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A Rare Kind of Care

AN AGENDA TO DELIVER HEALTH EQUITY FOR AUSTRALIANS LIVING WITH A RARE DISEASE

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About this report

This report has been prepared by the McKell Institute with support from Alexion, AstraZeneca's Rare Disease group.

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Acknowledgement of country

This report was written on the lands of the Darug and the Eora Nations. The McKell Institute acknowledges Aboriginal and Torres Strait Islander peoples as the Traditional Owners of Country throughout Australia and their continuing connection to both their land and seas.

The opinions in this paper are those of the authors and do not necessarily represent the views of the Expert Advisory Group that informed this research. Any remaining errors are the sole responsibility of the authors and the McKell Institute.

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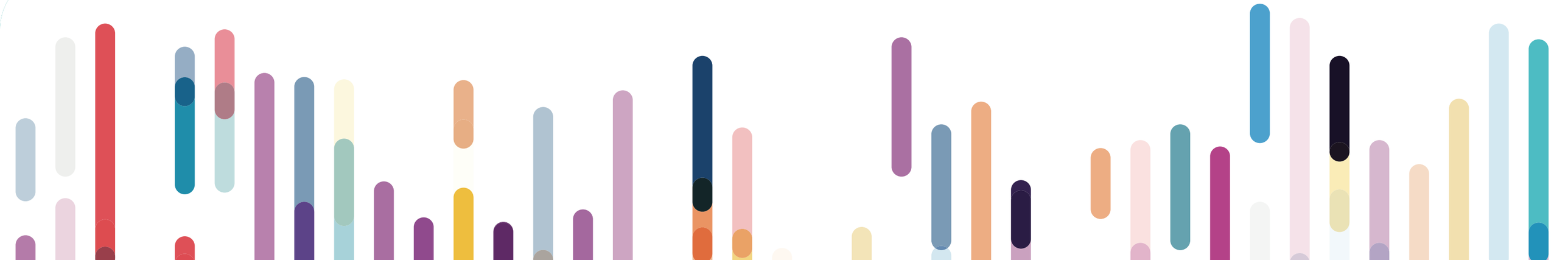


Executive Summary

An estimated two million Australians live with one or more of the over 7000 known rare diseases. Despite affecting so many in our community, people living with a rare disease are too often left behind by our health system. While efforts to provide more support for people living with rare disease (PLWRD) are underway, support remains fragmented, poorly coordinated, and inconsistent in its treatment of rare disease across Australia.

Rare disease patients and their families and carers face multiple inequities in relation to diagnosis, care, support and access to treatment. This report explores Australia's rare disease policy framework, shining a spotlight on the approach of state and territory governments, to identify gaps and propose new actions by all levels of government to improve health equity for people living with a rare disease so that no patient is left behind.

Part 1 briefly outlines the nature of rare disease in Australia, before describing the Australian Government's approach to rare disease policy. It notes that up to one-in-twelve Australians will experience a rare disease, including an estimated 350,000 children[1], and those Australians living with rare diseases interface with an array of disjointed health and support services.

Part 1 also notes that the Australian Government has established a national policy framework for rare disease in response to concerted advocacy campaigns from the rare disease sector. While this increased focus is welcome, the Australian Government's role in primary care is limited. There is further effort needed at the national level to coordinate and support responses within state and territory health systems.

Part 2 examines the policy approaches of state and territory governments for rare diseases. It maps the activities occurring within each jurisdiction against the three pillars of the Australian Government's National Strategic Action Plan for Rare Diseases[2]: *Awareness and Education, Care and Support, and Research and Data*. It also explores the 12 priorities that sit underneath these pillars and their alignment with state and territory policy settings. While it is beyond the scope of this report to evaluate the efficacy of each of the activities identified, this exercise provides a comprehensive mapping of Australian state and territory responses to rare disease, allowing for a comparative analysis of each state's approaches, and therefore, an identification of the gaps that exist across each jurisdiction.

Part 3 explores the gaps this analysis has identified. It highlights consistent and common gaps across the nation's policy landscape, noting a lack of coordination, workforce development, access to specialised care and the fragility of funding support for certain programs, and other significant policy gaps that are impeding Australian governments' delivery of health equity for people living with a rare disease. It notes that, while smaller jurisdictions sometimes have the weakest health ecosystems for rare disease patients, improvements can be made quickly with dedicated investment and prioritisation.

This analysis notes that much of the focus of state and territory governments has been on care and support. While this is critical, there has been a less consistent policy approach to both awareness and education, and research and data, in all jurisdictions.

This report then concludes with actionable short- and long-term recommendations, for both state and territory governments, and the Australian Government.

RECOMMENDATIONS FOR STATES AND TERRITORIES

For states and territories, this report recommends that each government develop and resource a dedicated Rare Disease Strategy and Action Plan for their jurisdiction, which align with the National Strategic Action Plan for Rare Diseases (NSAPRD).

These strategies must be developed through transparent and inclusive collaboration processes that incorporates patient perspectives, experience and aspiration for their health care. Engagement with people living with rare diseases (PLWRD) and their families and carers, patient organisations, clinicians and industry must be central to those strategies. Rare diseases are far too complex in their individual impacts for health departments or clinicians to alone determine what is needed. Policy must be shaped by those directly affected as empowering the patient voice will ensure outcomes that meet patient needs.

RECOMMENDATIONS FOR THE AUSTRALIAN GOVERNMENT

At the national level, this report recommends establishing an Office of Rare Disease, established under a Rare Disease Act. As a statutory agency, the Office of Rare Disease would coordinate services across portfolios and social services. It would guide rare disease policy development in collaboration with patients, oversee national implementation efforts including of the National Strategic Actional Plan for Rare Disease, support funding of innovative treatments and models of care and care coordination, and lead workforce development across jurisdictions. Its work can complement the newly created Genomics Australia.

In recent years, there has been some progress in the policy settings aimed to address gaps in treatment and care for Australians living with rare disease. However, it remains the case that Australia's rare disease policy architecture is patchy, inconsistent between jurisdictions, and suffering from a lack of coordination across health systems. This has resulted in poorer outcomes for people living with a rare disease.

A Note on the **Scope of this Report**

This report provides an overview of the Australian rare disease policy response, with a focus on states and territories, and to identify gaps and opportunities to improve the health ecosystem for people living with a rare disease. It follows the pillar and priority framework outlined in the National Strategic Action Plan for Rare Diseases. It begins by defining rare disease in the Australian context.

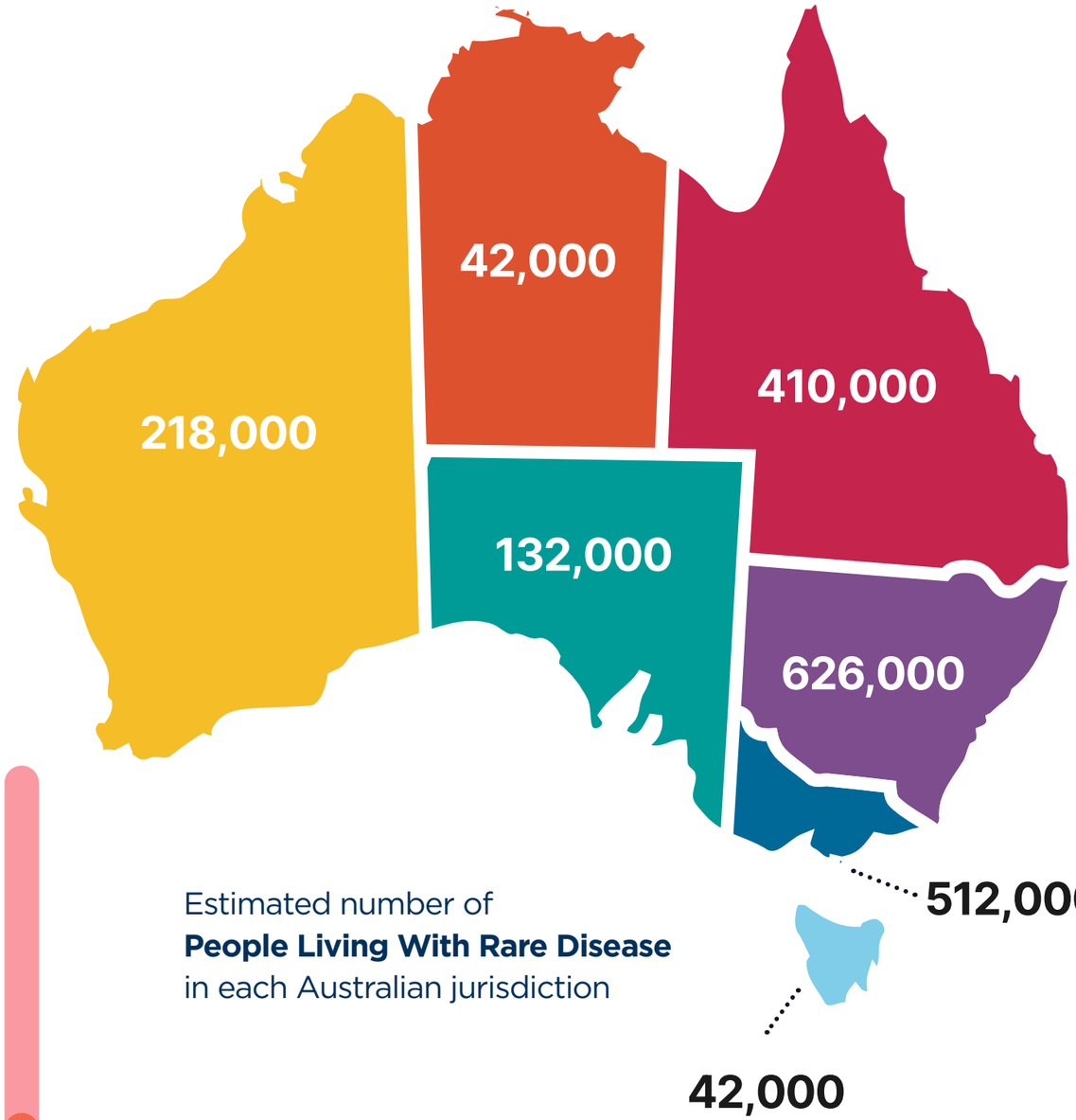
The report also identifies and spotlights a number of programs and policies responding to rare disease, including national, state and territory initiatives spanning awareness and education, care and support, and research and data. It complements existing resources, including the Rare Awareness Rare Education (RARE) Portal, which is being collaboratively developed by Rare Voices Australia alongside the rare disease sector.[3] While highlighting many of these programs, this report does not evaluate the efficacy of these programs.

This mapping exercise has been undertaken through a combination of in-depth interviews with practitioners and local experts in each jurisdiction who have visibility over existing programs, alongside extensive desktop research using publicly available materials. It builds on existing efforts to track rare disease policy activity and implementation, including the 2023 Rare Voices Australia (RVA) Implementation Status Report.

