DISEASE:
Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy

NAME:	Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy
DESCRIPTION:	A rare mitochondrial DNA depletion syndrome characterized by neonatal or infantile onset of hypotonia, failure to thrive, global developmental delay, and persistent lactic acidosis. The disease course is variable and ranges from intractable diarrhea and respiratory failure with fatal outcome in early infancy to a milder phenotype with survival into childhood. Additional reported features include sensorineural hearing loss, microcephaly, seizures, pigmentary retinopathy, and renal tubulopathy.
ORPHACODE:	255235
SYNONYMS:	mtDNA depletion syndrome, encephalomyopathic form with renal tubulopathy
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	RRM2B
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RELATED CONTENT

Related Genetic Tests

- Mitochondrial disorders (gene panel)
- Mitochondrial disorders, mitchondrial DNA based (Full sequencing of mtDNA genome)
- mtDNA depletion syndrome

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- ribonucleotide reductase regulatory TP53 inducible subunit M2B

Related Gene Panels

- mitochondrial disease, nuclear based (343 genes) - VUB
- mtDNA depletion syndrome (6 genes) - VUB