GENETIC TEST:
Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure (hot spot mutation - 1555A-G in MT-RNR1)

FULL NAME:	Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure (hot spot mutation - 1555A-G in MT-RNR1)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Targeted variant analysis
METHOD TECHNIQUE:	Bi-directional Sanger Sequence analysis Next Generation Sequencing (NGS)
RIZIV CODE:	565390-565401
EQA:	• Mitochondrial DNA (mtDNA) Metabolic Disorders, • Mitochondrial DNA (mtDNA) Metabolic Disorders, • Mitochondrial DNA (mtDNA) Metabolic Disorders

TURNAROUND TIME (MAXIMUM):	20 - 60 days
CREATED:	06 Aug 2019 - 13:11
CHANGED:	22 Jan 2024 - 09:57

RELATED CONTENT

Related Diseases

- Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- mitochondrially encoded 12S rRNA