

	virtus core carrier screen	virtus core+ carrier screen
Genes	3 genes Cystic Fibrosis (CFTR) Fragile X Syndrome (FMR1) Spinal Muscular Atrophy (SMN1)	6 genes Cystic Fibrosis (CFTR) Fragile X Syndrome (FMR1) Spinal Muscular Atrophy (SMN1) Alpha and Beta thalassemia (HBA1/HBA2 and HBB)
Testing location	Australia (Virtus Genetics)	Australia (Virtus Genetics)
Turnaround time	Please refer to our website for up to date "turnaround times".	Please refer to our website for up to date "turnaround times".
Sample type	Blood or cheek swab To have cheek swab test kits sent to their home, patients can call Virtus Genetics (1800 837 284) or email a copy of their request form to info@virtusgenetics.com.au. Kits will be available at Virtus collection sites.	Blood or cheek swab To have cheek swab test kits sent to their home, patients can call Virtus Genetics (1800 837 284) or email a copy of their request form to info@virtusgenetics.com.au. Kits will be available at Virtus collection sites.
Price	Bulk Billed Private Fee: \$340 for non Medicare Eligible Patients	Approx. \$150 out of pocket \$490 per person Medicare rebate available for eligible patients.

Test limitations: Carrier screening does not exclude the possibility of having a child affected by a genetic condition. There will remain a small residual risk of having a child affected by one or more of the screened genetic conditions. Test reports rely on currently available information in the medical literature at the time of reporting. A repeat specimen collection may be required in a minority of cases.



How do I arrange to be screened?

- STEP 1:** Discuss carrier screening with your doctor, including any known family history.
- STEP 2:** **Option 1**
Take the request form completed by your doctor to a Virtus Diagnostics collection centre. Specimen collections through other pathology providers may incur additional transport / handling fees from that provider.
- Option 2**
Request a cheek swab kit to be sent to your home. Call 1800 837 284 or email a copy of your request form to info@virtusgenetics.com.au. Kits will include everything you need to self-collect a specimen & instructions on how to post the kit back.
- STEP 3:** Results will be securely returned to your doctor.

To find out more information please email:

info@virtusgenetics.com.au

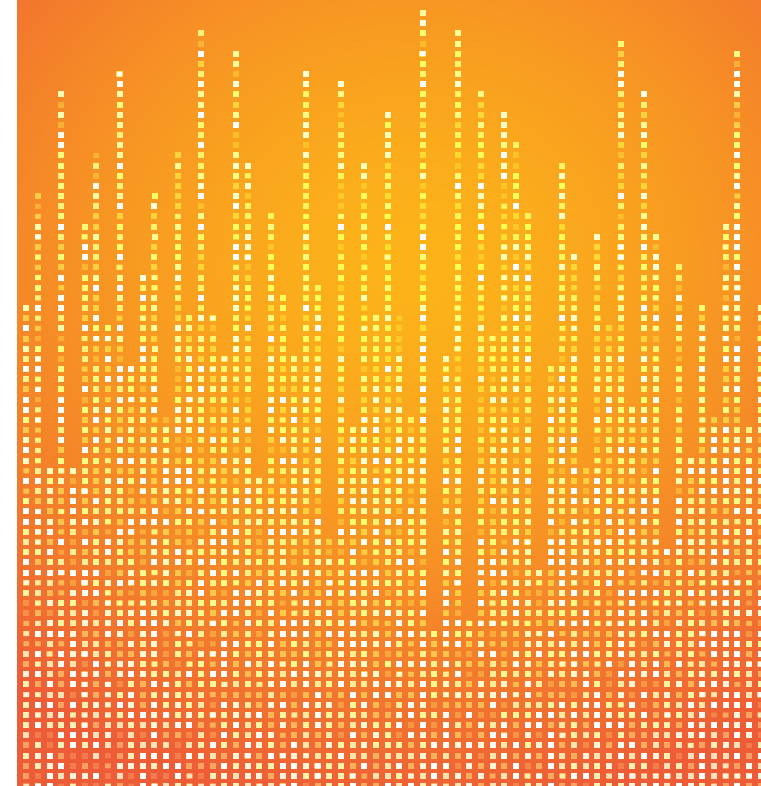
or phone: **1800 837 284**

PATIENT INFORMATION

virtus
core
carrier screen

virtus
core+
carrier screen

Supporting you
through genetic testing



What is the Core and Core+ Carrier Screen?

The Virtus Genetics Core Carrier Screens are a blood or cheek swab test that identifies individuals who are carriers of a genetic condition that can have a significant impact on the child's health and development. These childhood-onset conditions may lead to a shortened life expectancy, intellectual disability, and/or physical disability.

