addressing the 'Postcode Lottery'

The McKell Institute's 2025 report documents that Australians living with rare disease are subject to a postcode lottery – where access to medical support is significantly influenced by where they live. No state or territory has a current, dedicated state-wide rare disease strategy or action plan providing an overarching framework for supporting people living with rare disease. The report's mapping exercise confirms that policy responses across all three pillars of the Action Plan are patchy, inconsistent, and often dependent on the efforts of individual 'rare disease champions' rather than embedded systemic planning.

A national Registry directly addresses this inequity. By creating a single, connected data system accessible across jurisdictions, a Registry ensures that clinical and service data for patients in the Northern Territory is as visible and usable as data for patients in New South Wales. It removes the dependence on fragmented, state-level data collections and enables national monitoring of outcomes and access gaps.

Generating the Evidence Needed to Close Gaps

The McKell Institute identifies a lack of coordinated data collection as one of the most consequential gaps in Australia's rare disease policy framework. The report notes that the absence of national coordination is most noticeable in areas widely accepted as benefitting from national leadership, such as workforce development, research collaboration, and data collection. It observes that pockets of rare disease expertise are dispersed and are not organised to collaborate in a systemic way.

The WEF report reinforces this, finding that the challenge is not just to collect more data but to ensure it is meaningful and actionable. It sets out a theory of change: better data can generate evidence of need, guide investment, and drive innovation. Patient registries – described as structured databases that collect and track health information from individuals living with a specific condition – are identified as engines of innovation when well designed and when patient insights are captured systematically.

Economic case: diagnostic delay costs

The WEF's 2026 report cites a study[2] finding that delays in diagnosis across seven rare conditions led to avoidable costs of up to US$517,000 per patient. While these figures reflect a limited set of diseases, they illustrate how prolonged diagnostic journeys impose substantial economic and human costs that are echoed across many rare conditions. A national registry, by enabling earlier and more accurate identification of rare disease patients, directly reduces these avoidable costs.

Supporting the Broader Architecture of Rare Disease Reform

The McKell Institute's 2025 report recommends the establishment of an Office of Rare Disease under a Rare Disease Act, the implementation of Centres of Expertise, a reformed PBS pathway for rare disease therapies, and a national workforce strategy. Each of these reforms is dependent, to varying degrees, on the availability of coordinated national data.

An Office of Rare Disease, for example, would require a data infrastructure to oversee national implementation of the Action Plan, identify gaps, and report on progress. Centres of Expertise would rely on Registry data to identify patients, coordinate care pathways, and evaluate their own outcomes. PBS and HTA reform require real-world evidence that can only be generated from a systematic,

[2] EveryLife Foundation for Rare Diseases. (2023, September 14). The cost of delayed diagnosis in rare disease: A health economic study.

longitudinal data collection. A workforce strategy must be grounded in evidence of where specialist capacity is needed – which a Registry would provide.

The WEF report makes a similar observation at the global level, noting that rare disease progress depends on how effectively data can move across institutions and borders. A trusted data-sharing ecosystem allows information to flow securely between stakeholders while respecting privacy, ethics and sovereignty. The report identifies federated data systems as the preferred model – in which datasets stay where they are generated but insights travel freely – and calls for common governance frameworks to ensure data sharing is ethical, secure, and transparent.

Australia's Newborn Bloodspot Screening programme provides a domestic precedent for this kind of nationally guided, locally delivered model – recognising inconsistencies still exist which can provide learnings. The McKell Institute notes that the programme demonstrates how national coordination can promote alignment across jurisdictions while allowing for local implementation and flexibility, and identifies it as a precedent for how other areas of rare disease policy – such as genomics, access to therapies, or data sharing – might be tackled through a similar approach.

Patient Engagement and Person-Centred Data

The WEF report's second strategy for improving rare disease data systems is to strengthen patient engagement in data collection. It finds that efforts to improve the quality and comprehensiveness of rare disease data frequently overlook the individuals, families, and communities directly affected, who bring unique insights including natural history data, social and environmental determinants of health, quality-of-life measures, and day-to-day experiences with care and treatment.

