Open Peer Review on Qeios

Combined oxidative phosphorylation defect type 20

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined oxidative phosphorylation defect type 20. ORPHA:420728

Combined oxidative phosphorylation defect type 20 is a rare mitochondrial oxidative phosphorylation disorder characterized by variable combination of psychomotor delay, hypotonia, muscle weakness, seizures, microcephaly, cardiomyopathy and mild dysmorphic facial features. Variable types of structural brain anomalies have also been reported. Biochemical studies typically show decreased activity of mitochondrial complexes (mainly complex I).