DISEASE:
Achondrogenesis type 1B

NAME:	Achondrogenesis type 1B
DESCRIPTION:	A rare, lethal type of achondrogenesis characterized by severe micromelia with very short fingers and toes, a flat face, a short neck, thickened soft tissue around the neck, hypoplasia of the thorax, protuberant abdomen, a hydropic fetal appearance and distinctive histological features of the cartilage.
ORPHACODE:	93298
SYNONYMS:	Achondrogenesis, Parenti-Fraccaro type
XREF(S):	Orphanet MeSH OMIM ICD-10
ANALYTE(S):	SLC26A2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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