Virtus Genetic Screen (VGS)

Planning a Pregnancy: could you be a carrier of a common genetic condition? Carriers of commonly inherited conditions such as; Cystic Fibrosis (CF), Fragile X Syndrome (FXS) and Spinal Muscular Atrophy (SMA) are generally healthy adults with no family history of the disorder and consequently unaware of their carrier status. Preconception screening will allow you to find out if you and your partner are carriers and at risk of having a child with one of these conditions.

Genetic carrier screening gives individuals and couples information about their risk of having a child with a genetic condition. The VGS test is a simple blood test which can be arranged through your doctor. It will tell you if you are a carrier for three commonly inherited conditions: Cystic Fibrosis, Fragile X Syndrome and Spinal Muscular Atrophy.

What is the chance that I could be a carrier?

If you have a family history of one of these disorders, you and your partner may have a greater risk than the general population, of an affected child and you should both consider screening. If you have a new partner for a subsequent pregnancy, consideration should be given to having the new partner tested.

What causes these conditions?

Genetic conditions are caused by changes in genes, which provides the instructions for our bodies. Babies inherit one copy of each gene from each parent. Some people carry changes in a gene which can mean there is a chance that they may have a child with a genetic condition.

If the tests show that we are at risk of having a child with CF, FXS or SMA, what happens next?

If you or your partner are carriers of CF, FXS or SMA you will be offered genetic counselling. The genetic counsellor will talk to you about your results, options for further testing and can help with making decisions and providing support.

What is Cystic Fibrosis?

Cystic Fibrosis is an inherited disorder that permanently affects the respiratory (lungs) and digestive systems with its effect on sweat glands, saliva, tears and mucus. The thick mucus entraps bacteria causing gut infections, reduced pancreatic function and recurrent lung infections which may lead to irreversible damage. Lung failure is the major cause of death for someone with CF.

A person with CF requires medical treatment from birth and throughout their entire life. Daily physiotherapy is needed for their lungs and frequent medical treatments such as antibiotics are required to treat bacterial infections.

What is the chance that I could be a carrier?	Number of people with the condition	Number of people who are carriers of the condition
CYSTIC FIBROSIS CF	1 in 2500	1 in 20 to 1 in 30
FRAGILE X SYNDROME FXS	1 in 4000	1 in 150
SPINAL MUSCULAR ATROPHY SMA	1 in 6000 to 1 in 10,000	1 in 40 to 1 in 60

Despite modern treatment there is currently no cure for CF sufferers hence experiencing a reduced life expectancy, often needing extreme treatments like a heart and lung transplant.

As carriers of the CF gene are typically unaffected with no symptoms Male CF carriers frequently have an altered gene that is associated with infertility, and carrier rates for CF are higher in infertile couples.

What is Fragile X Syndrome?

Fragile X syndrome causes intellectual disability that ranges from mild, through moderate to severe. It affects both males and females, but males are more likely to be affected. Females usually have milder intellectual symptoms. Symptoms can include delays in talking, anxiety and hyperactive behaviour.

Fragile X affects both males and females with symptoms including learning difficulties, anxiety, autism and epilepsy and hyperactive behaviour. Males are more likely to be affected with physical indicators including large ears, a long face, a prominent jaw, forehead and flat feet. Females usually have milder intellectual symptoms however can develop premature ovarian failure before the age of 40 years.

There is no cure for Fragile X but some educational, behavioural and medical interventions can improve outcomes in some people with Fragile X.

What is Spinal Muscular Atrophy?

Spinal Muscular Atrophy (SMA) is a neuromuscular disorder characterised by loss of motor neurons and progressive muscle wasting. Age of onset and severity are variable, from severe forms causing death in early childhood to later onset forms which may not reduce life expectancy.

A drug has recently been released in Australia to help in the treatment SMA after an individual has been diagnosed with the disorder; however this medication is not a cure.

Family history of 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 child than the general population and you should both consider screening.

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

Accuracy of testing

The current screening tests detect most carriers 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.

While there is a range of providers offering CF this screen is the most comprehensive CF screen identifying over 1000 gene variants.

VGS Screening costs

VGS Screening costs Genetic screening is not covered by Medicare. For current test fees, please contact Virtus Diagnostics on 1800 090 325.