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

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*Źródło

orphanet