natera™ | Panorama™ Next-generation NIPT

Understanding Your Results

High risk for 22q11.2 deletion syndrome

What do my results mean?

Your results show that your pregnancy has a high risk for 22q11.2 deletion syndrome (formerly known as DiGeorge syndrome). **This result *does not* mean that your baby has 22q11.2 deletion syndrome.** The chance that your baby has 22q11.2 deletion syndrome is 50% (1 out of 2), and the chance that your baby does not have 22q11.2 deletion syndrome is also 50% (1 out of 2). To know for sure whether your baby has 22q11.2 deletion syndrome, you would need to have additional testing. You should not make decisions about your pregnancy based only on this Panorama result, as it is not certain that your baby has 22q11.2 deletion syndrome.

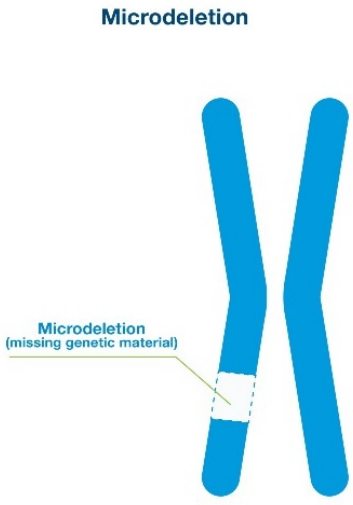
1 out of 2 pregnancies with this test result will have 22q11.2 deletion syndrome

What is 22q11.2 deletion syndrome?

22q11.2 deletion syndrome is a genetic condition that about 1 in 4,000 people are born with. 22q11.2 deletion syndrome can affect many different parts of the body. It can be very mild in some people and very severe in others. Many people with 22q11.2 deletion syndrome are born with heart defects, and they can also have defects in the roof of their mouth (palate). Some people with 22q11.2 deletion syndrome have trouble fighting infections (immune system problems), kidney problems, low calcium levels leading to seizures, and/or hearing loss. Many people with 22q11.2 deletion syndrome have developmental delay, learning problems, and/or mild intellectual disability. Children with 22q11.2 deletion syndrome are also more likely than other children to have attention-deficit/hyperactivity disorder (ADHD) or autism spectrum disorder.[1]

What causes 22q11.2 deletion syndrome?

22q11.2 deletion syndrome is usually not inherited and happens by chance. Parents cannot cause 22q11.2 deletion syndrome to happen by anything they do before or during pregnancy. 22q11.2 deletion syndrome happens when someone is missing a small piece of chromosome 22. 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 22q11.2 deletion syndrome are missing a small piece of one copy of chromosome 22. This small missing piece is called a microdeletion.



About 10% of the time, 22q11.2 deletion syndrome is inherited from a parent.[1] If a baby does have 22q11.2 deletion syndrome, testing can be performed to find out if it was inherited. This testing can tell parents the chance of 22q11.2 deletion syndrome happening again in another pregnancy. Most parents of children with 22q11.2 deletion syndrome learn that the chance of 22q11.2 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 22q11.2 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 22q11.2 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 22q11.2 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 22q11.2 deletion syndrome. If you have not had diagnostic testing during pregnancy, you should talk to your healthcare provider to make sure that you are delivering your baby in a tertiary care center. These hospitals have specialists who will watch the baby carefully while waiting to find out if they have 22q11.2 deletion syndrome or not. Changes to medical care and early intervention services can often improve the health and development of a child with 22q11.2 deletion syndrome.

Where can I find more information?

- International 22q11.2 Foundation 22q.org
- Chromosome 22 Central c22c.org
- Chromosome Disorder Outreach chromodisorder.org
- MedlinePlus medlineplus.gov/genetics/condition/22q112-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]. 22q11.2 deletion syndrome; [updated 2020 Sept 8; reviewed 2019 Dec 1; cited 2022 Feb 2]; [about 5 p.]. Available from: medlineplus.gov/genetics/condition/22q112-deletion-syndrome.

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