DISEASE:
Acromesomelic dysplasia, Maroteaux type

NAME:	Acromesomelic dysplasia, Maroteaux type
DESCRIPTION:	A rare autosomal recessive acromesomelic dysplasia characterized by severe dwarfism (adult height <120 cm), both axial and appendicular involvement (shortening of the middle and distal segments of limbs and vertebral shortening), and with normal facial appearance and intelligence. It is a less severe form than acromesomelic dysplasia, Grebe type and acromesomelic dysplasia, Hunter-Thomson type .
ORPHACODE:	40
XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	NPR2
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Short Stature (gene panel)
- Short stature/ Growth retardation/ (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- natriuretic peptide receptor 2

Related Gene Panels

- Growth retardation/short stature (genepanel) - UZA
- Short Stature (46 genes) - IPG