DISEASE:
Leber hereditary optic neuropathy

NAME:	Leber hereditary optic neuropathy
DESCRIPTION:	A rare hereditary optic neuropathy characterized by sudden onset, painless central vision loss, loss of retinal ganglion cells and optic atrophy.
ORPHACODE:	104
SYNONYMS:	LHON Leber optic atrophy
XREF(S):	Orphanet OMIM ICD-10 OMIM OMIM

ANALYTE(S):	NDUFS2 DNAJC30 MT-ATP6 MT-CO1 MT-CO3 MT-CYB MT-ND1 MT-ND2 MT-ND4 MT-ND4L MT-ND5 MT-ND6
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RELATED CONTENT

Related Genetic Tests

- Leber hereditary optic neuropathy (LHON) (hot spot mutations - p.Ala53Thr in MT-ND1; p.Arg340His in MT-ND4; p.Met64Val in MT-ND6)
- Leber hereditary optic neuropathy (LHON) (hot spot mutations - p.Ala53Thr in MT-ND1; p.Arg340His in MT-ND4; p.Met64Val in MT-ND6)
- Leber hereditary optic neuropathy (LHON) – (DNAJC30 gene)

Related Laboratories

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

Related Analytes

- DnaJ heat shock protein family (Hsp40) member C30
- mitochondrially encoded ATP synthase membrane subunit 6
- mitochondrially encoded cytochrome c oxidase I
- mitochondrially encoded cytochrome c oxidase III
- mitochondrially encoded cytochrome b
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 1
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 2
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 4
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 4L
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 5
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 6
- NADH:ubiquinone oxidoreductase core subunit S2

Related Gene Panels

- LHON (3 genes) - VUB
