

Panorama™ Prenatal Screen Common Questions



What is Panorama™?

Panorama is a non-invasive DNA screening test that can tell you important information about your pregnancy, as early as nine weeks of gestation. With Panorama, you can find out if your baby is at risk for chromosomal abnormalities, such as Down syndrome, and all that's required is a sample of your blood. If you so choose, you may also find out the gender of your baby.

How does the Panorama prenatal screen work?

During pregnancy, some of the DNA from the baby crosses into mom's bloodstream. DNA is organized in structures known as chromosomes, which carry the baby's genetic information. Panorama uses a blood sample from the mother to analyze the baby's DNA for certain chromosomal conditions that could affect the baby's health.

Panorama is a non-invasive prenatal test (NIPT). This means that Panorama is safe for you and your baby. To have the test done, your healthcare provider just draws a sample of blood from your arm. The sample is then sent to a lab for processing.

When will I get my results?

Most results will be returned within 7-10 days.

What kind of results will I get from the Panorama screening test?

The report sent to your healthcare provider will have one of these results:

- **LOW-RISK RESULTS:** A low risk result means the chance that your baby has one of the chromosome conditions Panorama screens for is very low.
- **HIGH-RISK RESULTS:** A high risk result means there is an increased risk that your baby has a particular condition. Your healthcare provider will talk to you about follow-up testing options, so you can determine if your baby is affected.

There is also a very small chance that no results will be obtained from your initial sample. In this case, we may recommend sending us another blood sample for re-testing.

Most women who have the Panorama screening test will discover their baby is at low risk for the conditions tested. This means that the chance of the baby having one of these conditions is very low, which can be reassuring. When the test result shows a high risk, it is important to talk with your healthcare provider about your next steps before making any decisions about your pregnancy.

What conditions does Panorama screen for?

Currently, the test screens for:

- **Trisomy 21:** This is caused by an extra copy of chromosome 21 and is also called Down syndrome. This is the most common cause of intellectual disability.

It may also cause certain birth defects of the heart or other organs and may cause hearing or vision problems.

- **Trisomy 18:** This is caused by an extra copy of chromosome 18 and is also called Edwards syndrome. This causes severe intellectual disability. It also causes serious birth defects of the heart, brain and other organs. Babies with Edwards syndrome usually pass away before one year of age.
- **Trisomy 13:** This is caused by an extra copy of chromosome 13 and is also called Patau syndrome. This causes severe intellectual disability. It also causes many serious birth defects. Babies with Patau syndrome usually pass away before one year of age.
- **Monosomy X** (also called Turner syndrome or 45, X): This is caused by a missing X chromosome and affects only girls. Girls with Monosomy X may have heart defects, hearing problems, minor learning disabilities and are usually shorter than average. As adults, they are often infertile.
- **Triploidy:** This condition is caused by having an extra set of 23 chromosomes (for a total of 69) and is associated with severe birth defects. A triploid pregnancy can cause serious complications for the mother, such as excessive bleeding after delivery and a risk of developing cancer. Babies with triploidy rarely make it to term and those that do usually pass away within a few months after delivery. It is important for the doctor to know about triploidy even if the mother miscarries so that she can be monitored for complications.
- **Klinefelter syndrome:** This is caused by an extra copy of the X chromosome, is also known as 47, XXY and only affects boys. Boys with Klinefelter syndrome may have learning disabilities, tend to be taller than average, and most men with this condition are infertile.
- **Triple X syndrome:** This is caused by an extra copy of the X chromosome, is also known as 47, XXX and only affects girls. Some girls with triple X syndrome have learning disabilities, some have emotional problems and most are taller than average.
- **XYY syndrome:** This is caused by an extra copy of the Y chromosome, is also known as 47, XYY and only affects boys. Boys with this condition tend to be taller

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than average and may have associated mild learning and behavioral difficulties.

- Panorama also screens **for five microdeletion syndromes.**

What are microdeletions? Which ones does Panorama screen for?

A small, missing (or “deleted”) piece of a chromosome is called a microdeletion. Unlike Down syndrome, which occurs more frequently in mothers who are 35 and older, microdeletions occur in pregnancies at the same rate for mothers of any age.

In many cases, there are no obvious ultrasound abnormalities that would suggest the fetus has a microdeletion. While many microdeletions have little impact on a child’s health and life, there are some that can cause intellectual disabilities and birth defects. Panorama screens for five microdeletions, all of which can be associated with serious health issues:

- **22q11.2 deletion syndrome / DiGeorge syndrome** (occurs in about 1 in 2,000 births)*
Babies born with 22q11.2 deletion syndrome often have heart defects, immune system problems, and mild-to-moderate intellectual disability. They may also have kidney problems, feeding problems, and/or seizures. Up to 25% of individuals with this syndrome develop schizophrenia in adulthood.
- **1p36 deletion syndrome** (occurs in about 1 in 5,000 births)
Babies born with 1p36 deletion syndrome have weak muscle tone, heart and other birth defects, intellectual disabilities, hearing loss and behavior problems. About half will have seizures.
- **Angelman syndrome** (occurs in about 1 in 12,000 births)
Babies born with Angelman syndrome often have delayed milestones (like sitting, crawling and walking), seizures, and problems with balance and walking. They also have severe intellectual disability and most do not develop speech.
- **Cri-du-chat syndrome**, also known as 5p minus (occurs in about 1 in 20,000 births)
Babies born with Cri-du-chat syndrome typically have low birth weight, small head size, and decreased muscle tone. Feeding and breathing difficulties are also common. They have moderate-to-severe intellectual disability.
- **Prader-Willi syndrome** (occurs in about 1 in 10,000 births)
Babies born with Prader-Willi syndrome have low muscle tone and problems with feeding and gaining weight. They also have intellectual disability. As children and adults, they

have rapid weight gain and often develop obesity-related medical problems.

Who should get the Panorama prenatal screen?

Many pregnant women want to know about the health of their baby. If you would like information about your baby’s health, talk with your healthcare provider. He or she will advise you as to what tests you might want to have to help give you peace of mind.

The Panorama prenatal screen is designed for women of any age and ethnicity who are at least 9 weeks pregnant. It cannot currently be used by women who are carrying more than one baby (twins or triplets), women who have used a donor egg or a surrogate, or those who have received a bone marrow transplant.

Some women have a higher chance for chromosomal abnormalities because of their age, family history or other screening test results. However, certain conditions such as the microdeletions that are on Panorama’s panel affect women of all ages at the same rate. Regardless of your age or family or reproductive history, Panorama can help determine your baby’s risk of being affected with a chromosome condition.

What are the alternatives to the Panorama prenatal screen?

Panorama is not the only screening test available during pregnancy. Older screening tests that measure hormones in a pregnant woman’s blood (called maternal serum screening tests) can also tell you if there is a high chance your baby has a chromosomal condition, such as Down syndrome. Maternal serum screening tests are less accurate than Panorama when screening for the conditions above. This means that serum screening tests are more likely than Panorama to miss certain chromosomal conditions and more likely than Panorama to indicate an abnormal chromosomal condition when none actually exists.

Panorama is a screening test; it is not a diagnostic procedure. This means that test results from Panorama only alert you if your baby is at risk for a chromosomal condition. To diagnose the baby – and know with certainty if the baby has a chromosomal condition – invasive diagnostic tests, such as chorionic villus sampling (CVS) or amniocentesis, can be done. Both of these tests have risks, including the small chance of miscarriage.

Speak with your healthcare provider if you have more questions about your testing options.

*Recent studies have shown incident rates as high as 1/992.