DISEASE:
Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies

NAME:	Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies
DESCRIPTION:	A rare mitochondrial DNA depletion syndrome characterized by congenital or early-onset lactic acidosis, hypotonia, and severe global developmental delay with feeding difficulties and failure to thrive. It is frequently associated with variable dysmorphic facial features. Additional manifestations include seizures, movement disorders, and cardiac and ophthalmologic anomalies, among others. Brain imaging may show generalized atrophy and white matter abnormalities.
ORPHACODE:	369897
SYNONYMS:	mtDNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	FBXL4
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RELATED CONTENT

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- Centrum Medische Genetica - UZ Brussel VUB

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Related Gene Panels

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