DISEASE:
Multiple epiphyseal dysplasia, Beighton type

NAME:	Multiple epiphyseal dysplasia, Beighton type
DESCRIPTION:	A rare primary bone dysplasia characterized by the association of multiple epiphyseal dysplasia, visual impairment (with early-onset progressive myopia, retinal thinning, and cataracts), and conductive hearing loss. Patients are of short stature and present brachydactyly, genu valgus deformity, and joint pain.
ORPHACODE:	166011
SYNONYMS:	Multiple epiphyseal dysplasia-myopia-deafness syndrome Multiple epiphyseal dysplasia-myopia-hearing loss syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	COL2A1
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