Rare Voices Australia

RArEST Rare Disease Awareness, Education, Support, and Training

Key learnings from Rare Disease Project ECHO®

Series 1: Session 4 - Tools for Diagnosis and Referral Pathways

Opening presentation

Dr Emma Palmer, Clinical Geneticist at Sydney Children's Hospitals Network, spoke on tools for diagnosis and referral pathways, using a real case as an example. You can find a recording of the presentation here.

For more on the diagnostic odyssey and tools, check out Lesson 5 in our Rare Disease 101 Australia module. You can register for this short, free CPD on the Medics for Rare Disease website.

Opening presentation discussion

Discussion included when a GP might use the tools highlighted in the presentation (FindZebra and PubCaseFinder). These are most useful in cases where a patient has a clear set of distinctive features. Additionally, they can help a GP find the right Human Phenotype Ontology (HPO) terms to describe their patient. Using these terms facilitates clear communication with other people involved in a patient's care. It is a good idea to include HPO terms in requests to testing laboratories as that can improve the quality of the lab's reporting.

Case presentation

A GP presented on a case in which parents had noticed some mild symptoms and signs in their child, but not suspected that they had a condition, genetic or otherwise. They had had a normal NIPT (non invasive prenatal test) in the pregnancy. This case raised the following questions for the involved practitioner:

- Does NIPT give false reassurance that a genetic condition is not likely?
- How can health professionals raise concerns about a child without fear?
- How can health professionals counsel patients and families about uncertainty?
- What are the referral pathways when families have concerns?

Case discussion

The case discussion covered:

- The importance of providing clear information to patients about what tests can and can't do. For example, Non-Invasive Prenatal Testing (NIPT) was developed as an alternative *screening* test for Down Syndrome, so it is less sensitive to other genetic changes and should not be considered a *diagnostic* test. For this reason, it is also sometimes called Non-Invasive Prenatal Screening (NIPS). A helpful booklet to share with families is available here
- Analogies to use with patients to discuss the need for different types of diagnostic tests, for example:
    - A sieve analogy where the NIPT/NIPS can be seen as a sieve with big holes, meaning it catches some things (e.g chromosomal aneuploidies like Down syndrome) but not much detail. Then subsequent tests are finer and finer sieves. It is best to start with

the big picture tests so more common/easy to detect things have a good chance of being picked up before we look more closely.
    - A 'Google' map analogy with a similar message, where we start by looking at the continents, then zoom into cities, then further in to the streets and houses.
- Strategies to reduce fear when talking with parents, which connected strongly to the importance of a **partnership approach**. For example:
    - Discussing how it is worth looking at common but less serious conditions, as well as rare but more serious conditions, just to make sure nothing is missed, including the opportunity to improve management or for targeted treatments.
    - Tailoring your communication based on **both** how concerned you are as a health professional, and how concerned the family is (i.e. a 'partnership' and 'person-centred' approach). RD101 Australia includes a lesson on effective approaches to which you can access for free here
    - Inviting the family to express what they are really worried about. It is important to normalise that *everyone* looks things up on the internet and people often come to appointments with specific concerns. An honest discussion can follow, based on a true understanding of each other's perspectives. The community talked about how this type of a conversation is easier when rapport and trust have already been established, for example with a GP.
- The high variability of testing times across jurisdictions

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

- Provide clear information to patients about what genetic tests can and can't do – to reduce the chance of confusion if a condition is not initially detected
- Use analogies to discuss the purpose and sensitivity of different genetic tests, such as sieves with finer and finer holes, or zooming in on a map
- A partnership approach can help reduce fear and manage uncertainty – it is important to practice honest and open communication tailored to each patient

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