CONDITION

Trisomy 13 syndrome

Trisomy 13 syndrome is a chromosome condition that affects development of the brain, heart, and other parts of the body.

WHAT IT IS
Chromosome condition affecting development before birth

BODY SYSTEM
Brain, heart, face, limbs, and other organs

MAIN SYMPTOMS
Developmental differences, heart problems, feeding difficulties, breathing problems

WHAT CAUSES IT
Extra chromosome 13, usually from random cell division

HOW IT IS DIAGNOSED
Chromosome testing after prenatal or blood sampling

MAIN TREATMENT
Individualised supportive care, procedures, and sometimes surgery

CAN IT BE CURED?
No - extra chromosome cannot be removed

IS IT AN EMERGENCY?
Sometimes - breathing trouble, blue colour, or unresponsiveness

LONG-TERM OUTLOOK
Often life-limiting; some children survive for years

Overview

Trisomy 13 syndrome is a chromosome condition present from conception. It is also called trisomy 13 or Patau syndrome.

Most people have two copies of chromosome 13 in each cell. A baby with trisomy 13 has three complete copies, or extra material from chromosome 13. Chromosomes are packages of DNA that carry instructions for growth and development.

The extra genetic material can affect many parts of the body before birth. Commonly affected areas include the brain, heart, face, eyes, kidneys, hands, and feet. The condition often causes major medical needs and profound developmental disability.

Many pregnancies affected by trisomy 13 end in miscarriage or stillbirth. Among babies born

alive, survival is often limited to days, weeks, or months. Some children live beyond their first birthday, and a smaller number survive for several years. The outlook can be different for children with mosaic or partial trisomy 13, but chromosome results alone cannot predict an individual child's course.

Families may face difficult decisions about tests, hospital treatment, surgery, feeding support, and comfort care. These choices should reflect the child's medical needs and the family's values, with clear information from the care team.

Symptoms

Trisomy 13 affects each baby differently. Not every baby has every feature, and the seriousness of each problem differs.

Possible physical and medical features include:

Babies who survive usually have significant delays in movement, communication, learning, and other areas of development. Hearing and vision may also be affected.

Some findings are seen on an ultrasound during pregnancy. Others become clear only after birth. Physical features can suggest trisomy 13, but they cannot confirm it without chromosome testing.

- Differences in brain development, sometimes including incomplete separation of the brain's two halves
- Congenital heart disease, meaning a heart problem present at birth
- Cleft lip, cleft palate, or both
- Very small or underdeveloped eyes
- Extra fingers or toes
- Differences in the scalp, abdominal wall, kidneys, or genital development
- Low muscle tone, poor growth, or difficulty feeding
- Breathing pauses, seizures, or problems controlling body temperature

Causes and risk factors

Trisomy 13 is caused by extra genetic material from chromosome 13. It is not caused by anything someone did during pregnancy.

There are several chromosome patterns:

The chance of a random trisomy rises with the age of the egg, so it is higher with increasing maternal age. However, trisomy 13 can occur in a pregnancy at any age.

Most cases are not inherited. If testing finds a translocation, chromosome testing may be offered to both biological parents. A genetics professional can explain whether there is an increased chance in a future pregnancy.

- Full trisomy 13: Every tested cell has three separate copies of chromosome 13. This is the most common form and usually results from a random error when an egg or sperm forms.
- Translocation trisomy 13: Extra chromosome 13 material is attached to another chromosome. This can happen as a new event, or it may be related to a balanced chromosome rearrangement in a parent.
- Mosaic trisomy 13: Some cells have an extra chromosome 13 and others do not. Effects can be less severe, but this is not always the case.
- Partial trisomy 13: Only part of chromosome 13 is present in an extra copy. The effects are influenced by which genes are included.

Diagnosis

During pregnancy, screening may show an increased chance of trisomy 13. Screening can include ultrasound findings, blood tests, or cell-free DNA testing. Cell-free DNA examines small DNA fragments mainly released by the placenta.

A screening result is not a diagnosis. False-positive and false-negative results can occur. Diagnostic testing is needed to determine whether the fetus has trisomy 13.

Diagnostic options during pregnancy include:

Because placental cells and fetal cells can occasionally have different chromosome patterns, further testing may be needed after an uncertain chorionic villus sampling result.

After birth, trisomy 13 is usually confirmed with a chromosome analysis called a karyotype, using a blood sample. This test can show an extra chromosome and help identify whether the pattern is full, mosaic, or caused by a translocation. Faster preliminary tests may be used, but a full chromosome analysis provides more detail.

A detailed ultrasound during pregnancy or examinations after birth can assess the brain, heart, kidneys, and other organs. These tests help plan care but do not determine exactly how long a child will live.

Genetic counselling can help you understand the result, its limits, and what it may mean for other pregnancies.

- Chorionic villus sampling , which tests a small sample from the placenta
- Amniocentesis , which tests cells in the fluid surrounding the fetus

Treatment and management

There is no treatment that can remove the extra chromosome. Care focuses on the child's comfort, medical problems, development, and the goals chosen by the family.

A newborn may need help with breathing, warmth, blood sugar, or feeding. Tests may assess the heart, brain, kidneys, hearing, and vision. Seizures, infections, reflux, and pain can be treated when they occur.

Feeding support may include changes to positioning, specialised bottles, or tube feeding. A feeding assessment can help identify swallowing problems and reduce the chance of milk entering the lungs.

