DISEASE:
Multiple mitochondrial dysfunctions syndrome type 2

NAME:	Multiple mitochondrial dysfunctions syndrome type 2
DESCRIPTION:	A rare mitochondrial disease characterized by infantile onset of severe regression after a period of normal development, epileptic encephalopathy, hypotonia, movement disorder, cardiomyopathy, hyperglycinemia, and lactic acidosis. Optic atrophy may also be present. Brain imaging findings are highly variable and include white matter abnormalities. The disease is typically fatal in infancy.
ORPHACODE:	401874
SYNONYMS:	BOLA3 deficiency MMDS2
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	BOLA3
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