CONDITION

Cleidocranial dysostosis

Learn how cleidocranial dysostosis affects the skull, collarbones, teeth, and other bones, including its genetic cause, diagnosis, treatment, and follow-up care.

WHAT IT IS
Rare genetic bone development condition

BODY SYSTEM
Bones and teeth

MAIN SYMPTOMS
Delayed tooth eruption, extra teeth, open fontanelles, underdeveloped collarbones

WHAT CAUSES IT
Inherited or new RUNX2 gene change

HOW IT IS DIAGNOSED
X-rays and RUNX2 genetic testing

MAIN TREATMENT
Coordinated dental and orthodontic care

CAN IT BE CURED?
No, individual effects can be managed

IS IT AN EMERGENCY?
Sometimes, with severe trouble breathing

LONG-TERM OUTLOOK
Many people lead active, independent lives

Overview

Cleidocranial dysostosis is a rare genetic condition that mainly affects the development of the skull, collarbones, and teeth. Current medical literature usually calls it cleidocranial dysplasia , or CCD. The condition is present from birth, although it may not be recognized until delayed tooth eruption or another skeletal feature becomes noticeable.

"Cleido" refers to the clavicles, or collarbones. "Cranial" refers to the skull. In CCD, the collarbones may be short, partly formed, or occasionally absent. The joints between skull bones may also close later than usual. Dental development is often affected, including delayed loss of baby teeth, delayed appearance of adult teeth, and extra teeth.

The features and their severity vary widely. Some people have only mild dental or skeletal differences. Others need coordinated dental, hearing, respiratory, or orthopedic care.

Intelligence is usually unaffected, and many people with CCD lead active, independent lives.

CCD does not have a single cure, but its individual effects can often be managed. Early dental planning is especially important because treatment may take place over several years.

Symptoms

CCD can affect several areas of the body. A person may have only some of the following features.

Skull and facial features

The soft spaces between a baby's skull bones, called fontanelles, may remain open longer than expected. The skull sutures-the joints where skull bones meet-may also close late. Small extra bones can form within these sutures; these are called Wormian bones.

Other possible features include:

These differences do not usually affect brain development. However, the shape of the jaw and palate can contribute to dental crowding, speech differences, snoring, or sleep-related breathing problems.

Collarbone and skeletal features

One or both collarbones may be underdeveloped. Some people can bring their shoulders unusually close together in front of the chest because the collarbones do not hold the shoulders as far apart as usual.

People with CCD may also have:

Bone density may be lower in some people, which can increase the risk of fractures later in life. Not everyone with CCD develops weak bones.

Teeth and mouth

Dental findings are among the most common and medically important features. Baby teeth may remain in place longer than expected. Adult teeth may develop normally but remain trapped within the jaw instead of coming through the gums.

Extra adult teeth, called supernumerary teeth, are common. These extra teeth can block the normal teeth from erupting. Crowding, an abnormal bite, cavities, and gum problems may follow.

Ear, sinus, and breathing symptoms

The shape of the face and skull can affect drainage and ventilation around the ears and sinuses. Some children experience repeated middle-ear infections, sinus infections, or

temporary hearing loss caused by fluid behind the eardrum.

A narrow upper airway or dental and jaw differences may contribute to snoring. Pauses in breathing during sleep, restless sleep, daytime tiredness, or behavior changes can be signs of obstructive sleep apnea and should be discussed with a clinician.

- A broad or prominent forehead
- A skull that appears shorter from front to back
- A flat or underdeveloped midface
- A low or broad bridge of the nose
- A high or narrow roof of the mouth
- A lower jaw that appears prominent compared with the midface
- Shorter-than-average height
- Narrow, sloping shoulders
- Short or tapered fingers
- Broad thumbs
- Flat feet or knees that angle inward
- Curvature of the spine, including scoliosis
- Differences in the pelvis or hip joints

Causes and risk factors

CCD is usually caused by a disease-causing change in the RUNX2 gene. This gene provides instructions for a protein that helps immature cells develop into bone-forming cells. It is also important for tooth development.

The condition follows an autosomal dominant inheritance pattern. This means a person generally needs a change in only one copy of the gene to have CCD. An affected parent can pass the altered gene to a child, but the severity in the child cannot be predicted from the parent's features.

Sometimes the gene change is new in the affected person, rather than inherited from either parent. This is called a de novo variant. A person with a new variant can still pass it to their children.

The main risk factor is having a parent or close family member with CCD or a known RUNX2 variant. The condition is not caused by something a parent did during pregnancy, and it is not

caused by diet, exercise, or other lifestyle choices.

A genetic counselor can explain inheritance, family testing, and reproductive options in a way that reflects a particular family's results and goals.

Diagnosis

Diagnosis is based on a combination of medical history, physical examination, dental findings, and imaging. A clinician may suspect CCD when a person has delayed tooth eruption together with characteristic skull or collarbone findings.

X-rays may show:

Dental panoramic X-rays can help an oral health team see teeth that are still inside the jaws. Because repeated imaging involves radiation, clinicians plan studies according to the information needed for treatment.

