Sonic Genetics

Service available through Sonic Healthcare practices:

Douglass Hanly Moir Pathology
Sullivan Nicolaides Pathology
Melbourne Pathology
Barratt & Smith Pathology
Capital Pathology
Clinipath Pathology
Bunbury Pathology
Clinpath Pathology
Hobart Pathology
Launceston Pathology
North West Pathology
Southern.IML Pathology

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 gene/mutation(s) detected, if known:

_____

_____

_____

Test requested

	Individual	Couple*
Beacon expanded carrier screen (♀>400 genes, ♂>350 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.

PARTNER SIGNATURE

X _____ Date

Requesting doctor

Name _____

Address _____

_____

_____

Phone _____ Provider No. _____

I have read the Doctor Privacy Consent section on the reverse of this request form. 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 _____

_____

_____

FOR THE PATIENT – Patient Privacy and Financial Consent

I have read the Patient Privacy and Financial Consent section on the reverse of this request form.

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 that I will pay in full prior to testing being performed.

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.

PATIENT SIGNATURE

X _____ 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 sample collection.

For pricing and terms and conditions, please refer to our website – sonicgenetics.com.au/rcs/beacon.

FOR THE COLLECTOR

Sample collection instructions

Please collect 1 x 4 mL dedicated whole blood EDTA tube.
Buccal swab only by pre-arrangement. Store all samples at room temperature.

I certify that 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 ☐ Buccal	Patient initials
Location code	Date collected / /	PAY CAT SGU
Collection type	Time collected :	

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Sonic Genetics

Expanded carrier screening

Information for patients

Purpose

Expanded 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.

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 best technology available to detect most 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 couples:

- A sample is collected and tested from the female partner first. If she is found to be a carrier of an autosomal recessive condition, a sample can be collected from the male partner for carrier screening at a later date. In approximately 70% of couples, the woman is a carrier and testing of the male partner is recommended. This option will therefore add several weeks' delay to getting a final report.
- Samples are collected from both partners and fully tested. This reduces the time taken to clarify the risk of having an affected child, and a joint report for the couple is provided.

Unaffected males are not tested for X-linked recessive conditions by this screen, as a man's children are not at high risk of developing such a condition.

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 be performed in early pregnancy.

Privacy

Your sample will be transported to Fulgent Genetics in the US for analysis. 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. Details about the collection, use and storage of your information can be found on the Sonic Genetics website, sonicgenetics.com.au/privacy-policy.

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

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, available on our website, 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 and 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 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 sent to your doctor in approximately 4–5 weeks from when the sample was received in the laboratory.

- If no mutations are found, this greatly reduces the chance of you being a carrier for the conditions included in the test and of having a child with the conditions tested.
- If a mutation is found, this means you are a carrier and you have an increased chance of having a child with a genetic condition. Your doctor may recommend that your partner be tested to determine if he or she is a carrier of the same condition.

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

Doctor Privacy Consent (Please sign overleaf to confirm that you agree with this statement)

By consenting overleaf:

- You expressly agree to consent to the disclosure of your information to Sonic Pathology Australia Pty Ltd and its relevant affiliated pathology practice(s) (sonicpathology.com.au/our-practices) (including Douglass Hanly Moir Pathology as the hub for this test), and their disclosures of the same to Fulgent Genetics, a CLIA-accredited US laboratory. You acknowledge that Australian Privacy Principle 8.1 will not apply to such disclosures to Fulgent Genetics and that Fulgent Genetics may not be bound by laws that provide the same level of protection for personal information afforded by the Australian Privacy Principles (APPs). You agree that Sonic Pathology Australia Pty Ltd and its affiliated pathology practice(s) will not be responsible for any breach of privacy by Fulgent Genetics, and you will not be able to seek redress under the APPs.

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

By consenting overleaf:

- You consent to the Beacon expanded 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 your 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 conditions, as not all mutations can be detected. You understand that being a carrier increases the likelihood of passing on these genes to your child but that determining the risk of your child being affected may be difficult if your partner's carrier status is unknown.
- You expressly agree and consent to the disclosure of your personal information and sample (including health information) to Sonic Pathology Australia Pty Ltd and its relevant affiliated pathology practice(s) (sonicpathology.com.au/our-practices) (including Douglass Hanly Moir Pathology as the hub for this test), and their disclosures of the same to Fulgent Genetics, a CLIA-accredited US laboratory, for the purpose of analysing and interpreting your sample and data. You acknowledge that Australian Privacy Principle 8.1 will not apply to such disclosures to Fulgent Genetics and that Fulgent Genetics may not be bound by the laws that provide the same level of protection for personal information afforded by the Australian Privacy Principles (APPs). You agree that Sonic Pathology Australia Pty Ltd and its affiliated pathology practice(s) will not be responsible for any breach of privacy by Fulgent Genetics, and you will not be able to seek redress under the APPs.
- You understand that the test requested is not eligible for a Medicare rebate and that you will pay in full for the screen before your sample is processed.