Resource on the State of the Art of Rare Disease Activities

2025 Report for Australia

National Mirror Group

Australia does not have a National Mirror Group (NMG) currently in place. However, an NMG is currently in development in Australia and will be a collaboration between Rare Voices Australia and the Rare Care Centre (Perth Children's Hospital).

A data contributing committee has overseen the collection of data for this report. Contributors are listed at the end of this report.

Definition of a Rare Disease

Australia uses the definition that a rare disease is defined as is one that affects less than five in 10,000 people. The national plan for Australia espouses this definition and there are no instances when a different definition is used. This definition does not cause any challenges in Australia, but it was noted that the biggest challenge is around raising awareness of rare disease in general.

Status Quo of any National Plan or Strategy for Rare Disease

What is the status quo?

Australia has an active, open-ended, National Action Plan, officially described as the 'National Strategic Action Plan for Rare Diseases'. The Action Plan was written to be an open-ended plan.

Elaboration and Adoption/How was the original plan elaborated?

Commissioned by the Australian Government, Rare Voices Australia (RVA) led the collaborative development of the plan, which is the first nationally coordinated effort to address rare disease in Australia. It is not enshrined in law but is an Australian Department of Health Government policy, providing a common language and shared direction for the whole rare disease sector in Australia.

RVA is the national peak body for people in Australia living with a rare disease. In 2018, the Australian Government commissioned RVA to lead the development of Australia's Action Plan for rare disease.

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The plan was launched in February 2020 by the Minister for Health, with bipartisan support. RVA continues to work with State and Federal Governments, as well as other stakeholders, in leading the collaborative implementation of the plan. The following stakeholder types are included in this group:

- Patients/people with lived experience of rare condition
- Health Ministry/Competent National Authority in charge of Health or Care
- Research Ministry/ Competent National Authority in charge of Research
- Social or Welfare Ministry/ Competent National Authority in charge of social affairs
- Research funders
- Researchers/clinicians from rare disease centres

For further information about the involvement of different stakeholders, please refer to the links below:

- Engaging with the rare disease community in Australia
- A stocktake of rare disease activities in Australia
- The evidence base for the National Strategic Action Plan for Rare Diseases in Australia

How is it Implemented and/or Monitored or Evaluated?

The same group that drafted/adopted Australia's Action Plan is also responsible for implementing and overseeing the plan. The group was specifically established for the purpose of drafting and implementing Australia's Action Plan for Rare Disease. The implementation/oversight group meets regularly.

During development of Australia's Action Plan for Rare Diseases eight key themes were identified. These themes are used by Australia's rare disease sector as measures of progress:

- Theme 1: National leadership, coordination and consistency
- Theme 2: Prioritisation of systematic building of knowledge, evidence and expertise
- Theme 3: A person-centred approach
- Theme 4: Rare disease measurement
- Theme 5: Sustainable systems and workforce
- Theme 6 Stakeholder collaboration
- Theme 7: State, national and international cross-sector (i.e. specifically across government sectors) collaboration
- Theme 8: Progression of early implementation wherever possible.

Implementation of the plan is the shared responsibility of all Australian rare disease stakeholders. Monitoring and evaluation of activity is undertaken by, and the responsibility of, those conducting activities in line with the Action Plan. However, as Australia's peak body for rare disease and leading the collaborative implementation of the Action Plan, RVA has taken steps to monitor and evaluate cross-sector implementation of the Action Plan.

In 2023, RVA conducted a sector-wide activity scan, inviting the rare disease sector to share their projects, initiatives and achievements. The activity scan captured hundreds of activities aligning with the three pillars (Awareness and Education, Care and Support, Research and Data) and priorities of the plan. These activities were categorised according to 5 key elements of progress: input, activities or processes, outputs, outcomes and impact. The activities were also mapped to the eight key themes in the plan. Results of this work are published in a status report here.

No funding or resources are committed to the plan itself but there are funds dedicated to deliver actions within the plan.

Research in National Plans or national research strategies relevant to rare disease

The scope of the Action Plan aligns with international rare disease plans. Additionally, RVA has relationships with many international rare disease initiatives including the International Rare Diseases Research Consortium (IRDiRC), Rare Disease International, Health Technology Assessment international (HTAi), EURORDIS, Asia Pacific Alliance for Rare Disease Organisations (APARDO), World Health Organisation and MetabERN.

