RArEST
Rare Disease Awareness,
Education, Support, and Training

National Recommendations for

Rare Disease Health Care

ACKNOWLEDGEMENT OF COUNTRY

We acknowledge the Traditional Owners of Country throughout Australia and recognise the continuing connection to lands, waters, and communities. We pay our respect to Aboriginal and Torres Strait Islander people, and to Elders both past and present.

DEVELOPMENT OF THE NATIONAL RECOMMENDATIONS FOR RARE DISEASE HEALTH CARE

Development of the National Recommendations for Rare Disease Health Care was led by the RArEST consortium which includes the University of New South Wales (NSW), Rare Voices Australia, University of Western Australia, and Macquarie University, with funding from the Australian Government.

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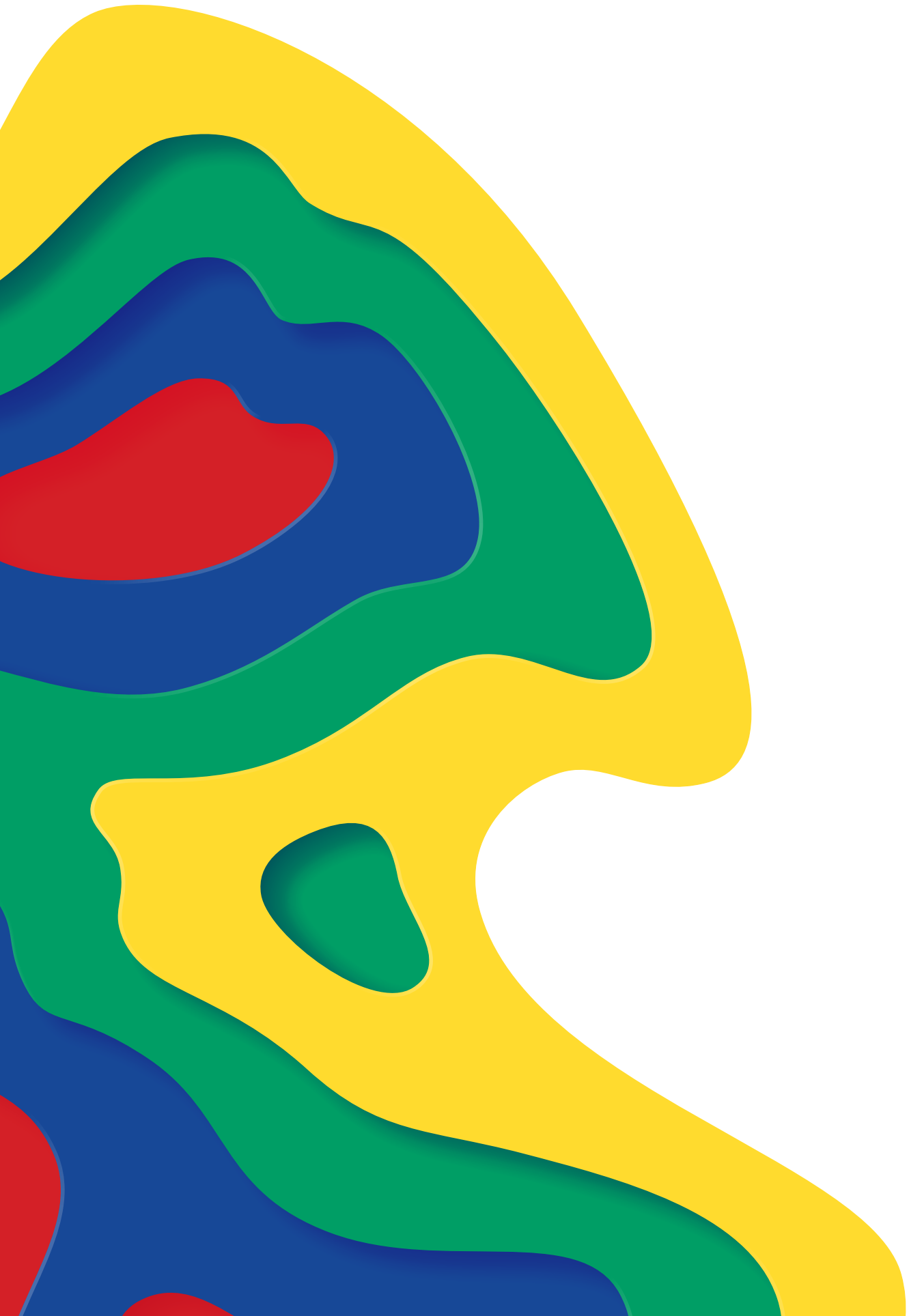

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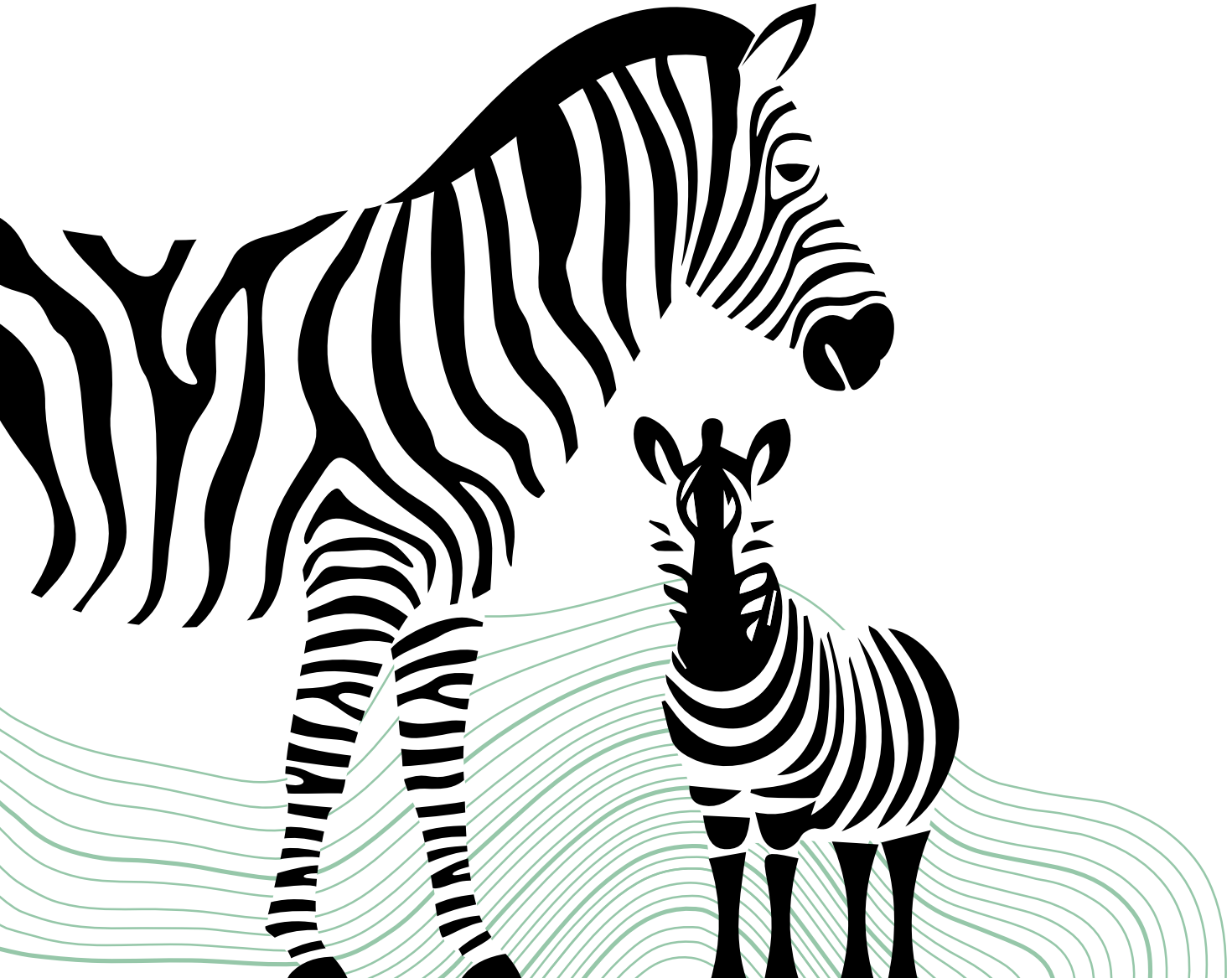
Executive Summary

An estimated **8% of Australians** (around 2 million people) **are living with rare disease**, so most health professionals will encounter people with diagnosed and undiagnosed rare diseases during their career.

The **eight recommendations** detailed in this document and summarised opposite can assist health professionals in Australia in providing high quality care for people living with rare and undiagnosed conditions, including their families and carers.

An online and navigable version is available at
www.rarevoices.org.au/national-recommendations



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.

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.

RECOMMENDATION 3

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

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.

RECOMMENDATION 5

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

RECOMMENDATION 6

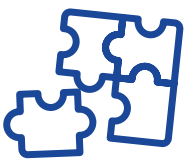
Promote integrated and coordinated care across the lifespan as people living with rare disease require a wide range of health and support services.

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.

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.



Introduction

CONTEXT

Any condition that affects fewer than 1 in 2,000 people is considered to be a rare disease.[1,2] Approximately 80% of rare diseases have a genetic basis,[3,4] but there are many other types of rare diseases, including cancers, infectious diseases and autoimmune diseases.[2] While individually rare, rare diseases are collectively common. There are over 7,000 known rare diseases, with that figure growing steadily as technologies evolve and knowledge improves.

An estimated 8% of Australians (around 2 million people) live with rare disease,[6] so most health professionals will encounter patients with diagnosed and undiagnosed rare diseases during their careers.

The sparsity of clinical knowledge about rare diseases, combined with their complexity, means appropriate training and support for health professionals are particularly important. Both are often lacking, making it harder for health professionals to care for people living with rare disease.[7]

Note: The impact of a rare disease extends beyond the individual person living with rare disease. Therefore, when this document refers to people living with rare disease, this includes their families, carers, and support people.

Most individual rare diseases do not have their own clinical guidelines. However, regardless of the rare disease, people have many common needs, strengths, and experiences that high quality rare disease care and support can address. These common needs, strengths, and experiences overlap with several goals for health care more broadly. However, a multifaceted and cross-sectoral approach is needed for people living with rare disease to bridge the health inequities they currently experience.

Small patient populations are one reason for these inequities. Often, limited information is available about a disease, including its cause, symptoms, diagnosis, progression, and treatment. There is also little perceived incentive for the public and private sectors to fund research, or for individual health professionals to gain specialist knowledge.[8] Resources can be pooled where commonalities exist, but rare diseases are highly heterogeneous. Additionally, there is also often limited awareness and understanding of most rare conditions in the wider community.[9]

As outlined in the Australian Charter of Healthcare Rights,[10] everyone deserves the same standard of care and consideration. Recognising the common challenges and opportunities for people living with rare disease and the health professionals dedicated to their care is a significant step towards this.

PURPOSE

The National Recommendations for Rare Disease Health Care (the Recommendations) provide recommendations for how health professionals in Australia can provide optimal care for people living with rare disease throughout their lifetime. Where available, the Recommendations highlight enabling resources, training, and frameworks for auditing to help support their implementation.

The Recommendations are:

- Aimed at health professionals;
- Aligned with the Australian Government's National Strategic Action Plan for Rare Diseases (Appendix 2.1);
- Based on national and international evidence and the input of people living with rare disease; and
- Disease agnostic and therefore also appropriate for people living without a diagnosis.

There are multiple barriers to delivering and receiving high quality care, which also need to be addressed. The responsibility for this does not rest on the shoulders of individual health professionals alone.[8] These barriers are listed below.

- **Time constraints:** Significant time pressures limit health professionals' availability for additional education, collaboration, research, and service improvements.
- **Lack of resources:** Inequitable or insufficient distribution of the resources needed to effectively manage rare diseases.
- **Systemic barriers:** Health systems and reimbursement structures that do not support the needs of people living with rare disease.
- **Limited populations:** The smaller population sizes of people living with rare disease make it difficult for many people to build a sufficient network of colleagues or patient organisations.

The Australian Government's National Strategic Action Plan for Rare Diseases (the Action Plan)[9] outlines the priorities, actions, and implementation steps required to achieve the best possible health and wellbeing outcomes for people living with rare disease. The Recommendations align with the Action Plan, which is centred around three interrelated pillars:

Figure 1: The three pillars of the National Strategic Action Plan for Rare Diseases.

How to use this document

A printable summary of the Recommendations and an infographic are available on Rare Voices Australia's website (rarevoices.org.au/national-recommendations), as a useful quick reference for people living with rare disease and health professionals alike.

Each recommendation provides an overview of why the recommendation is important, how to implement the recommendation, and suggests indicators of good practice categorised as:

ACTIONS	OUTCOMES	EVIDENCE
What health professionals can do	**What people living with rare disease experience**	**What evidence to monitor**

The Recommendations also embed quotes from people living with rare disease, to emphasise the importance of their lived experience in the co-development of these guidelines.

The following **Appendices** have been included after the recommendations to provide more context and help health professionals translate these recommendations into clinical practice. Hyperlinks throughout the document also link to relevant sections within this Appendix for ease of navigation.

APPENDIX	TITLE	CONTENT
Appendix 1	**Glossary and abbreviations**	Definition of terms and abbreviations used throughout this document
Appendix 2	**Enablers of good practice**	Key resourses and online courses
Appendix 3	**National and international sources**	Key national and international sources consulted when writing these recommendations
Appendix 4	**Primary preventive measures during pregnancy**	Preventive measures related to antenatal care
Appendix 5	**Development of the Recommendations**	Overview of the development process for these recommendations

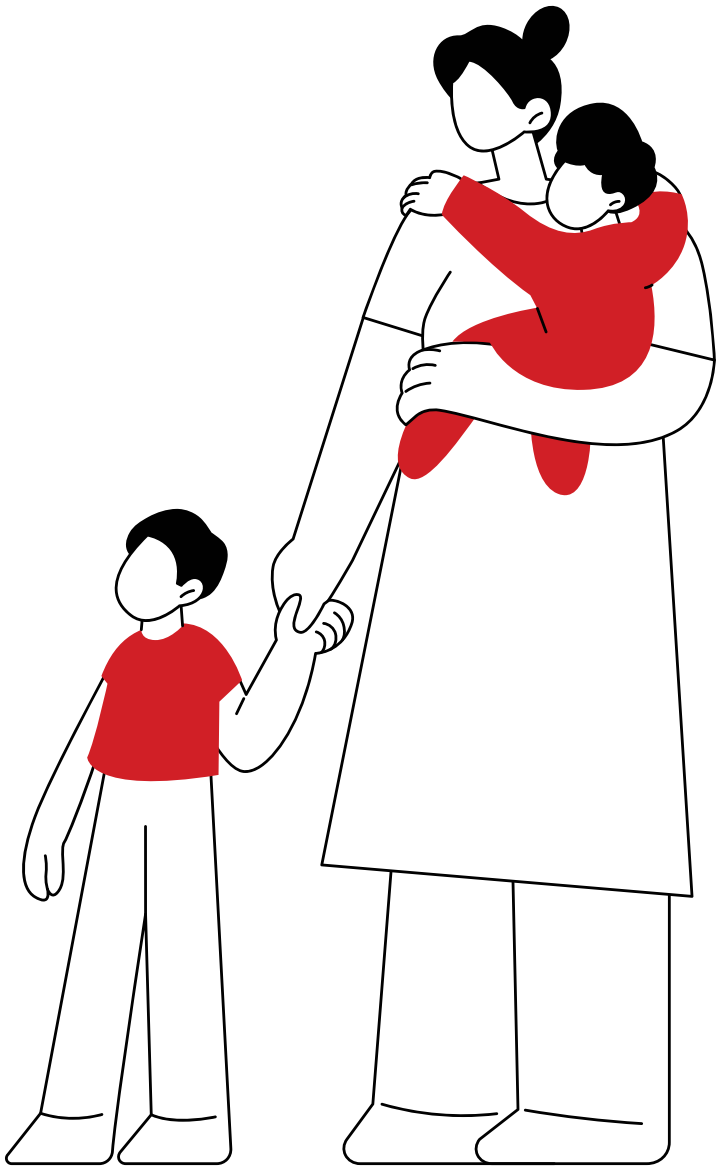
Next Steps

The Recommendations are a first step towards developing robust national guidelines for rare disease care. National guidelines for rare disease care must be evidence-based, with input from established rare disease networks and collaborations, including people living with rare disease, health professionals, and state and national health departments.

Development of the guidelines should be informed by further research in the following areas:

- The causes, prevention, treatment, and progression of rare diseases
- Models of care for people living with rare disease
- System level requirements for rare disease care.

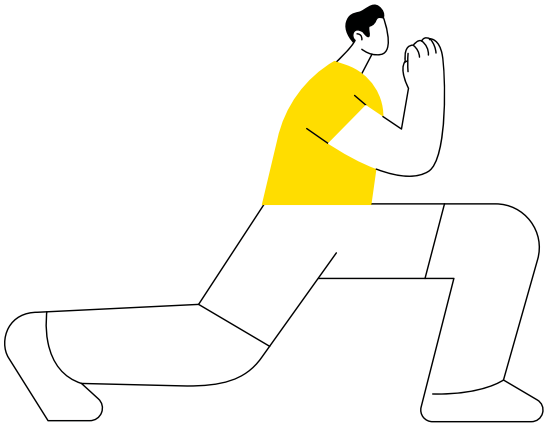
This work can be supported by harnessing and expanding existing networks and collaborations. This will help to ensure meaningful input from people living with rare disease, health professionals, and state and national health departments. The guidelines will need to be a living document to respond to the rapid changes and innovation in the rare disease sector.

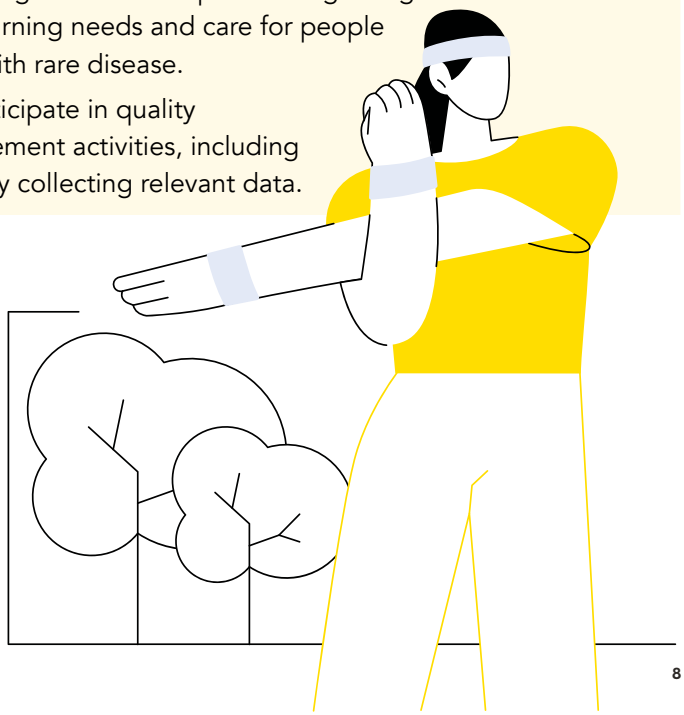


Summary of Recommendations

RECOMMENDATION	SUB-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 and services which could reduce the time and expense to access care.
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.
3. Engage in two-way knowledge sharing with colleagues and Centres of Expertise in and across jurisdictions 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.
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.