The key contribution of this project is the provision of centralised, jurisdiction-by-jurisdiction visibility of rare disease programs and policies, consolidated for the first time in the Appendix, and assessed against a progress framework advanced in Part 2 of this report.

The report has also been assisted by an expert advisory group and we are grateful for their input.

The findings, analysis and recommendations of this report are, however, the responsibility of the McKell Institute.



Estimated number of
People Living With Rare Disease
in each Australian jurisdiction

Key Findings

FINDING 1

While each disease may be 'rare', collectively, rare diseases affect up to 8 per cent of the Australian population, with this proportion multiplied when the impacts on family, friends and carers are taken into consideration.

FINDING 2

There is growing evidence of the disproportionate fiscal impact of rare disease, providing the opportunity for policymakers to consider innovative, cross-sector policy responses to effectively and efficiently meet the needs of Australians living with rare disease.

FINDING 3

Australians living with rare disease are subject to a "postcode lottery" due to patchy and inconsistent policy settings across each jurisdiction. These policy settings often result in a long journey to diagnosis, delayed or poor access to treatment, including specialist care and missed opportunities to receive early and innovative medicines, including through clinical trials.

FINDING 4

The publication of the National Strategic Action Plan for Rare Diseases, and the initiatives it champions, provide a foundation for more effective policy responses by all levels of government. Despite the efforts of patient groups and clinicians, implementation of the Action Plan has been patchy at the national level and has not been supported through the development of comprehensive state and territory rare disease policies and action plans.

FINDING 5

All aspects of rare disease policy, across health and social systems, need greater focus and resourcing from the national and state and territory governments to ensure health equity for Australians living with a rare disease.

FINDING 6

Successful programs exist, despite funding uncertainty and inconsistent government support and planning. Such programs should become the 'norm' rather than the exception so Australians living with a rare disease have access to world class support no matter where they live in Australia. There is an opportunity to learn from successful programs and models and replicating them across jurisdictions.

FINDING 7

The Australian government has a leadership role in coordinating and funding a national, best-practice response to rare diseases to address the significant inequities currently faced by Australians living with rare disease. The Australian government has a crucial role in improving affordable and timely access to innovative medicines and treatments for rare diseases through the Pharmaceutical Benefits Scheme (PBS) and other programs.

FINDING 8

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.

Recommendations in Detail

RECOMMENDATION 1

The Commonwealth should develop and legislate a Rare Disease Act, which legislates the establishment of an Office of Rare Disease (ORD). The ORD would be responsible for overseeing the implementation of the Action Plan, and coordinate reforms to rare disease policy and the delivery of health services for people living with a rare disease in Australia.

Chief amongst this report's recommendations is the passage of a Commonwealth Rare Disease Act, which would legislate the establishment of an Office of Rare Disease, or ORD. The ORD would be a central agency charged with overseeing Australian jurisdiction's response to the rare disease challenge.

A Rare Disease Act may also formalise different jurisdictions' role in responding to rare disease, and the process of its legislation may consider formally addressing the numerous policy gaps identified in this report. While an Act may not need to be a precondition for the establishment of an ORD, and an ORD may be established prior to the passing of an Act, the Act would help enshrine the ORD in Australia's health policy architecture.

RECOMMENDATION 2

Implementing the National Strategic Action Plan for Rare Disease, with a Priority on Forming the Implementation Group Required for its Implementation.

The Australian government should recommit to the full implementation of the National Strategic Action Plan and prioritise the forming of an implementation group to oversee the implementation of the plan. The group should bring together the federal health department, other agencies like the NDIS and Genomics Australia, representatives of the states and territories, patient organisations, clinicians and industry, as well as key data sharing agencies, and entities involved with the overseeing of rare disease registries.

RECOMMENDATION 3

Ensuring access to innovative medicines by reforming the PBS.

The Australian government plays a critical role in ensuring people living with a rare disease have access to innovative treatments through the PBS and the Life Saving Drugs Program. The Australian government must prioritise implementation of the recommendations of its HTA review, which examined the processes used for the listing of medicines on the PBS, and ensure faster and fairer access to rare disease therapies are delivered through those reforms. The Australian government should also expand the Life Saving Drugs Program for ultrarare disease by including medicines that help prevent significant disability and carer burdens alongside those that are lifesaving.

RECOMMENDATION 4

State and territory governments should develop rare disease strategies for their jurisdictions, aligned to the National Strategic Action Plan.

The consistent theme of this report is the fragmented and inadequate nature of health system support for people living with a rare disease. This reflects the absence of overarching prioritisation and planning within each state and territory for the provision of health care for rare disease patients.

As a first and urgent step, each state and territory should develop its own rare disease strategy in partnership with patients, clinicians and other stakeholders. Those strategies should align with the national Action Plan and include a detailed analysis of gaps in the diagnosis and care pathways. The strategies should include clear KPIs and processes for their implementation.

RECOMMENDATION 5

'Centres of Expertise' for rare disease should be expanded around the country, supported with Commonwealth funding.

This report highlights the efficacy of the Rare Care Centre in Western Australia, and its unique impact on West Australian children living with rare disease and other models around the country for individual diseases or segments of the population. They all point the vital need for increased capacity within state health systems for rare disease hubs or centres of expertise that can serve a cross-section of rare disease types to ensure patients have access to specialist care. Each state and territory will have different needs and capacities and both domestic and global exemplar models can provide guidance for the funding and development of such centres.

While the operation of these centres will fall primarily within the responsibilities of state and territory governments, the Australian Government should establish a competitive start-up funding pool to encourage their development and support centres with cross-jurisdictional reach (for example, serving indigenous communities or northern Australia).

Recommendations in Detail

RECOMMENDATION 6

Strengthening Australia's Diagnostics and Screening Framework.

A common experience for rare disease patients is an extended delay in diagnosis, often taking many years. Incorrect or delayed diagnosis can hinder treatment and exacerbate the impact of a disease – particularly in childhood. The creation of Centres of Expertise will enhance diagnostic processes to ensure patients receive the right treatment early in their disease progression.

There are also steps all governments can take to support the expansion and use of early genetic testing to improve diagnosis outcomes including:

- Work to prepare Australia for the use of whole genome sequencing as a part of the newborn screening programs;
- The use of genetic testing in emergency settings for undiagnosed acute medical issues;
- The inclusion of genetic counsellors on the MBS schedule; and
- Greater awareness and education of rare diseases among general practitioners and specialists to allow earlier identification and diagnosis of rare disease.

RECOMMENDATION 7

Expediting the establishment of the National One Stop Shop for clinical trials.

Clinical trials play a vital role in the development of new therapies for rare diseases and provide Australian patients with early access to innovative medicines and technologies.

Australia is a well-regarded environment for clinical trials due to its advanced health care system, research capacity and demographic diversity.

However, those advantages have been hindered by the absence of national and simplified processes for gaining approvals for clinical trial projects.

This is particularly profound for the development of rare disease therapies as a clinical trial for a rare disease might, intrinsically, involve a small patient pool spread across the nation at multiple locations. Given the absence of drug treatments for 95% of rare disease, access to clinical trials is typically the first opportunity for a potential life-transforming or life-saving drug therapy for many patients.

Rare disease trials can involve single patients in different locations and hospitals, exacerbating the impact of the requirement for different approvals at individual hospital and research institutions.

To address these problems, the Australian government and the states and territories have agreed to establish a National One Stop Shop for clinical trials. Funding has been allocated by the Australian Government and the implementation of this program must now be a priority for all jurisdictions. The unique challenges of rare disease trials should be reflected in the design of the NOSS.

RECOMMENDATION 8

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

This report has identified a lack of coherent workforce strategy in the rare disease space. Rare disease care is highly specialised and multi-disciplinary, and the disparate nature of care across the country creates significant gaps in care for many Australians based upon their geographic location. Workforce gaps exist across the health sector.

As governments develop and implement workforce strategies for health, they must ensure that those unique needs of rare disease patients are reflected in their planning.

Part One: Understanding Rare Disease in an Australian and Global context

Key points

1. While each disease may be rare, collectively, it is estimated that up to **8 per cent of Australians live with a rare disease** of some kind.
2. More than **7000 rare diseases** have already been identified and, as our knowledge of genetics expands, estimates suggest that the number of known rare diseases may increase to 10,000.
3. Australia is yet to have a **Rare Disease Act**, or **Office of Rare Disease**, and there are significant gaps in national leadership and coordination of rare disease policy and health services.

The Prevalence and Classification of Rare Diseases

Rare diseases collectively impact a significant portion of the global population, despite their individual rarity. "Whilst historically, the definition of a rare disease has varied, in 2024 a global panel of rare disease experts formally defined a rare disease as "a medical condition with a specific pattern of clinical signs, symptoms, and findings that affects fewer than or equal to 1 in 2000 persons living in any World Health Organization-defined region of the world."[4]

There is broad consensus that there are at least 7000 unique rare diseases,[5] with more recent estimates suggesting the number could exceed 10,000.[6] The number of known rare diseases continues to increase as disease discovery is enabled by emerging technologies and through genetic identification.[7]

It is estimated that "patients with rare disease" may comprise as much as 8 per cent of the population[8], with proportions far exceeding this when impacted family members and carers are taken into consideration.[9,10]

Rare Diseases in the Australian Context

In the Australian context, with a current population of approximately 27.5 million, the number of people living with rare disease — approximately 2 million — is similar to the combined population of South Australia and the Australian Capital Territory.[11]

This proportion is comparable to the prevalence of common chronic conditions such as diabetes or asthma,[12] underscoring the substantial impact of rare diseases on healthcare systems, patients, and their families.

Given the complexity and rarity of these conditions, affected individuals often experience challenges related to diagnosis, treatment, and access to specialised care.

The global imperative to address the inequities faced by people living with rare disease is growing

As documented by Wang et.al. in late 2024, rare disease is emerging as a global policy priority.[13] In late 2021, the United Nations adopted the first-ever UN Resolution on "Addressing the Challenges of Persons Living with a Rare Disease and their Families."[14] The Resolution affirms that addressing the needs of persons living with a rare disease is essential to advancing the 2030 Agenda for Sustainable Development.[15] Global advocacy for a World Health Assembly (WHA) Resolution on Rare Diseases saw a landmark resolution adopted by the WHA in May 2025 which provides a strong foundation and framework for international, regional and national action to support those living with a rare disease.

The Australian Government has supported these international resolutions and has an obligation to ensure they are translated into comprehensive implementation at the national level.

The Australian Government creates an enabling environment for the states and territories in relation to rare disease policy.

The Australian Government plays a crucial enabling role in shaping rare disease policy across the nation. While health service delivery largely rests with the states and territories, the Commonwealth—consistent with its responsibilities under the Constitution—is uniquely positioned to provide national leadership, promote coordination, and ensure consistency and equity across jurisdictions. Its role includes but extends beyond financing to include setting strategic direction, establishing national frameworks and standards, and fostering a policy environment where individual jurisdictions can align their efforts for greater collective impact. The Australian government also provides a crucial role through the provision of Medicare, the registration of treatments by the Therapeutic Goods Administration, supporting affordable medicines through the PBS and the Life Saving Drugs Program (LSDP) and treatments and diagnostic tools through the Medical Services Advsiory Committee (MSAC) process.

