# Spondyloepiphyseal dysplasia with metatarsal shortening

## Description

Spondyloepiphyseal dysplasia (SED) with metatarsal shortening (formerly called Czech dysplasia) is an inherited condition that affects joint function and bone development. People with this condition have joint pain that begins in late childhood or adolescence. The cartilage in their hips, knees, shoulders, and spine usually degenerates over time ( osteoarthritis), which may impair their mobility. Due to these severe joint problems, people with SED with metatarsal shortening may require joint replacement in early adulthood.

People with SED with metatarsal shortening often have shortened bones in their third and fourth toes, which make their first two toes appear unusually long. Affected individuals may also have abnormalities in the bones of the spine (vertebrae), including flattened vertebrae (platyspondyly), a reduction in the space between the vertebrae, or an abnormal curvature of the spine. Some people with SED with metatarsal shortening have progressive hearing loss.

## Frequency

SED with metatarsal shortening is rare; as of 2020, fewer than 15 families have been reported with this condition.

## Causes

SED with metatarsal shortening is caused by a particular variant (also called a mutation) in the *COL2A1* gene. The *COL2A1* gene provides instructions for making a protein that forms type II collagen, which is found in the clear gel that fills the eyeball (the vitreous) and in cartilage. Cartilage makes up much of the skeleton during early development. Most cartilage is later replaced by bone, except for the cartilage that continues to cover and protect the ends of bones and the cartilage that is present in the nose and external ears. Type II collagen is essential for the normal growth and development of bones and other connective tissues.

The variant that causes SED with metatarsal shortening replaces one protein building block (amino acid) known as arginine in the COL2A1 protein with another amino acid known as cysteine. This interferes 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 with metatarsal shortening

- 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

- Czech dysplasia, metatarsal type
- Progressive pseudorheumatoid dysplasia with hypoplastic toes
- SED with metatarsal shortening
- SED with metatarsal shortening, COL2A1-related
- Spondyloepiphyseal dysplasia with precocious osteoarthritis

## Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Spondyloepiphyseal dysplasia with metatarsal shortening (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1836683/)

Genetic and Rare Diseases Information Center

- Spondyloepiphyseal dysplasia with metatarsal shortening (https://rarediseases.info.nih.gov/diseases/10220/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- CZECH DYSPLASIA (https://omim.org/entry/609162)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28czech+dysplasia%5BTIAB%5D%29+OR+%28progressive+pseudorheumatoid+dysplasia+with+hypoplastic+t

oes%29+OR+%28spondyloarthropathy+with+short+third+and+fourth+toes%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

## References


- Burrage LC, Lu JT, Liu DS, Moss TJ, Gibbs R, Schlesinger AE, Bacino CA, Campeau PM, Lee BH. Early childhood presentation of Czech dysplasia. ClinDysmorphol. 2013 Apr;22(2):76-80. doi: 10.1097/MCD.0b013e32835fff39. Citation on PubMed (https://www.ncbi.nlm.nih.gov/pubmed/23448908)
- Gregersen PA, Savarirayan R. Type II Collagen Disorders Overview. 2019 Apr 25[ updated 2024 Oct 24]. In: Adam MP, Bick S, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, editors. GeneReviews(R) [Internet]. Seattle (WA): Universityof Washington, Seattle; 1993-2025. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK540447/ Citation on PubMed (https://www.ncbi.nlm.nih.gov/pubmed/31021589)
- Hoornaert KP, Marik I, Kozlowski K, Cole T, Le Merrer M, Leroy JG, Coucke PJ, Sillence D, Mortier GR. Czech dysplasia metatarsal type: another type II collagendisorder. Eur J Hum Genet. 2007 Dec;15(12):1269-75. doi: 10.1038/sj.ejhg.5201913.Epub 2007 Aug 29. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17726487)
- Kozlowski K, Marik I, Marikova O, Zemkova D, Kuklik M. Czech dysplasiametatarsal type. Am J Med Genet A. 2004 Aug 15;129A(1):87-91. doi:10.1002/ajmg.a.30132. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15266623)
- Marik I, Marikova O, Zemkova D, Kuklik M, Kozlowski K. Dominantly inheritedprogressive pseudorheumatoid dysplasia with hypoplastic toes. Skeletal Radiol.2004 Mar;33(3):157-64. doi: 10.1007/s00256-003-0708-z. Epub 2004 Jan 17. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/14730409)
- Tzschach A, Tinschert S, Kaminsky E, Lusga E, Mundlos S, Graul-Neumann LM. Czech dysplasia: report of a large family and further delineation of thephenotype. Am J Med Genet A. 2008 Jul 15;146A(14):1859-64. doi:10.1002/ajmg.a.32389. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18553548)


**Last updated December 19, 2024**