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Genetic Disease Carrier Screening

Below is information about OPTIONAL testing for carrier status of certain genetic conditions. Carrier screening will be offered to you in your pregnancy — either at your new OB visit or at a routine genetic counseling appointment. Please consider this information and let your provider know if you would like to proceed with this type of testing.

Most babies are born healthy. A small number are born with a rare genetic disease. This can happen when the biological mother and father are genetic disease carriers. Most of the time the mother and father do not have health problems, and therefore they do not know they are carriers. However, when both parents are carriers, there is a 25% chance the baby will have the condition, and have significant health problems. In these cases, testing is available for pregnancies to determine if a baby actually has the condition. There are screening tests available that can identify couples at risk for having a child with some of these genetically inherited conditions.

Genetic carrier testing is a blood test to check if someone is a genetic disease carrier or unlikely to be a carrier (some rare cases can be missed). Once someone is found to be a carrier, they will meet with a genetic counselor to discuss the option of carrier testing for their partner and genetic testing of a pregnancy.

Carrier screening can check your carrier status for a few basic diseases (smaller panel of 2-13) or a large number of diseases (larger panel up to 175+). No screening has a 100% detection rate, so some carriers will not be detected. Carrier screening can reduce, but not eliminate, the chance to have a child with a genetic disease. A person's ethnicity is sometimes used to guide their decision about which testing they would like to have done. Please see the table below.

Ethnicity	Disease (descriptions on back side)	Carrier frequency in high risk population	Pregnancy risk
All ethnicities	Spinal Muscular Atrophy	1 in 50	1 in 10,000
Caucasian	Cystic Fibrosis	1 in 28	1 in 3,136
Asian	Beta-Thalassemia Alpha-Thalassemia Cystic Fibrosis	1 in 44 1 in 30 1 in 94	1 in 7,744 1 in 3,600 1 in 35,344
Middle Eastern, Indian, Mediterranean, African, Hispanic	Beta-Thalassemia Sickle Cell Cystic Fibrosis	1 in 8 to 25 1 in 11 to 24 1 in 60 to 70	1 in 256 to 2,500 1 in 484 to 2,304 1 in 14,400 to 19,600
French Canadian, Cajun	Tay-Sachs Cystic Fibrosis	1 in 30 1 in 28	1 in 3,600 1 in 3,136
Ashkenazi Jewish	Tay-Sachs Familial Dysautonomia Cystic Fibrosis Canavan disease Bloom syndrome Fanconi Anemia C Gaucher disease Mucolipidosis IV Niemann Pick A	1 in 28 1 in 31 1 in 25 1 in 55 1 in 110 1 in 90 1 in 16 1 in 100 1 in 100	1 in 3,136 1 in 3,844 1 in 2,500 1 in 12,100 1 in 48,400 1 in 32,400 1 in 1,024 1 in 40,000 1 in 40,000

Questions to consider in choosing a smaller vs larger carrier screening panel:

- What is your ethnicity? (e.g., German, Asian Indian, Black, Ashkenazi Jewish)
- Do you have a family history of any of the above conditions?
- Do you have a family history of autism or intellectual disability?
- Would you prefer to test for diseases more common to your ethnicity? Or would you prefer to look for a larger number of diseases?

CONDITION DESCRIPTIONS

Spinal Muscular Atrophy (SMA) is a disorder that affects the control of muscle movement. There is a wide range of severity, but all types cause weakness and wasting of the muscles. Mild cases might require a person to use walking aids in their 30s, while severe cases can cause death in early childhood because the muscles used for breathing and swallowing are affected.

Cystic Fibrosis (CF) is a disorder that causes the buildup of thick mucus that can damage the lungs and digestive system, as well as other organs. Other health problems involving nutrient absorption, pancreatic issues, and male infertility can occur. Classic CF is a life altering and life shortening disease with average life expectancy into the 40s. The features and severity of CF vary among affected individuals, with very mild symptoms possible.

Beta-Thalassemia is a blood disorder that lowers a person's ability to carry oxygen, causing a lack of oxygen in the body. There are varying levels of severity, but more severe cases affect children within the first two years of life. They develop life-threatening anemia, do not grow at the expected rate and may have liver problems. Some have enlarged organs, including the heart, and misshapen bones. Affected individuals may need life-long blood transfusions.

Sickle cell disease is a blood disorder that changes the shape of a person's blood cells. This altered shape can cause episodes of severe pain, anemia, and infections. While symptoms vary between individuals, sickle cell anemia can cause growth and developmental delays, fatigue and shortness of breath in children. Symptoms may require hospitalization.

Tay-Sachs disease progressively destroys nerve cells in the brain and spine. Children with Tay-Sachs begin to show muscle weakness and delayed development by 3 to 6 months of age. Eventually, infants develop seizures, vision and hearing loss, intellectual disability, and become paralyzed. These children usually pass away within 3 to 5 years of life.

Familial Dysautonomia (FD) affects the body's ability to control involuntary actions such as digestion, breathing, blood pressure and body temperature. People with FD may have muscle weakness, poor growth and feeding difficulties in childhood. Older children may have episodes of vomiting, poor balance, and both kidney and heart problems. About one in three people with FD have learning disabilities. By adulthood, recurrent infection can cause lung damage and worsening vision as time goes on.

Canavan disease affects nerve cells in the brain. Children with Canavan seem healthy at birth, but start developing symptoms by 5 months of age. Symptoms included delayed motor skills, muscle weakness, an unusually large head and intellectual disability. Seizures and difficulty eating are common. Life expectancy varies but most children do not live past about age ten.

Bloom syndrome causes a greatly increased risk for cancer. People with Bloom syndrome are short in stature and have sun-sensitive skin. They also have a distinct set of facial features, learning disabilities, and an increased risk of infection. Males experience infertility, and females may have reduced fertility and early menopause. Life expectancy is decreased due to cancer risk.

Fanconi Anemia (FA) affects many parts of the body. People with FA have bone marrow failure resulting in severe anemia and other blood problems including cancer (leukemia). They may also have underdeveloped hands or forearms, heart defects, and many other organ problems. Hearing loss and infertility may occur. Life expectancy is reduced.

Gaucher disease is caused by a buildup of material that the body cannot process. Patients may experience an enlarged liver and spleen, anemia, easy bruising, lung disease, and bone issues such as pain, fractures, and arthritis. Very mild forms of this disease are possible and treatment is available.

Mucolipidosis IV causes developmental delays and vision loss that worsens over time. People with Mucolipidosis IV may have difficulty sitting, standing and controlling their hand movements, have limited speech, difficulty chewing and swallowing, and may become blind over time. Patients may have a shortened life span.

Niemann Pick type A affects the body's ability to breakdown and use fats. Harmful amounts of fats build up in the spleen, liver, lungs, bone marrow and brain. Infants with Niemann Pick A show signs shortly after birth and do not develop or gain weight as expected. They also have severe intellectual disability and typically do not survive past the first few years of life.

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