Spondyloperipheral dysplasia

Description

Spondyloperipheral dysplasia is a disorder that impairs bone growth. The signs and symptoms of this condition can vary among affected individuals. People with spondyloperipheral dysplasia typically have short stature, with a short torso, short arms and legs, and short fingers and toes (brachydactyly). These parts of the body are not proportional to one another (disproportionate short stature) in people with this condition. Affected individuals also tend to have flattened bones of the spine (platyspondyly) and inward- and upward-turning feet (clubfoot). Some people with spondyloperipheral dysplasia may also experience nearsightedness (myopia) or hearing loss.

Frequency

Spondyloperipheral dysplasia is very rare, though the exact prevalence is unknown.

Causes

Spondyloperipheral dysplasia is caused by variants (also called mutations) in the *COL2A1* gene. This gene provides instructions for making a protein that forms type II collagen. This type of collagen is found 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. Type II collagen is essential for the normal growth and development of bones and other connective tissues.

The variants in the *COL2A1* gene that cause spondyloperipheral dysplasia typically affect a region of the protein called the C-propeptide domain. These variants cause cells to produce an abnormal version of the COL2A1 protein, which interferes with the production of mature type II collagen molecules. A reduction in the amount of mature type II collagen in the body disrupts the normal development of bones and other connective tissues, leading to the skeletal abnormalities seen in people with spondyloperipheral dysplasia.

Learn more about the gene associated with Spondyloperipheral dysplasia

- COL2A1

Inheritance

This condition is 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

- Spondyloperipheral dysplasia, COL2A1-related

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)

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