



RARE

Rare Disease Awareness,
Education, Support, and Training

Taking Action Together Toolkit

**Improving Health and Wellbeing Outcomes
for Australians Living with a Rare Disease**

Companion document:

**Who's who: the people and organisations involved in
the Australian rare disease sector**

18 June 2024

People and organisations involved in the Australian rare disease sector

People living with a rare disease

People living with a rare disease are the most important stakeholders in the sector. They include:

- People with a diagnosis of a rare disease
- People with a suspected rare disease (undiagnosed diseases are often rare diseases)
- Family members and carers of people living with a rare disease
- People at high risk of developing, or having children who develop, a rare disease

Many people living with a rare disease share their lived experience to drive change (advocacy). This is often more effective or amplified when individuals work together towards shared goals.

People living with a rare disease can collaborate effectively by:

- Working with their rare disease group/organisation or peak body
- Meeting with their local member of parliament
- Sharing their personal story through public speaking, focus groups or co-design processes to raise awareness of an unmet need and/or support service design
- Advocating alongside their rare disease group or organisation or peak body
- Participating in all aspects of research to improve clinical care and scientific knowledge
- Provide comments on the need for and impact of new technologies, therapies, and medicines to support new services

Consumer-led organisations, peak bodies, and charities

Rare disease-specific consumer-led organisations, peak bodies and charities are often the first point of contact for advocacy, awareness, direct care, and support for people living with a rare disease, and often fill gaps in the health system. These groups are also known as patient groups, patient advocacy groups, rare disease organisations (RDOs), and consumer health organisations.¹ They may be umbrella groups (covering several different rare diseases) or focused on one rare disease only.

RDOs may partner with other consumer-led organisations, peak bodies or charities with other subject-matter or community expertise, for example, disability, cultural and linguistic diversity or Indigenous Australian health. It is important to ensure a mutual understanding of challenges that impact their communities, and the most appropriate ways of responding to those challenges. RDOs can understand, capture and communicate the lived experience of those impacted by a rare disease, and understand the unique challenges and needs that rare diseases impose on people living with them. This makes, rare disease organisations and their leaders an essential part of the sector.

Many RDOs in Australia partner with RVA for greater impact and to align with their strategies. RVA has developed [Engaged, Ethical and Effective. A Guide for Rare Disease Organisation Leaders in Australia](#) to support rare disease organisations in providing essential support and leadership.

Consumer-led groups/organisations, peak bodies, and charities can collaborate effectively by:

- Leading the development of policy frameworks
- Developing relationships and consulting with scientific and medical advisors involved in rare disease clinical care and research
- Forming and contributing to partnerships with medical and health systems researchers
- Participating in the collaborative development of clinical practice guidelines and recommendations
- Organising and participating in events to raise awareness of rare disease
- Providing representative input to policy consultations to ensure the priorities and needs of people living with a rare disease are heard
- Providing leadership and support for their community, for example to inform and drive research, policy and legislation
- Meeting with government and policymakers
- Developing ethical relationships with pharmaceutical companies involved in producing rare disease medicines and therapies.

Health care professionals

Australia's health workforce is made up of more than 600,000 people employed to deliver health services.² The [Australian Commission on Safety and Quality in Healthcare](#) maintains standards including the [Australian Charter of Healthcare Rights](#), and the [National Safety and Quality Health Service Standards](#), which set a nationally consistent level of quality and safety that all Australians can expect from their health care system and health care providers.

Health care professionals can collaborate effectively with the rare disease community by:

- Committing to a person-centred approach by recognising and prioritising lived experience to inform the best possible care
- Following the clinical practice guidance in the [National Recommendations for Rare Disease Health Care](#) to improve clinical care
- Generating data and evidence to improve best practice care and treatment
- Collaborating with other advocates to work towards policy change and promotion of health literacy and health equity
- Working with consumer-led organisations, peak bodies and charities to develop frameworks, health services, and support legislative review
- Developing ethical relationships with pharmaceutical companies involved in producing rare disease medicines and therapies to improve treatment and outcomes

Academics, researchers, and research-funders

Health researchers are increasingly generating evidence with consumer-led groups/organisations, people living with a rare disease, Departments of Health (Federal and State), and non-governmental institutions. Authentic partnerships and collaborations between all stakeholders in the rare disease sector are essential when conducting and translating research that brings lasting value.

