Opis choroby *

Definicja

A rare mitochondrial oxidative phosphorylation disorder characterized by a variable clinical phenotype including infantile onset of epileptic encephalopathy, hypotonia, global developmental delay, failure to thrive, complex movement disorder, and liver involvement, as well as childhood onset of severe myoclonus epilepsy, cognitive decline, progressive hearing and visual impairment, and progressive tetraparesis. Serum lactate may be increased, and brain imaging shows variable atrophy and white matter abnormalities.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD27
COXPD27

Kod ORPHA
477774

Kod OMIM
616672

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet