National Recommendations for Rare Disease Health Care

- **A rare disease is a health condition that affects fewer than 1 in 2,000 people[1,2]**
- **There are over 7,000 different rare diseases,[1,2] affecting around 8% of Australians[3]**
- **This means that although each condition is rare, an estimated 2 million people live with rare disease in Australia**

The National Recommendations for Rare Disease Health Care (The Recommendations) are:

- designed to help health professionals provide quality care for people living with rare disease
- aligned with the Australian Government's National Strategic Action Plan for Rare Diseases
- based on national and international evidence and the input of people living with rare disease;
- disease agnostic and therefore also appropriate for people living without a diagnosis

Who might find the Recommendations helpful?

The Recommendations are aimed at:

- **All health professionals** – to provide better care for people living with rare disease

They may be useful for:

- **People living with rare disease** – to help in partnering with their health care team to plan the type of health care that works best for them
- **Policy makers** – to help shape health care policy relating to rare disease

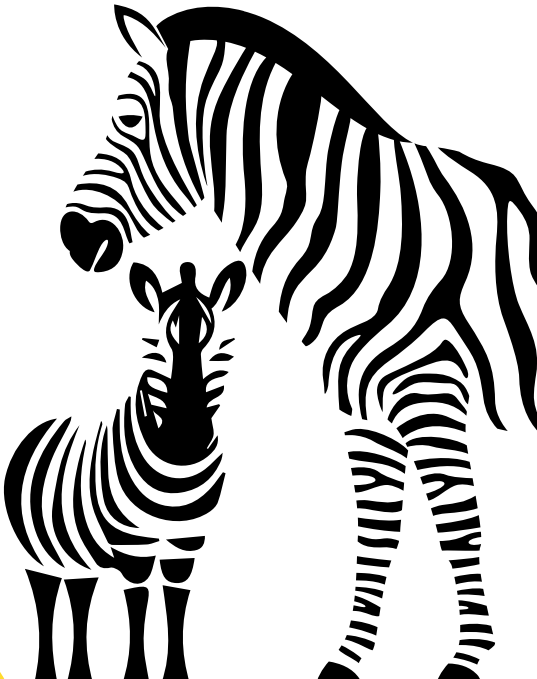
Who wrote the Recommendations?

The Recommendations were developed as part of the Rare Disease Awareness, Education, Support and Training (RArEST) Project, a collaboration between Rare Voices Australia, the University of New South Wales, Macquarie University, and the University of Western Australia, and funded by the Australian Government.

The Recommendations were written by a Working Group that included clinicians, academics, educators, and advocates with expertise in rare disease.

Input was received from:

- Patient advocacy and support groups
- People living with rare disease and their families/carers
- Doctors, nurses, and other health professionals



1. European Commission. Rare Diseases. EU research on rare diseases.
Available from: https://research-and-innovation.ec.europa.eu/research-area/health/rare-diseases_en [accessed Jan 2024];
2. EURORDIS - Rare Diseases Europe. What is a rare disease?
Available from: https://www.eurordis.org/information-support/what-is-a-rare-disease/ [accessed Feb 2024];
3. Elliot E, Zurynski Y. Aust Fam Physician 2015;44:630-33.

RArEST
Rare Disease Awareness, Education, Support, and Training

National Recommendations for Rare Disease Health Care

RECOMMENDATION 1

Deliver person-centred care that values diversity and lived experience as people living with rare disease are often experts in their own conditions and have changing, complex needs

1.1 Partner with people living with rare disease in diagnosis and care by sharing tailored information and education, facilitating shared decision making, and empowering them to be advocates and active participants in decision making

1.2 Recognise additional challenges faced by priority populations living with rare disease* and consider how to appropriately tailor care

1.3 Practice in a culturally safe manner with Aboriginal and Torres Strait Islander people

1.4 Practice in a culturally safe manner with people from culturally and linguistically diverse backgrounds

1.5 Link people living in regional, rural, and remote areas to resources which could reduce the time and expense to access care

RECOMMENDATION 2

Facilitate timely and accurate diagnosis as a rare disease diagnosis can lead to better clinical care, peer support, reproductive confidence, and access to services and clinical trials

2.1 Identify red flags that indicate someone may have a rare disease

2.2 Follow established protocols and pathways for timely and accurate diagnosis

2.3 Support people living with rare disease who remain on the diagnostic odyssey

RECOMMENDATION 3

Engage in two-way knowledge sharing with colleagues and Centres of Expertise as no one can be an expert in over 7,000 rare diseases

3.1 Consult with and refer people living with rare disease to rare disease experts and Centres of Expertise, including internationally

3.2 Facilitate systematic access to rare disease data collection, including access to rare disease registries and natural history studies