RECOMMENDATION	SUB-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.
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.
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.
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.





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Recommendations

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.

Recommendation 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.

Why this is important: Working in partnership with people living with rare disease has been shown to have benefits throughout the entire rare disease experience, including diagnosis, care, and research, for people living with rare disease as well as clinicians and researchers.[7]

> *"A specialist recently asked me what I expected from him in terms of my care needs. This was an approach that I had not experienced before from a health care professional. It made me feel as though he was interested and willing to listen, and that I was a part of and in control of my treatment program."*

People living with rare disease face uncertainty throughout their lives, which can cause additional challenges to health-related decision making.[9,12,13] Uncertainty is a feature of being undiagnosed. A diagnosis, while having benefits as outlined in Recommendation 2, can also create uncertainty.[13] Health professionals have not heard of most individual rare diseases and most rare diseases do not have formal management guidelines, optimal therapies, or easily accessible plain language information to refer to. This should be acknowledged by health professionals.

People living with rare disease often extensively research their symptoms or diagnosis to address these uncertainties. Often, people living with rare disease are more knowledgeable about their condition than the health professionals they engage with, given their personal investment and connections to rare disease communities and patient advocacy groups. Acknowledging that fact with people living with rare disease and being open to their suggestions, as well as assisting them by signposting reliable sources of information, can be helpful in guiding people's self-management skills, while also equipping them to be effective advocates.[7]

> *"One of the most helpful things was being consistently treated as part of our child's care team. They asked questions and listened, using this information to discuss decisions around treatment and management with our family. They supported efforts to become better informed by suggested research papers and other resources. It was really empowering, especially because you often feel so powerless in the face of a rare disease diagnosis."*

Being involved in making decisions for their own health care can result in higher levels of positive talk and self-esteem, and increased engagement and satisfaction, leading to people living with rare disease feeling empowered as active partners in their care.[14]

When taking a shared decision making approach, health professionals and people living with rare disease jointly participate in decisions, having discussed all options and their potential benefits and harms.[14] Shared decision making carefully considers each person's values, preferences, and circumstances, and thus is a central tenet of person-centred care. Shared decision making can be carried out in relation to screening, management, investigations, and research.[14]

> *"Rather than empowering myself and my family by providing us with the knowledge and tools to take ownership of my condition, I found that I was given disjointed and often incomplete information, and that I did not have in place the preventive measures to protect myself in an emergency."*

How to progress Recommendation 1.1: Partner with people living with rare disease in their care and support them in having active and informed partnerships with other health professionals. Empower them to be advocates and active participants in decision making. At the time of diagnosis and afterwards, proactively facilitate access to tailored, reliable, and understandable information about their rare disease. People can be supported both by providing credible information and equipping them to do effective research on their own. Explicitly address uncertainty with people living with rare disease, as this is a key step to building trust and expectation. These approaches all align with the preferences of people living with rare disease (see Figure 2).[15]

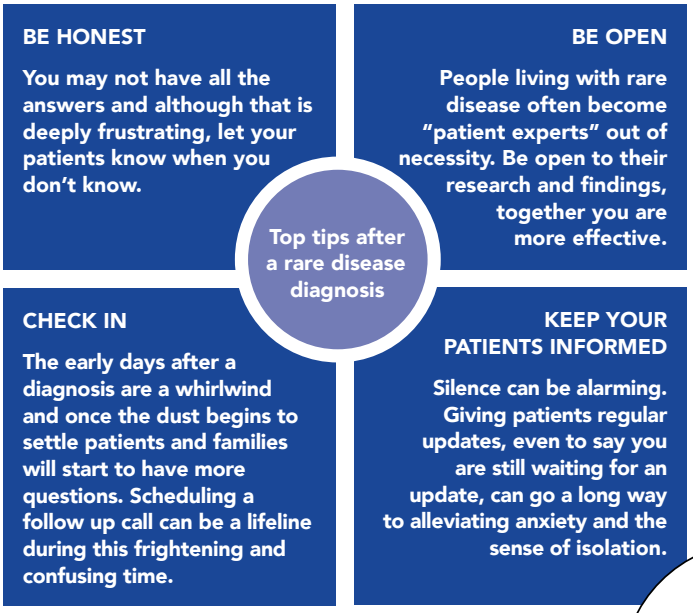
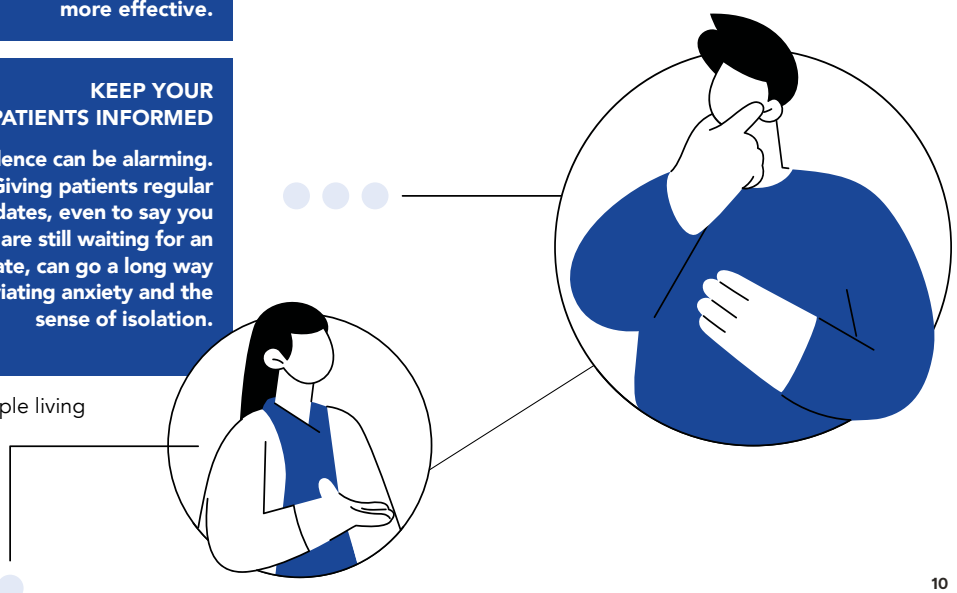


Figure 2: Tips for health professionals from people living with rare disease.[16]

The RARE Portal (Australian-based)[11] and the Genetic and Rare Diseases (GARD) Information Center (based in the United States of America [USA]) are key sources of reliable information.[17] The RARE Helpline is a new free service which aims to provide timely access to information and answer key questions that people living with a rare and complex disease often face.[18]
See Appendix 2.2 for more examples.

Patient advocacy groups (see Recommendation 4) can also be excellent sources of curated and tailored rare disease information and education.[19] Rare Voices Australia, the national peak body for Australians living with rare disease, has an A-Z Support Directory, which lists rare diseases and support groups/organisations where available.[20] Tips for assessing the credibility of support groups/organisations can be found in the A-Z Support Directory and in Engaged, Ethical and Effective: A Guide for Rare Disease Organisation Leaders in Australia on Rare Voices Australia's Online Education Portal (Appendix 2.3).[21]

Building health literacy can enable people living with rare disease to critically assess the array of rare disease information on the internet more broadly, which can be overwhelming and unreliable at times. An article by Healthgrades[22] discusses four key ways to do this (Appendix 2.4).



> *"Credible is the keyword there because I could look up anything online, I could put anything in Google. One thousand things pop up, but they all look legit. How do I know that's actually something that I should be following or finding out more about?"*

Encourage people living with rare disease to share their goals and give a clear plan of steps to take after the appointment. The Australian Commission on Safety and Quality in Health Care (ACSQHC)[23] have created tools to help clinicians in shared decision making with their patients (Appendix 2.5). Additionally, encourage and support people living with rare disease to use the Ask Share Know framework to improve the uptake of evidence-based practice and shared decision making (Appendix 2.5).[24]

It is important to recognise that rare diseases are commonly associated with a range of disabilities, including physical, sensory, and intellectual disabilities. People living with rare disease often report that health information is not presented in a way that meets their communication needs or that the setting is not accessible.[25,26]

Reasonable adjustments can help people with disabilities access and utilise health care. A reasonable adjustment is a change to an existing approach or process that is essential to ensure a person's access to a health service.[27] Making reasonable adjustments for people with disabilities means doing things differently to ensure people are not disadvantaged or harmed and is a means of avoiding direct and indirect discrimination, and meeting the National Safety and Quality Health Service Standards.[27-29]

> *"I reckon it's very important to be inclusive, because I think people can feel unincluded, especially people who've got the condition, and they kind of feel left out of the picture, but I think it's important for them to acknowledge the facts… in case they want to find out more about it."*

Examples of reasonable adjustments include:

- Using augmentative and assistive communication technologies
- Providing information in alternative formats, including Easy Read
- Allowing extra time
- Ensuring clinic rooms are accessible to those in a wheelchair.[30]

For example, assessment of capacity for informed consent for diagnostic genomic testing cannot be made unless reasonable adjustments have been made first.[31] Appendix 2.6 includes guides and toolkits for making reasonable adjustments.

It is also important to provide inclusive and individually appropriate rare disease health care for individuals who identify as gender and/or sexually diverse. For example, checking on people's preferences for personal pronouns and following up-to-date guidelines regarding family tree (pedigree) drawing which both respect individual differences and identities while maintaining clinically meaningful information (Appendix 2.5).

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

Why this is important: The Action Plan (Appendix 2.1) highlights that for the priority populations below, the common challenges faced by people living with rare disease are often exacerbated.

- **Australians living with an undiagnosed rare disease:** A large proportion of people suspected of having a rare disease remain on a diagnostic odyssey, which limits their access to appropriate care and supports. Recommendation 2 details positive actions health care professionals can take to facilitate a timely and accurate diagnosis.
- **Australians with an increased chance of developing a rare disease or of having a child with a rare disease:** See Recommendation 7 for actions that can be taken to promote screening choices and preventive options for rare diseases.

- **Aboriginal and Torres Strait Islander people:** Cultural safety is an approach that aims to address power imbalances between health professionals and patients to facilitate appropriate care, in an environment that is safe for everyone, including Aboriginal and Torres Strait Islander people.[32] Key principles of cultural safety, which are critical for ensuring that Aboriginal and Torres Strait Islander people living with rare disease have equitable access to high quality rare disease care, are provided in Recommendation 1.3.
- **People from culturally and linguistically diverse backgrounds:** Australia is a multicultural society, with 28% of Australians born overseas (representing almost every country in the world) and 23% speaking a language other than English at home.[33] Recommendation 1.4 details strategies for tailoring care for culturally and linguistically diverse people living with rare disease.
- **People experiencing socio-economic disadvantage:** Socio-economic disadvantage can significantly impact people's ability to access and navigate health and support services.[34] This can be compounded by the well documented financial impacts of living with rare disease. A key aspect of providing coordinated care for people living with rare disease is ensuring access to appropriate financial and disability supports as detailed in Recommendation 6.2.
- **People living in regional, rural, and remote areas:** People living further away from metropolitan centres have longer diagnostic odysseys and when they do travel for care, this can impact their education, employment, finances, and relationships, as partners and other children often stay at home. Recommendation 1.5 outlines how health professionals can reduce the barriers faced by people living with rare disease in regional, rural, and remote areas and thus improve wellbeing as well as support preventive measures, timely diagnosis and treatment, and reduced hospital presentations.

People from these priority populations often face more challenges in navigating health care systems and accessing appropriate care, including being diagnosed. There is often intersectionality across priority populations, creating further challenges.

For example, someone at an increased risk of developing a rare disease and living in a rural or remote area. The financial impacts of rare diseases are often acute for priority populations: significant out-of-pocket costs, including the cost of private appointments due to challenges accessing specialists in a timely manner in the public system, and additional costs of therapies, aids, and loss of income due to caregiver responsibilities.[35-37]

> *"I am one person and treat me as one person. Don't treat me only for my (rare) condition or for my mental health or based on my age."*

How to progress Recommendation 1.2: Priority populations require targeted activities to improve health and wellbeing outcomes so that no Australian living with rare disease is left behind. Monitoring the proportion of people living with rare disease from priority populations (for example, through practice audits) and their experiences with their care (for example, through patient reported outcome and experience measures and satisfaction surveys) can help tailor and improve the patient and family/carer experience.

If a particular gap is identified, a service that is more culturally safe, welcoming, inclusive, and accessible may be achieved through authentic co-design by partnering with people living with rare disease, peak bodies, and/or culturally appropriate advocacy services.

Learning from what has worked elsewhere is also helpful. There may be existing, evidence-based solutions to challenges of equitable access and care for other areas of health. For example, the Aboriginal Community Controlled Health services have been pioneering innovative models of care for decades (Appendix 2.7).[32]

It is also evident that the financial impacts of rare diseases are significant and can compound health inequalities further.[37] A truly intersectoral approach to rare disease care is ideal, whereby access to practical supports such as the National Disability Insurance Scheme (NDIS) and carer's supports are facilitated, as discussed in more detail in Recommendation 6.

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

Why this is important: Cultural safety is an approach that aims to address power imbalances between health professionals and their patients to facilitate appropriate care in an environment that is safe for patients and their families.[32] Aboriginal and Torres Strait Islander people experience unique challenges in accessing appropriate rare disease care. Factors include:[38]

- Historical and ongoing experiences of individual and institutional racism that have contributed to trauma and mistrust in the healthcare system
- A higher proportion of Aboriginal and Torres Strait Islander people live outside major metropolitan areas, reducing their access to many specialist services
- Many Aboriginal and Torres Strait Islander people utilise Indigenous languages as their primary language, and accessing trained interpreters and communicating clinical terms and concepts can be difficult
- Misalignment between biomedical approaches and Indigenous holistic understandings of health, which encompass physical, social, emotional, and spiritual wellbeing
- Significantly for rare diseases, there is a paucity of responsible rare disease research and appropriate genetic reference databases.

> *"To this day I still don't know if there is another Aboriginal person out there with what [son] has because I don't think they had that information."*

How to progress Recommendation 1.3: To provide culturally safe health care, health care professionals need to acknowledge and continually reflect on their own biases, positioning, attitudes, and assumptions, and how these may impact on the quality of care provided to patients. Culturally safe care considers questions of equity and access, integrates mental health, social and emotional wellbeing, and incorporates an understanding of the impacts of social determinants of health.[39]

Key aspects of culturally safe care for Aboriginal and Torres Strait Islander people living with rare disease include:

- Accessing culturally relevant information and resources for people living with rare disease. For example, information about rare diseases and genetic health care from the Indigenous Genomics Health Literacy Project (IG-HeLP)[40] and the universal medical translator Lyfe Languages rare disease module[41]
- Incorporating culturally safe approaches to clinician-person communication. For example, by applying the Clinical Yarning framework[42]
- Asking people if they would prefer co-consultation with culturally appropriate health advocates, and considering if rare disease care could be integrated into established and trusted services such as Aboriginal Community Controlled Health Organisations[32]
- Using resources such as Lyfe Languages to translate medical terminology into Indigenous languages to support culturally safe care by facilitating improved communication and understanding.

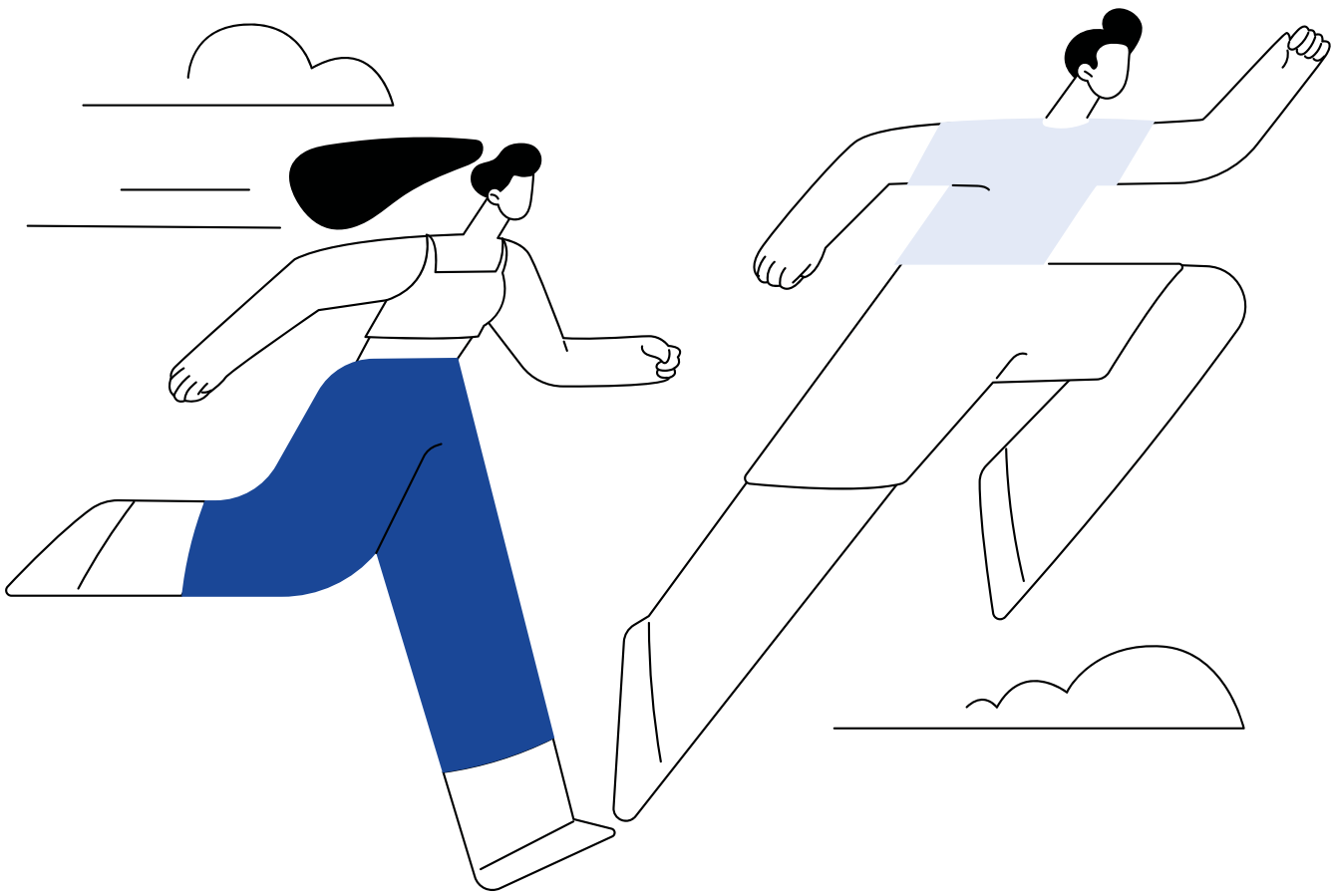
Relevant enablers of culturally safe care are provided in Appendix 2.7, including key rare disease resources tailored to Aboriginal and Torres Strait Islander communities. Culturally safe care is not only the responsibility of individual health professionals, but also of health institutions and structures. The Australian Institute of Health and Welfare has a monitoring framework for Cultural Safety in Health Care for Indigenous Australians (Appendix 2.7).[39]

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

Why this is important: There is no universal definition of cultural and linguistic diversity, which can relate to country of birth, ancestry, language spoken, or religious affiliation.[33] For people of culturally and linguistically diverse backgrounds, accessing appropriate health care services and support may be impacted by:

- Language barriers
- Different understandings of health and expectations of health services
- Limited knowledge of the Australian health system
- Prior negative experiences with health care, whether in Australia or internationally
- Limited understanding and/or accommodation of diverse needs by individual health professionals, organisations, and systems.[43]

How to progress Recommendation 1.4: Gather as much information as possible about an individual to understand and respond to their personal context and needs. Factors like language, migration experience, education, and socioeconomic status vary hugely across the culturally and linguistically diverse population. Offer and facilitate access to medical translators and interpreters. Additionally, refer and work alongside service providers that offer tailored services to culturally and linguistically diverse people, including for mental health, palliative care, and disability services (services signposted in Appendix 2.8).