Genetic testing can look for a disease-causing RUNX2 variant. Testing may confirm the diagnosis, clarify risk for relatives, or support family planning. A negative genetic test does not always rule out CCD if the clinical and X-ray findings are strongly suggestive.

Other conditions can cause delayed skull development, short stature, or dental abnormalities. A clinician may consider these possibilities before confirming the diagnosis. Additional assessment may include hearing tests, evaluation of breathing during sleep, or orthopedic imaging when symptoms suggest a need.

- Short, partly formed, or absent collarbones
- Delayed closure of skull sutures and fontanelles
- Extra bones within the skull sutures
- Adult teeth that have not erupted
- Extra teeth within the jaws
- Characteristic changes in the pelvis, hands, or other bones

Treatment and management

Care is tailored to the person's age, symptoms, and priorities. A coordinated team may include a primary care clinician, dentist, orthodontist, oral and maxillofacial surgeon, geneticist, ear-nose-and-throat specialist, audiologist, and orthopedic specialist.

Dental treatment

Dental management often begins in childhood and continues into adulthood. The team first maps the location of baby teeth, adult teeth, and extra teeth. Treatment may involve removing selected retained baby teeth or extra teeth that are blocking eruption.

Some adult teeth may then appear on their own. Others may need surgical exposure followed by orthodontic traction, in which braces gradually guide the tooth into place. The timing of these steps matters because jaw growth and tooth development continue through childhood and adolescence.

When natural teeth cannot be brought into a useful position, options may include bridges, removable dental appliances, or dental implants after growth is sufficiently complete. The best approach depends on bone structure, tooth position, oral health, and personal preferences.

Regular brushing with fluoride toothpaste, cleaning between teeth, professional dental care, and attention to diet can reduce cavities and gum disease during lengthy orthodontic treatment.

Bone and joint care

Many skeletal differences do not require treatment unless they cause pain or limit movement. Physical therapy may help with strength, posture, mobility, or joint symptoms. Scoliosis, hip problems, repeated fractures, or persistent pain may require orthopedic assessment.

A clinician may assess bone health when there are fractures or other risk factors for low bone density. Nutrition, weight-bearing activity, and treatment of any identified vitamin or hormonal problem may form part of a broader bone-health plan.

Hearing, speech, and breathing care

Repeated ear infections or concerns about hearing should prompt hearing assessment. Treatment depends on the cause and may include management by an ear-nose-and-throat specialist.

Speech therapy can help when hearing loss, palate shape, or dental position affects speech. Snoring with breathing pauses or daytime symptoms may lead to a sleep study, which records breathing and oxygen levels during sleep.

Surgery and procedural planning

Surgery is not routinely needed for an open fontanelle or underdeveloped collarbones when these findings are not causing problems. Procedures may be considered for specific functional or protective reasons.

Before anesthesia or surgery, the care team should know about the diagnosis. Dental abnormalities, facial structure, hearing needs, and possible neck or spine differences can affect planning.

Pelvic differences may make vaginal delivery more difficult for some pregnant people with CCD. Prenatal care should include early discussion of delivery planning with the obstetric team. The diagnosis alone does not determine how a baby must be delivered.

Complications

Potential complications depend on which parts of the body are affected. Dental crowding and unerupted teeth can contribute to cavities, gum disease, jaw cysts, difficulty chewing, or concerns about appearance. Long treatment periods can also affect school, work, and emotional well-being.

Repeated ear infections may cause temporary or persistent hearing problems. Hearing loss can affect speech and learning if it is not recognized. Sinus problems and obstructive sleep apnea can interfere with sleep and daytime functioning.

Spinal curvature, hip abnormalities, flat feet, or altered joint mechanics may cause pain or reduced mobility. Lower bone density may increase fracture risk in some people.

Routine symptoms such as slowly changing dental alignment, mild joint discomfort, or concerns about growth can usually be discussed at a scheduled appointment.

Seek prompt medical or dental care for facial swelling, fever with severe tooth pain, drainage from the gums, sudden hearing loss, a suspected fracture, or rapidly worsening pain.

Call emergency services for severe trouble breathing, blue or gray lips, loss of consciousness, or new weakness or loss of sensation after a head, neck, or back injury.

Follow up

Follow-up needs change with age. Children benefit from coordinated dental and orthodontic planning before extensive crowding or impaction makes treatment more difficult. The schedule for examinations and X-rays should be set by the dental team.

Routine care may include:

Adults should continue dental follow-up even after orthodontic treatment has ended. New pain, changes in bite, loose teeth, hearing problems, or sleep-related breathing symptoms should be evaluated rather than assumed to be part of CCD.

Keeping copies of genetic test results, dental images, and major treatment records can make it easier for new clinicians to understand prior care.

- Dental and orthodontic review
- Monitoring of growth, posture, spine, hips, and walking
- Hearing testing when age-appropriate or when concerns arise
- Review of ear infections, sinus symptoms, snoring, and sleep quality
- Assessment of pain, fractures, physical activity, and bone-health risks
- Support for speech, school participation, and emotional well-being
- Genetic counseling when family planning or testing is being considered

Sources

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