Sonic Genetics
Quality is in our DNA

Reproductive 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 reproductive carrier screen for **cystic fibrosis (CF)**, **spinal muscular atrophy (SMA)** and **fragile X syndrome (FXS)**. Please see overleaf for Medicare criteria.

Patient details

First name _____

Surname _____

Date of birth ____/____/____ Sex _____

Address _____

_____

_____

Phone (mobile) _____

Medicare No. ☐☐☐☐☐☐☐☐☐☐

PATIENT STATUS AT TIME OF SERVICE OR SPECIMEN COLLECTION
(Required by law for all patients) Was the patient a:

Private patient in a private hospital or approved day hospital?	☐ Yes	☐ No
Hospital patient in a recognised hospital?	☐ Yes	☐ No
Private patient in a recognised hospital?	☐ Yes	☐ No
Outpatient of a recognised hospital?	☐ Yes	☐ No

Hospital _____ Ward _____

Clinical information

☐ Pregnant ☐ Not pregnant

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

If yes, please provide details: _____

_____

_____ ☐ SD

Test/s requested

☑ **Reproductive carrier screen**
(♀CF, SMA, FXS, ♂CF and SMA only*)
*Sonic Genetics does not recommend reproductive carrier screening of men for fragile X syndrome.

Partner details (For information only – not a test request)

First name _____

Surname _____

Date of birth ____/____/____ Sex _____

☐ Pregnant ☐ Not pregnant

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.

DOCTOR SIGNATURE

X

Copy reports to

Name _____

Address _____

_____

_____

FOR THE PATIENT – Patient and Financial Consent

I confirm that I have been informed about the purpose, scope and limitations of the test.

If I do not fulfil the Medicare criteria

ACCOUNT STATEMENT
I understand that as the test requested is not eligible for a Medicare rebate, I will pay in full prior to the sample being tested.

If I do fulfil the Medicare criteria

MEDICARE ASSIGNMENT (Section 20A of the Health Insurance Act 1973):
I offer to assign my right to benefits to the Approved Pathology Practitioner who will render the requested pathology service(s) and any eligible pathologist determinable service(s) established as necessary by the practitioner.

PATIENT SIGNATURE

X

Practitioner's Use Only (Reason for patient being unable to sign)

For pricing, please refer to our website – www.sonicgenetics.com.au

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 initials	☐ 1 x 4 mL EDTA	Patient initials
Location code	Date collected / /	PAY CAT
Collection type	Time collected :	

Your doctor has recommended that you use one of the subsidiaries affiliated with Sonic Healthcare Limited, an Approved Pathology Authority. You are free to choose your own pathology provider. However, if your doctor has specified a particular pathologist on clinical grounds, a Medicare rebate will only be payable if that pathologist performs the service. You should discuss this with your doctor.

ISSUE JUNE 2020
SHG-REQ-0015-00.4

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Reproductive carrier screening

Information for patients

Purpose

Reproductive 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 one or more of 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.

Limitations

It is important to note that no reproductive 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 majority of relevant mutations for these three common inherited conditions. The interpretation of the result may also be dependent on ethnicity and family history.

Being a carrier increases the likelihood of passing on a mutation to your child, but your chance of having an affected child may be difficult to assess without a reproductive partner's carrier screen result.

Testing procedure

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

Testing can be performed on 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, a sample can be collected from the male partner and tested for the same condition.
- Samples are collected from both partners and tested at the same time.

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.

Family history

If you have a blood relative who is either a carrier or affected by any of the conditions screened, you have a greater chance that you will be a carrier. Where there are symptoms or a family history, a Medicare rebate may be available.

Please advise your doctor if you have a family history of inherited conditions before testing is arranged. 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.

Genetic counselling

Your doctor will be able to provide you with information and advice regarding this test. Your doctor may also recommend that you seek genetic counselling before or after the test.

A genetic counsellor is a professional who provides information and support to patients as they make decisions about their genetic health. We can provide contact details for genetic counselling services nationally. Once the genetic carrier screen is completed, we offer free telephone-based counselling to couples who have been identified as carriers of mutations which place them at high risk of having an affected child. This counselling service requires a referral from your doctor and is only available to couples that have paid Sonic Healthcare or one of its subsidiaries directly for the test.

Results

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

- 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.
- If the female is tested first and found to be a carrier of either cystic fibrosis or spinal muscular atrophy, a free-of-charge test of the relevant gene will be offered to her partner (full details will accompany the test report).

Your doctor can contact our genetic pathologists and senior medical scientists to discuss technical aspects of your result.

Patient and Financial Consent (Please sign overleaf to confirm that you agree with these statements)

By consenting overleaf:

- You consent to the Sonic Genetics reproductive carrier screen being performed and confirm that you have been informed about the purpose, scope and limitations of the test. Sources of information that you can access include my doctor, the Sonic Genetics website and brochures, a genetic counsellor and this request form. You have had the opportunity to ask questions and understand that you can request further information or genetic counselling.
- You understand that receiving a result indicating low risk of being a carrier does not guarantee that you are not a carrier of these disorders, as not all mutations can be detected. You understand that being a carrier increases the likelihood of passing on these genes to your offspring but that it is not guaranteed and, without a partner's test results, may be difficult to interpret. You also understand that genetic counselling may be recommended if you are shown to be a carrier.
- You understand that the test requested may not be eligible for a Medicare rebate and that you will pay in full for the screen prior to sample processing.

Medicare criteria

Indication	Item number	MBS rebate requirements
Cystic fibrosis	73348	Patient has a documented family history of CF mutation in a close biological relative. Can be ordered by a GP or specialist.
Cystic fibrosis	73349	Patient has a reproductive partner with a documented CF mutation. Can be ordered by a specialist only. Please refer to the Medicare Benefits Schedule for details.
Fragile X	73300	Can be ordered by a GP or specialist and subject to certain criteria. Please refer to the Medicare Benefits Schedule for details.

PRIVACY NOTE The information provided will be used to assess any Medicare benefit payable for the services rendered and to facilitate the proper administration of government health programs, and may be used to update enrolment records. Its collection is authorised by provisions of the Health Insurance Act 1973. The information may be disclosed to the Department of Health and Ageing or to a person in the medical practice associated with this claim, or as authorised/required by law.