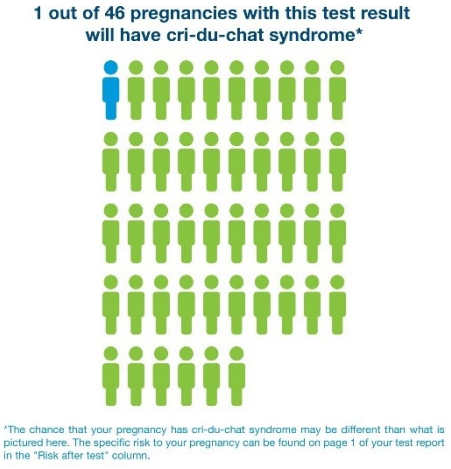
natera™ | Panorama™ Next-generation NIPT

Understanding Your Results

High risk for cri-du-chat syndrome

What do my results mean?

Your results show that your pregnancy has a high risk for cri-du-chat syndrome (also known as 5p- or 5p minus). **This result *does not* mean that your baby has cri-du-chat syndrome.** The specific chance that your baby has cri-du-chat syndrome can be found on page 1 of your test report under "Risk after test." To know for sure whether your baby has cri-du-chat 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 cri-du-chat syndrome.

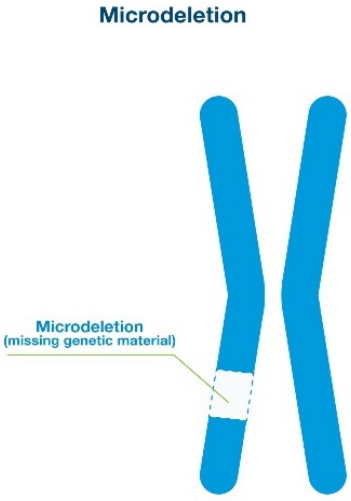


What is cri-du-chat syndrome?

Cri-du-chat syndrome is a genetic condition that about 1 in 20,000 or less people are born with. Babies with cri-du-chat syndrome have a high-pitched cry that many people think sounds like a cat. They also have intellectual disabilities, low birth weight, low muscle tone, and small heads (microcephaly).[1] Some babies with cri-du-chat syndrome will be born with heart defects and may have hearing and vision problems or curving of the spine (scoliosis). Feeding and breathing problems are common during infancy, and some babies do not survive their first year of life.[2]

What causes cri-du-chat syndrome?

Cri-du-chat syndrome usually is not inherited and happens by chance. Parents cannot cause cri-du-chat syndrome to happen by anything they do before or during a pregnancy. Cri-du-chat syndrome happens when someone is missing a small piece of chromosome 5. 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 cri-du-chat syndrome are missing a small piece of one copy of chromosome 5. This small missing piece is called a microdeletion.



About 10% of the time, cri-du-chat syndrome is inherited from a healthy parent (who does not have cri-du-chat syndrome).[1] If a baby does have cri-du-chat syndrome, testing can be performed to find out if it was inherited. This testing can tell parents the chance of cri-du-chat syndrome happening again in another pregnancy. Most parents of children with cri-du-chat syndrome learn that the chance of cri-du-chat 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 cri-du-chat 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 cri-du-chat 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 cri-du-chat 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 cri-du-chat syndrome. Changes to medical care and early intervention services can often improve the health and development of a child with cri-du-chat syndrome.

Where can I find more information?

- 5p- Society fivepminus.org
- National Organization for Rare Disorders rarediseases.org/rare-diseases/cri-du-chat-syndrome
- MedlinePlus medlineplus.gov/genetics/condition/cri-du-chat-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]. Cri-du-chat syndrome; [updated 2020 Sept 8; reviewed 2020 March 1; cited 2022 Jan 31]; [about 5 p.]. Available from: medlineplus.gov/genetics/condition/cri-du-chat-syndrome.
2. Ajitkumar A, et al. Cri Du Chat Syndrome. [Updated 2021 Sep 14]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-. Available from: ncbi.nlm.nih.gov/books/NBK482460. Accessed January 2022.

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