

GENETIC TEST:
Achondroplasia (hot spot mutation - p.Gly380Arg)

FULL NAME:	Achondroplasia (hot spot mutation - p.Gly380Arg)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis, Prenatal diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, Chorionic villi, Amniotic fluid, DNA
METHOD CATEGORY:	Targeted variant analysis
METHOD TECHNIQUE:	Bi-directional Sanger Sequence analysis
RIZIV CODE:	565390-565401
ACCREDITATION (ISO 15189):	2022-02-24 / 2026-02-23

EQA:	• Skeletal dysplasis , • Skeletal dysplasis
TURNAROUND TIME (MAXIMUM):	21 days
CREATED:	26 Jul 2019 - 12:45
CHANGED:	28 Sep 2023 - 09:30
URL:	https://www.chuliege.be/jcms/c_525267/fr/achondroplasie

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