Opis choroby *

Definicja

Combined oxidative phosphorylation defect type 14 is a rare mitochondrial disease due to a defect in mitochondrial protein synthesis characterized by neonatal or infancy-onset of seizures that are refractory to treatment, delayed or absent psychomotor development and lactic acidosis. Additional manifestations reported include poor feeding, failure to thrive, microcephaly, hypotonia, anemia and thrombocytopenia.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD14
COXPD14

Kod ORPHA
319519

Kod OMIM
614946

Kod ICD10
E88.8

Kod ICD11
5C53.23

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*Źródło

orphanet