DISEASE:
MERRF

NAME:	MERRF
DESCRIPTION:	A rare mitochondrial oxidative phosphorylation disorder characterized by myoclonic seizures, ataxia, generalized epilepsy, muscle weakness and ragged red fibers in the muscle biopsy.
ORPHACODE:	551
SYNONYMS:	Fukuhara syndrome Myoclonus epilepsy associated with ragged-red fibres
XREF(S):	Orphanet MedDRA ICD-10 OMIM MeSH

ANALYTE(S):	MT-TP MT-ND5 MT-TL1 MT-TK MT-RNR1 MT-TQ MT-TH MT-TS1 MT-TS2 MT-TF
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RELATED CONTENT

Related Genetic Tests

- Leigh / NARP Syndrome
- Myoclonic epilepsy associated with ragged-red fibers (MERFF) (full sequencing) (2nd tier)
- Myoclonic epilepsy associated with ragged-red fibers (MERFF) (hot spot mutation - m.8344A>G)
- Myoclonic epilepsy associated with ragged-red fibers (MERFF) (hot spot mutation - m.8344A>G) (1st tier)

Related Laboratories

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

Related Analytes

- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 5
- mitochondrially encoded 12S rRNA
- mitochondrially encoded tRNA-Phe (UUU/C)
- mitochondrially encoded tRNA-His (CAU/C)
- mitochondrially encoded tRNA-Lys (AAA/G)
- mitochondrially encoded tRNA-Leu (UUA/G) 1
- mitochondrially encoded tRNA-Pro (CCN)
- mitochondrially encoded tRNA-Gln (CAA/G)
- mitochondrially encoded tRNA-Ser (UCN) 1
- mitochondrially encoded tRNA-Ser (AGU/C) 2

Related Gene Panels

- Leigh syndrome (mtDNA / 37 genes) - VUB

Source URL: http://gentest.healthdata.be/disease/83