Combined oxidative phosphorylation defect type 17

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined oxidative phosphorylation defect type 17. ORPHA:369913

Combined oxidative phosphorylation defect type 17 is a rare, genetic, mitochondrial disorder due to a defect in mitochondrial protein synthesis characterized by infantile-onset of severe hypertrophic cardiomyopathy (that occasionally progresses to dilated cardiomyopathy) associated with failure to thrive, global development delay, muscular hypotonia, elevated serum lactate and complex I deficiency in skeletal muscle biopsy. Intellectual disability, pericardial effusion and a mild cardiac phenotype have been also reported.