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WORLD LEADING FERTILITY

Reproductive genetic carrier screening

Carrier screening is all about providing couples with reproductive choices.

Who should have the test?

Australian Obstetric Guidelines recommend that ANY couple who are planning or are in the first trimester of pregnancy should be offered reproductive genetic carrier screening. This test is for anyone regardless of family history, ethnicity or your own health.

What is genetic carrier screening?

Genetic carrier screening is a blood or saliva test which looks to see if a healthy reproductive couple has an increased chance of having a child with an inherited genetic condition. There are hundreds of inherited genetic conditions, some common and others very rare.

Common inherited genetic conditions include cystic fibrosis, spinal muscular atrophy and Fragile X syndrome.

Often, these genetic conditions are inherited from healthy parents who don't know they are carriers and there is no reported family history of the condition.

How are genetic conditions passed on?

Genetic conditions are health or developmental issues caused by genetic 'variants' which stop the gene from working properly. Sometimes, genetic conditions occur when genetic variants are passed on by healthy 'carrier' parents. All of us are 'carriers' of several 'recessive' genetic conditions – many of us will never discover this because carriers generally don't show signs of symptoms themselves. There are two main ways a genetic condition can be passed on from healthy parents which include autosomal recessive or X-linked inheritance (see next page).

What is tested?

Genetic carrier screening can look at a limited panel of common genetic conditions such as cystic fibrosis, spinal muscular atrophy and fragile X syndrome or can look at large panels of 500+ genetic conditions. The genetic conditions screened are generally childhood-onset which have a significant impact on health and development.

Carrier screening is not a personal health test and no test screens for all genetic conditions. If you have a personal or family history of a genetic condition that you would like to have testing for, please contact the Genea Genetic Counsellors as carrier screening might not be the most appropriate test for you.

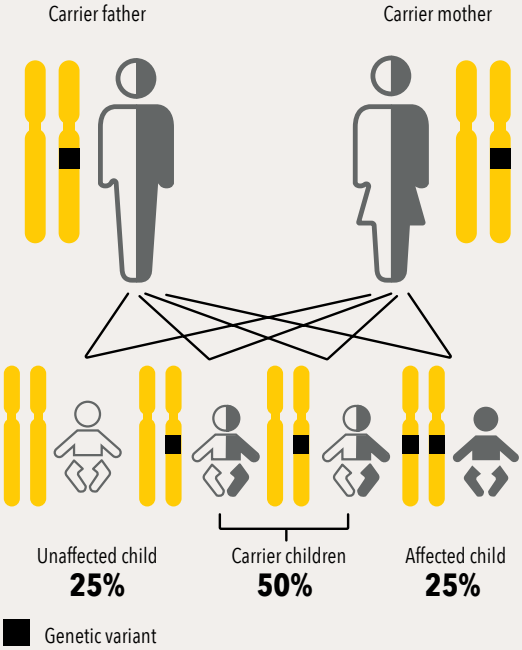
When to test

It's best to test before your fertility treatment, so you can confidently make reproductive decisions. If you have already fallen pregnant, you can still have carrier screening (the earlier in the pregnancy the better to allow time to make informed choices). Ideally both members of a couple are tested at the same time to determine the couple's combined reproductive risk. If you decide to do sequential screening (one partner at a time), ALWAYS TEST THE FEMALE PARTNER FIRST to rule out risk for X-linked conditions.

What happens if we receive a 'high-risk' result?

Your Genetic Counsellor will have a detailed discussion about your options which include:

- Genetic testing of embryos before a pregnancy via IVF (called preimplantation genetic testing or PGT)
- Testing for the condition during pregnancy from 12 weeks gestation via a procedure called a chorionic villus sampling (CVS) or amniocentesis
- Testing your baby for the condition at birth

