Combined oxidative phosphorylation defect type 14

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined oxidative phosphorylation defect type 14. ORPHA:319519

Combined oxidative phosphorylation defect type 14 is a rare mitochondrial disease due to a defect in mitochondrial protein synthesis characterized by neonatal or infancy-onset of seizures that are refractory to treatment, delayed or absent psychomotor development and lactic acidosis. Additional manifestations reported include poor feeding, failure to thrive, microcephaly, hypotonia, anemia and thrombocytopenia.