GENETIC CARRIER SCREENING

Genetic Carrier Screening is done by a simple blood draw and will determine if you carry the genes for certain genetic disorders. Some genetic disorders are more common among people of specific racial and ethnic heritage. These tests will help to identify if you are at risk of having a child affected by the most commonly screened inherited genetic disorders. This information allows you to make informed reproductive choices.

The following genetic carrier screening panel may be offered to you today

- **Cystic Fibrosis (CF)** - A blood test can determine if you are a carrier of CF. Both parents must carry the gene for this disease in order for the baby have a (25%) chance of being born with it. People affected by CF are now starting to live longer into their 30's, but it is still considered a serious disease. It is a life-threatening disorder that causes severe, progressive lung damage and nutritional deficiencies.
- **Spinal Muscular Atrophy (SMA) –** Both parents must be carriers in order for the baby to have a (25%) chance of being born with it. Carrier status can be detected by a blood test. Spinal Muscular Atrophy is a neuromuscular disorder that causes severe muscle weakness and progressive loss of voluntary muscle control. It is a life shortening disorder.
- **Fragile X –** Fragile X is the most common cause of inherited intellectual disability. A blood test can determine if you are carrier of Fragile X. Fragile X syndrome in its early stages can include delayed speech & developmental milestones which progress into moderate intellectual disability. Fragile X is caused by mutations in the fragile X gene located on the X chromosome and typically passed on from carrier mothers and more commonly affect boys. There are usually no life threatening health concerns associated with fragile X syndrome and treatment involves early intervention & management of symptoms.
- **Tay Sachs Disease** – Both parents must be carriers in order for the baby to have a (25%) chance of being born with it. Tay Sachs is caused by an enzyme deficiency that allows a harmful substance to build up in the brain, causing mental and physical deterioration, and it is generally fatal by age 5. This is more commonly found among people of Ashekenazic (Eastern European) Jewish, French Canadian or Louisiana Cajun descent. People of Ashekenazic descent are also at increased risk for a number of other, "related" genetic disorders.

Concord OBGYN uses Invitae Laboratories for personal genetic carrier screening. Carrier screening can be drawn at your visit today! The nurse will ask you to provide your email address. Invitae will do a benefits investigation with your specific insurance plan on your behalf and send an email to you with information about your coverage. Some plans cover genetic testing fully and other plans may be subject to deductibles and coinsurances. **Please be on the lookout for an email from Invitae within a few days of having your bloodwork drawn to receive your benefits investigation and choose your preference of insurance vs patient pay or to cancel your testing if you so choose.** Prior authorizations are obtained by Invitae on your behalf (if your insurance requires it) before processing your specimen.