Diastrophic dysplasia

Description

Diastrophic dysplasia is a disorder of cartilage and bone development. Affected individuals have short stature with very short arms and legs. Most also have early-onset joint pain (osteoarthritis) and joint deformities called contractures, which restrict movement. These joint problems often make it difficult to walk and tend to worsen with age. Additional features of diastrophic dysplasia include an inward- and upward-turning foot (clubfoot), progressive abnormal curvature of the spine, and unusually positioned thumbs (hitchhiker thumbs). About half of infants with diastrophic dysplasia are born with an opening in the roof of the mouth (a cleft palate). Swelling of the external ears is also common in newborns and can lead to thickened, deformed ears.

The signs and symptoms of diastrophic dysplasia are similar to those of another skeletal disorder called atelosteogenesis type 2; however, diastrophic dysplasia tends to be less severe. Although some affected infants have breathing problems, most people with diastrophic dysplasia live into adulthood.

Frequency

Although the exact prevalence of diastrophic dysplasia is unknown, researchers estimate that it affects about 1 in 500,000 newborns in the United States. This condition is more common in Finland, where it affects about 1 in 33,000 newborns.

Causes

Diastrophic dysplasia is one of several skeletal disorders caused by mutations in the *SLC26A2* gene. This gene provides instructions for making a protein that is essential for the normal development of cartilage and for its conversion to bone. 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 *SLC26A2* gene alter the structure of developing cartilage, preventing bones from forming properly and resulting in the skeletal problems characteristic of diastrophic dysplasia.

Learn more about the gene associated with Diastrophic dysplasia

- SLC26A2

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

- Diastrophic dwarfism
- DTD

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Diastrophic dysplasia (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0220726/)

Genetic and Rare Diseases Information Center

- Diastrophic dysplasia (https://rarediseases.info.nih.gov/diseases/6275/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- DIASTROPHIC DYSPLASIA; DTD (https://omim.org/entry/222600)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28diastrophic+dysplasia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D)

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