Kniest dysplasia

Kniest dysplasia is a disorder of bone growth characterized by short stature (dwarfism) with other skeletal abnormalities and problems with vision and hearing.

People with Kniest dysplasia are born with a short trunk and shortened arms and legs. Adult height ranges from 42 inches to 58 inches. Affected individuals have abnormally large joints that can cause pain and restrict movement, limiting physical activity. These joint problems can also lead to arthritis. Other skeletal features may include a rounded upper back that also curves to the side (kyphoscoliosis), severely flattened bones of the spine (platyspondyly), dumbbell-shaped bones in the arms and legs, long and knobby fingers, and an inward- and upward-turning foot (clubfoot).

Individuals with Kniest dysplasia have a round, flat face with bulging and wide-set eyes. Some affected infants are born with an opening in the roof of the mouth called a cleft palate. Infants may also have breathing problems due to weakness of the windpipe. Severe nearsightedness (myopia) and other eye problems are common in Kniest dysplasia. Some eye problems, such as tearing of the back lining of the eye (retinal detachment), can lead to blindness. Hearing loss resulting from recurrent ear infections is also possible.

Frequency

Kniest dysplasia is a rare condition; the exact incidence is unknown.

Causes

Kniest dysplasia 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 in 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 Kniest dysplasia 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 Kniest dysplasia.

Inheritance Pattern

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

- Kniest chondrodystrophy
- Kniest syndrome
- Metatropic dwarfism, type II
- Metatropic dysplasia type II
- Swiss cheese cartilage dysplasia

Diagnosis & Management

Genetic Testing Information

- What is genetic testing? [primer/testing/genetictesting]

Other Diagnosis and Management Resources

- GeneReview: Type II Collagen Disorders Overview [https://www.ncbi.nlm.nih.gov/books/NBK540447]
- MedlinePlus Encyclopedia: Clubfoot [https://medlineplus.gov/ency/article/001228.htm]
- MedlinePlus Encyclopedia: Retinal Detachment [https://medlineplus.gov/ency/article/001027.htm]
- MedlinePlus Encyclopedia: Scoliosis [https://medlineplus.gov/ency/article/001241.htm]

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Clubfoot [https://medlineplus.gov/ency/article/001228.htm]
- Encyclopedia: Retinal Detachment [https://medlineplus.gov/ency/article/001027.htm]
- Encyclopedia: Scoliosis [https://medlineplus.gov/ency/article/001241.htm]
- Health Topic: Bone Diseases [https://medlineplus.gov/bonediseases.html]
• Health Topic: Connective Tissue Disorders
  https://medlineplus.gov/connectivetissuedisorders.html

• Health Topic: Dwarfism
  https://medlineplus.gov/dwarfism.html

Genetic and Rare Diseases Information Center
• Kniest dysplasia
  https://rarediseases.info.nih.gov/diseases/6841/kniest-dysplasia

Additional NIH Resources
• National Institute of Arthritis and Musculoskeletal and Skin Diseases: Heritable Disorders of Connective Tissue
  https://www.niams.nih.gov/health-topics/heritable-disorders-connective-tissue

Educational Resources
• MalaCards: kniest dysplasia
  https://www.malacards.org/card/kniest_dysplasia

• Nemours Children's Health System
  https://www.nemours.org/services/skeletal-dysplasia/kniest.html?tab=about

• Orphanet: Kniest dysplasia
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=485

Patient Support and Advocacy Resources
• American Cleft Palate-Craniofacial Association
  https://cleftline.org/

• Human Growth Foundation
  https://www.hgfound.org/

• International Skeletal Dysplasia Registry, UCLA
  https://www.uclahealth.org/ortho/isdr

• Little People of America
  https://www.lpaonline.org/

• Little People UK
  https://littlepeopleuk.org/

• National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/kniest-dysplasia/

• Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/skeldysp.html
• Self Help for Hard of Hearing People (SHHH)
  https://www.hearingloss.org/
• The MAGIC Foundation
  https://www.magicfoundation.org/

Clinical Information from GeneReviews
• Type II Collagen Disorders Overview
  https://www.ncbi.nlm.nih.gov/books/NBK540447

Scientific Articles on PubMed
• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28kniest+dysplasia%5BTIAB%5D%29+OR+%28kniest+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3240+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM
• KNIEST DYSPLASIA
  http://omim.org/entry/156550

Medical Genetics Database from MedGen
• Kniest dysplasia

Sources for This Summary
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18175850
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10406661
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14644246

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