Combined oxidative phosphorylation defect type 24

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined oxidative phosphorylation defect type 24. ORPHA:444458

Combined oxidative phosphorylation defect type 24 is a rare mitochondrial oxidative phosphorylation disorder characterized by variable phenotype, including developmental delay with psychomotor regression, intellectual disability, epilepsy, Leigh syndrome, non-syndromic hearing loss, visual impairment and severe myopathy. Decreased activity of mitochondrial respiratory complexes and lactic acidosis are common findings, and diffuse cerebral atrophy may be associated.