The Action Plan is flexible and thus able to respond to changing policy contexts and opportunities. All three Pillars of the Action Plan are interrelated. Therefore, the strongest policy responses address priorities across multiple Pillars. Effective policy reform in one area will create change and momentum in other areas.

The Action Plan aligns with and is complemented by a range of continually changing policies including, National Health Genomics Policy Framework, Newborn Blood Spot Screening Policy, Chronic Conditions Framework, various Health and Medical Research policies, policies around Australia's Life Saving Drug Program and broader Health Technology Assessment landscape. RVA is closely engaged with various government departments responsible for these policies to ensure gaps in rare disease are considered and addressed.

Select Achievements of the National Plan

Five key achievements resulting from Australia’s Action Plan are:

1. Initial funding of AU$3.3 million directly in response to the launch of the National Strategic Action Plan in 2020. This funding was to progress implementation of Pillar 1 - Awareness and Education
2. Specific influence on policy reform, including on the Health Technology Assessment review and National Medicines Policy, Newborn Screening policy reform, and health and medical research policy reform
3. RARE Portal development/RArEST (Rare Disease Awareness, Education, Support and Training) Project. Further information is available here
4. Peak body recognition and growing profile, increased sector coordination and partnerships
5. Increased effective/impactful rare disease research partnerships and research policy advocacy

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Rare Disease Research Programmes and Funding

Australia's National Action Plan specifically addresses research. Research and Data is one of the three pillars within the Action Plan. There is evidence of the Action Plan for Rare disease influencing health and medical research funding policy and direction. RVA is on the Technical Reference Group for Australia's first National Health and Medical Research Strategy, which will be finalised by December 2025. RVA has published a report outlining Australia's Top 10 Rare Disease Research Priorities here.

The National Plan specifically addresses the following areas:

- National RD research investment, opportunities and funding calls
- International/transnational RD research investment, opportunities and funding calls
- Registries or registry catalogues for rare diseases
- Biobanks/biosample catalogues for rare diseases
- Ontologies, codification or data standardisation
- Diagnostics research ('solving unknown conditions')
- Basic research (e.g. cell lines, animal models etc)
- Clinical/Translational research
- Sociological (e.g. Quality-of-Life-related) or socio-economic research
- Rare disease treatments, including HTA

The Action Plan includes specific mention of the following initiatives/organisations: IRDiRC, EURORDIS, APARDO, European Reference Networks, and Asia-Pacific Economic Cooperation (APEC) Action Plan on Rare Diseases. Australia's top ten Recommendations for a National Approach to Rare Disease Data encourages alignment with IRDiRC's goals and priorities for 2017-2027, as well as the United Nations Resolution on Addressing the Challenges of Persons Living with a Rare Disease. The recommendations also highlight and align with the Joint Declaration of ten Key Principles for Rare Disease Patient Registries.

Australia has specific research programme/funding calls dedicated to rare disease. These research programmes/funding calls are funded by both public and private bodies and are launched regularly. In Australia, public funding for rare disease research is primarily delivered through two government initiatives: the Medical Research Future Fund (MRFF) and the National Health and Medical Research Council (NHMRC). To date, rare disease–specific funding calls have been offered exclusively through the MRFF. These include the Clinical Trials Activity grant stream for rare cancers, rare diseases, and unmet needs, which commenced in 2022 and remains ongoing, and the Genomics Health Futures Mission, which has explicitly identified rare disease as a core focus area. The latter includes dedicated investments and targeted initiatives to advance diagnostics, treatments, and understanding of rare genetic conditions and rare cancers, as well as to integrate genomic medicine into clinical practice.

In the private sector, several Australian rare disease organisations and research foundations provide funding for research into their specific conditions of interest, whilst some clinical services actively pursue larger philanthropic investments to support rare disease research.

