RArEST Rare Disease Awareness, Education, Support, and Training

Key learnings from Rare Disease Project ECHO®

Series 3: Session 2 (17 October 2024) – Achieving early diagnosis

Opening presentation

This session was presented by Dr Emma Weisz, a paediatrician with special interest in genetics and Senior Project Clinician at Melbourne Genomics Health Alliance, and Erin Crellin, PhD candidate at Melbourne Genomics Health Alliance, University of Melbourne, and Murdoch Children's Research Institute. Their presentation was about achieving early diagnosis in people with a rare disease, focussing on genomic testing in children aged <11 years with a suspected childhood syndrome.

Medicare-funded genomic testing that can be ordered by paediatricians is now available for children aged <11 years with a suspected monogenic condition. A suite of resources to support paediatricians and families are available on the Melbourne Genomics website, including:

- Questions to ask your child's paediatrician about genomics, and things a paediatrician may ask at the appointment
- Genomics and childhood syndromes, presented by two parents who share their experiences and insights into genomic testing. The resource covers relevant topics for families who are considering genomic testing, for example, timing, what testing can offer, and potential drawbacks of testing.
- Genomic testing for paediatricians, an all-in-one resource hub to support paediatricians with Medicare-funded genomic testing, including:
    - Step-by-step guide – a short guide with resources to support paediatricians through the testing process from start to finish
    - Ordering a genomic test – instructions and links to assist with ordering a genomic test
    - Ask a genetic expert – a service open to anyone in Australia, to ask questions of a genetic expert (usually a genetic counsellor) via phone or email.

Families who have undergone genomic testing have reported that they would like:

- To be given time and space to decide whether the time is right for genomic testing
- Written information that can be taken away
- Their health professional to be mindful of the words they use (positive, diversity affirming)
- Their health care provider to acknowledge the emotional impact of receiving a genetic diagnosis and proactively intervene
- Genetic experts to be involved as needed.

Families who have received genomic testing results have reported that they would like:

- To plan with their clinician how the results will be delivered
- Results not to be returned without prior warning
- Clinicians to prepare for the results disclosure appointment and provide:
    - Information about the condition
    - Supports for families
    - Resources to share with other family members/care team.

Rare disease presentation

Emma presented the case of a 3-year-old boy with severe developmental delay and atypical body characteristics including tapered fingers and below-average head circumference, height, and weight. A paediatrician had conducted Fragile X testing and a chromosomal microarray, along with an MRI of the child's brain. All tests were uninformative, meaning no explanations for the child's characteristics had been found.

The next step for the child is a genomic test, which can be ordered using the step-by-step guide from Melbourne Genomics Health Alliance.

Discussion with the community of clinical learning practice

The group discussion covered:

- General practitioners (GPs) can order Fragile X and CMA testing for specific conditions including autism, intellectual disability, and congenital conditions – something many families and GPs are not aware of.
- If a person has multiple health concerns, it is useful to book a long appointment (eg 60 minutes) rather than a shorter one with a GP.
- A number of new Medicare item numbers for genomic testing in different clinical specialities, including cardiac, oncology, renal, and mitochondrial disease are now available, and can be ordered by non-genetics specialists.
- Allied health professionals such as speech and language services can be excellent sources of knowledge and can support the family though the diagnostic odyssey. In an ideal world, the primary care provider, specialist, allied health providers, and education providers work together to support the family.
- Having reports from allied health professionals can be helpful to support parents when they speak to GPs about the possibility of diagnostic testing.
- Saliva samples can be used by many laboratories and is a simple and easy way to obtain a DNA sample for genomic testing. Saliva samples can also be used for Fragile X and chromosome microarray testing.

You may be interested in information on the diagnostic odyssey and diagnostic tools in the free Rare Disease 101 Australia e-learning module.

Key recommendations that Project ECHO® participants and experts shared relating to paediatric genomic testing were:

- Genomic testing can be useful to facilitate a more rapid diagnosis for children with suspected childhood conditions, however it requires planning and careful and informed conversations with families
- Resources are available to support clinicians and families with paediatric genomics testing, including Genomic testing for paediatricians
- GPS can help support and coordinate care for families undergoing genomic testing

Useful resources for clinicians and patients discussed this session were:

- Genomic testing for paediatricians
- Questions to ask your child's paediatrician about genomics
- Information about:
    - Chromosome microarray testing
    - Fragile X testing
- Mental health and wellbeing support for Australians living with a rare disease
- Facematch, which uses face-matching technology to help find a diagnosis for children
- National Recommendations for Rare Disease Health Care
- Next step roadmap for rare and undiagnosed diseases (Undiagnosed Diseases Network International)
- Rare Helpline
- Lyfe Languages