DISEASE:
Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency

NAME:	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency
DESCRIPTION:	A rare immune disease characterized by severely reduced mitochondrial DNA content due to DGUOK deficiency typically manifesting with early-onset liver dysfunction, psychomotor delay, hypotonia, rotary nystagmus that develops into opsoclonus, lactic acidosis and hypoglycemia.
ORPHACODE:	279934
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	DGUOK
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RELATED CONTENT

Related Genetic Tests

- Cholestasis (gene panel)
- Metabolic diseases with hepatic disorders (20 genes)
- Mitochondrial disorders (gene panel)
- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)
- mtDNA depletion syndrome

Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- deoxyguanosine kinase

Related Gene Panels

- Cholestasis (40 genes) - UCL
- Neuromuscular disorders (166 genes) - VUB
- mitochondrial disease, nuclear based (343 genes) - VUB

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