THE COUNSYL FORESIGHT CARRIER SCREEN

AT A GLANCE

- 175+ diseases with one simple test.
- Leading 'at risk' couple detection rate[1].
- Multiple peer reviewed publications validating test design, performance and clinical utility.
- Single and couple reporting with reproductive risk.
- Includes local genetic counselling to ensure you understand your results. Call 1300 268 6795 for counselling queries.
- Proven local track record in Australia since 2009.
- Over 1 million patients tested world-wide.

KNOW THE FACTS ABOUT GENETIC DISEASE

- Inherited genetic disorders account for more than 10% of deaths in childhood.
- Rare disorders, including diseases covered by the Counsyl Foresight Carrier Screen, affect up to 3% of babies.
- Each year many Australian couples are at risk of having a child with a preventable genetic disease.
- Most children with a recessive genetic disorder are born to parents with no prior family history.

HOW CAN I GET SCREENED?

The Counsyl Foresight Carrier Screen is prescribed by your doctor.

Results are available in 2 to 3 weeks from receipt of specimen in the Counsyl lab in California and are returned to your doctor.

The Counsyl Foresight Carrier Screen is currently not covered by Medicare or private health insurance in Australia.

You will need to pay for your test before your blood collection by calling 1800 822 999. You will be given a receipt number that should be written on your request form in the receipt box.

Attend your nearest Dorevitch Pathology collection centre to have your sample collected.

Price per person tested is $699*.

*Prices correct at May 2019 and are subject to change without notice.

1. Hogan, G., Vysostskaia, V., & Beauchamp, K (2018). Validation of an Expanded Carrier Screen that Optimises Sensitivity via Full-Exon Sequencing and Panel-wide Copy Number Variant Detection. *Clin. Chem.* 64:7, p1-11.

THE COUNSYL FORESIGHT CARRIER SCREEN CAN DETECT 175+ HEALTH CONDITIONS

Beta Thalassemia • Cystic Fibrosis • Spinal Muscular Atrophy
Bloom Syndrome • Canavan Disease • Familial Dysautonomia
Gaucher Disease • Niemann-Pick Disease • Tay-Sachs Disease

ABCC8-Related Hyperinsulinism • Adrenoleukodystrophy
Alpha Thalassemia • Alpha-Mannosidosis • Argininosuccinic Aciduria
Ataxia With Vitamin E Deficiency • Bardet-Biedl Syndrome
Biotinidase Deficiency • Carnitine Palmitoyltransferase IA Deficiency
Carbamoylphosphate Synthetase Deficiency • Cartilage-Hair Hypoplasia
Cystinosis • DFNB 1 Nonsyndromic Hearing Loss and Deafness
Delta-Sarcoglycanopathy • Duchenne/Becker Muscular Dystrophy
Familial Mediterranean Fever • Fanconi Anemia • Galactosemia
Glutaric Acidemia • Glycogen Storage Diseases
Hereditary Fructose Intolerance • Hereditary Thymine-Uraciluria
Infantile Refusum Disease • Isovaleric Acidemia
Joubert Syndrome 2 • Krabbe Disease
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency
Maple Syrup Urine Diseases • Megalencephalic Leukoencephalopathy
Medium Chain Acyl-CoA Dehydrogenase Deficiency
Metachromatic Leukodystrophy • Mucolipidosis IV
MYH-Associated Polyposis • Nemaline Myopathy
Neuronal Ceroid Lipofuscinosis • Ornithine Transcarbamylase Deficiency
Pendred Syndrome • Phenylalanine Hydroxylase Deficiency
Polycystic Kidney Disease • Primary Hyperoxaluria • Pycnodysostosis
Sickle Cell Disease • Smith-Lemli-Opitz Syndrome
Tyrosinemia • Usher Syndrome • Wilson Disease
X-Linked Juvenile Retinoschisis • Fragile X Syndrome

full list of diseases screened is available at
https://myriadwomenshealth.com/diseases

Limitations: As with any medical diagnostic test, genetic screening is risk reducing, rather than risk-eliminating. Results are based on probabilities, and as such, cannot diagnose or predict all disease.

Genomic Diagnostics
LEADING THE WAY TO IMPROVE HEALTH

FOR ANY QUESTIONS, TALK TO OUR FRIENDLY CUSTOMER CARE TEAM CALL

1800 822 999 between 9.00 am and 5.00 pm (EST)

OR EMAIL US AT
info@genomicdiagnostics.com.au

Specialist Diagnostic Services Pty Ltd ABN 84 007 190 043 APA trading as Dorevitch Pathology
IS-MKT-124. V1 July 2019

Genomic Diagnostics

Dorevitch PATHOLOGY

COUNSYL FORESIGHT CARRIER SCREEN

myriad® WOMEN'S HEALTH

THE COUNSYL FORESIGHT CARRIER SCREEN

WHAT IS THE COUNSYL FORESIGHT CARRIER SCREEN?

