RArEST
Rare Disease Awareness, Education, Support, and Training

National Recommendations for

Rare Disease Health Care

SUMMARY

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National Recommendations for Rare Disease Health Care

With over 7,000 different conditions, rare diseases are collectively common, affecting over 2 million Australians. These eight recommendations can help health professionals provide quality care for people living with rare and undiagnosed conditions, including families and carers.

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

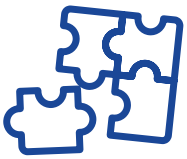
Promote integrated and coordinated care across the lifespan as people living with rare disease require a wide range of health and support services.

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

About rare disease

- A rare disease is one that affects fewer than 1 in 2,000 people.[1,2]
- There are over 7,000 known rare diseases,[1,2] and around 2 million Australians live with a rare disease.[3]
- Most rare diseases are genetic (caused by changes in a person's genetic makeup).[4,5]
- Other types of rare disease include cancers, infectious diseases, and autoimmune disorders.[2]

Caring for people living with rare disease: Because each rare disease is so rare, very few health professionals – if any – may have seen and treated people with each condition before. This makes it hard for health professionals to know how to provide the best care.

About the Recommendations: The National Recommendations for Rare Disease Health Care were written to help health professionals provide better care for people living with rare disease including:

- People who know which rare disease they have (diagnosed people)
- People who do not yet know which disease they have (undiagnosed people).

They may also be helpful for rare disease organisations and people living with rare disease, to help them partner with their health care professional to plan the type of health care that works best for them.

This is a **summary** of the Recommendations. The **full Recommendations** can be found on Rare Voices Australia's website. For each recommendation, resources are included to help health professionals put the recommendation into practice. These Enablers of Good Practice are included at the end of this Summary. They can also be found in Appendix 2 in the full Recommendations document.

This summary was written for many different audiences, including health professionals, both experienced and in training, people living with rare disease and their families, and health care executives and policy makers. Experienced health professionals will often already be practicing in alignment with many of the recommendations.

The Australian Government's National Strategic Action Plan for Rare Diseases (the Action Plan) outlines the priorities, actions, and steps required to achieve the best possible health and wellbeing outcomes for people living with rare disease.

It is recognised that there are many barriers to delivering and receiving high quality care, which need to be addressed. These include:

- **Lack of time:** Health professionals may not have enough time to take part in education, collaboration, or research, or time to think about and improve the way they deliver health care.
- **Lack of resources:** There may not be enough health care resources (for example, doctors, hospitals, clinics) and they may not be available to all people.
- **Problems with the health care system:** The health systems and methods of providing financial support may not meet the needs of people living with rare disease.
- **Small numbers of people with rare disease:** As few people in Australia live with each rare disease, it is hard to build networks of health professionals and patient organisations.

The responsibility for delivering rare disease health care does not rest on the shoulders of individual health professionals alone.

Indicators of good practice: Each Recommendation includes indicators of good practice. Health professionals can use these indicators to quickly learn what they can do to provide better care to their patients, how people living with rare disease experience good practice, and how they can see if they are providing care in a way that would be considered good practice.

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

How the Recommendations were written: The Recommendations were written as part of the Rare Disease Awareness, Education, Support and Training (RArEST) project, which was funded by the Australian Government. The Recommendations were written with the help of education experts, health professionals, and people living with rare disease.

The Recommendations have been reviewed and endorsed by:

The full Recommendations document has been endorsed by APNA according to approved quality standards criteria. Completion of this resource entitles eligible participants to claim 2 CPD hours

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

In this document we use the term 'people living with rare disease'. This refers to people who have a rare disease AND their families and carers.

Next steps

It is expected that the Recommendations will be the first step towards co-developing national guidelines for rare disease care and support. National guidelines will require:

- Further evidence to make them stronger
- Changes to be made to the health system
- Building on existing networks and collaborations
- Ongoing close partnerships with people living with rare disease.

