

DISEASE:
Kniest dysplasia

NAME:	Kniest dysplasia
DESCRIPTION:	Kniest dysplasia is a severe type II collagenopathy characterized by a short trunk and limbs, prominent joints and midface hypoplasia (round face with a flat nasal root).
ORPHACODE:	485
XREF(S):	Orphanet MeSH OMIM ICD-10
ANALYTE(S):	COL2A1
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Source URL: <http://gentest.healthdata.be/disease/840>

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