1

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

Why this is important: People living with rare disease in regional, rural, and remote areas face many challenges in accessing rare disease diagnoses and care, including:

- Limited local health infrastructure
- Few or no local health professionals with experience in rare and undiagnosed diseases
- Higher cost of living pressures
- Lack of other support networks and services
- Logistical and financial barriers to travelling for care, including transport, accommodation, childcare, and leave from work.[44]

These challenges can result in a longer diagnostic odyssey for people living further away from metropolitan centres.[37] When people living in regional, rural, and remote areas travel for care, this can impact their education, employment, finances, and relationships, as partners and other children often stay at home. Linking individuals with available services and resources that can help overcome these barriers can therefore improve wellbeing and support preventive measures, timely diagnosis and treatment, and reduce hospital presentations.

> *"My neurological team… are the people who do my prescriptions and order my tests for me because I still can't find a GP in a regional location willing to share my rare journey."*

How to progress Recommendation 1.5: At a systemic level, improving equity of access requires formal links between health providers in regions, metropolitan areas, and Centres of Expertise (see Recommendation 3). These links can be used to develop robust partnerships between individuals, carers, families, communities, and the public and private health sectors.

There are also actions that can be taken on an individual or practice level, targeting the five dimensions of access: availability, accessibility, accommodation, affordability, and acceptability.[45] For example:

- Mobile clinics and outreach programs
- Targeted training for local health professionals and carers
- Evolving technology such as point-of-care testing, teleradiology, e-Health, telehealth, and videoconferencing to help more people access expert rare disease care (for example, from a rare disease Centre of Expertise)
- Connecting people living with rare disease to external support services and funding such as Angel Flight,[46] the NDIS[47] and appropriate carer support and funding, for example through the Carer Gateway[48]
- Seeking advice and support from the Australian College of Rural and Remote Medicine (ACRRM).

For resources to support people living in regional, rural, and remote areas see Appendix 2.9.

Summary of Recommendation 1

The following actions can facilitate a more tailored and person-centred approach:

- Scheduling longer appointment times, so that people can discuss the aspects of their life that are impacting, or being impacted by, their rare condition
- Allowing significant people in a patient's life to attend appointments, as is culturally appropriate
- Offering flexible appointment formats such as telehealth and multidisciplinary clinics
- Tailoring communication styles to suit the socio-cultural preferences of the person and their family
- Being part of a cross-sector team with links between health, disability, education, and employment
- Building capacity for more community-based care.

Indicators of good practice

Actions

- Sharing decision making to develop a holistic care plan
- Valuing the input and expertise of people living with rare disease based on their experiences
- Responding appropriately and communicating effectively in situations involving clinical uncertainty
- Sourcing and providing rare disease information resources that are appropriate for each person's health literacy
- Being flexible in models of care
- Embedding cultural safety principles and reasonable adjustments in practice

Outcomes

- High scores in Patient-Reported Experience Measures (PREMS), such as those included in the Australian Hospital Patient Experience Question Set from the ACSQH,[49] which can be adapted to most settings
- People living with rare disease report that they have the desired level of involvement in decision making concerning their treatment and care
- Aboriginal and Torres Strait Islander, culturally and linguistically diverse, and disabled people living with rare disease report a positive health experience without discrimination

Evidence

- Rare disease-specific training, cultural safety and disability training that exposes health professionals to the voices of lived experience and co-designed best practice frameworks
- Adoption of flexible consultation models, accommodating families, translators, and culturally sensitive communication methods, such as yarning
- Accurate, appropriate, and culturally sensitive information provided to people living with rare disease
- Use of shared decision making tools
- Incorporating reasonable adjustments into all models of care

For useful tools and supports to help implement Recommendation 1, see Appendix 2.1-2.9.

2

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.

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

Why this is important: Around 2 million Australians have a rare disease and diagnostic delay is a major challenge for this population, with:

- Many people waiting an average of 5–7 years for a diagnosis, if one is ever made
- 4 out of 10 people living with rare disease in Australia reporting seeing more than six doctors and having at least one misdiagnosis.[50]

> *"My diagnostic odyssey took 56 years. I have multiple organ damage and left the job I loved due to illness. I receive no government support and my medical costs are high. If diagnosed and treated earlier, I would still be working, not so ill and having a much better quality of life!"*

How to progress Recommendation 2.1: Consider rare disease when diagnosing new and existing patients. This can be done by taking a family history, carefully reflecting on people's key clinical signs and symptoms, and listening openly to their concerns and suggestions.[9,51] It is important to recognise that rare diseases are often complex and involve multiple organ symptoms. Carefully review a person's current and previous medical history and be open to the possibility of connections between symptoms that at first may seem unrelated.

The 'Family GENES' mnemonic was developed by the Genetics in Primary Care Faculty Development Initiative (based in the USA) to help clinicians recognise when a person might have a genetic condition.[52] This tool can be adapted to the rare disease setting (Figure 3). It helps signpost that a referral to a diagnostic centre should be considered (Appendix 2.10a). It especially highlights the importance of being open to the possibility of connections between at first seemingly unrelated issues.

Family history
Multiple affected siblings or individuals in multiple generations. Remember that lack of a family history does NOT rule out genetic causes.

G **Group of congenital anomalies**
Common anatomic variations are common; but two or more anomalies are much more likely to indicate the presence of a syndrome with genetic implications.

E **Extreme or exceptional presentation of common conditions**
Early onset cardiovascular disease, cancer, or renal failure. Unusually severe reaction to infectious or metabolic stress. Recurrent miscarriage. Bilateral primary cancers in paired organs, multiple primary cancers of different tissues.

N **Neurodevelopmental delay or degeneration**
Developmental delay in the paediatric age group carries a very high risk for genetic disorders. Developmental regression in children or early onset dementia in adults should similarly raise suspicion for genetic aetiologies.

E **Extreme or exceptional pathology**
Unusual tissue history, such as pheochromocytoma, acoustic neuroma, medullary thyroid cancer, multiple colon polyps, plexiform neurofibromas, multiple exostoses, most paediatric malignancies.

S **Surprising laboratory values**
Markedly abnormal pathology results.

Other red flags for rare disease
Multiple visits to different specialists without a diagnosis. A long 'diagnostic odyssey'.

Figure 3: Family GENES tool. Adapted from the Genetics in Primary Care Faculty Development Initiative.[52]

Recommendation 2.2: Follow established protocols and pathways for timely and accurate diagnosis.

Why this is important: International experts recommend that all people with a suspected rare disease should receive a diagnosis within 1 year if they have a condition known in the medical literature,[53] to facilitate access to optimal management, treatments, supports, and if appropriate, genetic counselling.

> *"I was ill for 20 years before I was offered genetic testing–I didn't know this was part of the protocol. If I know why certain tests are being done (and the frequency of them) or why they have stopped looking for answers, I think I would feel less confused about my disease management."*

How to progress Recommendation 2.2: Instigate or refer people suspected of having a rare disease to appropriate diagnostic testing. This may be to the relevant specialty if symptoms are isolated to one body system, or to a rare disease Centre of Expertise or clinical genetics service if they are multisystem disorders. In time, information on Centres of Expertise will be available on the RARE Portal (Appendix 2.2).[11] A list of clinical genetics and metabolic services and up-to-date information on genetic tests and the informed consent process is provided by the Centre for Genetics Education (Appendix 2.2).[54] It is ideal to provide information about waiting times for such services, as these can be significant and often currently exceed 1 year.

Search engines such as PubMed[55] and Online Mendelian Inheritance in Man (OMIM)[56] may be helpful in constructing a differential diagnosis to help guide appropriate diagnostic testing or referrals. A growing number of search engines (Appendix 2.10b) incorporating artificial intelligence can be used to help diagnose rare diseases, such as the Orphanet Clinical Signs and Symptoms App,[57] Find Zebra,[58] and PubCaseFinder.[59] These list potential underlying conditions and/or genetic causes based on the phenotypic information entered (that is, observable characteristics).[60-62]

Diagnostic tests will vary based on a person's symptoms. Given that approximately 80% of rare diseases have a genetic basis, familiarity with genetic and genomic testing processes, which Medicare item numbers are available to fund genomic testing for what indications, and the informed consent processes is needed, with many educational resources on these topics provided by the Centre for Genetics Education (see Appendix 2.2).[54,63] Lesson 5 of the Rare Disease 101 Australia e-learning module (see Appendix 2.2) covers diagnosis and diagnostic tools, including examples of using the Family GENES mnemonic and rare disease search engines.[64]

Genetic and genomic testing is becoming more widely available in Australia, and is funded or subsidised under Medicare in several clinical situations. Diagnostic yield varies and can improve over time as new genes are discovered and technology improves. Re-testing or re-analysis of previous genetic or genomic testing results is an option that can be discussed with people who remain without a diagnosis. Moreover, as discussed in more detail below, referral to an undiagnosed disease program can be considered.

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

Why this is important: It is estimated that a diagnosis explaining all symptoms cannot be made for at least half the people assessed by health professionals as being likely to have a rare disease. This may be because the causative condition cannot be identified, due to challenges in recognising the full spectrum of clinical features associated with individual rare diseases, or because the rare disease is not yet discovered.[65]

People living with an undiagnosed rare disease cannot be provided with an accurate prognosis, and have little access to evidence-based treatment, Centres of Expertise, clinical trials, or patient advocacy groups. Moreover, a lack of diagnosis can be a roadblock to obtaining adequate funding from the NDIS.[9]

Remaining on the diagnostic odyssey can also have significant impacts on people's mental health and wellbeing. People living with rare disease may feel uncertainty, stigma, and frustration, especially if they perceive health professionals do not believe they have a real condition.[50,66,67]

RECOMMENDATION 2

2

> *"We have had to fight the entire length of our journey to ensure our daughter was diagnosed in a reasonable time frame, follow up and chase care and information. If you do not have the ability to do this, you get left behind and lost in a complex world of rare care."*

How to progress Recommendation 2.3: Identify which people in your practice have a suspected but undiagnosed rare disease and implement the following five steps.

1. **Deliver person-centred symptom-based care.** Even without an aetiological diagnosis, it is possible to practice integrated care based on a person's symptoms and their priorities for their own health care (see Recommendation 1). Where possible, people living with rare disease can be offered referral to Centres of Expertise for the group of conditions most closely aligned with their symptoms.
2. **Screen for mental health and wellbeing and facilitate appropriate supports.** Due to the uncertainty inherent in living without a diagnosis, the mental health and wellbeing of people living with rare disease should be carefully considered in each appointment. Those who are undiagnosed may have a greater need for referral to mental health services and care navigation than people with a diagnosis (Recommendation 5).[68]
3. **Facilitate practical and financial supports.** Practical supports such as the NDIS can still be accessed by people living with rare disease, and health professionals can play an important role by providing letters of support that delineate the impact of a person's symptoms. It is also important to check whether people with rare diseases, including family members and carers, are accessing the full range of financial support benefits that they are entitled to. The Disability Support Information section on the RARE portal includes information to help navigate current disability, carer and aged care supports and financial entitlements (Recommendation 6).[69]
4. **Connect to support groups for undiagnosed families.** Providing information on specific support organisations such as Syndromes Without A Name (SWAN) Australia[70] and, internationally, the Wilhelm Foundation[71] can help people with an undiagnosed rare disease access further supports (Recommendation 4; Appendix 2.11).
5. **Offer connection to undiagnosed disease programs.** Lastly, it is important to discuss a referral back to a clinical genetics service to review previous testing and see if more up-to-date testing may be helpful if an underlying genetic cause for a rare disease is considered. Genetics services can also help families who want to explore research diagnostics including undiagnosed disease programs (UDP). UDPs like the Australian Undiagnosed Disease Network (UDN-Aus)[72] and the Undiagnosed Diseases Network International (UDNI)[73] offer opportunities for clinicians to enrol undiagnosed families in cutting-edge clinical research (Appendix 2.12). This research boosts diagnostic yields by applying genomic reanalysis, international case 'matchmaking' and multi-omic tools such as RNA sequencing.[74] Some states also have UDPs that run as clinical services and partner with the UDN-Aus research network (Appendix 2.12).

Indicators of good practice

Actions

- Identifying people diagnosed with a rare disease, and accounting for the impacts of rare disease when formulating care plans
- Recognising signs and symptoms that might indicate a potential rare or genetic disease
- Coordinating and following-up with referrals to facilitate a timely diagnosis
- Providing ongoing support to people with an undiagnosed disease

Outcomes

- People living with rare disease report that their provider attentively listened to and addressed their concerns
- Thorough investigations and follow-up of potential diagnoses

Evidence

- Accurate coding of rare disease in patient records
- Identifying overlooked red flags in complex or undiagnosed cases
- A diagnosis or a referral to the undiagnosed disease pathway within 1 year of presentation
- Referrals to clinical genetics services if an underlying genetic rare disease is suspected
- Referrals to Centres of Expertise for that symptom group or group of conditions
- Health professional training in genetics, genomics and rare disease awareness and diagnosis

For useful tools and supports to help implement Recommendation 2, see Appendix 2.1, 2.2, 2.10-2.12.

3

RECOMMENDATION 3.

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

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

Why this is important: The existence of over 7,000 rare diseases and small patient numbers per rare disease creates a unique need for global cooperation and infrastructure to ensure the best outcomes for people living with rare disease. This information sharing can occur between individuals, and across large institutions, both nationally and internationally.

A Centre of Expertise provides expert structures for the management and care of people living with rare disease,[75] including access to multidisciplinary expertise in a single location. Although there is currently no consensus definition of a rare disease Centre of Expertise, commonly recognised features include:

- Multidisciplinary expertise in the diagnosis and management of rare diseases
- Provision of coordinated care
- Involvement in research, collaborations, and data sharing
- Involvement in education
- Engagement with people living with rare disease to continually refine and improve the model of care.

A centre may specialise in one or a group of related rare diseases. Centres of Expertise can facilitate improved care, a shorter diagnostic journey, and easier care coordination. This is supported by a person-centred model to empower people living with rare disease in realising their right to the highest attainable standard of physical and mental health.

Importantly, Centres of Expertise can also assist external health professionals and people living with rare disease by:

- Supporting high quality care, including timely diagnosis, appropriate treatments, and follow-up
- Sharing and connecting to specialised knowledge
- Facilitating peer support
- Providing access to research opportunities, resources, data, and expertise.

> *"We desperately need help with services other than medical so having the support of [a rare disease Centre of Expertise] will be amazing for us because no one knows much about the condition. I am finding it hard to get any support."*

How to progress Recommendation 3.1: Identify Centres of Expertise in rare diseases through online portals, including the RARE Portal in Australia, Orphanet in Europe, the Genetic and Rare Diseases Information Center (GARD) in the USA, or from patient advocacy groups (see Appendix 2.2, 2.3). Reach out to these experts and organisations for advice and discuss appropriate referrals, in line with the preferences of people living with rare disease.

Individual rare disease experts, in the absence of a Centre of Expertise, can be identified by checking the author list of up-to-date management guidelines or reviews (such as GeneReviews articles; see Appendix 2.2) or by joining a community of clinical learning practice (see Appendix 2.2, 2.13 and Recommendation 8.1). For many rare conditions, the relevant expert may be overseas, and identified through an international patient advocacy group (for which they may be a medical or scientific advisor) or key publications in the medical literature such as GeneReviews articles. Most experts are happy to be contacted for general advice and to discuss research opportunities.

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

Why this is important: There is currently no nationally coordinated effort to collect rare disease data in Australia.[9] Data for most rare diseases is not captured in either health information systems or registries and there is a need for a national strategy to collect, measure, build, and translate data.

A rare disease registry is a database set up to collect, store, retrieve, analyse, and disseminate information on people with a specific condition.[76,77] These collections of standardised information are used for a variety of specific purposes.[78] Natural history studies are pre-planned observational studies designed to track the course of a disease over time and identify demographic, genetic, environmental, and other variables that correlate with the disease's evolution and outcomes.[79]

Registries and natural history studies may have various aims, including but not limited to:[80]

- Revealing information about incidence and prevalence
- Improving understanding of how a condition evolves over time
- Describing epidemiology
- Facilitating public reporting and knowledge building about the impact of rare diseases
- Revealing differences in care practices and processes and the impact of both on people living with rare disease
- Identifying best practice and target areas for future improvement
- Linking participants to information on relevant clinical studies and trials
- Promoting connections between and networks of stakeholders
- Developing, evaluating, or improving therapies and other care interventions, such as providing evidence for management decisions.

Registries are especially critical for rare diseases with low prevalence and propensity for variation in treatment and outcomes.[77] They can:

- Provide health professionals and researchers with first-hand information about people with certain rare diseases, both individually and as a group, and over time
- Be cost-effective instruments to support clinical trials and translational research to improve quality of care, quality of life, and survival
- Act as support structures for people living with rare disease.