The guidelines will need to be a living document to respond to the rapid changes and innovations in the rare disease sector.

1

Recommendations

More details and resources to help with applying these recommendations and sub-recommendations are provided in the full 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.

SUB-RECOMMENDATIONS

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.

People living with rare disease often become experts in their own conditions and have complex needs that change over their lifetimes. Although there are shared symptoms and approaches for rare diseases, for a model of care to be effective and equitable, the individual needs and preferences of a person, their family, and community need to be recognised and addressed.

Person-centred care involves:

- Sharing decision-making, which can empower people living with rare disease to be active partners in their own care. With this approach, the health professional and people living with rare disease make decisions together, having discussed all the options.
- Partnering with people living with rare disease in both their diagnosis and care, including:
  - Recognising the uncertainty that comes with being undiagnosed or living with a rare condition
  - Acknowledging that people living with rare disease may know more about their condition than their health care team
  - Encouraging and supporting partnerships with other health professionals
  - Empowering people living with rare disease to be advocates and active participants in decision making.
- Sharing tailored, reliable, and understandable information once a diagnosis is found. Resources like those found on the Rare Awareness Rare Education (RARE) portal or via the RARE helpline can help reduce uncertainty and build health literacy.
- Encouraging people living with rare disease to share their goals and giving them a clear plan of steps to take after each appointment.

These approaches all align with the preference of people living with rare disease (see Figure 1).

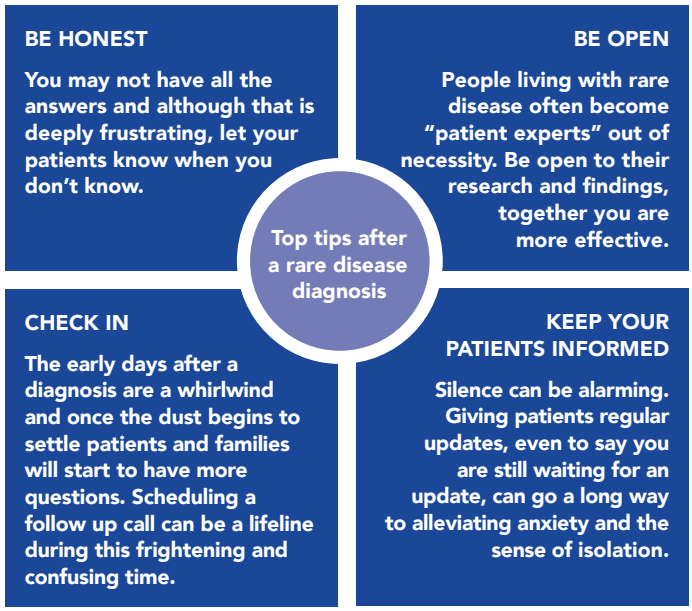


Figure 1: Tips for health professionals from people living with rare disease.[6]

As many people with rare disease also have a disability, health professionals can consider how to embed reasonable adjustments. For example, providing rare disease information in different formats such as Easy Read, allowing extra time for appointments, and having clinic rooms that are wheelchair accessible.

Measures to optimise flexibility in the delivery of rare disease care can include:

- Allowing more than one family member to attend appointments
- Considering new ways of caring for patients. For example, having a regular clinic in a place that is easy for people living with rare disease to get to, such as within existing community health spaces
- Using digital solutions such as telehealth to help more people access expert rare disease care (for example, from a rare disease Centre of Expertise), and making it possible for health professionals from different locations to join the one appointment
- Inviting other health professionals involved in the person's care to their appointments.

Person-centred care is particularly important for priority populations, as defined in the Action Plan. These populations include:

- People living with an undiagnosed rare disease
- People with an increased chance of developing a rare disease or having a child with a rare disease
- Aboriginal and Torres Strait Islander people
- People from culturally and linguistically diverse (non-English speaking) backgrounds
- People experiencing socioeconomic disadvantage
- People living in regional, rural, and remote areas.