Public funding calls for rare disease research often encompass broad categories and multiple disease areas, making it difficult to determine even an approximate value of the national investment. Moreover, despite efforts by the MRFF to highlight rare disease in its funding calls, comparatively few rare disease research programs are ultimately funded relative to other disease areas. RVA has raised this issue in its advocacy to the Health and Medical Research Office, calling for rare disease research funding to be more accurately tracked within their systems. These discussions remain ongoing. RVA is not aware of any available data on rare disease research funding within the private sector. However, with the forthcoming release of Australia's first ever National Health and Medical Research Strategy, there are recommendations to improve understanding of research funding sources across the country. Moving forward, RVA will leverage any opportunity to ensure that rare disease research funding is captured within these national data efforts.

The third pillar of the National Strategic Action Plan for Rare Diseases focuses on research and data. However, it was not accompanied by any dedicated research funding. RVA is advocating for the Australian Government's forthcoming National Health and Medical Research Strategy to explicitly recognise and prioritise investment in rare disease research across the entire research pipeline. Currently, the only specific policy mechanism supporting rare disease research funding comes through the MRFF, which has issued dedicated funding calls via the Clinical Trials Activity grants and the Genomics Health Futures Mission grants.

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Rare Disease Registration and Biobanking

Australia does not have a national nor regional rare disease registries. Establishing a rare disease registry was recommended within Australia's Action Plan but this was prevented due to a lack of funding. In Australia, there are several disease specific or group of rare disease registries, all of which have soft and non-recurrent funding and are almost exclusively supported through research and other grants. The greatest area of growth is in patient advocacy organisation-driven data collection. There are many individual patient advocacy organisations that have developed and maintain their own rare-disease specific registry.

There is no integrated, sustainable pan-rare disease specific biobanking nationally or state-based initiatives in Australia. There are a number of individual or group of rare disease specific biobanks that are supported through research granting and philanthropy. The majority of such initiatives are for more common and well-funded Rare Diseases.

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Organisation of Rare Disease Care

Currently, there is no centralised mechanism to identify rare disease expertise in Australia. In practice, many Australian rare disease organisations play a vital role in supporting patients and families to locate appropriate expertise. Some of these organisations maintain clinical directories, which may be either publicly accessible or restricted to members. Health professionals are also a key pathway through which patients and families identify rare disease expertise. However, even within the healthcare system, there is often limited knowledge and awareness of appropriate referral pathways for people living with rare diseases.

The RARE Portal, Australia's national resource for rare disease, provides condition-specific information and resources tailored to the Australian context. These information pages include guidance on the types of expertise that may be required for particular rare conditions, as well as a link to a directory of genetic counsellors. The National Health Services Directory offers a searchable national directory of health services, including hospitals, specialty clinics, and healthcare professionals by state or territory. However, this directory is not tailored specifically to the needs of people living with rare diseases.

One of RVA's strategic priorities is to drive the recognition and development of networked rare disease centres of expertise. RVA is engaged in ongoing advocacy with the Department of Health, Disability and Aged Care, promoting this model as a key solution to addressing the significant gaps in care and support for Australians living with rare diseases. RVA is also leading several projects aimed at building the evidence base to support this approach. RVA's Scientific and Medical Advisory Committee is developing draft guidelines for Australian networked rare disease centres of expertise. These guidelines are designed to recognise the diversity of health system structures and service delivery models across the country, and to enable the authentic development and integration of rare disease centres of expertise within these existing frameworks. Currently, there are two rare disease–agnostic centres of expertise operating/under development in Australia: The Rare Care Centre in Western Australia, and Rare Diseases New South Wales, based within the Sydney Children's Hospitals Network. In addition, there are a handful of rare disease–specific centres of expertise, including one for Prader-Willi Syndrome and the Lifespan Metabolic Medicines Clinic in Queensland.

Newborn Screening

National recommendations and state-based implementation of Newborn Bloodspot Screening (NBS) currently vary in Australia. The program is currently expanding to ensure that all babies born in Australia have access to the same screening tests. 32 conditions are screened consistently nationally, a further five conditions are currently being added and are inconsistently tested for between states. There are a further 18 conditions that are not targeted but may be incidentally detected when screening for a target condition. The list of conditions screened for in Australia and further information can be found here.

Australia has policies pertaining to newborn screening and they are outlined within the National Policy Framework for NBS, further information is available here. Note: the decision making pathway outlined in the framework is out of date, the updated decision pathway is detailed here.

