Combined oxidative phosphorylation defect type 21

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined oxidative phosphorylation defect type 21. ORPHA:420733

Combined oxidative phosphorylation defect type 21 is a rare mitochondrial disease characterized by axial hypotonia with limb hypertonia, developmental delay, hyperlactatemia, central nervous system anomalies visible on magnetic resonance imaging (e.g. corpus callosum hypoplasia, lesions of the globus pallidus) and multiple deficiency of the mitochondrial respiratory chain complexes in muscle tissue, but not in fibroblasts or liver.