Comprehensive Carrier Screen Request Form

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

Patient Details

Given Name: .....

Surname: .....

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

Address: .....

.....

Ethnic Group: .....

Phone (mobile): .....

Test/s Requested

☐ Comprehensive Carrier Screen (287 genes including 21 X-linked Disorders)

Clinical information MANDATORY

☐ Individual ☐ Couple **(Couples must present together with a separate request form filled for each)**

Is the patient: ☐ Pregnant ☐ Not pregnant

If the patient is pregnant, it is recommended that the partner is tested at the same time. Reduced cost applies for partner testing.

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

Conception Information:

☐ Natural ☐ IVF (Patient Egg) | Age at egg retrieval: _____Years
☐ IVF (Donor Egg) | Age at egg retrieval: _____Years

DOB of egg donor: _____/_____/_____

Date of egg transfer: _____/_____/_____

Is there a family history of any genetic disorder? ☐ Yes ☐ No

If yes, please provide details of mutation(s) detected, if known: .....

.....

Do you wish to merge this patient with a previously tested partner?

☐ Yes ☐ No

Partner details (MANDATORY for merged couple reports)

Given Name: .....

Surname: .....

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

Phone (mobile): ..... ☐ Pregnant ☐ Not pregnant

Is there a family history of any genetic disorder? ☐ Yes ☐ No

If yes, please provide details of mutation(s) detected, if known: .....

.....

Partner Consent and Signature

I consent for my information to be included on my partner's report

Partner Signature: X ..... Date: .....

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.
Please note that this test is not Medicare-rebated.

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: .....

Copy Reports To

Name: .....

Address: .....

.....

Clinic Phone: .....

FOR THE PATIENT – Patient Signature & Financial Consent

Price for Carrier Screen is $790 | Partner testing is $700

I confirm that I have been informed about the purpose, scope and limitations of the test. I understand that the test requested is not eligible for a Medicare rebate, and I will pay in full before the test is performed. I consent to my personal information and sample being sent to the Invitae accredited laboratory in the US for analysis and interpretation.

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)	☐ 2x 4mL EDTA	PAY CAT
	Date collected / /	
	Time collected :	

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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

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. A full list of the genes covered in our Comprehensive Carrier Screen can be downloaded from our website: (www.clinicallabs.com.au/doctor/specialists-services/obstetrics-gynaecology). 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. Please advise your doctor if you (or your partner/family) have any inherited or known genetic conditions before testing is arranged.

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.

Benefits, Risks and Limitations of Testing

Genetic carrier screening testing is highly reliable with a >99% accuracy. 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 test is risk-reducing, not risk-eliminating. Negative results do not guarantee that you or your offspring will be free from a genetic predisposition for a particular disorder. 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.
- To avoid medical decision delays, if the female partner is pregnant, it is recommended that both partners are tested at the same time.

Results

Results will be available to your doctor in ~3-5 weeks from the time the sample is received in the laboratory.

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.

Privacy

You give permission to Clinical Labs to transport your sample to Invitae accredited laboratories in the US for analysis and interpretation. privacy and data protection may be different in the US compared to Australia.

Genetic Counselling

For completed reports, Clinical Labs can offer one free consultation with a genetic counselor for positive cases.

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