



atelosteogenesis type 2

Atelosteogenesis type 2 is a severe disorder of cartilage and bone development. Infants born with this condition have very short arms and legs, a narrow chest, and a prominent, rounded abdomen. This disorder is also characterized by an opening in the roof of the mouth (a cleft palate), distinctive facial features, an inward- and upward-turning foot (clubfoot), and unusually positioned thumbs (hitchhiker thumbs).

The signs and symptoms of atelosteogenesis type 2 are similar to those of another skeletal disorder called diastrophic dysplasia; however, atelosteogenesis type 2 is typically more severe. As a result of serious health problems, infants with this disorder are usually stillborn or die soon after birth from respiratory failure. Some infants, however, have lived for a short time with intensive medical support.

Frequency

Atelosteogenesis type 2 is an extremely rare genetic disorder; its incidence is unknown.

Genetic Changes

Atelosteogenesis type 2 is one of several skeletal disorders caused by mutations in the *SLC26A2* gene. This gene provides instructions for making a protein that is essential for the normal development of cartilage and for its conversion to bone. Cartilage is a tough, 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. Mutations in the *SLC26A2* gene disrupt the structure of developing cartilage, preventing bones from forming properly and resulting in the skeletal problems characteristic of atelosteogenesis type 2.

Inheritance Pattern

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

- AO2
- Atelosteogenesis de la Chapelle type
- atelosteogenesis, type 2

- De la Chapelle dysplasia
- McAlister dysplasia
- Neonatal osseous dysplasia 1

Diagnosis & Management

These resources address the diagnosis or management of atelosteogenesis type 2:

- GeneReview: Atelosteogenesis Type 2
<https://www.ncbi.nlm.nih.gov/books/NBK1317>
- Genetic Testing Registry: Atelosteogenesis type 2
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1850554/>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Bone Diseases
<https://medlineplus.gov/bonediseases.html>
- Health Topic: Dwarfism
<https://medlineplus.gov/dwarfism.html>
- Health Topic: Respiratory Failure
<https://medlineplus.gov/respiratoryfailure.html>

Genetic and Rare Diseases Information Center

- Atelosteogenesis type 2
<https://rarediseases.info.nih.gov/diseases/8329/atelosteogenesis-type-2>

Additional NIH Resources

- National Institute of Arthritis and Musculoskeletal and Skin Diseases: Questions and Answers about Heritable Disorders of Connective Tissue
https://www.niams.nih.gov/Health_Info/Connective_Tissue/

Educational Resources

- Disease InfoSearch: Atelosteogenesis Type 2
<http://www.diseaseinfosearch.org/Atelosteogenesis+Type+2/641>
- Orphanet: Atelosteogenesis type II
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=56304

Patient Support and Advocacy Resources

- European Skeletal Dysplasia Network
<http://www.esdn.org/eug/Home>
- Human Growth Foundation
<http://hgfound.org/>
- Little People of America
<http://www.lpaonline.org/>
- Resource list from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/dwarfism.html>
- The Compassionate Friends
<https://www.compassionatefriends.org/>

GeneReviews

- Atelosteogenesis Type 2
<https://www.ncbi.nlm.nih.gov/books/NBK1317>

Genetic Testing Registry

- Atelosteogenesis type 2
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1850554/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28type+2%5BTIAB%5D+AND+atelosteogenesis%5BTIAB%5D%29+OR+%28type+ii%5BTIAB%5D+AND+atelosteogenesis%5BTIAB%5D%29+OR+%28atelosteogenesis+de+la+chapelle+type%5BTIAB%5D%29+OR+%28de+la+chapelle+dysplasia%5BTIAB%5D%29+OR+%28mcalister+dysplasia%5BTIAB%5D%29+OR+%28neonatal+osseous+dysplasia+1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- ATELOSTEOGENESIS, TYPE II
<http://omim.org/entry/256050>

Sources for This Summary

- GeneReview: Atelosteogenesis Type 2
<https://www.ncbi.nlm.nih.gov/books/NBK1317>
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