Spondyloperipheral dysplasia

Spondyloperipheral dysplasia is a disorder that impairs bone growth. This condition is characterized by flattened bones of the spine (platyspondyly) and unusually short fingers and toes (brachydactyly), with the exception of the first (big) toes. Other skeletal abnormalities associated with spondyloperipheral dysplasia include short stature, shortened long bones of the arms and legs, exaggerated curvature of the lower back (lordosis), and an inward- and upward-turning foot (clubfoot). Additionally, some affected individuals have nearsightedness (myopia), hearing loss, and intellectual disability.

Frequency

This condition is rare; only a few affected individuals have been reported worldwide.

Causes

Spondyloperipheral dysplasia is one of a spectrum of skeletal disorders caused by mutations in the \textit{COL2A1} gene. This gene provides instructions for making a protein that forms type II collagen. This type of collagen is found mostly in the clear gel that fills the eyeball (the vitreous) and in cartilage. 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. Type II collagen is essential for the normal development of bones and other connective tissues that form the body’s supportive framework.

Mutations in the \textit{COL2A1} gene interfere with the assembly of type II collagen molecules, reducing the amount of this type of collagen in the body. Instead of forming collagen molecules, the abnormal COL2A1 protein builds up in cartilage cells (chondrocytes). These changes disrupt the normal development of bones and other connective tissues, leading to the signs and symptoms of spondyloperipheral dysplasia.

Inheritance Pattern

This condition is probably inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- SPD
- spondyloperipheral dysplasia with short ulna
Diagnosis & Management

Genetic Testing Information
• What is genetic testing? /primer/testing/genetictesting

Other Diagnosis and Management Resources
• GeneReview: Type II Collagen Disorders Overview https://www.ncbi.nlm.nih.gov/books/NBK540447

Additional Information & Resources
Health Information from MedlinePlus
• Encyclopedia: Nearsightedness https://medlineplus.gov/ency/article/001023.htm
• Health Topic: Bone Diseases https://medlineplus.gov/bonediseases.html
• Health Topic: Connective Tissue Disorders https://medlineplus.gov/connectivetissuedisorders.html

Genetic and Rare Diseases Information Center
• Spondyloperipheral dysplasia https://rarediseases.info.nih.gov/diseases/4994/spondyloperipheral-dysplasia

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
• MalaCards: spondyloperipheral dysplasia https://www.malacards.org/card/spondyloperipheral_dysplasia
• Nemours Children's Health System: Skeletal Dysplasia https://www.nemours.org/services/skeletal-dysplasia.html?tab=about
• Orphanet: Spondyloperipheral dysplasia-short ulna syndrome https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1856
Patient Support and Advocacy Resources

- Human Growth Foundation
  https://www.hgfound.org/
- International Skeletal Dysplasia Registry, UCLA
  https://www.uclahealth.org/ortho/isdr
- Little People of America
  https://www.lpaonline.org/
- Little People UK
  https://littlepeopleuk.org/
- MAGIC Foundation
  https://www.magicfoundation.org/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/connect.html

Clinical Information from GeneReviews

- Type II Collagen Disorders Overview
  https://www.ncbi.nlm.nih.gov/books/NBK540447

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28spondyloperipheral+dysplasia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SPONDYLOPERIPHERAL DYSPLASIA
  http://omim.org/entry/271700

Medical Genetics Database from MedGen

- Spondyloperipheral dysplasia

Sources for This Summary
