

## Genetic Carrier Screening – What is it and Do I Need it?

Carrier screening is testing that's done to see if you or your partner carry a genetic mutation that could cause a serious inherited disorder in your baby. Some of the more common disorders screened for include cystic fibrosis, sickle cell disease, thalassemia, and Tay-Sachs disease. If you have a family history of any of these diseases below, please let us know.

### How is the screening done?

Your practitioner will ask you a lot of questions about your risk factors for genetic disorders at your preconception appointment or first prenatal visit. If you or your partner is found to be at high-risk of being a carrier for a certain disorder, your practitioner should offer to screen you for it.

Risk factors include having a family member with the inherited disorder (or a family member who's a known carrier) or being part of an ethnic group at increased risk for the disease.

If you opt for this kind of screening, you'll probably be asked to give a blood sample first. Then if you're found to be a carrier, your partner will be screened as well. (Both partners may be screened at the same time to get the results faster.)

### Here is more information on some of the carrier screening tests that are available.

- **Cystic fibrosis screening:** Cystic fibrosis (CF) is a life-threatening genetic disease. People with CF are prone to breathing difficulties (including lung infections and severe lung damage), digestive problems, and other complications. Some providers offer CF carrier screening to all patients. Others offer it only to those at highest risk, but anyone may request it. People at high-risk include Caucasians and people who have a relative who has CF or who is known to be a carrier.
- **Sickle cell screening:** Sickle cell disease is a debilitating red-blood-cell disorder. Depending on your family history and ethnic heritage, you may be offered a blood test to check for the genetic mutations that cause this condition. High-risk groups include people of African, Caribbean, South or Central American, Mediterranean, Indian, or Arabian descent. According to the National Institutes of Health, 1 in 12 African Americans carries the gene for this disorder.
- **Thalassemia screening:** Thalassemia encompasses a varied group of inherited blood disorders, including some that are relatively mild and others that may cause severe anemia and other serious problems. More than 2 million people in the United States carry the genetic trait for it. High-risk groups include people of Southeast Asian, Chinese, East Indian, African, Middle Eastern, Italian, Greek, and Turkish ancestry as well as anyone with a family history of the disease or a family member who is a known carrier.
- **Tay-Sachs screening:** Tay-Sachs is a fatal disease of the central nervous system. About 1 in 250 people in the United States carry the genetic trait for it. Among Central or Eastern European (Ashkenazi) Jews, French Canadians, and Cajuns, the carrier rate is about 1 in 27. Among Irish Americans it's 1 in 50. You should be offered screening if you belong to any of these groups or have a family history of the disease. (Ashkenazi Jews are also at risk for carrying the genes that cause two other severe nervous system disorders, familial dysautonomia and Canavan disease, and can be screened for those and a few others as well.)

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- **Fragile X:** Fragile X syndrome is the most common inherited form of mental retardation. The syndrome occurs in approximately 1 in 3,600 males and 1 in 4,000–6,000 females. Approximately 1 in 250 females carry the premutation. DNA-based molecular analysis is the preferred method of diagnosis for fragile X syndrome and its premutations. Prenatal testing for fragile X syndrome should be offered to known carriers of the fragile X premutation or full mutation. Women with a family history of fragile X-related disorders, unexplained mental retardation or developmental delay, autism, or premature ovarian insufficiency are candidates for genetic counseling and fragile X premutation carrier screening.
- **SMA:** Spinal muscular atrophy (SMA) is an autosomal recessive neurodegenerative disease that results from degeneration of spinal cord motor neurons leading to atrophy of skeletal muscle and overall weakness. In current practice, patients with a family history of SMA are being offered carrier screening for the survival motor neuron gene (SMN1) deletion mutations. Recent marketing and public awareness campaigns by laboratories and advocacy organizations are promoting widespread population-based carrier screening for SMA in the prenatal or preconception setting, regardless of family history. However, the American College of Obstetricians and Gynecologists' Committee on Genetics agrees that preconception and prenatal screening for SMA is not recommended in the general population at this time.
- **Canavan Disease:** Canavan disease is an inherited disorder that causes progressive damage to nerve cells in the brain. Infants with Canavan disease typically appear normal for the first few months of life. By age 3 to 5 months, affected infants begin having problems with development, including a delay in motor skills such as turning over, controlling head movement, and sitting without support. These infants typically also have weak muscle tone (hypotonia), unusually large head size (macrocephaly), abnormal posture, and intellectual disability. Feeding and swallowing difficulties, seizures, and sleep disturbances may also develop. While this condition occurs in people of all ethnic backgrounds, it is most common in people of Ashkenazi (eastern and central European) Jewish heritage. Studies suggest that this disorder affects 1 in 6,400 to 13,500 people in the Ashkenazi Jewish population. The incidence in other populations is unknown.
- **Expanded Carrier Screening of the Parents:** This screens potential parents for a number of genetic mutations that could affect their children, including those genetic diseases recommended for carrier testing by the American Congress of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG).
  - These panels have been expanded to include more advanced detection of Cystic fibrosis, Fragile X, Duchenne Muscular Dystrophy and Spinal Muscular Atrophy (SMA) carrier status.
  - With high accuracy, this panel offers multi-ethnic carrier screening. This helps doctors refine their understanding of the risk for each patient of passing on genetic diseases to the next generation.
  - Horizon 27 Panel tests for: Alpha Thalassemia, Batten Disease (Neuronal Ceroid Lipofuscinosis CLN3-Related), Beta-Hemoglobinopathies, Bloom Syndrome, Canavan Disease, Citrullinemia Type 1, Cystic Fibrosis, Duchenne Muscular Dystrophy, Familial Dysautonomia, Fanconi Anemia Group C, Fragile X Syndrome, Galactosemia, Gaucher

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Disease, Glycogen Storage Disease Type Ia, Isovaleric Acidemia, Medium Chain Acyl-CoA Dehydrogenase Deficiency, Methylmalonic Aciduria and Homocystinuria Type cbIC, Mucopolipidosis Type IV, Mucopolysaccharidosis Type 1 (Hurler Syndrome), Niemann-Pick Disease Types A/B, Polycystic Kidney Disease Autosomal Recessive, Rhizomelic Chondrodysplasia Punctata Type 1, Smith-Lemli-Opitz Syndrome, Spinal Muscular Atrophy, Tay-Sachs Disease (DNA only); Tyrosinemia Type 1, Zellweger Spectrum Disorders PEX1-Related.