



Sonic
Genetics

Quality is in our DNA

Expanded carrier screening

Information for patients



Screens more
than 400 genes

Informing your chance
of having a baby with
heritable genetic
conditions

Reproductive carrier screening provides information about your chance of having a child with a serious genetic condition. This test is becoming an essential part of pregnancy planning, and allows you to make informed decisions about your reproductive options and prenatal care. In Australia, the Beacon expanded carrier screen is available exclusively through Sonic Genetics.



What does the Beacon expanded carrier screen test for?

The Beacon expanded carrier screen tests more than 400 genes. These genes can cause genetic conditions that are called 'autosomal recessive' or 'X-linked'. Screening can identify if you are a carrier of a certain genetic condition that could affect your baby. This comprehensive screen can detect one in 20 couples at high risk of having an affected child. The decision to have this test is entirely your choice.

What is a carrier?

We all have two copies of most chromosomes, and each parent passes one copy of each chromosome to their child. The chromosomes are made of DNA, the chemical that encodes the genes in every cell of your body. A carrier is a person who has a genetic change, or mutation, in their DNA, but in most cases does not have any associated health problems. Carriers are, however, able to pass that mutation on to their children, who may then develop a genetic condition.

Carriers are usually not affected. The presence of one normal copy of the gene is sufficient to keep a carrier healthy. However, some women who are carriers of an X-linked condition can show signs of the condition.

Why test for these conditions?

In the past, the only clue that a healthy person was a carrier had been the diagnosis of a genetic condition in their affected child. That has now changed. We can examine the genes of a couple to see if they are a carrier and at increased risk of having an affected child before a woman becomes pregnant (preconception) or in early pregnancy.

If a couple is shown to be at increased risk of having an affected child, they can make an informed choice to accept that risk or consider a range of reproductive options such as IVF and prenatal testing to reduce that risk.

When both parents are carriers of the same faulty gene on their chromosomes (autosomal recessive)

There is one in four chance (or 25%) in every pregnancy that their child will inherit both copies of the faulty gene and be affected by the genetic condition. This applies to both boys and girls.

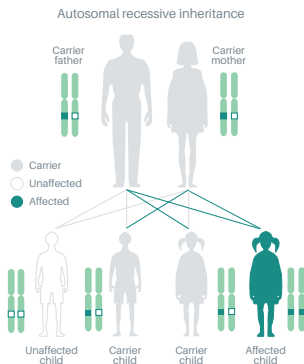
Examples of conditions like this include:

Cystic fibrosis (CF)

Affects the lungs and digestive system and is a life-limiting condition.

Spinal muscular atrophy (SMA)

Progressive muscular weakness and wasting leads to early childhood death without treatment.



Reproductive carrier screening

The Beacon expanded carrier screen looks for variations in many of the genes that cause serious genetic conditions affecting babies and children.

Most of these conditions are very rare. There is usually no family history of the condition and the only way to find out if you are a carrier is to have carrier screening.

Testing is available for unaffected adults before pregnancy (preconception) or in early pregnancy. It can be performed either individually or as a couple.

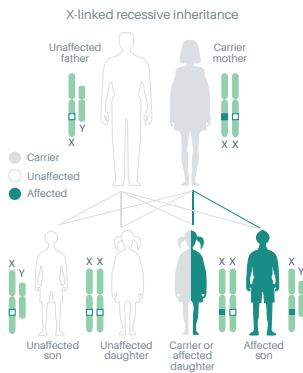
When the mother is a carrier of a faulty gene on her X chromosome (X-linked)

Males have only one X chromosome (inherited from the mother) while females have two. There is one in two chance (or 50%) in every pregnancy that her child will inherit the faulty gene on the X chromosome. Sons will be affected by the genetic condition. Daughters will be carriers of the condition, however, in some cases may also be affected by the genetic condition.

An example of a condition like this is:

Fragile X syndrome

The most common familial cause of intellectual disability in boys; girls can also be mildly affected.



- **Individual testing** – Females are tested for more than 400 genes; this includes more than 50 genes on the X chromosome. Males are tested for more than 350 genes as they do not need to be tested for the genes on the X chromosome.
- **Couple testing** – Both partners are tested at the same time to see if they carry the same faulty gene for any of more than 350 conditions. The woman is also tested for faults in the more than 50 genes on her X chromosome. Please note that testing as a couple gives you information about the risk you have, together, for your pregnancies.

Arranging a test

- 1 Your doctor will have completed an Expanded carrier screening request form, providing details regarding:
 - Your (and your partner's) family history of conditions that affect health, growth and development
 - Previous genetic test results (if any)

This information may impact on the interpretation of your risk of having an affected child.

- 2 Your blood sample can be taken at any of our pathology collection centres. No special preparation or booking is necessary.

- 3 Your sample is tested by Fulgent Genetics and results authorised by one of our genetic pathologists in Australia.

- 4 A report is delivered to your doctor, usually within 3–5 weeks of the laboratory receiving your sample.

Cost

The Beacon expanded carrier screen is not covered by Medicare and is privately billed.*

Full payment is required prior to sample processing.

*Correct at time of printing.

Please refer to www.sonicgenetics.com.au/rcs/beacon for current pricing.

Requests for cancellation of testing prior to sample processing may be possible. An administration fee may apply.

Genetic counselling

We strongly recommend that couples with an increased chance of having an affected child seek expert genetic counselling regarding the options available to them.

- Couples tested by Sonic Genetics and found to be at high risk of having an affected child will be offered genetic counselling free-of-charge,[^] upon referral from your doctor.

[^]Terms and conditions apply. Please refer to www.sonicgenetics.com.au/rcs/gc

Points to consider

- If you and your partner are both found to be carriers of the same faulty gene, or if you are a woman carrying a faulty gene on your X chromosome, there is a greatly increased chance of having a child with a condition due to that gene. It is important for you and your partner to understand what these results mean for your situation.
- Hearing about the carrier result may be unexpected. It may lead to a range of physical and emotional reactions, especially if there are decisions that need to be made. Genetic counsellors are experienced in providing support and information that will help you and your partner make an informed decision.
- We strongly recommend that carrier testing is performed before conception. The earlier that testing is performed, the more time you have to make your decision.
- When testing shows that there is an increased chance of having an affected child:
 - **If you are not pregnant**, you have time to think about the carrier test result when discussing your family planning options. You can consider testing the baby in pregnancy for the condition (prenatal testing). Another option is using IVF and then testing the embryo for the condition before implantation. However, not testing for the condition is also an option.
 - **If you are pregnant**, your options include prenatal testing or not testing for the condition. Prenatal testing and/or IVF are specialist services and may not be widely available by all providers.

Privacy

Your sample and personal information (including health information) are sent to Fulgent Genetics, a CLIA-accredited US laboratory, for the purposes of analysis and interpretation. 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).

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



Sonic Genetics is part of Sonic Pathology Australia, the largest pathology provider in the country. Our state-of-the-art laboratories and extensive network of collection centres serve each state and territory capital, as well as regional and rural Australia.

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

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