> *"We're always finding brick wall after brick wall... having to justify ourselves over and over again. A rare disease centre educates all departments in the right way to treat children, so we don't go through all that stress. A centre that helps clinical trials to be more cost effective and available in Australia, is important. A centre that looks after you."*

A key element of rare disease registries is interoperability with other registries through use of common standards for coding (classifying) rare diseases and symptoms, and for data storage and transfer. A further key element is the involvement of people living with rare disease in registry governance, adaptability, and sustainability. [81]

The Recommendations for a National Approach to Rare Disease Data: Findings from an Audit of Australian Rare Disease Registries is a key document, led by Rare Voices Australia, that reports the findings from a recent audit of rare disease registries in Australia. It lists all Australian rare disease registries, how they are funded, what they do with their data, their impact on patient outcomes, and strategic recommendations and associated implementation priorities for a national approach to rare disease data (Appendix 2.14).[81]

How to progress Recommendation 3.2: Educate people living with rare disease on the benefits of engaging with registries and natural history studies early in their health care journey. Facilitate enrolment by supplying appropriate information and resources to enable informed consent.

Information on registries and natural history studies can be found by contacting Centres of Expertise, national peak bodies, and research or advocacy groups. Considerable work has already been undertaken to further develop a growing understanding of registries and the national rare disease picture. This includes work on the National Alliance of Rare Disease Registries and the Recommendations for a National Approach to Rare Disease Data which can guide best practice (Appendix 2.14).[81]

Health professionals who diagnose a child with a rare disease can report the diagnosis to the Australian Paediatric Surveillance Unit at Kids Research[82] and the International Network of Paediatric Surveillance Units (see Appendix 2.14).[83] The RARE Portal (Appendix 2.2) contains links to rare disease registries, where available.[11]

Health professionals can also increase the visibility of rare diseases by entering, or advocating for the entry of, rare disease information and codes in electronic medical record data collection systems. Coding systems include the Systematized Nomenclature of Medicine Clinical Terms (SNOMED CT), ORPHAcodes, and the World Health Organization's International Classification of Diseases (ICD) system (Appendix 2.15).[84]

Recommendation 3.3: Align care with best practice guidelines and evidence.

Why this is important: Aligning with best practice can shorten the time to diagnosis and improve the quality of care both before and after diagnosis. The Action Plan recommends the use and development of clinical practice guidelines to support health professionals in identifying and diagnosing possible rare disease, as well as providing ongoing management.[9]

> *"Even when a patient is still on a diagnostic pathway, or there is no specific treatment or cure, there is always management and validation. These are two things that can make a profound difference to a person's quality of life. And every doctor is capable of giving it."*

How to progress Recommendation 3.3: In Australia, guidelines on some specific diseases can be found through Centres of Expertise, national peak bodies, research or advocacy groups, and key summary publications in the medical literature, searchable using medical databases such as PubMed. The RARE Portal links to both national and international guidelines, current evidence and best practice for a growing number of individual rare diseases, where available (Appendix 2.2).[11]

If no guidelines exist in Australia, international versions can be accessed. For example, individual disease pages on Orphanet[2] link to clinical practice guidelines if they are available, and guidelines can be found for over 800 rare genetic conditions on the international point-of-care resource GeneReviews (Appendix 2.2).[85]

Additionally, education and capacity building sessions for health professionals are key to building awareness leading to successful implementation of existing evidence and guidelines (Appendix 2.13). Health professionals can participate in guideline development, for example, by engaging with rare disease and patient advocacy groups (Recommendation 4).

Indicators of good practice	
Actions	▪ Knowing where to find expert advice for people living with rare disease ▪ Facilitating access to expert care ▪ Participating in the creation and dissemination of new knowledge and practices applicable to rare disease
Outcomes	▪ People living with rare disease feel they have access to the best possible expertise related to their rare disease
Evidence	▪ Involvement of Australian or international rare disease experts ▪ Alignment of care plans with best practice guidelines, where available ▪ Interactions with rare disease and patient advocacy groups and sharing of relevant information with people living with rare disease ▪ Use of databases of rare disease expertise ▪ Referrals to Centres of Expertise ▪ Connection of patient data to clinical registries related to rare disease

For useful tools and supports to help implement Recommendation 3, see Appendix 2.2, 2.3, 2.13-2.15.

4

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.

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

Why this is important: Rare disease and patient advocacy groups, also called patient support organisations or consumer health organisations, play a critical role in supporting people living with rare disease. These organisations:

- Connect people living with a particular rare disease or umbrella group of rare diseases with one another for practical and emotional support
- Are a credible source of information about a rare disease or group of diseases
- Can drive research and advocate for better access to care, support, and treatments.

> *"Seeing others who have felt our struggles, who know our pain and share our hopes for better care gives us a sense of comfort and strength that is hard to put into words."*

In this rapidly changing landscape, it is difficult for health professionals to keep up with the available information on management, therapeutics, research, and clinical trial opportunities. Patient advocacy groups are often important sources of expertise about conditions or groups of conditions. They can provide health professionals and people living with rare disease with deep, person-centred knowledge that can improve care and outcomes.[19] These groups will often know of local and international research opportunities, including the availability of disease-specific clinical quality registries (Recommendation 3.2).

Connections between health professionals and patient advocacy groups are also mutually beneficial. Health professionals can contribute scientific and medical advice that strengthens the rigour, reliability, and credibility of patient advocacy groups. This in turn means they are better able to support people living with rare disease and other health professionals. It also increases the visibility of rare diseases and the growth of clinical networks with relevant experience.

How to progress Recommendation 4.1: Identify patient advocacy groups that are relevant to your patients. Rare Voices Australia is the national peak body for Australians living with rare disease and advocates for policy as well as health, disability, and other systems for people with a rare disease. It hosts the national RARE helpline (Appendix 2.2).[18]

There are several directories of patient support and advocacy groups, such as Rare Voices Australia's A-Z Support Directory (Appendix 2.3).[20] People living with rare disease have asked for introduction and connections to these groups as early as possible after a diagnosis. These groups can be very helpful for the person with the rare disease diagnosis as well as their family and support people.

If no group exists in Australia for a particular rare disease, international groups may be able to provide support; these may be identified through international portals such as Orphanet or GARD (Appendix 2.2). In some cases, people living with rare disease may be interested in setting up a patient advocacy group for their condition. If so, Engaged, Ethical and Effective: A Guide for Rare Disease Organisation Leaders in Australia (Rare Voices Australia) is a good starting point.[21]

Engaging with these groups can take many forms, including:

- Referring people living with rare disease to them
- Utilising information from their websites
- Joining a scientific or medial advisory board
- Reviewing the accuracy of patient-facing information.

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

Why this is important: Limited knowledge and data are common features of rare diseases, making research critical to the current and future care of people living with rare disease. Reflecting this, Research and Data comprise one of three key Pillars in the Action Plan (Appendix 2.1).[9]

Rare disease research can include:

- Clinical trials, including for advanced therapy medicinal products (advanced therapies), which are medicines for human use that are based on genes, tissues, or cells (see also Recommendation 4.3)
- Undiagnosed disease programs (see Recommendation 2.3; Appendix 2.12)
- Non-therapeutic studies (such as into quality of life or service utilisation).

For many people, clinical trials are the only way to access treatment. Research increases understanding of a condition, informs new treatments, and benchmarks outcomes and service provision.

> *"As a parent of a child with rare disease, you want to know WHAT you can do to help in an impossible situation. Providing information and access to things such as clinical trials is important early on. No matter what the long-term outcome, a lot of peace can come from knowing you did everything you could as a parent."*

How to progress Recommendation 4.2: Health professionals have a key role to play as gatekeepers to research and clinical trials for people living with rare disease. Best practice in rare disease care can only be achieved by embedding data collection and connection to research and clinical trials into routine practice.

The first step is identifying opportunities for clinical trials, including through Centres of Expertise, patient advocacy groups and databases like Australian Clinical Trials[86] and ClinicalTrials.gov (international) (Appendix 2.16).[87] Health professionals can then link the family with the clinical trial team. Often, rare disease registries (see Recommendation 3.2) are set up with a key aim of linking rare disease cohorts to specific research and clinical trial opportunities, and thus are another reason to discuss and facilitate access to such registries for people living with rare disease.

Health professionals also have an important role to play in facilitating shared decision making around research, clinical trial participation, and clinical quality registries. Discussions may include uncertainty of treatment outcomes, risks and benefits in comparison to available treatments or no action, and explaining inclusion and exclusion criteria. Resources to inform such discussions are provided in Appendix 2.16 and 2.17.

A challenging aspect of connecting people living with rare disease to opportunities is simultaneously educating them about the importance of research and managing expectations. Health professionals should be prepared to discuss people's disappointment and frustration when a clinical trial is not the right fit or is unavailable in Australia, or a treatment is not effective.

It is important to ensure sufficient time and access to expert information so that people do not feel rushed into making quick decisions, especially as access to some clinical trials may exclude them from future trials or therapies.

Undiagnosed rare disease programs are typically led by researchers and academics. Health professionals have an important role in referring patients to these programs, usually via clinical genetics services (see Recommendation 2.3). Timely referral to undiagnosed rare disease programs is vital for those living with an undiagnosed condition, given they currently represent at least 50% of all people living with rare disease.[88]

Recommendation 4.3: Facilitate access to advanced therapies.

Why this is important: Advanced therapies, also known as advanced therapy medicinal products, are innovative therapies based on genes, tissues, or cells.[89] These therapies aim to prevent, change the course of, or potentially cure some diseases[90] and are often aimed at rare diseases. There are only a few advanced therapies available at the time of writing. However, this field of medicine is undergoing rapid growth and many more are likely to be available soon. Without assistance from their health care team, people living with rare disease may not be aware of, or able to, access these products.

Examples of advanced therapies include:

- Genetic technologies such as the CRISPR-Cas9 gene editing technology. Genetic manipulation technology has been used successfully in clinical trials, and the first CRISPR-based therapy, which upregulates useful compensatory fetal haemoglobin and reduces red cell sickling in sickle cell disease, has now been approved by regulatory authorities in the United Kingdom, United States, and European Union.[91-94] Gene therapy is another mechanism for editing genes, and uses vectors to introduce working versions of genes. It has been used successfully in inherited disorders such as spinal muscular atrophy and eye diseases causing blindness.[95,96]
- Biological therapies such as Chimeric Antigen Receptor (CAR) T-cell therapy. CAR-T is a form of personalised immunotherapy in which T cells from an individual are re-engineered in a laboratory to produce chimeric antigen receptor proteins on their surface.[97] When reintroduced into the patient, these receptor proteins bind to antigens on cancerous cells allowing targeted cell killing. This has been used for leukaemia, lymphoma, and multiple myeloma.

> *"Even just knowing that there's a clinical trial out there is actually useful for your mental health—knowing that there's hope."*

How to progress Recommendation 4.3: As advanced therapies are usually delivered in Centres of Expertise (see Recommendation 3), referring people with rare disease to these centres is critical. In many circumstances, these therapies will only be accessible through clinical research and trials.

Health professionals have a role in educating and supporting people living with rare disease by proactively exploring the evidence for, and availability of, advanced therapies. This should continue before, during, and after receiving new therapies (Appendix 2.17).

A potential challenge is that people living with rare disease who feel they have few options may seek unregulated and unproven cell-based therapies. These may be ineffective and/or pose the risk of serious side effects.[98]

If a health professional is concerned that a person is considering unregulated advanced therapies and/or is travelling abroad for treatment, they could:[99]

- Discuss the risks and benefits of the therapy in a non-judgemental way
- Discuss how to report side effects of the therapy, if they experience any
- Advise they check regulations in the country they are considering travelling to
- Link them to the Therapeutic Goods Administration advice on buying medicines and medical devices online[100]
- Link them to the Smartraveller advice on going overseas for a medical procedure[101]
- Provide resources about these options (Appendix 2.17 and 2.18).

As lack of access or delays in accessing effective treatments such as advanced therapies can contribute to poor mental health and wellbeing, people living with rare disease and their families/carers should be monitored and psychosocial supports provided if needed (see Recommendation 5).

Indicators of good practice	
Actions	▪ Linking people living with rare disease and their families/carers with rare disease and patient advocacy groups ▪ Connecting people living with rare disease to research opportunities, rare disease registries, and clinical trials ▪ Supporting people who are participating in clinical trials and accessing novel therapies
Outcomes	▪ People living with rare disease and their families/carers are encouraged to connect with support groups, expertise, and research opportunities related to their rare disease
Evidence	▪ Interactions with rare disease and patient advocacy groups and sharing of relevant information with people living with rare disease ▪ Capture of rare disease information and codes in electronic medical record data collection systems ▪ Engagement with emerging and advanced therapies ▪ Use of clinical trials databases ▪ Enrolment in clinical registries and natural history studies ▪ Referrals to Centres of Expertise

For useful tools and supports to help implement Recommendation 4, see Appendix 2.1, 2.16-2.18.

5

RECOMMENDATION 5.

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

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

Why this is important: People living with rare disease have an increased risk of developing mental health conditions.[102] In a 2022 survey of people living with or caring for someone with a rare disease, over 90% reported feeling anxious, stressed, or experiencing low mood.[68] High rates of stigma, isolation, uncertainty, and negative educational and financial impacts were reported, leading to a high prevalence of mental health conditions.[12]

People living with rare disease and their families/carers face particular mental health challenges compared to people with other chronic conditions, including the following experiences that were reported during the consultation to develop the Action Plan:[9]

- Feelings of isolation and loneliness
- Prolonged stress
- Ongoing and anticipatory grief
- Devastation and loss of hope.

These feelings can be caused by many factors, including:

- Diagnostic delay
- High burden of care
- Disease progression and emerging symptoms
- Limited proven treatments
- Treatments not working or clinical trials failing to progress
- Lack of knowledge about the condition to provide adequate understanding, including amongst health professionals.

> *"For me, the biggest challenge I face living with [rare disease] is the stigma and judgement around it. I feel weak or judged whenever I need to take a day for me. I also feel there needs to be more awareness of rare diseases in general. A lot of the time, I have to educate health professionals about my condition. I think there is also a need for more support financially and mentally for rare diseases."*

How to progress Recommendation 5.1: Learn about the mental health and wellbeing impacts of living with rare disease. A lesson has been dedicated to the importance of mental health impacts within the free e-learning module, Rare Disease 101 Australia. (Appendix 2.2)[64] additionally, mental health impacts are often highlighted in patient stories profiled on rare disease and patient advocacy websites, including Rare Voices Australia's website (Appendix 2.19).[103]

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

Why this is important: People living with rare disease consistently highlight the need for mental health and wellbeing to be integrated into their care. This builds trust, destigmatises these issues and facilitates appropriate care. Yet, a 2022 survey of more than 1000 people living with rare disease reported that nearly 50% of people had never been asked about their mental health.[2]

> *"I have had to find my own psychologist and ask my GP for a mental health plan… they're not even recognising you need mental health support services."*

How to progress Recommendation 5.2: Health professionals should ask about mental health and wellbeing at each appointment, check if the person needs any additional support, and show sensitivity to people and their family/caregivers. It is also helpful to validate their emotional and psychological responses to the many difficult experiences of living with rare disease.

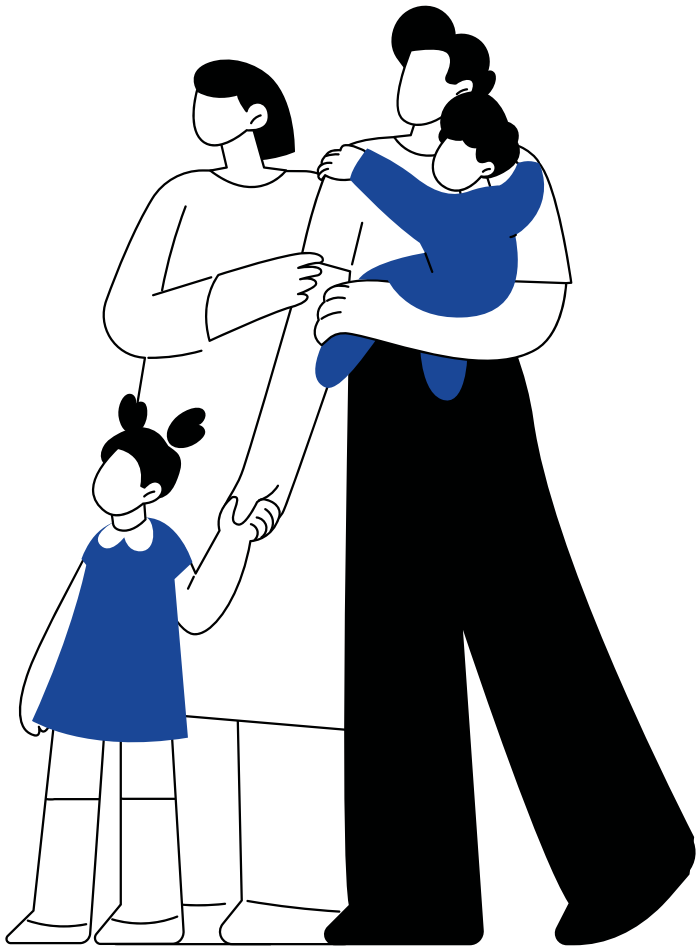
Refer people living with rare disease, including their family members, carers, and support people, to additional support where required. Suggested topics for discussion include:

- Developing a mental health care plan and referring to a psychologist
- Identifying and connecting to appropriate support groups, considering cultural safety and accessibility
- Identifying and connecting to supports to help with practical issues including financial assistance, such as social workers, carer resources, or disability support organisations. The Disability Support Information section in the RARE portal includes information about the NDIS, and aged care, disability and carer supports and resources[11]
- Signposting digital mental health and wellbeing resources
- Ensuring resources are culturally safe and appropriate, and helping navigate access where access to services may be costly or have wait lists.

> *"A mental health care plan should be implemented post-diagnosis…ensuring that the patient [isn't] struggling to come to terms with their rare disease, leading to social isolation. Sometimes you need to talk to a person that's not a family member or a friend or a carer."*

A list of recommended services and resources for people living with rare disease, including for carers, the extended family, and siblings, is provided in Appendix 2.19 and 2.20. Rare Voices Australia, as part of the RArEST project, have developed educational resources to help support people with rare disease (Appendix 2.19).

When referring to mental health professionals, try to identify those who have experience working with people living with rare disease or have undergone relevant training. The RARE Portal (Appendix 2.2) provides links to national and state-based organisations and services.



Recommendation 5.3: Deliver strengths-based and trauma-informed care.

Why this is important: When people are living with rare disease, much of the focus and language is on their deficits and problems. A strengths-based approach identifies the unique strengths, abilities, and resources the person and their support network possess. This can help identify ways that people living with rare disease can optimally deal with challenges, meet their health care goals, and function as integrated whole people.[104] Recent studies on strengths-based approaches for people with chronic illness have found such approaches lead to increased activation and positive health outcomes.[105-107]

Trauma-informed care is a related approach that:[108]

- Considers what has happened to a person rather than what is 'wrong' with them
- Focuses on doing no harm
- Understands the effects of trauma
- Is sensitive to a person's comfort and focuses on working with them.

