



# RESEARCH AND DATA

## VISION:

The best possible health and wellbeing outcomes for Australians living with a rare disease.

## CRITICAL ENABLERS:

Multi-stakeholder involvement and engagement.

Collaborative governance and leadership.

State, national and international partnerships.

High quality, comprehensive collection, and effective use, of rare disease data.

## HOW WE ACHIEVE PROGRESS:

- Further develop RVA as the national peak organisation for rare diseases.
- Build on existing strengths to formalise centres of excellence throughout Australia.

## WHY IS THIS IMPORTANT?

- In Australia, data for most rare diseases is not captured in either health information systems or registries and there is no coordinated strategy to collect, measure, build and translate data that does exist. Australia needs a national, coordinated, and systematic approach to the collection and use of rare diseases data. This will enable the accumulation of knowledge about rare diseases. For key decision-makers at all levels, greater knowledge can facilitate more responsive and appropriate services for people living with a rare disease.
- For many rare diseases, there are a number of barriers to effective research and no active research programs. Depending on the specific rare disease, research priorities can be different. Investment into all types of research is needed: from data collection and registries to basic research and clinical trials. People living with a rare disease need research into diagnostics (including genomics); new health technologies; precision/ personalised medicine; and care and support.
- Research into rare diseases must address existing gaps and the coordination of research projects must be prioritised. Improved policy settings, and national and international collaborations, will help to drive strong research and innovation for all rare diseases. Research into rare diseases needs to inform evidence-based policy across all systems, extending beyond health.
- For many people living with a rare disease, participation in a clinical trial may be the only way to access treatment.
- While research may not lead to better outcomes for people currently living with a rare disease, participating in research may drive change for future generations.





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## PRIORITIES

**Enable coordinated and collaborative data collection to facilitate the cumulative knowledge of rare diseases.**

- Health information systems identify and measure rare diseases and undiagnosed rare diseases.
- The Australian Institute of Health and Welfare (AIHW) re-establishes the Australian National Congenital Anomalies Register, including rare disease coding (Orphacodes). Accelerate, extend and nationalise rare disease coding already underway in Western Australia.
- Provide for rare disease codes in patient records, including an undiagnosed disease code. This code could raise a flag or alert to health professionals leading to appropriate care and support data collection for rare diseases, as well as service planning.
- Undertake broad epidemiological surveillance of rare diseases to support decision-makers to access the information they need.
- Build on existing newborn screening and congenital anomalies data collections, to further develop Australia's monitoring of rare diseases and undiagnosed rare diseases.
- Establish a dedicated Rare Disease Office within the AIHW that publishes periodic national reports on the epidemiology of rare diseases and undiagnosed rare diseases in Australia.
- Improve rare disease data collection and use.
- Develop a national approach to person-centred rare disease registries to support national standards, best practice and minimum data sets.

**Develop a national research strategy for rare diseases to drive all types of research for rare diseases.**

- Develop a national research strategy for rare diseases, to keep pace with genomic advancements, precision medicine and innovation.
- Undertake a national stakeholder consultation process to set agreed priorities for a national research strategy for rare diseases.
- Address evidence gaps in areas that are important to people living with a rare disease.
- Support collaborative research into rare diseases in Australia and internationally.
- Foster an environment conducive to clinical trials for rare diseases taking place in Australia.
- Investigate and promote options that enable Australians living with a rare disease to participate in Australian and international clinical trials.

**Ensure research into rare diseases is collaborative and person-centred.**

- Provide people living with a rare disease or an undiagnosed rare disease with the opportunity and support to participate in research.
- Enable researchers, funders and policy-makers to access the voice of people living with a rare disease in driving and delivering research into rare diseases.

**Translate research and innovation into clinical care; clinical care informs research and innovation.**

- Support partnerships between researchers and clinicians in research into rare diseases.
- Identify, leverage and enhance existing capability and infrastructure to ensure appropriate and experienced resourcing is available within clinical teams that deliver rare disease care.
- Identify and enhance existing capacity within clinical centres to ensure appropriate capability is available to support the operation of clinical trials.
- Support clinical teams to collect and input data, contributing to research and evidence-building.

