CHST3-related skeletal dysplasia

Description

CHST3-related skeletal dysplasia is a genetic condition characterized by bone and joint abnormalities that worsen over time. Affected individuals have short stature throughout life, with an adult height under 4 and a half feet. Joint dislocations, most often affecting the knees, hips, and elbows, are present at birth (congenital). Other bone and joint abnormalities can include an inward- and upward-turning foot (clubfoot), a limited range of motion in large joints, and abnormal curvature of the spine. The features of *CHST3*-related skeletal dysplasia are usually limited to the bones and joints; however, minor heart defects have been reported in a few affected individuals.

Researchers have not settled on a preferred name for this condition. It is sometimes known as autosomal recessive Larsen syndrome based on its similarity to another skeletal disorder called Larsen syndrome. Other names that have been used to describe the condition include spondyloepiphyseal dysplasia, Omani type; humero-spinal dysostosis; and chondrodysplasia with multiple dislocations. Recently, researchers have proposed the umbrella term *CHST3*-related skeletal dysplasia to refer to bone and joint abnormalities resulting from mutations in the *CHST3* gene.

Frequency

The prevalence of *CHST3*-related skeletal dysplasia is unknown. More than 30 affected individuals have been reported.

Causes

As its name suggests, *CHST3*-related skeletal dysplasia results from mutations in the *CHST3* gene. This gene provides instructions for making an enzyme called C6ST-1, which is essential for the normal development of 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. Mutations in the *CHST3* gene reduce or eliminate the activity of the C6ST-1 enzyme. A shortage of this enzyme disrupts the normal development of cartilage and bone, resulting in the abnormalities associated with *CHST3*-related skeletal dysplasia.

Learn more about the gene associated with CHST3-related skeletal dysplasia

- CHST3

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Autosomal recessive Larsen syndrome
- CDMD
- Chondrodysplasia with multiple dislocations
- Humero-spinal dysostosis
- SED with luxations, CHST3 type
- SED, Omani type
- Spondyloepiphyseal dysplasia with congenital joint dislocations
- Spondyloepiphyseal dysplasia, Omani type

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Larsen-like syndrome, B3GAT3 type (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3278404/)

Genetic and Rare Diseases Information Center

- CHST3-related skeletal dysplasia (https://rarediseases.info.nih.gov/diseases/13169/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- SPONDYLOEPIPHYSEAL DYSPLASIA WITH CONGENITAL JOINT DISLOCATIONS; SEDCJD (https://omim.org/entry/143095)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28chst3-related+skeletal+dysplasia%5BTIAB%5D%29+OR+%28humero-spinal+dysostosis%5BTIAB%5D%29%29+OR+%28%28larsen+syndrome%5BTIAB%5D%29+AND+%28CHST3%5BTIAB%5D%29+AND+%28autosomal+recessive%5BTIAB%5D%29%29+OR+%28%28spondyloepiphyseal+dysplasia%5BTIAB%5D%29+AND+%28omani+type%5BTIAB%5D%29%29+OR+%28%28SED%5BTIAB%5D%29+AND+%28omani+type%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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