



## Patient Details

Given Name: .....  
 Surname: .....  
 Date of Birth: ..... Gender: .....  
 Address: .....  
 .....  
 Ethnic Group: .....  
 Phone (mobile): .....

## Test/s Requested

Comprehensive Carrier Screening (up to 569 genes, including X-linked conditions)

## 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.**

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 combine this patient's results with a previously tested partner?

Yes  No

## Partner details (MANDATORY for combined 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 the patient's decision-making future plans regarding the genetic carrier screening. 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 Comprehensive Carrier Screening is \$595 | Partner testing is \$595

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)	<input type="checkbox"/> 2x 4mL EDTA	PAY CAT
	Date collected / /	
	Time collected :	



## Information for patients

This consent form reviews the benefits, risks and limitations of undergoing DNA testing for the genetic condition(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 analyses specific gene changes, called mutations, that can increase your likelihood of having a child with a genetic condition. You may use this information to inform family planning decisions. A full list of the genes covered in our Comprehensive Carrier Screening test can be downloaded from our website ([antenatal.clinicallabs.com.au/patient/resources](http://antenatal.clinicallabs.com.au/patient/resources)). 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. 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 result outcomes:

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

**No mutations detected (Negative):** A negative result indicates that no gene mutation was identified. This significantly 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 conditions 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

The Comprehensive Carrier Screening test is highly reliable with a >99% accuracy rate. 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 recognise 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 other 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 condition. 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 ~3-6 weeks from the time your 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 authorisation 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 combined, you are authorising 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

If you and your partner are both carriers, Clinical Labs can offer you a consultation with an experienced genetic counsellor, upon your doctor's request.