

DISEASE:
Spondyloepimetaphyseal dysplasia, aggrecan type

NAME:	Spondyloepimetaphyseal dysplasia, aggrecan type
DESCRIPTION:	Spondyloepimetaphyseal dysplasia, aggrecan type is a new form of skeletal dysplasia characterized by severe short stature, facial dysmorphism and characteristic radiographic findings.
ORPHACODE:	171866
SYNONYMS:	SEMD, aggrecan type
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>ACAN</u>
CREATED:	13 May 2019 - 01:02
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