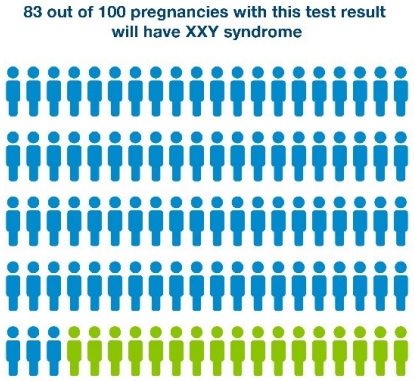
natera™ | Panorama™ Next-generation NIPT

Understanding Your Results

Suggestive of XXY syndrome

What do my results mean?

Your results show that your pregnancy has a high risk for XXY syndrome (also known as Klinefelter syndrome). **This result *does not mean* that your baby has XXY syndrome.** The chance that your baby has XXY syndrome is 83%, and the chance that your baby does not have XXY syndrome is 17%. To know for sure whether your baby has XXY 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 XXY syndrome.

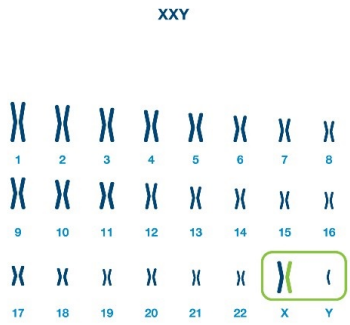


What is XXY syndrome?

XXY syndrome is a genetic condition that about 1 in 650 males are born with. People with XXY syndrome are usually born healthy and without birth defects. They are expected to live a normal lifespan and usually have normal intelligence. Some children with XXY syndrome have low muscle tone (hypotonia) and learning problems. Most people with XXY syndrome are taller than average. They usually have low levels of testosterone, which means that they can have problems with puberty and having children.[1]

What causes XXY syndrome?

XXY syndrome usually happens by chance. Parents cannot cause XXY syndrome to happen by anything they do before or during a pregnancy. XXY syndrome happens when a person has an extra sex chromosome. 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. Sex chromosomes are labeled either X or Y. Most people have two X chromosomes or one X and one Y chromosome. People with XXY syndrome have two X chromosomes and one Y chromosome.



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 XXY 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 XXY 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 XXY 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 XXY syndrome. Changes to medical care and early intervention services can often improve the health and development of a child with XXY syndrome.

Where can I find more information?

- The Focus Foundation thefocusfoundation.org
- Association for X and Y Chromosome Variations genetic.org
- MedlinePlus medlineplus.gov/genetics/condition/klinefelter-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]. Klinefelter syndrome; [updated 2020 Sept 8; reviewed 2019 April 1; cited 2022 Jan 20]; [about 5 p.]. Available from: medlineplus.gov/genetics/condition/klinefelter-syndrome.

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