The report emphasises that registries and data platforms must be participatory and equitable to build trust and fairness, and that involving patients as co-creators in governance and implementation enhances legitimacy, reduces duplication, and increases the likelihood that the data generated will be used in decision-making. This aligns with the McKell Institute's finding that rare diseases are far too complex in their individual impacts for health departments or clinicians alone to determine what is needed, and that policy must be shaped by those directly affected.

An Australian national rare disease Registry, designed with substantive patient involvement from the outset, would operationalise this principle. It would capture patient-reported outcomes and experiences alongside clinical data, ensuring that the Registry's governance reflects the communities it serves, including Aboriginal and Torres Strait Islander peoples, regional and remote populations, and other priority groups.

Policy Position

1. Medicines Australia supports the establishment of a national, coordinated, and systematically governed rare disease Registry, underpinned by improved rare disease ICD coding, common data standards, and linkages across health and disability datasets.
2. The development of a Registry should be overseen by the federal government, such as the Australian Digital Health Agency, amongst others.
3. The Registry should be designed to enable clinical care coordination, clinical research, generation of real-world evidence (RWE) to support HTA evaluation, and system planning; incorporating patient-reported outcomes and experiences; and ensuring culturally safe approaches for priority populations.
4. Governance should be collaborative, with multi-stakeholder oversight (patients, clinicians, researchers, government, and industry), transparent data stewardship, privacy-preserving linkage, and alignment to international common data models to enable global collaboration for ultra-rare cohorts.

5. Implementation should prioritise interoperability with hospital electronic health records (EHRs), pathology and genomics systems, NDIS, newborn screening programmes, and existing disease-specific registries; adopt standard terminologies (e.g., Orphanet/ORPHAcodes mapped to ICD); and invest in workforce capability for coding and data quality.
6. Real-world data generated could be used in HTA (consistent with HTA Review recommendations 27–31), managed access programmes, and post-market evaluations to improve timeliness and confidence in funding decisions for rare and precision therapies.

Benefits

Benefits for Government and Health System Stewards

A Registry would provide the evidence base for responsible, equitable policymaking and resource allocation. It would:

- **Clarify prevalence and unmet need** across geographic and demographic groups, including Aboriginal and Torres Strait Islander peoples and regional and remote communities, enabling targeted investment rather than ad hoc responses.
- **Strengthen HTA** by generating Australian real-world evidence on clinical effectiveness, safety, and utilisation to support managed access arrangements and post-market reviews.
- **Inform national service and workforce planning** by identifying diagnostic bottlenecks, outcome variation, and gaps in integrated care across jurisdictions.
- **Reduce waste by revealing the true cost of rare disease** to the health system – currently invisible due to the absence of rare disease coding – and identifying where intervention delivers savings.
- **Enable international data collaboration** for ultra-rare conditions through alignment with common data models, supporting Australia's attractiveness as a market for innovation.
- **Improve accountability** through public reporting on outcomes, access, and system performance.

Benefits for People Living with a Rare Disease, Families, and Carers

For those directly affected, a Registry would reduce uncertainty and improve care experiences. It would:

- **Shorten the 'diagnostic odyssey'** through better data linkage, and clinician feedback loops. Diagnostic delay across rare conditions has been associated with avoidable costs of up to US$517,000 per patient (WEF, 2026).
- **Enable person-centred care planning** by making care pathways, outcomes, and service options visible, and embedding patient-reported outcomes alongside clinical data.
- **Improve equitable access** by identifying inequity to accessing genomic testing, specialist care, and mental health support – particularly for populations dispersed across large geographic areas.
- **Expand clinical trial participation** through Registry-enabled patient matching, critical given that 95% of rare diseases have no approved treatment (McKell Institute, 2025).
- **Strengthen patient advocacy** with credible, aggregated evidence to support policy and funding submissions.

Benefits for Clinicians and Care Teams

A Registry would equip clinicians with the tools and evidence to deliver safer, more consistent care. It would:

- **Support clinical decision-making** through linked data on diagnostics, treatments, and outcomes, reducing practice variation.

- **Improve coding accuracy**, reflecting true clinical complexity and enabling appropriate case-mix funding.
- **Drive quality improvement** through outcome feedback loops and multidisciplinary care model evaluation.
- **Streamline research participation** for small cohorts through integrated data capture and standardised terminologies.
- **Build workforce capability** in genomics and rare disease recognition, in line with the Action Plan's workforce strategy priority.

