Non-Invasive Prenatal Screening

Non-invasive prenatal screening

The St. Cloud Perinatal Clinic offers non-invasive prenatal screening (NIPS) to all pregnant women. This is a screen that uses maternal blood to help us identifiy women who may be at an increased risk for having a child with Down syndrome, Trisomy 18, Trisomy 13 or other chromosome problems.

What conditions are screened for?

- Down syndrome is a medical problem that results from having too much genetic information. Individuals with this medical problem have an extra chromosome 21. Children with this medical problem have characteristic facial features, learning problems within the range of intellectual disability, and an increased risk for birth defects.

- Trisomy 18 and Trisomy 13 also result from having too much genetic information but these medical problems are more severe than Down syndrome. Many babies or children with these medical problems die during the pregnancy or within the first year of life due to birth defects and respiratory problems. Those that survive have serious learning problems and are usually unable to walk or talk.

- Sex chromosome abnormalities are a group of genetic conditions caused by the loss of or the presence of an extra sex chromosome (X or Y). The clinical features of these conditions are variable. Common sex chromosome abnormalities include Turner syndrome and Klinefelter syndrome.

- Additional chromosome problems can also be screened for as well as limited microdeletion syndromes. Detection rates for these medical problems are limited. Screening for these conditions is not recommended for the general population.

Risk associated with maternal age

Every woman has a chance of having a child with a chromosome problem like Down syndrome, Trisomy 18 and Trisomy 13. This risk increases with increasing maternal age. For example:

Maternal Age at Delivery	Chance for Having a Child with an Extra Chromosome
20	0.2%
25	0.2%
30	0.3%
35	0.5%
40	1.5%

Who is considered increased risk?

A woman is at an increased risk for having a child with an extra chromosome problem if:

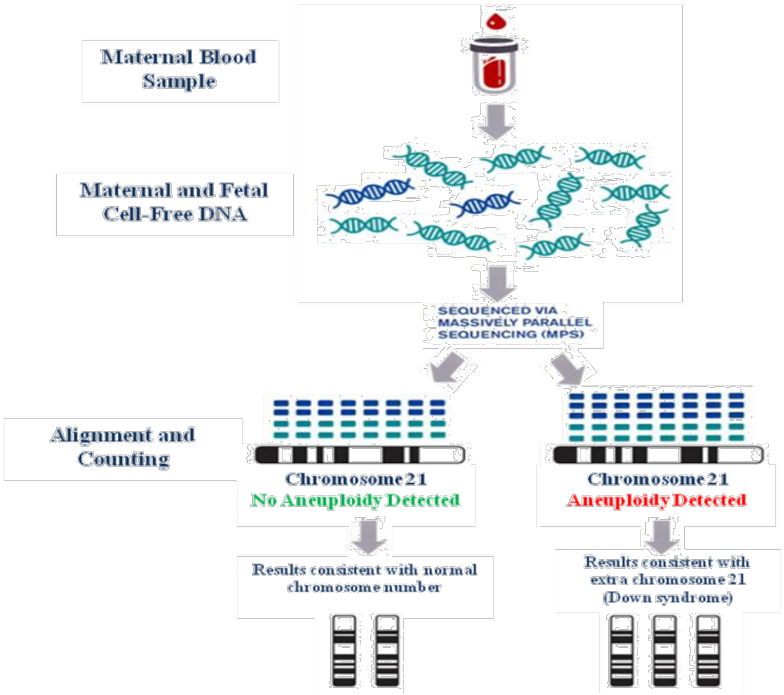
- She will be 35 years or older at delivery
- Ultrasound reveals findings associated with chromosome problems
- She has an increased risk for having a child with a chromosome problem based on first trimester, sequential, integrated, or quadruple screen.
- She has had a previous pregnancy affected with a chromosome problem

How does non-invasive prenatal screening work?

Non-invasive prenatal screening is a test that measures the amount of genetic material in a pregnant woman's blood. This test is performed as early as 10 weeks of pregnancy, through a maternal blood draw.

During pregnancy, maternal blood contains fragments of our genetic material, or DNA, from the mother and fragments of DNA from the baby.

These fragments are free floating in the blood and come from cells that have broken down (as part of the normal process of cells wearing out and dying). These fragments are isolated from the blood and it is determined which chromosome each fragment came from. The amount of material from each chromosome is then examined. If there is too much DNA from chromosome 13, 18, or 21, this suggests that there is an extra chromosome. Since mom is unaffected, this suggests that the baby may have an extra chromosome.



NIPS identifies

- > 99% percent of babies with Down syndrome (Trisomy 21)
- ~ 98% of Trisomy 18
- ~91% of Trisomy 13
- ~95% of Turner syndrome (Monosomy X)

What will the results tell you?

Your results will tell your doctor whether or not extra chromosome 13, 18 or 21 or sex chromosome abnormalities are highly suspected in this pregnancy. A positive result does not mean that your baby has a medical problem. In the case of a positive result, genetic counseling is available to discuss what the results mean to you and your baby, as well as to discuss further testing options for your pregnancy.

Limitations of testing

No test can guarantee a baby will not have any medical issues. Non-invasive prenatal screening only looks for Down syndrome, trisomy 13, trisomy 18 and sex chromosome problems. This screen does not test for all genetic and non-genetic problems that may be present in a baby. If the test result is consistent with two copies of each chromosome indicating a negative result, it does not eliminate the risk for these conditions for the baby. False positives and false negatives can occur. False positives are more likely to occur in low risk populations.

Other options

If you are not interested in non-invasive prenatal screening, there are other screening and diagnostic options available for chromosome problems.

First trimester screening and quad serum screening can tell you the chance of your pregnancy (for example, 1 in 50 or 1 in 5,000) having a certain chromosome problem but they do not provide a definitive answer.

Invasive procedures (such as a CVS or amniocentesis) can provide a more definitive answer, but they have a small risk of complications, including miscarriage.

Non-invasive prenatal screening has higher detection rates and lower false positive rates compared to other screening tests and does not carry the risk of complications that an invasive procedure can have.

Please feel free to ask your health care provider if you have further questions about this screening.

Why have non-invasive prenatal screening?

Some parents do non-invasive prenatal screening because they want more information about the risks for a chromosome problem specific to their current pregnancy. If additional testing confirms that their baby has a chromosome problem, they might find it helpful to use this information to plan and prepare for their child's condition. Others in this situation may choose to not continue a pregnancy.

There is no prenatal treatment or cure when we learn that a child has a chromosome problem during pregnancy but we do follow these pregnancies more closely. This information may also impact plans for delivery. In some cases, early treatment can have great benefits for long term health.

Please feel free to ask your health care provider if you have further questions about this screening.

Insurance coverage

You should check with your insurance company to see if they cover this screening. You may need to provide specific information to your insurance plan regarding the testing being performed. **CPT Codes: 81420.** Your doctor or genetic counselor will help you determine which diagnostic codes apply to you.