



HORIZON™ CARRIER SCREENING

Knowing Matters



Rachel and John are planning a family. Rachel has a cousin with Cystic Fibrosis and she discusses carrier screening for this condition with her health care provider. Her carrier screening results indicate that she is a carrier of Cystic Fibrosis. If John is also a carrier, their chance to have a child with Cystic Fibrosis is 1 in 4, or 25%. To better understand their risk, John decides to pursue Horizon carrier screening, and his test results are negative for Cystic Fibrosis.

The couple discusses their results with a genetic counselor. The counselor explains that John's negative screening result for Cystic Fibrosis indicates that there is still a small residual chance that he is a carrier. Therefore, the chance for them to have a child with this condition is low but not zero. Rachel shares her carrier status with her family members in order to give them the option to be screened. Rachel and John successfully conceive and end up having a healthy baby girl.

HorizonTM carrier screening is a simple test that checks to see if you are a carrier of one or more autosomal recessive or X-linked genetic conditions. The majority of people are carriers of at least one genetic condition. Most carriers are healthy; however, they can have a risk to have a child with a genetic condition. Having carrier screening allows you to make more informed reproductive decisions by helping you understand your carrier status. You can have Horizon carrier screening before or during pregnancy.

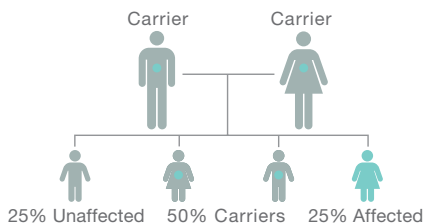
Inheritance of Genetic Conditions

GENETIC CONDITIONS. Autosomal recessive and X-linked conditions are genetic conditions. Genetic conditions are caused by a change, or a mutation, in a particular gene or pair of genes that cause that gene or pair of genes to work improperly or not work at all. When genes do not work correctly, serious health problems in babies and children can result.

INHERITANCE. Autosomal recessive conditions and X-linked conditions are inherited differently.

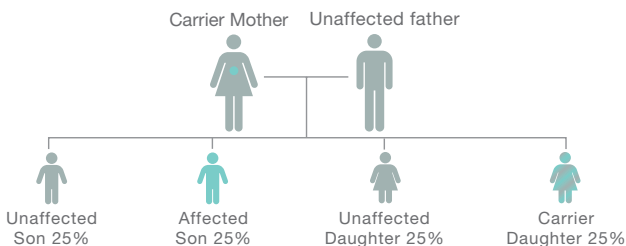
AUTOSOMAL RECESSIVE INHERITANCE

Both the mother and the father have to be carriers of a specific autosomal recessive condition to be at risk to have a child with that condition. If a woman and her partner are both carriers of the same condition, they have a 1 in 4, or 25%, chance in each pregnancy to have a child affected with the condition.



X-LINKED INHERITANCE

Typically, only females can be carriers of X-linked conditions. If a woman is a carrier of an X-linked condition, she has a 1 in 2, or 50%, chance in each pregnancy of passing her gene mutation on to the child. If the child is a girl, she has a 50% chance of being a carrier. If the child is a boy, he has a 50% chance of being affected with the condition.



Is Carrier Screening Right for Me?

WHO SHOULD HAVE HORIZON CARRIER SCREENING?

Anyone can have Horizon carrier screening. The American Congress of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG) recommend carrier screening for certain conditions.

WHEN SHOULD I HAVE HORIZON CARRIER SCREENING?

Horizon can be done before or during pregnancy. Some people may want to have more information about the chance to have a baby with a genetic condition before pregnancy. Others may have screening during a pregnancy.

WHAT ARE THE BENEFITS OF HAVING HORIZON CARRIER SCREENING?

Horizon can help you and your partner learn about the chance to have a child with a genetic condition. Many people do not know they are a carrier of an inherited genetic condition until they have an affected child.

WHAT DOES IT MEAN TO BE A CARRIER?

A carrier of a genetic condition is someone who has a change (or mutation) in one gene copy of a pair of genes. A carrier is usually healthy because the other copy of the gene works normally. Carriers do, however, have a risk of having a child affected with a specific condition.

