Opis choroby *

Definicja

A rare mitochondrial oxidative phosphorylation disorder characterized by microcephaly, global developmental delay, spastic-dystonic movement disorder, intractable seizures, optic atrophy, autonomic dysfunction, and peripheral neuropathy. Serum lactate is increased, and muscle biopsy shows decreased activity of mitochondrial respiratory complexes I and III. Brain imaging reveals progressive cerebellar atrophy and delayed myelination.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD29
COXPD29

Kod ORPHA
478029

Kod OMIM
616811

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet