DISEASE:
Acromesomelic dysplasia, Grebe type

NAME:	Acromesomelic dysplasia, Grebe type
DESCRIPTION:	A rare autosomal recessive acromesomelic dysplasia characterized by severe dwarfism at birth, abnormalities confined to limbs, severe shortening and deformity of long bones, fusion or absence of carpal and tarsal bones, ball shaped fingers and, occasionally, polydactyly and absent joints. As seen in acromesomelic dysplasia, Hunter-Thomson type and acromesomelic dysplasia, Maroteaux Type, facial features and intelligence are normal.
ORPHACODE:	2098
SYNONYMS:	Chondrodysplasia, Grebe type
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	BMPR1B GDF5
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Analytes

- bone morphogenetic protein receptor type 1B
- growth differentiation factor 5