Many people living with rare disease have had traumatic experiences associated with diagnostic delay, misdiagnosis, having their symptoms disbelieved, the diagnosis of a severe and/or life-limiting condition without treatment options, challenges accessing clinical trials or failure of research efforts, and/or the burden of care associated with a rare condition. Trauma can cause protective responses within the body that may then impact many aspects of mental and physical health.[109]

> *"I have consistently experienced my normal responses to ongoing pain, fatigue and long-term uncertainty being pathologised as an additional health condition."*

A trauma-informed approach, as detailed below, can improve engagement, treatment adherence, and health outcomes for people living with rare disease, as well as provider and staff wellness.[110]

Trauma-informed and strengths-based care recognises that health and illness are best understood in light of the person's unique situation, their past and current experiences, culture, relationships, and the wider community context. These approaches are relevant to the delivery of person-centred and culturally appropriate and safe care (see Recommendation 1).

How to progress Recommendation 5.3: In order to adopt a strengths-based approach the focus shifts from only considering the negative to considering what is currently working well, what the person does best, and what resources people have available.

This can be as simple as asking what the person is most proud of, what they do best, or how they have dealt with challenging situations in the past. A health professional can then work with the person to incorporate effective strategies into their management plan to help them deal more effectively with their life, health, and health care challenges.[111,112]

A trauma-informed approach starts with being aware that a person living with rare disease may well have experienced health care-related trauma. Signs might include:

- Having intrusive thoughts
- Avoiding seeing health professionals
- Negative changes in cognition or mood
- Hypervigilance.

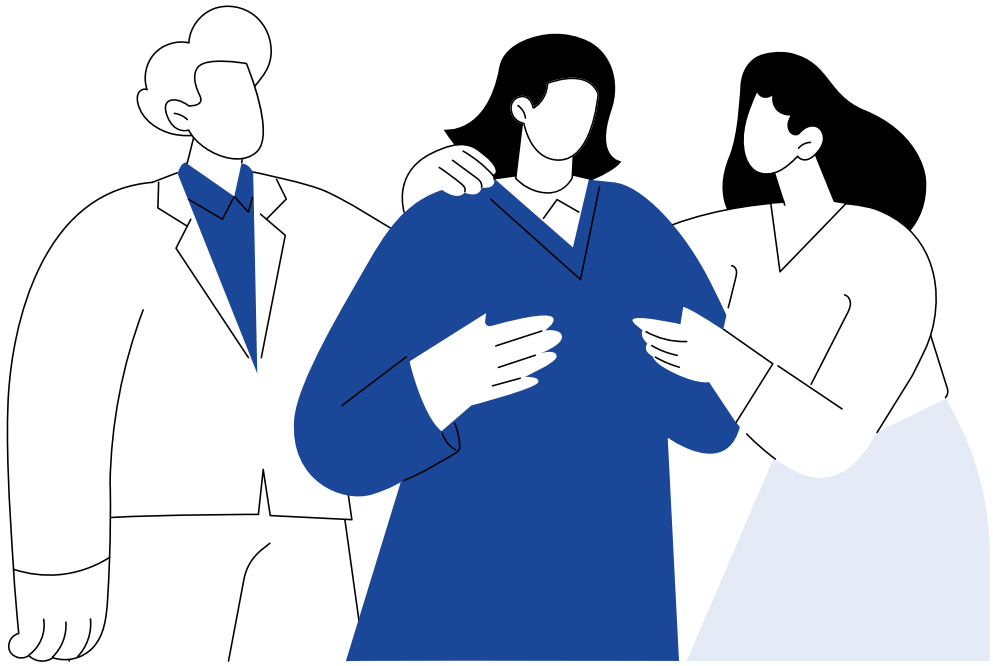
Other actions for practicing trauma-informed care include:[108]

- Embedding physical and emotional safety into your practice
- Building trust by being reliable and responsive to the needs of people living with rare disease
- Collaborating with people living with rare disease and providing choice
- Respecting diversity in all its forms.

Resources to support strengths-based and trauma-informed care are provided in Appendix 2.21.

Indicators of good practice	
Actions	▪ Understanding the impact that a rare disease can have on mental health and wellbeing ▪ Supporting the mental health and wellbeing of people living with rare disease ▪ Supporting the mental health and wellbeing of carers and families, including siblings, of people living with rare disease ▪ Considering strengths-based and trauma-informed principles in therapeutic relationships
Outcomes	▪ People living with rare disease feel supported in their mental health and wellbeing ▪ People living with rare disease report feeling safe and supported following interactions with health professionals
Evidence	▪ Discussion and proactive addressing of psychological and social needs, including practical and financial supports ▪ Use of mental health care plans ▪ Referrals to psychology services ▪ Use of appropriate mental health resources

For useful tools and supports to help implement Recommendation 5, see Appendix 2.2, 2.19-2.21.



6

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.

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

Why this is important: People living with rare disease live with multidimensional health and social care needs that can impact every aspect of their life, such as education, employment, finances, family, and community relationships. Integrated care involves coordinating and connecting health services as well as other sectors, including disability, education, and employment. This requires information sharing, clear pathways through these systems, and support in navigating significant life stage transitions.[113]

> *"It can be very difficult to navigate this rare condition within the community, the education department and coordinating all aspects of his care and NDIS funding. It can be very overwhelming at times trying to advocate correctly for the right services."*

Using a multi-sectoral model reduces the amount of time people living with rare disease spend re-telling their story, attending appointments in multiple locations, and searching and advocating for assistance to meet their needs. Importantly, it also reduces the burden to accurately remember and relay all that was discussed with another care provider.[114] Thus, the opportunity for error and duplication is limited.

> *"I share critical health information between my specialists. This is something I don't feel qualified to do. I am worried that when the information is shared by me, it is not taken seriously."*

How to progress Recommendation 6.1: Multi-sectoral teams can consist of, but are not limited to:

- Physicians (for example, paediatrician, clinical geneticist)
- General practitioner
- Rural generalist
- Aboriginal health worker
- Clinical nurse consultant
- Senior school teacher
- Paediatric nurse
- NDIS support coordinator
- Mental health clinical nurse specialist
- Welfare officer
- Genetic counsellor
- Department of Communities and Justice coordinator.

Such teams would ideally be linked to a rare disease Centre of Expertise (see Recommendation 3). However, many rare conditions lack an easily accessible Centre of Expertise, and many people remain without a diagnosis. In these situations, health professionals can work with people living with rare disease to map out their team (for example, by developing a complex care plan). They can also establish collaborative relationships with such teams, for example, by joining a community of clinical learning practice. This is discussed in Recommendation 8.

To establish and maintain multidisciplinary teams, it is recommended that critical information such as test results, clinical plans, and condition progress are shared. Additionally, it is helpful for copies to be provided to the person and their key carers/support people so they can also share them with organisations outside of health. To keep team members updated on clinical changes and management plans, it is advisable to include all members of the care team in written communication in a timely manner. Open communication between

metropolitan specialists and rural and remote doctors is particularly important given the time-poor nature of these latter professionals. Consideration could be made to including a dedicated advice line in service planning for specialist rare disease services/Centres of Expertise.

Resources to facilitate this include (Appendix 2.22):

- Digital platforms, for example, My Health Record and HealthLink
- Medical, health care or patient 'passports', which can help convey key information about the person's condition, routine and emergency management, and preferences for care and communication[115]
- Personalised Complex Care Plans, shared between all practitioners, the individual, and their family and/or support people, and ideally uploaded onto hospital electronic medical record portals.[116] Such plans are most likely to be beneficial when they are accessible (for example, available in digital and paper based formats) and easy to keep updated.[117]

Recommendation 6.2: Facilitate care coordination for each person living with rare disease.

Why this is important: Ideally, each person living with rare disease would have a dedicated care coordinator in their multidisciplinary care team. A care coordinator manages care across multiple sectors, taking the lead on scheduling, organising, and managing all aspects of a person's health care. They can also be involved in:[118]

- Assessing and screening for care needs and identifying people at risk for adverse outcomes
- Facilitating delivery of evidence-based care
- Ensuring timely and appropriate referral to specialist, allied health, and support services
- Linking to appropriate social care, disability support, and education/employment support services. For example, NDIS coordination, and carer's, disability, sibling, and aged care support and funding
- Facilitating continuity of care between health professionals and across care settings
- Providing timely and consistent education and information
- Participating in activities to improve care coordination and optimise outcomes for individuals and services.

Establishing one person to provide support can improve access to care. It can also reduce the mental load and time burden of living with, or caring for someone with, a rare disease.

Currently, care coordination between providers and services for people living with rare disease is lacking.[114] People living with rare disease often fill these gaps by juggling multiple roles, including that of advocate, case manager, and medical navigator.

> *"I coordinate my care. I research my symptoms, stay up-to-date with new diagnostic and treatment advances, plan my visits to different doctors, monitor my symptoms and test results. I ensure I take my medication. I advocate for myself. It is like having a full-time job on top of my other life commitments."*

Recent evaluations have shown that care coordination services can reduce inpatient admissions, emergency department presentations, and travel.[119] Additionally, simply recognising the burden of care carried by people living with rare disease and their families/carers builds therapeutic rapport.

How to progress Recommendation 6.2: Primary health practitioners can play a key role in care coordination as the central clinician, for example, facilitating delivery of evidence-based care. There are specific Medicare item numbers (for example items 721 and 723), which can help primary practitioners hold longer appointments in order to develop a management plan and put into place a team care arrangement.

Services within tertiary hospitals can provide care coordination in a different sense, including assistance with scheduling and organising appointments, especially where extensive inpatient and outpatient care is required. They can also assist with making sure care is coordinated between different health care settings, linking to appropriate social care, disability support, and education/employment support services, for example, NDIS coordination, and carer's, disability, sibling, and aged care support and funding, and providing information and education.

> *"A rare disease care plan needs to be created, an essential database … that holds all of our medical scans and documentation that can be easily accessed by not only the specialists, your GP and the patient. We want to have equal access to our records. This allows for transparency...of care, handover between professionals and allows the rare disease patient to be part of their medical journey."*

Multidisciplinary clinics and Centres of Expertise, for example the Rare Care Clinical Centre of Expertise for Rare and Undiagnosed Diseases, Perth Children's Hospital, Western Australia, and Rare Diseases NSW, Randwick Health and Innovation Precinct (Appendix 2.23), can also assist, with more rare disease Centres of Expertise being launched in other areas soon. If no care coordinator or Centre of Expertise is available, health care professionals should consider which members of the existing health care team could assist with care coordination.

Other useful services to consider include (Appendix 2.23):

- Complex Care Hub at the Royal Children's Hospital in Melbourne[120]
- Complex Needs Coordination Team (CoNeCT) in Western Australia
- Connected Care Program in Queensland[121]
- Kids Guided Personalised Services (KidsGPS) at the Sydney Children's Hospitals Network[122,123]
- Koorliny Mort (Perth Children's Hospital).[124]

For people with more complex situations, NDIS funding may be available for specialist support coordination to help with coordinating effective community and clinical appointments and support services.[125]

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

Why this is important: Key health and life transitions that require support include entering school, university, employment, adult care, and end of life care. Periods of transition can be challenging for anyone. For people living with rare disease, who often have complex needs, there is a significant risk that transitions will negatively impact health outcomes, including mental health and wellbeing. Successful transitions are those that cause the least disruption and are respectful to the person's health and wellbeing.

> *"A smooth, simple transition as possible for the wellbeing and the continuous care of the patient is really important."*

Support from family members who understand treatment requirements is crucial for people living with rare disease. However, young people transitioning into adulthood often lose this level of support. Without adequate preparation, young people can disengage.[126]

> *"Why does becoming an adult change everything? I am still the same person with the same symptoms, just one year older."*

A collaborative and comprehensive model of care between clinicians, patients, carers, and services may prevent these impacts and reduce the fear a person living with rare disease may feel when transitioning and learning to manage their own health needs.

> *"Between the age of eight and 38 I didn't see a doctor. I've got a superb GP now. I was just open and said, 'You're the first doctor I've seen in 30 years'. And he just said, 'I don't doubt it after being through what you've been through.'"*

For life-limiting conditions, early engagement with palliative care may improve quality of life and increase the likelihood of successful transition.

How to progress Recommendation 6.3: Begin planning for the transition from paediatric to adult health services in the early teenage years and ensure services are available, in addition to support from a general practitioner or rural generalist. Work towards the long-term goals of the young person managing their condition to the best of their ability, interacting positively with adult health services, and engaging with appropriate cross-sector organisations. Many states have dedicated transition care networks or services, listed in Appendix 2.24. Appendix 2.25 lists key principles for transition that can be used to plan a coordinated and person-centred transition to adult health services.[127]

Paediatric to adult transition services can provide guidance and support to move across health services and prevent feelings of fear, uncertainty, and abandonment that may arise during this process, as well as provide skills training and resources to empower young people living with rare disease.

Refer people with life-limiting conditions to palliative care early to allow time to build relationships and trust with their palliative care team. Understanding the multifaceted role of palliative care, including perinatal palliative care, aids in knowing when to refer and how to discuss these services with people living with rare disease. Relevant palliative care services and resources are listed in Appendix 2.26.

Indicators of good practice

Actions

- Assisting people living with rare disease in navigating social support services, the NDIS, financial and disability supports, education, and employment
- Involving people living with rare disease in the development of a holistic inter-sectoral care plan
- Ensuring timely written communication from the clinician to the primary carer and person living with rare disease to facilitate better management of complex needs
- Supporting people living with rare disease during transitions (for example, from paediatric to adult care, initiating palliative care)
- Working with multidisciplinary teams to offer coordinated care to people living with rare disease
- Taking steps towards providing equitable care that is accessible to all Australians

Outcomes

- People living with rare disease report efficient communication among their health care team members
- People living with rare disease feel their care is continuous and well coordinated
- People living with rare disease are accessing appropriate cross-sectoral services and financial and disability supports

Evidence

- An assigned, dedicated care coordinator or complex care plan for people living with rare disease
- Proactive planning for transitions to adult care, palliative care, and other related services
- Engagement with and integration of cross-sectoral services including employment, education, and welfare services to provide comprehensive support to people living with rare disease, including the NDIS and carers, respite, and financial assistance
- Referrals to multidisciplinary clinics or Centres of Expertise
- Referrals to paediatric to adult transition services
- Referrals to palliative care

For useful tools and supports to help implement Recommendation 6, see Appendix 2.2; 2.22-2.26.

7

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.

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

Why this is important: Health promotion is the process of encouraging and enabling people to increase control over, and to improve their health.[128] Rare diseases can have a range of causes, including genetic, autoimmune, infectious agents, and environmental factors (such as asbestos exposure).

Delivery of relevant primary prevention measures can reduce the incidence of rare diseases, whereas delivery of secondary to quaternary prevention measures through early diagnosis, evidence-based action, and person-centred communication can ameliorate or minimise the manifestations and burden of rare diseases.

Population- and individual-based preventive interventions for rare diseases may include:

- Enabling reproductive genetic counselling and preimplantation genetic diagnosis (primary prevention)
- Facilitating early, rapid, and accurate diagnoses of rare diseases, for example, through prenatal and newborn screening (secondary prevention)
- Coordinating with a Centre of Expertise (Recommendation 3.1) and implementing best management guidelines to reduce or prevent complications of rare disease (tertiary prevention)
- Protecting individuals from interventions that are more likely to cause harm, such as providing accurate information on the risks of advertised interventions with no evidence of therapeutic effect. For example, stem cell tourism (quaternary prevention).

How to progress Recommendation 7.1: Understand and work in partnership with people living with rare disease and their families to promote the importance of preventive measures for rare diseases and the best opportunities for action. Such measures are listed below.

- **Comprehensive prenatal and pregnancy care:** This includes screening and advice which could reduce the chance of, or result in, early detection and management of a range of rare diseases. A checklist may be helpful to guide the screening process, such as the PLaN checklist from the Royal Hospital for Women, Sydney.[129] As part of this process, referral to appropriate specialists may be required, including to a maternal fetal medicine unit and/or clinical genetics service. More details are provided in Appendix 2.27 and Appendix 4.
- **Antenatal testing and screening:** This should be offered to all women as part of their first trimester screening. Additional prenatal testing should be offered to those who require further testing (Appendix 2.27 and Appendix 4).
- **Registration of congenital anomalies:** Fetuses and babies with congenital anomalies (live births, stillbirths, and terminations of pregnancy) should be reported to the relevant registry (Appendix 2.14). This allows for the collection of high-quality data for primary prevention of rare diseases impacted by medicinal drugs, food/nutrition, lifestyle, health services, and environmental pollution.
- **Newborn screening programs:** These programs should be offered to all parents. The newborn bloodspot test screens babies for a range of conditions, including phenylketonuria, congenital hypothyroidism, and cystic fibrosis. Newborn hearing screening can detect hearing loss. Early action can lead to better results for the baby or prevent serious illness (Appendix 2.28).

- **Routine child health nurse developmental checks:** These checks should be undertaken as they can detect early signs of a rare disease (Appendix 2.28).
- **Appropriate immunisations:** Facilitation of all immunisations according to the National Immunisation Program Schedule is important to prevent rare infectious diseases such as measles, polio, and tetanus.[130] Additional immunisations as described in the Australian Immunisation Handbook (Appendix 2.29) should be offered where appropriate to people with medical conditions (for example, people with Down syndrome, premature babies, and immunocompromised individuals).[130] Additional immunisations should also be offered to those with environmental risk factors (such as farm animal handlers or people living in areas where Mycobacterium ulcerans is present).
- **Occupational health and safety:** Occupational history is important to understand if people may have been historically or currently exposed to environmental agents that can cause rare diseases (such as asbestos, industrial chemicals, dusts, or metals) (Appendix 2.30).

Individual-based secondary prevention strategies can also ameliorate or minimise the manifestations and burden of rare diseases through early diagnosis and action.

Families with several members who have had specific cancers (often breast, ovarian, or bowel cancer) or individuals with multiple cancers diagnosed at an unusually young age should be offered a referral to a family cancer clinic.[131] The eviQ guidelines from Cancer Institute NSW can help guide risk assessment and referral, as well as provide plain English fact sheets for families (Appendix 2.30).[132]

Genetic counselling and information on individual risk, including cascade testing and relevant management, should be offered to relatives at risk of inherited genetic conditions. See the Centre for Genetics Education website for details of local clinical genetics services (Appendix 2.2).

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

Why this is important: Reproductive testing and technologies include reproductive carrier testing, preimplantation genetic testing, and prenatal ultrasound, screening and genetic testing. These tests and technologies are increasingly able to provide couples with personalised information about the chance of having a baby with a rare congenital condition. This includes genetic conditions such as cystic fibrosis and conditions that may not have a genetic aetiology such as isolated congenital heart disease.

