



RAReST

Rare Disease
Awareness, Education,
Support, and Training



Key learnings from Rare Disease Project ECHO®

Series 3: Session 1 (19 September 2024) – Mental Health and Wellbeing

Opening presentation

Louise Healy, Rare Voices Australia's Education and Advocacy Manager, gave a presentation on the mental health and wellbeing challenges that people living with a rare disease often experience, along with resources that can help address these challenges.

People with a rare disease and their families and carers experience higher levels of anxiety, stress, low mood and emotional exhaustion compared with the general population. Poorer mental health and wellbeing is influenced by many factors, including:

- Feelings of isolation and loneliness
- A prolonged diagnostic journey and misdiagnoses
- Needing to educate health professionals
- Not being taken seriously by health professionals
- Lack of treatment options
- Ongoing and anticipatory grief
- Poor care coordination
- Loss of hope.

However, very few people living with a rare disease and their families/carers are offered mental health support. Health professionals can help support the mental health and wellbeing of people living with a rare disease by:

- Following the [National Recommendations for Rare Disease Health Care](#), especially [Recommendation 5: Mental health, social and emotional wellbeing](#)
- Learning about the mental health needs and challenges of people with lived experience of rare disease and what is most helpful by viewing the [Supporting the mental health and wellbeing of people living with a rare disease](#) video case studies
- Reducing stigma by discussing and normalising the mental health and wellbeing challenges experienced by people living with a rare disease
- Practicing shared decision-making and partnering with families
- Referring people living with a rare disease and their families/carers to a mental health professional.

Resources and information to support people living with a rare disease include:

- [Digital mental health fact sheet](#): A resource developed for Australians living with a rare disease, including their families and carers. It provides information about

mental health and wellbeing and links to digital mental health supports. The fact sheets are also available in [Easy Read](#) and [Plain language](#) formats

- [Rare disease support organisations](#): Contact details for a range of rare disease support groups
- [Rare Portal](#): Evidence-based rare disease information for the Australian context
- [RARE Helpline](#): A service navigation and information helpline that provides information and support for people living with a rare disease
- [Mental health and rare disease resources](#) for consumers and health care professionals living in Western Australia
- [Resources for priority populations](#) identified in the [National Strategic Action Plan for Rare Diseases](#).

Rare disease presentation

Stephanie Broley, Senior Genetic Counsellor at King Edward Memorial Hospital and Perth Children's Hospital presented the case of a 2-year-old girl with a rare genetic epilepsy condition. Despite trialling multiple anti-epileptic medications, the child has experienced many episodes of status epilepticus, and has complex care needs, with severe global developmental delay, feeding difficulties, failure to thrive, and minimal social interaction. She has increased muscle tone and is not able to crawl or stand independently.

This ultra-rare condition has impacted the child and her family in many ways. The condition's rarity has made it difficult to obtain information about management and prognosis, while a focus on urgent medical care has led to difficulties with bonding and engagement and placed stress on family relationships.

The child's minimal developmental progress, and uncertainty about the level of function and communication she might achieve, along with the prospect of lifelong care, has placed a great deal of stress on the family. In addition, with complex care needs, family life revolves around the child, and she requires full time care by her mother. Together, these affect parental coping and adjustment, and cause anger, frustration, grief, loss, constant worry, uncertainty, avoidance of social networks, and relationship issues.

Discussion with the community of clinical learning practice

The group discussed:

- The limitations of the current Australian fee-for-service model.
- The proposed establishment of rare disease Centres of Expertise across Australia to support rare disease families and provide more integrated healthcare.
- The frustrations of families and caregivers in dealing with the current system.
- That innovative ideas are needed to address the many challenges.

Key recommendations that Project ECHO® participants and experts shared for supporting the mental health and wellbeing of people living with a rare disease were:

- Being aware of the mental health challenges associated with rare disease
- Applying Section 5 of the [National Recommendations for Rare Disease Health Care](#)
- Sharing the resources below with families and carers.

Useful resources for clinicians and patients discussed this session were:

- [Digital mental health fact sheet](#), also in [Easy Read](#) and [Plain language](#) formats
- [Supporting the mental health and wellbeing of people living with a rare disease](#) video case studies
- [Rare disease support organisations](#)
- [Rare Portal](#)
- [RARE Helpline](#)
- [Mental health and rare disease resources](#) for consumers and health care professionals living in Western Australia
- [Resources for priority populations](#)
- Family/carer support courses run by [Kiind](#) in WA

You may be interested in information on mental health and wellbeing in the free [Rare Disease 101 Australia e-learning module](#).