Genetic Carrier Screening Request Form

GENE ACCESS *Reproductive Carrier Screening for cystic fibrosis (CF), spinal muscular atrophy (SMA) and Fragile X syndrome (FXS)*

ASHKENAZI JEWISH CARRIER SCREENING

AUSTRALIAN Clinicallabs

VIC NSW SA NT 1300 453 688 | Western Australia 1300 367 674

FOR THE DOCTOR

Patient Details

Given Name: .....

Surname: .....

Date of Birth: .....Gender: .....

Address: .....

.....

Ethnic Group: .....

Phone (mobile): .....

Medicare No. ☐☐☐☐ ☐☐☐☐☐ ☐

Test/s Requested

☐ Gene Access Carier Screen (CF, SMA and FXS) **$350**

or by individual test:

☐ CF **$150** ☐ SMA **$195** ☐ FXS **$100**

☐ Ashkenazi Jewish Carrier Screen** **$330**

Clinical information MANDATORY

☐ Pregnant ☐ Not pregnant

Gestational Age: .....Weeks: .....Days: .....

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

If yes, please provide details: .....

.....

Patient Status at Time of Service or Collection

Was the patient a: MANDATORY FOR ALL PATIENTS REQUIRED BY LAW

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
Out-patient of a recognised hospital?	☐ Yes ☐ No

Hospital ..... Ward .....

Partner details (For information only)

Given Name: .....Gender: .....

Surname: .....

Date of Birth: .....

☐ Pregnant ☐ Not pregnant If primary patient is positive for a particular condition, partner can be tested for free.

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

If yes, please provide details: .....

.....

.....

Requesting Doctor MANDATORY

The test must be requested by the clinician responsible for managing a patient's decision-making future plans regarding the reproductive carrier screen.

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.....Date: .....

Please see overleaf for Medicare criteria*

Copy Reports To

Name: .....

Address: .....

.....

Clinic Phone: .....

FOR THE PATIENT – Patient Signature & Financial Consent

I confirm that I have been informed about the purpose, scope and limitations of the test. I understand that the test requested may not be eligible for a Medicare rebate and I may receive an account which I will pay in full.

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..... Date: .....

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

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: .....

Collector Signature: X..... Date: .....

Staff ID/Location code Collection type (stamp)	☐ 1x4 mL EDTA	PAY CAT
	Date collected / /	
	Time collected :	

Your doctor has recommended that you use one of the subsidiaries affiliated with Australian Clinical Labs, 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.

**Note: Ashkenazi Jewish Genetic Carrier Screening includes testing for Diseases such as Tay Sachs, Canavan, Niemann-Pick, Cystic Fibrosis, Bloom Syndrome, Fanconi Anaemia, Familial Dysautonomia and Mucolipidosis IV

Information for patients

This consent form reviews the benefits, risks and limitations of undergoing DNA testing for the genetic disorder(s), as advised by your doctor. This is a voluntary test and you may wish to seek genetic counselling prior to signing this form.

Purpose

Reproductive and Ashkenazi Jewish Genetic Carrier Screening analyzes specific changes, called mutations, that can increase your likelihood of conceiving a pregnancy with a hereditary condition. You may use this information to inform family planning decisions. Disease descriptions, prognoses, and treatment options are available to you through our website. Most of the conditions tested for are inherited in an autosomal recessive manner, meaning that both parents have to carry a mutation in the same disease gene in order to be at risk of having an affected child.

Test Results and Interpretation

If you have a family history of one of the conditions, it is your responsibility to inform the laboratory of the specific gene mutation(s) present in your family. Screening for the diseases on our panel may significantly reduce the likelihood of being a carrier but does not eliminate the possibility of being a carrier. Most of the conditions on the panel are inherited in an autosomal recessive manner, meaning that both parents have to carry a mutation in the same disease gene in order to be at risk of having an affected child. Disease severity can be variable, even within family members with the same mutations. Due to varying modes of inheritance, there are a few diseases on the panel (e.g. Fragile X) that can be transmitted when only one parent is a carrier. The following describes the possible results outcomes:

Carrier (Positive): A positive result indicates that a gene mutation has been identified and therefore you are a carrier of this disorder. You may be identified as a carrier for more than one disorder. Carriers usually do not experience symptoms of the disease.

