

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

*Źródło

orphanet