Genetic Carrier Screen

Cystic Fibrosis, Fragile X-Syndrome, Spinal Muscular Atrophy

GENETIC CARRIER SCREEN

Virtus Genetics Carrier Screen tests for Cystic Fibrosis (CF), Fragile X-Syndrome (FXS), and Spinal Muscular Atrophy (SMA)

Carrier screening is a test performed to determine whether your patient may carry a gene mutation that may affect their chance of having a child with a genetic condition.

Your patient can be screened prior to becoming pregnant, or, if your patient is already pregnant then we recommend screening prior to 12 weeks.

It is advised that your female patient be screened first and her partner tested only if she is found to be a carrier. However, an advantage of screening as a couple is that if a positive screen occurs it does alleviate the anxiety while waiting for the second result.

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*

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 FXS but some educational, behavioural and medical interventions can improve outcomes in some people.

For further information please visit *fragilex.org.au*

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*

WHY TEST FOR THESE CONDITIONS?

Even without a family history of a genetic disease, you can still be a carrier. When two people are carriers of the same disease, they can unknowingly have a child with a serious health problem.

We now examine genes of couples to see if they have an increased risk of having an affected child.

These three conditions combined are amongst the most commonly carried mutations in European populations.

MODE OF INHERITANCE

	Mode Of Inheritance	Number of People with the condition	Number of people who carry the condition
CF	**Autosomal Recessive.** Occurs when a baby inherits a gene change from both parents.	1 in 2500	1 in 25
FXS	**X-Linked.** Females who are carriers of a Fragile X gene that is increased in size are at risk of an affected baby.	1 in 4000	1 in 50
SMA	**Autosomal Recessive.** Occurs when a baby inherits a gene change from both parents.	1 in 6000 to 1 in 1000	1 in 50

VirtusGenetics

Genetic Carrier Screen

Cystic Fibrosis, Fragile X-Syndrome, Spinal Muscular Atrophy *(continued)*

WHY CHOOSE VIRTUS GENETICS OVER OTHER LABORATORIES?

Our specialists Genetics Laboratories have developed as centres of excellence. Our technology and research in the area of reproductive health is recognised with national and international expertise available to consult with our referring clinicians.

CYSTIC FIBROSIS

NextGeneration Sequencing data is used to screen for >99% of disease causing variants (to date 336 disease causing variants in the CFTR2 data base).

SPINAL MUSCULAR ATROPHY

The common deletions are detected(95% of cases). NextGeneration Sequencing data is used for rarer DNA variants known to cause SMA.

FRAGILE X

Repeat size is determined; normal, intermediate, pre-mutation and full expansion repeats are detected.

HOW DO I ORDER THIS TEST ON MY PATIENT?

After discussion with your patient complete our General Pathology request form. These are supplied by our Laboratory or can be downloaded from our website.

Ensure all patient details are complete, including all relationships to your patient and any family history. This information is essential and will assist your patients Medicare eligibility.

Always indicate pregnancy status on request.

Sample Collection

This test is performed using a simple blood test. The sample is collected in an EDTA tube.

You may choose to collect your patients' sample at your clinic and have our courier call to collect and deliver to our genetics laboratory. Alternatively you can refer your patient to one of our collection centres.

HOW MUCH DOES THIS TEST COST?

This test is **NOT** covered by Medicare or any Private Health Care Funds.

In some cases for CF and FXS testing, where there are symptoms of family history, a medicare rebate may apply.

For prices & rebate information visit our website: www.virtusgenetics com.au or phone our accounts department on 1800 090 325.

MY PATIENT IS A CARRIER

If your patient is found to be a carrier of these conditions it is important that they know they have choices available to them. Making informed decisions about their reproductive options is paramount. Our genetic counselling service can assist during this time to;

- Assist in interpreting results.
- Learn more about the genetic conditions their child may inherit.
- What to expect when continuing with their pregnancy.
- IVF options including pre-implantation genetic diagnosis (PGD), possible sperm or egg donation.
- Adoption.

VIRTUS GENETICS COUNSELLING SERVICE

A positive result can be an emotional and confusing time for your patient. We strongly recommend that couples with an increased change of having an affected child seek expert genetic counselling regarding options available.

Virtus Genetics offers a national, comprehensive reproductive genetic counselling service. This service is available to our referring doctors and their patients.

Medical Genetic consultations with Dr Coman can be facilitated rapidly either face to face or via Skype.
Phone: 1800 409250
Fax: 07 3319 6617
Email:reception@drdavidcoman.com.au

For further information contact our laboratory on 1800 837 284

VirtusGenetics