Mitochondrial myopathy with reversible cytochrome C oxidase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Mitochondrial myopathy with reversible cytochrome C oxidase deficiency. ORPHA:254864

A rare, genetic, mitochondrial oxidative phosphorylation disorder characterized by a potentially life-threatening, severe myopathy manifesting in the neonatal to early infantile period, followed by marked, spontaneous improvement of muscular function by early childhood. Associated biochemical findings include lactic acidosis and a transient, marked decrease in respiratory chain activity.