It is important that health care is culturally safe for Aboriginal and Torres Strait Islander people, and those from culturally and linguistically diverse backgrounds. To provide culturally safe care, health professionals must be flexible and open to tailoring the care they provide. Useful resources are listed in the Enablers of Good Practice. For example, the translation web platform Lyfe Languages, which includes culturally appropriate information on rare diseases.

People living with rare disease in rural, regional, and remote areas do not have the same access to care as people in cities. Many strategies can be used to improve access, such as testing that happens at or near the place they receive their health care, virtual clinics, telehealth, mobile clinics, and outreach programs. Linking individuals with available services and resources can improve wellbeing and support preventive measures, enhance timely diagnosis and treatment, and reduce hospital presentations.

It is also important to ensure care is safe and inclusive for individuals who are gender and sexually diverse. Further information is included in the Enablers of Good Practice at the end of this Summary.

RECOMMENDATION 1

1

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

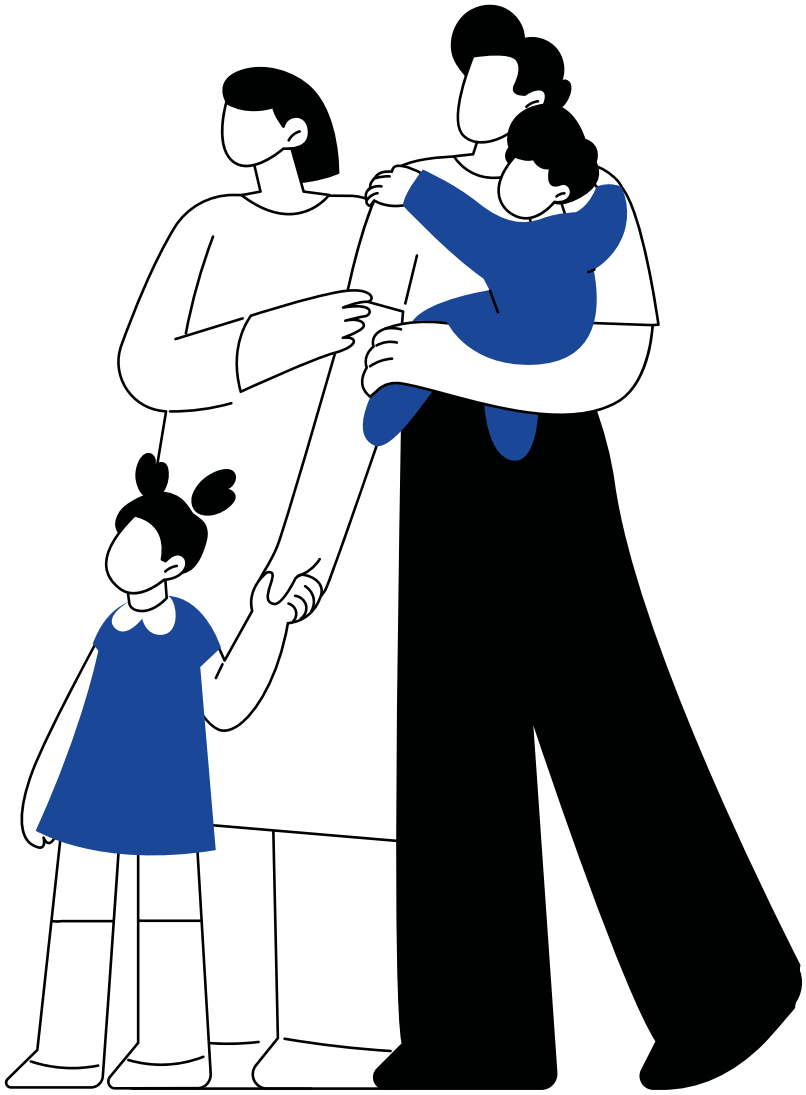
- High scores in Patient-Reported Experience Measures (PREMS), such as those included in the Australian Hospital Patient Experience Question Set from the Australian Commission on Safety and Quality in Health Care,[7] which can be adapted to most settings
- People living with rare disease report that 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

