CONDITION

D trisomy syndrome

D trisomy syndrome is a chromosome condition usually caused by an extra copy of chromosome 13.

WHAT IT IS
A chromosome condition involving extra chromosome 13 material

BODY SYSTEM
Brain, heart, face, eyes, limbs, kidneys, and digestion

MAIN SYMPTOMS
Heart defects, facial clefts, extra digits, developmental disability

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

HOW IT IS DIAGNOSED
Chromosome testing before or after birth

MAIN TREATMENT
Supportive care, symptom treatment, and selected surgery

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

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

LONG-TERM OUTLOOK
High infant mortality; some children live for years

Overview

D trisomy syndrome is an older name usually used for trisomy 13. Trisomy means that cells contain three copies of a chromosome instead of the usual two. Chromosome 13 carries genes that guide development before birth.

The name comes from an older system that grouped chromosomes 13, 14, and 15 as "D-group" chromosomes. Modern care usually names the chromosome involved. If an old medical record says only "D trisomy," the original chromosome report can help confirm the diagnosis.

Extra chromosome 13 material can affect the brain, heart, face, eyes, limbs, kidneys, digestive system, and other organs. The effects are often serious, but they vary between pregnancies and children.

Many affected pregnancies end in miscarriage or stillbirth. Many babies born alive have complex medical needs and do not survive infancy, although some live for several years or longer. The chromosome pattern and medical problems provide useful information but cannot predict one child's future with certainty.

D trisomy syndrome is genetic, not an infection. The usual form is not caused by anything a parent did during pregnancy.

Symptoms

Signs may be detected on a prenatal ultrasound or suspected after birth from a combination of physical features and medical problems.

Possible features include:

Some babies have holoprosencephaly, in which the front part of the developing brain does not divide in the usual way. This can affect feeding, muscle control, facial development, and seizure risk.

Heart defects may disrupt blood flow and oxygen levels. A baby may breathe quickly, tire during feeds, gain weight slowly, or develop a blue or grey colour around the lips or skin.

Mosaic or partial trisomy 13 may cause a different combination of features. Mosaic means only some cells carry the extra chromosome. Partial means only part of chromosome 13 is present in an extra copy. These forms may be less severe, but this is not guaranteed.

- Poor growth before or after birth
- Cleft lip or cleft palate
- Small or underdeveloped eyes
- Extra fingers or toes
- Areas where scalp skin did not fully form
- Structural heart defects
- Differences in brain development
- Kidney or digestive tract abnormalities
- Low muscle tone, feeding difficulty, or breathing problems
- Seizures and major developmental disability

Causes and risk factors

Most cases result from a random error when an egg or sperm forms. The chromosomes fail to separate correctly, leaving an extra copy of chromosome 13. This is called nondisjunction.

The chance of this chromosome error rises with the age of the egg, although it can happen at any parental age.

Less often, extra chromosome 13 material is attached to another chromosome. This is called a translocation. It may arise for the first time or be inherited from a parent with a balanced translocation.

A balanced translocation usually does not cause health problems because the expected amount of genetic material is present but arranged differently. However, a carrier's eggs or sperm can contain an unbalanced amount of chromosome material.

Mosaic trisomy 13 usually begins with a random cell-division error after fertilisation. Some cells then have the usual chromosome number while others have an extra chromosome 13.

The chance of recurrence depends on the chromosome pattern and may be higher if a parent carries a balanced translocation. A genetics professional can interpret the result and arrange parental testing when appropriate.

Diagnosis

During pregnancy, ultrasound, blood screening, or cell-free DNA screening may estimate the chance of trisomy 13. Cell-free DNA screening examines small pieces of placental DNA in the pregnant person's blood.

Screening cannot confirm the condition, and a high-chance result does not always mean the fetus has trisomy 13. Diagnostic testing is needed for certainty.

Prenatal diagnostic options include chorionic villus sampling, which tests placental tissue, and amniocentesis, which tests the fluid surrounding the fetus. Both collect cells for chromosome analysis and have benefits and risks that should be discussed before the procedure.

After birth, diagnosis is usually confirmed with a blood test called a karyotype. It can identify full trisomy, mosaicism, or a translocation. Other chromosome tests may be used if a smaller extra section is suspected.

Further assessments may check the heart, brain, kidneys, hearing, vision, breathing, and feeding. These tests do not confirm the chromosome condition but help identify support needs.

Treatment and management

There is no treatment that removes the extra chromosome material. Care focuses on comfort, manageable medical problems, development, and informed family choices.

Early care may include support for breathing, temperature, feeding, and low blood sugar. Some babies can feed by mouth with positioning or specialist help; others need milk through a tube into the stomach.

Heart, digestive, cleft, or other surgery may be considered for selected children. Decisions are individual because surgery may involve a difficult recovery and may not change problems caused by the chromosome condition. The expected benefit, burden, family goals, and child's overall health all matter.

