CONDITION

Trisomy 18 syndrome

Trisomy 18 is a chromosome condition that affects growth, development, and many organs.

WHAT IT IS
Chromosome condition caused by an extra chromosome 18

BODY SYSTEM
Whole body, especially heart, brain, kidneys, and growth

MAIN SYMPTOMS
Poor growth, feeding problems, heart defects, developmental disability

WHAT CAUSES IT
Usually a random chromosome-separation error

HOW IT IS DIAGNOSED
Chorionic villus sampling, amniocentesis, or postnatal chromosome testing

MAIN TREATMENT
Supportive, symptom-directed care and developmental therapies

CAN IT BE CURED?
No, chromosome change remains lifelong

IS IT AN EMERGENCY?
Sometimes, with severe breathing problems or unresponsiveness

LONG-TERM OUTLOOK
Serious lifelong disability with high early mortality

Overview

Trisomy 18 is a chromosome condition. It is also called Edwards syndrome or Edward's syndrome.

Chromosomes are packages of genetic material inside most cells. People usually have two copies of chromosome 18. In trisomy 18, some or all cells have a third copy or extra piece of this chromosome.

The extra genetic material changes how the body develops. It can affect growth, the brain, heart, kidneys, digestive system, hands, feet, and other parts of the body. Most children have major developmental disabilities and complex medical needs.

Many affected pregnancies end in miscarriage or stillbirth. Among babies born alive, many die

during the first days, weeks, or months of life. Some children live beyond their first birthday, and a smaller number live for years. Survival and medical needs cannot be predicted exactly from the diagnosis alone.

Symptoms

Signs may first appear during pregnancy. An ultrasound may show slow growth, too much amniotic fluid, a heart difference, or unusual development of the brain, hands, feet, or other organs. These findings can also have other causes.

Possible signs after birth include:

Not every child has every feature. Children with mosaic or partial trisomy 18 may have fewer or less severe effects, but this is not guaranteed.

Feeding can take a long time because sucking and swallowing may be difficult. Some children have reflux, where stomach contents move back toward the throat. Others may breathe pauses, need oxygen, or have repeated chest infections.

Children who live longer usually develop more slowly than other children. Some learn to smile, recognize familiar people, communicate preferences, sit with support, or use adapted equipment. Their abilities differ widely.

- Low birth weight and poor growth
- A small head or lower jaw
- Clenched hands with overlapping fingers
- Feet with rounded soles, sometimes called "rocker-bottom" feet
- Heart, kidney, or digestive system differences
- Weak muscle tone, breathing problems, or difficulty feeding
- Delayed development and significant intellectual disability

Causes and risk factors

Trisomy 18 usually begins with a random error when an egg or sperm cell forms. Chromosome 18 does not separate as expected, leaving an extra copy after fertilization. This is called nondisjunction .

This random event is not caused by something a parent ate, did, or failed to do. It is not an infection, and it cannot be caught from another person.

There are three main chromosome patterns:

The chance of trisomy 18 increases with the age of the egg, so it is higher in pregnancies at older maternal ages. However, it can occur at any reproductive age.

Most cases are not inherited. Rarely, partial trisomy 18 results from a parent having a balanced chromosome rearrangement. The parent may be healthy because no important genetic material is missing or extra, but the rearrangement can affect a pregnancy.

- Full trisomy 18: Nearly every cell has three complete copies of chromosome 18. This is the most common pattern and usually causes severe effects.
- Mosaic trisomy 18: Only some cells have an extra chromosome 18. The effects depend partly on which cells and organs are involved.
- Partial trisomy 18: Cells have an extra section of chromosome 18 rather than a whole extra chromosome.

Diagnosis

Prenatal screening estimates the chance that a pregnancy has trisomy 18. Screening may use ultrasound, blood tests measuring pregnancy-related substances, or cell-free DNA testing. Cell-free DNA examines small fragments of placental DNA in the pregnant person's blood.

A high-chance screening result does not confirm trisomy 18. False-positive and false-negative results can occur.

A prenatal diagnosis requires testing cells from the pregnancy. Chorionic villus sampling , or CVS, takes a small sample from the placenta. Amniocentesis tests cells from the fluid surrounding the fetus. A laboratory then examines the chromosomes using a karyotype or another chromosome test.

Because CVS tests placental tissue, uncommon results may require clarification with amniocentesis. This can happen when the placenta contains cells with trisomy 18 but fetal cells do not, or when mosaicism is suspected.

After birth, trisomy 18 is confirmed with a blood chromosome test. Testing can identify whether the pattern is full, mosaic, or partial. Parents may also be offered chromosome testing if a structural rearrangement is found.

Genetic counselling can help you understand what the result means, what it cannot predict, and whether there may be implications for relatives or future pregnancies.

Treatment and management

There is no treatment that removes the extra chromosome. Care focuses on comfort, health problems, development, and the goals of the child and family.

After a prenatal diagnosis, a maternal-fetal medicine team can assess fetal growth and organ development. Families may discuss pregnancy options, delivery planning, newborn treatment, and comfort-focused care. Available options are also affected by local law and access to services.

After birth, decisions should be individualized. Trisomy 18 alone does not show exactly which treatments will help a particular child. Care may include:

Palliative care does not necessarily mean stopping other treatment. It can be provided alongside medical or surgical care and can help with comfort, decision-making, and support at home.