Resources for health professionals

For useful tools and supports to help implement Recommendation 1, see Enablers of Good Practice Sections 2.1-2.9. A useful resource is Rare Disease 101 Australia. This is a free e-learning module developed by the RArEST team and hosted on the Medics for Rare Disease website. Lesson 4 explains the importance of respectful communication with all Australians. It includes case studies and resources to help health professionals deliver person-centered care and shared decision making.



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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.

SUB-RECOMMENDATIONS

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.

Steps to achieving a quick and correct diagnosis include:

- Recognising that rare diseases are often complex and involve multiple organ symptoms. Carefully review a person's current and previous medical history. Be open to the possibility of connections between symptoms that at first may seem unrelated. Figure 2 shows 'red flags' that can indicate a person might have a genetic condition.
- Following established protocols and pathways. This may involve referring the person for tests or to other health professionals. For example, to specialists in rare or genetic conditions, such as a clinical geneticist or a rare disease Centre of Expertise (See also Recommendation 3.1). The RARE Portal and RARE Helpline can direct to these experts.
- Using online tools and databases (such as Orphanet's Clinical Signs App, Find Zebra, and PubCaseFinder, described in the Enablers of Good Practice) to link symptoms and test results to a diagnosis.

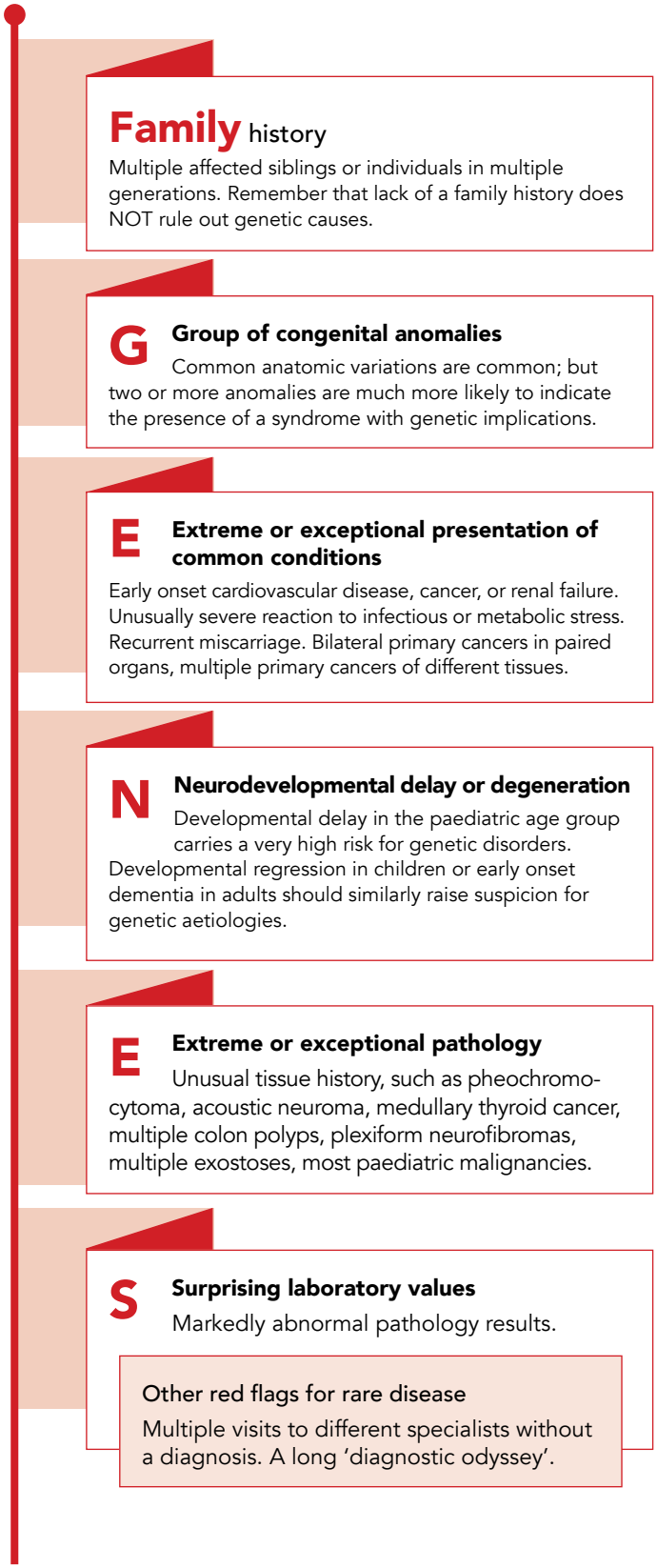

Figure 2: Family GENES tool. Adapted from the Genetics in Primary Care Faculty Development Initiative[8]

People with a suspected but undiagnosed rare disease need:

- Medical care that acknowledges and treats their known symptoms. This may involve referral to specialist health professionals
- To be asked about their mental health and supports offered if needed
- To be offered help in accessing practical and financial support, for example, the National Disability Insurance Scheme (NDIS)
- To be connected with support groups for undiagnosed families
- To be connected with undiagnosed disease pathways. For example, an Undiagnosed Diseases Program, which can usually be accessed through clinical genetics units.

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

Resources for health professionals