This is where a long-standing and fundamental gap in rare disease policy becomes evident. The absence of consistent national leadership and coordination has led to fragmented, duplicative, and often isolated initiatives for rare disease patients. This challenge is at the heart of theme one of the National Strategic Action Plan for Rare Diseases (the "Action Plan"): "The need for national leadership, coordination and consistency." In the face of growing unmet needs, a multitude of stakeholders—clinicians, researchers, community organisations, and governments—are initiating activities that, while well-intentioned, are often disconnected. This results in unnecessary duplication, missed opportunities for collaboration, and avoidable gaps in care and support. Fundamentally it means that appropriate access to care can depend on where a patient lives and on other characteristics such as age.

Rare Voices Australia's 2023 *Status Report: Implementing the National Strategic Action Plan for Rare Diseases* starkly highlights this issue. The report documented hundreds of rare disease-related activities occurring across the country that could be aligned with the Action Plan's pillars and priorities. However, despite efforts from non-government organisations such as Rare Voices Australia, the proliferation of fragmented and uncoordinated activities reflects an incomplete investment in theme one. Specifically, the absence of a nationally endorsed implementation plan, backed by funding and governance mechanisms, has meant that there is no clear pathway for ensuring progress, accountability, or sustainability.

The National Health Reform Agreement (NHRA) offers a potential mechanism to address this. The 2020–25 NHRA sets the terms of intergovernmental collaboration on public hospital funding and broader health system reform. It introduces six long-term reform streams and allows states and territories to trial new funding models and care pathways. Significantly, it also supports the delivery of new, high-cost therapies—an area critical to the rare disease community, where patients often have few treatment options available.

The NHRA also reaffirms governments' shared commitment to Medicare principles, ensuring that public hospital services are delivered equitably, based on clinical need and not geography. For people living with rare diseases, this promise is particularly meaningful—but so far, rare diseases have received no mention in the NHRA. There is now a clear opportunity to embed rare disease more explicitly in future intergovernmental agreements, using the NHRA as a vehicle to drive national consistency and ensure that jurisdictions are equipped and incentivised to respond within a coherent policy framework. As a minimum, the NHRA should require states and territories to develop rare disease strategies and action plans that align with the national action plan.

There is a broad Commonwealth policy architecture for rare disease

Although Australia does not have dedicated Office of Rare Disease or Rare Disease Act, there are numerous programs that are relevant to Australians living with rare disease, that can be deployed to specifically name, target and support implementation against unmet rare disease need.

These include:

POLICY	RELEVANCE TO RARE DISEASE
National Strategic Framework for Chronic Conditions	Overarching policy document for chronic conditions. Aims to help Australians live healthier lives through prevention and management.[16]
Health Technology Assessment (HTA) Policy and Methods Review	Evaluates Australia's HTA approach to ensure access to safe, effective, and affordable health technologies in a timely manner through the PBS.
2020–25 National Health Reform Agreement (NHRA)	Signed by all Australian governments, this agreement aims to improve health outcomes and maintain a sustainable health system, but does not adequately focus on rare disease
National Health Reform Agreement (2011)	Agreement between Commonwealth and state governments to enhance health outcomes and system sustainability.

The National Strategic Action Plan for Rare Diseases is the most significant of these policies

The National Strategic Action Plan for Rare Diseases,[17] launched with bipartisan support by the Australian Government in 2020, is central to this landscape, and is the only contemporary rare disease strategy or policy document in Australia.

The Action Plan is one of 11 developed under the National Strategic Framework for Chronic Conditions (NSFCC),[18] which was endorsed in 2017 by the Council of Australian Government (COAG) Health Council with an overarching vision that "all Australians live healthier lives through effective prevention and management of chronic conditions."[19]

Commonwealth support for Australians living with rare disease remains characterised by theme one of the Action Plan: "the need for national leadership, coordination and consistency".[20] Under this theme, participants in the Action Plan consultation process consistently called for three components (see p.44):

- A national plan for Australia that is in line with global standards
- An annual implementation plan
- Ongoing stewardship and policy sustainability.[21]

While the Action Plan satisfies the first of these components, the latter two remain elusive, despite the best efforts of non-government bodies such as Rare Voices Australia and Australian Genomics.

Australians living with rare disease often rely on untargeted support

Many Australians living with rare disease have access to Commonwealth support, but often it part of the broader social welfare framework rather than being targeted to their specific needs. Many Australians living with rare disease require highly personalised care and support, which some Commonwealth programs fail to accommodate.

The National Disability Insurance Scheme (NDIS) also supports some Australians living with rare disease, noting that access to the NDIS for Australians living with a rare disease is often dependent on having a diagnosis, which can take many years[22] and, even with a diagnosis, is only guaranteed for a small percentage of the up to 10,000 rare diseases.[23] Pathways for the undiagnosed are required[24].

As the McKell Institute has previously identified, Australians living with disability caused by rare diseases often fall in the gap between the Commonwealth-funded NDIS, and the state funded primary care systems. This gap can be confusing and challenging for Australians living with rare disease to navigate. Due to the novelty and complexity of many rare diseases, Australians experiencing these conditions often need highly individualised care and support, which the NDIS may not always be in the best position to accommodate.

Rare Voices Australia has submitted 18 disability-related submissions to the Commonwealth, or similar documents, since the Action Plan was published, indicating that this remains an area requiring significant attention for Australians living with rare disease.[25]

COMMONWEALTH PROGRAM	DESCRIPTION IN RELATION TO ITS SUPPORT OF AUSTRALIANS EXPERIENCING RARE DISEASE[26]
Medicare Benefits Schedule (MBS)	Supports patient care payments, including Chronic Disease Management plans. Changes effective from 1 July 2025.[27]
Life Saving Drugs Program	Funds essential medicines for patients with rare and life-threatening diseases.
Pharmaceutical Benefits Scheme (PBS)	Helps pay for medicines for rare diseases.
Therapeutic Goods Administration (TGA)	Has an orphan drug program that waives application fees for new medicines for rare diseases.
Medical Research FundingScheme (PBS)	The Medical Research Future Fund (MRFF) and the National Health and Medical Research Council (NHMRC) support research into rare diseases, including Clinical Trials Activity for rare cancers, rare diseases, and unmet needs.

The table below outlines key national policies and frameworks that shape healthcare and support systems—none of which are specifically designed for people living with rare diseases.

The Commonwealth has jurisdictional coverage of medicines policy

While the Commonwealth does not have sole control of the planning of primary care, it has jurisdiction for Australia's medicines policy and ensuring access to affordable treatments in a timely way. For Australians living with rare disease, therapeutics and medicines can be expensive and inaccessible without Commonwealth support.

Australia's medicines framework allows for TGA-approved medicines to be listed on the PBS or the LSDP. The pace of PNS listing of some medicines, however, is at times in conflict with the expectations of Australians living with rare disease. Many Australians are missing out on innovative treatments available in other countries because the PBS fails to recognise the often-unique challenges of listing rare disease medicines. There is a critical need for PBS reform to address these issues.

Part Two: Assessing State and Territory Approaches to Rare Disease

Key points

1. Given the primary role state and territories have in the provision of health care in Australia, their **approach to rare disease policy is critical**.

2. There is **little uniformity in the support** and approach of state and territory governments provided to people living with a rare disease.

3. While many jurisdictions have exceptional individual programs, there is a lack of consistency across the country and within individual jurisdictions, resulting in some **Australians receiving poorer care simply because of where they live**.

The role of the states and territories in rare disease

One of the key challenges facing people living with a rare disease is the lack of consistency in how care is approached across and within different states and territories, as well as the disparities between public and private healthcare systems. In some hospitals, patients may have access to innovative treatments that are not listed on the PBS, while similar options may not be available in other hospitals, cities, or states. Likewise, some states support rare disease centres of expertise (either for individual diseases or broader therapeutic areas) while in others access to such specialised care centres is unavailable. 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.

Australia's size, geography and federation exacerbate rare disease policy challenges

Challenges commonly associated with Australia's health system are exacerbated in the ctonext of care for rare disease patients. Small population numbers for individual rare diseases dispersed over large geographical areas impacts the ability of Australians living with rare disease to access specialist care and treatments; connect with others, including patient groups, for support; and participate in research projects, including clinical trials.

> An example shared through this report's consultation process was of a research group based in one jurisdiction that had received funding to investigate a rare disease that no one living in that jurisdiction had yet been diagnosed with.[28]

When an individual in another jurisdiction received a diagnosis for this rare disease,[29] the researchers asked the relevant genetics service if they could facilitate that individual's participation in this study. The efforts to make this a reality - to advance our understanding of this particular rare disease, and the care, including treatment, and support of the many rare diseases with which symptoms are shared - were unfunded and there was no mechanism for sharing patient data and specimens across borders.[30]

Queensland's population is dispersed over a large area and has the highest mainland percentage of its population living outside of its capital city, in contrast to all other Australian states and territories except Tasmania.[31,32]

Western Australia also faces challenges associated with remote service provision, and in several local government areas (LGAs), a significantly higher proportion of Aboriginal and Torres Strait Islander people (for example, between 37.6 per cent and 84.5 per cent of the population in Murchison and Ngaanyatjarraku, in comparison to 3.3 per cent of the state population.)[33]

All of Tasmania is considered regional or remote under the Australian Standard Geographic Classification. A recent report that investigated options for improving diagnostic pathways for rare disease in regional Australia highlighted the following barriers to accessing care and diagnosis in Tasmania: dispersed population, geographical inequity, transport disadvantage, lack of access to after-hours and emergency care, socio-economic and educational disadvantage, low education levels contributing to low health literacy, high barriers to accessing primary care, difficulties in recruitment and retention of healthcare professionals, shortages in specialists, limited ability to establish multidisciplinary teams, and a consumer lack of awareness.[34] These findings are applicable to other regional or remote locations across Australia.

These jurisdiction-specific dynamics give rise to what is commonly referred to as "postcode lottery" in rare diseases, where access to medical support of people living with rare disease is significantly influenced by where they live.

Rare disease policy at a state and territory level is patchy, but WA has been a leader

No state or territory in Australia has in place a current and dedicated state-wide rare disease strategy or action plan which provides an overarching framework for supporting people living with a rare disease and improving the broader health and research ecosystem for rare diseases.

Western Australia is the only jurisdiction to have previously published a targeted rare disease policy: the WA Rare Diseases Strategic Framework 2015-2018.[35] This policy was developed by the Office of Population Health Genomics (OPHG), which is situated within the Public Health Division of the WA Department of Health. It was established in 2001[36] "to help optimise the health benefits of genomic knowledge for the population of Western Australia".[37] The OPHG develops public policy that draws on stakeholder engagement and collaboration, and research outcomes, a methodology that the OPHG itself believes has made a significant difference to the effective implementation of those policies within the state.[38]

Western Australia's *Rare Care Centre* is also a leading example of best practice care for children living with rare disease.

