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There's a lot of information out there, but when it comes to you and your future family, it's definitely up to you to take the reins and make the decision to find out even more about it.

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— Laura M.
Counsyl patient

innovative DIAGNOSTICS

Innovative Diagnostics is a medically managed service practice led by a group of doctors and supported by a dedicated team of senior and experienced industry professionals. We are forging a new path and aspire to set industry benchmarks in doctor and patient experiences.

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Family Prep/Carrier Screen Learn About



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DIAGNOSTICS

THE FAMILY PREP SCREEN

WHAT IS THE FAMILY PREP SCREEN?

The Family Prep Screen, as prescribed by your doctor, can detect over 100 health conditions that can be passed unknowingly from parent to child, 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

A full list of diseases Counsyl screens for is available at www.counsyl.com/diseases.

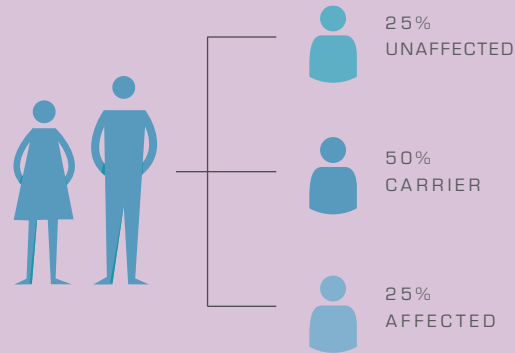
WHO SHOULD BE SCREENED?

Physician societies recommend that pregnant women or anyone planning a pregnancy should be offered carrier screening. Counsyl provides a simple screening panel which ensures the most comprehensive analysis for all individuals independent of ethnic background.

Couples can choose to screen both partners simultaneously. This is a good option if getting all of the answers quickly is your priority.

I DON'T HAVE A FAMILY HISTORY OF DISEASES — SHOULD I STILL BE SCREENED?

Even without a family history of diseases, 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 with recessive genetic diseases are born to couples with no known family history of that disease.



Autosomal Recessive Inheritance

WHAT IS CARRIER SCREENING?

A carrier screen analyzes a person's genes in order to determine if that person is a recessive genetic disease carrier. A screen is able to detect many, but not all, carriers of a disease.

WHAT IS A RECESSIVE DISEASE AND WHAT IS A CARRIER?

Recessive diseases are caused by changes (called mutations) in 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.

A carrier is someone who has only 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.

Some of the diseases on the Family Prep Screen are inherited differently — only the female needs to be a carrier to have a baby at risk. 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 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 Family Prep Screen 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.

HOW CAN I GET SCREENED?

The Counsyl Family Prep Screen is a simple blood test prescribed by your physician. Results become available in approximately 2 weeks.