Sonic Genetics

Information for patients

Expanded carrier screening

Reproductive carrier screen for more than 400 genetic conditions

Reproductive carrier screening, either prior to conception (preferred) or in early pregnancy, can identify 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.

The expanded carrier screening panel available through Sonic Genetics 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. One in 20 couples will be shown to have a high chance of having a child with one of the conditions covered by this test. You and your doctor can use this information to help you make your own choices about pregnancy planning.

Why test for these conditions?

In the past, the only clue that a healthy person was a carrier was the diagnosis of a genetic condition in their affected child. Reproductive carrier screening can examine the genes of a couple before a woman becomes pregnant (preconception), or in early pregnancy, to see if they are carriers and at increased chance of having an affected child.

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

What is a carrier?

A carrier is a person who has a genetic change, or mutation, in their DNA and does not have any associated health problems. Carriers are usually unaffected because there is a second normal copy of most genes, and this is sufficient to keep the person healthy. However, there are exceptions with mutations on the X chromosome.

If a child inherits an abnormal gene from each parent, the child will have no normal copy of the gene and be affected. Again, there are some exceptions with the X chromosome.

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

There is a one-in-four chance (25%) in every pregnancy that their child will inherit a copy of the faulty gene from each parent and be affected by the genetic condition. This applies to both boys and girls. Examples of conditions like this include cystic fibrosis and spinal muscular atrophy.

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

There is a one-in-two chance (50%) in every pregnancy that her child will inherit the faulty gene on the X chromosome. Sons will be affected by the genetic condition because males have only one X chromosome (inherited from the mother). Females have two X chromosomes and so her daughters will be carriers of the condition and probably unaffected. However, some female carriers of X-linked conditions may be affected. An example of a condition like this is fragile X syndrome.

How common are these conditions?

The 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 procedure

Reproductive carrier screening can be performed for individuals or couples.

- **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; they do not need to be tested for the genes on the X chromosome as they are assumed to be unaffected.
- **Couple testing** – Both partners are tested simultaneously 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 chance you have, together, for your pregnancies.

Speak to your doctor about Sonic Genetics' online resources that explain the various options for carrier screening available to couples.

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 have a high chance will be offered genetic counselling at no additional cost,^ upon referral from your doctor.

^Terms and conditions apply.
Please refer to sonicgenetics.com.au/patient/rcs/gc

How you will receive your test results

Your doctor will discuss the report with you and let you know if any other investigations are recommended. In your initial consultation with your doctor, we strongly advise that you discuss what you would do with the possible results, prior to the test being ordered.

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

For further information, please refer to our website, sonicgenetics.com.au/besp

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)
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. Your result is reported back to your doctor, usually within 3–5 weeks of the laboratory receiving your blood sample.
5. Couples tested by Sonic Genetics and found to have a high chance of having an affected child will be offered genetic counselling at no additional cost,^ upon referral from your doctor.

^Terms and conditions apply.
Please refer to sonicgenetics.com.au/patient/rcs/gc

Cost

Please refer to our website for current pricing, sonicgenetics.com.au/becs. A Medicare rebate is not available for the expanded carrier screen and this test is privately billed.
Full payment is required prior to sample processing.~

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

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).

SONIC HEALTHCARE

Sonic Genetics is Sonic Healthcare's centre of excellence in genetic pathology testing. As the largest pathology provider in the country, Sonic Healthcare's state-of-the-art laboratories and extensive network of collection centres serve each state and territory capital, as well as regional and rural Australia.

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For further information, please refer to our website, sonicgenetics.com.au or call us on 1800 010 447

Sonic Healthcare
Level 22, 225 George Street, Sydney NSW 2000, Australia
sonichealthcare.com.au

SHG-MKT-0070.00.6
*Correct at time of printing | Sept 2022