Australia does not have any known formal collaborations with other countries concerning newborn screening decisions. However, Genomic Screening Consortium for Australian Newborns (GenSCAN) has awarded funding to five research projects to explore how genomics might be incorporated within newborn screening programs. GenSCAN has links to Newborn Screening programs in England, Canda and the USA.

Diagnostics

The Human Genetics Society of Australasia (HGSA) maintains a list of clinical genetic services across Australia and the wider Australasia region. This list does not include specific laboratories and the tests that they perform but it includes clinical genetic services, which are the access point to receive testing. The list of clinical genetic services is available to view here.

Public funding for genetic testing is a mix of federal and state-based funding. State-based funding is organised variably between states and is a mix of funding via dedicated centres of excellence/clinical services or their equivalents. At a federal level, funding is provided by the Medicare Benefits Schedule (MBS) but only for a limited range of testing possibilities. More information is available here (tests have to be searched for individually).

There are no formal, standardised, agreements for cross-border testing at a national or state level. There is no specific mention of cross-border testing within the National Action Plan for Rare Disease.

There are a number of state-based research and clinical initiatives to address undiagnosed patients/people with currently undiagnosable conditions in Australia. A nationally funded research project, UDN-Aus, is completing and further funding for this project has not yet been secured. Multiple individual institutions in Australia participate in Undiagnosed Diseases Network International (UDNI) and Undiagnosed Hackathons. Some examples of state-based programs are listed below:

- Undiagnosed Diseases Program-Western Australia (UDP WA), first program in Australia, commenced 2016
- RD Now, Victoria
- GeneAdd, New South Wales

Australia does not have a single, mandatory, national genetic counselling policy, but rather a professional self-regulatory framework managed by the Human Genetics Society of Australasia (HGSA). This framework sets standards, competency frameworks, and a code of ethics, with the ultimate aim of ensuring patient safety and quality of care. Government-funded services are primarily delivered by public hospitals, while access is managed through physician referrals to Medicare-funded clinical genetics services, although proposals are underway to enhance Medicare coverage for genetic counselling services. The Independent Health and Aged Care Pricing Authority determines reimbursement for publicly funded genetic counselling, further information is available here. Access to clinical genetics and genetic counselling is partly governed by the Medicare Benefit Scheme.

National Alliances of Rare Disease Patient Organisations

Rare Voices Australia (RVA) is the National Alliance for rare disease patient organisations in Australia. RVA formally partners with over 100 rare disease partner organisations, collectively called 'RVA Partners'. These organisations are required to meet the following criteria:

- Can demonstrate a representative perspective for a rare disease(s) community
- Must be a consumer-led, not-for-profit group/organisation with no commercial incentive

There are several benefits to becoming an RVA Partner organisation including, bespoke mentorship on advocacy and strategy, access to RVA's education program, and an invitation to the biennial National Rare Disease Summit. RVA also formally partners with individuals and research groups. For more information about RVA, click here.

RVA is involved in setting strategic direction for rare disease research. In 2025, RVA, with guidance from its Scientific and Medical Advisory Committee, partnered with experts at The Kids Research Institute Australia to conduct a modified James Lind Alliance Priority Setting Partnership Project. This process identified Australia's top ten rare disease research priorities, which were recently published. The full report can be accessed here. Beyond this project, RVA actively leads research advocacy. RVA works closely with the National Health and Medical Research Office and contributes to key consultations on research and data strategies and plans. This ensures that rare disease remains visible and prioritised in national policy and research funding initiatives.

RVA provides several opportunities for RVA partners to build their capacity and provides educational activities. An example is the RVA Partner Forum. This forum provides an opportunity for leaders of rare disease organisations to come together, exchange ideas and learnings. Additionally, RVA partners can access to RVA's Education Program, which contains short courses and resources across several topics, including rare disease advocacy, understanding Australia's rare disease landscape and writing effective government submissions.

