

Złożony defekt fosforylacji oksydacyjnej typu 4

Kod Orpha: 254925 Kod OMIM: 610678

Opis choroby *

Definicja

Combined oxidative phosphorylation defect type 4 is a rare mitochondrial disorder due to a defect in mitochondrial protein synthesis characterized by a neonatal onset of severe metabolic acidosis and respiratory distress, persistent lactic acidosis with episodes of metabolic crises, developmental regression, microcephaly, abnormal gaze fixation and pursuit, axial hypotonia with limb spasticity and reduced spontaneous movements.

Neuroimaging studies reveal polymicrogyria, white matter abnormalities and multiple cystic brain lesions, including basal ganglia, and cerebral atrophy. Decreased activity of complex I and IV have been determined in muscle biopsy.

Dane

Klasyfikacja

Choroba

Synonimy

COXPD4

COXPD4

Kod ORPHA

254925

Kod OMIM

610678

Kod ICD10

E88.8

Kod ICD11

5C53.23

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie www.orphanet.pl