DISEASE:
Hypochondroplasia

NAME:	Hypochondroplasia
DESCRIPTION:	A primary bone dysplasia with micromelia characterized by disproportionate short stature, mild lumbar lordosis and limited extension of the elbow joints.
ORPHACODE:	429
XREF(S):	Orphanet ICD-10 OMIM MedDRA
ANALYTE(S):	FGFR3
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Hypochondroplasia (full sequencing)
- Hypochondroplasia (full sequencing)
- Hypochondroplasia (hot spot mutation - p.Asn540)
- Hypochondroplasia (hot spot mutation - p.Asn540Lys)
- Hypochondroplasia (Hotspot mutation p.(Asn540Lys))
- Hypochondroplasia (hot spot mutations - p.Asn540; p.Ile538; p.Lys650 FGFR3)
- Short Stature (gene panel)
- Short stature/ Growth retardation/ (gene panel)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Gent

Related Analytes

- fibroblast growth factor receptor 3

Related Gene Panels

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