Benefits for Industry and Research Partners

A Registry is foundational to timely, evidence-based market access and sustained research investment. It would:

- **Generate fit-for-purpose real-world evidence** to complement clinical trial data in PBAC and MSAC submissions, i.e. second order benefits and patient reported outcomes, particularly for ultra-rare therapies where traditional trial datasets are insufficient.
- **Enable horizon scanning** through earlier visibility of patient populations and unmet clinical needs.
- **Create a predictable evidence pathway** for rare indications, reducing regulatory and access uncertainty and strengthening Australia's position in global launch sequencing.
- **Support repurposing and combination assessments** through linked outcome data and common data models.
- **Facilitate international data collaboration** through alignment with Orphanet/ORPHAcodes and global registry standards.

Implementation Considerations and Recommendations

National Rare Disease Minimum Dataset

To ensure feasibility, consistency and early value, the Registry could adopt a tiered data model that distinguishes core national data from disease-specific and therapy-specific modules.

Tier 1 (Core National Dataset) could be mandatory across all participating registries and datasets and could include at a minimum: patient demographics, Aboriginal and Torres Strait Islander status, confirmed diagnosis using ORPHAcodes mapped to ICD-10-AM/ICD-11, genomic testing status, key diagnostic timepoints, and consent flags for longitudinal follow-up, data linkage and re-contact.

Tier 2 (Disease Group Modules) could capture condition-specific clinical detail and could align with existing mature registries wherever possible to avoid duplication. Tier 3 (Therapy-specific Modules) could support post-market monitoring, managed access arrangements and HTA requirements for rare and precision therapies.

Existing disease registries could be mapped to the Tier 1 dataset as a first implementation step, enabling immediate participation without re-collection of data already captured.

Coding and Standards

Adopt and mandate rare disease identifiers (e.g., ORPHAcodes mapped to ICD-10-AM/ICD-11), with national guidance, training, and auditing to improve hospital and NDIS coding accuracy. The WEF report identifies ORPHAcodes and ICD-11 rare disease extension codes as the key international standards that make it possible to identify individual conditions in health records with greater precision. Western Australia's Register of Developmental Anomalies demonstrates that implementation is achievable within Australian systems.

Data Architecture

The Commonwealth should partner with the states and territories to design and implement a workforce strategy specific to rare disease.[3]

Build a federated, privacy-preserving data linkage framework, integrating hospital, primary care, pathology and genomics, NDIS, newborn screening, and disease-specific registries; ensure interoperability using common data models and Fast Healthcare Interoperability Resources (FHIR)-based Application Programming Interface (API)s. The WEF report recommends federated data systems as the preferred model, in which datasets stay where they are generated but insights travel freely, giving patients and institutions confidence in how their data is used. The WEF observes that achieving this vision requires both technical and institutional alignment, including common governance frameworks that ensure data sharing is ethical, secure, and transparent.

Governance and Ethics

Establish multi-stakeholder governance with strong patient representation, transparent data stewardship arrangements, and nationally consistent consent models suitable for longitudinal research, routine data linkage and care. Governance arrangements should include a National Data Access Committee with defined membership, decision-making criteria and service-level timeframes for data access decisions.

Consent frameworks should explicitly support longitudinal participation, re-use of data for research, health system planning, HTA evaluation and post-market monitoring, and routine linkage to administrative datasets. Clear, predictable data access pathways are essential to ensure the Registry delivers public benefit and is actively used by clinicians, policymakers, researchers and industry.

Quality and Utility

Invest in data quality processes, patient-reported outcome and experience measures, and where possible public reporting dashboards; embed learning health system cycles to translate insights into practice and policy.

Funding and Sustainability

Provide dedicated, multi-year funding for build and operations, aligned with HTA review timelines; build on existing national infrastructure (e.g., Australian Institute of Health and Welfare, state data assets) and international partnerships; ensure independent evaluation and published KPIs.

The McKell Institute's 2025 report notes that it is common for rare disease activities to be funded in short-term cycles, which risks programmes ending when funding expires. Sustained, multi-year funding with clear governance is essential. Predefined quality standards, sensitivity analyses, and agreed methodologies are required to ensure that real-world evidence generated is robust and credible. Access arrangements must be explicit and timely; a government-run registry offers limited value if industry access is restricted or delayed.

