CONDITION

Ellis-van creveld syndrome

Ellis-van Creveld syndrome is a rare inherited condition affecting bone growth, fingers, teeth, nails, and often the heart.

WHAT IT IS
Rare inherited condition affecting bone growth and several organs

BODY SYSTEM
Bones, teeth, nails, heart and circulation

MAIN SYMPTOMS
Short limbs, extra fingers, abnormal teeth, abnormal nails

WHAT CAUSES IT
Inherited EVC or EVC2 gene changes

HOW IT IS DIAGNOSED
Physical features, X-rays, and genetic testing

MAIN TREATMENT
Heart, breathing, dental, bone, and developmental care

CAN IT BE CURED?
No - genetic changes cannot be reversed

IS IT AN EMERGENCY?
Sometimes - severe breathing difficulty needs emergency care

LONG-TERM OUTLOOK
Many reach adulthood; heart and breathing problems shape outlook

Overview

Ellis-van Creveld syndrome is a rare genetic condition present from birth. It mainly affects how the bones, teeth, nails, and heart develop.

It is a type of skeletal dysplasia, meaning it changes bone and cartilage growth. People usually have short arms and legs, a narrow chest, and short adult height. Extra fingers are common, usually beside the little fingers. Teeth and nails may be small, missing, or shaped differently.

Many babies are also born with a structural heart problem, called a congenital heart defect. The severity of heart and breathing problems has a major effect on early health and long-term outlook.

Intelligence is usually not affected. However, a child may need support with movement,

feeding, speech, school access, or the emotional effects of visible physical differences.

The condition is lifelong. Treatment cannot reverse its genetic cause, but heart care, breathing support, dental treatment, surgery, rehabilitation, and practical support can improve health and independence.

Symptoms

Features vary between people, including within the same family. Some are detected during pregnancy or at birth, while dental, growth, and joint concerns may become clearer later.

Common features include:

The upper lip may be joined to the gum in places, and the gums may have notches or extra tissue. These differences can affect feeding, speech, dental alignment, and denture fitting.

Heart defects are common. A baby with a significant defect may breathe quickly, tire or sweat while feeding, gain weight slowly, or develop a blue-gray color around the lips.

A narrow chest can limit lung expansion. Some newborns have serious breathing difficulty; others have milder problems or repeated chest infections.

Less common features involve the spine, hips, kidneys, urinary system, or genitals. Not everyone has every feature.

- Short arms and legs, especially the forearms and lower legs
- Short stature continuing into adulthood
- An extra finger beside each little finger
- Small hands and feet with short fingers or toes
- A long, narrow chest caused by short ribs
- Knees that angle inward, or knock knees
- Small, thin, ridged, or missing nails
- Missing, unusually shaped, or widely spaced teeth
- Teeth present at birth or appearing unusually early

Causes and risk factors

Ellis-van Creveld syndrome is usually caused by harmful changes in the EVC or EVC2 gene. These genes help control signals that guide the development of bones, teeth, and other

tissues before birth.

The condition has an autosomal recessive inheritance pattern. A child usually receives one changed copy of the same gene from each biological parent. Parents carrying one changed copy generally do not have the syndrome.

When both parents carry a disease-causing change in the same gene, each pregnancy has:

The chances are the same in every pregnancy, regardless of what happened previously.

A family history increases the chance that both parents carry the same change. The chance may also be higher when biological parents are closely related because they are more likely to share rare variants. However, many families have no known history before a child is diagnosed.

Nothing a parent did during pregnancy caused the condition.

- A 1 in 4 chance of a child having the condition
- A 1 in 2 chance of a child being an unaffected carrier
- A 1 in 4 chance of a child having neither familial gene change

Diagnosis

The condition may be suspected before birth if an ultrasound shows short limbs, extra fingers, a narrow chest, or a heart defect. Because these findings occur in other genetic conditions, ultrasound alone may not confirm the diagnosis.

After birth, assessment includes a physical examination and review of growth, limbs, nails, teeth, chest, and heart. X-rays may show characteristic changes in the hands, legs, ribs, pelvis, and other bones.

Genetic testing can confirm the diagnosis by finding disease-causing changes in both copies of EVC or EVC2 . A panel may examine several genes linked to skeletal conditions. If two changes are not found, specialists may review the clinical evidence and consider broader testing.

Assessments for related health problems may include:

Testing biological parents can show whether they carry the identified variants and clarify the chance of recurrence in another pregnancy.

- An echocardiogram to examine the heart
- Oxygen measurements and breathing assessments
- Dental and jaw imaging
- Kidney or urinary tract ultrasound when indicated

- Growth, feeding, movement, and developmental assessments

Treatment and management

There is no treatment that corrects the underlying genetic changes. Care addresses each person's heart, breathing, bones, teeth, development, and daily needs.

A newborn with a small chest or lung problems may need oxygen, breathing support, feeding support, or treatment for respiratory infections.

A pediatric cardiology team monitors congenital heart defects. Some defects require surgery; others can be observed or managed with medicines. Long-term heart follow-up may be needed even after repair.

