Combined oxidative phosphorylation defect type 9

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined oxidative phosphorylation defect type 9. ORPHA:319509

Combined oxidative phosphorylation defect type 9 is a rare mitochondrial disease due to a defect in mitochondrial protein synthesis characterized by initially normal growth and development followed by the infantile-onset of failure to thrive, psychomotor delay, poor feeding, dyspnea, severe hypertrophic cardiomyopathy and hepatomegaly. Laboratory studies report increased plasma lactate and alanine, abnormal liver enzymes and decreased activity of mitochondrial respiratory chain complexes I, III, IV, and V.