Key learnings from Rare Disease Project ECHO®

Series 4, Session 2
Coordinating Care when Systems Fail: Your Roadmap for Rare Disease Management
7 October 2025

Presentation: Coordinating care when systems fail

Janna Linke (Founding Director of Connective Tissue Disorders Network Australia (CTDNA))

CTDNA (Connective Tissue Disorders Network Australia)

CTDNA supports Australians with heritable connective tissue disorders through advocacy, education, system navigation and research. Conditions include Ehlers-Danlos syndromes, Marfan syndrome, Loeys-Dietz syndrome, Stickler syndrome and many others.

Challenges in diagnosis and care

Despite recognisable phenotypes and available genomic testing, patients often face years of misdiagnosis, fragmented care and dismissal. Hypermobile Ehlers-Danlos Syndrome (hEDS) is now believed to affect 1 in 500 people but remains underdiagnosed due to the lack of a genetic marker. The average diagnostic delay for hEDS is 22 years.

System Limitations

Short GP appointments, high out-of-pocket costs and underutilisation of care plans (e.g. chronic condition, mental health, home medicine reviews) contribute to inequity. Many patients qualify for but lack formal care plans, often because they were not initiated by their health care team:

- Chronic condition plans (40% uptake)
- Mental health treatment plans (25% uptake)
- Home medicine reviews (4% uptake)

Reporting requirements for NDIS and Centrelink also place financial strain on patients. GPs can help by:

- Proactively initiating care plans
- Incorporating reporting into NDIS plans
- Using simplified GPCCMP processes (as of July 2025) to trigger allied health rebates

The simplified GPCCMP process now triggers allied health rebates, though access remains capped at five sessions. Creative use of these sessions, for example, to support NDIS applications, can improve outcomes.

Additional Strategies

- **Home Medicine Reviews** help manage polypharmacy and clarify drug-related symptoms.
- **Eating Disorder Plans** may be relevant due to GI symptoms and disordered eating.
- Group education models, similar to diabetes educators, could support rare disease care.
- **The SPIDER tool**, developed for hEDS/HSD, helps patients and clinicians prioritise care based on the most pressing needs. For example, addressing mental health first may improve engagement with allied health services.
- GPs can support patients by identifying relevant specialists, engaging with advocacy groups (e.g. CTDNA, Ehlers-Danlos Society, SWAN), and using resources like the Rare Portal and Rare Helpline.

Addressing Gender Bias

hEDS disproportionately affects women, who frequently report being dismissed or misdiagnosed with anxiety. Carers, often mothers, also face bias when advocating for their children. There is a critical need for medical education that addresses gendered experiences in rare disease care.

Case presentation and discussion

Dr Jason Lam (GP, Sport and Exercise Medicine Registrar, CTDNA Scientific Advisory Committee Member)

Jason presented the case of a 42-year-old woman with a complex history including POTS, asthma, hip dysplasia, fibromuscular dysplasia, and recurrent episodes of head and neck pressure and pain, tachycardia, and throat spasms. Despite multiple specialist reviews (ENT, neurology, rheumatology, clinical pharmacology and general physician), no unifying diagnosis was made. Jason suspected hypermobile Ehlers-Danlos Syndrome and stylojugular syndrome and confirmed diagnosis of hEDS. Additional issues were slow gut transit, allergy-like symptoms and multiple chemical sensitivities suggesting MCAS, revisiting of prior imaging showed missed elongated styloid process leading to diagnosis of stylojugular syndrome accounting for her head and neck pain. The patient's medication regime and autonomic symptoms were not suitable and required review and revision.

Discussion

The presentation and case prompted discussion around diagnostic complexity, care fragmentation, and ideally co-located expertise.

- Participants discussed the underutilisation of care plans and the challenges of organising case conferences.
- Caution with Medication Reviews - reviewer needs expertise with the conditions and medications (many of which are off label), and access to clinical documentation so they can understand the rationale.
- The need for dedicated centres of expertise for rare and complex conditions was raised. Jason suggested co-located expertise and multidisciplinary clinics as a potential solution.
- CTDNA is actively advocating for multidisciplinary clinics for heritable connective tissue disorders.
- Participants with lived experience resonated with the frustration of being told their symptoms are due to anxiety.
- The group agreed that it was important for GPs to validate patient concerns and address the gender bias in diagnosis and care.

Further resources

Clinical and educational tools

- A GPs guide to hypermobile Ehlers-Danlos syndrome (AusDoc)
- Healthed podcast series – CTDNA
- The Spider Questionnaire – triage tool for hEDS/HSD
- AskShareKnow – shared decision-making support
- Top Tips for Safe Health Care – Australian Commission on Safety and Quality in Health Care

Care planning resources

- Care plans for health professionals – Services Australia
- GP Chronic Condition Management Plan (GPCCMP)
- Eating Disorder Treatment and Management Plan

Patient advocacy and specialist directories

- Connective Tissue Disorders Network Australia (CTDNA)
- EDS Australia – Specialist Directory
- Australian POTS Foundation
- Syndromes Without a Name (SWAN)

Educational resources from the RArEST project including:

- National Recommendations for Rare Disease Health Care: recommendation 2
- 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.