STATE POLICY CASE STUDY

WA's Rare Care Centre and Pilbara Hub

The Rare Care Centre, established by the Child and Adolescent Health Service (CAHS) in 2022, serves as a Clinical Centre of Expertise for Rare and Undiagnosed Diseases in Western Australia.

This initiative addresses the complex challenges faced by children and families affected by rare and undiagnosed conditions, ensuring equitable access to specialised care and support services. It is entirely philanthropically funded through a network of philanthropy.* The prospective economic assessment shows the most conservatively assessed return on investment (ROI) is 4.75 to 1, poising it for sustainable funding through delivering systems-wide efficiencies, savings and increased productivity.

Clinical Services and Support Programs

The Rare Care Centre provides two primary streams of clinical service: a state-wide cross-sector care coordination service and a dedicated Nurse Navigator program.

> These services are designed to streamline patient care pathways, enhance coordination across multiple sectors, and improve the overall healthcare experience for affected families.

In addition, the Centre supports various complementary initiatives, including education and workforce capacity-building, digital data integration, device innovation, global partnerships, program evaluation and research, peer support networks, and advocacy efforts to raise awareness and enhance systemic support.

Expansion into Regional Western Australia – The Pilbara Hub

To extend its reach beyond metropolitan Perth, the Rare Care Centre has expanded into regional Western Australia with the establishment of the first of its remote regions Hubs (Pilbara Hub). This expansion is made possible through the existing philanthropic network and with the addition of new donors, working together with multiple Health System Providers, and first catalysed through the generous support of the McCusker Charitable Foundation via the Channel 7 Telethon Trust. The Pilbara Hub is based in the north-west of the state, ensuring improved access to specialised services for children and families in remote areas. By decentralising care and building remote region capacity, the Hub aims to address disparities in healthcare accessibility and enhance support for those living with rare and undiagnosed diseases in rural and remote communities. Economic modelling shows the remote region (Pilbara Hub) hub is anticipated to show a greater ROI than the metropolitan-based alone model.

Cross-Sector Care Coordination Approach

The Rare Care Centre's model of care is underpinned by a cross-sector care coordination framework, as highlighted in the European Journal of Medical Genetics publication, 'Rare Care – Cross Sector Care Coordination'. Co-authored by Clin/Prof Gareth Baynam of the RVA Scientific and Medical Advisory Committee, the paper outlines a holistic approach that integrates multiple service sectors, including healthcare, education, welfare, disability support, and social services. This model has been instrumental in improving the health and wellbeing of children with rare and undiagnosed diseases by fostering a more comprehensive and seamless support system. The Rare Care Centre (RCC) and its Pilbara Hub represent a significant advancement in the provision of care for children with rare and undiagnosed diseases in Western Australia, its model is being adopted and adapted internationally in the UK, EU, North America, Asia and Africa., Wales is now government funded to implement their Rare Care Centre, the Democratic Republic of Congo operates an RCC model, and China is partnering with the RCC to adapt the model for implementation in multiple hospitals, such as Peking Union Medical College Hospital, as parts of its governments national approach to rare disease healthcare. Through an integrated, cross-sector approach and regional outreach initiatives, the Centre is making substantial progress in addressing healthcare inequities and enhancing support systems for affected families. Continued investment in these programs will be critical to sustaining and expanding these essential services, ensuring that all children, regardless of location, receive the care they need.

> The development of innovative models, such as Western Australia's Rare Care Centre and the Pilbara Hub, highlight efforts to decentralise services and improve access for patients in remote and underserved areas.

However, without sustained funding and commitment, these initiatives risk being short-lived in Australia whilst sustained in other countries, and gaps in care will persist, particularly in smaller and regional areas.

* *Philanthropic Network: McCusker Charitable Foundation via the Channel 7 Telethon Trust, Angela Wright Bennett Foundation, Stan Perron Charitable Foundation, Perth Children's Hospital Foundation (PCHF), with its major Rare Care Centre funding partner, Mineral Resources Limited. On expansion of the Rare Care Centre to include the Pilbara Hub, addition to the network of HanRine Community Foundation, BHP, Rio Tinto, and other partners via the PCHF.*

Genomics policy is relevant to rare disease, but is, alone, is an insufficient policy response

Genomics is a powerful technology that is one component of a cohesive response to the breadth of unmet needs across the rare disease domain. The policy response to rare disease can benefit from genomic policy, but like other health domains that benefit from genomics, such as cancer, cardiovascular disease and infectious disease, cannot be adequately served by being subsumed under broader genomics strategies.

A comprehensive policy response requires dedicated rare disease policy and including rare disease as a priority within relevant policies, strategies and frameworks across the breadth of government sectors. With 80 per cent of rare diseases being genetic in origin, a policy area highly relevant to rare disease is the provision of genetic services and research. At the national level, the National Health Genomics Policy Framework 2018-2021, outlined Commonwealth objectives and actions in this area.[39] The framework was supported by an associated implementation plan, for the period 2018-2021.[40] The Australian government has established a new genomics agency, Genomics Australia, which health minister, the Hon. Mark Butler MP, has indicated "will work with states and territories to implement an updated National Health Genomics Policy Framework."[41]

Some jurisdictions have developed state policies which complement this policy framework, which are summarised in the table below. However, as the table demonstrates, implementation has not been consistent.

TABLE 2.1 GENOMICS POLICIES ACROSS AUSTRALIA

JURISDICTION	POLICY NAME	ADDITIONAL CONTEXT
New South Wales	NSW Health Genomics Strategy: A comprehensive strategy to integrate genomics into healthcare[42]	Supported by two subsequent Implementation Plans: NSW Health Genomics Strategy Implementation Plan 2018-20 NSW Health Genomics Strategy Implementation Plan (2021-2025)
Victoria	Genetic and genomic healthcare for Victoria 2021[43]	
Queensland	Queensland Health Genomics and Precision Medicine Strategic Roadmap 2021-2026[44]	May be supported by the State-wide Genetic Health Queensland Service Plan 2017-2022[45] Queensland Genomics was a Queensland Government initiative that ran from 2016 – 2021, and has not received ongoing funding[46]
Western Australia	WA Genomics Strategy 2022-2032: Towards Precision Medicine and Precision Public Health[47]	Supported by Strategic Framework for Rare Diseases 2015-18, OPHG Strategic Plan, as well as an internal Implementation Plan
South Australia	South Australian Clinical Genomics Plan 2022[48] (not available to the public)	
Tasmania	-	-
Australian Capital Territory	-	-
Northern Territory	-	-

STATE POLICY CASE STUDY

The Genomic Autopsy Study: Advancing Research in Perinatal Loss

The Genomic Autopsy Study was a national research initiative based in Adelaide, South Australia, aimed at understanding the genetic factors contributing to stillbirth and perinatal death (encompassing 13 weeks' gestation to 28 days post-delivery).[49]

In recognition of its potential impact, the study received **$3.4 million in funding** from the Australian Government's **Medical Research Future Fund (Genomics Health Futures Mission)** in July 2020, for the period to December 2023.[50]

> Findings from the study have demonstrated that genomic investigations can identify a cause of death in **over 50 per cent of cases**, particularly when congenital abnormalities are present, highlighting that congenital anomalies are a large class of mainly rare diseases, and the largest class of rare disease in children.

However, identifying causes for stillbirths without congenital anomalies remains a significant challenge, highlighting the need for further research and technological advancements.[51]

The study has gained international recognition, with a journal article[52] and research briefing published in **Nature Medicine**, emphasising the value of genomic autopsy in providing answers for families affected by pregnancy loss.

> This breakthrough approach is paving the way for new clinical guidelines and shaping future medical interventions.

Additionally, the research team, led by **Professor Christopher Barnett and Professor Hamish Scott**, was **highly commended by Research Australia** for their contributions to the field.iv

By embedding genomic autopsy services within the South Australian **Women's and Children's Health Network (WCHN)**[54] this initiative is ensuring that families in South Australia and beyond have access to cutting-edge genetic diagnostics, providing much-needed answers to affected families while informing medical advancements that inform family planning and could help prevent future losses.

Congenital anomalies data collection is being undertaken by jurisdictions, some under legislative requirement

Congenital anomalies data collections, sometimes still referred to as 'birth defects' data collections, represent organised, or system-based, rare disease activity. Congenital anomalies are a large class of mainly rare diseases.[55] Reflecting their life-long nature, in a whole of state data linkage study, they were the most frequent group of rare diseases across all ages. They are important to mention, in the context of rare disease policy, as, both in Western Australia and internationally, establishing these collections, and embedding rare disease data coding (Orphacodes or International Classification of Diseases 11th Revision (ICD-11)) have represented the foundation for a more strategic approach to rare disease.[56]

Each state and territory in Australia submits data to the Australian Institute of Health and Welfare (AIHW) to support the preparation of the report *Congenital Anomalies in Australia*.[57] The state and territory data sources are detailed on the AIHW website, and a high-level summary is provided below, along with the title of the supporting legislation, if any.[58]

TABLE 2.2 CONGENITAL ANOMALIES DATA COLLECTIONS AND SUPPORT LEGISLATION
(Excerpt from AIHW table "State and territory congenital anomaly data sources")

JURISDICTION	COLLECTION NAME	RELEVANT LEGISLATION
New South Wales	NSW Register of Congenital Conditions	NSW Public Health Act 2010
Victoria	Victorian Congenital Anomalies Register (VCAR)	Nil
Queensland	Congenital Anomaly Linked File (CALF)	Nil
Western Australia	Western Australian Register of Developmental Anomalies (WARDA)	WA Register of Developmental Anomalies Regulations 2010
South Australia	South Australian Birth Defects Register (SABDR)	South Australian Health Care Act 2008, and South Australian Health Care Regulations 2008
Tasmania	Tasmanian Perinatal Data Collection Tasmanian Admitted Patient Data Collection	Nil
Australian Capital Territory	ACT Perinatal Data Collection ACT Admitted Patient Data Collection ACT Perinatal Deaths Data	Nil
Northern Territory	Congenital Anomalies Register NT	NT Public and Environmental Health Act, and Public and Environmental Health Regulations 2014

Our approach to assessing and mapping rare disease policies across Australia

While this report highlights ongoing major gaps in Australia's rare disease policy landscape, it is also clear that support for people living with a rare disease has received attention from some parts of national and state health systems in recent years. These developments are, however, patchy and often the result of patient and clinician action rather than system-wide planning or needs assessment.

Accordingly, identifying the responses to Australia's rare disease challenge is a complicated exercise that requires a degree of methodological consideration. This report has engaged in both direct interviews with subject matter experts and local experts across each state and territory, and has undertaken a rigorous review of publicly available programs. It has also been guided by an expert advisory panel of clinicians and patient representatives.