For useful tools and supports to help health professionals act on Recommendation 2, see Enablers of Good Practice Sections 2.1, 2.2, 2.10-2.12. Lesson 5 of the free online e-learning module Rare Disease 101 Australia explains how it is important for health professionals to recognise and acknowledge that many people with rare disease are on a 'diagnostic odyssey'. This is the long and often complex path to a diagnosis. It also provides tools that health professionals can use to help them find a diagnosis. Lesson 6 explains genetics (the study of genes), genomics (the study of all of our DNA), and what an undiagnosed disease program is and how to access one.

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.

SUB-RECOMMENDATIONS

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.

No one can be an expert in over 7,000 rare diseases, so sharing of knowledge between health professionals can be very helpful. This may involve:

- Consulting with and referring people living with rare disease to rare disease experts and Centres of Expertise, both in and out of Australia. These can be identified through the RARE Portal in Australia, and through international portals such as Orphanet and the Genetic and Rare Diseases Information Center (GARD; see Enablers of Good Practice).
  - A rare disease Centre of Expertise is a place that specialises in managing and caring for people with rare disease. Many different types of health professionals with expertise in rare disease work at these centres. Although there is no common definition of a rare disease Centre of Expertise, most will be:
    - Focussed on diagnosing and managing rare diseases and providing coordinated care
    - Involved in research, collaboration, data sharing, and education
    - Working with people living with rare disease to continually refine and improve the way they are cared for.
- Encouraging people with rare disease to connect with registries and natural history studies. This gives them the chance to help grow the knowledge base about their own rare disease. Centres of Expertise, rare disease organisations, and research and advocacy groups can provide information on registries and natural history studies. More information on current Australian rare disease registries and natural history studies can also be found in Recommendations for a National Approach to Rare Disease Data: Findings from an Audit of Australian Rare Disease Registries.
- Using clinical practice guidelines and standards of care where possible. These may be from Australia or other countries (see Enablers of Good Practice).

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

Resources for health professionals

For useful tools and supports to help health professionals act on Recommendation 3, see Enablers of Good Practice Sections 2.2, 2.3, 2.13-2.15. Lesson 7 of the free online e-learning module Rare Disease 101 Australia outlines the importance of coordinated care across the lifespan.

