Kniest Dysplasia

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

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1. Introduction

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.

2. Frequency

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

3. 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.

3.1. The gene associated with Kniest dysplasia

- 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

- Kniest chondrodystrophy

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

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