PRENATAL TESTING

Prenatal testing can be divided into two basic groups: screening tests and diagnostic tests. Screening tests include quadruple screen, **noninvasive prenatal screening or testing (NIPS or NIPT)**, and ultrasound. These tests do not make a definitive diagnosis of trisomy 13, trisomy 18, or related disorders.

Screening tests:

The quadruple screen is an older test, requires a blood sample from the mother, and looks at specific hormones and proteins.

The NIPS or NIPT (cell-free DNA) test is a newer screening test that also requires a blood sample from the mother. This test looks for fetal DNA fragments that have crossed the placenta and are in the mother's bloodstream. The fetal DNA in the mother's serum is measured. This test is typically offered at 10 weeks or later. It is important to ask the doctor or genetic counselor how likely it is that a positive result predicts a syndrome **(positive predictive value PPV)**.

Diagnostic tests:

Diagnostic tests include **chorionic villus sampling (CVS)** at 10-12 weeks or **amniocentesis** at 15-16 weeks. Both are considered invasive tests and have less than 1% of pregnancy loss over normal risk for pregnancy loss.

CVS is performed in the first trimester and involves taking a small sample of the developing placenta for the study. Due to this, CVS carries a slightly higher risk than amniocentesis. The benefit of CVS is that the procedure is performed earlier in pregnancy, and thus the result is back sooner (12-13 weeks of pregnancy compared to 16-17 weeks for an amniocentesis). Knowing early is important for families who would consider termination, as abortion laws are changing rapidly and are becoming increasingly more restrictive depending on the state.

Amniocentesis can be performed any time after 15 weeks and involves obtaining a sample of the fluid surrounding the developing fetus. Both the CVS and amniocentesis use ultrasound to guide the needle during sampling. These tests are most often performed by maternal- fetal medicine (MFM) specialists, or "high-risk OB's." While there are rare exceptions, CVS and amniocentesis are generally considered very accurate. Risks of both procedures include miscarriage, premature rupture of membranes, infection, or bleeding from the placenta; however, these are typically very rare.

Other Tests During Pregnancy:

If a genetic difference is suspected, the pregnancy is typically considered a high-risk pregnancy and will be followed by both a general obstetrician-gynecologist (Ob-Gyn) and a maternal-fetal medicine (MFM) specialist. They can perform the ultrasound mentioned above looking for changes in the organs (often called a "level 2 ultrasound"). Heart defects are commonly associated with trisomy, and they may either perform or recommend a special heart ultrasound of the fetus called a fetal **echocardiogram.**

There are a number of other complications that can arise during pregnancy. **Fetuses** (babies inside the womb) with trisomy are at an increased risk for miscarriage and stillbirth. The risk for loss during pregnancy decreases as the pregnancy progresses.

Some mothers may require additional testing in the third trimester of pregnancy. These tests are typically performed along with fetal growth ultrasounds done every 3 to 4 weeks in the third trimester. They may include: fetal non-stress testing (NST), measurement of the fluid around the baby (amniotic fluid indexes, or AFI), and/or a biophysical profile (BPP). Each office has their own protocol for performing these tests.

Glossary

AMNIOCENTESIS — a prenatal diagnostic technique where a needle is placed into the uterus and a small amount of amniotic fluid is withdrawn usually for performing testing.

CHORIONIC VILLUS SAMPLING (CVS) — a prenatal diagnostic technique where a small amount of placental tissue (chorionic villus) is taken for performing testing.

ECHOCARDIOGRAM — an ultrasound study of the heart, if prenatal a fetal echocardiogram.

NONINVASIVE PRENATAL SCREENING (NIPS) — maternal blood sample obtained at 10-11 weeks gestation to study cell- free DNA derived from the fetus

POSITIVE PREDICTIVE VALUE (PPV) — the chance that a positive result of a screening test will predict a condition, for example, trisomy 18 or trisomy 13.

ULTRASOUND — a procedure using sound waves to look at tissues and organs; also called a sonogram.