TO WATCH A BRIEF VIDEO about Horizon, text **“Horizon” to 67076.**



Horizon Logistics and Results

HOW IS CARRIER SCREENING PERFORMED?

A small blood or saliva sample is required. The type of sample you give will depend on what your doctor recommends in his/her practice. Results are returned to your doctor in about 2 weeks.

WHAT DOES A NEGATIVE RESULT MEAN?

A negative result means that no mutations for the conditions screened for were found. While a negative result significantly lowers your chances to be a carrier, carrier screening cannot detect all disease-causing mutations.

WHAT ARE MY REPRODUCTIVE OPTIONS IF I AM A CARRIER?

For autosomal recessive conditions, you and your partner need to be carriers of the same condition to have a risk of having an affected child. If you and your partner are both carriers of the same condition, or you are a carrier of an X-linked condition, the following reproductive options may be considered.

- Natural conception
- Prenatal testing such as amniocentesis or CVS for the specific condition
- In Vitro Fertilization (IVF) with Preimplantation Genetic Diagnosis (PGD)
- Use of sperm or egg donor who is not a carrier for the condition
- Adoption



Conditions Horizon Screens

HORIZON SCREENS FOR UP TO 274 GENETIC CONDITIONS. Your doctor will recommend the right screen for you based on your ethnic background and other factors. It may include a few or all of the conditions available on Horizon. Below are the conditions on the Horizon 27 (Standard Pan-Ethnic Panel).

- Alpha Thalassemia
- Batten Disease (Neuronal Ceroid Lipofuscinosis, CLN3-Related)
- Beta-Hemoglobinopathies
- Bloom Syndrome
- Canavan Disease
- Citrullinemia, Type I
- Cystic Fibrosis
- Duchenne Muscular Dystrophy
- Familial Dysautonomia
- Fanconi Anemia, Group C
- Fragile X Syndrome
- Galactosemia
- Gaucher Disease
- Glycogen Storage Disease, Type IA
- Isovaleric Acidemia
- Medium Chain Acyl-CoA Dehydrogenase Deficiency
- Methylmalonic Aciduria and Homocystinuria, Type cblC
- Mucopolysaccharidosis, Type IV
- Mucopolysaccharidosis, Type I (Hurler Syndrome)
- Niemann-Pick Disease, Types A/B
- Polycystic Kidney Disease, Autosomal Recessive
- Rhizomelic Chondrodysplasia Punctata, Type I
- Smith-Lemli-Opitz Syndrome
- Spinal Muscular Atrophy
- Tay-Sachs Disease
- Tyrosinemia, Type I
- Zellweger Spectrum Disorders, PEX1-Related



LEARN MORE

To learn more about Horizon, visit www.horizonscreen.com. You will find FAQs, condition fact sheets, and information about how the conditions Horizon screens for are inherited.

GENETIC COUNSELING SERVICES

Do you have questions about conditions screened for by Horizon? Would you like to speak to someone about your results? Natera's genetic counselors provide phone information sessions to you at no additional cost. Call (650) 249-9090 to schedule a session.

OTHER PRODUCTS AVAILABLE

Spectrum,[™] Natera's preimplantation genetic screening and diagnosis product, can screen IVF-created embryos before implantation to increase the likelihood of transferring healthy embryos. If you and your partner screen positive for a condition on Horizon, Spectrum may be used to test embryos for that condition.

Panorama,[™] Natera's non-invasive prenatal screen, can be used for women who are at least 9+ weeks pregnant to screen the fetus for common chromosomal abnormalities including Down Syndrome, Edwards Syndrome, Patau Syndrome, Klinefelter syndrome, Triple X syndrome, and Turner Syndrome. Finding out your baby's gender is optional.

Anora,[™] Natera's miscarriage test, helps determine if a miscarriage was caused by a chromosomal abnormality in the fetus. This test can be used for women who have had one or multiple miscarriages and can help reduce the emotional burden of the loss by helping to explain why it occurred.

The tests described have been developed and their performance characteristics determined by the test laboratory. They have not been cleared or approved by the U.S. Food and Drug Administration (FDA).

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201 Industrial Road, Suite 410
San Carlos, CA 94070
1-650-249-9090
Fax 1-650-730-2272
www.natera.com

