Auriculo-condylar syndrome

Description

Auriculo-condylar syndrome is a condition that affects facial development, particularly development of the ears and lower jaw (mandible).

Most people with auriculo-condylar syndrome have malformed outer ears ("auriculo-" refers to the ears). A hallmark of this condition is an ear abnormality called a "question-mark ear," in which the ears have a distinctive question-mark shape caused by a split that separates the upper part of the ear from the earlobe. Other ear abnormalities that can occur in auriculo-condylar syndrome include cupped ears, ears with fewer folds and grooves than usual (described as "simple"), narrow ear canals, small skin tags in front of or behind the ears, and ears that are rotated backward. Some affected individuals also have hearing loss.

Abnormalities of the mandible are another characteristic feature of auriculo-condylar syndrome. These abnormalities often include an unusually small chin (micrognathia) and malfunction of the temporomandibular joint (TMJ), which connects the lower jaw to the skull. Problems with the TMJ affect how the upper and lower jaws fit together and can make it difficult to open and close the mouth. The term "condylar" in the name of the condition refers to the mandibular condyle, which is the upper portion of the mandible that forms part of the TMJ.

Other features of auriculo-condylar syndrome can include prominent cheeks, an unusually small mouth (microstomia), differences in the size and shape of facial structures between the right and left sides of the face (facial asymmetry), and an opening in the roof of the mouth (cleft palate). These features vary, even among affected members of the same family.

Frequency

Auriculo-condylar syndrome appears to be a rare disorder. More than two dozen affected individuals have been described in the medical literature.

Causes

Auriculo-condylar syndrome can be caused by mutations in either the *GNAI3* or *PLCB4* gene. These genes provide instructions for making proteins that are involved in chemical signaling within cells. They help transmit information from outside the cell to

inside the cell, which instructs the cell to grow, divide, or take on specialized functions.

Studies suggest that the proteins produced from the *GNAI3* and *PLCB4* genes contribute to the development of the first and second pharyngeal arches, which are structures in the embryo that ultimately develop into the jawbones, facial muscles, middle ear bones, ear canals, outer ears, and related tissues. Mutations in these genes alter the formation of the lower jaw: instead of developing normally, the lower jaw becomes shaped more like the smaller upper jaw (maxilla). This abnormal shape leads to micrognathia and problems with TMJ function. Researchers are working to determine how mutations in these genes lead to the other developmental abnormalities associated with auriculo-condylar syndrome.

In some people with the characteristic features of auriculo-condylar syndrome, a mutation in the *GNAI3* or *PLCB4* gene has not been found. The cause of the condition is unknown in these individuals.

Learn more about the genes associated with Auriculo-condylar syndrome

- GNAI3
- PLCB4

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is typically sufficient to cause the disorder. In some cases, an affected person inherits the mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

Some people who have one altered copy of the *GNAI3* or *PLCB4* gene have no features related to auriculo-condylar syndrome. (This situation is known as reduced penetrance.) It is unclear why some people with a mutated gene develop the condition and other people with a mutated gene do not.

Other Names for This Condition

- Auriculocondylar syndrome
- Dysgnathia complex
- Question-mark ear syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Auriculocondylar syndrome 1 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4551996/)

- Genetic Testing Registry: Auriculocondylar syndrome 2 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3553404/)

Genetic and Rare Diseases Information Center

- Auriculocondylar syndrome (https://rarediseases.info.nih.gov/diseases/9798/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- AURICULOCONDYLAR SYNDROME 1; ARCND1 (https://omim.org/entry/602483)
- AURICULOCONDYLAR SYNDROME 2A; ARCND2A (https://omim.org/entry/614669)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28auriculo-condylar+syndrome%5BTIAB%5D%29+OR+%28auriculocondylar+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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