All women should have access to individualised, evidence-based preconception, antenatal, and perinatal care that offers screening for, and management of, risk factors for genetic and non-genetic rare diseases.[133,134]

On average, each person is a carrier of about two different genetic variants linked to inherited genetic conditions. In addition, a significant proportion of rare congenital conditions are preventable with non-genetic measures, such as nutrition, weight and lifestyle advice, folic acid and iodine supplementation, immunisation status review, medication review, and optimisation of chronic medical conditions.

> *"Previous genetic testing for my condition came back inconclusive, but this time the 'gene fault' was found. This was great news for us, because it meant that health professionals can use this information for in vitro fertilisation when we are ready."*

How to progress Recommendation 7.2: All couples should be supported in their preparation for a healthy pregnancy according to guidelines from the Royal Australian and New Zealand College of Obstetricians and Gynaecologists[135] and the Royal Australian College of General Practitioners.[136]

This includes (Appendix 2.31 and Appendix 4):

- Stabilisation of any medical problems
- Discussion of reproductive genetic carrier screening
- Updating vaccination status
- Lifestyle recommendation regarding health, weight, folate supplementation, and substance use.[133,137]

For couples concerned about medication or potential teratogens, consultation with or referral to a specialist teratogen or drug information service[138] (Appendix 2.27) is recommended.

A family history should be taken from couples planning a pregnancy, with referrals made to clinical genetics teams for genetic counselling if a history of inherited conditions is identified. Reproductive options for couples who are at an increased chance of having a child with a genetic and/or rare condition should be referred to a genetics and maternal-fetal medicine team. Information on reproductive choices, screening and testing, as well as contact details for local genetics services can be found at the Centre for Genetics Education (Appendix 2.31).[54]

Preconception and early pregnancy screening and care should be non-directive to allow people to make informed reproductive choices and access health care in line with their wishes and values. Supportive counselling is important, as many people choose to have screening or testing to help them prepare to deliver their baby in an appropriate centre, rather than to inform decisions about continuing a pregnancy.

As the availability of prenatal and perinatal treatment for rare diseases is increasing, connection with a Centre of Expertise is important to ensure the most up-to-date advice and treatment is offered (see Recommendation 3).[139] Families may value speaking to someone with lived experience of a rare condition whilst making reproductive choices, so connection to patient support and advocacy groups can be helpful (see Recommendation 4).

Indicators of good practice	
Actions	▪ Practicing preventive medicine and health promotion related to rare diseases ▪ Offering, or supporting the offering of, screening and testing including preconception carrier screening
Outcomes	▪ People living with rare disease report that matters relating to sensitive topics, such as heritable genetic conditions and the privacy of genetic information, are handled with sensitivity
Evidence	▪ Data contributed to congenital anomaly registers ▪ Adherence to best practices for antenatal care, immunisations, and screening programs ▪ Facilitation or support of cascade testing offered to family members, where appropriate

For useful tools and supports to help implement Recommendation 7, see Appendix 2.2, 2.14, 2.27-2.31, and Appendix 4.

8

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.

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

Why this is important: Awareness of the needs of people living with rare disease, and approaches to support them, is an essential skill for all health professionals. However, rare diseases are not a focus of most training curricula. Engaging with tailored professional development can enhance awareness, understanding, and expertise, leading to improved outcomes for people living with rare disease.

> *"The first thing [my health professional] did when I had my initial consultation, and I explained my condition, was she went and researched therapies for [my symptoms] and specifically she researched my disease and ... what the progression looks like... To me [that] is a dedicated professional with buy-in... and it's something they may not come across every day, so why wouldn't you want to be more educated on that..."*

Competencies that are relevant to the care of people living with rare disease can be considered in two categories: those that are common across all fields of clinical care, including some competencies that need additional attention within the context of rare disease; and competencies that are specific to the management of patients with rare disease (see Figure 4).

Extensions of competencies relevant to all aspects of clinical care

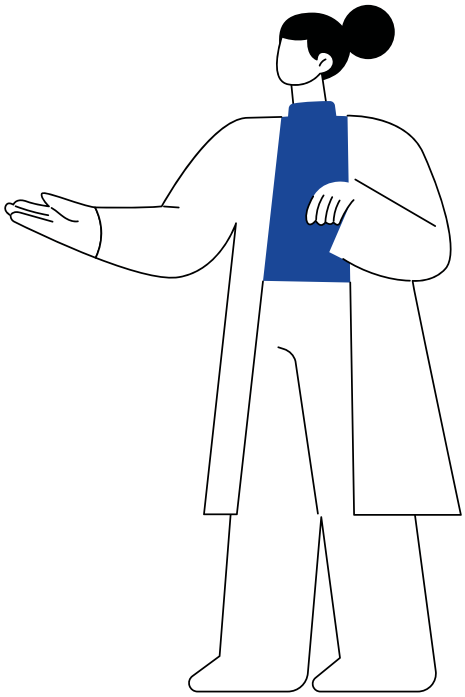
- A patient-centred approach
- Holistic care
- Effective communication strategies

Rare disease-specific competencies

- Skills in accessing up-to-date information on specific rare diseases
- Accessing peer networks to share practice
- Utilising CPD activities specific to rare disease

Figure 4: Examples of competencies relevant to the care of people living with rare disease. CPD, continuing professional development

Tailored Continuing Professional Development (CPD) activities are required to develop these competencies.



How to progress Recommendation 8.1: CPD plans should be regularly reviewed and targeted towards skills or expertise that may help health professionals meet the needs of people living with rare disease. Relevant CPD activities may include:

- Participating in conferences specific to rare disease
- Engaging in peer-to-peer learning
- Rare disease-specific online learning
- Interprofessional and interdisciplinary learning
- Investing time in reviewing practice performance and measuring outcomes.

The Rare Disease 101 Australia module[64] (Appendix 2.13) is an example of a highly relevant source of CPD. Other key areas that could be considered are trauma-informed care, mental health training, and strengths-based communication practice.

> *"I'd like to see [health professionals] go through an annual education in rare diseases ...to give them ... the skills to look at pathways and ... professional conduct with rare disease patients and ...how to connect them to rare medical specialist networks rather than just saying 'it's a little bit too hard, it's out of my scope of work'"*

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

Why this is important: Rare disease is often associated with a high degree of uncertainty, which can have an impact on health professionals and people living with rare disease.[13] While developing these recommendations, people living with rare disease have shared that by not addressing some common challenges, health professionals are not providing them with high quality care.

Reflection, as an essential part of professional development, involves consciously thinking about and analysing our own and others' experiences, actions, and decisions to gain insight, improve performance, and enhance personal and professional development. Reflective practice can play a significant role in maintaining the wellbeing of health professionals.

Reflection is particularly important for clinicians who treat people with rare disease because these conditions present unique challenges that may require innovative or unconventional solutions. It is particularly important to reflect on the lived experience of rare disease, and the range of challenges faced by patients and families, in order to be responsive to their needs.

> *"I wasn't aware that I had a rare disease until an intern specialist told me that the hospital only had a few patients like me... Having a doctor who recognised me as a rare disease patient and one who was willing to go on that journey with me would have resulted in a very different illness experience and health outcomes."*

How to progress Recommendation 8.2: CPD plans should include strategies to develop skills in listening to and acknowledging the illness experience from the perspective of people living with rare disease.[140] As a means to develop their reflective skills more broadly, clinicians may choose to use models such as Kolb's cycle of experiential learning.[141]

Health professionals should also reflect on the impact of uncertainty or case complexity on their own professional practice, identifying where additional support may be needed from colleagues or formal supports. Participation in peer-to-peer learning modules, such as a community of clinical learning practice, for example, Rare Disease Project ECHO® (Appendix 2.13) can be a useful approach to facilitate discussion, reflection, and improvement in practice.

For reflection to be truly meaningful, it must be accompanied by the development of concrete plans for next steps to address any areas of need.

8

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

Why this is important: Quality improvement involves continuously assessing and improving practices, and seeking feedback from patients, colleagues, and other stakeholders. Seeking such feedback will help to ensure health professionals are providing the best possible care for patients with rare disease. Feedback from these sources will augment routinely collected data and will often help to contextualise and interpret the data in a meaningful manner. The need for further quality improvement is highlighted by the following:

- A 2021 EURORDIS Rare Barometer survey, which showed patients with rare diseases rated their health care experience 2.5 out of 5—much worse than those with other chronic illnesses[15]
- A 2016 survey of parents in Australia and New Zealand, which found that a majority (54%) were dissatisfied with health professionals' level of knowledge and awareness of their rare disease.[142]

How to progress Recommendation 8.3: Options range from relatively simple methods to gather feedback from relevant parties to more formal processes such as '360-degree evaluation'. Some health contexts may have established mechanisms to collect patient experience and outcome data that individual clinicians can use. Reviewing performance and measuring outcomes are key areas in many college and society CPD requirements, for example, the Royal Australian College of General Practitioners (RACGP; Appendix 2.32).[143]

It is important to embed these feedback processes into routine practice to increase acceptability and gradually reduce any anxieties around them. Seeking feedback may be daunting for some practitioners, so mechanisms to support these processes need to be developed. Mechanisms also need to be developed to support clinicians in meaningfully addressing feedback within a culture that values feedback as a development tool. Data must be confidential and only used for the purpose for which it is collected.

Clinical audits are another important aspect of quality improvement. Health professionals should periodically review their patients who have existing rare disease diagnoses, including the need for additional support. Clinicians should also periodically review their patient cohorts to identify those who may have an undiagnosed rare disease to evaluate their needs, and tailor care accordingly.

Clinical audits can be supported using appropriate clinical codes as mentioned in Recommendation 4. Participating collaboratively in research and clinical trials that include measures of patient experience can also be a source of data to support quality improvement activities.

CPD enabling audits, such as those offered by the RACGP[143] (Appendix 2.32) are an option when a more structured clinical audit is desired.

Indicators of good practice	
Actions	▪ Engaging in self-reflective practice to identify areas for professional development relating to rare disease and acting to develop these areas ▪ Participating in regular evaluations of clinical care for people living with rare disease ▪ Applying quality improvement principles to continually improve care of people living with rare disease
Outcomes	▪ People living with rare disease report that health professionals demonstrate a willingness to acknowledge the limitations of their knowledge and seek opportunities to learn about best practice care for their rare disease
Evidence	▪ Capture of rare disease information and codes in electronic medical record data collection systems ▪ Uptake of rare disease-focused professional development and quality improvement

For useful tools and supports to help implement Recommendation 8, see Appendix 2.13, 2.32.

Appendices

APPENDIX 1: GLOSSARY AND ABBREVIATIONS

Glossary

Advanced therapies / Advanced therapy medicinal products: Medicines for human use that are based on genes, tissues, or cells

Cascade testing: The process by which biologically-related family members are informed about a genetic condition that has been discovered within their family, followed by stepwise testing of family members.

Coding: Medical coding is the transformation of health care diagnosis, procedures, medical services, and equipment into universal medical alphanumeric codes. Many individual rare diseases still lack a specific code in the most commonly used medical coding systems, leading to under recognition of the importance of rare conditions.

Congenital anomaly: A structural or functional change that occurs while an individual is in the womb.

CRISPR-Cas9: A technique used to edit the genome, which was adapted from a naturally occurring genome editing system present in bacteria. CRISPR-Cas9 is short for clustered regularly interspaced short palindromic repeats and CRISPR-associated protein 9.

Diagnostic odyssey/journey: The time between when a symptom or feature of a rare disease is first noted to when a final diagnosis is made. Often called an odyssey because for many people it can take years for a diagnosis to be reached and may involve several incorrect diagnoses.

Gene therapy: A medical therapy that treats or prevents disease by correcting the underlying genetic problem.

Genetic/genomic testing: Analysis of a person's genetic material (DNA) to identify variants that could be associated with a condition or the chance of developing a condition.

Genomic reanalysis: Re-examination of DNA sequence data from a previous genomic test.

Health promotion: A process that encourages and enables people to increase control over, and to improve, their health.

Natural history study: A preplanned observational study that tracks the course of a disease over time, usually in the absence of any therapeutic intervention.

People living with rare disease: Individuals with a rare disease, as well as their family, carers, and support people.

Phenotype: An individual's observable traits, such as eye colour, height, and blood type, which is influenced by both the person's genetic makeup and environmental factors.

Registry: A dedicated database capturing information about people with specific conditions or types of conditions.

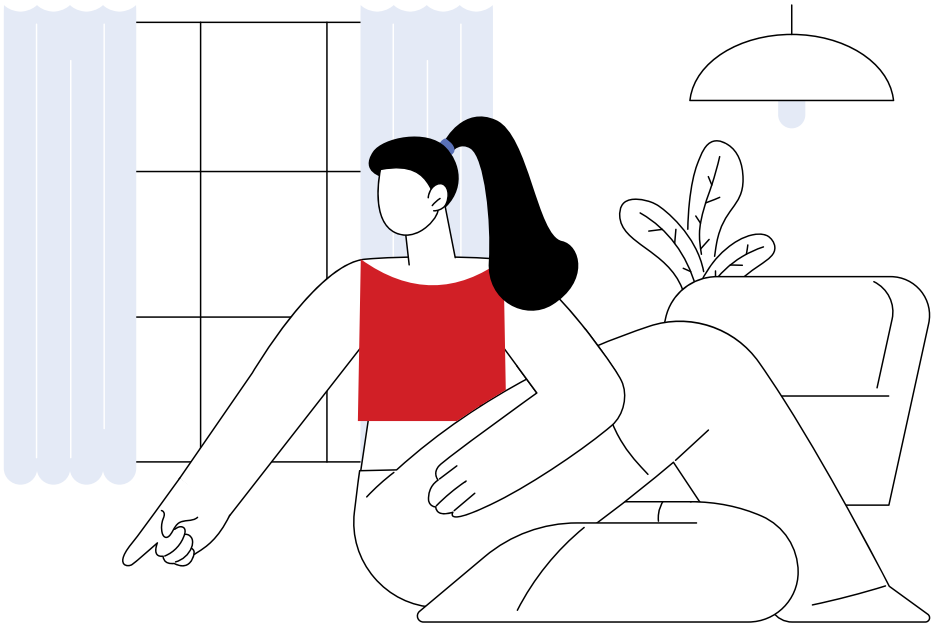
Undiagnosed disease program: A research program, often a collaboration between multiple researchers and institutes, that utilises the latest genomic technologies and emerging analytic tools to find diagnoses for undiagnosed individuals.

Variant: A permanent change in the DNA sequence that makes up a gene. These may be either inherited from a biological parent or occur spontaneously during a person's lifetime (either before or after they are born).

Variant of uncertain significance: A genetic variant identified by genetic or genomic testing that may or may not cause disease. The pathogenicity is unknown because insufficient data is available.

Abbreviations

ACSQHC	Australian Commission on Safety and Quality in Health Care
CAR-T	Chimeric antigen receptor T-cell
CPD	Continuing Professional Development
GARD	Genetic and Rare Disease Information Center
HIV	Human immunodeficiency virus
MBS	Medicare Benefits Schedule
NDIS	National Disability Insurance Scheme
RACGP	Royal Australian College of General Practitioners
RACP	Royal Australasian College of Physicians
RARE	Rare Awareness Rare Education
RArEST	Rare Disease Awareness, Education, Support and Training
TORCH	Congenital infections of toxoplasmosis, syphilis, hepatitis B, rubella, cytomegalovirus, herpes simplex, and others
UDNI	Undiagnosed Diseases Network International
UDP	Undiagnosed Diseases Program



APPENDIX 2: ENABLERS OF GOOD PRACTICE

2.1 The National Strategic Action Plan for Rare Diseases

National Strategic Action Plan for Rare Diseases (Australian Government): Outlines a comprehensive, collaborative, and evidence-based approach to achieving the best possible health and wellbeing outcomes for Australians living with rare disease.

2.2 Curated information portals on rare and genetic conditions

RARE Portal (Rare Voices Australia): A digital library of verified rare disease information, services, and resources. A living website in ongoing development, with new information added regularly.

RARE Helpline (Rare Voices Australia): A free helpline that:

- Supports people to connect with existing reliable information
- Provides resources in response to specific needs
- Helps to increase health literacy and engagement with care and support services
- Assists in connecting people with existing health services and/or professionals where possible.

Rare Disease Australia 101 e-learning module (RArEST and Medics4RareDiseases): A series of short, free, and interactive e-learning modules for health professionals in the Australian context, focusing on person-centred approaches to common challenges for rare diseases. The modules are accredited for Continuing Professional Development by the RACGP.

Genetic and Rare Diseases Information Center (GARD) (USA National Institutes of Health): Provides information about rare and genetic diseases, including resources and organisations that help support the needs of children and adults living with rare disease.

Orphanet (French National Institute of Health and Medical Research and European Commission): Includes a range of information to improve knowledge on rare diseases, including a high-quality dataset related to rare diseases and orphan drugs.

GeneReviews (University of Washington): An international point-of-care resource for clinicians, providing clinically relevant and medically actionable information for inherited conditions in a standardised journal-style format, covering diagnosis, management, and genetic counselling for patients and their families.

Centre for Genetics Education (NSW Health): Provides a range of information about genetic conditions, including fact sheets on genetic conditions and online learning programs for health professionals, patients and caregivers.

Rare Disease Database (National Organization for Rare Disorders; USA): Information on over 1200 different rare conditions.

MedlinePlus Genetics (USA National Institutes of Health): Includes a database of over 1300 health conditions with a genetic basis.

2.3 Rare disease patient advocacy and support groups and guides

A-Z Support Directory (Rare Voices Australia): Links to support groups for a wide range of rare diseases.

Engaged, Ethical and Effective: A Guide for Rare Disease Organisation Leaders in Australia (Rare Voices Australia): The Guide is for both established rare disease organisations and people interested in setting up an organisation. For existing rare disease organisations, the Guide can be used in several ways to complement the current status, resources, and development phase of an organisation. For those investigating how to establish an organisation, this is an easy to follow resource that covers the key areas to consider. Each section contains information about the topic, links to relevant resources, and a checklist or self-evaluation tool for identifying current strengths and areas for development.

- Chapter 1: Introduction to Rare Diseases, Rare Voices Australia, the Australian Landscape and the National Strategic Action Plan for Rare Diseases
- Chapter 2: Rare Disease Organisation Strategy
- Chapter 3: The Vital Roles of Rare Disease Organisations
- Chapter 4: Ethical Rare Disease Organisations
- Chapter 5: Governance Foundations
- Chapter 6: Funding Your Rare Disease Organisation
- Chapter 7: Community Engagement
- Appendices: Assessment Tools and Planning Template.

2.4 Health literacy guides

Health literacy and rare diseases: 4 ways to close the gap (Healthgrades Marketplace): An overview of four ways to close the health literacy gap and improve care and quality of life for people with rare diseases.

