Carrier Screening in Pregnancy for Common Genetic Diseases

Although most people have healthy babies, with every pregnancy there is a 3-4% chance to have a baby born with problems. The following are a few common, serious disorders that can occur even without a family history. You can have carrier screening (a simple blood test) before the baby is born to determine if you *carry* the genes that cause the disorders shown below.

What is a carrier?
A carrier is a person who has a gene that increases the risk to have children with a specific genetic disease. People do not know if they are carriers until they have a blood test or an affected child. Some disorders occur only if both parents are carriers and other disorders occur only when the mother is a carrier.

What is carrier screening?
Carrier screening involves a blood test from one or both parents to determine if they carry a specific gene that increases the risk for that disorder. If you turn out to be a carrier, prenatal testing such as amniocentesis or chorionic villus sampling (CVS) is available to determine if your unborn baby is affected. All testing is optional and you can choose which disorder(s) for which you want to be tested.

Disease	Cystic Fibrosis (CF)	Fragile X Syndrome	Spinal Muscular Atrophy (SMA)
Symptoms of Disease	*Most common inherited disease in North America.* A chronic disorder that primarily involves the respiratory, digestive and reproductive systems. Symptoms include pneumonia, diarrhea, poor growth and infertility. Some people are only mildly affected, but individuals with severe disease may die in childhood. With treatments today, people with CF can live into their 20's and 30's. CF does not affect intelligence.	*The most common inherited cause of mental retardation.* Fragile X syndrome is a disorder that causes mental retardation, autism, and hyperactivity. It affects both boys and girls, although boys are usually more severely affected than girls. Women who are carriers are at risk to have a child with mental retardation.	*Most common inherited cause of infant death.* SMA destroys nerve cells that affect voluntary movement. Infants with SMA have problems breathing, swallowing, controlling their head or neck, and crawling or walking. The most common form of SMA affects infants in the first months of life and can cause death between 2 and 4 years of age. Less commonly the disease starts later and people can survive into adulthood. SMA does not affect intelligence. There is no cure or treatment.
Inheritance	If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with cystic fibrosis.	If a mother is a carrier, there is up to a 50% chance to have a child fragile X syndrome.	If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with SMA.
General Population Carrier Frequency	1 in 25 Caucasians 1 in 26 Ashkenazi Jewish 1 in 46 Hispanics 1 in 65 African Americans ~1 in 90 Asian	1 in 260 females in North America Occurs in all ethnic backgrounds	1 in 35 Caucasians 1 in 41 Ashkenazi Jewish 1 in 117 Hispanics 1 in 66 African Americans 1 in 53 Asian
Have you ever had testing for this condition? **(please circle one)**	YES NO Not Sure	YES NO Not Sure	YES NO Not Sure
Do you want this testing or more information?	YES NO	YES NO	YES NO

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