Key learnings from Rare Disease Project ECHO®

Series 4, Session 1
When Common Symptoms Point to Uncommon Diseases: A GP's Detective Toolkit
16 September 2025

Presentation 1: PRECISE genomics project

Dr Alan Ma (Clinical Geneticist, Sydney Children's Hospital Westmead)

PRECISE

PRECISE (Practitioner Readiness Education Capabilities with Implementation Science and Evaluation) is an MRFF-funded initiative supporting genomics integration into primary care. It equips GPs with knowledge, tools and confidence for genomic medicine in everyday practice.

Findings

In scoping reviews and co-design workshops with GPs and patients across Australia, they identified:

GP-reported barriers

- Insufficient genomics knowledge
- Time constraints in consultations
- Unclear referral pathways
- Ethical concerns around testing

Patient perspectives

- Need for empathy and advocacy
- Recognition of their lived expertise
- Accessible patient-facing resources

Resources

Based on these insights, PRECISE created:

- Point-of-care tools (fact sheets on non-invasive prenatal testing, carrier screening, cancer genetics)
- Referral decision aids (genetic testing tables with Medicare eligibility)
- Educational content (webinars and e-learning modules on Medcast)

Presentation 2: When common symptoms point to uncommon diseases- A GP's detective toolkit

Dr Claire Bowden (GP, Rare Care Centre WA)

The diagnostic journey

Many rare diseases have an underlying genetic cause. GPs face several challenges in diagnosing and caring for people living with a rare disease, including:

- Diagnostic uncertainty: Rare diseases often masquerade as common conditions, leading to delayed or missed diagnoses.
- Fragmented care pathways: Patients frequently bounce between providers without a coordinated approach, prolonging the diagnostic odyssey.
- Limited access to genetic services: GPs may be unsure when and how to refer, and patients face long wait times or out-of-pocket costs.
- Ethical complexity: Decisions around testing, disclosure, and family implications require nuanced conversations that GPs may feel ill-equipped to lead.

The red flags approach

This approach identifies clinical clues suggesting underlying rare conditions, helping GPs recognise when further investigation is needed (a version is included in the National Recommendations for Rare Disease Health Care)

Chromosomal Microarray (CMA) testing
CMA is a first-line genetic test for detecting copy number variants. Medicare-rebated for:

- Developmental delay
- Intellectual disability
- Autism spectrum disorder
- Congenital anomalies

GPs can order CMA but should be aware that results may reveal variants of uncertain significance or incidental findings requiring specialist interpretation and counselling.

GPs play a key role in:

- Recognising red flags that warrant referral (e.g., global developmental delay, dysmorphic features)
- Initiating conversations with families about genetic testing and supporting them through the genetic testing and diagnostic process
- Coordinating genetic tests and/or referrals to clinical genetic services or other specialist (e.g. paediatricians, neurologists) who can organise further testing.

Case presentation and discussion

Claire presented the case of a young patient who initially appeared well, but began showing signs of predominately gross motor developmental delay, hypotonia, recurrent viral illnesses and pectus excavatum. Despite normal prenatal screening and newborn tests, the child's condition deteriorated over time. The red flags prompted metabolic and genetic testing, which confirmed a diagnosis of a rare neurodegenerative condition.

Discussion

The group discussed the case presentation and whether normal screening tests during pregnancy give false reassurance to patients and clinicians.

- Using the term "non-diagnostic" result rather than "normal" or "negative" better reflects the limitations of screening tests and avoid implying certainty.
- The genetic variants causative of this condition would not be included in the Medicare-rebated 3 condition carrier screening, and would only be included in expanded carrier screening panels, which are available privately and cost ≥ $1500.
- Most conditions detected by expanded carrier screening conditions are X linked or autosomal recessive and often there is no family history.
- The patient's mother was offered expanded carrier screening but declined due to cost and lack of family history. This became a source of distress after the diagnosis, highlighting the burden placed on individuals to make complex decisions without systemic support.
- Better education is needed on what prenatal and carrier screening can and cannot detect.
- Health literacy resources and time are required to help clinicians consent and support families through genetic testing.
- Having GPs embedded in tertiary, multidisciplinary teams helps empower the GP.
- Point of care genetic counselling advice plus Medicare items that enable GPs to bill for long appointments and join MDT discussions with rare disease experts would overcome many barriers of time and reimbursement.

Further resources

Clinical genetics information for clinicians

- PRECISE portal (more resources will be uploaded soon.
- Dunlop, K. L. A., et al. (2025). Building capacity for genomics in primary care: a scoping review of practitioner attitudes, education needs, and enablers. Frontiers in medicine, 12, 1577958
- Guide to accessing clinical genetics services

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.