Opis choroby *

Definicja

A rare, genetic neurological disorder defined by early-onset of neurologic symptoms, biphasic clinical course, unique MRI features (incl. extensive, symmetrical, deep white matter abnormalities), and increased lactate in body fluids. The severe form is characterized by delayed psychomotor development, seizures, early-onset hypotonia, and persistently increased lactate levels. The mild form usually presents with irritability, psychomotor regression after six months of age, and temporary high lactate levels, with overall clinical improvement from the second year onward.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD12
LTBL
COXPD12
Złożony defekt fosforylacji oksydacyjnej typu 12
Combined oxidative phosphorylation defect type 12
LTBL

Kod ORPHA
314051

Kod OMIM
614924

Kod ICD10
E88.8

Kod ICD11
-

---

*Źródło

orphanet