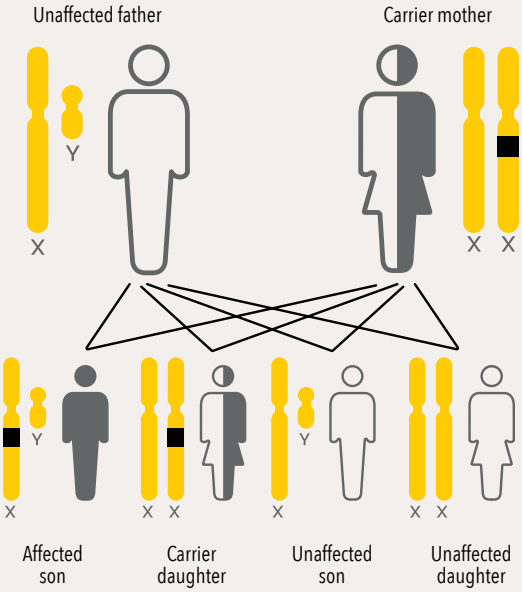


Autosomal recessive inheritance

We all have two copies of each gene, one inherited from our mother and the other inherited from our father. Being a carrier of a condition means that one copy of a gene has a variant (i.e. not working properly) and the other copy is working.

If a child inherits a faulty copy of the same gene from both parents, they will not have a working copy of the gene and will develop the condition.

If a couple are carriers of the same recessive condition, each of their children (males or females) will have a 1 in 4 (or 25%) chance of being affected with that condition. Cystic fibrosis is an example of a recessive condition.



X-linked inheritance

Genes are packaged on our chromosomes. X-linked inheritance is when an altered gene is carried on the X chromosome. Females have two X chromosomes and males have an X and Y chromosome. As males have one copy of the X chromosome, they tend to be more severely affected by X-linked conditions. Female carriers of X-linked conditions are generally unaffected.

Sons of a female carrier each have a 50% (or 1 in 2) chance of being affected with an X-linked condition.

Fragile X syndrome is an example of an X-linked condition.

Reproductive carrier screening options

Some tests are covered by Medicare (limited gene panels) and others have out-of-pocket costs (expanded carrier screening).

Test details	Three gene screen (limited screen)	500+ Gene screen Comprehensive carrier screen or Couples carrier screen
What is tested	3 common genetic conditions · Cystic fibrosis (CF) · Fragile X syndrome (FXS), · Spinal muscular atrophy (SMA)	3 common conditions and over 500 childhood onset severe genetic conditions
Cost	Bulk Billed – Medicare rebate if eligible	Out-of-pocket costs
Timeframe	Within 2 weeks	Approximately 6 weeks
Who is tested	Biological female first	Both members of reproductive couple screened at same time
Report	Female reported first and if a carrier, male reproductive partner then tested	**Comprehensive carrier screening** – Each partner will receive an individual carrier screen report as well as a combined couples report. Or **Couples carrier screening** – combined couples report (high or low reproductive risk report only)
Chance of high-risk result	Around 1 in 240 (0.4%) reproductive couples tested will be reported as increased risk of having a child affected with one of the three conditions screened.	Around 1 in 50 (~2%) couples tested will be reported as increased risk of having a child affected with one of the 500+ conditions screened.

Whilst testing is accurate for the genes screened, all results are reported as low risk, not no risk.
Your reported results are based on current knowledge and reporting guidelines at the time of testing.
Information may become available in the future which may change the interpretation of results.
You should seek updated advice about carrier screening options when planning subsequent pregnancies.

Data from: Archibald A, Smith M, Burgess T, et al. Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: outcomes of 12,000 tests. Genet Med. 2018; 20(5):513-526

Westemeyer M, Saucier J, Wallace J, et al. Clinical experience with carrier screening in a general population: support for a comprehensive pan-ethnic approach. Genet Med. 2020; 22(8):1320-1328

How to order

Genea's recommended carrier screening provider is Eugene who offer a range of carrier screening options as well as expert genetic counselling support throughout the testing process. To access carrier screening through Eugene, please use the QR code and follow the steps below.

Step 1

Order the test via the QR code below or click here eugenelabs.com/pages/genea

Step 2

Provide your health history, consent and doctors details and a saliva collection kit will be sent to your home. Provide a sample and post it back.

Step 3

Eugene will contact you with the results. A summary will be sent back to you and your fertility specialist.

Step 4

If an increased reproductive risk is reported, the Genea Genetic Counselling team will be in contact to discuss your reproductive treatment options.

For any questions, please contact the Genea Genetic Counselling team on **02 8484 6548** or email **genetic.counsellors@genea.com.au**

Comprehensive Eugene Kit –
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