Some children are considered for procedures or surgery, such as treatment for a heart defect, cleft lip or palate, or feeding difficulty. Whether an intervention is appropriate involves more than the diagnosis alone. The likely benefit, discomfort, recovery, other medical problems, and the family's goals all matter.

Families may choose treatment aimed at prolonging life, comfort-focused care, or a combination. Comfort-focused care is active care. It can include warmth, feeding when safe and comfortable, relief of pain or breathlessness, family contact, spiritual support, and help creating memories.

For children who live longer, care may involve several specialists. Physical, occupational, feeding, speech, hearing, and vision services may support comfort and development. Therapy cannot correct the chromosome difference, but it may help a child use their abilities and interact with others.

A written care plan can record which treatments are wanted during a health crisis. It can also be reviewed as the child's needs and the family's priorities change.

Complications

Breathing problems are common. They may result from pauses in breathing, airway differences, infection, heart disease, or reduced control of breathing by the brain.

Swallowing difficulties can cause poor weight gain, dehydration, or aspiration. Aspiration means food, milk, or saliva enters the airway and lungs. This can lead to coughing, breathing

trouble, or repeated chest infections.

Heart defects may lead to low oxygen levels, poor circulation, or heart failure. Seizures, kidney problems, hearing loss, vision loss, reflux, constipation, and changes in the spine or joints may also need care.

A child's risks are shaped by their particular organ differences, not only by the label of trisomy 13.

Prevention

Trisomy 13 usually cannot be prevented. Most cases happen because of a random chromosome error, not because of food, exercise, medicines, stress, or something a parent did.

Prenatal screening can estimate the chance of the condition, but it does not prevent it. Diagnostic testing can provide more certain information during pregnancy.

If a parent carries a balanced chromosome translocation, a genetics specialist can discuss reproductive options. These may include testing during pregnancy or, in some circumstances, testing embryos created through in vitro fertilisation. No option can guarantee a pregnancy without a chromosome condition.

Living with it and what to expect

The early course is often uncertain. Some babies die during pregnancy or shortly after birth. Others become stable enough to go home, sometimes with oxygen, tube feeding, medicines, monitoring equipment, or community nursing support.

Children who survive usually have profound developmental disability and ongoing medical needs. Development tends to be slow. Some children learn to recognise familiar voices, express preferences, smile, interact, or use limited movements and communication. Skills and medical needs differ greatly.

Health may remain fragile over months and years. Breathing infections, seizures, feeding problems, and heart complications can lead to repeated appointments or hospital stays. Some children have periods of relative stability between illnesses.

Mosaic or partial trisomy 13 may be linked with longer survival or fewer medical problems, but neither term guarantees a milder course. The percentage of affected cells in a blood sample does not show what is happening in every organ.

Routine follow-up may include growth and nutrition checks, review of breathing and heart

symptoms, seizure care, hearing and vision assessment, dental care, and support for movement and communication. Plans should be based on the child's current needs rather than assumptions about the diagnosis.

Caring for a child with trisomy 13 can involve grief, uncertainty, and meaningful family experiences at the same time. Perinatal palliative care, hospice services, social workers, counsellors, faith communities, and condition-specific family groups may offer practical and emotional support.

When to get medical help

Seek emergency help if your baby or child:

If your child has an agreed emergency or palliative care plan, follow it and contact the named service. If you are unsure and the situation appears life-threatening, call emergency services.

Contact the child's healthcare team promptly for new feeding difficulty, repeated vomiting, fewer wet nappies, increasing sleepiness, worsening seizures, fever, signs of pain, or a change in breathing that is not immediately life-threatening.

Routine appointments are the right place to discuss growth, equipment, therapies, sleep, constipation, hearing, vision, or changes to long-term care goals.

During pregnancy, a high-chance screening result is not usually an emergency. Arrange timely follow-up with your maternity team to discuss diagnostic testing and specialist assessment.

- Is struggling to breathe, has a sudden major change in breathing, or stops breathing
- Becomes blue, grey, or unusually pale
- Is unresponsive or much harder to wake than usual
- Has a prolonged seizure or repeated seizures outside their care plan
- Has severe choking or a suspected blocked airway

Common questions

Is trisomy 13 the same as Patau syndrome?

Yes. Patau syndrome is another name for trisomy 13. Some families and professionals prefer the chromosome-based name because it describes the underlying condition.

Can a baby with trisomy 13 survive?

Some can. Many affected pregnancies do not reach birth, and many liveborn babies die during

the first weeks or months. A minority live beyond one year, and some live for several years. No test can predict an individual baby's lifespan with certainty.

Did I cause my baby's trisomy 13?

No. Full trisomy 13 usually comes from a random error as an egg or sperm forms. It is not caused by ordinary activities, stress, or something eaten during pregnancy.

Can a prenatal screening result be wrong?

Yes. Ultrasound, blood screening, and cell-free DNA estimate chance; they do not confirm trisomy 13. Chorionic villus sampling or amniocentesis with chromosome testing is needed for a prenatal diagnosis.

Can trisomy 13 happen again in another pregnancy?

It can, but the chance is not the same for every family. It is influenced by the chromosome pattern and parental age. A translocation may raise the chance if a biological parent carries a balanced rearrangement. Genetic counselling can provide an estimate based on the actual test results.

Does mosaic trisomy 13 mean the condition will be mild?

Not necessarily. Mosaic means the extra chromosome is found in some cells but not others. The effects depend partly on which tissues contain those cells, and a blood test cannot measure every organ.

Sources

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