Genetic Carrier Screening

The majority of babies are born healthy; however, there is a 3–5% risk in every pregnancy for health problems. The disorders listed below are serious conditions that can occur—sometimes without a family history of the disorder. Carrier screening can be performed to determine if you are at risk of having a child affected with one of these disorders.

Carrier screening consists of a blood test (for one or both parents) to determine if the individual carries a gene that increases the risk for having a child with a specific disorder. A carrier is a person who has the disorder trait but is not affected by the disorder. Individuals do not know that they are carriers until they have a specific blood test, or sometimes, an affected child.

When carrier testing is done *prior to conception,* it provides a wider range of reproductive choices if a risk is identified. These choices may include the use of donor egg or sperm, in-vitro fertilization with pre-implantation genetic diagnosis, or adoption. If a risk is identified *during a pregnancy*, testing can be done through amniocentesis or chorionic villi sampling (CVS) to determine if the baby is affected. Regardless of what you may choose, you can use the information to prepare for the birth of a child with a genetic disorder. A genetic counselor can explain more about the specific disorder and all the choices available to you.

All testing is optional. You can choose to be tested for all, some, or none of the following disorders.

Disease	**Cystic Fibrosis (CF)** *The most common fatal genetic disease in North America.*	**Spinal Muscular Atrophy (SMA)** *The most common inherited cause of infant death.*	**Fragile X syndrome (FXS)** *The most common inherited cause of intellectual disability.*
Symptoms	CF is a chronic disorder that causes the body to produce abnormally thick mucus, leading to life-threatening lung infections, digestion problems, diarrhea, poor growth, and infertility. Symptoms range from mild to severe. The average lifespan is 37 years. CF does not affect intelligence.	SMA is a severe, often fatal disorder in which muscles involved in many essential functions, such as breathing, eating, crawling and walking, stop functioning. The most common form of SMA affects infants and causes death by two years of age. Less common forms begin later, with many individuals surviving to adulthood. SMA does not affect intelligence.	FXS is a disorder that causes intellectual disability, autism, and hyperactivity. FXS affects boys and girls; however, boys typically have more severe symptoms.
Inheritance	*Autosomal recessive* When both parents are carriers, there is a 1 in 4 (25%) chance the child will be affected.	*Autosomal recessive* When both parents are carriers, there is a 1 in 4 (25%) chance the child will be affected.	*X-linked dominant* If the mother is a carrier, there is *up to* a 1 in 2 (50%) chance the child will be affected.
Carrier Frequency	1 in 25 Caucasians 1 in 46 Hispanics 1 in 65 African Americans 1 in 92 Asians 1 in 24 Ashkenazi Jews	1 in 35 Caucasians 1 in 117 Hispanics 1 in 66 African Americans 1 in 53 Asians 1 in 41 Ashkenazi Jews	1 in 260 females (all ethnic groups)

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