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Cleidocranial dysplasia

Description

Cleidocranial dysplasia is a condition that primarily affects development of the bones and teeth. Signs and symptoms of cleidocranial dysplasia can vary widely in severity, even within the same family.

Individuals with cleidocranial dysplasia usually have underdeveloped or absent collarbones, also called clavicles ("cleido-" in the condition name refers to these bones). As a result, their shoulders are narrow and sloping, can be brought unusually close together in front of the body, and in some cases can be made to meet in the middle of the body. Delayed maturation of the skull (cranium) is also characteristic of this condition, including delayed closing of the growth lines where the bones of the skull meet (sutures) and larger than normal spaces (fontanelles) between the skull bones that are noticeable as "soft spots" on the heads of infants. The fontanelles normally close in early childhood, but they may remain open throughout life in people with this disorder. Some individuals with cleidocranial dysplasia have extra pieces of bone called Wormian bones within the sutures.

Affected individuals are often shorter than other members of their family at the same age. Many also have short, tapered fingers and broad thumbs; flat feet; knock knees; short shoulder blades (scapulae); and an abnormal curvature of the spine (scoliosis). Typical facial features include a wide, short skull (brachycephaly); a prominent forehead; wide-set eyes (hypertelorism); a flat nose; and a small upper jaw.

Individuals with cleidocranial dysplasia often have decreased bone density (osteopenia) and may develop osteoporosis, a condition that makes bones progressively more brittle and prone to fracture, at a relatively early age. Women with cleidocranial dysplasia have an increased risk of requiring a cesarean section when delivering a baby, due to a narrow pelvis preventing passage of the infant's head.

Dental abnormalities are very common in cleidocranial dysplasia and can include delayed loss of the primary (baby) teeth; delayed appearance of the secondary (adult) teeth; unusually shaped, peg-like teeth; misalignment of the teeth and jaws (malocclusion); and extra teeth, sometimes accompanied by cysts in the gums.

In addition to skeletal and dental abnormalities, people with cleidocranial dysplasia may have hearing loss and are prone to sinus and ear infections. Some young children with this condition are mildly delayed in the development of motor skills such as crawling and walking, but intelligence is unaffected.

Frequency

Cleidocranial dysplasia occurs in approximately 1 per million individuals worldwide. It is likely underdiagnosed because

many affected individuals have mild signs and symptoms.

Causes

Cleidocranial dysplasia is usually caused by mutations in the *RUNX2* [<https://medlineplus.gov/genetics/gene/runx2/>] gene. This gene provides instructions for making a protein that is involved in the development and maintenance of teeth, bones, and cartilage. Cartilage is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone (a process called ossification), except for the cartilage that continues to cover and protect the ends of bones and is present in the nose, airways, and external ears.

The RUNX2 protein is a transcription factor, which means it attaches (binds) to specific regions of DNA and helps control the activity of particular genes. Researchers believe that the RUNX2 protein acts as a "master switch," regulating a number of other genes involved in the development of cells that build bones (osteoblasts) and in the development of teeth.

The *RUNX2* gene mutations that cause cleidocranial dysplasia reduce or eliminate the activity of the protein produced from one copy of the *RUNX2* gene in each cell, decreasing the total amount of functional RUNX2 protein. This shortage of functional RUNX2 protein interferes with the normal development of bones, cartilage, and teeth, resulting in the signs and symptoms of cleidocranial dysplasia. In rare cases, individuals with a deletion of genetic material that includes *RUNX2* and other nearby genes may experience additional features, such as developmental delay, resulting from the loss of these genes.

In about 30 percent of individuals with cleidocranial dysplasia, no mutation in the *RUNX2* gene has been found. The cause of the condition in these individuals is unknown.

Learn more about the gene associated with Cleidocranial dysplasia

RUNX2 [<https://medlineplus.gov/genetics/gene/runx2/>]

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Some affected individuals inherit the mutation from one affected parent. Often the parent is mildly affected, and in some cases had not previously been recognized as having the disorder. Other cases result from new mutations in the gene. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

- Cleidocranial dysostosis
- Dento-osseous dysplasia


- Marie-Sainton syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Cleidocranial dysostosis [<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0008928/>] 

Genetic and Rare Diseases Information Center

- Cleidocranial dysplasia [<https://rarediseases.info.nih.gov/diseases/6118/index>] 

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) [<https://rarediseases.org/>]


Clinical Trials

- ClinicalTrials.gov [[https://clinicaltrials.gov/search?cond=%22Cleidocranial dysplasia%22](https://clinicaltrials.gov/search?cond=%22Cleidocranial+dysplasia%22)] 

Catalog of Genes and Diseases from OMIM

- CLEIDOCRANIAL DYSPLASIA 1; CLCD1 [<https://omim.org/entry/119600>]

Scientific Articles on PubMed

- PubMed [<https://pubmed.ncbi.nlm.nih.gov/?term=%28Cleidocranial+Dysplasia%5BMAJR%5D%29+AND+%28cleidocranial+dysplasia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>] 

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MEDICAL ENCYCLOPEDIA

Cleidocranial dysostosis [<https://medlineplus.gov/ency/article/001589.htm>]

Genetics [<https://medlineplus.gov/ency/article/002048.htm>]

Understanding Genetics

What is the prognosis of a genetic condition? [<https://medlineplus.gov/genetics/understanding/consult/prognosis/>]

How can gene variants affect health and development?
[<https://medlineplus.gov/genetics/understanding/mutationsanddisorders/mutationscausedisease/>]

What does it mean if a disorder seems to run in my family?
[<https://medlineplus.gov/genetics/understanding/inheritance/runsinfamily/>]

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