The [Australian Code for the Responsible Conduct of Research 2018](#) provides a foundation for building trust and credibility with communities when conducting research. Specific guidelines for genomic research have also been developed. The [Guidelines for Community Involvement in Genomic Research](#) were developed by Involve Australia, a partnership between patient support and advocacy groups, patients and carers, people who involve community in their research, interested members of the public, genomic researchers, and health care professionals.³

Universities, professional colleges and associations are responsible for health professional training and continuing education, to ensure health professionals have the knowledge and skills to care for people living with a rare disease and keep abreast of the latest advances in evidence-based practice and medical technologies.

Academics, researchers, and research funders can collaborate effectively with the rare disease community by:

- Ensuring people living with a rare disease are considered, and their data captured, when conducting medical, health and health systems research
- Inviting people living with a rare disease to be part of lived experience panels, sometimes referred to as stakeholder reference groups or consumer panels
- Evaluating the suitability of research based on community needs and preferences
- Forming partnerships to apply for and implement research grants, such as the [Rare Disease Awareness Education Support and Training \(RAREST\) Project](#), to improve awareness, knowledge, clinical care and support for people living with a rare disease
- Improving health professionals' knowledge of rare disease by developing educational resources for families and health professionals, for example Rare Disease 101 Australia¹

Pharmaceutical and biotechnology industry

The pharmaceutical industry in Australia comprises research-based, generic, consumer, and medical device companies. [Medicines Australia](#) provides information about the types of pharmaceutical and biotechnology companies and their roles. Medicines Australia's [Working Together Guide](#) outlines the types of collaborations that can take place when working with the pharmaceutical and biotechnology industry, and principles that should underpin effective collaboration, including:

1. Respect for independence
2. Achieving and maintaining public trust
3. Open communication
4. Confidentiality
5. Accountability
6. Successful collaborations that focus on health consumers and their carers

The pharmaceutical and biotechnology industry can collaborate effectively with the rare disease community by:

- Researching and developing new therapies, including funding and running clinical trials, to expand treatment options
- Navigating the regulatory process to bring new treatments to the market and onto the Pharmaceutical Benefits Scheme
- Joining RVA's [Round Table of Companies](#) to discuss and progress topics related to the development of treatments for rare diseases
- Sponsoring clinical trials that are initiated by health care providers or government-run organisations (for examples, hospitals) to trial new treatments
- Sponsoring conferences and/or educational events
- Conducting focus groups with people living with a rare disease to gain insights into their lived experience of rare disease and treatments

Governments and policy makers

Australian governments, at Federal and State levels, play a key role in both regulating and delivering health care for people living with a rare disease. There are several different sectors within the government, including Services Australia, the National Disability Insurance Scheme, and the Department of Health and Aged Care.

The Australian Government's [Public Service Academy](#) outlines the principles that policymakers consider for policy development and encourages wide involvement of all stakeholders, using evidence and data to inform decisions.

Governments and policymakers can collaborate effectively with the rare disease community for by:

- Funding, regulating, and delivering health care and targeted medical research into rare diseases to improve treatment, for example:
 - Funding the development of the [National Strategic Action Plan for Rare Diseases](#) (Action Plan)
 - Offering the clinical trials for rare diseases
- Being involved in rare disease events, for example the Rare Disease Day Parliamentary Event and the biennial National Rare Disease Summit to raise awareness of rare disease
- Appointing people with rare disease expertise to advisory groups to ensure the needs and priorities of people living with a rare disease are heard
- Developing and implementing policy aligned with the Action Plan to ensure it is responsive to rare disease

References

1. Medics 4 Rare Diseases. Rare Disease 101 - Australia (course) [internet]. Available from: <https://learn.m4rd.org/> [accessed 2023 Jul].
2. Australian Institute of Health and Welfare. Health workforce [internet]. Available from: <https://www.aihw.gov.au/reports/workforce/health-workforce> [accessed 2024 Apr].
3. Involve Australia. Guidelines for Community Involvement in Genomic Research [internet]. Available from: <https://www.australiangenomics.org.au/involve-australia-guidelines-for-community-involvement-in-genomic-research/> [accessed 2024 Apr].