Through this process, this report synthesises this array of policy or system-level responses (referred to here as "initiatives", for brevity) to rare disease; catalogues this response by category and by state; and can therefore identify consistencies as well as gaps across jurisdictions. While this report might omit individual highly localised programs and responses to rare disease, or those provided through patient support groups, it has worked to identify policy approaches by state and territory governments, or programs within their publicly funded health systems.

The three 'policy pillars' that structure rare disease policy responses

The National Strategic Action Plan for Rare Diseases (the Action Plan) is the most comprehensive strategic framework for setting and guiding rare disease policy in Australia. The Action Plan put forward a 'policy pillar' framework, which provides a structure for assessing and cataloguing existing and future policy and system-level responses relating to rare disease.[59] This policy review follows the three policy pillars as a means of establishing a methodology to identify gaps in Australia's response.

TABLE 2.3 THE THREE POLICY PILLARS UNDER THE NATIONAL STRATEGIC ACTION PLAN FOR RARE DISEASES.

PILLAR 1	**Awareness and Education**
PILLAR 2	**Care and Support**
PILLAR 3	**Research and Data**

Policy and system-level responses to rare disease are multifaceted. There are, of course, the direct responses to patient need: treating and caring for those experiencing rare disease. However, holistic responses to rare disease are more than direct treatment pathways and also include the overarching health framework. Awareness and education, as well as the collection of data and research efforts (including for clinical trials) are all critical policy responses to rare disease.

The three policy pillars have individual sub-priorities

As outlined in the Action Plan, each pillar includes a set of defined priorities. These priorities provide a framework for policy analysis, and allow policy reviewers to cross-check the extent to which each state and territory is pursuing the relevant objectives.

THE DETAILED POLICY PILLARS, AND THEIR CORRESPONDING PRIORITIES, ARE AS FOLLOWS:

PILLAR 1	AWARENESS AND EDUCATION
Priorities	**Priority 1.1:** Increase every Australian's awareness of rare diseases including, where applicable, relevant prevention measures.
	Priority 1.2: Ensure Australians living with a rare disease have access to information and education that enables them to be active participants in their rare disease journey.
	Priority 1.3: Develop a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics.

PILLAR 2	CARE AND SUPPORT
Priorities	**Priority 2.1:** Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family-centred.
	Priority 2.2: Ensure diagnosis of a rare disease is timely and accurate.
	Priority 2.3: Facilitate increased reproductive confidence.
	Priority 2.4: Enable all Australians to have equitable access to the best available health technology.
	Priority 2.5: Integrate mental health, and social and emotional wellbeing, into rare disease care and support.

PILLAR 3	RESEARCH AND DATA
Priorities	**Priority 3.1:** Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning.
	Priority 3.2: Develop a national research strategy for rare diseases to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps.
	Priority 3.3: Ensure research into rare diseases is collaborative and person-centred.
	Priority 3.4: Translate research and innovation into clinical care; clinical care informs research and innovation.

Mapping each jurisdiction's action towards each pillar priority area of the national Action Plan

This report has undertaken a comprehensive review of the rare disease and associated policies and system-level initiatives undertaken by state and territory governments. While no state or territory is a signatory to the National Action Plan, we have used the plan as the benchmark for assessing policy performance as it provides a framework accepted and developed by Australia's leading rare disease experts, advocates and patient representatives.

This analysis shows that the policy pillars advanced in the National Action Plan have not been achieved, either collectively or by any individual jurisdiction. It should be emphasised that the analysis does not offer a qualitative evaluation of all identified initiatives, or of progress towards the priorities of the Action Plan. However, as a starting point for reviewing the policy frameworks for the rare disease health ecosystem, it establishes the presence of policy initiatives relevant to the Action Plan.

While there is no identified hierarchy in terms of the delivery of Action Plan priorities, it is understandable, in a resource-constrained state health policy environment, that much of the investment states and territory governments have made in the rare disease space has been on care and support for patients, rather than Pillars 1 and 3. This also more closely aligns to state and territory responsibilities in the provision of health care within the federation.

The following table reflects this—state and territory activity is strongest in pillar 2 policies and activities. It is important to note that priorities 1.3 and 3.2 both refer to the development of national strategies, which are outside the direct scope of state and territory responsibility. It should also be noted that some initiatives address multiple priorities. In most cases, initiatives have been assigned to the dominant priority, with an exception being for the clinical genetics services, which contributes to both priority 2.2 (ensure diagnosis of a rare disease is timely and accurate) and priority 2.3 (facilitate increased reproductive confidence). Lastly, our analysis should be interpreted as inferring that a particular priority has been achieved in full – in many cases there will be opportunities to further improve the comprehensiveness and effectiveness of performance.

Our framework for assessing adherence towards the National Action Plan

This report has undertaken a broad mapping exercise of initiatives underway in each jurisdiction and applied these activities to the priority areas adopted as part of the National Strategic Action Plan on Rare Disease. Often, progress cannot be measured in a binary manner — routinely, there is evidence of some progress, even if it is isolated to one project or disease area. Accordingly, this report adopts a 5-point assessment framework, which allows each jurisdiction's performance against each Priority Area to be assessed. Qualifications and assessments are included in the framework, with a complete list of programs assessed available in the Appendix. Disclaimer: any omissions of programs underway are the error of the authors.

SYMBOL	STATUS	DETAIL
	Inadequate Actions Taken	While there may be occasional or individual projects that have aligned with the Priority, there is no demonstrable systematic approach to achieving the Priority area yet in place.
	Some Action Underway	There is evidence that there are steps established to achieve this Priority area.
	Notable Progress	There is evidence that the jurisdiction is making notable progress to some extent towards achieving the priority area.
	Close to meeting the priority area, with improvement still possible.	While there is significant activity evident, there may be room for further improvement
	Implemented Priority Area	The programs within the jurisdiction fully meet the Priority.

PILLAR 1	AWARENESS AND EDUCATION							
PRIORITIES	**ACT**	**NSW**	**NT**	**QLD**	**SA**	**TAS**	**VIC**	**WA**
1.1 – Increased Awareness	★	★	★	★	★	◆	➤	◆
Qualifications & Notes	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	RD focus in Tasmania's 20-year preventative health strategy.	Victoria's *The Science Within Us* program is a notable program focusing on genetics education.	WA Health has paid attention to RD, and include RD information in its ***Healthy WA*** information package.
ASSESSMENT	There has only been piecemeal and occasional efforts by governments to raise RD awareness. While civil society has led these efforts, state and Commonwealth focus on this area remains patchy and slow.							
1.2 – Ensure PLWRD have access to information and education to enable active participation in their disease journey	★	◆	★	★	➤	★	★	◆
Qualifications & Notes	No discernible qualifying activity.	The NSW Governments Centre for Genetics Education provides detailed information.	No discernible qualifying activity.	No discernible qualifying activity.	SA has a suite of support programs which aren't exclusive to RD, but can provide further information on genetics. Also established a digital animation project for younger children experiencing rare conditions.	No discernible qualifying activity.	No discernible qualifying activity.	The Rare Care Centre provides wholistic support for many WA children living with rare disease, although these services don't extend to all adults.
ASSESSMENT	Access to timely and accurate information within each jurisdiction remains inconsistent.							
1.3 - Develop national rare disease workforce strategy	★	★	★	★	★	★	★	★
Qualifications & Notes	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.
ASSESSMENT	No jurisdiction has taken a meaningful approach to workforce development, including the Commonwealth.							

SYMBOL KEY

★ Inadequate Actions Taken ➤ Some Action Underway ◆ Notable Progress
❖ Close to meeting the priority area, with improvement still possible. ✔ Implemented Priority Area

PILLAR 2	CARE AND SUPPORT							
PRIORITIES	**ACT**	**NSW**	**NT**	**QLD**	**SA**	**TAS**	**VIC**	**WA**
2.1 – Providing Care & Support for PLWRD	★	◆	★	★	★	➤	◆	✔
Qualifications & Notes	No discernible qualifying activity.	Rare Diseases NSW - Comprehensive Centre of Expertise for Rare and Undiagnosed Diseases - Driving research, care, and cure NSW Government - The Sydney Children's Hospitals Network - Kids rare diseases	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	Improving diagnostic pathways for rare diseases in regional Australia program.	Rare Disease Now	Multiple initiatives, including Rare Care Centre, and the WA Disease Strategic Framework
ASSESSMENT	**While each state has support services for people living with a range of conditions, rare disease specific programs are less common. NSW and VIC have meaningful programs, while WA's *Rare Care Centre* remains best practice.**							
2.2 – Ensure Diagnosis of Rare Disease is Timely & Accurate	➤	◆	➤	➤	➤	➤	➤	◆
Qualifications & Notes	Some activity, such as the Canberra Clinical Genomics program	Multiple initiatives, documented in Appendix.	Modest activity, including the Northern Territory Clinical Genetics Service.	Multiple initiatives, documented in Appendix	Multiple initiatives, documented in Appendix	Multiple initiatives, documented in Appendix	Multiple initiatives, documented in Appendix	Multiple initiatives, documented in Appendix
ASSESSMENT	**While governments have increasingly recognised the need to better focus on diagnostics for PLWRD, there remain nation-wide barriers to timely diagnosis. There are many individual services, however, that have been established, representing progress on this area. This could be improved by newborn diagnostics, and better genetic testing, as well as more dedicated state-specific services for PLWRD. While there are diagnostic pathways for PLWRD in each state, it can at times be complex, slow, and cumbersome.**							
2.3 - Facilitate increased reproductive confidence	★	★	★	★	★	★	★	★
Qualifications & Notes	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.
ASSESSMENT	**This analysis of programs has found little evidence of systematic approaches designed to achieve this priority area.**							

SYMBOL KEY

★ Inadequate Actions Taken ➤ Some Action Underway ◆ Notable Progress
◆ Close to meeting the priority area, with improvement still possible. ✔ Implemented Priority Area

