GENETIC TEST:
Hypochondroplasia (hot spot mutation - p.Asn540Lys)

FULL NAME:	Hypochondroplasia (hot spot mutation - p.Asn540Lys)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Mutation screening and sequence analysis of selected exons
METHOD TECHNIQUE:	Bi-directional Sanger Sequence analysis
RIZIV CODE:	565390-565401
EQA:	• Skeletal dysplasis
TURNAROUND TIME (MAXIMUM):	2 months
CREATED:	07 Aug 2019 - 11:17
CHANGED:	22 Jun 2022 - 10:35

URL:	https://labogidsmedgen.uza.be/analyses/hypochondroplasie

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Source URL: http://gentest.healthdata.be/genetic_test/538

RELATED CONTENT

Related Diseases

- Hypochondroplasia

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- fibroblast growth factor receptor 3