



Preconceptual Counseling

To maximize the chances of delivering a healthy baby, Women's Fertility Center offers and recommends certain screening tests to assess your risk for passing on various genetic diseases, before you begin trying to conceive.

Based on your family history and ethnicity, we begin by identifying which diseases you are most at risk for being a carrier of, and recommend screening the female partner first for those diseases. If genetic screening identifies that she is a carrier of a particular disease, we then test the male partner. If both partners are carriers of a genetic disease, then there is a significant risk of passing on that disease, and we advise that you consult with a genetic counselor prior to pregnancy.

Genetic testing during pregnancy may also be necessary, if the carrier state of both partners is not determined until after conception has occurred. Chromosomal testing during pregnancy includes chorionic villus sampling (CVS), usually done between 10 and 12 weeks of gestation; or amniocentesis which is performed between 16 and 18 weeks of pregnancy.

We offer screening for the following genetic disorders:

Cystic Fibrosis (CF)

Cystic fibrosis is a disease that causes the body to produce abnormally thick mucus that leads to life-threatening lung infections. Obstruction in the pancreas and intestine may also occur, causing poor growth and development in children, and leading to a shortened life span. The median age of survival for someone with cystic fibrosis is 33 years. Screening for CF is recommended for all women who are attempting to conceive. For Caucasians, approximately 1 in 25 people carry the gene for CF (the "carrier rate.") However, Hispanic and African American women are also at risk with a carrier rate of 1 in 46 and 1 in 65, respectively, and, therefore, should be screened.

Sickle Cell Disease

Sickle cell disease is a blood disorder characterized by anemia and episodic pain. The anemia is due to abnormal hemoglobin production which results in red blood cells becoming stiff and sickle shaped. This disease is most common in people of African descent with a carrier rate of one in 12. People from South America, Central America,

India, Saudi Arabia, Italy, Greece and Turkey are also at risk for being a carrier of sickle cell disease. The carrier state is easily diagnosed by performing a hemoglobin electrophoresis.

Fragile X Syndrome

This syndrome is the most commonly inherited form of mental retardation. It is a result of a mutation on the fragile X gene (FMR1). Anyone with a family history of Fragile X Syndrome or a family history of undiagnosed mental retardation should be screened for this disease.

Alpha Thalassemia

This is another blood disorder in which there is an abnormal production of hemoglobin causing anemia. Symptoms vary depending on the number of abnormal globin genes (between one and four). It is frequently inherited in people from southeastern Asia and China with a carrier rate of 1 in 30. Screening can be accomplished by checking a complete blood count and a hemoglobin electrophoresis. Then, the carrier state is confirmed by molecular genetic testing.

Beta Thalassemia

Beta thalassemia is an inherited blood disorder in which there is a decrease in hemoglobin A production. Symptoms vary depending on whether a child inherits the mild, intermediate or severe form of this disease, although anemia is present in all cases. It is a commonly inherited disorder in people of Mediterranean descent (Greeks, Italians) with a carrier rate of 1 in 10. In addition, people from Africa, the Middle East and Asia are also at an increased risk of being carriers. Screening can be accomplished by checking a complete blood count and a hemoglobin electrophoresis. The carrier state is then confirmed by molecular genetic testing for thalassemia.

Tay Sach's Disease

Tay Sach's disease is a progressive neurodegenerative disorder characterized by severe mental retardation and seizures and is fatal. It is caused by a deficiency in the enzyme hexosaminidase A. Tay Sach's disease occurs most commonly in Ashkenazi Jews with a carrier rate of 1 in 25. Non-Jewish people of French Canadian background are also at an increased risk for Tay Sach's disease when compared to the risk for the general population (1 in 250).

Canavan's Disease

Canavan's disease is another progressive neurologic disease due to a deficiency in the enzyme aspartoacyltransferase. Children with Canavan's disease die by age 10. This disease occurs most commonly in Ashkenazi Jews who have a carrier rate of 1 in 40.

Familial Dysautonomia

This disease is due to a deficiency in the enzyme dopamine beta hydroxylase and is characterized by an abnormally functioning central nervous system. The median age of survival is 30. In people of Eastern European Jewish descent, 1 in 30 are carriers.

Fanconi Anemia

Fanconi anemia is a disorder in which there is a decrease in the production of all types of blood cells and abnormal bone development. People with this disease may have skeletal abnormalities, mental retardation and are more susceptible to cancer. Death usually occurs by age 20. One in 87 people of Ashkenazi Jewish ancestry are carriers of this disease.

Bloom Syndrome

Bloom syndrome is due to an increased number of chromosomal breaks. This disease causes growth retardation, an increased susceptibility to infections and in some, mental retardation. There is a predisposition to cancer and death occurs early (in the teens or early twenties). The carrier rate is 1 in 100 in people of Eastern European Jewish descent.

Gaucher's Disease

This disorder is due to a deficiency in the enzyme galactosidase. Anemia, osteoporosis and enlargement of the liver and spleen may occur, although symptoms are highly variable. There is also an increased risk of cancer. Death may occur anytime from age six to eighty. This is another disease that is most common in Ashkenazi Jews with a carrier rate of 1 in 10.

Niemann-Pick Disease

Niemann-Pick disease is due to a deficiency in the enzyme sphingomyelinase which leads to an accumulation of sphingomyelin. Children with type A disease usually die by age 3. Those with Type B have a less extreme form of the disease and may survive to adulthood. One in 75 Ashkenazi Jews are carriers of Type A Niemann-Pick disease.