Spondyloepiphyseal dysplasia congenita

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 \( \text{COL2A1} \) 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 \( \text{COL2A1} \) gene is essential for the normal development of bones and other tissues that form the body’s supportive framework (connective tissues). Mutations in the \( \text{COL2A1} \) gene interfere with the assembly of type II collagen molecules, which prevents bones and other connective tissues from developing properly.
Inheritance Pattern

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

Diagnosis & Management

Genetic Testing Information

• What is genetic testing? 
  [link]
• Genetic Testing Registry: Spondyloepiphyseal dysplasia congenita
  [link]
• Research Studies from ClinicalTrials.gov
  [link]

Other Diagnosis and Management Resources

• GeneReview: Type II Collagen Disorders Overview
  [link]
• MedlinePlus Encyclopedia: Clubfoot
  [link]
• MedlinePlus Encyclopedia: Lordosis
  [link]
• MedlinePlus Encyclopedia: Retinal Detachment
  [link]
• MedlinePlus Encyclopedia: Scoliosis
  [link]
Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Clubfoot
  https://medlineplus.gov/ency/article/001228.htm
- Encyclopedia: Lordosis
  https://medlineplus.gov/ency/article/003278.htm
- Encyclopedia: Retinal Detachment
  https://medlineplus.gov/ency/article/001027.htm
- Encyclopedia: Scoliosis
  https://medlineplus.gov/ency/article/001241.htm
- Health Topic: Bone Diseases
  https://medlineplus.gov/bonediseases.html
- Health Topic: Connective Tissue Disorders
  https://medlineplus.gov/connectivetissuedisorders.html
- Health Topic: Dwarfism
  https://medlineplus.gov/dwarfism.html

Genetic and Rare Diseases Information Center

- Spondyloepiphyseal dysplasia congenita
  https://rarediseases.info.nih.gov/diseases/4987/spondyloepiphyseal-dysplasia-congenita

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

- Johns Hopkins Medicine
  https://www.hopkinsmedicine.org/health/conditions-and-diseases/spondyloepiphyseal-dysplasia-congenita
- KidsHealth from the Nemours Foundation
- MalaCards: spondyloepiphyseal dysplasia congenita
  https://www.malacards.org/card/spondyloepiphyseal_dysplasia_congenita
• Nemours Children's Health System
  https://www.nemours.org/services/skeletal-dysplasia/spondyloepiphyseal.html?tab=about

• Orphanet: Orphanet: Spondyloepiphyseal dysplasia congenita
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=94068

Patient Support and Advocacy Resources
• American Cleft Palate-Craniofacial Association
  https://cleftline.org/

• 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/

• National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/spondyloepiphyseal-dysplasia-congenital/

• Resource List from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/dwarfism.html

• The MAGIC Foundation
  https://www.magicfoundation.org/

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=%28Osteochondrodysplasias%5BMAJR%5D%29+AND+%28%28spondyloepiphyseal+dysplasia+congenita%5B5BTIAB%5D%29+OR+%28sedc%5B5BTIAB%5D%29+OR+%28sed+congenita%5B5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM
• SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA
  http://omim.org/entry/183900
Medical Genetics Database from MedGen

- Dysplasia, Spondyloepiphyseal

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10743764

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/25735649

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8157027

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22791362

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17509551

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21924244

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