DOUGLASS HANLY MOIR PATHOLOGY

Information for Doctors

Reproductive carrier screening

Medicare rebate now available

Cystic fibrosis (CF), spinal muscular atrophy (SMA) and fragile X syndrome (FXS) are three of the most common serious genetic disorders in Australia. Reproductive carrier screening can identify couples at high reproductive risk for these inherited disorders, and allows couples to make informed decisions about their reproductive options and prenatal care.

The importance of reproductive carrier screening

RANZCOG recommends that information about reproductive carrier screening be offered to every woman, either prior to conception (preferred) or in early pregnancy.[1] CF, SMA and FXS affect one in 1,000 babies; this is greater than the combined risk of trisomies 21, 18 and 13 in a woman under the age of 30 years. Most carriers have no family history and are unaware that they are carriers.

Disorder	Frequency of affected child	Frequency of carriers
CF	1 in 3,500	1 in 30
SMA	1 in 10,000	1 in 50
FXS	1 in 4,000 males 1 in 6,000 females	1 in 330

Data from: Archibald A, Smith M, Burgess T, et al. *Genet Med.* 2018; **20**(5):513–526

Targeted screening

- **Cystic fibrosis (CF)** is the most common inherited disorder in Europeans. It affects respiratory and gastrointestinal function, resulting in progressive lung disease, recurrent respiratory tract infection, pancreatic insufficiency and male infertility.
  The test detects 50 of the most common mutations responsible for cystic fibrosis.
- **Spinal muscular atrophy (SMA)** is the most common genetic cause of mortality in children under two. It is characterised by progressive symmetric muscle weakness and atrophy that can be complicated by respiratory, orthopaedic and nutritional comorbidities.
  The test detects the deletions of the SMN1 gene which account for approximately 96% of the mutations in this gene.
- **Fragile X syndrome (FXS)** is the most common form of inherited intellectual disability, developmental delay and behavioural abnormalities, including autism.
  The test detects expansions of the CCG triplet repeat in the FMR1 gene which account for >99% of the mutations in this gene.

Testing procedure

This test has a Medicare rebate which, subject to criteria being met, may cover all or part of the cost. There are two options for carrier testing:

- A sample is collected and tested from the female partner first. If she is found to be a carrier of CF or SMA, a sample can be collected from the male partner for carrier testing for the same disorder. Reproductive carrier screening of an unaffected male for FXS is not required, as this is an X-linked disorder.
- Samples are collected from both partners and tested simultaneously. This allows additional time to consider the issues arising from the test and make informed choices about the couple's reproductive plans.

Information for Doctors

Testing is performed in Australia at one of our NATA-accredited laboratories.

Reproductive carrier screening

Online patient education course

Sonic Genetics offers an online educational course to support doctors and their patients who are considering reproductive carrier screening. This is an RACGP- and ACRRM-approved CPD activity for general practitioners.

The intent of this course is to supplement pre-test discussions with your patient. Complete your doctor registration at patientedu.sonicpathology.com.au

Reporting results

Results indicate whether a mutation was found, and the implication for the patient.

Result	Interpretation
Carrier for CF or SMA	Individual is at increased risk of having affected children. Testing of reproductive partner is recommended.
Carrier for FXS (females)*	Individual is at increased risk of having affected children. This result also has potential medical implications for the individual being tested.
Carrier status for CF, SMA and FXS unlikely	A mutation was not detected but the possibility that the patient is a carrier cannot be excluded. The risk of having an affected child is greatly reduced but not eliminated.

*Note that carrier testing of males for FXS is not undertaken routinely as part of the reproductive carrier screen. If clinically indicated, on the basis of family history or clinical features, this can be undertaken on specific request. Due to the predictive nature of this testing, genetic counselling is strongly recommended. Medicare rebates may also apply in such circumstances.

Our clinical and scientific experts have selected the mutations most relevant to these disorders and use the most appropriate technology to detect them. However, the test does not detect every mutation that can cause CF, SMA and FXS, or mutations in other genes responsible for other disorders. If no mutation is found, the risk of the patient being a carrier for these three disorders is greatly reduced; however, the possibility cannot be eliminated.

Genetic counselling

If both reproductive partners are found to be carriers for CF or SMA, or if the woman is found to be a carrier for FXS, it is important to discuss what it might mean for them individually and as a couple.

Genetic counsellors are experienced in providing support and information that help patients make an informed decision.

Genetic counselling is recommended, and is available at no additional cost^ if both partners are carriers of the same autosomal recessive disorder, or the female partner is a carrier for an X-linked disorder. At least one partner must have had carrier testing through Sonic Genetics.

1. Prenatal screening and diagnostic testing for fetal chromosomal and genetic conditions C-Obs 59 [Internet]. RANZCOG. July 2018. <www.ranzcog.edu.au/resources/statements-and-guidelines-directory> (Keyword C-Obs 59)

Arranging a test

1. Complete a Reproductive carrier screening request form or use your standard pathology request form. Please indicate if the patient is already pregnant or has a family history of any of the disorders they are being screened for.
2. Your patient can have a blood sample taken at any Douglass Hanly Moir Pathology collection centre.
3. Results are usually provided within two weeks of the laboratory receiving the sample, and are available electronically via Sonic Dx or downloaded to your practice management system; fax and hard copy reports are also available.
4. Couples tested by Sonic Genetics and found to be at high risk of having an affected child will be offered genetic counselling at no additional cost^ (details on how to refer eligible couples will accompany the results).

Cost

Medicare-rebated testing is available for:

- a female who is pregnant or planning pregnancy, to identify carrier status for CF, SMA or FXS or
- the male reproductive partner of a female identified as a carrier of CF or SMA, to determine reproductive risk for the same condition.

Testing can also be requested for a private fee if Medicare criteria are not met.

For further information regarding this test, please visit sonicgenetics.com.au/rcsd

^Terms and conditions apply.
Please refer to sonicgenetics.com.au/rcs/gc

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For further information, please refer to our website, sonicgenetics.com.au or call us on 1800 010 447

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