San Diego Perinatal Genetic Screening Consent

Carrier Screening

Up to 1 in 3 people are carriers of a genetic disease and most carriers do not have a family history of this disease. So when a baby is born with a genetic disease, it is usually a surprise. The ability to test parents to see if they are carriers for genetic diseases is rapidly changing. As your doctors we want to provide you with up to date recommendations.

The technology exists to test to see if you are a carrier for over 200 diseases. Some of these diseases can impact your baby's life expectancy, quality of life or intellectual ability. Early intervention with some conditions can actually improve life expectancy or reduce symptoms. Carrier testing does not need to be repeated in subsequent pregnancies. Carrier testing ideally should be done before pregnancy or as early in the pregnancy as possible in order for patients to have the most options available.

Certain ethnic groups are at higher risk for certain conditions. For example, African Americans are at higher risk for sickle cell anemia, Ashkenazi Jews are at higher risk for Tay-Sachs and Asians are at a higher risk for alpha thalassemia. However, many people do not accurately know their family heritage and therefore we support a pan-ethnic approach to carrier screening (this means offering everything to everyone). Additional testing and counseling would be recommended if any test is positive.

Currently, we are recommending/offering carrier screening for the most common genetic diseases - Cystic Fibrosis, Spinal Muscular Atrophy, Fragile X Syndrome and Hemoglobinopathies.

Cystic Fibrosis: Cystic fibrosis is a genetic disorder that affects the lungs, pancreas, kidneys and intestines. Symptoms can include excessive mucus production and frequent lung infections, bowel blockages, fatigue and poor weight gain. There is no cure but early detection and intervention can improve the outcome for these children.

Spinal Muscular Atrophy: Spinal muscular atrophy is a genetic neuromuscular disorder that results in progressive muscle wasting and early death. This blood test will determine if you are a gene carrier. Your partner also needs to be a carrier in order for your baby to be at risk. Early treatment of this condition can greatly improve the outcome for your baby and in some cases prevent nerve degeneration.

Fragile X: Fragile X is the most common inherited cause of mental retardation. This blood test will determine if you are a gene carrier. Your partner also needs to be a carrier in order for your baby to be at risk. There is no current cure for this but early therapy can help with learning disabilities.

Hemoglobin Electrophoresis: this blood test screens for genetic conditions that cause anemia, such as thalassemia and sickle cell. This is a standard prenatal test which will be included in your initial OB bloodwork.

If your carrier test is negative, your doctor will review your results with you at your next prenatal visit.

If your carrier test is positive, you will be called by a genetic counselor who will review the results with you and help you make arrangements to have your partner tested. If your partner is not a carrier, then your baby is at very low risk for having the condition and no further testing is recommended. If your partner is also a carrier, we recommend that you have formal genetic counseling to review your risk and all of your additional testing options.

If you have any questions, please let us know so we may address your concerns during this visit.

Patient Consent

I have read this information and have discussed carrier screening with my provider. I have had all of my questions answered. After being informed of my options:

☐ I decline to have any carrier testing done.

☐ I choose to have carrier testing for Fragile X, Spinal Muscular Atrophy, Cystic Fibrosis and a Hemoglobin Electrophoresis.

☐ I choose to have expanded carrier testing done. I understand that this increases the likelihood that something will come back positive and my partner will need to be tested as well.

Patient Signature: _____ Date: _____

☐ I consent to have a genetic counselor call me with a positive test result. I understand that they will offer testing for my partner. I can choose formal in person genetic counseling if I desire further discussion or counseling.

Patient Signature: _____ Date: _____