Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD9
COXPD9

Kod ORPHA
319509

Kod OMIM
614582

Kod ICD10
E88.8

Kod ICD11
5C53.23

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*Źródło

orphanet