Sonic Genetics

Information for Doctors

Reproductive carrier screening

Free testing for male partners of identified carriers of CF or SMA

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 conditions, 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 the equivalent to 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.

Condition	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. Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: outcomes of 12,000 tests. *Genet Med.* 2018; **20**(5):513-526

Targeted screening

- **Cystic fibrosis (CF)** is the most common inherited disorder in Caucasians. It affects respiratory and gastrointestinal function, resulting in progressive lung disease, recurrent respiratory tract infection, pancreatic insufficiency and male infertility.
  The test detects the 50 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 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

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. Sonic Genetics provides this service free-of-charge. 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.

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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 conditions 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 conditions is greatly reduced, however the possibility cannot be eliminated.

Genetic counselling

If both parents 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 free-of-charge^ if at least one partner is tested by Sonic Genetics and both are found to be carriers.

Genetic counselling is also available through a number of public and private providers nationally; for a list of these services, please visit sonicgenetics.com.au/counsellingservices.

1. Genetic carrier screening C-Obs 63 [Internet]. RANZCOG, March 2019. [Accessed May 2021]. <www.ranzcog.edu.au//statements-guidelines/obstetrics/genetic-carrier-screening-(C-Obs-63)>

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

Arranging a test

1. Complete a Reproductive carrier screening request form or use your local pathology request form. Please note 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 of our pathology collection centres.
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 free-of-charge^ (details on how to refer eligible couples will accompany the results).

Cost

Please refer to our website for current pricing, sonicgenetics.com.au/rcsd.

The cost of carrier screening is generally not Medicare-rebatable, except in some cases of CF or FXS testing where there are symptoms or a family history. Male reproductive partners of women who are identified to be carriers of CF or SMA are provided screening for that condition at no cost.

^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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