Key learnings from Rare Disease Project ECHO®

Series 4, Session 3
Two-Way Learning: Integrating Cultural and Clinical Expertise in Rare Disease Care
28 October 2025

Presentation 1: Integrating Cultural and Clinical Expertise in Rare Disease Care

Katrina Connolly (Indigenous Health Worker, Parent, Advocate)

Katrina shared her journey as a mother of a child with a rare genetic condition and as an Indigenous health worker in the remote community of Yarrabah. Her presentation illuminated the layered challenges faced by First Nations families navigating rare disease care from diagnosis to daily life, and called for culturally safe, family-centred models that honour lived experience.

Emotional impact of diagnosis

Katrina's son is the only child in Yarrabah with his rare condition, which is also rare in Aboriginal and Torres Strait Islander populations. The diagnosis brought clarity but revealed how limited and fragmented the support systems were for families in remote communities. She described the emotional toll of constant hospital visits, uncertain prognosis, and the fear of the unknown.

Referral pathways in Yarrabah

Access to paediatric services in Yarrabah depends on referrals to Cairns Base Hospital, where patients are triaged by need. Katrina was fortunate to be seen during a visiting clinic but emphasised that most families face long delays and complex navigation. She highlighted how geographic isolation, lack of local specialists, and rigid referral systems leave families feeling invisible and unsupported.

Cultural disconnect in care models

The widely used social and emotional wellbeing (SEWB) framework centres individual wellbeing while neglecting the broader family unit. Katrina called for models that reflect Aboriginal values of interconnectedness, where healing involves carers, siblings, and extended kin. She noted that even as a health worker, she struggled to access culturally safe genetic counselling. After her son's diagnosis, she was told the only relevant information available was from families in the United Kingdom. She called for sustained peer support for carers and culturally safe advocates who understand the realities of remote Indigenous families.

Digital health barriers

Telehealth has helped bridge some gaps, but Katrina noted persistent issues with connectivity, digital literacy, and cultural safety in online platforms. Many communities lack NBN access, and phone consults are often the only option. She stressed the importance of having Indigenous health workers present during telehealth appointments to support understanding and trust.

Visual storytelling and co-designed resources

A turning point came when Katrina collaborated with the Australian National University's National Centre for Indigenous Genomics to co-design a visual resource that placed her son at the centre of a culturally resonant diagram. The artwork captured her journey from parent to health worker, surrounded by family, community, and external services. Katrina described this as the first time she felt truly heard and emphasised the power of visual storytelling for First Nations families navigating complex health information.

Katrina offered practical guidance for clinicians working with First Nations families:

- Use plain language and visual aids to explain genetics
- Respect cultural beliefs about illness and recognise they can be compatible with other models of health and medicine
- Include Aboriginal health workers early in the care team
- Recognise historical trauma and systemic mistrust
- Coordinate care to reduce travel and repetition
- Ask, listen, and problem-solve together – nonattendance doesn't mean disinterest

Discussion

- Participants expressed deep appreciation for Katrina's honesty and clarity. Many described her visual resource as one of the most powerful depictions of rare disease impact they'd seen.
- The ANU-developed videos and fact sheets are important as best-practice genetic health literacy tools. National Centre for Indigenous Genomics team members spoke about the co-design process behind these resources, highlighting Katrina's leadership and the group's commitment to creating meaningful, culturally safe tools.
- Peer support is critical for families navigating rare disease, especially in remote communities.
  - Katrina noted that there were no Indigenous-specific or Indigenous-led peer support groups or networks in her region.
  - NDIS support workers or organisations like Mission Australia sometimes help connect families.
- Participants reflected on the challenges Katrina described such as referral delays, lack of paediatric specialists, NDIS navigation, and the emotional burden of coordinating care alone.

Further resources

Culturally safe genetic literacy tools

- NCIG Genetic Resources – Australian National University
- What is a Rare Genetic Condition – NCIG Videos
- FAQ – NCIG Resources

Educational resources to support timely diagnosis from the RArEST project including:

- National Recommendations for Rare Disease Health Care: recommendation 2 which includes Red Flags for Rare Disease and AI assisted diagnostic platforms
- Rare Disease 101 Australia, an RACGP accredited e learning module
- RACGP check on Rare Diseases [Unit 607]

Read previous summaries of presentations from the Rare Disease Project ECHO series here.

Send us questions, discuss presenting a case, or let us know your go-to resources by emailing us at RareDiseasesNSW@unsw.edu.au.