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Kniest Dysplasia

National Cancer Institute

Source

National Cancer Institute. *Kniest Dysplasia*. NCI Thesaurus. Code C125594.

A rare, autosomal dominant inherited bone growth disorder caused by mutations in the COL2A1 gene. It is characterized by short stature (dwarfism) and other skeletal abnormalities, round, flat face with bulging and wide-set eyes, myopia and retinal detachment that can lead to blindness.