RArEST Rare Disease Awareness, Education, Support, and Training

Key learnings from Rare Disease Project ECHO®

Series 2: Session 4 (26 October 2023) – Coordinating and connecting care for people living with a rare disease

Opening presentation

Professor Michelle Farrar, Paediatric Neurologist at Sydney Children's Hospitals Network (SCHN), gave a presentation on coordinating and connecting care for people living with rare diseases, with a focus on Duchenne Muscular Dystrophy (DMD).

- DMD is an X-linked recessive condition affecting 1:3500 male births.
- The condition is characterised by progressive deterioration of skeletal muscle tissue and function, with most individuals in a wheelchair by the age of 12 years, and median survival of 30 years.
- Glucocorticoids are the only disease-modifying drugs available but are not curative.
- Michelle presented the case of Toby, a 13-year-old non-ambulatory boy with DMD and recent history of racing heart and breathlessness. Toby's family express an interest in discussing clinical trials with his treating team.

Rare disease presentation

- The multidisciplinary DMD clinic at SCHN coordinates care for individuals with DMD across multiple specialities including rehabilitation, orthopaedics, cardiology, endocrinology, psychosocial, primary/emergency care, respiratory, and sleep medicine.
- Parents of children with DMD and other rare neurological conditions often enquire about the possibility of advanced neurotherapeutics as treatment for their children.
- In general, parents interested in these approaches have:
    - An unmet need and urgency to know more about and trial these agents
    - Strong desire to seek information about advanced therapeutics
    - Interest in partnerships with their clinicians
    - A high tolerance of risk and uncertainty that is driven by hope; one challenge is managing their expectations and ensuring they understand the complexities and limitations of treatments and clinical trials.

Discussion with the community of clinical learning practice

- Reliable, credible information is important, and families generally want a single source of information they can trust and rely on.
- A good relationship with families is critical, so they feel comfortable discussing alternative options they are pursuing, for example, going overseas for a medical procedure (medical tourism).
- It is important to look after families' psychosocial and mental health, including their hopes, and manage expectations around clinical trials, particularly as clinical trial opportunities in Australia are often limited.
- Consideration must be given to whether patients and families are able to cope with the requirements of a clinical trial.

Toby's story – next steps

- Toby's family was referred to ClinicalTrials.gov to look for suitable clinical trials.
- Toby was enrolled in an Australian DMD registry. Enrolling patients in registries has many benefits for patients and for the community as a whole:
    - Families can be contacted about local clinical trials and research opportunities
    - Registries facilitate clinical trial readiness by compiling lists of potentially suitable clinical trial participants that can be shared with organisations looking to conduct trials in Australia.
- Although Toby's family were very interested in participating in a clinical trial, access to trials can be challenging. This is due to many factors, including:
    - Limited places in many trials
    - Often strict eligibility criteria
    - Once enrolled, clinical trials can be a substantial time and emotional commitment.
- The process of obtaining informed consent is important so families understand what is involved and their expectations are managed.
- The Advanced Therapies handbook co-developed by Michelle, her team at SCHN, and families, and supported by funding from Cerebral Palsy Alliance, is now available on the SCHN website. These resources and videos will help families understand, search for, and participate in clinical trials of advanced therapies.

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

- Suggested approaches to respond in a coordinated and connected way to new and emerging therapies:
    - Recognise research is part of clinical care in rare diseases
    - Build knowledge
    - Empower communication
    - Share decision making
    - Value supportive care
    - Connect with patient advocacy groups
    - Support psychosocial wellbeing.
- Patient advocacy and patient advocacy groups are valuable to help patients access trials and new therapies.

Useful resources for clinicians and patients discussed this session were:

- Clinicaltrials.gov: Database of international clinical trials
- SCHN video: Explains how to search for clinical trials on the Australian Clinical Trials website
- RARE Portal: Information about research and clinical trials.

The Advanced Therapies Handbook from Sydney Children's Hospitals Network is a new resource designed to help families learn about advanced therapies being used to treat some rare conditions. It includes resources and a suite of educational videos.

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