

## What is Carrier Screening?

Carrier Screening is a way to see whether you carry certain gene mutations that do not affect you, but can affect your baby. If both you and your partner are carriers of the same genetic disorder, your baby has a significant chance of suffering from a serious genetic disease.

### This is performed using a simple blood test.

Virtus Reproductive Carrier screening will allow you to find out if you are a carrier of three of the most common inherited conditions:

- Cystic Fibrosis (CF)
- Fragile X Syndrome (FXS)
- Spinal Muscular Atrophy (SMA).

We strongly recommend that couples with an increased chance of having an affected child seek expert genetic counselling regarding the options available to them

## Virtus Genetics Counselling

Virtus Genetics offers a comprehensive genetic counselling service that is available to our referring doctors and their patients.

**For further information on our Genetic Counselling phone our laboratory on 1800 409 250**

*The Royal Australian and New Zealand College of Obstetricians and Gynaecologists recommend all pregnant women, or anyone planning pregnancy, be provided with information and have access to screening for chromosomal and genetic conditions.*

VirtusGenetics 

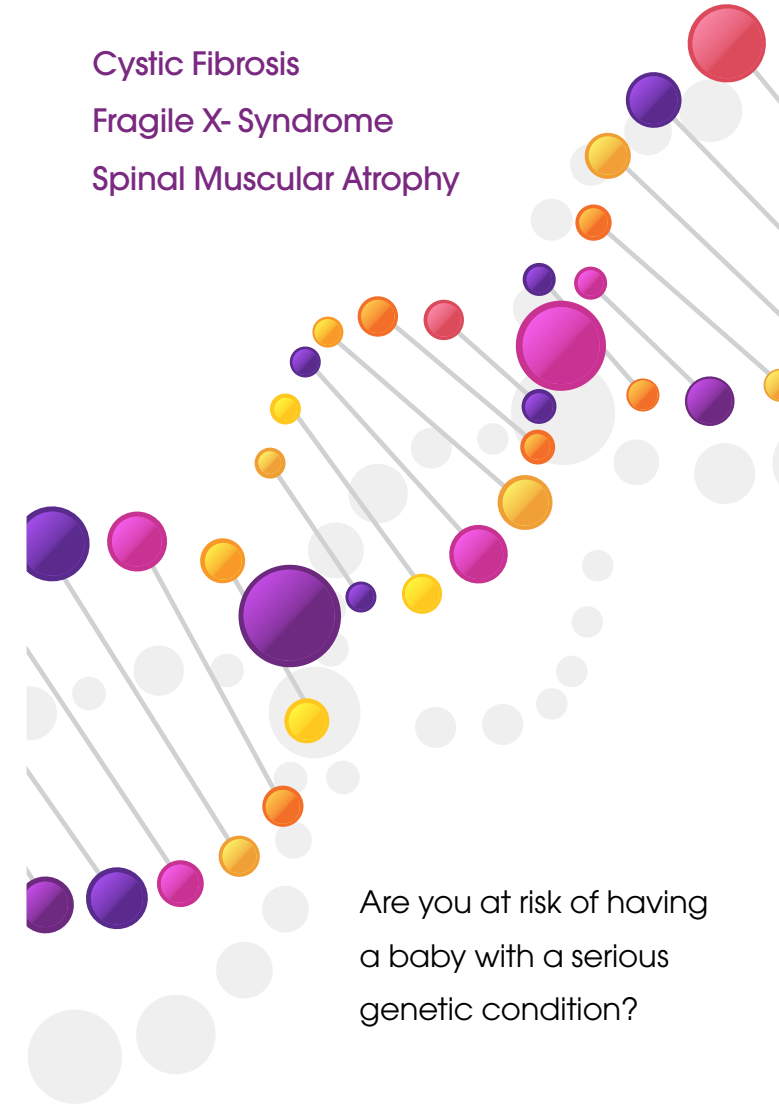
General Enquiries: 1800 837 284  
[www.virtusgenetics.com.au](http://www.virtusgenetics.com.au)

# Genetic Carrier Screen

Cystic Fibrosis

Fragile X-Syndrome

Spinal Muscular Atrophy



Are you at risk of having a baby with a serious genetic condition?

