DISEASE:
Achondrogenesis type 2

NAME:	Achondrogenesis type 2
DESCRIPTION:	A rare, lethal type of achondrogenesis, and part of the spectrum of type 2 collagen-related bone disorders, characterized by severe micromelia, short neck with large head, small thorax, protuberant abdomen, underdeveloped lungs, distinctive facial features such as a prominent forehead, a small chin, a cleft palate (in some) and distinctive histological features of the cartilage.
ORPHACODE:	93296
SYNONYMS:	Achondrogenesis, Langer-Saldino type
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	COL2A1
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