PILLAR 2	CARE AND SUPPORT							
PRIORITIES	**ACT**	**NSW**	**NT**	**QLD**	**SA**	**TAS**	**VIC**	**WA**
2.4 – Enabling all Australians to have equitable access to the best health technology	★	➤	★	➤	➤	➤	➤	◆
Qualifications & Notes	No discernible qualifying activity.	Some activity, including the Kids Advance Therapeutic program	No discernible qualifying activity.	Multiple initiatives, listed in Appendix.	Some progress, noting the Review of the South Australian Clinical Genomics Plan, 2022.	Some progress under the Tasmanian Rare and Undiagnosed Rare Disease Network.	Some progress under the Genetic and Genomic Healthcare for Victoria 2021 program	LaunchR initiative is a standout program
ASSESSMENT	**Jurisdictions that have shown some progress on this have done so within broader rare disease policies and programs. It is unclear, however, the extent to which the identified programs have accelerate the realisation of this priority area. It should be noted that the Commonwealth carries considerable responsibility in this area. The Pilbara Hub of the Rare Care Centre supports access in remote WA.**							
2.5 – Integrate mental health support into rare disease care and support	★	★	★	★	◆	★	◆	❖
Qualifications & Notes	No discernible qualifying activity	No discernible qualifying activity	No discernible qualifying activity	No discernible qualifying activity	Rare and Undiagnosed Condition Parent Support Group (RUC) is consistent with this priority.	No discernible qualifying activity	Genetic Support Network of Victoria is consistent with this priority.	Rare Care Centre provides a central service, which can help in identifying support for PLWRD.
ASSESSMENT	**While most jurisdictions do not have a dedicated service providing mental health support for PLWRD, there is some progress in Victoria, NSW, and WA. WA has partnered with the federally funded Mindspot GP program to increase access to mental health support and upskill mental health service providers in RD. It has also built mental health support into RD clinical trials. Its Rare Care Centre is staffed with mental health clinicians.**							

SYMBOL KEY

★ Inadequate Actions Taken ➤ Some Action Underway ◆ Notable Progress
❖ Close to meeting the priority area, with improvement still possible. ✔ Implemented Priority Area

PILLAR 3	RESEARCH AND DATA							
PRIORITIES	**ACT**	**NSW**	**NT**	**QLD**	**SA**	**TAS**	**VIC**	**WA**
3.1 – Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning.	★	◆	★	★	◆	◆	◆	❖
Qualifications & Notes	No discernible qualifying activity.	Progress, focused on the NSW Register of Congenital Conditions.	No discernible qualifying activity.	No discernible qualifying activity.	SA Birth Defects Register is a notable program.	Progress seen in the Tasmanian Biorepository for Genomic Research into Disease.	Notable progress under the CareSync exchange.	While there may be some room for improvement still, WA's suite of programs represents the closest to full implementation of this objective.
ASSESSMENT	States have established a number of impactful programs which aid in achieving this objective. WA's Register of Developmental Anomalies with its implementation of rare disease coding (Orphacodes).							
3.2 – Develop a national research strategy for rare diseases to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps.	★	★	★	★	★	★	★	❖
Qualifications & Notes	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	-
ASSESSMENT	This has not been sufficiently met by the Commonwealth, nor state jurisdictions. WA, as with other areas, is the most advanced in terms of this objective. But this national approach remains outstanding. WA has a Collaborative Centre for Excellence for RD Research and Innovation, a Rare Disease Research Consortium with a coordinating Framework, Charter and series of flagships aligned to the National Plan. This brings together all WA Universities, multiple medical research institutes, public and private health service providers, industry and the community. WA is a leader in the International Rare Disease Research Consortium and ,is together with RVA, leading the National Australian Mirror Group for ERDERA.[60]							

SYMBOL KEY

★ Inadequate Actions Taken ➤ Some Action Underway ◆ Notable Progress
❖ Close to meeting the priority area, with improvement still possible. ✔ Implemented Priority Area

PILLAR 3	RESEARCH AND DATA							
PRIORITIES	**ACT**	**NSW**	**NT**	**QLD**	**SA**	**TAS**	**VIC**	**WA**
3.3 - Ensure research into rare diseases is collaborative and person-centred.	★	★	★	★	★	★	➤	◆
Qualifications & Notes	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	No discernible qualifying activity.	Tasmania's *Measuring the Impact of Rare Diseases* research project adopted this focus, but was a singular project, and not a systematic approach.	No discernible qualifying activity.	Although WA's *Rare Care Centre* is an outstanding program and a national leader, on this specific objective, there remains deeper person-cantered research focus that could be improved.
ASSESSMENT	**States have established a number of impactful programs which aid in achieving this objective. WA's Register of Developmental Anomalies with its implementation of rare disease coding (Orphacodes).**							
3.4 – Translate research and innovation into clinical care; clinical care informs research and innovation.	★	❖	★	◆	◆	◆	◆	❖
Qualifications & Notes	No discernible qualifying activity.	Rare Diseases NSW - Comprehensive Centre of Expertise for Rare and Undiagnosed Diseases - Driving research, care, and cure.	No discernible qualifying activity.	-	Embedding of genomic autopsy within WCHN services: Genomics Autopsy study.	NOT YET AVAILABLE TO PUBLIC Tasmanian Clinical Genomics Network.	Murdoch Children's Research Institute (MCRI)	Office of Population Health Genomics
ASSESSMENT	**There has been progress in this domain in recent years. As the Appendix shows, many jurisdictions have established a suite of programs and projects that have better linked clinical care and research. While progress remains required, there are positive steps being made towards this objective.**							

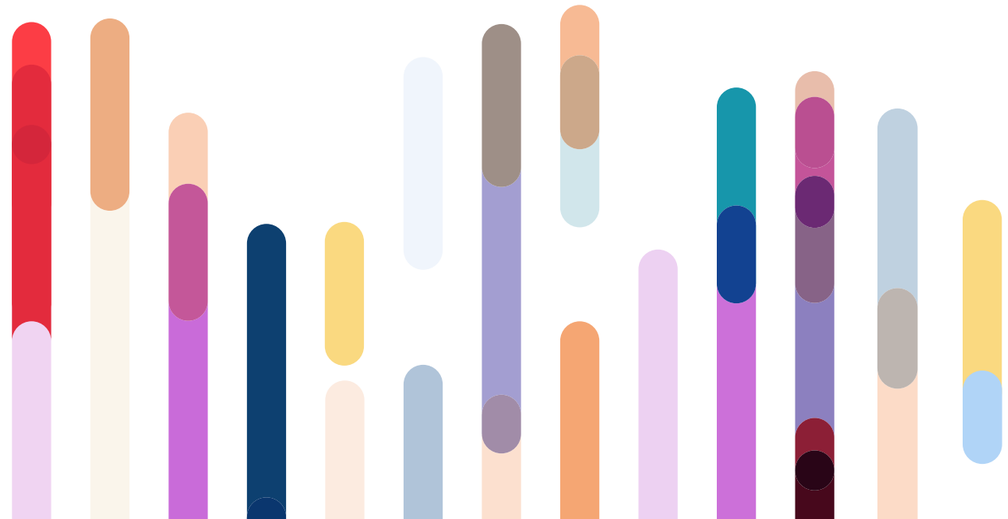
SYMBOL KEY

★ Inadequate Actions Taken ➤ Some Action Underway ◆ Notable Progress
❖ Close to meeting the priority area, with improvement still possible. ✔ Implemented Priority Area

Part Three: Identifying gaps in Australia's Rare Disease Policy Framework

Key points

1. While there is **important activity occurring in the rare disease policy space**, as Part 2 has demonstrated there are gaps in the responses of all governments.
2. **Strong rare disease policy planning** allows initiatives to be integrated within and across systems makes for lasting, impactful improvements.
3. While many states have exceptional individual programs, there is a **lack of consistency** across the country, meaning some Australians receive inadequate care simply due to where they live.
4. **Many programs in Australia demonstrate best practice** and could be scaled and replicated around the country.



As the mapping exercise in Part 2 demonstrated, there is a wide variety of activity and programs aimed at meeting each of the three pillars of the National Strategic Action Plan occurring across the Federation. There are, however, clear gaps that have emerged through this exercise that can guide future policy responses, both at a State and Territory level, and at a Commonwealth level.

GAP 1

National coordination

The first identified gap relates to a common and long-standing theme in the rare disease sector, and Theme #1 of the National Strategic Action Plan for Rare Diseases (the Action Plan): "The need for national leadership, coordination and consistency".[61] In the absence of this, and in the context of escalating unmet needs, the patchwork of rare disease programs are disconnected from one another. Despite the best of intentions, the absence of national coordination means that there is unnecessary duplication of effort, avoidable gaps in activities and, for many initiatives, lost opportunities to gain economies of scale.

This is evidenced in Rare Voices Australia's 2023 Status Report: Implementing the National Strategic Action Plan for Rare Diseases,[62] which "captured hundreds of activities" that can be aligned with the pillars and priorities of the Action Plan. Despite the best efforts of national bodies, such as Rare Voices Australia and Australian Genomics, the proliferation of disconnected activities, however well-intentioned, reflects incomplete implementation of Theme #1 and the absence of an annual implementation plan, ongoing stewardship and policy sustainability (see p.44).[63]

The severity of this gap is most keenly felt by Australians living with rare disease and their families and carers. Whilst these individuals are able to access various services to manage symptoms and support daily living available across the health system, rarely are these customised for the unique needs and circumstances of their disease. Indicative of this is the nature of services identified within the Rare Awareness Rare Education (RARE) Portal.[64] The overwhelming majority of resources and services available in each state and territory are not specific to rare disease.[65]

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—critical areas for improving rare disease outcomes. It means that pockets of rare disease expertise are dispersed and are not organised to collaborate in a systemic way.

Without stronger national leadership and sustained funding, Australians living with rare disease will continue to fall through the gaps in rare disease and related policy.

NATIONALLY COORDINATE POLICY

CASE STUDY

The Newborn Bloodspot Screening Program

The Newborn Bloodspot Screening (NBS) program illustrates how a nationally guided approach can be effectively implemented at the jurisdictional level.

Although the program is administered by states and territories, its overarching policy direction is now set at the national level. In 2018, the former Australian Health Ministers' Advisory Council (AHMAC) endorsed the *Newborn Bloodspot Screening – National Policy Framework*. [i] While the decision-making pathway outlined in that document has since been superseded, the framework set clear expectations for consistency, equity, and evidence-based decision-making. [ii]

The NBS program demonstrates how national coordination can promote alignment across jurisdictions while allowing for local implementation and flexibility.

> It provides 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 nationally guided, locally delivered model.

Genomics Australia offers another emerging example of national infrastructure to support jurisdictional implementation.

GAP 2

State and territory implementation strategies

There are two key national policy frameworks relating to rare disease - the *National Strategic Action Plan for Rare Diseases*, and the National Health Genomics Policy Framework 2018-2021. The former is not supported by implementation strategies in any jurisdiction, and the latter has been supported, to varying degrees, by only some jurisdictions (see Table X). This is despite growing recognition that adequately addressing rare disease policy areas is complex and requires dedicated system responses.[66]

This absence of state- and territory-level implementation strategies means that policy responses remain reactive rather than proactive, with initiatives often emerging in an ad-hoc manner rather than as part of a cohesive plan and these are often the result of the work of frustrated patient groups or clinicians. Rare disease policy is frequently overshadowed by other health and social priorities, leading to inconsistent levels of care, support, and research investment across jurisdictions, despite growing Australian evidence that rare disease care takes up a significantly greater proportion of resources than the significant percentage of the population directly impacted.[67,68] It hinders coordination between healthcare providers, social services, researchers, and policymakers. It means there is limited capacity to address specific regional challenges, such as service access in rural and remote areas or workforce shortages in specialist fields.

