RArEST Rare Disease Awareness, Education, Support, and Training

# Key learnings from Rare Disease Project ECHO®

*Series 1: Session 8: Rare Disease Research*

**Opening presentation**

Dr Falak Helwani, Research and Evaluation Manager at Rare Voices Australia, spoke on rare disease research.

**Opening presentation discussion**

Falak shared how research is a key priority for people living with rare disease to:

- Understand the mechanisms of disease and advance diagnosis
- Develop new treatments
- Develop better models of rare disease care, both broadly and for individual rare diseases
- Improve the health and social care systems that people living with a rare disease (PLWRD) need and rely on

She shared that:

- Limited data, knowledge and high levels of uncertainty are the driving forces behind patient-led advocacy for, and involvement in, research
- PLWRD often want to be involved in research even where there is no tangible/immediate benefit for them, but they can see the potential for benefits to future generations
- PLWRD can feel empowered through contributing to research
- Timely access to research and clinical trials can be life-changing and, in some cases, life-saving

She emphasised that health care professionals are the gatekeepers to research and clinical trials for PLWRD and suggested that health care professionals can:

- Actively scope potential clinical trials and research opportunities for their patients and know where to refer to ensure timely access to potentially life-changing or life-saving clinical trials and research opportunities
- Attend relevant rare disease specific conferences and meetings to keep abreast of best practice and build networks
- Allow time in consultations for discussing research and supporting shared and informed decision making
- Contribute to registries to improve the profile of rare diseases and improve natural history data

**Case presentation**

A metabolic physician presented on a family with two children impacted by an ultra-rare and life-limiting childhood onset condition. Intensive discussions supported the family in making an informed decision to travel overseas to access a clinical trial that was potentially life-saving for one child who met inclusion criteria for the trial.

Challenges included:

- How to support decision making when little is known about the long-term impacts of a novel therapy in a clinical trial
- How to support families where access to clinical trials may be limited (e.g. due to restrictions in inclusion criteria, geographical location)
- How to deal with the 'roller coaster' of emotions (hope and despair) that may be encountered related to clinical trials

**Case discussion**

The case discussion covered:

- The importance of building trust with families and connecting them wherever possible to experts or centres of expertise that can advise on clinical trials and research
- The importance of setting reasonable expectations about clinical trials, including restricted enrolments and what cannot be known or predicted
- How health care professionals can keep themselves up to date with advances in research and clinical trials, as well as reliable sources of point-of-care information that can assist with discussions with PLWRD

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

- Do your own research to build your network and increase your patient's opportunities to access research through referrals to specialist services, including clinical genetics for undiagnosed disease programs
- Build patient health literacy around research
- Understand the impact of uncertainty on PLWRD (uncertainty drives PLWRD to do their own research to find out what can help)
- Manage the balance between hype and safe and beneficial research involvement
- Support patients in decision making and consent processes to participate in research/clinical trials

**Useful resources** for clinicians and patients include:

- The **Rare Awareness Rare Education (RARE)** Portal: www.rareportal.org.au. This website contains current, reliable and straightforward information and resources for all rare disease stakeholders customised for the Australian context, including a dedicated research section
- **Clinical trials websites**: www.clinicaltrials.gov (International)
  www.australianclinicaltrials.gov.au (Australia)
  This short video provides a step-by-step guide for how to register for a clinical trial for a condition or group of conditions: www.schn.health.nsw.gov.au/research/clinical-trials
- **Undiagnosed Disease Network Australia** www.udnaus.org
- The **Australian Paedatric Surveillance Unit** (APSU) is a national resource to facilitate active surveillance of uncommon, rare childhood diseases: www.apsu.org.au

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