Combined oxidative phosphorylation defect type 7

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined oxidative phosphorylation defect type 7. ORPHA:254930

Combined oxidative phosphorylation defect type 7 is a rare mitochondrial disease due to a defect in mitochondrial protein synthesis characterized by a variable phenotype that includes onset in infancy or early childhood of failure to thrive and psychomotor regression (after initial normal development), as well as ocular manifestations (such as ptosis, nystagmus, optic atrophy, ophthalmoplegia and reduced vision). Additional manifestations include bulbar paresis with facial weakness, hypotonia, difficulty chewing, dysphagia, mild dysarthria, ataxia, global muscle atrophy, and areflexia. It has a relatively slow disease progression with patients often living into the third decade of life.