

Opis choroby *

Definicja

Combined oxidative phosphorylation defect type 21 is a rare mitochondrial disease characterized by axial hypotonia with limb hypertonia, developmental delay, hyperlactatemia, central nervous system anomalies visible on magnetic resonance imaging (e.g. corpus callosum hypoplasia, lesions of the globus pallidus) and multiple deficiency of the mitochondrial respiratory chain complexes in muscle tissue, but not in fibroblasts or liver.

Dane

Klasyfikacja

Choroba

Synonimy

COXPD21

COXPD21

Kod ORPHA

420733

Kod OMIM

615918

Kod ICD10

E88.8

Kod ICD11

5C53.23

*Źródło

orphanet