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.

SUB-RECOMMENDATIONS

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 therapy medicinal products.

Curative treatment is available for less than 5% of rare diseases. However, research into rare diseases and their treatment is progressing rapidly.

To help people living with rare disease access and benefit from this research and treatment:

- Learn from, contribute to, and connect people living with rare disease to rare disease and patient advocacy groups. These groups (also known as patient support organisations) support and provide information to people living with rare disease. They can also drive research and work for better access to care, support, and treatments.
  - 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. For example, many organisations organise events for siblings and groups for parents/carers.
- Find, join, and connect people with rare disease to research. This research includes studies to improve our knowledge of the causes of rare diseases, how they may progress, and how to optimally care for and treat them.
- Help people living with rare disease to safely access innovative treatments. For example, advanced therapies, which are medicines based on genes, tissues, or cells.

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

Resources for health professionals

For useful tools and supports to help health professionals act on Recommendation 4, see Enablers of Good Practice Sections 2.1, 2.16-2.18. Lesson 8 of the free online e-learning module Rare Disease 101 Australia is about the importance of partnering with rare disease and patient advocacy groups.

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.

SUB-RECOMMENDATIONS

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.

People living with rare disease have an increased risk of developing mental health issues, including anxiety, isolation, loneliness, stress, grief, and loss of hope. Many people living with rare disease experience trauma during their health journey, which can further impact their mental health.

It is critical that health professionals act early and often to help people living with rare disease and their families/carers with the mental heath challenges they face. Health professionals can offer management and support in line with the preferences of each individual.

Health professionals can:

- Be aware of and learn about the way rare diseases can impact mental health and wellbeing
- Ask sensitively about mental health and wellbeing at all appointments
- Let the person know it's okay to feel the way they do
- Recommend suitable resources, support, and referrals, particularly to mental health professionals who have worked with people living with rare disease or who have had relevant training
- Ask whether people living with rare disease are receiving appropriate financial support (for example, disability, aged care, and carer's supports). If not, help them to access this funding, which may involve referring to social workers where possible. The Disability Support Information section in the RARE portal includes key information about the NDIS, aged care, disability and carer supports and resources.
- Deliver strengths-based and trauma-informed care
  - A strengths-based approach focuses on the unique strengths, abilities, and resources of an individual and their support network
  - Trauma-informed care is a related approach that considers what has happened to a person rather than what is 'wrong' with them. It focuses on building trust and partnerships.

Rare Voices Australia, as part of the RArEST project, have developed educational resources to help support the mental health of people living with rare disease and their families/carers. These resources are included in the Enablers of Good Practice Section 2.19.

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

Resources for health professionals

For useful tools and supports to help health professionals act on Recommendation 5, see Enablers of Good Practice Sections 2.2, 2.19-2.21. Lesson 3 of the free online e-learning module Rare Disease 101 Australia focusses on mental health and wellbeing in people living with rare disease.

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.

SUB-RECOMMENDATIONS

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.

People living with rare disease see a wide range of health and support services, so assistance in accessing integrated and coordinated care across their lifespans is helpful. This involves coordinating and connecting health services with other sectors including disability, education, and employment (cross-sectoral care).

People with rare diseases can be encouraged to give consent for uploading their health care records to My Health Record to facilitate sharing of up to date health care records within their multi-sector teams. These teams may include people from a range of different professions (for example, physicians, general practitioners (GPs), rural generalists, nurses, genetic counsellors, health workers, teachers, and welfare officers).

Primary health practitioners (for example, GPs and rural generalists) can play an important central role in care coordination. For example, GPs can make sure evidence-based care is provided. Specific Medicare item numbers (for example, items 721 and 723) can be used for longer appointments to help with chronic and complex care planning and management. Funding may be available within the NDIS to help coordinate community and clinical appointments and support services for people with more complex needs.

