Sonic Genetics
Quality is in our DNA

Preconception Carrier Screening | Request Form

FOR THE DOCTOR

This test should be requested by the doctor responsible for managing a patient's decision-making regarding the preconception carrier screen.

Patient details

First name _____ Surname _____

Date of birth ____/____/____ Sex _____

Address _____

_____

_____

Phone (mobile) _____

Test/s requested

☐ **Preconception carrier screening panel**
Cystic fibrosis (CF, 50 mutations), spinal muscular atrophy (SMA), Fragile X syndrome (FXS)

☐ CF ☐ SMA ☐ FXS

Clinical information REQUIRED

Is there a family history of CF, SMA or FXS? ☐ Yes ☐ No

If yes, please provide details: _____

_____

_____

☐ Pregnant ☐ Not pregnant

☐ Tick here and complete section below if partner sample is also being analysed. Please note that a separate request form will need to be completed.

Partner details

Please complete to enable results to be linked if necessary.

First name _____ Surname _____

Date of birth ____/____/____ Sex _____

Address _____

_____

Phone (mobile) _____

Is there a family history of CF, SMA or FXS? ☐ Yes ☐ No

If yes, please provide details: _____

_____

Requesting doctor

Name _____

Address _____

_____

_____

Phone _____ Provider No. _____

I confirm that this patient has been counselled about the purpose, scope and limitations of the test and has given consent.

Signature ✕ DOCTOR SIGNATURE Date

Copy reports to

Name _____

Address _____

_____

FOR THE PATIENT – Patient and Financial Consent

I have read and agreed to the Patient and Financial Consent section on the reverse of this request form.

Signature ✕ PATIENT SIGNATURE Date

Patient sample collection

Please make sure to bring this form with you on the day of your sample collection. **Medicare benefits do not apply.**

For pricing, please refer to our website – www.sonicgenetics.com.au
For any enquiries, please contact Sonic Genetics on 1800 010 447.

FOR THE COLLECTOR

I certify I established the identity of the patient named on this request, collected and immediately labelled the accompanying specimen(s) with the patient's name, DOB and date/time of collection.

Collector's name: _____

Signature ✕ COLLECTOR SIGNATURE Date

Staff ID/Location code Collection type (stamp)	☐ 1 x 4 mL EDTA Date collected / / Time collected :	PAY CAT

Sonic Genetics
Quality is in our DNA

Preconception Carrier Screening

Information for patients

WHAT IS PRECONCEPTION CARRIER SCREENING?

Preconception carrier screening provides information on your risk of having a child with a serious genetic disorder. Our test will tell you if you are a carrier for three genetic disorders:

- **Cystic fibrosis (CF)** is the most common life-limiting genetic condition in Australia.
- **Spinal muscular atrophy (SMA)** is the most common genetic cause of death in children under the age of two.
- **Fragile X syndrome (FXS)** is the most common form of inherited intellectual disability.

People are usually unaware that they are carriers and often do not have a history of these conditions in their family. These conditions do not have a cure, but early treatment and supportive care may improve quality of life. If you are a carrier, there are several options available for you to explore and these can be discussed with your genetic counsellor or doctor.

TESTING PROCEDURE

Your sample may be collected by your doctor or at any Sonic Healthcare pathology collection centre.

It is important to note that no preconception carrier screen is able to detect all possible disease-causing mutations. However, our clinical and scientific experts have selected the most common mutations and best technology available to detect the vast majority of relevant mutations for these three most commonly inherited conditions. The interpretation of the result may also be dependent on the findings of other investigations.

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

RESULTS

Your results will be delivered to your doctor, typically within two weeks.

- If no mutations are found, this greatly reduces the chance that you are a carrier.
- If a mutation is found, this means that you are a carrier and you have an increased chance of having a child with a genetic disorder. If you are a carrier, your doctor may recommend that your partner also be tested to determine if he or she is a carrier of the same disorder.

FAMILY HISTORY

Please advise your doctor if you have a family history of any of these conditions. If there is a relevant family history, please provide as much information as possible, including any results from any previous testing that may have been performed for other family members.

SONIC GENETICS

We are part of Sonic Healthcare, Australia's largest pathology provider and the third largest pathology provider in the world. We employ highly qualified genetic pathologists, genetic scientists and molecular biologists and operate out of fully accredited laboratories throughout Australia, using state-of-the-art equipment.

PATIENT AND FINANCIAL CONSENT (Please read and then sign the Patient and Financial Consent section overleaf)

I consent to the Sonic Genetics preconception carrier screen being performed and confirm that I have been informed about the purpose, scope and limitations of the test. Sources of information that I can access include my doctor, the Sonic Genetics website and brochures, a genetic counsellor and this request form. I have had the opportunity to ask questions and understand that I can request further information or genetic counselling.

Futhermore, I understand that receiving a result indicating low risk of being a carrier is no guarantee that I am not a carrier of these disorders, as not all mutations can be detected, and all pathology tests have biological limitations. I understand that being a high-risk carrier increases the likelihood of passing on these genes to my offspring but that it is not guaranteed and, without a partner's test results, may be difficult to interpret. I also understand that genetic counselling may be recommended if I am shown to be a carrier.

Finally, I understand that the test requested is not eligible for a Medicare rebate and that I will receive an account which I will pay in full.