A demonstrative example is that of Rare Diseases NSW. This is an important and valuable service which provides the foundation for more comprehensive services in NSW. Yet it acknowledges: "While we work towards establishing a comprehensive centre that unites fragmented rare disease activities across New South Wales, we currently operate as an informal network of experts based at the Randwick Health and Innovation Precinct."[69] A NSW Rare Disease Strategy could transform this initiative from an aspiration into a reality, leading to immense benefits for people living with rare disease and the health and social systems more broadly.

The consequence, for Australians living with rare disease, is that patients and families affected by rare diseases are left navigating a fragmented system. Put more practically: they may receive world-class medical care from their hospital-based specialist, yet not be able to access the National Disability Insurance Scheme (NDIS) due to bureaucratic barriers; they may have access to brilliant peer support facilitated by a rare disease organisation, yet may be battling with their local primary school over the installation of a wheelchair ramp. Individuals and families living with rare disease have a full life of largely unmet needs; the states and territories can meet these needs with thoughtful, comprehensive policy responses.

Dedicated state and territory rare disease strategies and actions plans, aligned with the National Action Plan, would provide clarity, drive funding commitments, and embed accountability measures to ensure rare disease initiatives are effectively integrated into broader health and social systems. Importantly, as with other areas of health policy, these plans must address the full spectrum of unmet needs across the entire journey of the person and their family—not only those related to genomics and emerging technologies, but also the social, psychological, clinical, and practical supports required to ensure equitable care and quality of life for all Australians living with a rare disease.

GAP 3

An over-reliance on 'Rare Disease Champions'

An additional recurring theme of the consultations undertaken as part of this project was the important role that rare disease 'champions' play in progressing rare disease initiatives. Interviewees in both Western Australia and New South Wales attributed the notable difference in the presence and cohesiveness of rare disease activities between their jurisdiction and others to the efforts of rare disease champions.

A compelling case study demonstrating the impact of individual champions comes from Tasmania. In 2019, Dr Mathew Wallis became the state's first full-time Clinical Geneticist, joining the Tasmanian Health Service's Tasmanian Clinical Genetics Service and the University of Tasmania. Since then, Dr Wallis has played a catalytic role in advancing rare disease policy and care across the state. Notably, he was instrumental in the formation of the Tasmanian Rare and Undiagnosed Diseases Network — a cross-sector initiative that brings together people with lived experience, clinicians, researchers, and other stakeholders. This collaborative network exemplifies how leadership grounded in both clinical expertise and a commitment to lived experience can drive systemic progress in a complex and often under-recognised area of health.

It is well-established that Tasmanians must travel long distances to access certain health services, often outside the state.[70] A resident clinical geneticist has made a significant difference for Tasmanians living with rare disease.

Dr Wallis' role as a Tasmanian rare disease champion is evident in the proliferation of systemic rare disease work emerging from Tasmania, putting the state on the 'rare disease map'. This is in contrast to the state of play in Tasmania prior to his arrival, when the report "A stocktake of rare disease activities in Australia: Key activities, initiatives and programs occurring in Australia in the area of rare diseases, to inform the development of the National Strategic Action Plan for Rare Diseases" was prepared by Rare Voices Australia as a companion document to the Action Plan.[71]

At that time, it was noted only that "[d]ata is collected on congenital anomalies in... Tasmania" (p.29) and that a Clinical Trials Unit had been established in the years prior (p.34). While this list is not comprehensive, and no doubt there are other factors and individuals contributing to the building momentum in Tasmania, it helps demonstrate the significant impact that a rare disease champion, together with the teams of people they lead, can have on advancing outcomes for people living with rare disease.

Despite the marked difference that 'rare disease champions' can make for people living with rare disease in their jurisdiction, the fact that they are needed is reflective of Gaps 1 and 2, described in detail above. Locally, this reliance presents a key person risk, whilst nationally, excellence in one jurisdiction unmatched in others does result in significant inequities for Australians living with rare disease.

GAP 4

A lack of 'systems integration'

Throughout the consultations, a recurring theme was the criticality of system integration to drive real change for people living with rare disease. In this context, the need for system integration is juxtaposed against two main concepts: short-term funding and the proliferation of disconnected activities. While the latter concept has been explored above (See Gaps 1 and 2), the impact of short-term funding is explored further below.

It is common for rare disease activities to be funded in short-term cycles, whether that be in line with the election cycle, grant-funding periods, or as a result of philanthropic donations. As highlighted in the UDN-Aus case study presented earlier in this Report, when the funding period ends, so does the program.

However, there are examples of short-term projects being rapidly integrated into local systems by visionary individuals or health systems. Two relevant examples include the Undiagnosed Disease Program WA, which has been run and funded as a clinical service since 2015,[72] and the recent embedding of genomic autopsy as standard of care in the South Australian Women's and Children's Health Network following the success of the Genomic Autopsy Study and the strong, evidence-based advocacy of Professor Barnett.

GAP 5

Workforce development

While progress has been made in areas of research, diagnostics, and care, the development of a coordinated rare disease workforce remains a significant and largely unmet need across the Australian health system. Priority 1.3 of the National Strategic Action Plan— "Develop a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics"—has not yet been fully realised. Healthcare professionals across the country are navigating increasing complexity in diagnosis and care, particularly as genomics and precision medicine become more embedded in practice. However, without a nationally coordinated and well-supported approach to workforce development, there is a risk that capacity will remain uneven, and expertise concentrated in metropolitan hubs, limiting access to timely and accurate care for people living in regional and remote areas.

This is not a reflection of individual clinician capability or commitment—in fact, many professionals go above and beyond in their efforts to support patients with rare diseases. Rather, it highlights the need for structural investment in education, training, and sustained program funding to support a growing, multidisciplinary workforce. The experience of Rare Voices Australia in piloting a targeted strategy for the rare metabolic workforce[80] demonstrates how a strengths-based and collaborative approach can help address this gap in a constructive and inclusive way.

Workforce development is also closely tied to funding continuity. Short-term or project-based funding models often place pressure on skilled clinical and non-clinical staff such as care navigators, who may be unable to remain in roles due to financial insecurity. This disrupts both continuity of care and long-term

CASE STUDY

WA's Leadership in 'Systems Integration'

Western Australia (WA) has been at the forefront of rare disease research, practice, policy and infrastructure for five decades. The WA Cerebral Palsy Register (1975)[73] and the WA Congenital Malformations Registry (1980)[74] laid the foundation for the Western Australian Register of Developmental Anomalies (WARDA), supported by legislation since 2011.[75]

Genetic Health WA (GHWA) has provided state-wide genetic diagnostics and counselling for over 30 years, covering paediatric, cancer, and obstetric genetics. Additionally, the WA Data Linkage System, established in 1995, remains a global leader in population health research.[76]

> WA Health is the only health system that is a member of the International Rare Disease Research Consortium, the global multistakeholder body harmonising and advancing rare disease research.

The Rare Care Centre has a leadership role in the World Health Organisation Global Network for Rare Disease, the Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease and is on the Steering Committee for the Lancet Commission on Rare Disease.

The Office of Population Health Genomics (OPHG), founded in 2001, drives genomic policy and research, including the WA Genomics Strategy 2022-2032[77] and the WA Rare Diseases Strategic Framework 2015-18[78]—the only state-level rare disease policy in Australia.[79]

As a result of integrating rare disease activity within the system, including in legislation, there has been a proliferation of activity, over an extended period of time, making meaningful improvements to the lives of Western Australians with rare disease.

> With ongoing advancements and strong policy support, WA continues to be a global leader in rare disease healthcare.

knowledge-building. A sustainable, national funding model—backed by coordinated state and federal investment—would help retain expertise, ensure equitable access to care, and support the full integration of genomics and other innovations into mainstream services.

The optimal use of existing workforces is also vital. For example, the growth in genetic testing has increased the need for access to genetic counselling services. Despite a strong existing workforce, access to counselling is limited by the non-inclusion of these services on the Medicare Benefits Schedule which means that for many Australians, affordable access to genetic counselling is only available through overstretched public hospital systems with unacceptable waiting times. Including these services on the MBS would unlock the workforce in a way they can support the more comprehensive and higher impact use of genetic testing.

GAP 6

Adequate funding for hospital-based treatment and care

A core challenge in Australia's current response to rare diseases is the fragmented and inconsistently funded approach to hospital-based care and coordination. While state and territory governments frequently shoulder the cost of delivering novel treatments and managing complex cases within the hospital system, the absence of a sustainable national funding framework limits the system's capacity to provide equitable and coordinated care. Rare disease patients often require ongoing, multidisciplinary care—including input from medical specialists, allied health professionals, mental health practitioners, and genetic counsellors—but in many regions, particularly outside major metropolitan centres, access to these services remains limited or unavailable.

Hospitals are not always funded or resourced to offer the integrated care that rare disease patients need. Without dedicated support for roles like care navigators, interagency partnerships, multidisciplinary teams (MDTs), and telehealth-enabled outreach, care delivery can become siloed, reactive, and burdensome for families. Even in well-resourced hospitals, the cost burden of rare and emerging therapies, comprehensive diagnostic testing, and integration of genomic technologies is frequently absorbed within existing budgets, contributing to delays, care rationing, and missed opportunities for early intervention. Further, the lack of rare disease coding (e.g. Orphacodes) means the system does not track rare disease costs, nor does it have a means to show enormous savings from interventions – burying efficiency opportunities and threatening healthcare sustainability. This is limiting both clinical outcomes, and efficiencies within the system.

This gap underscores the need for a nationally coordinated model of care that supports the delivery of specialised, patient-centred services—regardless of a patient's postcode or age. The Rare Care Centre in Western Australia provides a strong example of what this could look like: a hub offering a single point of care for children with a rare disease and their families, led by generalists (nurses, general paediatricians, primary care providers, and clinical geneticists), underpinned by a care coordination ethos, and supported by MDTs, specialist medical and allied health input, accessible telehealth and a digital data ecosystem that is AI-empowered. Calls for similar models in other jurisdictions, such as the proposed Tasmanian Rare Care Centre, reflect a clear appetite among stakeholders for solutions that address fragmentation and improve patient outcomes.[81]

To make such models sustainable and scalable, Commonwealth leadership is critical. A national funding partnership could help close existing service gaps, support the expansion of coordinated care models, and ensure that all Australians living with a rare disease have access to comprehensive, multidisciplinary hospital-based care—no matter where they live.

GAP 7

Access to innovative medicines for rare diseases

For many rare diseases access to innovative medicines can provide treatment for both the underlying causes and symptoms of the condition. While generally not cures for genetic conditions, those medicines can be lifesaving or prevent severe disability and mitigate against impacts on a patient's quality of life.

Australia has an excellent healthcare system, and, at the federal level, this is supported by the two pillars of Medicare and the Pharmaceutical Benefits Scheme (PBS). The PBS helps Australians access medicines at affordable prices, through government reimbursement of the costs to the supplier.

