Sonic Genetics

Information for patients

Medicare rebate available

# Reproductive carrier screening

Cystic fibrosis, spinal muscular atrophy and fragile X syndrome

Reproductive carrier screening, either prior to conception (preferred) or in early pregnancy, can identify your chance of having a child with a serious genetic condition.

This test is becoming an essential part of pregnancy planning, and allows you to make informed decisions about your reproductive options and prenatal care.

The reproductive carrier screening panel available through Sonic Genetics will tell you if you are a carrier for any of three genetic conditions:

- Cystic fibrosis (CF)
- Spinal muscular atrophy (SMA)
- Fragile X syndrome (FXS)*

## Why test for these conditions?

CF, SMA and FXS are three of the most common serious inherited conditions. People are usually unaware that they are carriers and often do not have a history of these conditions in their family.

- CF is the most common life-limiting genetic condition in Australia.
- SMA is the most common genetic cause of death in children under the age of two.
- FXS* is the most common form of inherited intellectual disability.

These conditions do not have a cure, but early treatment and supportive care may improve quality of life.

*Sonic Genetics does not recommend routine reproductive carrier screening for FXS in males.

## What is a carrier?

A carrier is a person who has a genetic change, or mutation, in their DNA, but in most cases does not have any associated health problems. Carriers are able to pass that mutation on to their children, who may then develop a genetic condition.

CF and SMA are recessive conditions, which means that both parents must pass on a mutation for their children to be affected. A couple who are both carriers for the same condition has a one-in-four chance (25%) of having a child that will be affected by that condition.

FXS is an X-linked condition, which means that the mutation is found on the X chromosome. Female carriers of FXS have a one-in-two chance (50%) of passing on the mutation to their children, who in turn may develop FXS.

## What if CF, SMA or FXS runs in your family?

If you have a blood relative who is either a carrier or affected by one of these conditions, you have a greater chance that you will be a carrier. It is very important that you share this information with your doctor before testing is arranged.

A targeted diagnostic test based upon the specific mutation detected in your family may be more appropriate than this screen.

## How common are these conditions?

| 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<br>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

## Testing procedure

Reproductive carrier screening can be performed for individuals or couples. There are two options for carrier testing of couples:

- 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 condition. Reproductive carrier screening of an unaffected male for FXS is not required, as this is an X-linked condition.
- 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.

The best time to find out about your chance of having a child with a serious genetic condition is before you conceive. However, carrier screening can still be performed in early pregnancy.

Speak to your doctor about Sonic Genetics' online resources that explain the various options for carrier screening available to couples.

## Genetic counselling

We strongly recommend that couples with an increased chance of having an affected child seek expert genetic counselling regarding the options available to them.

Couples tested by Sonic Genetics and found to have a high chance will be offered genetic counselling at no additional cost,^ upon referral from your doctor.

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

### How you will receive your test results

Your doctor will discuss the report with you.

If you are a CF or SMA carrier, your doctor will recommend that your partner also be tested to see if they are a carrier of the same condition.

For further information, please refer to our website, sonicgenetics.com.au/rcs/3p

## Arranging a test

1. Your doctor will have completed a Reproductive carrier screening request form or a standard pathology request form, providing details regarding:
   - Your (and your partner's) family history of conditions that affect health, growth and development
   - Previous genetic test results (if any)

2. Your blood sample can be taken at any of our pathology collection centres. No special preparation or booking is necessary.

3. Your result is reported back to your doctor, usually within two weeks of the laboratory receiving your blood sample.

4. Couples tested by Sonic Genetics and found to have a high chance of having an affected child will be offered genetic counselling at no additional cost,^ upon referral from your doctor.

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

## 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.

Your doctor can also request reproductive carrier screening for a private fee if Medicare criteria are not met.

SONIC HEALTHCARE

Sonic Genetics is Sonic Healthcare's centre of excellence in genetic pathology testing. As the largest pathology provider in the country, Sonic Healthcare's state-of-the-art laboratories and extensive network of collection centres serve each state and territory capital, as well as regional and rural Australia.

Douglass Hanly Moir Pathology
Sullivan Nicolaides Pathology
Melbourne Pathology
Capital Pathology
Clinipath Pathology
Bunbury Pathology
Clinpath Pathology
Hobart Pathology
Launceston Pathology
North West Pathology
Southern.IML Pathology


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

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

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