Ongoing management may include:

Palliative care is not limited to the end of life. It can accompany medical or surgical treatment to relieve symptoms, plan for changes, and support the child and family.

Some families choose mainly comfort-focused care, while others choose selected procedures or intensive treatment. Choices may change as the child's health and the family's goals change.

- Treatment for seizures, heart failure, infections, pain, or reflux
- Hearing aids, glasses, or other sensory support
- Physical, occupational, feeding, and communication therapy
- Equipment for positioning, mobility, breathing, or feeding
- Coordinated paediatric, genetics, cardiology, neurology, and palliative care

Complications

Breathing pauses, airway problems, aspiration, and chest infections can occur. Aspiration means milk, food, saliva, or stomach contents enter the airway.

Feeding problems may cause dehydration, poor growth, or choking. Reflux can cause discomfort and worsen breathing or feeding.

Heart defects may lead to low oxygen, poor circulation, or heart failure. Heart failure means the heart cannot pump effectively enough for the body's needs; it does not mean the heart has stopped.

Other complications include seizures, hearing or sight loss, kidney problems, constipation, scoliosis, joint stiffness, and pressure injuries from limited movement.

Children who survive beyond infancy usually have significant developmental disability. They may communicate through expressions, sounds, movements, eye gaze, or assistive devices rather than speech.

Prevention

D trisomy syndrome usually cannot be prevented. Most cases result from a random chromosome-separation error, not diet, exercise, stress, medicines taken as directed, or something a parent did.

Folic acid before and during early pregnancy helps prevent certain brain and spinal cord defects, but it does not prevent trisomy 13.

Chromosome testing after a diagnosis can help estimate the chance in a future pregnancy. If a translocation is found, parental testing may show whether either parent carries a balanced rearrangement.

People planning another pregnancy may discuss prenatal screening, diagnostic testing, donor eggs or sperm, or in vitro fertilisation with chromosome testing of embryos. These are personal reproductive options, not guaranteed prevention.

Living with it and what to expect

The course is often uncertain. Ultrasound findings and chromosome results cannot show exactly how long a child will live or which abilities they will develop.

Many affected pregnancies do not reach birth. Many live-born babies die during the first weeks or months, often because of breathing problems, heart defects, infections, or several complications together. Some children, including some with full trisomy 13, survive for years.

Children who live longer usually need continuing help with feeding, movement, communication, and daily care. Development is much slower than usual, but families may recognise individual responses to voices, touch, music, routines, and familiar people.

Periods of stability may be followed by illness or new feeding, breathing, or seizure problems. Regular reviews can monitor growth, comfort, hearing, vision, heart function, kidney health, mobility, and equipment needs.

Home nursing, respite care, therapies, social work, disability support, and palliative care may reduce practical and emotional strain. Written plans for seizures, breathing problems, hospital care, and emergency decisions can help families and clinicians respond consistently.

When to get medical help

Seek emergency help if a baby or child has:

Arrange prompt medical assessment for new or worsening breathing trouble, repeated vomiting, a sudden drop in feeding, fewer wet nappies, fever or other signs of infection, increased sleepiness, or a clear change in seizure pattern. A displaced feeding or breathing tube also needs timely advice.

Routine follow-up should review growth, feeding, development, hearing, vision, heart and kidney problems, movement, comfort, and equipment. Planned reviews do not replace urgent care for sudden changes.

- Severe difficulty breathing, gasping, or long pauses in breathing
- Blue, grey, or very pale skin that does not quickly improve
- Choking with an inability to breathe or make sounds
- A seizure lasting longer than the child's emergency plan allows
- Unresponsiveness or unusual difficulty waking
- Cold, mottled skin with marked weakness or other signs of severely reduced circulation

Common questions

Is D trisomy syndrome the same as trisomy 13?

Usually, yes. It is an older term commonly used for trisomy 13. Because the D group also included chromosomes 14 and 15, check an old diagnosis against the original chromosome report when possible.

Did something I did cause this?

Usually not. Most cases result from a random error when chromosomes separate in an egg or sperm. Everyday activity, stress, or food eaten during pregnancy does not cause it.

Can a prenatal blood test tell me for certain?

No. Cell-free DNA and other blood tests are screening tests. Confirmation requires chromosome testing from chorionic villus sampling, amniocentesis, or testing after birth.

How long can a baby with D trisomy syndrome live?

There is no exact answer. Many affected pregnancies end before birth, and many live-born babies die during infancy. Some children live for several years or longer.

Can a child with this condition learn or communicate?

Most children who survive have major developmental disability. Some communicate comfort, distress, preferences, or enjoyment through sounds, expressions, movement, eye gaze, or communication aids.

Can it happen again in another pregnancy?

It can, but the chance varies depending on whether the condition resulted from full trisomy, mosaicism, or a translocation. The chromosome report and, when needed, parental testing provide the most useful estimate.

Sources

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