



## 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 *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.

## **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?  
[/primer/testing/geneticTesting](#)
- Genetic Testing Registry: Spondyloepiphyseal dysplasia congenita  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2745959/>

### Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22spondyloepiphyseal+dysplasia+congenita%22>

### Other Diagnosis and Management Resources

- GeneReview: Type II Collagen Disorders Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK540447>
- MedlinePlus Encyclopedia: Clubfoot  
<https://medlineplus.gov/ency/article/001228.htm>
- MedlinePlus Encyclopedia: Lordosis  
<https://medlineplus.gov/ency/article/003278.htm>
- MedlinePlus Encyclopedia: Retinal Detachment  
<https://medlineplus.gov/ency/article/001027.htm>
- MedlinePlus Encyclopedia: Scoliosis  
<https://medlineplus.gov/ency/article/001241.htm>

## **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  
<https://kidshealth.org/en/parents/dwarfism.html>
- MalaCards: spondyloepiphyseal dysplasia congenita  
[https://www.malacards.org/card/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](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%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>

#### Catalog of Genes and Diseases from OMIM

- SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA  
<http://omim.org/entry/183900>

## Medical Genetics Database from MedGen

- Dysplasia, Spondyloepiphyseal  
<https://www.ncbi.nlm.nih.gov/medgen/20916>

### **Sources for This Summary**

- Dahiya R, Cleveland S, Megerian CA. Spondyloepiphyseal dysplasia congenita associated with conductive hearing loss. *Ear Nose Throat J.* 2000 Mar;79(3):178-82.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/10743764>
- Silveira KC, Bonadia LC, Superti-Furga A, Bertola DR, Jorge AA, Cavalcanti DP. Six additional cases of SEDC due to the same and recurrent R989C mutation in the COL2A1 gene--the clinical and radiological follow-up. *Am J Med Genet A.* 2015 Apr;167A(4):894-901. doi: 10.1002/ajmg.a.36954. Epub 2015 Mar 3.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25735649>
- Spranger J, Winterpacht A, Zabel B. The type II collagenopathies: a spectrum of chondrodysplasias. *Eur J Pediatr.* 1994 Feb;153(2):56-65. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/8157027>
- Terhal PA, van Dommelen P, Le Merrer M, Zankl A, Simon ME, Smithson SF, Marcelis C, Kerr B, Kinning E, Mansour S, Hennekam RC, van der Hout AH, Cormier-Daire V, Lund AM, Goodwin L, Mégarbané A, Lees M, Betz RC, Tobias ES, Coucke P, Mortier GR. Mutation-based growth charts for SEDC and other COL2A1 related dysplasias. *Am J Med Genet C Semin Med Genet.* 2012 Aug 15;160C(3):205-16. doi: 10.1002/ajmg.c.31332. Epub 2012 Jul 12.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/22791362>
- Xia X, Cui Y, Huang Y, Pan L, Wu Y, Zhang P, Jin B. A first familial G504S mutation of COL2A1 gene results in distinctive spondyloepiphyseal dysplasia congenita. *Clin Chim Acta.* 2007 Jul; 382(1-2):148-50. Epub 2007 Apr 14.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17509551>
- Zhang Z, He JW, Fu WZ, Zhang CQ, Zhang ZL. Identification of three novel mutations in the COL2A1 gene in four unrelated Chinese families with spondyloepiphyseal dysplasia congenita. *Biochem Biophys Res Commun.* 2011 Oct 7;413(4):504-8. doi: 10.1016/j.bbrc.2011.08.090. Epub 2011 Sep 6.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21924244>

---

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/spondyloepiphyseal-dysplasia-congenita>

Reviewed: April 2016

Published: August 17, 2020

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services