No mutations detected (Negative): A negative result indicates that no gene mutation was identified. This reduces but does not eliminate the possibility of being a carrier.

No Call: A "no call" describes the inability to confidently report a positive or negative result using stringent quality-control guidelines.

Homozygote or compound heterozygote: This result indicates the presence of two disease-causing mutations, which would typically indicate that you are affected now or may be affected in the future. However, some of the disorders in this panel may be mild and variable in severity and therefore you may not experience clinically significant symptoms. Alternatively, in rare cases, asymptomatic individuals may possess two disease-causing mutations on the same chromosome, potentially requiring further testing for yourself or your family.

Results will be available to your doctor in 7-10 business days from the time the sample is received in the laboratory.

Benefits, Risks and Limitations of Testing

Genetic carrier screening testing is highly reliable. As with all medical screening tests, there is a chance of a false positive or false negative result. A "false positive" refers to the identification of a gene mutation that is not present. A "false negative" is the failure to recognize a mutation that indeed exists. Result interpretation is based on currently available information in the medical literature and scientific databases. While this information may change in the future, Clinical Labs does not routinely reanalyse test results or issue new test reports.

Benefits: Your carrier screening results may help you and your partner make more informed family planning decisions, particularly if screening is performed prior to conceiving a pregnancy. Your results may also benefit family members. If you test positive, your relatives are more likely to test positive for the same mutation(s), thereby discovering previously unknown risks, which would ultimately help them make more informed family planning decisions.

Limitations: This test is designed to detect DNA mutations associated with genetic disease. It cannot detect every mutation associated with each disease, nor does it analyse all known genetic diseases. The genetic carrier screening testing is risk-reducing, not risk-eliminating. Negative results do not guarantee that you or your offspring will be healthy. Some biological factors, such as a history of bone marrow transplantation or recent blood transfusions limit the ability to provide accurate results. Diagnostic errors may occur due to sample mix-up or contamination.

When to Test?

Ideally screening is performed prior to conception to offer greater reproductive choice. However, genetic screening can also be performed in early pregnancy.

Test Procedure

- A sample is collected from the Female partner first. If she is found to be a carrier, a sample can be collected from the partner to be tested for the same condition.
- Samples can be collected from both partners at the same time to test for the genetic carrier conditions.

Legal Agreement

You give permission to Clinical Labs, its contractors and assignees to perform genetic testing on the sample you provided and to disclose the results of the testing to your requesting doctor. You are not an insurance company or an employer attempting to obtain information about an insured person or an employee. You take full responsibility for all possible consequences if you share your test results with others. You agree to hold and assign harmless Clinical Labs, its employees, contractors and successors from any and all liability arising from your disclosure, whether intentional or inadvertent, of your Medical Information and test results to any third parties for diagnostic or any other purposes.

Confidentiality

By signing this informed consent, you provide authorization for your results to be disclosed to your doctor and other entities involved in providing this service to you such as Australian Clinical Labs. You understand that, should you be found to be a carrier for one of the conditions tested for, your results may be shared with Australian Clinical Labs's appointed genetic counselling service who will contact your doctor to discuss the results. Your doctor may elect to have the genetic counsellor contact you directly to discuss your results and if requested, assist with arranging testing of your partner. You understand that test results will not be released to you directly, they will only be made available via your doctor. If both you and your partner are being tested simultaneously, or if your results are subsequently merged, you are authorizing the release of your results to your partner via their doctor, which may include sensitive medical information. Your results may become part of your partner's medical record, which is available to your partner's doctor and other covered entities.

*Medicare criteria

Indication	Item number	MBS rebate requirements
Cystic fibrosis	73345–73350	Can only be ordered by a specialist or consultant physician and subject to certain criteria. 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.

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