Combined oxidative phosphorylation defect type 15

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined oxidative phosphorylation defect type 15. ORPHA:319524

Combined oxidative phosphorylation defect type 15 is a rare mitochondrial disease due to a defect in mitochondrial protein synthesis characterized by onset in infancy or early childhood of muscular hypotonia, gait ataxia, mild bilateral pyramidal tract signs, developmental delay (affecting mostly speech and coordination) and subsequent intellectual disability. Short stature, obesity, microcephaly, strabismus, nystagmus, reduced visual acuity, lactic acidosis, and a brain neuropathology consistent with Leigh syndrome are also reported.