DISEASE:
Acromesomelic dysplasia, Hunter-Thompson type

NAME:	Acromesomelic dysplasia, Hunter-Thompson type
DESCRIPTION:	A rare autosomal recessive acromesomelic dysplasia characterized by severe dwarfism (adult height approximately 120 cm) with abnormalities limited to the limbs (affecting the lower limbs more than upper limbs, with middle and distal segments being the most affected), severe shortening, absence or fusion of tubular bones of hands and feet and large joint dislocations. As seen in acromesomelic dysplasia, Grebe type and acromesomelic dysplasia, Maroteaux type, facial features and intelligence are normal.
ORPHACODE:	968
SYNONYMS:	Acromesomelic dwarfism
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	GDF5
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Analytes

- growth differentiation factor 5