3.3 Align care with best practice guidelines and evidence

RECOMMENDATION 4

Respond to the inherent uncertainty of rare disease, by facilitating connections with rare disease and patient advocacy groups, research including clinical trials, and new therapies and technologies as fewer than 5% of rare diseases have a curative treatment but knowledge is rapidly expanding

4.1 Learn from, contribute to, and connect people living with rare disease to rare disease and patient advocacy groups

4.2 Find, participate in, and connect people living with rare disease to research, including clinical trials and research studies in rare diseases

4.3 Facilitate access to advanced therapies

*Priority populations are: people living with an undiagnosed rare disease; people with an increased chance of developing a rare disease or of having a child with a rare disease; Aboriginal and Torres Strait Islander people; people from culturally and linguistically diverse backgrounds; people experiencing socio-economic disadvantage; people living in regional, rural, and remote areas

RECOMMENDATION 5

Recognise and support mental health, social and emotional wellbeing needs as living with rare disease affects all facets of people's lives

5.1 Be aware of the mental health and wellbeing impacts of living with rare disease

5.2 Ask about mental health and wellbeing at all appointments and recommend appropriate resources, support, and referrals

5.3 Deliver strengths-based and trauma-informed care

RECOMMENDATION 6

Promote integrated and coordinated care across the lifespan as people living with rare disease require a wide range of health and support services

6.1 Advocate for and deliver an integrated and cross-sectoral model of care

6.2 Facilitate care coordination for each person living with rare disease

6.3 Facilitate successful transitions at key points, including to adult and end-of-life care

RECOMMENDATION 7

Facilitate health promotion, reproductive choices, and preventive measures for both genetic and non-genetic rare diseases as some rare diseases may be preventable or their impact reduced through these measures

7.1 Apply the principles of health promotion and prevention where relevant to rare diseases, including infectious diseases, cancers, and autoimmune disorders

7.2 Facilitate understanding of and access to testing and technologies to support reproductive confidence

RECOMMENDATION 8

Engage in relevant continuing education, reflective practice, and quality improvement as knowledgeable and skilled health professionals can greatly improve outcomes for people living with rare disease

8.1 Engage in continuing professional development on multiple aspects of rare disease care

8.2 Engage in reflective practice regarding your learning needs and care for people living with rare disease

8.3 Participate in quality improvement activities, including routinely collecting relevant data

Read the full Recommendations at:
rarevoices.org.au/national-recommendations
Read more about the RArEST Project at
rarevoices.org.au/rarest-project

Explore the full recommendations to find:

- Explanations for why each sub-recommendation is important
- Quotes from people living with rare disease
- Practical suggestions for progressing the sub-recommendations across a range of health care contexts
- Indicators of good practice, including suggested outcomes, evidence, and actions associated with implementing each of the recommendations
- Enablers of good practice – links to key resources, tools, and training

Next steps

The Recommendations are a first step towards developing national guidelines for rare disease care and support. This will require:

- Further research into rare disease and effective models of care
- Implementing system-level requirements
- Building on existing networks and collaborations for effective co-development
- Ongoing close partnerships with people living with rare disease

Want to know more?

To learn more about the RArEST Project, and to view and download the full Recommendations, visit Rare Voices Australia's website

- RArEST Project: https://rarevoices.org.au/rarest-project
- National Recommendations for Rare Disease Health Care: https://rarevoices.org.au/national-recommendations

To learn more about rare disease

- View the free, interactive Rare Disease 101 Australia e-learning modules for health professionals, developed by the RArEST team, which embed the lived experiences of people living with rare disease: https://learn.m4rd.org
- Visit the Rare Awareness Rare Education (RARE) Portal: https://rareportal.org.au
- View resources from Rare Disease Project ECHO®: https://rarevoices.org.au/rare-disease-project-echo
- Contact the RARE Helpline: https://rareportal.org.au/rare-helpline

For more information about the National Strategic Action Plan for Rare Diseases: https://www.health.gov.au/resources/publications/national-strategic-action-plan-for-rare-diseases

---

The National Recommendations for Rare Disease Health Care have been endorsed by

rare voices AUSTRALIA®

Human Genetics Society of Australasia

Australian Genomics

Macquarie University – Australian Institute of Health Innovation

ACCYPN – Australian College of Children & Young People's Nurses

acnp – Australian College of Nurse Practitioners – Endorsed

ACRRM

Australian Rheumatology Association

Rural Doctors Association of Australia

The National Recommendations for Rare Disease Health Care have been officially recognised as an Accepted Clinical Resource by The Royal Australian College of General Practitioners

RACGP – Accepted clinical resource

Read the full Recommendations at:
rarevoices.org.au/national-recommendations
Read more about the RArEST Project at
rarevoices.org.au/rarest-project

RArEST
Rare Disease Awareness, Education, Support, and Training