



Virtus Diagnostics offers a complete range of general pathology services as well as specialist fertility and genetic testing.

Virtus Diagnostics conducts over a million tests each year across the following disciplines: general pathology, fertility pathology and advanced genetic testing.

We continually innovate and expand this range and with over 200 scientists, many that are internationally renowned in their field, you can be assured you are accessing world class diagnostics services.

Virtus Diagnostics is committed to supporting patients' health throughout their lifetime, and in particular, during their reproductive life. For women this includes general gynaecology and for both men and women it includes general health, fertility planning and conception, through to infertility treatment.

Our service offers:

- Specialists in clinical genetics
- A comprehensive range of tests
- Fast turnaround of results
- Highly skilled and experienced scientists, with access to the latest technology
- Convenient collection centres, and
- Personalised service

Virtus Diagnostics Collection Centres

The network of Virtus Diagnostics collection centres are across NSW, QLD, VIC & TAS. Please refer to virtusdiagnostics.com.au for the complete list of our collection centres.

Virtus Diagnostics General enquiries
1800 837 284

Virtus Diagnostics Accounts enquiries
1800 090 325

virtusdiagnostics.com.au

Preconception Genetic Screen



2MAY2018

Peace of mind for a healthy baby

The Preconception Genetic Screen is a blood test that you can have before you become pregnant, to help determine your likelihood as a couple of having a baby with a genetic disorder that can negatively impact the baby's health.

There are more than 3,000 inherited disorders that are individually rare, but collectively affect approximately 1% of births.



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.

Genes provide the instructions for the development of proteins and how they function. Proteins are the building blocks of life. They perform many critical functions in the body, including growth and development from the time of conception, reproduction, fighting infection, building muscle and healing wounds.

What is genetic variant?

We all have some level of variation in our genes, and much of this variation is normal.

However, some variants mean that important proteins do not function, or do not function as they should. These genetic variants may cause disease.

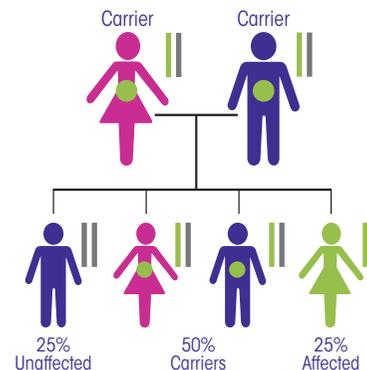
For example, a defect gene (called CFTR) that controls the movement of salt and water in and out of cells causes cystic fibrosis.

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. Research in our laboratory has shown that 1 in 16 of our male patients carries a variant causing cystic fibrosis and 1 in 23 of our female patients carries the same variant. In most cases our patients are unaware they carry this variant.

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. Some of the more common diseases covered include cystic fibrosis, adrenal hyperplasia, adrenoleukodystrophy and phenylketonuria.

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.

The test is performed in the Virtus Health Genetics Laboratory using Next Generation Sequencing - the most advanced 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 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 diagnosis 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?

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.

Please contact Virtus Diagnostics for costings for individuals and couples and a complete list of the tests.