

Preconception carrier screening

Information for patients

Arranging a test

- 1 Your doctor will have completed a Preconception Carrier Screening Request Form. Make sure you tell your doctor if you are already pregnant or have a family history of any of the disorders you are being screened for, so that this can be noted on the request form.
- 2 Your blood sample can be taken at any Douglass Hanly Moir Pathology collection centre. No special preparation or booking is necessary.
- 3 Your sample is tested at one of our NATA-accredited laboratories supervised by a genetic pathologist.
- 4 Your result is reported back to your doctor, usually within two weeks of the laboratory receiving your blood sample.

Cost*

Preconception panel (CF, SMA and FXS)	\$400
CF and SMA <u>or</u> FXS	\$400
CF only	\$310
SMA only	\$220
FXS only	\$101.30

*Prices correct at time of printing.
Please refer to www.sonicgenetics.com.au/pricing for current price.

The cost of carrier screening is generally not Medicare-rebatable except in some cases of FXS testing where there are symptoms or a family history.

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

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Preconception carrier screening provides information on your risk of having a child with a serious genetic disorder. This test is becoming an essential part of pregnancy planning, and allows you to make informed decisions about your reproductive options and prenatal care.

What does preconception carrier screening test for?

Our test will tell you if you are a carrier for three genetic disorders:

- › 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 commonly 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.

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, however, able to pass that mutation on to their children, who may then develop a genetic disorder.

CF and SMA are recessive disorders, 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 disorder have a 25% risk of having a child that will be affected by that disorder.

FXS is an X-linked disorder, which means that the mutation is found on the X chromosome. Female carriers of FXS have a 50% risk 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 disorders, 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.

What is the chance that you could be a carrier?

Condition	People with the condition	Carriers of the condition
CF	1 in 3,000 births	1 in 25
SMA	1 in 6,000–10,000 births	1 in 35
FXS	1 in 7,000–11,000 people	1 in 250
Data from:	www.cysticfibrosis.org.au www.fragilex.org.au www.smaaaustralia.org.au	

Testing procedure

Preconception 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 and tested for the same disorder. Testing of the male is not required for FXS as this is an X-linked disorder.
- › Samples are collected from both partners and tested.

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

How will you get your results?

Your results will be delivered to your doctor who will discuss them 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 disorder.

Finding out that you are a carrier of a genetic disorder 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.