A growing number of care coordination services are available to support people living with rare disease to manage their care. Currently, these services are generally based in children's hospitals (see Enablers of Good Practice).

These services can help with:

- Organising appointments and referrals to specialists, allied health professionals (for example, physiotherapists) and support services
- 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. Details can be found in the Disability Support Information page on the RARE Portal.
- Providing information and education.

Care coordination can also be facilitated by rare disease Centres of Expertise (listed in the RARE Portal). 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.

Health professionals can offer assistance to facilitate successful transition between different life stages. For example:

- Moving from paediatric (child) to adult care
- Entering school, university, and employment
- End of life care.

Planning for the transition from paediatric to adult health services is best started in the early teenage years. Primary health practitioners (for example, the GP) and specialist services are important supports. People with life-limiting conditions require early referrals to palliative care services to allow time for them to build relationships and trust with their palliative care team. Specialist transition and palliative care services, suggestions for successful transition frameworks, and how to start discussions about palliative care can be found in the Enablers of Good Practice Sections 2.24-2.26.

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

Resources for health professionals

For useful tools and supports to help health professionals act on Recommendation 6, see Enablers of Good Practice Sections 2.22-2.26. Lesson 7 of the free online e-learning module Rare Disease 101 Australia is about the importance of care coordination across the lifespan. It includes relevant case studies, and links to useful resources and frameworks.

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.

SUB-RECOMMENDATIONS

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.

Health promotion is the process of encouraging and helping people to increase control over, and to improve, their health. As some rare diseases can be prevented, it is important to help people understand and access appropriate screening and technologies.

This may include:

- Providing thorough prenatal and pregnancy care. This includes working with individuals and couples to make shared decisions about tests and screens that can be done before and during pregnancy
- Newborn screening programs and child health nurse checks
- Offering immunisation and occupational health and safety screens.

It is also important that health professionals help with collecting data, such as contributing to registries for rare diseases, and registries for conditions that develop before the baby is born (congenital anomalies).

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

Resources for health professionals

For useful tools and supports to help health professionals act on Recommendation 7, see Enablers of Good Practice Sections 2.14, 2.27-2.31, and Appendix 4 from the full Recommendations document.

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.

SUB-RECOMMENDATIONS

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.

Continuing to learn, reflect, and improve is important, particularly for health professionals who care for people with rare diseases. This is because being managed by a health professional who is knowledgeable and actively involved in care can improve outcomes for people living with rare disease.

For health professionals this may involve:

- Taking part in ongoing learning in rare diseases and in aspects of clinical practice that are relevant to rare disease. For example, attending conferences, learning from colleagues, and online learning.
  - The free e-learning module, Rare Disease 101 Australia was designed by the RArEST team to share the lived experiences of people with rare disease. It also offers solutions, resources, and frameworks to address common challenges faced by people living with rare disease.
  - The Royal Australian College of General Practitioners (RACGP) *check* interactive case on rare disease (Unit 607) was developed by the RArEST team and includes case-based learning on rare disease.
  - Both Rare Disease 101 Australia and *check* are accredited for RACGP Continuing Professional Development points. Other educational resources, including free e-learning modules, are included in the Enablers of Good Practice section.
- Reflecting on how they care for people living with rare disease and identifying further learning needs.
- Asking for feedback from people living with rare disease, colleagues, and other people they work with. This may be done by setting up ongoing methods of capturing feedback or regularly reviewing patients with undiagnosed disease to identify ways of helping them better.

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

Resources for health professionals

For useful tools and supports to help health professionals act on Recommendation 8, see Enablers of Good Practice Sections 2.13, 2.32.

Appendices

APPENDIX 1: ABBREVIATIONS

CPD	Continuing Professional Development
GARD	Genetic and Rare Disease Information Center
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
UDNI	Undiagnosed Diseases Network International

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 continuing professional development (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.

