Spondyloperipheral dysplasia

Description

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 *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 *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.

Learn more about the gene associated with Spondyloperipheral dysplasia

- COL2A1

Inheritance

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

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Spondyloperipheral dysplasia (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0796173/)

Genetic and Rare Diseases Information Center

- Spondyloperipheral dysplasia-short ulna syndrome (https://rarediseases.info.nih.gov/diseases/4994/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- SPONDYLOPERIPHERAL DYSPLASIA (https://omim.org/entry/271700)

Scientific Articles on PubMed

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

References

- Zabel B, Hilbert K, Stoss H, Superti-Furga A, Spranger J, Winterpacht A. Aspecific collagen type II gene (COL2A1) mutation presenting as spondyloperipheraldysplasia. Am J Med Genet. 1996 May 3;63(1):123-8. doi:10.1002/(SICI)1096-8628(19960503)63:13.0.CO;2-P. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/8723097)
- Zankl A, Zabel B, Hilbert K, Wildhardt G, Cuenot S, Xavier B, Ha-Vinh R,Bonafe L,

Spranger J, Superti-Furga A. Spondyloperipheral dysplasia is caused bytruncating mutations in the C-propeptide of COL2A1. Am J Med Genet A. 2004 Aug30;129A(2): 144-8. doi: 10.1002/ajmg.a.30222. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15316962)

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