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 \textit{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 \textit{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

- \textit{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

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

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
  https://www.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+2%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+AND+english%5Bl%29+AND+human%5Bmh%5D+AND+%22last+3600+days%22+AND+publications

Catalog of Genes and Diseases from OMIM

• ATELOSTEOGENESIS, TYPE II
  http://omim.org/entry/256050
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301493

  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/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9475095
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