Dental care often begins early and may include protecting enamel, treating decay, managing teeth present at birth, orthodontics, and replacing missing teeth. Dentures, bridges, or implants may be considered at suitable ages.

Orthopedic care addresses bone and joint concerns. Physical therapy can support strength, balance, and mobility. Braces or surgery may be considered if leg alignment, joint instability, pain, or walking problems affect daily life. Not every bone difference needs surgery.

Occupational therapy can help with hand function, self-care, writing, and adapted equipment. Speech or feeding therapy may help when oral, dental, breathing, or feeding differences cause difficulty.

Counseling, peer support, school accommodations, and practical planning can help with accessibility barriers, unwanted attention, and body-image concerns.

Complications

The most serious complications usually involve the heart and lungs. A major heart defect can cause heart failure, abnormal rhythms, or increased pressure in the lung arteries. A narrow chest may restrict lung growth and make respiratory infections harder to tolerate.

Possible complications include:

Some newborns do not survive severe heart and lung complications. For those who pass through infancy without major respiratory problems, the outlook may be much better. Adults may still need care for heart defects, dental problems, and mobility.

Short stature, extra fingers, and many nail or dental differences do not directly threaten health, but they can affect daily activities, confidence, and social experiences.

- Severe breathing problems during infancy
- Repeated respiratory infections
- Feeding difficulty and slow weight gain
- Heart failure or lasting effects of congenital heart disease
- Pain or reduced mobility from leg and joint alignment
- Tooth decay, chewing problems, and dental infection
- Speech difficulties related to teeth or oral structure

Prevention

Ellis-van Creveld syndrome cannot be prevented through diet, exercise, medicines, or pregnancy behavior. It results from inherited gene changes present from conception.

Genetic counseling can help families understand inheritance and testing options. If familial variants are known, carrier testing may be available for adult relatives. Prenatal diagnostic testing or testing embryos created through in vitro fertilization may also be possible.

These options provide information or reproductive choices; they do not prevent or cure the condition in someone who already has it.

Living with it and what to expect

Short stature and the basic bone-growth pattern remain throughout life, although surgery or therapy may improve alignment, movement, comfort, and function. Dental needs often continue for many years as childhood teeth are lost and adult teeth develop.

The first months may be the most medically intensive for babies with a narrow chest or serious heart defect. Some need prolonged hospital care or surgery, while others have stable breathing and mainly need planned specialist appointments.

Children commonly have regular heart, orthopedic, dental, and growth reviews. School adjustments may include accessible furniture, extra time between classes, handwriting support, or protection from unnecessary physical strain.

Intelligence is usually typical. Many people attend mainstream education, work, form relationships, and live independently or with limited practical support. Adaptations may be useful at home, in vehicles, and at work.

Because the condition is rare, long-term information is limited. Heart health, lung function,

mobility, dental health, and early complications influence each person's course. Regular follow-up can identify treatable problems but cannot guarantee a particular outcome.

When to get medical help

Call emergency services for:

A baby also needs urgent assessment for repeated pauses in breathing, marked chest pulling with each breath, or being too breathless to feed.

Seek prompt medical advice for new or worsening rapid breathing, poor feeding, sweating during feeds, unusual tiredness, swelling, reduced urine, fever with breathing symptoms, or poor weight gain. These may indicate a heart, lung, or infection-related problem.

Contact a dentist promptly for facial swelling, severe tooth pain, pus, fever with dental symptoms, or difficulty swallowing.

Routine follow-up is appropriate for stable concerns such as growth, leg alignment, mobility, dental development, school access, or equipment needs. Do not automatically assume new symptoms are part of Ellis-van Creveld syndrome.

- Severe or rapidly worsening breathing difficulty
- Blue-gray lips, tongue, or skin
- Collapse, loss of consciousness, or difficulty waking
- Severe chest pain or sudden marked weakness

Common questions

How tall do people with Ellis-van Creveld syndrome grow?

Most have marked short stature because long-bone growth is affected. Final height varies and may be influenced by family height, bone changes, nutrition, major illness, and complications.

Does Ellis-van Creveld syndrome affect intelligence?

Intelligence is usually not directly affected. Some children may reach milestones later because of serious illness, hospital stays, limited movement, or speech and learning-access needs.

Can someone with the condition walk?

Many people learn to walk. Knock knees, short limbs, joint differences, pain, or surgery may affect timing, balance, speed, and endurance. Physical therapy, mobility aids, or orthopedic treatment can help.

Can extra fingers be removed?

Sometimes. Surgery may be considered if an extra finger affects function or causes practical concerns. Timing and likely benefit depend on its bones, joints, nerves, and blood supply. Removal is not always medically necessary.

Can people with Ellis-van Creveld syndrome have children?

Some adults can have biological children. A person with the condition usually passes one changed gene copy to every child. The chance of a child having the syndrome depends on whether the other biological parent carries a harmful change in the same gene. Genetic counseling and carrier testing can clarify this.

Is Ellis-van Creveld syndrome the same as dwarfism?

It is one cause of disproportionate short stature, sometimes described using the broad term dwarfism. The specific name also reflects its characteristic finger, nail, tooth, chest, and heart differences.

Sources

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