Spondyloepimetaphyseal Dysplasia, Strudwick Type

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Spondyloepimetaphyseal dysplasia, Strudwick type is an inherited disorder of bone growth that results in short stature (dwarfism), skeletal abnormalities, and problems with vision. This condition affects the bones of the spine (spondylo-) and two regions (epiphyses and metaphyses) near the ends of long bones in the arms and legs. The Strudwick type was named after the first reported patient with the disorder.

Keywords: genetic conditions

1. Introduction

People with this condition have short stature from birth, with a very short trunk and shortened limbs. Their hands and feet, however, are usually average-sized. Affected individuals may have an abnormally curved lower back (lordosis) or a spine that curves to the side (scoliosis). This abnormal spinal curvature may be severe and can cause problems with breathing. Instability of the spinal bones (vertebrae) in the neck may increase the risk of spinal cord damage. Other skeletal features include flattened vertebrae (platyspondyly), severe protrusion of the breastbone (pectus carinatum), an abnormality of the hip joint that causes the upper leg bones to turn inward (coxa vara), and an inward- and upward-turning foot (clubfoot). Arthritis may develop early in life.

People with spondyloepimetaphyseal dysplasia, Strudwick type have mild changes in their facial features. Some infants are born with an opening in the roof of the mouth (a cleft palate) and their cheekbones may appear flattened. Eye problems that can impair vision are common, such as severe nearsightedness (high myopia) and tearing of the lining of the eye (retinal detachment).

2. Frequency

This condition is rare; only a few affected individuals have been reported worldwide.

3. Causes

Spondyloepimetaphyseal dysplasia, Strudwick type 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 the clear gel that fills the eyeball (the vitreous) and cartilage. Cartilage is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and 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.

Most mutations in the *COL2A1* gene that cause spondyloepimetaphyseal dysplasia, Strudwick type interfere with the assembly of type II collagen molecules. Abnormal collagen prevents bones and other connective tissues from developing properly, which leads to the signs and symptoms of spondyloepimetaphyseal dysplasia, Strudwick type.

3.1 Learn more about the gene associated with Spondyloepimetaphyseal dysplasia, Strudwick type

- COL2A1

4. 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.

5. Other Names for This Condition

- Dappled metaphysis syndrome
- SED Strudwick
- SEMD, Strudwick type
- SMED, Strudwick type
- SMED, type I
- Spondylometaepiphyseal dysplasia congenita, Strudwick type
- Spondylometaphyseal dysplasia (SMD)
- Strudwick syndrome

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