natera™ | Panorama™ Next-generation NIPT

Understanding Your Results

High risk for 1p36 deletion syndrome

What do my results mean?

Your results show that your pregnancy has a high risk for 1p36 deletion syndrome. **This result *does not* mean that your baby has 1p36 deletion syndrome.** The specific chance that your baby has 1p36 deletion syndrome can be found on page 1 of your test report under "Risk after test." To know for sure whether your baby has 1p36 deletion syndrome, you would need to have additional testing. You should not make decisions about your pregnancy based only on this Panorama result, as your baby most likely *does not* have 1p36 deletion syndrome.

1 out of 6 pregnancies with this test result will have 1p36 deletion syndrome*

*The chance that your pregnancy has 1p36 deletion syndrome may be different than what is pictured here. The specific risk to your pregnancy can be found on page 1 of your test report in the "Risk after test" column.

What is 1p36 deletion syndrome?

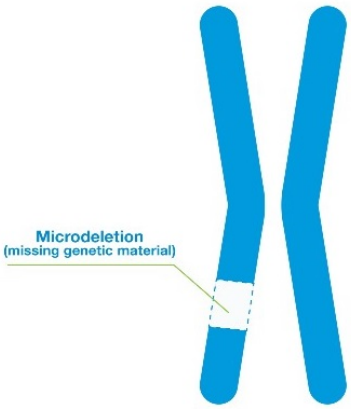
1p36 deletion syndrome is a genetic condition that about 1 in 5,000 or less people are born with. People with 1p36 deletion syndrome have severe intellectual disability and most do not talk or only learn a few words. More than half of people with 1p36 deletion syndrome have seizures, and most have low muscle tone (hypotonia) and problems swallowing. Some people with 1p36 deletion syndrome have heart, kidney, or other birth defects, and they can have behavior problems.[1]

What causes 1p36 deletion syndrome?

1p36 deletion syndrome is usually not inherited and happens by chance. Parents cannot cause 1p36 deletion syndrome to happen by anything they do before or during a pregnancy.

1p36 deletion syndrome happens when a person is missing a small piece of chromosome 1. Chromosomes are tiny structures inside our cells that hold our genes and genetic material. Most people have 46 chromosomes that come in 23 pairs in every cell of their bodies. People who have 1p36 deletion syndrome are missing a small piece of one copy of chromosome 1. This small missing piece is called a microdeletion.

Microdeletion



About 20% of the time, 1p36 deletion syndrome is inherited from a healthy parent (who does not have 1p36 deletion syndrome).[1] If a baby does have 1p36 deletion syndrome, testing can be performed to find out if it was inherited. This testing can tell parents the chance of 1p36 deletion syndrome happening again in another pregnancy. Most parents of children with 1p36 deletion syndrome learn that the chance of 1p36 deletion syndrome happening again is small.

What can I do next?

You should talk to your healthcare provider about these results. They usually will refer you to a genetic counselor and/or a maternal-fetal medicine specialist to talk about your options for further testing.

If you want to find out if your baby has 1p36 deletion syndrome during pregnancy, you can have a test called a CVS (chorionic villus sampling) or an amniocentesis. These tests are diagnostic and will tell you for sure if your baby has 1p36 deletion syndrome or not. Both tests have a small risk of miscarriage.

If you do not want these tests during pregnancy, the baby can be tested after birth using blood from the umbilical cord. This testing is also diagnostic and will tell you for sure if your baby has 1p36 deletion syndrome.

It is important to have diagnostic testing either during pregnancy or when the baby is born to know for sure if the baby has 1p36 deletion syndrome. Changes to medical care and early intervention services can often improve the health and development of a child with 1p36 deletion syndrome.

Where can I find more information?

- Unique (Understanding Rare Chromosome and Gene Disorders) rarechromo.org
- Chromosome Disorder Outreach chromodisorder.org
- MedlinePlus medlineplus.gov/genetics/condition/1p36-deletion-syndrome
- CVS marchofdimes.org/find-support/topics/planning-baby/chorionic-villus-sampling
- Amniocentesis marchofdimes.org/find-support/topics/planning-baby/amniocentesis

NEVA is always available to help you learn about your results. You can connect with Natera's Educational Virtual Assistant (NEVA) by logging into the patient portal at my.natera.com.

If you would like to discuss your results with a Natera genetic counselor, you can schedule a free information session at naterasession.com, by texting SESSION to 636363, or by calling 1.877.476.4743. Please select **Panorama Non-Invasive Prenatal Chromosome Screening Post-Test** as the appointment type.

You can find a local genetic counselor through the National Society of Genetic Counselors at findageneticcounselor.nsgc.org.

1. MedlinePlus [Internet]. Bethesda (MD): National Library of Medicine (US); [updated 2022 Jan 19]. 1p36 deletion syndrome; [updated 2020 Sept 8; reviewed 2014 Jan 1; cited 2022 Feb 1]; [about 5 p.]. Available from: medlineplus.gov/genetics/condition/1p36-deletion-syndrome.

13011 McCallen Pass, Building A Suite 100 | Austin, TX 78753 | natera.com