RArEST Rare Disease Awareness, Education, Support, and Training

Key learnings from Rare Disease Project ECHO®

Series 2: Session 1 (27 July 2023) – Supporting patients before, during, and after a rare disease diagnosis

Opening presentation

Clare Stuart, Policy and Advocacy Manager, Mito Foundation, provided a brief background to mitochondrial disease.

- Mitochondrial diseases are conditions affecting mitochondrial structure and/or function. Mitochondria are the 'energy packs' of our cells.
- At least 1 in 200 individuals or >120,000 Australians may carry genetic variants that put them at risk for developing mitochondrial disease.
- An accurate diagnosis can take years, decades, or generations, and patients and families need support before, during, and after a diagnosis.

Rare disease presentation

Michelle Hemmings, Patient Pathways Telehealth Nurse, Mito Foundation, discussed David*, a 40-year-old man living in a regional area with a 20-year history of hearing loss, cardiac issues, and progressive symptoms of fatigue, muscle weakness, and exercise intolerance. In 2021, David had a stroke which he did not fully recover from. He saw a large number of individual health providers and health care services, and care was not coordinated across services or time.

David received a diagnosis of the mitochondrial condition Mitochondrial Encephalomyopathy, Lactic Acidosis and Stroke-like episode (MELAS) after his brother Tom*, who lived in a metropolitan area, also had a stroke, and subsequently received a genetic diagnosis of MELAS. Even after receiving a diagnosis, David was struggling to access appropriate specialist and local services.

When David's family contacted the Mito Foundation, the Foundation's Telehealth Nurse was able to:

- Support the family to navigate care pathways to build a health care team including a specialist metropolitan mitochondrial disease service and a supportive and empowered general practitioner (GP).
- Educate the local regional hospital team and develop an emergency care plan which could be shared on the hospital electronic record.
- Assist David with access to National Disability Insurance Scheme (NDIS) and financial support (Disability Support Pension and insurance).

A particularly challenging aspect was restoring David's confidence in health services.

Discussion with the community of clinical learning practice

The discussion covered:

- The importance of helping individuals and families recognise that they may have 'red flags' for a rare condition such as mitochondrial disease. This may help them in having conversations with health care professionals if they are on a diagnostic odyssey.

*pseudonym

- The importance and value of the GP in considering and facilitating diagnosis, synthesising, translating, and explaining findings to patients and families, helping to develop management plans, coordinating care, following up with specialists, acting as a central repository for information, and providing ongoing holistic care to the whole person over years (including preventive health care, mental health care, care of their other illnesses, etc).
- Acknowledgement that while GPs are well placed to perform this central role, they are already overworked and not paid for time that is not patient-facing.
- The need for systemic change to improve communication and information dissemination and create networks to facilitate connections with specialists in rare disease care.
- As approaches to rare disease diagnosis and care are not currently taught in the medical curriculum, supports are needed to help health care professionals 'think rare' i.e. recognise the potential red flags for rare disease and determine next steps to access a diagnosis.
- The need for patients to have a rare disease management plan/passport for when they seek emergency care that is agreed on by the patient, their family, and their expert and local teams, and recognised by health care services. Surveys suggest that many people with mitochondrial disease do not have a general management plan and even less have an acute care plan.
- The value of personalised health care folders for families to store management plans and summarise their health care needs in the one place.

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

- Consider mitochondrial disease when a patient has an unexplained constellation of signs and symptoms which may include a 'common' disease with 'atypical' features, involving at least three organ systems and/or any 'red flag' symptom in the 'Maybe It's Mito' diagram (see Figure 1 below) especially if they have recurrent setbacks/flare ups with infections.
- Health care providers can reach out to Centres of Expertise and patient organisations (via the RARE Portal link below) to access appropriate educational materials, emergency and coordinated care plans, supports, and resources. Many such services and organisations have care coordinators or nurse specialists/consultants who can offer invaluable support to people living with rare disease and their local health care providers.
- To improve continuity of care, health care providers should ensure they copy the person living with rare disease into their communications so they can add to their own communications folder, as well as email/mail copies to all relevant health care providers.

Useful resources for clinicians and patients discussed at this session were:

- Rare Voices Australia: RARE Portal. Current, reliable, and straightforward information and resources about rare diseases, customised for Australia. Information for many conditions, including mitochondrial disease, is under development. Includes emergency management plans and links to relevant rare disease organisations and Centres of Expertise.
- Mito Foundation:
    - Managing Mito. Includes resources, services, and handy links on managing day-to-day life with mitochondrial disease.
    - For health professionals. A guide on mitochondrial disease for health professionals. Training modules about diagnosing mito (from Arterial Education) and managing patients with mito (from Australian Dr Group) is also available.
- Rare Voices Australia/RArEST project: Red flags for rare diseases. A helpful overview of the key red flags pointing to a rare disease more generally.
- Rare Disease 101 Australia free elearning module: Lessons on the diagnostic odyssey.

- US National Institutes of Health: GeneReviews. A point-of-care resource for clinicians, providing clinically relevant and medically actionable information for inherited conditions, covering diagnosis, management, and genetic counseling for patients and their families.
- Templates for health care passports/folders, including for people with intellectual disability or other conditions impacting communication and understanding.
  - Council for Intellectual Disability: My Health Matters
  - South-Eastern Sydney Local Health District: Admission 2 Discharge (A2D).

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.

Figure 1. Red flags for mitochondrial conditions. Reproduced from *Mitochondrial Disease Health Professionals Guide* with permission from Mito Foundation