

FACTS ABOUT NON-INVASIVE PRENATAL SCREENING (CELL-FREE FETAL DNA SCREENING)

Non-invasive prenatal screening (NIPS) is a blood test done during pregnancy. This screening test measures small bits of fetal DNA to check for certain genetic conditions in the developing baby. It is also called cell-free fetal DNA screening.

HEALTH NOTES



For more information

Visit kp.org

- Read about the NIPS in the Health encyclopedia.

Call Kaiser Permanente Medical Advice

- Talk to your medical advice nurse 24 hours a day, 7 days a week by calling **(703) 359-7878** or **1-800-777-7904**.
TTY: **(885) 632-8278**.

If you would like to speak with a Genetics Counselor, call: **(301) 702-5250**.

DNA is the genetic information we inherit from our parents. DNA is present in most cells of the body and is also found in your blood. When you are pregnant, a small amount of the DNA in your blood comes from the developing baby. Specialized blood testing can identify pregnancies that have an unusual pattern of fetal DNA in the mother's blood. This is a sign that the pregnancy has a much higher chance for certain birth defects.

How is NIPS done?

Non-invasive prenatal testing is done by a blood test taken at 10 weeks or later in pregnancy.

What can NIPS tell me?

Non-invasive prenatal screening is able to tell you whether your pregnancy is at low risk or high risk for Down syndrome (trisomy 21) and two other chromosome problems, called trisomy 18 and trisomy 13. NIPS can find almost all pregnancies with Down syndrome and trisomy 18, and most pregnancies with trisomy 13. NIPS can also detect many sex chromosome abnormalities.

Who can have NIPS?

All pregnant women can have non-invasive prenatal screening, including those pregnant with twins.

What if my NIPS result is low risk?

Most women who have NIPS will have a low risk result. A low risk result is very reassuring that your baby does not have Down syndrome, trisomy 18, or trisomy 13. Rarely, a baby with one of these three conditions will have a low risk result and be missed by this test.

A low risk result does not guarantee that the baby has no health problems or birth defects. Keep in mind that this test **does not** provide information about other chromosome problems, birth defects, or genetic disorders.

What if my NIPS result is high risk?

A high risk result usually means that your baby has Down syndrome, trisomy 18, or trisomy 13. With a high risk result, the chance that the baby actually has one of these three conditions may be as high as 99%. However, healthy babies have occasionally been reported to have a high risk result. If you have a high risk result, you will be offered more testing to verify whether or not the developing baby actually has a chromosome problem.

Important things to consider

Non-invasive prenatal screening:

- has no risk for miscarriage,
- may not identify all pregnancies with Down syndrome, trisomy 18, trisomy 13 or sex chromosome abnormalities,
- does not test for all chromosome problems,
- does not test for other genetic conditions or other birth defects,
- may signal a high risk in a healthy pregnancy (false-positive result),
- requires additional testing to verify a diagnosis of Down syndrome, trisomy 18, or trisomy 13, and
- is unable to give any result in a small percentage of pregnancies.

What follow-up testing will I be offered if my result shows a high risk?

If you have a high risk result you will be offered either CVS or amniocentesis.

- Chorionic villus sampling (CVS) is done between 10 and 13 weeks in pregnancy. A small sample of the developing placenta is taken by passing a thin needle through the lower abdomen or using a flexible plastic tube through the vagina.
- Amniocentesis (amnio) is usually done between 15 and 20 weeks in pregnancy. A small amount of the fluid that surrounds the baby is taken by passing a thin needle through the abdomen.

Both CVS and amniocentesis accurately diagnose chromosome problems by examining fetal cells under a microscope. However, both of these tests have a small risk (<1/300) for miscarriage.

How long does it take to get results from NIPS?

Results from NIPS usually take about one to two weeks. You will receive a phone call or a secure message when your result is ready. In a small number of pregnancies the test is unable to give any result.

Before you decide about NIPS, think carefully about whether or not you want to know this information during your pregnancy. Knowing about a chromosome problem can be useful for some people. You might use the information to decide whether or not to continue the pregnancy. You might find the information helpful to prepare for a child with special needs. But not everyone wants to learn about chromosome problems during the pregnancy.

It's up to you to decide whether or not you want this type of information during pregnancy. Our genetic counselors are available to help if you have questions.

More about...

Down syndrome is a chromosome problem associated with mild to moderate intellectual disability, a characteristic facial appearance, and weak muscle tone in infancy. Babies with Down syndrome also have a higher chance to be born with a physical birth defect, such as a heart defect or intestinal problem.

Trisomy 18 and Trisomy 13 are two different chromosome problems associated with severe intellectual disability and medical problems in many parts of the body. For either condition, survival beyond the first year of life is uncommon.

The information presented here is not intended to diagnose health problems or to take the place of professional medical care. If you have persistent medical problems, or if you have further questions, please consult your doctor or member of your health care team.

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