Some families choose mainly comfort-focused care. Others consider intensive care, surgery, or longer-term medical support. The likely benefits, burdens, recovery, and effect on the child's comfort should be discussed for each proposed intervention.

- Support with breast, bottle, tube, or gastrostomy feeding
- Oxygen or other breathing support
- Treatment of infections, seizures, reflux, constipation, or pain
- Assessment and selected treatment of heart or digestive system differences
- Physical, occupational, speech, hearing, and vision support
- Mobility, seating, and communication equipment
- Palliative care for symptom relief and family support

Complications

Congenital heart defects are common and can affect blood flow, breathing, feeding, and growth. Some children develop heart failure, meaning the heart cannot pump blood effectively enough for the body's needs.

Swallowing difficulties can allow milk, food, or saliva to enter the airway. This is called aspiration and may lead to coughing, low oxygen levels, or pneumonia. Tube feeding can improve nutrition for some children, but it does not remove every aspiration risk.

Other possible complications include breathing pauses, repeated infections, seizures, hearing or vision loss, kidney problems, reflux, constipation, and curvature of the spine. Limited

movement may contribute to stiff joints or pressure injuries.

Medical procedures can be harder on a child with several organ problems. Before an operation, the team may assess breathing, heart function, nutrition, and the expected effect on comfort and daily life.

Prevention

Trisomy 18 usually cannot be prevented. It most often results from a random chromosome-separation error, not from a parent's behaviour or health choices.

Prenatal screening does not prevent the condition. It can identify a higher chance of trisomy 18 and allow diagnostic testing if wanted.

After an affected pregnancy, genetic counselling may help explain the chance in a future pregnancy. That chance is influenced by parental age and by whether testing found full, mosaic, or partial trisomy 18. If a chromosome rearrangement is present, parental chromosome testing may provide more specific information.

Options for future pregnancies can include prenatal screening, diagnostic testing, or assisted reproduction with embryo testing. These are personal choices rather than required steps.

Living with it and what to expect

The early course is often medically fragile. A baby may need help with breathing, maintaining body temperature, or feeding. Families may need to make difficult decisions while learning what the child's specific health problems are.

Many babies with trisomy 18 die in infancy, including some receiving intensive treatment. Others become medically stable enough to live at home. Children who survive longer usually need ongoing help with feeding, movement, communication, and personal care.

Development generally continues at an individual pace rather than stopping completely. Children may form strong relationships, respond to voices and touch, show enjoyment, and develop ways to communicate. Significant developmental disability and physical dependence are expected, especially with full trisomy 18.

Health needs can change over months and years. Feeding, breathing, seizures, spinal curvature, and equipment needs may require regular review. Routine follow-up may involve primary care, cardiology, nutrition, therapy, hearing, vision, dentistry, and palliative care.

Home nursing, respite care, early-intervention services, school supports, and parent

communities can reduce practical strain. Plans for emergencies and end-of-life care can also give families more control. These plans can be reviewed whenever the child's condition or the family's goals change.

When to get medical help

Arrange routine follow-up if feeding is becoming harder, weight gain slows, constipation or reflux persists, or equipment no longer fits. New changes in movement, hearing, vision, sleep, or alertness also deserve assessment.

Seek prompt medical advice if your child has more coughing or choking during feeds, fewer wet diapers, repeated vomiting, a fever, increasing sleepiness, new swelling, or a noticeable change in breathing.

Call emergency services for:

If your child has an individualized emergency or comfort-care plan, keep it available and follow the instructions in that plan.

- Severe trouble breathing, gasping, or long breathing pauses
- Blue, grey, or unusually pale lips or skin
- Unresponsiveness or difficulty waking
- A first seizure, a seizure lasting more than five minutes, or repeated seizures without recovery
- Sudden collapse or another change covered by the child's emergency plan

Common questions

Is trisomy 18 always fatal?

No. Many affected pregnancies do not reach birth, and many liveborn babies die in infancy. However, some children live for years. A chromosome result cannot predict an exact lifespan for one child.

Can a prenatal screening test be wrong?

Yes. Screening estimates chance; it does not make a diagnosis. CVS or amniocentesis with chromosome testing is used to confirm a prenatal result. After birth, diagnosis is confirmed with a blood chromosome test.

Did I cause my baby's trisomy 18?

Almost certainly not. Most cases result from a random error as an egg or sperm forms. Ordinary activities, stress, foods, and routine medicines are not known to cause trisomy 18.

What is the difference between full, mosaic, and partial trisomy 18?

Full trisomy means nearly all cells have an extra complete chromosome 18. Mosaic trisomy affects only some cells. Partial trisomy involves an extra section of chromosome 18. These patterns can influence severity, but they do not predict every health need.

Can a child with trisomy 18 have surgery?

Sometimes. Surgery may be considered for selected heart, feeding, digestive, or other problems. Decisions are based on the child's health, the purpose and burden of treatment, the chance of benefit, and the family's goals.

Can people with trisomy 18 learn and communicate?

Yes, although development is usually significantly delayed. Some children communicate through facial expressions, sounds, signs, switches, or other adapted methods. Therapy and suitable equipment may help them take part in family, school, and community life.

Sources

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