

# Extended Genetic Carrier Screen

Testing for 553 Autosomal and X-linked diseases

## AN ESSENTIAL START TO PLANNING A FAMILY

In July 2018, 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.

Virtus Genetics offers the most comprehensive preconception carrier screening in Australia, offering testing for 553 Autosomal and X-linked inherited disease.

*For a complete list of all Genetic Diseases screened visit our website at [www.virtusgenetics.com.au](http://www.virtusgenetics.com.au)*

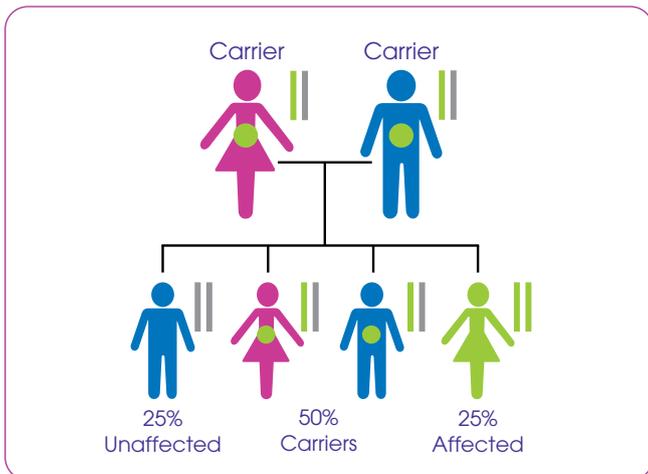
## WHY SCREEN FOR THESE CONDITIONS PRIOR TO BECOMING PREGNANT?

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.

There are hundreds of inherited genetic disorders that can cause serious health problems. We can now examine genes of individuals and couples to see if they have an increased risk of having an affected child.

The Extended Genetic Carrier Screen can identify genetic disorders in the family by reviewing a number of factors which include:

- Family members with learning difficulties
- Family members with a genetic disorder
- History of miscarriage
- Stillbirth pregnancy



## WHEN SHOULD I REFER MY PATIENT FOR TESTING?

The ideal time for screening is prior to your patient becoming pregnant.

It allows both yourself as the referring clinician and your patient time to consider the results and make informed choices about their future plans.

If your patient is already pregnant then we recommend screening be done as early into the pregnancy as possible. This is not ideal, as there are limited choices for your patient with fewer options available.

## INDIVIDUAL VS COUPLES SCREENING.



### Individual Screening

Testing the woman first is recommended. This includes testing for X-linked recessive disorders; the genes responsible for these disorders are not routinely tested in men, as their children are not expected to be affected.

If a patient is tested individually and found to be a carrier for an autosomal recessive disorder, it is then necessary to test the partner to determine the risk to their child.



### Couples Screening

An advantage of screening as a couple is that if a positive screen occurs from individual testing, it does alleviate the anxiety while waiting for a second result.

# Extended Genetic Carrier Screen

Testing for 553 Autosomal and X-linked diseases (*continued*)

## WHY CHOOSE VIRTUS GENETICS?

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.

The Virtus Genetics Extended Genetic Carrier Screen is for all ethnic groups. It includes all genes of interest to those of Ashkenazi Jewish ancestry as well as diseases screened for by Newborn screening programs.

Mendelian disorders are individually rare but collectively affect 1-2% of births in the UK. They account for 20% of infant mortality and 18% of paediatric hospitalisation.

All DNA data remains in Australia with Virtus Genetics and is available for re-analysis should further genetic information become available in a particular family.

## HOW DO I ORDER AN EXTENDED GENETIC CARRIER SCREEN 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 relationship 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 of CF and FXS testing, where there are symptoms of family history, a Medicare rebate may apply.

For prices and rebate information visit our website on [www.virtusgenetics.com.au](http://www.virtusgenetics.com.au) or phone our accounts department on 1800 090 325.

## MY PATIENT IS A CARRIER, WHAT DO I DO NOW?

If your patient is found to be a carrier of a genetic condition 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;

- Help with interpreting results.
- Learn more about the genetic conditions their child may inherit.
- What to expect when continuing with their pregnancy.
- IVF options including preimplantation genetic diagnosis (PGD) and 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 chance 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 409 250

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