

Kniest dysplasia is one of several forms of dwarfism that is caused by a change (mutation) in a gene known as COL2A1. This gene is involved in the production of a particular protein that forms type 2 collagen, which is essential for the normal development of bones and other connective tissue. Changes in the composition of type 2 collagen lead to abnormal skeletal growth and, thus, to a variety of dwarfing conditions known as skeletal dysplasias. Kniest Syndrome is a rare disorder that affects males and females in equal numbers. Newborns with this disorder are clearly in distress so that x-rays may be ordered. Diagnosis may frequently be made on the basis of the radiographs developed, while a bone biopsy usually clarifies more ambiguous cases.