

DISEASE:

Short stature-advanced bone age-early-onset osteoarthritis syndrome

NAME:	Short stature-advanced bone age-early-onset osteoarthritis syndrome
DESCRIPTION:	A rare, primary bone dysplasia characterized by proportional short stature, early cessation of bone growth, accelerated skeletal maturation, variable presence of early-onset osteoarthritis and osteochondritis dissecans, and normal endocrine evaluation. The variable dysmorphic features include mild to relative macrocephaly, frontal bossing, midfacial hypoplasia, flat nasal bridge, brachydactyly, broad thumbs, and lordosis.
ORPHACODE:	435804
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ACAN
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RELATED CONTENT

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