Genetic Carrier Screening
Frequently Asked Questions

1. What is genetic carrier screening?

Carrier screening is a genetic test that can inform you about whether you carry a DNA change (variant or mutation) that has the potential to cause a genetic disorder in your child. Typically, if you are a carrier of a DNA change, you are likely to be in good health and not exhibit any symptoms associated with the genetic disorder you carry.

2. Why is genetic carrier screening important?

If an individual carries a DNA change, their children may be susceptible to developing a genetic disorder. The majority of genetic disorders targeted by carrier screening are recessive, although a few are X-linked. For a child to be at risk of inheriting a recessive genetic disorder, both parents must be carriers of the same recessive genetic disorder. In the case of a woman being a carrier of an X-linked genetic disorder, each pregnancy carries a potential risk of up to 1 in 2 (50%) chance of being affected by the X-linked genetic disorder.

3. What if both parents are carriers for a recessive disorder?

If both parents are carriers of the same autosomal recessive condition, there is a 1 in 4 (25%) chance of having an affected child in each pregnancy. Being aware of your carrier status can aid you and your physician in making informed decisions regarding pregnancy planning.

4. Who should have genetic carrier screening?

Anyone can be a carrier of a recessive or X-linked genetic condition, including healthy individuals with no family history of a genetic disorder.

5. What if my carrier screening results are positive?

We understand the importance of screening your partner to assess the risk of recessive genetic disorders in your children. In the event of a positive result, HNL Lab Medicine will conduct carrier screening for the same genetic disorder in your reproductive partner with no additional charges. If you qualify for no-charge carrier screening, your report will indicate your eligibility.

6. What if I have a family history of genetic disorders?

It is important to discuss your health concerns with your doctor, especially if you have a family history of a specific genetic disorder. Your doctor may refer you to a genetic counselor to assess your family history and determine if additional genetic testing is necessary.

7. When should genetic carrier screening be done?

It is best to do carrier screening before getting pregnant, but it can also be done during pregnancy if needed. Finding out your carrier status before pregnancy helps you make informed decisions about the best reproductive options for you, including whether to consider advanced reproductive technologies.

8. What happens once my testing is ordered?

Once you and your doctor decide on which carrier screening tests to pursue, your doctor will place an order for these tests. You can then go to any HNL Lab Medicine Patient Service Center to have your blood drawn. Results for carrier screening are available within 7-14 days and can be accessed on MyHNLAccess.com, the HNL Lab Medicine patient portal.

9. Which genetic disorders should I be screened for?

The American College of Obstetricians and Gynecologists (ACOG) recommends offering carrier screening for cystic fibrosis and spinal muscular atrophy to all individuals who are pregnant or planning to become pregnant. Additionally, carrier screening for Fragile X syndrome should be offered to women with a personal or family history of Fragile X-related disorders, intellectual disability, unexplained ovarian insufficiency, or an elevated FSH level before age 40. ACOG also advises offering screening for specific blood conditions, such as alpha thalassemia, beta thalassemia, and sickle cell anemia, to individuals at increased risk based on their ancestry.

10. What are some of the genetic disorders HNL Lab Medicine's carrier screening can provide information on?

HNL Lab Medicine offers carrier screening for 46 recessive and X-linked genetic disorders. Some of the most common conditions include cystic fibrosis, spinal muscular atrophy, Fragile X, and sickle cell disease.

Commonly Screened Conditions

HNL Lab Medicine

Condition	Description	Carrier Frequency
Cystic Fibrosis	Characterized by the buildup of thick, sticky mucus that can damage many of the body's organs. The most common symptoms include progressive damage to the respiratory system and chronic digestive system problems, but symptom severity can vary widely. With modern treatments and management strategies, many people with cystic fibrosis now live well into adulthood.	Caucasian: 1 in 25 Ashkenazi Jewish: 1 in 24 Hispanic: 1 in 58 African American: 1 in 61 Asian: 1 in 94
Spinal Muscular Atrophy (SMA)	Characterized by weakness and wasting (atrophy) in muscles involved in movement. There are many different types of SMA, which differ in age of onset and severity of muscle weakness. The most common form of SMA is type I, where muscle weakness is evident within the first few months of life, and patients often do not survive past early childhood.	Caucasian: 1 in 35 Ashkenazi Jewish: 1 in 41 Asian: 1 in 53 African American: 1 in 66 Hispanic: 1 in 117
Fragile X	Causes a range of developmental problems, including learning disabilities and cognitive impairment. Usually, males are more severely affected than females. Female carriers of Fragile X may experience premature ovarian failure.	General population: 1 in 259 women
Sickle Cell Disease	Sickle cell disease affects hemoglobin, the molecule in red blood cells that delivers oxygen to cells throughout the body. Characteristic features of this disorder include a low number of red blood cells (anemia), repeated infections, and periodic episodes of pain, though the severity of symptoms varies from person to person.	African American: 1 in 10

Questions?
Contact Us at 877-402-4221

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