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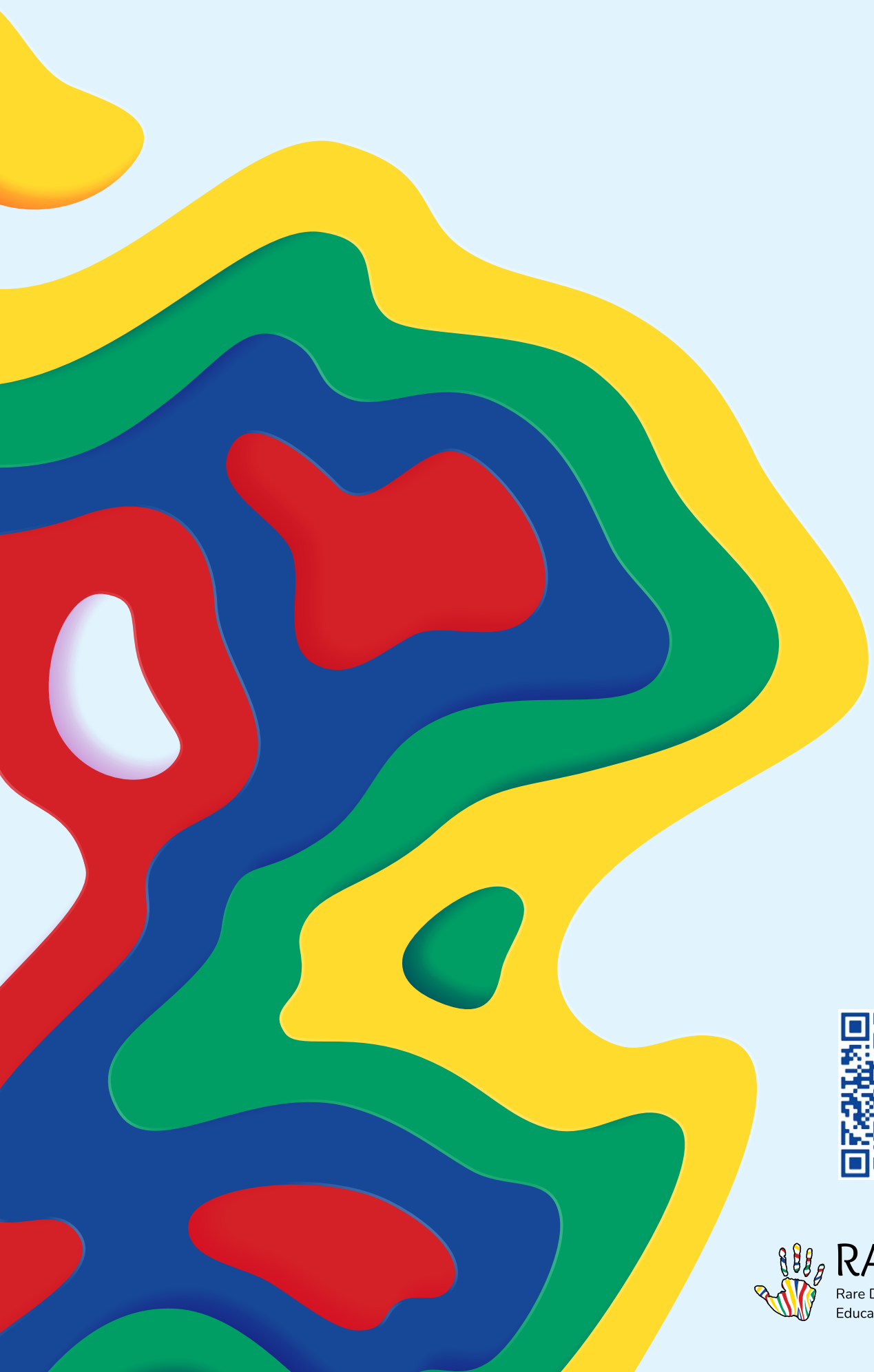

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