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

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*Źródło

orphanet