Measuring Impact and Accountability

The success of the National Rare Disease Registry could be assessed against clear, publicly reported measures that reflect meaningful impact for people living with rare disease. These could include coverage of diagnosed populations, completeness and timeliness of core data, reductions in diagnostic delay for selected conditions, use of Registry-derived evidence in HTA and funding decisions, and equity indicators disaggregated by geography, socio-economic status and Aboriginal and Torres Strait Islander status.

[3] McKell Institute, A Rare Kind of Care. (2025, August). 'Recommendation 8', page 14

Short-term milestones (within two years) could focus on implementation of the national minimum dataset, integration of established registries and publication of the first national rare disease data report. Longer-term measures should assess impact on access, outcomes and system efficiency.

Conclusion

Rare disease continues to pose significant challenges to patients and the families and carers who support them, the healthcare professionals that treat them as well as the scientific community who investigate their conditions. We all play an integral part to ensure the needs of rare disease populations are specifically addressed, just as for any vulnerable and underserved component of our community.

A national rare disease Registry is essential to transform fragmented data into actionable insight that improves diagnosis, care, and access for every Australian living with a rare condition.

While there are long-term reforms required to realise the objectives of the National Strategic Action Plan on Rare Disease, there are also shorter-term actions that state, territory and Australian governments can pursue.[4]

The McKell Institute's 2025 report confirms that no jurisdiction has achieved full implementation of Priority 3.1 of the National Strategic Action Plan, and that the resulting inequities – delayed diagnoses, missed treatment opportunities, and care determined by postcode – fall squarely on patients. The WEF's 2026 report reinforces that the case for investment in rare disease data systems is economic and scientific as well as moral, with benefits accruing to patients, health systems, governments, and industry alike.

A well-designed Registry – adopting ORPHAcodes mapped to ICD, implementing a national minimum dataset, and enabling privacy-preserving linkage across clinical, genomic, disability, and social care systems – makes small populations visible, strengthens HTA with real-world evidence, and creates the data infrastructure on which further reform depends.

This is a practical commitment to leave no patient behind: earlier diagnosis, coordinated care, and equitable access to effective treatments, regardless of where a person lives or how rare their condition may be.

[4] McKell Institute, A Rare Kind of Care. (2025, August). 'Finding 8', page 11

Sources

1. National Strategic Action Plan for Rare Diseases (2020). Australian Government Department of Health, Disability and Ageing. Available at: www.health.gov.au
2. The New Frontier report (2022). House of Representatives Standing Committee on Health, Aged Care and Sport. Commonwealth of Australia. Available at: https://www.aph.gov.au/Parliamentary_Business/Committees/House/Former_Committees/Health_Aged_Care_and_Sport/Newdrugs/Report
3. Full Recommendations of the Health Technology Assessment Policy and Methods Review (2024). Australian Government Department of Health, Disability and Ageing. Available at: https://www.health.gov.au/our-work/hta-review
4. Rare Voices Australia & Monash University. Recommendations for a National Approach to Rare Disease Data. Available at: https://rarevoices.org.au/wp-content/uploads/2023/08/RecommendationsforaNationalApproach_RareDiseaseData_August2023.pdf
5. McKell Institute (2025). A Rare Kind of Care: An Agenda to Deliver Health Equity for Australians Living with a Rare Disease. Available at: https://mckellinstitute.org.au/wp-content/uploads/2025/09/McKell-Institute-2025-A-Rare-Kind-of-Care-copy.pdf
6. World Economic Forum (2026). Making Rare Diseases Count: How Better Data Can Unlock a Multitrillion-Dollar Opportunity. Available at: https://www.weforum.org/publications/making-rare-diseases-count-how-better-data-can-unlock-a-multitrillion-dollar-opportunity/
7. Rare Voices Australia (2023). Implementation Status Report: Implementing the National Strategic Action Plan for Rare Diseases. Available at: https://rarevoices.org.au/wp-content/uploads/2023/05/Implementing-the-National-Strategic-Action-Plan-for-Rare-Diseases_May-2023-Status-Report.pdf