CHST3-related skeletal dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. CHST3-related skeletal dysplasia. ORPHA:263463

CHST3-related skeletal dysplasia is a very rare bone disorder characterized clinically by short stature of prenatal onset; dislocation of the knees, hips or elbows; club feet; limitation of range of motion of large joints; progressive kyphosis; and occasional scoliosis. In a few patients, minor heart valve dysplasia has also been described. Intellect, vision and hearing are normal.