Lethal infantile mitochondrial myopathy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Lethal infantile mitochondrial myopathy. ORPHA:254857

Lethal infantile mitochondrial myopathy is a rare mitochondrial oxidative phosphorylation disorder characterized by progressive generalized hypotonia, progressive external ophthalmoplegia and severe lactic acidosis, which results in early fatality (days to months after birth). Patients may present with lethargy and areflexia and may associate additional features, such as cardiomyopathy, renal dysfunction, liver involvement and seizures.