Spondyloepiphyseal dysplasia congenita

Description

Spondyloepiphyseal dysplasia congenita is an inherited bone growth disorder that results in short stature (dwarfism), skeletal abnormalities, and problems with vision and hearing. This condition affects the bones of the spine (spondylo-) and the ends (epiphyses) of long bones in the arms and legs. Congenita indicates that the condition is present from birth.

People with spondyloepiphyseal dysplasia congenita have short stature from birth, with a very short trunk and neck and shortened limbs. Their hands and feet, however, are usually average-sized. Adult height ranges from 3 feet to just over 4 feet. Abnormal curvature of the spine (kyphoscoliosis and lordosis) becomes more severe during childhood. Instability of the spinal bones (vertebrae) in the neck may increase the risk of spinal cord damage. Other skeletal features include flattened vertebrae (platyspondyly); an abnormality of the hip joint that causes the upper leg bones to turn inward (coxa vara) ; a foot deformity called a clubfoot; and a broad, barrel-shaped chest. Abnormal development of the chest can cause problems with breathing. Arthritis and decreased joint mobility often develop early in life.

People with spondyloepiphyseal dysplasia congenita have mild changes in their facial features. The cheekbones close to the nose may appear flattened. Some infants are born with an opening in the roof of the mouth (a cleft palate). Severe nearsightedness (high myopia) is common, as are other eye problems that can impair vision. About one quarter of people with this condition have hearing loss.

Frequency

This condition is rare; the exact incidence is unknown. More than 175 cases have been reported in the scientific literature.

Causes

Spondyloepiphyseal dysplasia congenita 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 cartilage and in the clear gel that fills the eyeball (the vitreous). The *COL2A1* gene is essential for the normal development of bones and other tissues that form the body's supportive

framework (connective tissues). Mutations in the *COL2A1* gene interfere with the assembly of type II collagen molecules, which prevents bones and other connective tissues from developing properly.

Learn more about the gene associated with Spondyloepiphyseal dysplasia congenita

- 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

- SED congenita
- SED, congenital type
- SEDc
- Spondyloepiphyseal dysplasia, congenital type

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Spondyloepiphyseal dysplasia congenita (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2745959/)

Genetic and Rare Diseases Information Center

- Spondyloepiphyseal dysplasia congenita (https://rarediseases.info.nih.gov/diseases/4987/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Spondyloepiphyseal dysplasia congenita%22)

Catalog of Genes and Diseases from OMIM

- SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA; SEDC (https://omim.org/entry/183900)

Scientific Articles on PubMed

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

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