What do the Core Carrier Screens test for?

Core includes

- **Cystic fibrosis (CF)** - CF is associated with respiratory and digestive problems. Approximately 1 in 25 individuals are carriers.
- **Spinal muscular atrophy (SMA)** - SMA is associated with muscle problems throughout the body. Approximately 1 in 40 individuals are carriers.
- **Fragile X syndrome** - Fragile X syndrome is an X-linked condition and the most common known cause of inherited intellectual disability. Approximately 1 in 250 women are carriers.

Core+ also includes

- **Alpha and Beta thalassemia** - Alpha and beta thalassemia are inherited blood disorders.

Who should be screened?

Carrier screening may be suitable regardless of your family history, genetic ancestry, or age. Most children born with a genetic condition will not have a family history of the condition. Nevertheless, if you are aware of a family history of a genetic condition, it is important to discuss this with your doctor.

Unlike chromosomal conditions (e.g. Down syndrome) that occur more commonly with advanced maternal age, the risk of being a carrier of a genetic condition does not vary with age. We are born with our genetic variants, usually inherited from our parents, and these do not change over time.

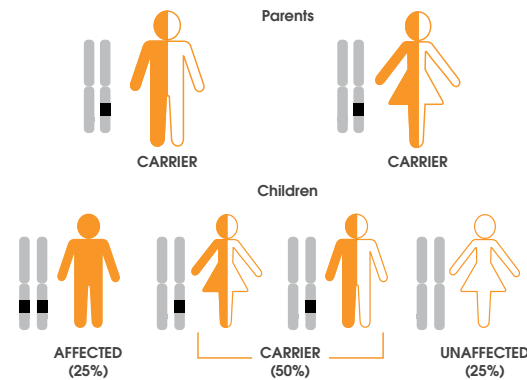
When should I be screened?

The ideal time for reproductive couple carrier screening is when planning a pregnancy. Screening before becoming pregnant gives couples the widest array of reproductive options. Carrier screening is also commonly used in early pregnancy.

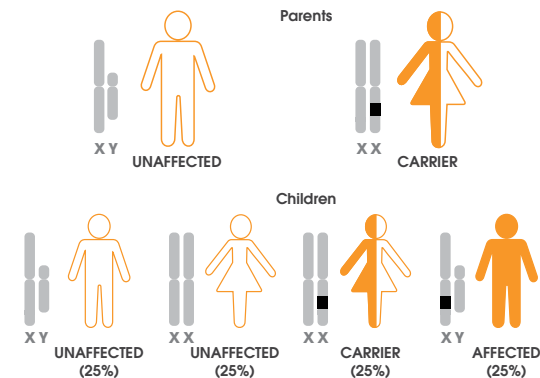
Why should I consider Core Carrier Screening if I have no family history?

Individuals often have no knowledge that they are carriers of a genetic condition. Carriers will have one healthy copy of the gene that works well enough to prevent them from developing symptoms of the condition. In contrast, children affected by these conditions have no working copies of the gene. Inheritance of these genetic conditions can happen in two main ways: Autosomal recessive and X-linked inheritance.

- **Autosomal recessive conditions:** If both the male and female individuals are found to be carriers of a variant in the same gene, there is a 1 in 4 (25%) chance of having a child affected by that genetic condition. Most of the screened conditions are autosomal recessive.



- **X-linked conditions:** Female carriers of an X-linked condition (caused by a gene on the X chromosome) have up to a 1 in 2 (50%) chance of having a child affected by that genetic condition, irrespective of the male reproductive partner's carrier status.



How will these results help me and my partner when planning a family?

Carrier screening results will inform your reproductive decision making. Couples identified as being at high risk of having a child with a genetic condition will have the option of screening embryos with preimplantation genetic testing (PGT) before pregnancy, or the option of testing a baby during pregnancy. Having this information allows couples to make reproductive choices right for them to ensure they have the highest chance of having a healthy family.

Virtus Genetics is a leading provider of PGT in Australia. A Medicare rebate for PGT is now also available for couples at risk of having children affected by a genetic condition.

What results should I expect?

There are two potential screen results:

• LOW RISK

This means that you were not found to be a carrier. You are at low risk of having a child affected with the genetic conditions screened by this test. This is a screening test and there remains a small residual risk of being a carrier. In this scenario, particularly when the female is screened first, partner testing is not required.

• CARRIER

This means that you were found to be a carrier of one or more conditions. For autosomal recessive conditions, carrier screening for these conditions is then recommended in your reproductive carrier.

If both you and your partner are carriers of the same condition, this means that there is at least a 1 in 4 chance of each pregnancy or child being affected by that genetic condition. A complimentary genetic counselling service is available for all reproductive couples who receive such a result.