## Foresight® Carrier Screen

# Genetic screening for inherited conditions



Myriad genetics

Why is this screen important?

# Carrier screening can help you plan and prepare

The Foresight® Carrier Screen can help your healthcare provider determine if you could pass serious inherited health conditions on to your child. A small sample of your saliva or blood is all that's needed. Results are ready in two to three weeks on average.



#### Inherited conditions are common

Individually, inherited conditions are rare, but collectively the conditions included in the Foresight Carrier Screen affect up to 1 in 300 pregnancies. That's higher than the incidence of Down syndrome.  $^{1,2}$ 



#### Family history doesn't tell the whole story

Knowing your family history can tell you a lot about your health and the health of your baby. But many of us are carriers of inherited conditions and simply don't know it. In fact, 88% of individuals who are carriers for cystic fibrosis, spinal muscular atrophy, and fragile X syndrome have no known family history.<sup>3</sup>



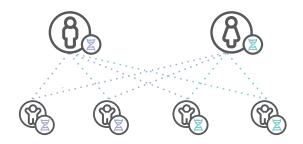
#### Inherited conditions don't discriminate

Anyone in any ethnic or racial group can have a baby with an inherited condition. Medical societies now recognize the advantages of offering screening for the same set of diseases to all patients, regardless of ancestry.

How it works

# Your genes are your family's blueprint

Babies inherit half their genes from each parent. Those genes pass along family characteristics like hair and eye color. Sometimes they also pass on inherited conditions.



#### It's normal to be a carrier for an inherited condition

Being a carrier means that you inherited a normal gene from one parent and a gene with an irregularity, also called a mutation, from the other. As long as you have one normal copy of a gene, you typically don't have any symptoms.

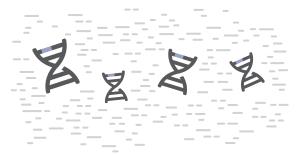
#### It's important to know if you're both carriers

If a mutation in a gene is found in the screening test for one of the parents, it will be important to have the reproductive partner screened as well. If both parents have a mutation in the same gene, there's a 1 in 4 (25%) chance for every pregnancy that your baby will inherit the mutation from both parents and develop symptoms of the associated condition.

There are also a few conditions where only the patient whose egg will be or has been fertilized needs to carry a mutation for the baby to be at risk of developing symptoms.

Conditions

## We can look for a variety of serious conditions



The Foresight Carrier Screen assesses your carrier status for serious conditions you may have heard of, as well as several others that fall into the following categories:

- Conditions where early treatment can make a difference (such as cystic fibrosis)
- Conditions that cause intellectual disability (such as fragile X syndrome)
- Conditions that shorten lifespan (such as Bloom syndrome)
- Conditions where there are **limited to no treatment options available** (such as Tay-Sachs disease)

A full list of conditions screened is available at **myriad.com/womens-health/diseases** 

Next steps

# You have your results. What happens next?

If you find out that you carry a mutation in a gene, it's essential that your partner is screened to make sure that your partner does not carry a mutation in the same gene. If you are both carriers, there are important steps you can take.

#### Perform prenatal diagnosis

Chorionic villus sampling (CVS) or amniocentesis can determine if an inherited condition was passed on to your child.

#### Prepare for delivery

Depending on your results, you may choose to seek additional support to help plan and prepare. In some cases, early treatment can make a big difference. You might start by speaking with a specialist or one of our genetic counsellors.

#### Explore other family building options

If you're not currently pregnant, your results can help you determine whether you want to consider an in vitro fertilization (IVF) procedure where embryos are screened for genetic disease before implantation. Other options include adoption or sperm or egg donation.

For more information visit

myriad.com/womens-health/patient-foresight

Australia's Most Experienced Expanded Carrier Screen

### Foresight® Carrier Screen

#### At a glance

- Proven local track record in Australia since 2009.
- 175+ diseases with one simple and cost-effective test.
- Saliva and blood based collection options.
- Order in clinic or by phone kit delivered overnight.
- Latest Next Generation Sequencing technology.
- Industry leading 'At Risk Couple' detection rate of 4.5%.<sup>1</sup>
- Single and Paired reporting with Reproductive Risk.
- Tandem Reflex minimises unnecessary testing and costs.
- Results matched with screened donor egg and sperm.
- Over 1.2 million patients tested world-wide.
- More than twenty peer-reviewed publications.
- Includes local genetic counselling and clinical support.

More about Myriad Genetics

### Why Myriad?

### We are dedicated to helping you make smart choices about your health, your family and your future

The Foresight Carrier Screen uses advanced science and technology to detect mutations associated with serious diseases.

### You will get screening results that you and your healthcare provider can trust

Our screens are designed to be accurate and comprehensive. Whether your results are positive or negative, you can feel confident that you are as informed as possible when making decisions for you and your family.

#### Support when you need it

We want you to have all the support you need from ordering through to reporting. Every Foresight Carrier Screen includes on-demand consultations with our **local** genetic counsellors.

### Your partner in planning

### The Foresight® Carrier Screen can detect 175+ health conditions including

Alpha Thalassemia • Beta Thalassemia • Cystic Fibrosis
21-OH Congenital Adrenal Hyperplasia • Spinal Muscular Atrophy
Bloom Syndrome • Fragile X • Canavan Disease • Familial Dysautonomia
Gaucher Disease • Niemann-Pick Disease • Tay-Sachs Disease

ABCC8-Related Hyperinsulinism • Adrenoleukodystrophy Alpha-Mannosidosis • Argininosuccinic Aciduria • Ataxia-Telangiectasia Ataxia With Isolated Vitamin E Deficiency • Bardet-Biedl Syndrome Biotinidase Deficiency • Carnitine Palmitoyltransferase IA & II Deficiency Carbamoylphosphate Synthetase I Deficiency • Cartilage-Hair Hypoplasia Cohen Syndrome • Congenital Muscular Dystrophy • Cystinosis • 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 II, IIIA, III Gamma & 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

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.

Tvrosinemia • Usher Svndrome • Wilson Disease



Counsyl Australia donates a percentage of sales from the Foresight® Carrier Screen to the Mission Massimo Foundation for local medical research into the prevention, diagnosis and treatment of childhood leukodystrophies.



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