2.5 Patient partnerships, shared decision making, and respecting diversity

Partnering with patients in their own care (Australian Commission on Safety and Quality in Health Care): A guide to effective partnerships with all patients, including key resources and supports to deliver person-centred care.

Supportive resources on shared decision making (Australian Commission on Safety and Quality in Health Care): Tools and resources to help consumers and health professionals make shared decisions together.

- These include resources addressing:
  - "What are my options?"
  - "What are the possible benefits and harms of those options?"
  - "How likely are each of those benefits and harms to happen to me?"
- Two-minute videos on:
  - What shared decision making is
  - Challenging common myths about shared decision making (such as the time it will take)
  - Patient decision aids that can help in clinical practice.
- The Partnering with patients in their own care standards is a useful guide.
- The Supportive resources on shared decision making tools can also be adapted to the rare disease context, for example those recommended by the Australian Commission on Safety and Quality in Health Care.

Ask, Share, Know (AskShareKnow): This website provides downloadable point-of-care resources to share with people living with rare disease to facilitate shared decision making using the Ask, Share, Know questions.

Standardized pedigree nomenclature update centered on sex and gender inclusivity: A practice resource of the National Society of Genetic Counselors: This practice resource provides up-to-date recommendations for inclusive communication and family trees (pedigrees) which respect sexual and gender diversity.

2.6a Guides and resources to support making reasonable adjustments for people living with rare disease with intellectual disability

Comprehensive Health Assessment Program (CHAP) tool (UniQuest Pty Ltd): A tool for health practitioners (for example, general practitioners or rural generalists) to guide annual health checks for people with intellectual disability. A dedicated Medicare Benefits Schedule (MBS) item (707) is available for these checks.

National Roadmap for Improving the Health of People with Intellectual Disability (Australian Government Department of Health and Aged Care): Highlights that best-practice models of care need to be developed that are person-centred, trauma-informed, and enable reasonable adjustments.

Reasonable adjustments (Australian Commission on Safety and Quality in Health Care): A suite of resources explaining the legal framework behind reasonable adjustments, practical how-to guides for clinicians, and posters and resources that can be used in clinical settings.

GeneEQUAL (NSW Health, UNSW Sydney): Includes an Educational Toolkit developed in partnership with NSW Health and the Centre for Genetics Education explaining how to deliver more inclusive genetic health care. Includes step-by-step guides and resources to help health professionals follow three key principles, namely making reasonable adjustments, and delivering person-centred and trauma-informed care; videos demonstrating how to put those principles into action; and Easy Read booklets on genetics and genomic medicine.

Just Include Me (Council for Intellectual Disability): Free online training to help health professionals provide person-centred care.

2.6b Easy Read resources

Easy Read is the presentation of text in an accessible, easy to understand format, where every concept is accompanied by a clear image. It is often useful for and preferred by people with learning disabilities and may also be beneficial for people with other conditions affecting how they process information, people who do not speak English as a first language, or those who have low health literacy.

Easy Read information relevant to rare disease health care can be found at the following websites.

The Council for Intellectual Disability (Council for Intellectual Disability):

- Includes health guides on a range of topics, including common procedures like blood tests and a guide to the Medicare funded annual health care checks for people with intellectual disability.
- My Health Matters is an Easy Read folder created to improve communication between people with an intellectual disability and their health professionals.

GeneEQUAL (NSW Health, UNSW Sydney): Includes Easy Read booklets on genetics and genomic medicine.

Department of Developmental Disability Neuropsychiatry (3DN) (UNSW Sydney): Easy Read booklets on mental health.

2.7 Supporting and providing culturally safe health care for Aboriginal and Torres Strait Islander people

Lyfe Languages Program (Project Y): A universal medical translator that translates complex medical terminologies into Indigenous languages and includes resources to facilitate culturally appropriate rare disease healthcare, connecting ancient knowledge with new technologies.

Rare Disease Resources for Aboriginal and Torres Strait Islander Community (Rare Voices Australia): A collection of helpful links for Aboriginal and Torres Strait Islander people who live with rare diseases.

Australian Indigenous HealthInfoNet (Edith Cowen University): Information and evidence on many aspects of Aboriginal and Torres Strait Islander health and wellbeing, including information to help support health professionals to critically reflect on their practice and learn how to deliver safe, accessible, and responsive health care that is free from racism.

Aboriginal and Torres Strait Islander Health Curriculum Framework (Australian Government Department of Health): Details five core competencies for health care professionals providing culturally safe health care.

Clinical Yarning e-learning program (Western Australian Centre for Rural Health): A free online program developed to improve the effectiveness of communication of all health clinicians who work with Aboriginal and Torres Strait Islander people.

13Yarn (Australian Government, Lifeline): Culturally appropriate mental health crisis support and referral for Aboriginal and Torres Strait Islander people.

Palliative Care and End of Life Guidelines (Australian Indigenous Health InfoNet): Guidance for providing palliative and end-of-life care.

Indigenous Program of Experience in the Palliative Approach (Program of Experience in the Palliative Approach): Grassroots approach to breaking down barriers to palliative care.

Gwandalan (Australian General Practice Accreditation Limited, in partnership with Palliative Care South Australia): Supports palliative care for Aboriginal and Torres Strait Islander communities by providing e-learning modules, webinars, and other resources for frontline staff.

2.8 Supporting people from culturally and linguistically diverse backgrounds

Medical Translating and Interpreting Service (Australian Government Department of Home Affairs): A service for private medical practitioners. There are also state-based health translating services.

Embrace Multicultural Mental Health (Mental Health Australia): Mental health resources and information for people from culturally and linguistically diverse backgrounds.

National Ethnic Disability Alliance (NEDA): NEDA is a national disabled people's organisation that advocates federally for the human rights of culturally and linguistically diverse people living with disability.

Multilingual resources (Palliative Care Australia): Resources in 21 languages.

Cultural and linguistic diversity considerations in palliative care (CareSearch Australia): Guides to providing culturally sensitive care to culturally and linguistically diverse communities.

Rare Disease Resources for the Multicultural Community (Rare Voices Australia): A collection of helpful links for multicultural people living with rare diseases.

2.9 Supporting people in regional, rural, or remote areas

Services Australia Assistance for Isolated Children Scheme (Australian Government): A group of payments for parents and carers of children who are unable to attend a local state school due to geographical isolation, disability, or special needs.

Healthdirect rural and remote health (Australian Government Department of Health and Aged Care): A collection of health-related resources for people living in rural and remote areas.

Australian College of Rural and Remote Medicine (Australian College of Rural and Remote Medicine): Supports Continuing Professional Development for rural and remote medical practitioners. Information on resources and training is available on their website.

Ronald McDonald Houses (Ronald McDonald Houses Charity): A home-away-from home for seriously ill children and their families.

Royal Flying Doctor Service of Australia (Royal Flying Doctor Service): Offers 24-hour aeromedical emergency services for people living in rural and remote areas in the Australian Capital Territory, New South Wales, Northern Territory, Queensland, South Australia, and Western Australia, as well as primary health services and non-emergency transportation in all states and territories.

Angel Flight (Angel Flight Australia): A not-for-profit organisation that coordinates non-emergency flights to assist people living in rural and remote areas to access specialist medical treatment that would otherwise be unavailable because of the distance and high travel costs.

Rare Disease Resources for Regional, Rural and Remote Communities (Rare Voices Australia): A collection of helpful links for people who have rare diseases and live in regional, rural, or remote parts of Australia.

2.10a Tools to support recognition of people living with rare and or genetic conditions

Genetic red flags: clues to thinking genetically in primary care practice (University of Washington): Original red flags for genetic conditions from Whelan et al., (2004).[52]

A Diagnostic Odyssey – Red Flags in the Red Sand (Medicus): Case study applying red flags in an Australian context.

2.10b Artificial Intelligence-assisted diagnostic tools

Orphanet Clinical Signs and Symptoms App (Orphanet): An online tool that uses clinical signs and/or symptoms entered by the user to search for a rare disease.

PubCaseFinder (Database Center for Life Science): A web-based clinical decision support system that uses human phenotype ontology-based phenotypic similarities to provide ranked lists of genetic and rare diseases that represent the most likely differential diagnoses.

FindZebra (FindZebra): An online tool to assist with the diagnosis of rare diseases that uses freely available, high quality curated information on rare diseases, and open source information retrieval software.

2.11 Support organisations for people living with an undiagnosed rare disease

Syndromes Without a Name (SWAN) Australia: A national organisation providing information, support, connection, and advocacy to families caring for a child with an undiagnosed or rare genetic condition.

Wilhelm Foundation: A global organisation aiming to facilitate diagnoses for children and adolescents.

2.12 Undiagnosed disease programs

The Australian Undiagnosed Disease Network (UDN-Aus) (Murdoch Children's Research Institute): A national initiative to support genomic reanalysis and further multiomic research-based testing.

Australian Functional Genomics Network (Murdoch Children's Research Institute): A national collaboration aiming to connect clinicians and researchers to investigate the functional impact of genetic variants in genomic medicine.

Undiagnosed Diseases Network International (UDNI): An international network working collaboratively and globally to:

- Provide diagnoses for patients who have eluded diagnosis by clinical experts
- Contribute to standards of diagnosis by implementing additional diagnostic tools
- Foster research into the aetiology and pathogenesis of novel diseases
- Disseminate those research results broadly and rapidly.

Australian members of the UDNI can be found at their website.

Undiagnosed Disease Program (Genetic Services of Western Australia): Western Australian program aimed at finding a diagnosis for the undiagnosed.

GeneAdd (Sydney Children's Hospitals Network): A clinical research service at Sydney Children's Hospitals Network, NSW.

Rare Disease Flagship (Murdoch Children's Research Institute): A clinical research service at the Royal Children's Hospital and Murdoch Children's Research Institute, Victoria.

2.13 Educational resources and clinical communities of practice for health professionals on genomics and rare disease

Rare Disease Australia 101 e-learning module (Medics4RareDiseases): A series of short, free, and interactive e-learning modules for health professionals in the Australian context, focussing on person-centred approaches to common challenges for rare diseases. Rare Disease 101 is accredited for RACGP CPD points and includes:

- Lesson 1: Introduction to Rare Disease
- Lesson 2: Understanding the Common Challenges
- Lesson 3: Mental Health and Wellbeing
- Lesson 4: Respectful and Effective Communication
- Lesson 5: The Diagnostic Odyssey and Diagnostic Tools
- Lesson 6: Genomics 101
- Lesson 7: Coordinated Care Across the Lifespan
- Lesson 8: Patient Advocacy Groups
- Lesson 9: Accessing Research and New Therapies

Rare Disease Project ECHO® (Rare Voices Australia, UNSW Sydney, the University of Western Australia and Macquarie University): A free, innovative video conferencing 'hub-and-spoke' outreach model connecting community providers or practices to a multidisciplinary team. Accredited for RACGP CPD points.

check RACGP CPD Solution (RACGP): *Check* is an RACGP CPD learning activity that is produced 11 times a year by the RACGP. All cases are written by expert clinicians and reviewed by subject matter experts. Each unit comprises approximately five clinical cases with answers, followed by multiple-choice questions, as well as references and resources. Unit 607 (December 2023) is dedicated to rare disease.

HealthPathways (Streamliners): An online tool providing concise point-of-care information on the clinical assessment, management, and referral pathways for a range of conditions. Primarily designed for general practice but can also be used by specialist, allied health, and other health teams in over 40 regions in Australia and New Zealand.

Clinical Genomics for Physicians e-learning module (Royal Australasian College of Physicians [RACP]): This resource, which can be found by searching for 'clinical genomics' on the RACP website home page, aims to introduce Australasian medical specialists to genomics, help them appropriately discuss genomics with patients and their families, and refer patients for genomic testing.

Genomics in General Practice Second (2023) Edition (Royal Australian College of General Practitioners): A suite of concise summaries on various clinical topics in genetics and genomics.

Guidelines for Community Involvement in Genomics Research (Australian Genomics): These guidelines for health professionals directly involved in genomics research projects may also be a useful resource for educating people living with rare disease around how they should expect to be included in research.

Rare diseases – new approaches to diagnosis and care (Medicine Today): An overview of common challenges for people living with rare disease and clinical resources to implement quality rare disease care in clinical practice.

Rare disease management in general practice (HealthEd): Podcast on rare disease care (accredited for RACGP CPD points)

Practical Medical Genomics (UNSW Sydney): A short course run at regular intervals providing specialised knowledge and skills to help health professionals confidently integrate genetics and genomics into daily practice.

2.14 Registries and registry-linked resources relevant to rare disease

Recommendations for a National Approach to Rare Disease Data: Findings from an Audit of Australian Rare Disease Registries (Rare Voices Australia and Monash University): This report explores the landscape

of Australian rare disease registries and databases and includes strategic recommendations and implementation priorities for a national approach to rare disease data collection, co-developed with Rare Voices Australia's Scientific and Medical Advisory Committee and industry representatives through Rare Voices Australia's Round Tale of Companies.

RARE Portal (Rare Voices Australia): Contains links to rare disease registries, where available.

The Australian Paediatric Surveillance Unit at Kids Research (affiliated with the University of Sydney and the Sydney Children's Hospitals Network with a close relationship with the Royal Australasian College of Physicians): A national resource to facilitate active surveillance of rare childhood diseases, complications of common diseases, or adverse effects of treatment.

The International Network of Paediatric Surveillance Units: A network of paediatric surveillance units aiming to advance knowledge of uncommon childhood infections and disorders.

Australian Congenital Anomalies Monitoring System (Australian Institute of Health and Welfare): Captures data on major congenital anomalies from all Australian states and territories except the Northern Territory.

Australian Pregnancy Register for Women on Antiepileptic Medications (The Royal Melbourne Hospital Neuroscience Foundation): An independent, observational register that collects information about pregnant women with epilepsy, treated and untreated, to assist in determining which anti-epileptic medications are safest for the baby while protecting the mother from seizures.

2.15 Rare disease coding

Orphanet Nomenclature for Coding and Associated Tools (European Commission): Information on the importance of rare disease coding and coding guidance.

2.16 Clinical trials

Information about clinical trials for patients and carers (Australian Clinical Trials Alliance): Includes a consumer involvement pack with information for patients and carers who are considering being involved in health research.

Consumer Involvement and Engagement Toolkit (Australian Clinical Trials Alliance): Provides practical advice for researchers and research organisations wishing to conduct patient-centred clinical trials.

Australian Clinical Trials (Australian Government Department of Health and Aged Care): Search for clinical trials being run in Australia and New Zealand; search and/or set an alert to be notified when new clinical trials start. The Consumer Guide to Clinical Trials explains what a clinical trial is, some potential benefits and risks, and questions to ask when considering joining a trial. This video from Sydney Children's Hospitals Network explains how to search for clinical trials on the Australian Clinical Trials website.

ClinicalTrials.gov (USA National Institutes of Health): A comprehensive database of clinical trials in the USA and around the world.

2.17 Advanced therapy medicinal products

Kids Advanced Therapeutics (Sydney Children's Hospitals Network): A program to improve access to advanced therapies (gene therapy, somatic cell therapy, tissue engineering, bacteriophage therapy) for children in Australia.

Gene therapy (Healthdirect; Australian Government Department of Health and Aged Care): Brief overview and links to further information about gene therapy.

Australia's Cell and Gene Catalyst (AusBiotech): Overview of the Cell and Gene Catalyst, an initiative aimed at accelerating the development and commercialisation of cell and gene therapies in Australia.

Approved Cellular and Gene Therapy Products (USA Food and Drug Administration): A list of cell and gene therapy products that are licensed for use in the USA.

Phage Australia (Phage Australia): A national network of phage researchers and clinician scientists developing phage therapy as a treatment for infectious diseases.

The Australian Stem Cell Handbook (National Stem Cell Foundation of Australia, Stem Cells Australia): A publication for patients and carers, with answers to common questions about stem cells, their use in medicine, and their promise for future therapies.

Stem cell treatments and regulation – a quick guide for consumers (Australian Government Department of Health and Aged Care): Detailed information about stem cell therapy, including potential risks.

Brain Aid (University of NSW and Sydney Children's Hospitals Network): A suite of co-designed psychoeducational videos and resources about clinical trials and advanced therapy medicinal products.

2.18 Resources to guide discussions about online and overseas research and therapies

Therapeutic Goods Administration (Australian Government Department of Health and Aged Care): Advice on buying medicines and medical devices online.

Smartraveller (Australian Government Department of Foreign Affairs and Trade): Advice on travel to overseas countries.

2.19 Mental health in people living with rare disease

Applying Mental Health First Aid in a Rare Disease Context (Rare Voices Australia): A companion resource developed in consultation with people living with rare disease to assist those working with the rare disease community to complement Mental Health First Aid Training.

Living with a rare disease: Digital mental health resources (RArEST Project): A fact sheet with digital mental health information for people living with rare disease.

Rare Disease Project ECHO® (Rare Voices Australia, UNSW Sydney, the University of Western Australia and Macquarie University): In the Mental Health and Wellbeing session, Louise Healy from Rare Voices Australia talks about supporting the mental health and wellbeing of people living with rare disease. A summary of key learnings from the session is available.

Head to Health (Australian Government Department of Health and Aged Care): Information about mental health and wellbeing, including digital resources, resources for building coping skills, and pathways for accessing mental health professional support.

Kids Helpline (Australian Government): A free 24/7, confidential and private counselling service for children and young people aged 5 to 25 years. Counselling and support is provided via the phone, web and email.

13Yarn (Australian Government, Lifeline): Culturally appropriate mental health crisis support and referral for Aboriginal and Torres Strait Islander people.

Intellectual Disability Mental Health Connect (Department of Developmental Disability Neuropsychiatry (3DN), UNSW Sydney, and the Council for Intellectual Disability): Aims to help people with intellectual disability get the right services and support for their mental health. It has information for people with intellectual disability, their supporters, and professionals. The website is aimed at people who live in New South Wales. However, people who do not live in New South Wales may still be able to use the information.

Embrace Multicultural Mental Health (Mental Health Australia): Mental health resources and information for people from culturally and linguistically diverse backgrounds.

2.20 Resources for carers and family members

Disability Support Information (Rare Voices Australia): A guide to existing disability, aged care, and carers supports and services.

Carers Gateway website and call centre (Australian Government Department of Social Services): An entry point for carers to access practical information and advice, online supports, and services in their local area.

Siblings Australia (Siblings Australia): A not-for-profit Australian organisation providing resources to support siblings of children and adults with disability.

2.21 Strengths-based and trauma-informed care

Strengths-based Approach: Practice Framework and Practice Handbook (United Kingdom Department of Health and Social Care): Detailed guidance on strengths-based social work with adults, individuals, families, and communities.

Strengths-based Nursing and Healthcare: (Canada, Ingram School of Nursing): This is a framework for health professionals to consider in the delivery of health care. It includes four foundational pillars and eight values and aims to help transform a depersonalized and fragmented health care system into a personal and collaborative model that fosters opportunities for self-healing, engenders hope, and enables patients to draw upon their strengths even in the most difficult circumstances.

