DISEASE:
NARP syndrome

NAME:	NARP syndrome
DESCRIPTION:	A clinically heterogeneous progressive condition characterized by a combination of proximal neurogenic muscle weakness, sensory-motor neuropathy, ataxia, and pigmentary retinopathy.
ORPHACODE:	644
SYNONYMS:	Neurogenic muscle weakness-ataxia-retinitis pigmentosa syndrome Neuropathy-ataxia-retinitis pigmentosa syndrome
XREF(S):	Orphanet OMIM ICD-10 MedDRA
ANALYTE(S):	MT-ATP6
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Leigh / NARP Syndrome
- Neuropathy, Ataxia, and Retinitis Pigmentosa (NARP) syndrome (hot spot mutation - m.8993T>C/G)

Related Laboratories

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

Related Analytes

- mitochondrially encoded ATP synthase membrane subunit 6

Related Gene Panels

- Leigh syndrome (mtDNA / 37 genes) - VUB