Otospondylomegaepiphyseal dysplasia

Description

Otospondylomegaepiphyseal dysplasia (OSMED) is a condition characterized by skeletal abnormalities, distinctive facial features, and severe hearing loss. The term " otospondylomegaepiphyseal" refers to the parts of the body that this condition affects: the ears (oto-), the bones of the spine (spondylo-), and the ends (epiphyses) of long bones in the arms and legs. The features of this condition significantly overlap those of two similar conditions, Weissenbacher-Zweymüller syndrome and Stickler syndrome type III. All of these conditions are caused by mutations in the same gene, and in some cases, it can be difficult to tell the conditions apart. Some researchers believe they represent a single disorder with a range of signs and symptoms.

People with OSMED are often shorter than average because the long bones in their legs are unusually short. Other skeletal features include enlarged joints; short arms, hands, and fingers; and flattened bones of the spine (platyspondyly). People with the disorder often experience back and joint pain, limited joint movement, and arthritis that begins early in life.

Severe high-frequency hearing loss is common in people with OSMED. Typical facial features include protruding eyes; a flattened bridge of the nose; an upturned nose with a large, rounded tip; and a small lower jaw. Almost all affected infants are born with an opening in the roof of the mouth (a cleft palate).

Frequency

This condition is rare; its prevalence is unknown. Only a few families with OSMED worldwide have been described in the medical literature.

Causes

OSMED is caused by mutations in the *COL11A2* gene. This gene provides instructions for making one component of type XI collagen, which is a complex molecule that gives structure and strength to the connective tissues that support the body's joints and organs. Type XI collagen is found in cartilage, a tough but flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears. Type XI collagen is also part of the inner ear

and the nucleus pulposus, which is the center portion of the discs between vertebrae.

The *COL11A2* gene mutations that cause OSMED disrupt the production or assembly of type XI collagen molecules. The defective collagen weakens connective tissues in many parts of the body, including the long bones, spine, and inner ears, which impairs bone development and underlies the other signs and symptoms of this condition.

Learn more about the gene associated with Otospondylomegaepiphyseal dysplasia

- COL11A2

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Chondrodystrophy with sensorineural deafness
- Insley-Astley syndrome
- Mega-epiphyseal dwarfism
- Nance-Insley syndrome
- Nance-Sweeney chondrodysplasia
- OSMED
- Oto-spondylo-megaepiphyseal dysplasia

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Otospondylomegaepiphyseal dysplasia, autosomal dominant (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1848488/)
- Genetic Testing Registry: Otospondylomegaepiphyseal dysplasia, autosomal recessive (https://www.ncbi.nlm.nih.gov/gtr/conditions/C5551484/)

Genetic and Rare Diseases Information Center

- Otospondylomegaepiphyseal dysplasia (https://rarediseases.info.nih.gov/diseases/4130/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA, AUTOSOMAL RECESSIVE; OSMEDB (https://omim.org/entry/215150)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28otospondylomegaepiphyseal+dysplasia%5BTIAB%5D%29+OR+%28oto-spondylo-megaepiphyseal+dysplasia%5BTIAB%5D%29%29+OR+%28%28OSMED%5BTIAB%5D%29+AND+%28collagen*%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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