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Prenatal Genetic Screening

In 2007, ACOG (American College of OB/GYN) recommended that all women seeking prenatal care be offered screening for chromosomal abnormalities regardless of age. Women are born with all the eggs they will have in their lifetimes and as women age, their eggs age with them. Since older eggs are more likely to be chromosomally abnormal, an older mother is more likely to have a fetus with a chromosomal abnormality compared to a younger mother. However, fetal chromosomal abnormalities may occur in women of any age. Down's Syndrome (Trisomy 21) is the most common chromosomal abnormality found during pregnancy and is characterized by mental retardation, short stature and other physical abnormalities. There are many other chromosomal abnormalities that can now be screened for and diagnosed during pregnancy. This document will review the current testing options for you. It is important to remember that these tests are optional and many patients decline prenatal genetic screening.

Screening Testing:

Screening tests do not give a positive (abnormal) or negative (normal) result but they will place patients in either high or low risk categories, which mean that some patients in low risk groups may still have a chromosomally abnormal fetus and most patients in high-risk groups will still have a chromosomally normal fetus. These tests are non-invasive and there is no risk to the fetus.

1. First trimester Screen (Nuchal Translucency)
 - a. Performed between 12-14 weeks gestation
 - b. Maternal blood draw testing three fetal blood proteins combined with an ultrasound measurement of the fetal neck.
 - c. Screens for three chromosomal abnormalities (90% sensitive for Down's Syndrome) but does not screen for potential spinal defects.
2. Second trimester Screen (Quadruple Screen)
 - a. Performed between 16-20 weeks gestation
 - b. Maternal blood draw testing four fetal proteins
 - c. Screens for three chromosomal abnormalities (80% sensitive for Down's Syndrome) as well as for potential spinal defects (including spina bifida)

Diagnostic Testing:

Diagnostic tests give positive (abnormal) or negative (normal) results. These tests are invasive and unfortunately may have complications including miscarriage.

1. Amniocentesis
 - a. Performed at 16 weeks gestation
 - b. Involves removing amniotic fluid via a needle placed through the abdomen and uterus
 - c. Miscarriage risk 1/300 (0.3%)
2. Chorionic Villous Sampling (CVS)
 - a. Performed at 12 weeks gestation
 - b. Involves removing chorionic villi (placenta) via a needle placed through the abdomen and uterus
3. Miscarriage risk 1/100 (1%)

I request to have the following prenatal testing performed. I acknowledge that it is my responsibility to determine if my insurance carrier will cover the cost of testing (especially if I am under 35 years old) and that I will be financially responsible for any costs not covered by my insurance.

- First Trimester Screen
- Second Trimester Screen
- Amniocentesis
- Chorionic Villous Sampling
- I decline all prenatal chromosomal testing
- I am currently undecided about prenatal chromosomal testing

Patient Name: _____

Signature: _____ Date: _____

