Preconception Genetic Screen

What is a gene?

Genes are inherited from your biological parents – one set from each – and carry information from both parents that will contribute to your appearance, and is why you may have similar physical characteristics to them. But more than that, every gene has a special function to perform in the body.

What is a genetic variant?

We all have some level of variant in our genes, and much of this variant is harmless. In fact, most people are unaware that this variant exists. However some variants mean that important proteins do not function, or do not function as they should. These genetic variants may cause disease.

Why should I consider genetic testing?

There are many recessive variants that do not affect your health, but can cause serious diseases in your children if your partner carries a variant in the same gene. Genetic testing gives us the ability to look for these variants in your genes.

A person is called a carrier when they have a recessive (or silent) genetic variant that does not cause any health issues. If you and your partner both carry the same recessive variant then there is a 1 in 4 chance, in each pregnancy, that you will have a child affected by that genetic disorder.

How does the Preconception Genetic Screen work?

The Preconception Genetic Screen determines your carrier status for 590 diseases. It does this by looking at variants in your DNA in 552 genes. This screen covers genes known to cause diseases in early childhood.

If you are aware of a family history of specific gene variants you should discuss this with your fertility specialist in order to determine if this screen will test for that variant.

It is recommended that the female is screened first and a partner will only need to be tested if she is found to be a carrier.

The test is performed in the Virtus Health Genetics Laboratory using Next Generation Sequencing – the most advance laboratory technology available for sequencing.

Should my partner be tested?

It is likely that the majority of people will have 4-5 DNA changes (variants that may cause disease) in the genes tested. Both you and your partner need to be screened to determine if you both carry variants in the same gene, and are therefore at risk of having a child with a specific disease. We recommend screening at the same time.

What if I test positive?

If you and your partner are identified as carrying a variant from the same gene, your fertility specialist will explain the clinical options available to you and arrange genetic counselling for both of you. Counselling will discuss the clinical symptoms of the disease and available diagnostic options.

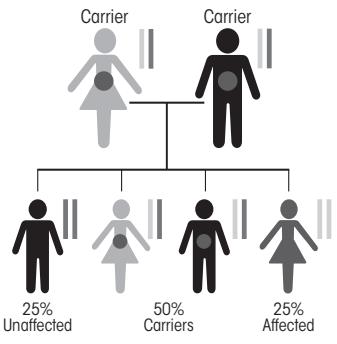
The information you obtain can then be used in planning future pregnancies and deciding on any possible forms of diagnostic intervention (for example preimplantation genetic testing or prenatal diagnosis).

Variants of pathological significance, only carried by one parent, will also be reported and discussed as this information may have significance in your extended family.

How much does it cost?

The test costs $750 per person or $1400 per couple. If either of you have a family history of one of the diseases being tested for, you should inform your treating clinician because you may be entitled to reduced cost testing.

Chances of being a carrier



For a complete list of the tests that the Preconception Genetic Screen covers go to qfg.com.au/preconception