Facts about………

**Carrier Screening Options**

**WHAT IS CARRIER SCREENING?**

Carrier screening determines if a couple is at an increased risk of having a baby with a specific inherited disease. Genetic disorders are caused by changes within genes called mutations. Every individual has two copies of each gene, one from each parent. A carrier is a person who has one normal copy of a gene and one abnormal copy. Carriers have no symptoms and usually no family history of the specific condition. If both parents are identified as carriers of the same abnormal gene for the disease, there is a 25% chance with each pregnancy to have an affected child with that disease.

Carrier screening requires a small sample of blood and the results may take several weeks. Testing is highly accurate and a normal test result leads to a significantly reduced chance of being a carrier however, it cannot completely exclude possible carrier status. If both parents are found to be carriers of the same disease, prenatal diagnosis can be performed to determine whether or not the fetus is affected.

**WHAT ARE THE DISEASES?**

**Ashkenazi Jewish Genetic Panel (AJGP)**

Currently, there are 11 conditions that are screened for individuals of Ashkenazi Jewish descent. The diseases are: Tay-Sachs Disease, Canavan Disease, Cystic Fibrosis, Familial Dysautonomia, Gaucher Disease, Niemann-Pick Disease, Bloom syndrome, Mucolipidosis Type IV, Fanconi Anemia, Glycogen Storage Disease Type 1A and Maple Syrup Urine Disease.

**Cystic Fibrosis (CF)** *most frequent in Caucasians*

A disease in which thick mucus accumulates in the respiratory and digestive tracts leading to a variety of respiratory and digestive illnesses. Intelligence is normal. Severely affected individuals may die in childhood, but some people are only mildly affected. Average lifespan is around 30 years. Approximately 1 in every 25 people of Caucasian descent is a carrier of CF.

**Sickle Cell Disease (SCD)** - *most frequent in individuals of African descent*

A disease that affects red blood cells and can cause painful episodes, anemia, organ damage, increased risk for infections, lung problems, leg ulcers and strokes. Approximately 1 in every 10 people of African descent is a carrier of SCD.

**Spinal Muscular Atrophy (SMA)**

SMA is a severe neurological disease that destroys the nerves responsible for muscle control of breathing, swallowing, head and neck control, walking and crawling. There is no treatment and death usually occurs in early childhood. Approximately 1 in 40 to 1 in 60 people is a carrier for SMA. (Recent data suggests carrier risk for individuals of Hispanic descent may be significantly lower. No published reports are available at this time.) Screening is currently offered to those with a family history of SMA. [Reference ACOG Committee Opinion May 2009 #432]

**Thalassemia**- *most frequently in individuals of Italian, Greek, Middle Eastern, Southeast Asian, and African ancestry*

Thalassemias are a group of disorders that cause a deficiency of red blood cells leading to anemia. There are two main types called alpha and beta. The most severe form of alpha thalassemia results in fetal or infant death. The most severe form of beta thalassemia can cause organ enlargement, poor growth, and changes in the bones. Heart failure and infections are the leading causes of death among children with the most severe form of beta thalassemia. The carrier frequency of thalassemias differs between the ethnic groups listed above.

**DECIDING ON CARRIER SCREENING**

After learning about carrier screening some people decide to have testing and others decide against it. The cost of screening varies and may or may not be covered by your insurance company/policy. It is important to ask your health insurance company about its policies as some plans may not cover carrier screening or may require that you obtain a referral prior to testing.