



RAReST

Rare Disease
Awareness, Education,
Support, and Training



Key learnings from Rare Disease Project ECHO®

Series 2: Session 5 (23 November 2023) – Innovative approaches to rare disease prevention and care

Opening presentation

Libby Massey, Director, Research, Clinical Services and Education at MJD Foundation (MJDF), gave a presentation on innovative approaches to delivering culturally sensitive and people-centred care to individuals with Machado-Joseph Disease (MJD) in remote areas.

About MJD

Machado-Joseph Disease (MJD) is a late-onset, autosomal dominant neurodegenerative condition characterised by:

- Progressive loss of mobility, hand dexterity, arm movements, trunk and head control, and speech and swallowing
- Progressive incontinence
- Sleep and visual disturbances.

The average age of onset is 36 years, with a mean life expectancy of 20 years after symptoms start, which may be as early as 5 years of age or as late as 80 years. Due to genetic anticipation, subsequent generations develop the disease earlier in their lives, and may experience more severe disease with more rapid symptom progression.

MJDF approach to rare disease care in remote Aboriginal and Torres Strait Islander communities

The MJDF is a small not-for-profit organisation that supports Aboriginal and Torres Strait Islander families who live with MJD and spinocerebellar ataxia type 7 (SCA7) across the Northern Territory and Far North Queensland. The organisation is staffed by allied health, nursing, and community service professionals and supported by an operations team.

The MJDF has a strong focus on overcoming the barriers of remote service delivery so their clients can stay living in and being cared for by their community if they choose. To do this, the MJDF operates five programs to support their clients:

- Research
- Education
- Clinical services; this includes genetic counselling, MJD therapy, and recently, community-based ataxia clinics in partnership with a leading Australian neurologist. These clinics support MJDF clients, help to support and upskill local primary health care providers and community service and therapy teams, and contribute to MJD research.
- Community services; these are delivered by a multidisciplinary team including Aboriginal Health and Community workers, allied health professionals, and family support workers.
- Advocacy.

A key aspect of the MJDF approach is the bicultural 'Our Way' service delivery model. Under this model, the needs and priorities of MJDF clients drive the development of programs and enable the delivery of genuinely person- and family-centred supports. Kinship, culture, language, and relationships based on trust are central tenets of this model.

Rare disease presentation

Libby presented the cast of Ernest*, a 45-year-old man with severe MJD, who is non-ambulatory, and experiencing severe dysarthria requiring the use of low and high-tech augmentative and alternative communication. Ernest has urinary incontinence, significant constipation, disturbed sleep, visual disturbances, and low mood with bouts of depression.

Ernest is struggling with symptoms of MJD but does not feel comfortable using Western medication, preferring to use 'bush' medicine.

Discussion with the community of clinical learning practice

Strategies to support people in remote areas and who are from Aboriginal or Torres Strait Islander communities like Ernest include:

- Taking time to understand and respect the individual, along with their personal and community priorities and ways of knowing and doing.
- Pairing non-indigenous health providers with indigenous health/support workers to build trust and cultural safety.
- Being respectful of each client's cultural beliefs around why their family has MJD, and working with this rather than discounting it.
- Holding clinics in a place that is most welcoming and safe for each individual. Libby mentioned most clinical encounters with the MJDF team are outdoors. She pointed out that people have often had traumatic experiences with 'western' style health clinics, which may place them in a situation they do not feel culturally safe to be in, for example being asked to be in a closed space with people they are forbidden to be close to.
- Having a journal, log, or 'passport' into which health care professionals can record notes at each encounter. The client then uses this journal to inform the next health care professional of their condition, treatment history, and how to best communicate with them. This would be particularly helpful in remote communities where rapid turnover within the health care workforce makes information exchange complex.
- Providing medications in single-dose sachets rather than more complex multiday packs.
- Using drawings and images to convey important concepts.

Ernest's story – next steps

The team spent time to understand why Ernest was not keen to use the proposed medications, which centred on his concerns about artificial chemicals and preference for therapies that were more in line with natural processes in his body. For example, the team explained that melatonin is a natural hormone involved in the sleep cycle. This helped allay some of his concerns and he was willing to trial melatonin.

Melatonin proved to be symptomatically helpful for Ernest. As a leader in his community, he was happy to share his positive view of this medication with his community. Discussions around continence were held with male practitioners, which increased Ernest's comfort with the conversations.

*pseudonym

Key recommendations that Project ECHO® participants and experts shared for supporting remote Aboriginal and Torres Strait Islander communities, which are also relevant to other remote situations and/or culturally and linguistically diverse communities were:

- Ensure discussions around treatment recommendations align with traditional concepts of health and wellness
- Acknowledge and accommodate differences in language and health literacy
- Understand local disease perceptions and knowledge
- Document and embed knowledge in the 'system', especially where the health workforce is casual, there is limited continuity of care, and/or the person has difficulty communicating
- Develop and utilise a patient 'passport' or similar to share information among health professionals to combat the gap created by a casual workforce.

Useful resources for clinicians and patients discussed this session were:

- [Clinical Yarning](#) website – a patient-centred framework to improve communication in Aboriginal health care.
- [13YARN](#) – a website and help line to support Aboriginal and Torres Strait Islander people. The website includes a range of fact sheets to provide information and assistance during challenging times.

You may be interested in reading [previous summaries](#) of presentations on culturally appropriate rare disease care, for example Session 6 of the first Rare Disease Project ECHO series, which included a presentation by Danielle Headland, Senior Health Promotion Officer at WA Health, Aboriginal Research Officer at Telethon Kids Institute, and Language Champion at Lyfe Languages.

There is also information on culturally safe and appropriate rare disease care in the free Rare Disease 101 Australia e-learning module, which you can access via the [RArEST webpage](#).

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