DISEASE:
Isolated ATP synthase deficiency

NAME:	Isolated ATP synthase deficiency
DESCRIPTION:	Isolated ATP synthase deficiency is a rare, genetic, mitochondrial oxidative phosphorylation disorder that may present with a wide range of symptoms (including muscular hypotonia, hypertrophic cardiomyopathy, psychomotor delay, encephalopathy, peripheral neuropathy, lactic acidosis, 3-methylglutaconic aciduria) and clinical syndromes (including NARP and MILS).
ORPHACODE:	254913
SYNONYMS:	Isolated mitochondrial respiratory chain complex V deficiency
XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM ICD-10

ANALYTE(S):	MT-ATP6 ATP5MK MT-ATP8 ATPAF2 ATP5F1E ATP5F1A ATPAF1 ATP5F1D
CREATED:	13 May 2019 - 01:02
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Related Genetic Tests

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- Centrum Medische Genetica - UZ Brussel VUB

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- ATP synthase F1 subunit alpha
- ATP synthase F1 subunit delta
- ATP synthase F1 subunit epsilon
- ATP synthase membrane subunit k
- ATP synthase mitochondrial F1 complex assembly factor 1
- ATP synthase mitochondrial F1 complex assembly factor 2
- mitochondrially encoded ATP synthase membrane subunit 6
- mitochondrially encoded ATP synthase membrane subunit 8

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