## CYSTIC FIBROSIS (CF)

Cystic Fibrosis (CF) is an inherited disorder which affects the respiratory and the digestive system. Thick mucus builds up, trapping bacteria leading to recurrent infections. Thick mucus in the gut makes digestion of food difficult. CF patients require medical treatment from birth and throughout their entire life. Daily physiotherapy is needed in most cases, as is medication, to treat lung infections and to assist digestion.

**For further information please visit [cysticfibrosis.org.au](http://cysticfibrosis.org.au)**

## FRAGILE X SYNDROME (FXS)

Fragile X syndrome (FXS) causes intellectual disability that ranges from mild, through moderate to severe. FXS affects both males and females with symptoms including learning difficulties, anxiety, autism, epilepsy and hyperactive behaviour. There is no cure for Fragile X but some educational, behavioural and medical interventions can improve outcomes in some people.

**For further information please visit [fragilex.org.au](http://fragilex.org.au)**

## SPINAL MUSCULAR ATROPHY (SMA)

Spinal Muscular Atrophy (SMA) is a neuromuscular disorder characterised by loss of motor neurons and progressive muscle wasting. Age of onset and severity is variable, from severe forms causing death in early childhood to later onset forms which may not reduce life expectancy. There is no cure for SMA, however there are treatments and interventions available.

**For further information please visit [smaaustralia.org.au](http://smaaustralia.org.au)**

## What causes these conditions?

Genetic conditions are caused by changes in genes, which provide instructions for our bodies. Babies inherit one copy of each gene from each parent.

<b>Cystic Fibrosis</b>	Occurs when a baby inherits a gene change from both parents
<b>Fragile X Syndrome</b>	Only females who are carriers of a Fragile X gene that is increased in size are at risk of an affected baby. They have a 50% chance of passing this on to their baby.
<b>Spinal Muscular Atrophy</b>	Occurs when a baby inherits a gene change from both parents

## Why test for these conditions?

Even without a family history of genetic disease, you can still be a carrier. When two people are carriers of the same disease, they can unknowingly have a child with serious health issues. We can now examine genes of couples planning pregnancy or those in early stages of pregnancy to see if they have an increased risk of having an affected baby.

## What is the chance I could be a Carrier?

	Number of people with the condition	Number of people who are carriers of the condition
<b>Cystic Fibrosis (CF)</b>	1 in 2500	1 in 25
<b>Fragile X Syndrome (FXS)</b>	1 in 4000	1 in 50
<b>Spinal Muscular Atrophy (SMA)</b>	1 in 6000 to 1 in 10000	1 in 50

## I have a family member with CF, FXS, or SMA

If you have a family history of one of these disorders, you and your partner may have a greater risk of affecting your baby and you should both consider screening.

If you have a new partner for a subsequent pregnancy, consideration should be given to having your new partner tested.

## When should I be screened?

It is recommended that the female is screened first and her partner tested only if she is found to be a carrier. You can be screened prior to becoming pregnant or if you are already pregnant then we recommend screening before 12 weeks.

## Accuracy of testing

The current screening tests detect most gene changes but they cannot detect every possible change in these genes that may cause the disorder. There is still a small chance that you may be a carrier.

## How much does this screen cost?

This test is not covered by Medicare or your private health fund. Visit our website [virtusgenetics.com.au](http://virtusgenetics.com.au) for more information. In some cases of CF or FXS testing, where there are symptoms of family history, you may be eligible for a Medicare rebate.

## I want to be screened – what do I do now?

Once you've made the decision to be screened your doctor will complete the Virtus Genetics request form and arrange a simple blood test. It takes approximately 10-15 days for your test to be processed. Your results are sent back to your referring doctor.

## My partner and I are carriers

### – What happens now?

It is important to know you have options. Your doctor will discuss the results with you, guide you through various options and offer genetic counselling. Knowing your carrier status before or early in your pregnancy gives you time to learn about the disorder and make informed decisions about your reproductive options and prenatal care.