CAPITAL WOMEN'S CARE®
"Working Together for Women's Health"

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GENETIC TESTING IN PREGNANCY

There are many options and tests available during your pregnancy for genetic screening. All pregnant women, regardless of their age, have the option to pursue all available testing.

The following is a brief overview of the currently available tests along with their respective risks and benefits.

A. Screening Tests for Baby

These tests only screen for genetic abnormalities and/or Neural Tube Defects (NTDs). These tests are not diagnostic tests - they will only give you a risk assessment for the disorders described (i.e. increased risk/decrease risk compared to the general population)

1. Maternal Blood Fetal Plasma Cell Free DNA testing (cFDNA) / NIPT (Non Invasive Prenatal Test)

This is a simple non-invasive test that involves a blood draw from the mothers arm. It tests the baby's DNA directly out of the mother's blood. This test is done at or after 10 weeks of pregnancy. It looks for the risk of Down Syndrome (Trisomy 21), Trisomy 18 and Trisomy 13. It can also test the sex chromosomes on the baby. It does NOT test all of the chromosomes. The predictive value (accuracy) can be as high as 99%. If the result is normal, no further testing is generally indicated. If the result shows a high risk of a problem with any of the tested chromosomes, a diagnostic test such as amniocentesis or CVS (see below) is recommended.

Benefits: Non-invasive
Highly accurate

Drawbacks: Must be at or above 10 weeks to do the test

2. First Trimester Screen / NT (Nuchal Translucency) Sonogram

This is a non-invasive test done between 11-13 weeks and 6 days of pregnancy that is done to check the risk of Down Syndrome, Trisomy 18 and Trisomy 13. It involves a sonogram to look for the nuchal fold (back of the neck of the baby) thickness and some blood from the mother. It has an accuracy rate of approximately 80-85%.

Benefits: No miscarriage risk as the test is non-invasive.
Can be performed at any maternal age.
Improved accuracy over the quadruple screen
Allows for a limited evaluation of major anatomical abnormalities earlier than the 20 week sonogram.

Drawbacks: Must be done between 11-13 weeks 6 days
80-85% accurate in its predictive value.
Only tests for the three disorders mentioned above.
Separate "AFP ONLY" test needs to be drawn at 15-20 weeks for neural tube defect risk (i.e. Spina Bifida)

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3. The Quad Screen

This test is a simple blood test that is done between 15 - 21 weeks of pregnancy. It tests for the risk of Down Syndrome, Trisomy 18, Trisomy 13 and Open Neural Tube (Spinal) Defects such as spina bifida. It is approximately 70% accurate.

Benefits:	No miscarriage risk as the test is non-invasive. Good negative predictive value. Can be done between 15 - 21 weeks.
Drawbacks:	Only 70% accurate in its predictive value. Only covers the disorders mentioned above. Not as accurate as the 1st trimester screen or NIPT / cfDNA.

B. Diagnostic Genetic Testing for Baby

The risk of having a child with a chromosomal abnormality (i.e.; Down Syndrome) rises significantly after age 35. There are two currently available FDA approved tests for chromosomal testing, which are almost 100% accurate and evaluate all 46 chromosomes.

1. CVS Testing - a biopsy of the placenta done between 10-12 weeks.

Benefits:	Early diagnosis
Drawbacks:	Approximately 1/100 risk of miscarriage. Only tests for chromosomes (not neural tube defects like Spina Bifida)

2. Amniocentesis - a needle is placed into the amniotic sac and a small amount of fluid is withdrawn and sent for evaluation.

Benefits:	Tests for both chromosomal abnormalities and neural tube defects. Lower risk of miscarriage at approximately 1/600
Drawbacks:	Done a few weeks later in pregnancy at 15-17 weeks,

C. Carrier Screening Genetic Testing for Mother

Testing that is specifically related to family history, patient history, and other genetic testing. These tests may vary by patient and will be recommended based on your risk after consulting with one of the doctors.

Some examples:	Cystic Fibrosis	Jewish Heritage Screening
	Spinal Muscular Atrophy	Sickle Cell Carrier Screening (Hemoglobin electrophoresis)