



RAReST

Rare Disease
Awareness, Education,
Support, and Training



Key learnings from Rare Disease Project ECHO®

Series 2: Session 2 (24 August 2023) – Communication and Empowerment

Rare disease presentation

Manjekah Dunn, a junior doctor at Sydney Children’s Hospital Westmead, gave a presentation about patient communication and empowerment, using the example of Tracey*, a 43-year-old woman who presented at the emergency department with left ankle pain.

Tracey had been diagnosed with the rare neurodevelopmental condition [CASK-associated syndrome](#), epilepsy, type 2 diabetes mellitus, and asthma. She was accompanied by a support worker from her group home.

After learning about Tracey’s rare disease, the treating clinicians made several adjustments to her care, including quickly learning about the condition by looking up the [RARE portal entry on CASK-associated conditions](#), giving Tracey the option to include her support person in the consultation, speaking directly to Tracey, using simple language, and providing an Easy Read version of the discharge summary for Tracey to keep and to share with her health care providers, using the [Council for Intellectual Disability Easy Read health letter templates](#).

Box 1 below highlights some of the main ways care was tailored to be more person-centred for Tracey.

Discussion with community of clinical learning practice

The discussion covered:

- The requirement of all Australian health professionals to make reasonable adjustments (see resources list below) for people with disabilities, as Australia is a signatory to the [UN Convention on the Rights of Persons with Disabilities](#) and aligns with the [Australian Charter of Healthcare Rights](#).
- The value of reasonable adjustments, such as using simple language, repeating the main points/take home messages, checking understanding using the Teach Back technique (see resources list below), and offering an accessible written summary (ideally in simple English or Easy Read).
- Why it is important to ask people if they want a support person with them rather than assuming yes or no, especially given the frequent lived experience of trauma for people with disabilities, as highlighted in the recent [Royal Commission into Violence, Abuse, Neglect and Exploitation of People with Disability](#).
- How important it is to be open if you have never heard of a rare condition (which is often the case given there are over 7000 rare conditions!) but also to be curious and review information that may be shared by the person living with rare disease, or use key information websites such as the [RARE portal](#) (see resources list below).
- The value of connecting people with others who have the same condition to help them feel less alone, which can be done via rare disease patient organisations.

*pseudonym

- The value of using social stories to explain what is going to happen, for example with a child with a rare disease. Social stories are a way of describing a scenario the person will encounter using pictures and drawings. Templates for social stories are freely available online (see resources list below). People with autism often value a social story to help reduce anticipatory anxiety. For people of all ages, using common and simple language is helpful.
- Considering how the health care environment you work in may impact people with autism or people with intellectual disability, and adjust where possible, for example, finding a quiet spot or a room away from the noise and bustle.

Key recommendations that the Project ECHO® participants and experts shared were:

- Speak to the person, not their condition, and make reasonable adjustments so they can equitably access and utilise care.
- Speak directly to people with intellectual disability (not just their support person) using short, simple sentences.
- Consider why you are writing a health letter, and that it should be understandable and useful. Write in plain English and, if preferred by an individual, include an Easy Read version.

Useful resources for clinicians and patients shared this session were:

- The [RARE portal](#), which provides information about individual rare diseases and where to find reliable information.
- [GeneReviews](#), a point-of-care resource giving information and management guidelines on rare genetic conditions.
- [Say Less, Show more](#) resources for children with intellectual disability.
- Guide to the [teach-back technique](#).
- [My Health Matters](#) folder for people with intellectual disability.
- [GeneEQUAL Educational toolkit](#): Communicating with people who have intellectual disability including tips on reasonable adjustments, person-centred care, and trauma informed care.

Thank you to all who attended. Please do not forget to let us know how we did via [a short survey](#), so we can continue to improve.

Send us questions, discuss presenting a case, or let us know your 'go to' resources by emailing us at rarest@unsw.edu.au

Box 1: Tailoring care to meet the needs and preferences of individuals with intellectual disability.

- Make reasonable adjustments (a change to an existing approach or process which is essential to ensure a person's access to a service):
 - Different appointment times (e.g. out of hours)
 - Longer appointments
 - Meet in a quiet room to minimise distractions.
- Improve patient communication:
 - Speak directly to the person with intellectual disability rather than their support person
 - Allow extra time to explain things well, even within a busy emergency department
 - Use the teach-back technique (asking the person to repeat what they have just been told) to ensure they understand what has been explained to them
 - Leave space for questions, including assuring the person it's okay to ask questions
 - Use short simple sentences
 - Provide written accessible information, e.g. in Easy Read or simple English format.
- Enhance patient empowerment:
 - Provide respectful health care, by using respectful and strengths-based language, respecting choices and opinion, and demonstrating an understanding of the patient's rare condition
 - Give choice and consent, e.g. giving the option of including a support person rather than assuming the patient would like them included, giving the patient an opportunity to ask questions, and seeking consent before examinations and investigations
 - When providing a diagnosis and health care plan, understand the patient's pre-existing medical conditions, explain the diagnosis and management plan, and follow up with general practitioner and support systems.