Understanding Optional Prenatal Testing

There are several genetic tests available to all pregnant women.
It's a personal decision as to whether or not you would choose any of these.

Maternal Carrier Screening

This genetic test determines if the mother carries certain genes that cause genetic disorders. These are typically recessive genes. The tests include screening for disorders such as cystic fibrosis, hemoglobinopathies, and spinal muscular atrophy. If the woman is positive, the father of the baby can be screened to assess his genes. This test does not need to be repeated in subsequent pregnancies because it evaluates the mother's genes not the fetus'.

Non Invasive Prenatal Testing (NIPT) - performed after 10 weeks

Blood test that determines the risk of the 3 most common fetal trisomies (abnormal chromosomes). It screens for Trisomy 21, Trisomy 18 and Trisomy 13. There is an option to determine the sex of the baby. It's 99% accurate.

Ultrascreen

A combination of a maternal blood test (10-13 weeks) plus an ultrasound (11 to 14 weeks). It determines the same 3 trisomies as the NIPT, but not the sex of the baby. It may also detect several other rare chromosomal abnormalities. There is a 91% detection rate.

Alpha Fetoprotein(AFP) - performed between 15 and 21 weeks

Maternal blood test to detect neural tube defects such as spina bifida or anencephaly. It should be done in addition to genetic screening.

Quad Screen - performed between 15 and 21 weeks

Maternal blood test that includes the AFP test as well as screening for trisomy 21. There is a higher false positive rate.

*If any of the above screening tests are positive, a diagnostic test may be indicated. These tests, such as chorionic villus sampling (CVS) and amniocentesis are not performed at MomDoc. They are performed at a high-risk OB office.

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