Alaska Women's Health PC

Everyone's Welcome and Everyone's Heard

GENETIC SCREENING & CARRIER TESTING INFORMATION

Genetic Screening and Carrier Testing is an important part of family planning, as it helps define your risk of having a child with a genetic disease.

Genetic screening allows you to know if you are at an increased risk of some genetic abnormalities such as Down syndrome or Trisomy 18.

Screening for Neural Tube Defects will identify if you are at an increased risk for open neural tube defects such as Spina Bifida.

Carrier testing will identity if you are a "carrier" of a recessive genetic disease such as Cystic Fibrosis. Typically, carriers are healthy individuals, but when two parents are carriers of the same genetic disease, they have a 1 in 4 (25%) chance of having an affected child.

Genetic Screening:

-Combined Screening:
This test combines the results of a nuchal translucency ultrasound and a simple blood test to look for Down Syndrome and Trisomy 18. Testing is performed between 11w0d and 13w6d of pregnancy and may include an additional blood draw between 15w0d and 22w.

-Noninvasive Prenatal Testing (NIPT) or Cell Free DNA
This is a blood test that uses cell free fetal DNA to screen for increased risk of Down Syndrome, Trisomy 13, Trisomy 18, Turners Syndrome, and sex chromosome abnormalities. Testing can be performed after 11w0d of pregnancy and anytime thereafter. Generally, this testing is performed on women who are at an increased risk of aneuploidies.

-MSAFP/Quad Screening
This is a blood test that measures particular substances in your blood. This test can detect an increased risk of neural tube defects, Down Syndrome, and Trisomy 18. Testing is performed as early as 15w0d of pregnancy. It may be recommended in conjunction with the testing listed above.

Carrier Testing:

-The most common carrier testing is for Cystic Fibrosis, Spinal Muscular Atrophy, Duchenne Muscular Dystrophy, and Fragile X.

-Carrier screening is available at any time during pregnancy. You may be screened prior to having a child.

-ACOG, the American College of Obstetricians and Gynecologists, recommends that all women of reproductive age be screened for Cystic Fibrosis as it is one of the most common life-threatening recessive conditions

-If you are a carrier of a particular recessive disorder, testing your partner will help determine your baby's risk of a genetic disorder

-If both you and your partner are carriers for the same disorder, additional testing can be done to determine if your baby will have a genetic disorder

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