Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome. ORPHA:391348

Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome is a rare, genetic, mitochondrial oxidative phosphorylation disorder characterized by intrauterine growth retardation, microcephaly, hypotonia, vision impairment, speech and language delay and lactic acidosis with reduced respiratory chain activity (typically complex I). Additonal features may include macrocytic anemia, tremor, muscular atrophy, dysmetria and mild intellectual disability.