



RAReST

Rare Disease
Awareness, Education,
Support, and Training



Key learnings from Rare Disease Project ECHO®

Series 2: Session 3 (28 September 2023) – Balancing patient advocacy, family empowerment, and multidisciplinary care

Opening presentation

Rebecca Saad, Clinical Nurse Consultant at the Epidermolysis bullosa service at Sydney Children's Hospital Network, spoke about balancing patient advocacy, family empowerment, and multidisciplinary care.

- EB is a rare inherited skin disease that leads to skin fragility, blistering, and wounds.
- Presentation and severity varies depending on the affected gene and inheritance pattern.
- In addition to the skin, EB can impact other organs including the heart, eyes, and gastrointestinal tract, and often leads to malnutrition.
- As a result, EB requires specialist multidisciplinary management.

Case presentation

Rebecca described a 5-year-old boy with severe EB. His parents had disengaged early from the local hospital and were noncompliant with his care plan including wound management and nutritional intake. The child was minimally handled, interacted with, or played with during his first year of life and had poor progression through his developmental milestones.

The usual approach to caring for a child with EB includes:

- Family engagement
- Family education
- Upskilling local providers
- Communication
- Multidisciplinary care.

In this situation, the usual approach was not leading to the desired outcomes. The EB team used multiple additional approaches to support the family and encourage compliance with the child's care plan, however by the age of 5, the child had stunted growth, severe malnutrition with micronutrient and vitamin deficiencies, poor weight gain, anaemia and iron deficiency, multiple wounds, and multiple antibiotic resistance. The parents were noncompliant with wound care, appointment attendance, and did not trust their local health care team.

When the child's weight became critical, a new approach was taken that involved:

- Altering previous family advocacy techniques to prioritise the child's best interests (person-centered care over family-centered care).
- Completion of child protection and wellbeing reports (person-centered care over family-centered care).
- The EB service assuming a primary and wholistic management role rather than using a multitiered care approach (care by a Centre of Expertise).
- Ensuring family capacity was included in management and planning (applying a strengths-based approach).

- Including regular revision of goals and monitoring outcomes from all members of the multidisciplinary team (MDT) and the family (partnership between the Centre of Expertise, other health care professionals and the family).
- Use of and promotion of the EB care plan in Powerchart for all teams involved in care and emailed to local health care team (promoting clinician education and empowerment).

The child underwent gastrostomy surgery with several additional procedures carried out while under general anaesthetic. This was followed by extended inpatient stay, support and management from the MDT, repeated education for the parents, and regular hospital reviews, resulting in the child gaining weight.

Discussion with attendees

The discussion covered:

- The challenges arising when English is a second language, and when traditional spiritual and cultural practices affect a person's care.
- The potential value of translating care plans into families' native languages, acknowledging this can be costly and difficult to fund.
- The possibility of trialling strategies that are quite different from usual clinical care, for example, involving the family's spiritual and cultural leader.
- The importance of psychological support for the family and delivering information in a way that the family will absorb, particularly the mother who is likely to be suffering from post-traumatic stress disorder (PTSD). Many approaches can be used to relax families and reduce stress, including sitting on the grass, holding warm or cold objects, and background music.
- The potential benefit of respite care for the mother, for example [Bear Cottage](#) or [Allowah](#), acknowledging that it may take some time for her to be able to leave her son for extended periods of time.

Key recommendations that the Project ECHO® participants and experts shared during of this discussion were:

- Consider different approaches for challenging individuals, such as using a person-centred rather than a family-centred approach, and assuming primary responsibility for a person's care when the usual multitiered approach is not working
- Recognise the value of a Centre of Expertise like the EB service in managing people with complex rare diseases
- Recognise the psychological impact of complex rare conditions on the family and use different strategies and settings to help families with trauma to better absorb information
- Ensure care is culturally appropriate.

Useful resources for clinicians and patients discussed this session were:

- Respite care:
 - [Bear Cottage](#)
 - [Allowah](#).
- [EB services](#) at Sydney Children's Hospital, Randwick.

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