University Park
OBSTETRICS AND GYNECOLOGY, LLC

Genetic Testing Options
Patient Consent Form

The American College of Obstetrics and Gynecology (ACOG) and the Society for Maternal and Fetal Medicine (SMFM) recommend ALL women be offered options for genetic testing. Genetic counseling and testing is recommended for ALL women but especially those who will be **35 years old or greater** at the time of delivery.

There are many chromosome abnormalities that can affect your baby but there are a few select ones that are able to be screened for with special tests that are available today. The most common is Trisomy 21 or Downs Syndrome which is a disorder that leads to birth defects and mental retardation. Trisomy 18 and 13 are two other chromosomal defects that can lead to severe morbidity and mortality and are always fatal.

Babies can also be affected by open neural tube defects. The neural tube is that which ultimately develops into the spinal cord and the brain. This normally closes as the baby develops in the embryonic stage of development but failure to do so results in an abnormal development of the brain or open spinal defects known as spina bifida. The severity depends on the size of the defect as well as the location, sometimes being a fatal anomaly.

In general these risks increase with family history, personal history, and age of the mother especially women 35 years and older. The majority of these cases however are in women under 35 years old as there is a significantly more number of women in this age group seeking pregnancy. Classically women 35 years old or more at the time of delivery were offered amniocentesis but today there are many other safer less invasive options although that remains the gold standard for the diagnosis.

Non-invasive screening tests using blood samples and ultrasounds are now available to women of all ages. These tests are completed in the 1st and 2nd trimesters and provide information of your risk for carrying a baby with any of the above genetic defects including Trisomy 21, 18, 13 and neural tube defects.

Given that these are screening tests they do have limitations including false positives and negatives. They are designed to quantify risk in terms of low risk and high risk. Low risk often leads to conservative management and no further testing whereas high risk results in recommendations for further diagnostic testing including chorionic villous sampling (CVS) which is a needle biopsy of the placental tissue or amniocentesis where a needle is placed into the amniotic sac and fluid removed for analysis.

An additional important point to remember is that not all genetic disorders or fetal anomalies can be detected by tests available today. These include but are not limited to autism, non-specific mental retardation, some genetic disorders, and some types of genetic birth defects.

Screening Options:

Integrated Screening (mostly for women <35 years of age)

- Between 10 and 13 6/7 weeks an initial blood test is drawn looking at proteins and pregnancy hormones
- An ultrasound between 11 and 13 6/7 weeks looking at the fetal anatomy and nuchal translucency (neck thickness)
- A second blood test between drawn 16 and 18 weeks
- These results are then all combined to give you an assessment of risk associated with Trisomy 21, 18, 13, as well as open neural tube defects
- Increased risk does NOT mean there is a problem with your baby, but it does mean that more testing is recommended and referral to a specialist would be initiated. You will be referred to high risk Obstetricians known as Maternal Fetal Medicine doctors. They will assist us in making sure we have a definitive diagnosis for your baby
- Likewise, if the screening comes back low risk this does NOT guarantee that your baby is completely normal as there are limitations and false negatives to the tests as described above

Non-invasive Prenatal Testing (NIPT) (mostly for women >/= 35 years of age)

- Chorionic Villous Sampling (CVS) and Amniocentesis are an immediate invasive option rarely done as an initial workup test
- A blood test done at 10 weeks for various proteins and pregnancy hormones collecting cell-free fetal DNA from a maternal blood sample
    - Trisomy 21, 18, 13
    - Some aneuploidies like 45X (Turners), 47 XXY (Kleinfelters), 47 XXX, 47 XYY etc.
- This test again is for screening purposes and does not replace diagnostic tests like CVS and amniocentesis, but it does decrease the need for these more invasive tests in most women
- The 2nd trimester blood test is still recommended for screening for neural tube defects as this test does not detect that
- An NT screen is also still recommended as this screens for other fetal anomalies and birth defects

Diagnostic Options like CVS and Amniocentesis will be further discussed by Maternal Fetal Medicine specialists if that service is needed.

The following table explains some of these tests, their limitations as well as risks for fetal loss in the event they are necessary to be performed:

	Integrated Screen (<35yo)	CVS	Amnio	NIPT (>/= 35yo)
When Performed	Part 1 10-13.6w Part 2 16-18w	10-12 weeks	16-21 weeks	Anytime after 10 weeks
T21 Detection Rate	92%	98%	99%	99%
T18 Detection Rate	90%	99%	99%	98%
T13 Detection Rate	Unknown	99%	99%	80%
Open Neural Tube Detection Rate	80%	Unable	95%	Unable
Risk to Baby	0%	1:100 loss	1:200 loss	0%
False Positive Rate	5%	Near 0%	Near 0%	<1.0%

Genetic counseling is always available if desired. The timing of these tests is very important so if they are desired you will have to decide early on and let the MD or CNM know your desires. They are usually covered tests but often are applied to deductibles meaning you may be responsible for some or all of the cost, please contact your insurance company for more information, we will also attempt to do this for you but there is no guarantee that the information given to us by your insurer is accurate.