However, applications for the PBS reimbursement of rare disease therapies can take too long and face barriers which can deny Australian patients access to treatments available overseas, and in the face of progressive deterioration and co-morbidity compounding. Innovative medicines, on average, take 466 days from registration to listing.[82] Rare disease therapies can require multiple rounds of submissions to the PBAC which delays access to patients. These challenges have been identified in earlier work by the McKell Institute including its 2014 report, *Funding Rare Disease Therapies in Australia – Ensuring equitable access to health for all Australians* and a 2020 update of that report. The need for reform was also a central focus of the House of Representatives Standing Committee on Health's landmark report, The New Frontier in 2021.

The government has recognised that the health technology assessment (HTA) processes used to assess applications for PBS listing need reform. It commissioned a major review of HTA systems which was released by the federal health minister in 2024. Its 50 recommendations are being considered by an HTA Implementation Advisory Group. To ensure health equity for Australians living with a rare disease, it is essential these reforms are implemented and the often unique issues faced by rare disease medicines are recognised in that process.

Specifically, the following reforms would ensure greater health equity for rare disease patients for applications submitted for PBS listing:

STREAMLINING THE PROCESS AND ENGAGING PATIENTS.

The HTA review recommended that 90 percent of applications for innovative medicines should be listed on the PBS within 6 months of registration. For lifesaving medicines for ultrarare diseases, it recommended a four-week timeline.

A system that delivered these outcomes would improve equitable access for rare disease patients. It is also vital that patients and their family and carers be empowered to participate in the HTA process. Considerable progress has been made in developing an Enhanced Consumer Engagement Process and this must remain a priority for the federal government.

HTA processes should also permit patient and clinician participation early in the assessment of applications through

stakeholder workshops, which would bring together applicant companies, patients, clinicians, the health department, and the Pharmaceutical Benefits Advisory Committee (PBAC). Such stakeholder workshops should become the standard rather than the exception for rare diseases where the complexity of new treatments and the existing knowledge base of the department and PBAC may not be as developed as for more common disease.

EXPANDING THE LIFE SAVINGS DRUG PROGRAM (LSDP)

The LSPD provides reimbursement for the cost of medicines for ultrarare diseases where those medicines would not meet the PBAC's normal cost-effective evaluation criteria and are lifesaving.

While the LSDP plays an important role in supporting medicines for a small number of diseases, there remains a gulf between PBS listed medicines and the LSDP for medicines that are not deemed cost-effective and cannot meet the stringent criteria of being lifesaving. This means that many ultrarare disease patients are not able to access medicines which could be lifechanging by reducing disability and significantly improve their quality of life.

The HTA review recognised the need for a more streamlined application pathway, however as part of the review of the LSDP eligibility criteria recommended by the HTA review, the government should support the expansion of the LSDP to include lifechanging medicines which prevent significant disability for ultrarare diseases.

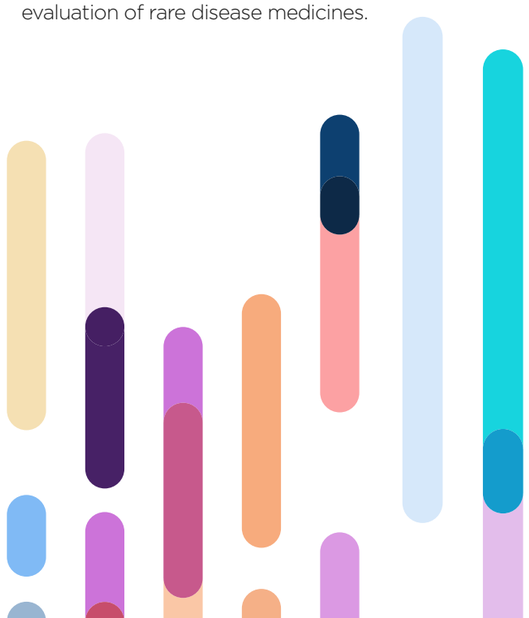
Preserving the LSDP as a separate funding program and providing faster access to the LSDP, as recommended by the HTA review, would also ensure more equitable access for patients with ultra-rare disease, who often have no alternative treatment options.

RECOGNISING THE LONG-TERM AND BROADER BENEFITS OF MEDICINES FOR RARE DISEASE PATIENTS.

Rare disease medicines often require lifelong usage, starting in childhood. These types of medicines struggle to demonstrate cost-effectiveness because the current HTA system fails to recognise their long-term benefits via the application of discount rates. The discount rate, at 5 percent, is higher than for most other comparable nations. Reducing the discount rate to 1.5 percent for lifelong rare disease medicines would improve the likelihood of PBS listing.

In assessing new medicines, the PBAC considers the impact of those treatments on the lives of patients and their productivity. It also considers the impact on the federal health budget. What those evaluations do not routinely consider are other benefits, such as for carers who are often parents who leave full-time employment to support their child reducing workforce productivity. They also don't consider the benefits for other government spending programs, like the NDIS, which may benefit from reduced spending if a medicine is listed on the PBS and is able to prevent or reduce disability.

Incorporating these second order benefits as a feature of HTA evaluations would ensure a fairer evaluation of rare disease medicines.



Conclusion

Australians are fortunate to have access to world leading health care. However, for Australians living with rare disease, gaps in treatment, care and long-term policy support which arise from the nature of rare disease are evident.

While individual 'rare diseases' might be uncommon, the incidence of rare disease is not: it is estimated that up to one in eight Australians experience a rare disease of some form. As this report has outlined, care, support and treatment for those Australians vary considerably depending on the jurisdiction in which they live.

At the heart of the gaps in Australia's rare disease policy framework is a lack of coordination between jurisdictions, especially between states and the Commonwealth. As this report has highlighted, there are many impactful initiatives and programs underway across the country that are providing care, support and therapy for Australians living with rare disease. A patchwork of policy settings and programs, however, means that navigating this ecosystem can be challenging for patients, as well as practitioners and policymakers.

This report has advanced eight recommendations aimed at strengthening Australia's rare disease policy framework. Chief amongst these is the enactment of a Rare Disease Act, which establishes an Office of Rare Disease (ORD). A centralised body such as the ORD would be better placed to address the numerous issues identified in this report including in relation to workforce coordination and ensuring funding consistency.

While there has been a considerable improvement in government support for Australians living with rare disease in recent years, gaps in care, treatment and coordination remain significant. However, from this report it can be seen that a comprehensive policy response to the rare disease healthcare challenge is possible. Nevertheless, without such further co-ordinated and consistent effort, the danger is that rare disease challenges become insurmountable and unfairly, all current and future Australians living with rare disease will not have access to the best care, support, and treatment available.

Appendix
A Catalogue of Rare Disease Policies by Pillar and State

PILLAR 1	AWARENESS AND EDUCATION							
PRIORITIES	**ACT**	**NSW**	**NT**	**QLD**	**SA**	**TAS**	**VIC**	**WA**
Priority 1.1: Increase every Australian's awareness of rare diseases including, where applicable, relevant prevention measures.						20-Year Preventive Health Strategy (in development)	The Science Within Us	
Priority 1.2: Ensure Australians living with a rare disease have access to information and education that enables them to be active participants in their rare disease journey.		NSW Government - Health - Centre for Genetics Education			An Australian first digital and animation pathway for clinical genetics appointments			
Priority 1.2: Develop a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics.								

PILLAR 2	CARE AND SUPPORT							
PRIORITIES	**ACT**	**NSW**	**NT**	**QLD**	**SA**	**TAS**	**VIC**	**WA**
Priority 2.1: Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family-centred.		Rare Diseases NSW - Comprehensive Centre of Expertise for Rare and Undiagnosed Diseases - Driving research, care, and cure NSW Government - The Sydney Children's Hospitals Network - Kids rare diseases				Improving diagnostic pathways for rare diseases in regional Australia	Project Coordinator (Rare Disease) (RDNow)	Rare Care Centre Rare Care Centre's Pilbara Hub WA Rare Diseases Strategic Framework 2015-18
Priority 2.2: Ensure diagnosis of a rare disease is timely and accurate.	Canberra Clinical Genomics	NSW Health Genomics Strategy NSW Health Genomics Strategy Implementation Plan 2018-20 NSW Health Genomics Strategy Implementation Plan (2021-2025) NSW Government - NSW Health Pathology - Genomics - Rare Diseases Genomics service - Statewide Sequencing service	Northern Territory Clinical Genetics Service (c/o VCGS)	Genetic Health Queensland Whole Genome Sequencing pilot helps patients and families of rare disease Achieving Equity in Genomic Health (AEGH) for Indigenous Australians	SA Clinical Genetics Service Doubling of the genetic workforce in South Australia	Tasmanian Clinical Genetics Service The importance of a resident clinical geneticist	Victorian Clinical Genetics Services Rare Disease Now (RDNow)	Genetic Health Western Australia (GHWA) Western Australian Undiagnosed Diseases Program
Priority 2.3: Facilitate increased reproductive confidence.								
Priority 2.4: Enable all Australians to have equitable access to the best available health technology.		Kids Advanced Therapeutics		Indigenous Genomics Health Literacy Project (IG-HeLP) Queensland Children's Hospital ACTION Centre - Research Theme - Rare Diseases (NOT AVAILABLE TO PUBLIC) Queensland Health Genomics and Precision Medicine Strategic Roadmap 2021-2026	Review of the South Australian Clinical Genomics Plan 2022	Tasmanian Rare and Undiagnosed Diseases Network (TRUDN)	Genetic and genomic healthcare for Victoria 2021	LaunchR
Priority 2.5: Integrate mental health, and social and emotional wellbeing, into rare disease care and support.					Rare and Undiagnosed Condition Parent Support Group (RUC)		Genetic Support Network of Victoria	

PILLAR 3	RESEARCH AND DATA							
PRIORITIES	**ACT**	**NSW**	**NT**	**QLD**	**SA**	**TAS**	**VIC**	**WA**
Priority 3.1: Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning.		NSW Register of Congenital Conditions			South Australian Birth Defects Register (SABDR)	A Tasmanian Biorepository for Genomic Research into Disease (Tas-Bio-GRiD)	CareSync Exchange: Victoria's secure patient health information sharing system	Western Australian Register of Developmental Anomalies Health (Western Australian Register of Developmental Anomalies) Regulations 2010 Western Australian Data Linkage System
Priority 3.2: Develop a national research strategy for rare diseases to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps.								
Priority 3.3: Ensure research into rare diseases is collaborative and person-centred.						Measuring the impact of rare diseases in Tasmania, Australia		
Priority 3.4: Translate research and innovation into clinical care; clinical care informs research and innovation.		Rare Diseases NSW - Comprehensive Centre of Expertise for Rare and Undiagnosed Diseases - Driving research, care, and cure		Genomics Research Centre (GRC)	Embedding of genomic autopsy within WCHN services: Genomics Autopsy study	Tasmanian Clinical Genomics Network	Murdoch Children's Research Institute (MCRI)	Office of Population Health Genomics

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