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

NAME:	Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure
ORPHACODE:	168609
SYNONYMS:	Mitochondrial isolated neurosensory deafness with susceptibility to aminoglycoside exposure Mitochondrial isolated neurosensory hearing loss with susceptibility to aminoglycoside exposure Mitochondrial isolated sensorineural deafness with susceptibility to aminoglycoside exposure Mitochondrial isolated sensorineural hearing loss with susceptibility to aminoglycoside exposure Mitochondrial non-syndromic neurosensory deafness with susceptibility to aminoglycoside exposure Mitochondrial non-syndromic neurosensory hearing loss with susceptibility to aminoglycoside exposure Mitochondrial non-syndromic sensorineural hearing loss with susceptibility to aminoglycoside exposure
XREF(S):	Orphanet
ANALYTE(S):	TRMU MT-CO1 MT-ND4 MT-RNR1 MT-TS1
CREATED:	13 May 2019 - 01:02
CHANGED:	01 Jul 2019 - 06:57

RELATED CONTENT

Related Genetic Tests

- Leigh / NARP Syndrome
- Mitochondrial disorders, mitchondrial DNA based (Full sequencing of mtDNA genome)
- Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure (hot spot mutation - 1555A-G in MT-RNR1)
- Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure (hot spot mutation - 1555A-G in MT-RNR1)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- mitochondrially encoded cytochrome c oxidase I
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 4
- mitochondrially encoded 12S rRNA
- mitochondrially encoded tRNA-Ser (UCN) 1
- tRNA mitochondrial 2-thiouridylase

Related Gene Panels

- Leigh syndrome (mtDNA / 37 genes) - VUB

Source URL: http://gentest.healthdata.be/disease/279