Sonic Genetics
Quality is in our DNA

Powered by fulgent

Expanded 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 Beacon expanded carrier screen. This screen is not suitable for patients seeking Medicare-rebated testing.

Patient details

First name

Surname

Date of birth / / Sex

Address

Phone (mobile)

Clinical information

☐ Pregnant ☐ Not pregnant

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

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

Test requested

	Individual	Couple*
Beacon expanded carrier screen (♀ 327 genes, ♂ 299 genes)	☐ G741	☐ G742

***Couples must present together with a separate request form each.**
Complete partner details below to enable results to be linked.

Please note partner MUST SIGN consent in order for a merged couple report to be provided.

Partner details (For merged couple report only)

First name

Surname

Date of birth / / Sex

Address

Phone (mobile)

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

Signature ✕ PARTNER SIGNATURE Date

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.

I confirm that I have been informed about the process, scope and limitations of the test. I understand that the test requested is not eligible for a Medicare rebate and that I will pay in full prior to testing being performed.

I consent for my personal information and sample to be sent to Fulgent Genetics, a CLIA-accredited US laboratory, for analysis and interpretation.

I also understand that a couple report will not be produced unless my partner provides consent for their results to be shared with me and my healthcare provider.

Signature ✕ PATIENT SIGNATURE Date

Patient sample collection and payment

Please make sure to bring this request form with you on the day of your sample collection.

Payment is required at the time of specimen collection.

For information on cancellation terms and conditions, 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.

☐ I have checked 'Patient and Financial Consent' and 'Partner details' signatures are complete.

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 **SGU**

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Expanded Carrier Screening

Information for patients

Purpose

Reproductive carrier screening is a genetic test used to identify carriers of serious childhood onset inherited conditions. Carriers have genetic changes within their DNA that are unlikely to cause any health problems for them, but increase their risk of having a child with a genetic condition.

Research has shown that most people are a carrier for at least one condition; however, they are often not aware of their carrier status because they do not have symptoms or a family history of disease.

Any adult who wishes to know their carrier status can access these screening tests through their doctor. These screens are particularly useful to couples considering pregnancy. Carrier screening is generally not recommended for minors unless there has been genetic counselling and consent through a clinical geneticist.

Limitations

It is important to note that no genetic 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 the inherited conditions included in the test. The interpretation of the result may also be dependent on the findings of other investigations, your ethnicity and relevant 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 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 of an autosomal recessive disorder, a sample can be collected from the male partner for carrier screening. In approximately 70% of couples, the woman is a carrier and testing of the male partner is recommended.
- Samples are collected from both partners and tested. This reduces the time taken to clarify the risk of having an affected child, and a joint report for the couple is provided. There is a discount for couples being tested together.

Males are not tested for X-linked recessive disorders by this screen, as a man's children are not at high risk of developing such a disorder.

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.

Privacy

DNA from your sample will be extracted in our NATA-accredited laboratory in Sydney and then transported to Fulgent Genetics in the US for analysis and interpretation. This means that your personal information will be subject to the privacy and data protection rules of Fulgent Genetics in the US, which may be different from those of Australia.

A couple report will not be produced unless your partner provides consent for their results to be shared with you and your healthcare provider.

Family history

If you have a blood relative who is either a carrier or affected by any of the conditions screened, there is a greater chance that you will be a carrier. Where there are symptoms or a family history, a Medicare rebate may be available. Rebated testing must be requested on a separate pathology request form and be performed in Australia.

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 give your doctor as much information as possible, including any results from any previous testing that may have been performed for other family members (please refer to the information brochure and state the exact mutation of interest, if known). A targeted test based upon the specific mutation detected in your family may be more appropriate.

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 their subsidiaries directly for the test.

Results

Your results will be sent to your doctor in approximately 3–4 weeks from when the sample is received in the laboratory.

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

Patient and Financial Consent (Please read and then sign the Patient and Financial Consent section overleaf)

I consent to the Beacon expanded 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.

Furthermore, I understand that receiving a result indicating low risk of being a carrier is no guarantee that I am not a carrier of these conditions, as not all mutations can be detected, and all pathology tests have biological limitations. I understand that being a carrier increases the likelihood of passing on these genes to my child but that determining the risk of my child being affected may be difficult if my partner's carrier status is unknown.

Finally, I understand that the test requested is not eligible for a Medicare rebate and that I will pay in full for the screen before my sample is processed.

Sonic Genetics

We are part of Sonic Healthcare which is 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. The DNA is extracted in a NATA-accredited laboratory in Australia by Douglass Hanly Moir Pathology Pty Limited (ABN 80 003 332 858, a subsidiary of Sonic Healthcare Limited) and transported to Fulgent Genetics in the US for analysis.