



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.

Causes

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

Genetic Testing Information

- What is genetic testing?
[/primer/testing/geneticTesting](#)
- Genetic Testing Registry: Atelosteogenesis type 2
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1850554/>

Other Diagnosis and Management Resources

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

Additional Information & Resources

Health Information from 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: Heritable Disorders of Connective Tissue
<https://www.niams.nih.gov/health-topics/heritable-disorders-connective-tissue>

Educational Resources

- Orphanet: Atelosteogenesis type II
https://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/>
- Human Growth Foundation
<http://hgfound.org/>
- Little People of America
<https://www.lpaonline.org/>
- Little People UK
<https://littlepeopleuk.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/>

Clinical Information from GeneReviews

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

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>

Catalog of Genes and Diseases from OMIM

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

Sources for This Summary

- Bonafé L, Mittaz-Crettol L, Ballhausen D, Superti-Furga A. Atelosteogenesis Type 2. 2002 Aug 30 [updated 2014 Jan 23]. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Ledbetter N, Mefford HC, Smith RJH, Stephens K, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from <http://www.ncbi.nlm.nih.gov/books/NBK1317/>
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20301493>
- Hästbacka J, Superti-Furga A, Wilcox WR, Rimoin DL, Cohn DH, Lander ES. Atelosteogenesis type II is caused by mutations in the diastrophic dysplasia sulfate-transporter gene (DTDST): evidence for a phenotypic series involving three chondrodysplasias. *Am J Hum Genet.* 1996 Feb;58(2): 255-62.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/8571951>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1914552/>
- Newbury-Ecob R. Atelosteogenesis type 2. *J Med Genet.* 1998 Jan;35(1):49-53. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/9475095>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1051187/>
- Rossi A, Superti-Furga A. Mutations in the diastrophic dysplasia sulfate transporter (DTDST) gene (SLC26A2): 22 novel mutations, mutation review, associated skeletal phenotypes, and diagnostic relevance. *Hum Mutat.* 2001 Mar;17(3):159-71. Erratum in: *Hum Mutat* 2001;18(1):82.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11241838>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/atelosteogenesis-type-2>

Reviewed: February 2008

Published: May 14, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services