Genetic Screening Consent Form

The American College of OBGYN (ACOG) recommends "information about genetic carrier screening should be provided to every pregnant woman."

Testing is available to help you determine your risks of having a baby with certain genetic conditions. This can occur even in the absence of any family history of these disorders, as disorders can her inherited in a variety of patterns.

For dominant disorders, the condition is caused by inheritance of a *single* abnormal gene. These disorders do not skip generations and are more likely to have been detected in a prior affected family member, as there is a 50% chance of having an affected child.

Recessive disorders, require inheritance of *two* abnormal copies of a gene, one from the mother and one from the father. These genes can pass through many generations without detection since there is a 25% chance of having an affected child *if both* parents are carriers of the mutation.

X-linked disorders are inherited through the X chromosome. Women have two copies of the X chromosome and as such, women are often asymptomatic, and are primarily carriers of a mutation. Men have only one copy of the X chromosome, so males are more severely affected and display more symptoms.

The following are 3 conditions that can be screened for through genetic testing available in our office from a blood sample. Please continue reading and consider whether you desire any, none, or all of these tests. Then please indicate your selections and sign below.

Cystic Fibrosis (CF)

Cystic fibrosis is the most common autosomal recessive condition in the non-Hispanic white population. It is a disease that primarily affects the lungs, pancreas and intestinal system but does not affect intelligence. Males with CF often are infertile. The current average survival is 42 years. Carrier frequencies in non-Hispanic white and Ashkenazi Jewish populations are about 1 in 25. So the chances of having a child with CF in those groups are about 1 in 2,500 births. In other ethnic groups, the carrier frequency is less, so the chance of having a child with CF for Hispanic couples is about 1 in 8,500 and 1 in 15,000 for African American couples. All newborns are tested for CF at birth.

Spinal Muscular Atrophy (SMA)

Spinal muscular atrophy is a rare autosomal recessive disease characterized by death of spinal cord cells that leads to death of skeletal muscle and overall weakness. The genetics of SMA are very complex, and affected individuals can have symptoms that vary significantly, and range

from mild muscle weakness to death in infancy. The incidence of SMA is approximately 1 in 6,000-10,000 live births. Carrier frequencies in most populations are estimated at 1 in 40-60 individuals.

Fragile X Syndrome (FXS)

Fragile X syndrome is an X-linked disorder. Though it is uncommon, it is the most common *inherited* form of intellectual disability. The syndrome occurs in approximately 1 in 3,600 males and 1 in 4,000–6,000 females. Intellectual impairment ranges from learning disabilities to severe cognitive and behavioral disabilities. Males are more significantly affected than females. Carrier screening is recommended for women with a family history of fragile X-related disorders, intellectual disability suggestive of FXS or premature menopause before age 40.

Ethnicity Considerations

Certain ethnic populations are at increased risk of additional genetic disorders. Please let us know if you have Ashkenazi Jewish, French Canadian. Mediterranean or Cajun ancestry, as additional genetic counseling and further screening options may be considered.

Based on the information above, I would like the following testing: (select all that apply)

_____ No genetic screening

_____ Cystic fibrosis (CF) screening

_____ Spinal muscular atrophy (SMA) screening

_____ Fragile X syndrome (FXS) screening

Cost of testing is dependent on your insurance carrier and deductible. (Please initial) _____

My signature below indicates I understand the above and have had all questions answered to my satisfaction.

_____ _____

Patient Signature Date

5/25/2017