Trauma-Informed Care: Best Practice Guidelines (USA Trauma-Informed Care Implementation Resource Center): Website with videos and guidance on implementing trauma-informed care.

2.22 Resources to support information sharing between clinical teams and people living with rare disease

My Health Record (Australian Government; Australian Digital Health Agency): A safe and secure place to keep key health information.

HealthLink (HealthLink Group Limited): Enables the secure electronic delivery of pathology and radiology results, referrals, clinical documents, and discharge summaries between health care professionals.

My Health Matters folder (Council for Intellectual Disability): A communications folder for people with intellectual disability.

The A2D (Admission to Discharge; South Eastern Sydney Local Health District): Folder for people with challenges with verbal communication.

Julian's Key (Queensland Health): A hospital passport for people with intellectual disability.

The Hospital Passport (Developmental Disability Western Australia): A health care passport for people with neurodevelopmental conditions.

2.23 Care coordination services

Paediatric hospitals

Connected Care Program (Queensland Health)

Complex Care Hub (Royal Children's Hospital in Melbourne).

Kids Guided Personalised Services (Kids GPS) Care Coordination (Sydney Children's Hospitals Network)

Connect Care Program for Kids (Perth Children's Hospital)

Rare Care Clinical Centre of Expertise for Rare and Undiagnosed Diseases (Perth Children's Hospital)

Rare Diseases NSW (Randwick Health and Innovation Precinct)

Koorliny Mort (Perth Children's Hospital), a care coordination program for Aboriginal children in Western Australia.

Adult hospitals

Complex Needs Coordination Team (CoNeCT) (Government of Western Australia).

Chronic disease GP Management Plans and Team Care arrangements (Australian Government, Services Australia): A guide for medical practitioners about supporting patients needing chronic disease management, including appropriate Medicare item numbers.

Support coordinators (NDIS): A guide to the different levels of support coordination that may be available under the NDIS including Level 3: Specialist support coordination. This level is for people with more complex situations that require a higher level of support. A specialist support coordinator can assist people to manage challenges in their support environment and ensure a consistent delivery of service.

2.24 Transition care services

New South Wales

NSW Transition Care Network Service (NSW Health Agency for Clinical Innovation): A New South Wales service offering practical help for young people with chronic health conditions transitioning to adult services.

Examples of services (NSW Health Agency for Clinical Innovation): List of transition clinics and services available in New South Wales to support young people, their families, and carers through transition.

Trapeze (Sydney Children's Hospitals Network): Transition from paediatric to adult health for young patients with chronic conditions.

Western Australia

Western Australia Child and Adolescent Health Service (Perth Children's Hospital, Western Australia): Transition to adult health care provides information and resources for adolescents and their families.

Victoria

Transition Support Service (Royal Children's Hospital in Melbourne, Victoria): Assists young people with chronic medical conditions and/or disabilities and their parents and carers to transition and transfer to adult care from the age of 15 onwards.

The Royal Melbourne Hospital Young Adults Transition Service (Royal Melbourne Hospital, Victoria): Provides specialist consultation to adults with congenital disabilities who require ongoing monitoring and care with regards to their physical wellbeing.

Queensland

Nurse Navigator Service (Queensland Health): Helps families of children with complex conditions navigate services across the health care system, including related to transition.

2.25 Transition care resources

Transition Care Network (Agency for Clinical Innovation, NSW Health): Key principles and resources to support successful transition from paediatric to adult services in New South Wales and the Australian Capital Territory.

Healthy WA: My Health in My Hands (WA Health): A three-minute animation aimed at young people residing in Western Australia aged 12 to 18 years to provide support as they become more independent with their health care. Includes a transition readiness checklist for young people and a transition readiness checklist for parents and carers of young people.

2.26 Palliative care resources and services

Palliative care: general

An Overview to Family Meetings and Difficult Conversations (Palliative Care Australia): A guide to holding family meetings and difficult conversations about palliative care.

Palliative Care in Acute Care (Care Search): Guidance to help non-palliative care health professionals provide appropriate palliative care at the end of life.

Palliative care: Aboriginal and Torres Strait Islander people

Palliative Care and End of Life Guidelines (Australian Indigenous Health InfoNet). Guidance for providing person-centred care.

Indigenous Program of Experience in the Palliative Approach (Program of Experience in the Palliative Approach): Grassroots approach to breaking down barriers to palliative care.

Gwandalan (Australian General Practice Accreditation Limited, in partnership with Palliative Care South Australia): Supports palliative care for Aboriginal and Torres Strait Islander communities by providing e-learning modules, webinars, and other resources for frontline staff.

Palliative care: people with intellectual disability

Palliative Care Easy Read resources (NSW Health and NSW Centre for Intellectual Disability): Easy Read resources for people with intellectual disability.

Palliative care: people with culturally and linguistically diverse backgrounds

Multilingual resources (Palliative Care Australia): Resources in 21 languages.

Cultural and linguistic diversity considerations in palliative care (CareSearch Australia; funded by the Australian Government Department of Health and Aged Care): Guides on providing culturally sensitive care to culturally and linguistically diverse communities.

2.27 Resources related to antenatal care and pregnancy planning

Pregnancy Care Guidelines (Australian Government Department of Health and Aged Care): Highlights specific approaches to pregnancy care for a range of groups, with a focus on improving the experience of antenatal care for Aboriginal and Torres Strait Islander women, migrant and refugee women, and women with severe mental illness.

Preparing for your healthy pregnancy (Australian Government Department of Health and Aged Care): Fact sheet with information and tips on preparing for a healthy pregnancy.

Planning for Pregnancy (Royal Australian and New Zealand College of Obstetricians and Gynaecologists): Fact sheet with information about preparing for pregnancy.

MotherSafe (Royal Hospital for Women Foundation): A comprehensive counselling service for women and their health care professionals concerned about exposures during pregnancy and breastfeeding.

Australian Pregnancy Register for Women on Antiepileptic Medication (The Royal Melbourne Hospital Neuroscience Foundation): An independent, observational register that collects information about pregnant women with epilepsy, treated and untreated, to assist in determining which anti-epileptic medications are safest for the baby while protecting the mother from seizures.

Information about pregnancy and health conditions:

- Living with Epilepsy (Epilepsy Action Australia)
- Pregnancy when you have diabetes (Diabetes Australia)
- Are you a woman with diabetes thinking about having a baby? (National Diabetes Services Scheme)
- Having a healthy baby. A guide to planning and managing pregnancy for women with type 1 diabetes (National Diabetes Service Scheme)
- Having a healthy baby. A guide to planning and managing pregnancy for women with type 2 diabetes (National Diabetes Service Scheme)
- Subclinical hypothyroidism and hypothyroidism in pregnancy (Royal Australian and New Zealand College of Obstetricians and Gynaecologists)
- Thyroid disease in the perinatal period (Australian Family Physician).

2.28 Newborn screening and early infantile checks

Newborn bloodspot test (Australian Government Department of Health and Aged Care): Information about newborn bloodspot screening.

Newborn hearing test (Australian Government Department of Health and Aged Care): All Australian states and territories offer a type of hearing screening (using either an automated auditory brainstem response or transient evoked otoacoustic emissions).

Routine child health nurse developmental checks (Healthdirect, Australian Government Department of Health and Aged Care): These checks can detect early signs of a rare disease, as well as other health and developmental conditions. Health checks are recommended at 1 month, 18 months, 2 years, 3 years, and 4 years of age (or before school commencement).

2.29 Immunisation guidelines

Australian Immunisation Handbook (Australian Government Department of Health and Aged Care): The Australian Immunisation Handbook provides clinical guidelines for health care professionals and others about the safest and most effective use of vaccines in their practice.

2.30 Environmental and rare cancer resources

EviQ guidelines (Cancer Institute NSW): Guidelines that can help guide risk assessment and referral for rare cancers, as well as plain English fact sheets for families.

Rare Cancers Australia (Rare Cancers Australia): A not-for-profit organisation aiming to improve awareness, support, and treatment of Australians with rare and less common cancers.

Asbestos support and advocacy groups (Australian Government Asbestos Safety and Eradication Agency): Listing of non-government organisations providing asbestos support and advocacy.

2.31 Reproductive carrier screening, genomic and genetic testing

Reproductive carrier screening (Royal Australian College of General Practitioners): Education about reproductive carrier screening in general practice.

Thinking about having a baby or currently pregnant? (Victorian Clinical Genetics Service): Information about reproductive carrier screening for couples.

Prepair carrier screening (Victorian Clinical Genetics Service): Information about reproductive carrier screening for health professionals.

Understanding genetics (NSW Health Centre for Genetics Education): Information about basic and more complex genetic topics for individuals and health professionals.

Genetic Services listing (NSW Health Centre for Genetics Education): Search for genetic services across Australia.

2.32 Continuing professional development-enabling audits

CPD at the RACGP (RACGP): Information about continuing professional development at the RACGP.

APPENDIX 3: NATIONAL AND INTERNATIONAL SOURCES

Strategies and action plans

Australian Government, National Strategic Action Plan for Rare Diseases (February 2020).

Canadian Organization for Rare Disorders, Canada's Rare Disease Strategy (May 2015).

Reports, statements, and proposals

EURORDIS-Rare Diseases Europe, Position Paper: Achieving Holistic Person-Centred Care to Leave No One Behind (April 2019).

EURORDIS-Rare Diseases Europe, Recommendations from the Rare 2030 Foresight Study (February 2021).

Genetic Alliance UK, Good Diagnosis: Improving the experiences of diagnosis for people with rare conditions (February 2022).

The McKell Institute, Disability & Rare Disease: Towards Person Centred Care for Australians with Rare Diseases (October 2019).

Rare Diseases International, Five Proposals on Global Networking to Strengthen Health Systems for People Living with a Rare Disease [document currently in draft form, personal communication].

United Nations, Addressing the challenges of persons living with a rare disease and their families (December 2021).

Education frameworks

Royal College of Physicians and Surgeons of Canada, CanMEDS (2015).

European Union of Medical Specialists, Syllabus for residents and trainees in Rare and Undiagnosed Diseases (December 2020).

Human Genetics Society of Australasia, Core Capabilities in Genetics & Genomics for Medical Graduates (April 2022).

Royal Australian College of General Practitioners, RACGP 2022 Curriculum core competency framework (2022).

APPENDIX 4: PRIMARY PREVENTIVE MEASURES DURING PREGNANCY

Primary prevention measures related to pregnancy can reduce the incidence of rare diseases. Several primary prevention measures are listed below.

- Educating and supporting all women in their preparation for conception and pregnancy by prescribing folic acid supplementation at least 4 weeks prior to conception and for the first 12 weeks of pregnancy.[144]
- Advising women who are pregnant, planning pregnancy or breastfeeding to take an iodine supplement of 150 micrograms (µg) each day.[145]
- Educating women of child-bearing age about the benefits of vaccination against rubella, measles, mumps, varicella, and hepatitis B prior to conception to prevent the development of congenital abnormalities.[144]
- Modifying diet and lifestyle risk factors associated with adverse pregnancy outcomes, such as smoking, alcohol consumption, and being overweight or obese. Lifestyle modification examples include smoking cessation and alcohol programs for pregnant women.
- Identifying and acting on factors that increase the chance of mothers having babies with rare congenital anomalies, including epilepsy, diabetes, and thyroid disorders, and providing specific preconception and pregnancy care.
  - For most women with diabetes, epilepsy, BMI>30 kg/m^2 or who have had a baby with a previous neural tube defect, 5 mg of folic acid/day should be taken for at least one month before conception and throughout the first trimester.[144]
  - Diabetes treatment should be reviewed before (or early in) pregnancy to have the best control.[146-149]
  - Women at high risk of gestational diabetes should have an early oral glucose tolerance test to identify and manage this if present.
  - Epilepsy treatment should be reviewed before pregnancy to have the best control on the lowest but still effective dose of suitable medication.[150,151]
  - Women with a personal history of thyroid disease, type 1 diabetes or symptoms of thyroid disease should be checked. Overt hypothyroidism should be treated in pregnancy (noting pregnancy-specific reference ranges).[145]
- Testing women with risk factors for deficiencies for Vitamin B12, iron, and Vitamin D, and prescribing supplementation if they are deficient.[144]
- Educating women about the recommended dietary and supplement restrictions in pregnancy (for example, avoiding mercury-containing fish).[144,152]
- Discussing TORCH infections (toxoplasmosis, other [for example, syphilis, varicella, mumps, parvovirus, HIV, listeriosis], rubella, cytomegalovirus and herpes simplex), including methods to reduce exposure and transmission.[144]
- Screening, and where required, treating women for infectious diseases in pregnancy, including HIV, hepatitis B, hepatitis C, and syphilis.[144,153]
- Screening for medication use in pregnancy and advising on cessation or avoidance of potentially teratogenic medications.[144]

APPENDIX 5: DEVELOPMENT OF THE NATIONAL RECOMMENDATIONS FOR RARE DISEASE HEALTH CARE

The Recommendations were developed as part of the Rare Disease Awareness, Education, Support and Training (RArEST) Project, which was awarded $1.9 million in funding from the Australian Government.

Research

Initial research was conducted to identify relevant standards, frameworks, and other publications. These are listed in Appendix 3. These documents were used to shape initial drafts. Subsequent versions were cross-checked to ensure continued alignment.

Consultation

The Recommendations were developed in close consultation with education experts, health professionals, and people living with rare disease. The following groups were consulted on the framing, development, and endorsement of the Recommendations.

RArEST Project Steering Committee	Health, education, policy, and rare disease experts from: ▪ UNSW Sydney, Faculty of Medicine and Health ▪ Rare Voices Australia ▪ University of Western Australia, School of Paediatrics and Child Health ▪ Rare Care Clinical Centre of Expertise for Rare and Undiagnosed Diseases, Perth Children's Hospital ▪ Macquarie University, Australian Institute of Health Innovation
RArEST Project Stakeholder Reference Group	Twelve people with lived experience of rare disease, with representation from priority populations, including: ▪ Aboriginal and Torres Strait Islander people ▪ People experiencing socioeconomic hardship ▪ People from culturally and linguistically diverse backgrounds ▪ People living in regional, rural, or remote communities ▪ People suspected of having an undiagnosed rare disease
RArEST Project (Stream 2) Health Professional Education and Training Working Group	Health and education professionals, including nurses, general practitioners, and medical specialists from: ▪ UNSW Sydney ▪ Rare Voices Australia ▪ University of Western Australia, School of Paediatrics and Child Health ▪ Rare Care Clinical Centre of Expertise for Rare and Undiagnosed Diseases, Perth Children's Hospital ▪ Australian Genomics

This document was also reviewed by the following organisations:

- Australian College of Children & Young People Nurses
- Australian College of Nurse Practitioners
- Australian College of Rural and Remote Medicine
- Australian Genomics
- Australian Primary Health Care Nurses Association
- Australian Rheumatology Association
- Human Genetics Society of Australasia
- The Royal Australian College of General Practitioners
- Rural Doctors Association of Australia

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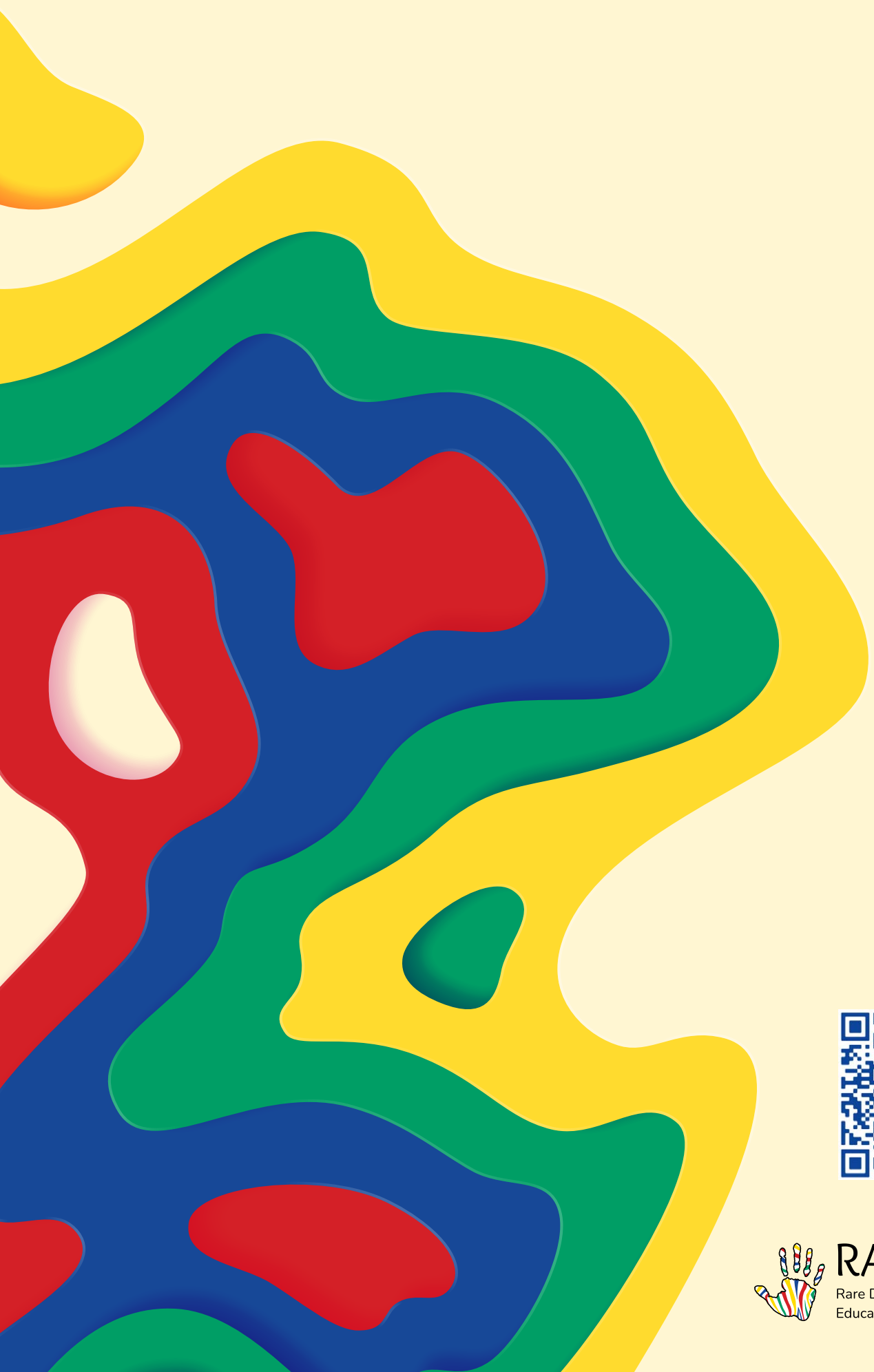

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