Each year, many parents are taken by surprise when their child is born with a life-threatening inherited genetic condition. These parents are healthy individuals who carry a faulty version of a critical gene that predisposes their child to a serious genetic condition.

The medical community has long recognised that early detection can allow carrier couples to conceive healthy children, but there has never been a single comprehensive test for carrier status - until now.

The Counsyl Foresight Carrier Screen, as requested by your doctor, can help provide you with genetic information about dozens of serious genetic conditions many of which you may have heard of.

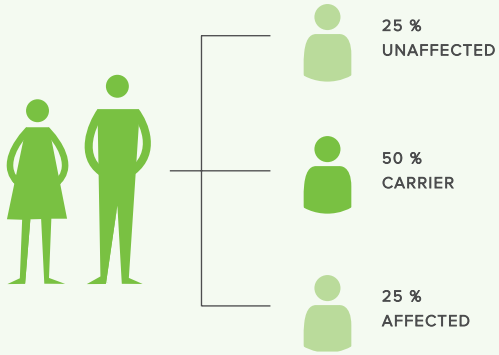
- **Cystic Fibrosis** - affecting the lungs and pancreas, requiring lifelong treatment or lung transplantation.
- **Fragile X Syndrome** - the leading inherited cause of intellectual disabilities and autism.
- **Tay-Sachs Disease** - a metabolic disorder that often causes death within the baby's first few years.
- **Sickle Cell Anemia** - affecting the blood's ability to carry oxygen to all parts of the body.
- **21-OH Congenital Adrenal Hyperplasia** - a serious, prevalent inherited condition affecting hormone levels.

WHO SHOULD BE SCREENED?

Numerous medical societies recommend that pregnant women, or anyone planning a pregnancy, should be offered carrier screening. The test provides a simple screening panel which ensures a comprehensive analysis for all individuals independent of ethnic background. It also allows couples to screen both partners simultaneously. This is a good option if getting results as quickly as possible is your priority.

I DON'T HAVE A FAMILY HISTORY OF DISEASE.

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 life-long health issues. In fact, 4 out of 5 children born with a preventable genetic condition have no family history of that disease.



Autosomal Recessive Inheritance

WHAT IS A CARRIER?

A carrier is someone who has one gene with a mutation and one gene that is unaffected. Carriers are typically symptom-free and do not know that they carry a mutation.

WHAT IS CARRIER SCREENING?

Think of it as an ultrasound for your DNA. Carrier screening is a way to see whether you carry certain gene mutations that do not affect you, but can affect your children. If both you and your partner are carriers of the same genetic disorder, your child has a significant chance of suffering from a serious genetic disease. The only way to know your carrier status is testing.

WHAT IS A RECESSIVE DISEASE?

Recessive diseases are caused by changes (called mutations) in both copies of a person's genes. Every person has two copies of each gene, one inherited from each parent. A recessive disease occurs when both copies of the same gene have a mutation. Cystic fibrosis is an example of a recessive disease. Some of the diseases on the Counsyl Foresight Carrier Screen have a different inheritance pattern - for example, when only the female needs to be a carrier to have a baby at risk. This is called X-linked inheritance. Fragile X Syndrome is a significant example of this.

WHAT IF I FIND OUT I AM A CARRIER?

It is important for you to know that you have options. When two parents are carriers of the same genetic disease, their children have a 1 in 4 (or 25%) chance of having that disease. For certain diseases, such as Fragile X Syndrome, only the mother needs to be a carrier for the child to have a high risk. Your medical professional is available to guide you through the various options to find out what is best for you. Knowing your carrier status before or early in your pregnancy gives you time to learn about the disorder and prepare.

WHAT IF I AM NOT A CARRIER?

Generally, no follow-up testing is suggested for the diseases screened. It is important to understand that no screen is able to identify every carrier of every disease. You should also know that while the test covers a lot of information, we cannot screen for all possible birth defects and genetic diseases. Speak to your healthcare provider if you have special concerns due to family history or other factors.

EARLY TESTING ALLOWS PREVENTION

The earlier you know your carrier status, the more options you have available. Many couples who are at high risk as both parents carry a mutation choose IVF with pre-implantation genetic diagnosis (PGD).

COMPLEMENTARY TO PRENATAL TESTING

It is advised to take the Counsyl Foresight Carrier Screen before a pregnancy. The test differs from tests such as Nuchal Translucency Screening, Chorionic Villus Sampling and Amniocentesis that are performed during the pregnancy. Couples who are carriers may have further testing in pregnancy (prenatal diagnosis). Prenatal testing is a specialised type of testing that is most commonly used to diagnose chromosome problems such as Down Syndrome. Preconceptional and prenatal tests have complementary roles in ensuring a healthy pregnancy.