Prenatal Testing Algorithm

Although the majority of pregnancies result in deliveries of healthy babies, every woman has a background risk for having a baby with a chromosome abnormality, birth defect or other genetic condition. During your pregnancy, there are multiple testing options that can provide you with additional information about the chance that your baby has one of these conditions. Your healthcare provider can help you determine which testing for chromosome abnormalities, like Down syndrome, might be most appropriate for you.

Some women choose to have no specific testing for chromosome abnormalities like Down syndrome

Screening tests don't tell for certain about chromosome abnormalities like Down syndrome, but do provide a more "individual" risk assessment.

If you want to know for sure about chromosome abnormalities, you can opt for diagnostic testing

Least Information → **Most Information**

	No testing	Traditional Serum Screen	Non-Invasive Prenatal Testing (NIPT)	CVS/AMNIO
BENEFITS	• Less anxiety for women who may worry about receiving an increased risk test result • No difficult decisions to make in case of abnormal results	• Non-invasive blood draw from mother • May be performed in first and/or second trimester with or without an ultrasound • If performed in 2nd trimester, screens for certain birth defects like spina bifida	• Non-invasive blood draw from mother • Screens for common chromosome abnormalities as early as 9 weeks • Optional additional testing for other genetic conditions • High detection rate • Few screen positive test results. Most women are reassured by low risk results	• Definitive results • Would be offered if screening test is high risk. • Ability to plan for baby's care in case of abnormal results
RISKS/LIMITATIONS	• Inability to plan medically, financially & emotionally • Missed opportunity to engage with specialists & community support resources prior to delivery	• Not diagnostic; does not tell "for sure" • Compared to CVS/amnio, does not screen for all chromosome abnormalities • Has a higher false positive rate and misses more high risk pregnancies, when compared to NIPT	• Not diagnostic; does not tell "for sure" • Compared to CVS/amnio, does not screen for all chromosome abnormalities • May not be able to report results in a small number of patients	• Small risk of miscarriage • Amnio results not available until 2nd trimester • Possible results of uncertain significance

☐ **I do not want screening or diagnostic testing**

☐ **I want maternal serum screening as described by my healthcare provider**

☐ **I want NIPT**
- ☐ **With 22q only**
- ☐ **With full microdeletion panel**

☐ **I want diagnostic testing, either CVS or amniocentesis**

☐ **I need more information before making a decision**

Patient Name _____ **Physician Name** _____

Patient Signature _____ **Date** _____

1. NIPT is a screening test and will only cover certain chromosome abnormalities. **Diagnostic testing (CVS, amnio, or blood work after delivery) is necessary to confirm these results and/or detect other problems.**
2. A low-risk screening test result tells me that the risk the baby will be born with one of these conditions is extremely small; it does not, however, rule the risk out completely.
3. A high-risk screening test result still requires confirmation with CVS, amino, or testing the baby after delivery.
4. Regardless of low-risk results on screening or negative diagnostic tests, there is still a 3-4% baseline risk of birth defects/genetic disorders in any pregnancy.
5. Women with pregnancies considered high risk due to ultrasound abnormalities, family history, advanced maternal and/or paternal age should discuss their risks with their doctor/genetic counselor to determine which tests are appropriate for their situation.

* ACOG and SMFM Joint Committee Opinion 2015: Any patient may choose cell-free DNA analysis as a screening strategy for common aneuploidies regardless of her risk status. The patient choosing this testing should understand the limitations and benefits of this screening paradigm in the context of alternative screening and diagnostic options.

NAT-PrenatalTestingAlgorithm_A_2018_04_09_NAT-801817