

# TRISOMY 13

## OVERVIEW

Trisomy 13, also called Patau Syndrome, is a life-threatening chromosomal abnormality that affects the way a baby's major organs develop during pregnancy.

Trisomy means *three chromosomes*. Chromosomes carry a person's genes, and are inside every cell in the body. Normally, an embryo gets one set of chromosomes (numbered 1 to 23) from each parent, for a total of 46 chromosomes. In Trisomy 13, there is an extra copy of chromosome 13. This extra chromosome causes major defects to multiple organs including the heart, brain and kidneys, as well as defects in facial features and limbs.

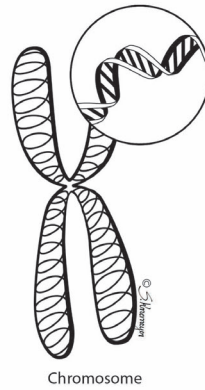
It is hard to predict how long a baby with Trisomy 13 will live. The medical problems associated with Trisomy 13 will cause most babies to die before they are born, or to die within a few days or weeks after birth. A small percentage of these babies will live more than one year. Babies who do live usually have severe mental, physical and chronic medical problems.

Trisomy 13 is a rare and random birth defect, with no definite cause. While mothers of any age can have a child with Trisomy 13, the risk of having a child with the condition increases as a woman gets older.

## HOW TRISOMY 13 IS FOUND

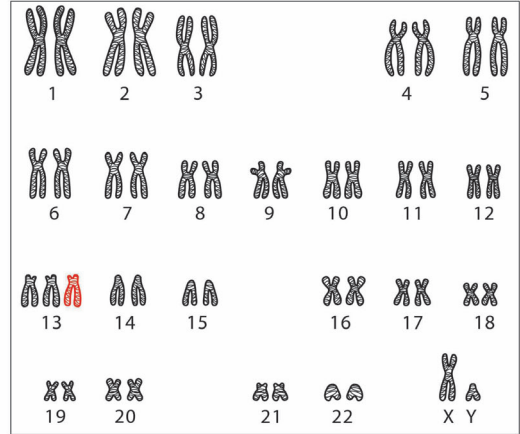
Your doctor may offer you a blood test early in your pregnancy to screen for certain conditions in the baby. Your blood test may indicate an increased risk for Trisomy 13, however further tests are needed to confirm the diagnosis. Trisomy 13 can also be found during pregnancy when a routine ultrasound shows an unusual appearance to the head, face or limbs. Your doctor may order more tests to confirm the diagnosis, including:

- **Chorionic Villous Sampling (CVS).** This test takes a small sample of the placenta which will be tested for chromosomal abnormalities. This test can only be done late in the first trimester.
- **Cell-Free Fetal DNA testing:** Your doctor can take a sample of your blood to look for copies of fetal (baby) DNA. This is also a screening test. A positive result should be confirmed with an additional test.
- **Amniocentesis.** A small sample of the amniotic fluid surrounding the baby in the womb is taken and tested for chromosomal problems. This test is usually done in the middle of the second trimester.
- **MRI (magnetic resonance imaging):** This type of imaging gives your doctors more detailed pictures of your baby's organs.
- **Testing the baby:** After birth, a test is done with a sample of your baby's blood to look for extra chromosomes.



DNA, which contains genes

Normally, each cell has 23 pairs of chromosomes. In trisomy 13, there is an extra copy of chromosome 13.



## SYMPTOMS AND CAUSES OF TRISOMY 13

Trisomy 13 occurs at the time of conception. Normally, egg and sperm cells each contain 23 chromosomes. During normal fertilization of the egg, one copy of each chromosome (which carry the parents' genes) is passed from each parent to the embryo, to make a complete set of 23 pairs (46 chromosomes). The chromosomes contain genes, made of DNA, which provides instructions to the embryo for how to grow and develop into a baby.

However, sometimes an egg or sperm cell will contain extra chromosomes. These extra chromosomes also get passed to the embryo and disrupt how the cells grow and develop.

With Trisomy 13, there is an extra copy of chromosome 13. This causes birth defects. The extra copies can appear in three different ways – which can impact the severity of the condition:

- **There are three copies of chromosome 13 in each cell instead of two.** This extra copy causes the classic features of Trisomy 13, and is often called free or full Trisomy 13. This is the most common type of Trisomy, occurring in about 75 percent of all cases.
- **A portion of chromosome 13 is attached to another chromosome (translocation).** This translocation, or misplacing of a portion of the chromosome, can lead to symptoms that range from mild to severe, depending on how much extra chromosome 13 is present. Often, this translocation is present in a parent, who shows no symptoms, but is at risk for passing the abnormal chromosome onto future children as well. This is found in about 20 percent of Trisomy 13 cases.
- **There are three copies of chromosome 13 in only some of the cells.** This rare type of the condition is referred to as mosaic Trisomy 13. Physical symptoms of mosaic Trisomy 13 may be milder than the full version.

