GENETIC TEST:
Achondroplasia (FGFR3 hot spot mutation - p.Gly380)

FULL NAME:	Achondroplasia (FGFR3 hot spot mutation - p.Gly380)
DESCRIPTION:	Alias: ACH
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Mutation confirmation, Post-natal Diagnosis, Pre-implantation genetic diagnosis, Prenatal diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, Amniotic fluid, Chorionic villi, Cell culture
METHOD CATEGORY:	Targeted variant analysis

METHOD TECHNIQUE:	Bi-directional Sanger Sequence analysis
RIZIV CODE:	565390-565401
EQA:	• DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger , • DNA Sequencing - Sanger
TURNAROUND TIME (MAXIMUM):	3 months (10 working days for prenatal diagnosis)
CREATED:	26 Jul 2019 - 12:45
CHANGED:	01 Mar 2023 - 16:26
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:Achondroplasie&&&&&1039&COLLE…

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