# **Presentation title** Reproductive carrier screening: making it work in your practice

## **Introduction** The purpose of reproductive carrier screening (RCS) is to identify couples at high risk of having a child with a severe autosomal recessive (AR) or X-linked (XL) disorder, allowing them to make informed reproductive decisions. RCS is now considered a mainstream test, offered through a number of public and private laboratories across Australia. The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) recommend that any practitioner providing reproductive care should offer information about carrier screening to women who are planning a pregnancy or who are in early pregnancy. While the merit of this requirement is not in dispute, the challenge lies in providing information about the purpose, scope, cost and outcomes of different carrier screening options to meet this professional obligation in the context of a busy general or specialist practice.

## **Aims** To discuss the clinical aspects of RCS testing and identify practical ways of providing relevant information and appropriate pre-test counselling.

## **Methods** Through a structured, albeit informal, workshop, a genetic pathologist and a genetic counsellor will address the information and counselling objectives related to RCS, discuss relevant molecular mechanisms and limitations of testing, and circumstances where Medicare-funded testing (or free male-partner testing) may be available. The conveners will also introduce a new online course for patients and discuss its utility in allowing practitioners to meet their professional and ethical obligations regarding RCS.

**Results**Participants at the workshop will learn what information and options about RCS must be provided, and how to achieve that goal in their practice.

## **Discussion** The growing complexity of genetic testing can overload both the provider and the recipient of this information. The workshop will identify the key objectives that must be met for the responsible provision of carrier screening and, by providing easy-access resources, assist clinicians in meeting these objectives.

**Learning outcomes**

1. Pre-test counselling and addressing challenges in providing information about the purpose, scope, cost and outcomes of different carrier screening options
2. Options for screening: basic RCS (e.g. testing for CF/SMA and FXS only) versus expanded carrier screening (including 100s of genes)
3. Testing pathways: sequential testing versus testing both members of the couple at the same time
4. Molecular mechanisms, importance of obtaining information related to family history and understanding test limitations
5. Becoming familiar with resources available to assist with provision of RCS, including exposure to an online educational tool developed for patients.

**How will delegates be involved throughout this presentation?**

* Opportunity for delegates to share their experience with RCS
* Opportunity to access the online learning tool
* Interactive Q&A