Navigating the NIPS Screening Process

Society for
Women's Health Research

Screening Process

NIPS can be conducted as early as 10 weeks into the pregnancy through a blood sample. Most NIPS panels will screen for trisomy 21 (**Down syndrome**), trisomy 18 (**Edwards syndrome**), and trisomy 13 (**Patau syndrome**). You may discuss with your health care provider if screening for sex **chromosome abnormalities**, **microdeletions**, or rare **aneuploidy** analysis are appropriate for your pregnancy.

Screening results usually arrive 1-2 weeks after the blood sample is taken. It can be beneficial to familiarize yourself with next steps after results come back.

To make the most of your consultation appointments, see the **Talking to Your Health Care Provider** section of the SWHR Noninvasive Prenatal Screening Resource Guide for Women.

Interpreting the Screening Results

If you get a negative screening result:
A negative result means that the screening **did not detect an increased risk** for the condition(s) screened. For a condition like Down syndrome, in which NIPS is a very reliable predictor, it is very unlikely (but not impossible) that women who receive a negative result will have a baby with Down syndrome.

If you get a positive screening result:
A positive result indicates that there is an **increased risk or suspicion** that the pregnancy is affected with a chromosomal abnormality. Because **genetic screening** is not definitive, it is important to consider a diagnostic test to confirm if the condition is present in the baby. Typical diagnostic tests include:

- **Chorionic villus sampling** – A procedure that involves taking a sample of tissue from the **placenta** for testing. Depending on the location of the placenta, tissue is taken by a tube is inserted through the vagina or a small needle inserted through the belly.
- **Amniocentesis** – A procedure that involves using a small needle to take a sample of the amniotic fluid from the sac that surrounds the **fetus** for testing.

DIAGNOSTIC TESTS TO CONFIRM A PRENATAL GENETIC CONDITION

	CHORIONIC VILLUS SAMPLING (CVS)	AMNIOCENTESIS
WHEN	Typically between 10 and 13 weeks of pregnancy	Typically between 15 and 20 weeks of pregnancy (ideally), but can be performed any time after 15 weeks
WHY	Can confirm if a fetus has a chromosomal condition	Can confirm a number of indicators of the fetus's health, including chromosomal conditions and neural tube defects
WAIT TIME	Typically 1-2 weeks	Typically 1-2 weeks, depending on the type of analysis being done

Although the chances are low, both CVS and amniocentesis come with a risk for bleeding, miscarriage, or preterm labor, so it is important to discuss the risks and benefits of these procedures with your health care provider or **genetic counselor**. Your health care team will be there to support you if the results from diagnostic testing come back positive. They can help walk you through expectations going forward, share useful resources, and connect you with community and support organizations for you and your family.

When you receive a positive NIPS result, your health care provider may refer you to a genetic counselor. A genetic counselor or maternal-fetal medicine specialist can help you assess your baby's risk of having a certain condition, discuss your options, and talk with you about any concerns you may have.

If you get an indeterminate result:
Sometimes screening results will not come back as positive or negative, meaning the result is indeterminate. This *may or may not* mean a chromosomal disorder is present. Naturally, these results can be confusing, so it is important to discuss your results with your health care provider or a genetic counselor. You will likely need to follow up with additional procedures, such as a fetal ultrasound and diagnostic testing.

Screening Accuracy

With any screening, it is possible to get results that do not represent what is actually happening with your baby.

A **false-negative** occurs when the screen shows a negative result, but the condition is actually present in the fetus.

- False-negative results are uncommon, with rates as low as 1 in 10,000.[8]

A **false-positive** occurs when the screen shows a positive result, but the condition is not actually present in the fetus.

- The likelihood of a false-positive result is affected by several factors, including the conditions screened and if the pregnancy is considered high risk for those conditions.
- Often, the false-positive rate of screenings for trisomy 13, 18, or 21 is <1%.

It is important to remember that NIPS is just one of many tools that can be used to assess your and your baby's health during your pregnancy. Your health care provider will likely share your NIPS results in combination with results from other routine tests and screens, such as a prenatal ultrasound. Ultrasound results can provide additional context to NIPS results and may provide additional information about the overall health of the baby.

Microdeletions are typically associated with rare conditions; thus, screening has an increased chance of returning false-positive results.