DISEASE:
Autosomal dominant otospondylomegaepiphyseal dysplasia

NAME:	Autosomal dominant otospondylomegaepiphyseal dysplasia
DESCRIPTION:	A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by craniofacial dysmorphism (midface hypoplasia, depressed nasal bridge, small nose with upturned tip, cleft palate, Pierre Robin sequence), bilateral, pronounced sensorineural hearing loss, and skeletal/joint anomalies (including spondyloepiphyseal dysplasia, arthralgia/arthropathy), in the absence of ocular abnormalities.
ORPHACODE:	166100
SYNONYMS:	AD OSMED Stickler syndrome type 3 Stickler syndrome, non-ocular type
XREF(S):	Orphanet MeSH OMIM ICD-10
ANALYTE(S):	COL2A1 COL11A2
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