



**RAReST**

Rare Disease  
Awareness, Education,  
Support, and Training



## Key learnings from Rare Disease Project ECHO®

### Series 1: Session 1 - Rare Disease Facts and Figures and the Importance of Lived Experience

#### Opening presentation

Nicole Millis, CEO of Rare Voices Australia, presented on the topic of Rare Disease: The Narratives Behind Facts and Figures.

You can find a recording of the presentation [here](#).

#### Case presentation

A GP presented a case of a woman who received a rare cardiological diagnosis later in life. The presenter asked for advice on:

- Communicating with patients when there is clinical uncertainty in management of the rare disease
- Involving the patient in shared decision making, particularly where the options for management are not clear
- Providing resources for patients on their rare disease—education and support

#### Case discussion

The discussion focused on how important it was to recognise the key theme of *uncertainty* in rare disease care. To acknowledge that when a patient or carer receives a rare diagnosis, with limited or no treatment options currently available, that this is outside most people's perceptions of health care and has a profound impact. That not only does this situation result in discomfort for patients and carers, but also for many health care workers. That this situation may be especially challenging for people from culturally and linguistically diverse communities who may commonly have the expectation that 'the doctor is always right'.

A key experience that was shared from a participant was as followed...

*"I was incredibly moved when my GP said, 'I know nothing about your condition, but I can ask, I can advocate, and I can beg.'"*

There was agreement that this honesty and openness to shared discovery about a rare disease between the health care provider and patient or carer was very refreshing. Also, that although people living with a rare disease are often referred to as experts in the rare disease, they are also beginners and learners about the rare disease, and that that journey would be less isolating and frightening if shared with their health care providers.

The multi-faceted impacts of rare diseases were discussed, including impacts on self-identity and mental health and wellbeing. This led to a recognition of the importance of a multidisciplinary approach – for example here involving the GP, who had an excellent knowledge of the patient's past health care and treatment, a psychiatrist, specialist cardiologist and cardiology nursing team, and the patient herself.

The importance of an authentic continuing relationship between a health care provider and person living with a rare disease was also discussed. It was discussed that it is important that people living with rare disease know that their health care providers will check back in with them and are flexible to revisit key decisions along a shared decision-making path.

**Key recommendations** that the Project ECHO® participants and experts shared at the end of this case-based discussion were:

- To acknowledge and think about approaches to uncertainty,
- To consider multidisciplinary approaches and shared decision making, and
- The importance of honesty and an ongoing partnership between those living with a rare disease and their health care providers to deliver person-centered care.

**Some key resources** discussed over this ECHO session were

- [National Strategic Action Plan for Rare Diseases](#)
- [Rare Voices Australia](#) website which will host the [RARE Portal](#)
- [RD 101 Australia](#) e-learning platform for rare disease training for medical professionals: first two lessons now available on introduction to Rare Disease and Understanding the Common Challenges with 8 new lessons becoming available over the next 6 months. Lesson 4 focuses on effective communication with many key issues that were discussed today. The full UK version and other modules are available too
- Rare Revolution Magazine [Top 10 Tips](#) for RARE Healthcare Providers
- [Family GENES](#) concept for red flags for rare disease

**Thank you to all who attended. Please don't forget to let us know how we did so we can continue to improve!**

Survey link: [Qualtrics Survey | Qualtrics Experience Management](#)