RVA does not conduct any formal surveys about patient engagement in research. However, RVA contributes rare disease peak body expertise to a number of national consultations working to improve patient engagement in health and medical research more broadly. These include the National Statement on Consumer and Community Involvement in Research (under revision) and the National Statement in Ethical Conduct in Human Research. RVA was also involved in a review and evaluation of Involve Australia's Guidelines for Community Involvement in Genomic Research, which aims to provide genomic researchers in Australia with practical information for involving community members effectively and meaningfully in research projects. More information about these guidelines can be found here. In addition to this, RVA has a formal Research Partnerships program which acts as a conduit to over 100 RVA Partners to link researchers with the right consumer expertise. As part of this formal program, RVA consultancy to researchers includes:

- Provision of broad rare disease stakeholder knowledge and expertise as Australia's national peak body for Australians living with a rare disease, including a strong track record in effective rare disease advocacy and policy influence
- Support to align research with priorities in the Action Plan
- Support to identify opportunities for translation of research outcomes into measurable impact, including rare disease policy reform to achieve the best possible health and wellbeing outcomes for Australians living with a rare disease
- Provision of rare disease consumer expertise
- Guidance on a person-centred approach to research co-design and implementation
- Links to a network of over 100 rare disease patient groups and other rare disease stakeholders (in most cases, RVA can provide these links without a formal research partnership if this is all that is required)
- Assistance convening focus groups to facilitate meaningful consumer engagement
- Support leveraging external grant funding for rare disease research

More broadly, there are significant barriers to meaningful consumer and community involvement (CCI) in research in Australia. These include the absence of a national framework for remuneration, which undervalues contributions; the lack of diversity and representative participation, which limits inclusivity; and consultation fatigue, where the same individuals are repeatedly asked to contribute without sufficient support. Importantly, there is also a lack of accountability for researchers to report on CCI, which diminishes transparency and weakens incentives for genuine engagement. Many of these issues are being increasingly recognised and there is work underway to address these barriers in Australia. For people living with a rare disease, these barriers are even more pronounced. Poor health and complex care needs can restrict participation, whilst carers often face severe time and energy constraints, making it difficult to engage despite their desire to be involved and contribute critical insights.

Information Resources for Rare Disease

Australia has a national helpline and portal for rare disease. The Rare Awareness Rare Education (RARE) portal and the RARE Helpline are available to all Australians living with a rare disease. The RARE Portal is a national resource for rare diseases, that includes an online library of current, reliable and straightforward rare disease information. It includes a comprehensive collection of national, state and territory based rare disease information, resources, services and supports relevant to all Australian rare disease stakeholders. The RARE Portal is being collaboratively developed and customised for the Australian context.

The RARE Helpline aims to provide timely access to information and answer key questions people living with a rare and complex disease often face. The RARE Helpline is staffed by RVA personnel, including those with qualifications in psychology, social work, and Mental Health First Aid. The RARE Helpline can:

- Support people to connect with existing reliable information
- Provide resources that respond to people’s specific needs
- Help to increase people’s health literacy and engagement with care and support services
- Assist in connecting people with existing health services where possible

The RARE helpline is supported by public funding. It is part of a time-limited project, called the Navigator Project, which is funded by the national Department of Health, Disability and Aged Care. Further information about this project can be found here.

Training and Education

There are training and education activities in rare disease in Australia. Topics included in these activities are awareness raising and diagnostics. A selection of initiatives are listed below:

- Rare Disease Awareness Support and training project (recently concluded)
- Rare ECHO

- Global Nursing Network Rare Diseases – Resource Hub
- Global Nursing Network Rare Disease – Rare Disease Resources for Educators
- Palliative Care Australia - Think Rare, Think Care, Think Together
- Medics4Rare Disease 101 (Australia version)

Orphan Medicinal Products (OMPs)

Between January 2018 and June 2023, 124 drugs received Orphan Drug Designation in Australia. Out of these, 90 were registered, and 48 were reimbursed. For more information about access to orphan drugs in Australia, click here. Australia does have an Early Access Programme/Expanded Access programme to help patients access medicinal products for rare conditions. Further information about the programme, Australia's Lifesaving Drug Progam, is available here. Australia is involved in formal efforts/initiatives with other countries to support access to medicines and therapies for rare diseases including the Rare Disease Moonshot and IRDiRC's Regulatory Scientific Committee.

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The Data Contributing Committee of Australia, which provided this 2025 data (correct as of the end of November 2025) in the context of the Resource on the State of the Art of Rare Disease Activities, is composed of the following individuals:

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