Babies with Trisomy 13 are often born with one or more of the following conditions:

- Small head size (microcephaly)
- Heart problems – about 80 percent of children with Trisomy 13 will have a heart defect, although this is usually a more mild defect.
- Severe brain abnormality (holoprosencephaly), which can affect the baby's ability to breathe on his/her own
- Spinal cord defects (*spina bifida*)
- Skin/scalp defects on the head (cutis aplasia)
- Very small or poorly developed eyes (microphthalmia)
- Extra fingers or toes, unusually formed hands and feet
- Weak muscles (*hypotonia*)
- An opening in the lip (*cleft lip*) or the roof of the mouth (*cleft palate*)
- Abnormally formed ears and hearing problems
- Problems with kidneys and bladder function
- Some abdominal organs located outside the body in a sac (omphalocele)
- Scoliosis (a sideways curve in the spine)
- Severe developmental delay

As children with Trisomy 13 age, they may be unable to speak or walk. They may have problems breathing, or eating or drinking by mouth.

While no one knows what triggers the chromosomal defects leading to Trisomy 13, they do know that it is **not caused by anything that the parents have done before or during pregnancy.**

In rare cases, there are inherited genetic factors that could increase the risk of a child developing Trisomy 13. Genetic testing and counseling is advised for parents who already have had a child with the condition and are thinking about having a second child.

## **BETWEEN DIAGNOSIS AND DELIVERY**

Advances in prenatal testing are helping doctors to diagnose several fatal and life-threatening conditions early in pregnancy. Following a diagnosis of Trisomy 13, you will continue to receive care from your obstetrician, who will discuss the different options you have for managing your pregnancy. You may be referred to the Ohio Fetal Medicine Collaborative (OFMC) for additional information and help.

Through the OFMC, you can see a doctor who specializes in the care of high-risk patients. You may also meet with specialists from Nationwide Children's Hospital to learn more about how your baby will be cared for when he or she is born. A fetal nurse coordinator can also help answer your questions and concerns, guide you through your pregnancy and prepare you for what to expect.



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**“Palliative care for children is the active total care of the child’s body, mind and spirit, and also involves giving support to the family.**

**It begins when illness is diagnosed, and continues regardless of whether or not a child receives treatment directed at the disease.”**

*– World Health Organization*

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## **PERINATAL PALLIATIVE CARE**

When prenatal testing shows that a baby has a life threatening or life-limiting condition, many parents feel overwhelmed at the thought of preparing for the possible death of their child.

Perinatal palliative care is designed to help families navigate a confusing and heartbreaking situation, and cope with the challenges of expecting a newborn with a potentially fatal illness. You will be faced with many difficult decisions following your baby’s diagnosis. The goal of perinatal palliative care is to provide compassionate and expert care starting from the time of diagnosis. While there are no cures treatments for many of the conditions commonly referred for palliative care, there are many things the team can do to help support you, your baby, and your family.

## **WHO IS PART OF A PERINATAL PALLIATIVE CARE TEAM?**

Perinatal palliative care is provided by a multi-disciplinary team of specialists including doctors, nurse practitioners, nurses, social workers, and clergy. The team is committed to supporting you and your family.

You and your family will meet with members of the palliative care team before delivery, and have additional visits as needed during your pregnancy and after delivery. Palliative care is highly individual and there is no one right path for every family. We will help guide you through making decisions about how to best care for your baby, decisions that fit with your goals and values. We will explain the different medical treatments that may be chosen with a focus on keeping your baby comfortable, and allowing you to spend quality time together.

Many families find the time between diagnosis and delivery very emotionally difficult. Coordinators can help connect you with counseling services and support groups.

## WHAT HAPPENS WHEN YOUR BABY IS BORN

You should discuss your plans for labor and delivery with your obstetrician. The specifics of the care your baby receives after birth will depend on what you and the team have discussed during your prenatal palliative care consult. Depending on your wishes, the neonatal team can be present at delivery. There are a variety of treatments and medicines that can be provided for comfort and support. The plan for how to care for your baby can evolve over time, as well.

## Lifelong Considerations

While the time with your baby with Trisomy 13 may be limited, we know that every minute is precious to you. If your baby lives and is able to leave the hospital, we will help teach you how to support and care your baby at home, with a focus on preventing problems, maximizing your child's abilities, and prevent pain and suffering. Physical and occupational therapy may be needed. About 1 in 20 babies with Trisomy 13 will live beyond the first year, and there have been rare cases of babies with Trisomy 13 who survive into childhood.

## Future Pregnancies

Most parents who have one child with Trisomy 13 do not have another with the condition. However, in some cases, Trisomy 13 can be inherited, and your doctor may recommend genetic counseling to discuss risks for a future pregnancy.

## ADDITIONAL RESOURCES

- **March of Dimes** — <http://www.marchofdimes.com/baby/chromosomal-conditions.aspx>
- **Chromosome Disorder Outreach** — <http://www.chromodisorder.org/>
- **Support Organization for Trisomy** — <http://trisomy.org/>
- **Living with Trisomy 13** — <http://www.trisomy13archive.com/>

