Rare Voices Australia

RArEST Rare Disease Awareness, Education, Support, and Training

Key learnings from Rare Disease Project ECHO®

Series 1: Session 3 – Rare Diseases and Communication

Opening presentation

Stephanie Broley, Senior Genetic Counsellor at the Rare Care Centre, Genetic Services WA, spoke on the topic of Rare Disease and Communication. A recording of the presentation can be found here.

There is an accompanying lesson on communication as part of RArEST's Rare Disease 101 module. You can register for this short, free CPD at the Medics for Rare Disease Website.

Discussion after the presentation

Working with translators

Discussion included how to effectively work with translators to support communication of complex and nuanced information. This underscores the benefits of partnerships. For example, if there is no direct translation, a health professional can work with a translator to find another way to explain a concept.

Follow up after diagnosis

Receiving a diagnosis can be overwhelming, so follow up with the family about their wellbeing and further questions after an appointment is important. Some genetic counsellors have this as part of their routine practice. It is key that genetic counsellors and clinical geneticists communicate well with a family's usual clinical team, including their General Practitioner (GP) to facilitate appropriate follow up.

The use of analogies

If used well, analogies are a good way to help families understand genetics. Stephanie Broley frequently uses the recipe analogy: for example, that a change in the DNA code is similar to a change in a recipe, which can have different impacts on the finished meal (or protein!). She recommends that whatever one people choose, it is important to be consistent. Factsheets are available to help clinicians have discussion about genetics from the Centre for Genetics Education and Australian Genomics. There is also an upcoming Rare Disease 101 Australia lesson on genomics.

Case-based discussion

Presentation: A GP presented a case of a child who was diagnosed with an ultra-rare condition at 18 months. There are many people involved in their care, including:

- GPs
- General paediatrician
- Speech pathologists
- Dietician
- Occupational therapist
- Orthotist
- At least 7 different hospital clinics

The GP is concerned that there may be late or missed management of issues due to the number of people involved. For example, when the GP referred the child to a service for one issue, they referred the child to yet another service for a separate issue. The GP is not sure if this was opportunistic or a planned review. Additionally, this did not reflect the parents' main concern.

An additional issue is the high burden that coordinating between the many providers and funders of care puts on the family, particularly in terms of time.

The presenter asked for advice on care coordination:

- How to support the family
- What is the scope of the GP role
- What could be slipping through the cracks

The discussion covered:

- Some care coordination supports that might be helpful include social workers at hospitals, clinical liaison nurses and dedicated care coordinators
- The mother, in particular, is under a lot of stress and this also needs to be addressed
- Due to system failures, the burden of record keeping and coordination often falls on parents/carers who are already overwhelmed
- The parents were disempowered when providers focused on a different issue to their main concern

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

- NDIS funding can be used for a support coordinator for a family, and this would be a good first step. There are a variety of state-based organisations that offer peer support to families to better understand their rights under the NDIS, for example Kindred in NSW and Kalparrin in WA.
- Supporting the family's wellbeing is key. A genetic counsellor could assist with this, or a counsellor with some understanding of complex medical issues. Carer's gateway is one first point of contact for practical carer support. Additionally, an increasing number of children's hospitals offer a care navigation service for complex, chronic, childhood-onset health conditions such as:
    - NSW - KidsGPS service through the Sydney Children's Hospitals Network
    - QLD - Connected Care and Nurse Navigator programs
    - WA - Rare Care Centre for families with a child with a rare complex condition
- That families/patients request that all their reports go to all relevant providers, including their GP.

Thank you to all who attended. Please do not forget to let us know how we did so we can continue to improve via this link and please let us know about any of your 'go to' resources you would like to share by emailing us at RAREST@unsw.edu.au