natera™ | Panorama™ Next-generation NIPT

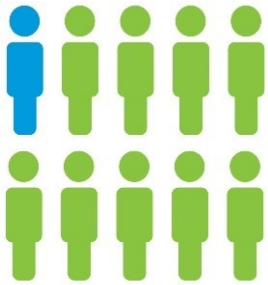
Understanding Your Results

High risk for Angelman syndrome

What do my results mean?

Your results show that your pregnancy has a high risk for Angelman syndrome. **This result *does not* mean that your baby has Angelman syndrome.** The chance that your baby has Angelman syndrome is 10% (1 out of 10), and the chance that your baby does not have Angelman syndrome is 90% (9 out of 10). To know for sure whether your baby has Angelman 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 Angelman syndrome.

1 out of 10 pregnancies with this test result will have Angelman syndrome



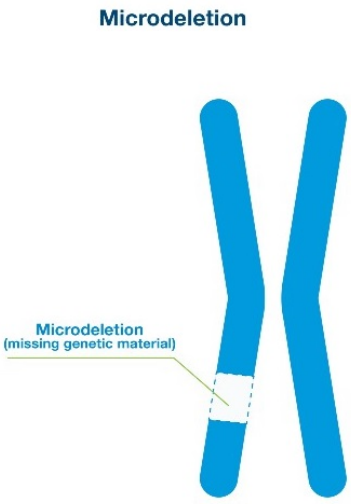
What is Angelman syndrome?

Angelman syndrome is a genetic condition that about 1 in 12,000 or less people are born with. People with Angelman syndrome have severe intellectual disability and often will never learn to talk. They can also have seizures, small heads (microcephaly), and problems with balance and walking (ataxia). Most children with Angelman syndrome have trouble sleeping and are very excitable, though these problems often get better as they get older. People with Angelman syndrome usually have a normal lifespan.[1]

What causes Angelman syndrome?

Angelman syndrome is usually not inherited and happens by chance. Parents cannot cause Angelman syndrome to happen by anything they do before or during a pregnancy. Angelman syndrome is usually caused by a missing piece of chromosome 15. 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. Most people who have Angelman syndrome are missing a small piece of one copy of chromosome 15. This small missing piece is called a microdeletion. People can also have Angelman syndrome if both of their chromosome 15 copies come from the same parent. This is called uniparental disomy. Usually, we receive one copy of each chromosome from each of our parents. If someone receives both copies of chromosome 15 from the same parent, they can have Angelman syndrome, even if no pieces of the chromosome are missing.

If a baby does have Angelman syndrome, testing can be performed to learn if it was inherited from a healthy parent (who does not have Angelman syndrome). This testing can tell parents the chance of Angelman syndrome happening again in another pregnancy. Most parents of children with Angelman syndrome learn that the chance of Angelman 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 Angelman 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 Angelman 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 Angelman 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 Angelman syndrome. Changes to medical care and early intervention services can often improve the health and development of a child with Angelman syndrome.

Where can I find more information?

- Angelman Syndrome Foundation angelman.org
- FAST (Foundation for Angelman Syndrome Therapeutics) cureangelman.org
- MedlinePlus medlineplus.gov/genetics/condition/angelman-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]. Angelman syndrome; [updated 2020 Sept 8; reviewed 2015 May 1; cited 2022 Jan 31]; [about 5 p.]. Available from: medlineplus.gov/genetics/condition/angelman-syndrome.

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