

Information for patients

Reproductive carrier screening

Cystic fibrosis, spinal muscular
atrophy and fragile X syndrome



Reproductive carrier screening can identify your chance of having a child with a serious genetic condition, either prior to conception (preferred) or in early pregnancy. This test is becoming an essential part of pregnancy planning, and allows you to make informed decisions about your reproductive options and prenatal care.

Our test 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.

Arranging a test

- 1 Your doctor will have completed a Reproductive carrier screening request form or a standard pathology request form. Make sure you tell your doctor if you are already pregnant or have a family history of any of the conditions you are being screened for, so that this can be noted on the request form.

- 2 Your blood sample can be taken at any of our pathology collection centres. No special preparation, prepayment 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 free-of-charge[^], upon referral from your doctor.

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

Cost

Please refer to our website for current pricing, sonicgenetics.com.au/rcs/3p.

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.

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 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. 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 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 risk of having a child with a serious genetic condition is before you conceive. However, carrier screening can still be performed in early pregnancy.

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 he or she is a carrier of the same condition.

Finding out that you are a carrier of a genetic condition can raise important concerns around pregnancy planning and care. Depending on your result and the result of your partner, your doctor may refer you to a genetic counsellor to discuss these issues further.

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

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

Barratt & Smith 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

Sonic Healthcare

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