# 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 typically 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 spinal curvature. 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. This type of collagen is found mostly 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 development of bones and other connective tissues that form the body's supportive framework.

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 *COL2A1* gene variant 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
- 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%5BTIA

B%5D%29+OR+%28progressive+pseudorheumatoid+dysplasia+with+hypoplastic+toes%29+OR+%28spondyloarthropathy+with+short+third+and+fourth+toes%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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