How do we arrange to be screened?

STEP 1: Discuss carrier screening with your doctor, including any known family history.

STEP 2: **For both options, specimens from the female and male partner must be collected at the same time.**

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 and instructions on how to post the kit back for you and your partner.

STEP 3: Results will be securely returned to your doctor.

Number of screened genes

390 in females

361 in males (excluding X-linked conditions)

Test fee: Test will be privately billed. A Medicare rebate for carrier screening is available for eligible patients and will cover part of the test fee. Visit virtusgenetics.com.au for up-to-date test fees. Medicare eligibility and criteria apply.

Turnaround time: Please refer to our website for up to date "turnaround times".

Testing laboratory: Virtus Genetics, based in Australia.

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.

PATIENT INFORMATION

duo
carrier screen

To find out more information please email:
info@virtusgenetics.com.au
or phone: 1800 837 284

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What is the Duo Carrier Screen?

The Duo Carrier Screen is a blood or cheek swab test that identifies couples at risk of having a child with 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. Although most of these conditions are very rare, collectively they occur in more than 1 in 200 live births.

The Duo Carrier Screen evaluates your genetic risk as a COUPLE, rather than separately determining each person's risk. Test results will show the carrier status for each partner regarding several common genetic conditions, including cystic fibrosis, spinal muscular atrophy, fragile X syndrome, and alpha and beta thalassemia. However, carrier status for other tested genes will only be disclosed if there is a risk to your offspring or to your own health. The test result is therefore specific to the COUPLE being tested - if either person changes partners or a sperm/egg donor is used, repeat carrier screening will be required.

Who should be screened?

The Duo Carrier Screen 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.

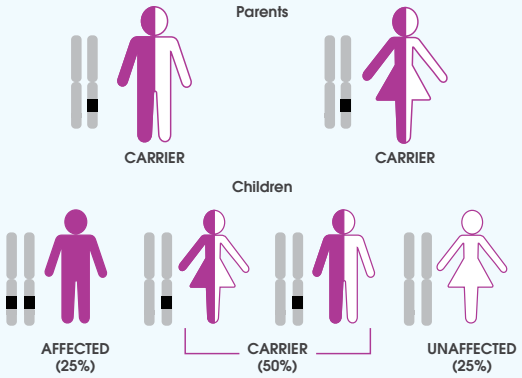
When should we be screened?

The ideal time for Duo 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.

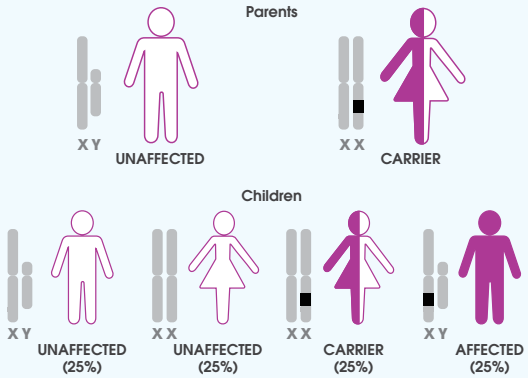
Why should we consider Duo Carrier Screening if we have no family history?

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 us 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 we expect?

There are different potential screen results:

- **LOW RISK**

  This means that you and your partner are at low risk of having a child with the genetic conditions screened by this test. You and your partner may individually be carriers of one or more genetic conditions, but you are not both carriers of the same condition. This is a screening test and there remains a small residual risk of having a child affected by these genetic conditions.

- **HIGH RISK**

  This means that you and your partner are at high risk of having a child with a genetic condition. This usually occurs when both you and your partner are carriers of the same condition. It usually 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 couples who receive such a result.

- **IMPLICATIONS FOR YOUR OWN HEALTH**

  It is very important to be aware that genetic testing can sometimes have significant implications for your own health, such as revealing an undiagnosed medical condition or increased risk of developing a medical condition. These results may also have implications for your other family members.