Opis choroby *

Definicja

A rare mitochondrial disease characterized by early onset of hypertrophic cardiomyopathy and variable neurologic symptoms including global developmental delay, hypotonia, intellectual disability, visual impairment, and seizures. Lactic acidosis is present in all patients. Muscle biopsy usually shows decreased activity of mitochondrial complexes I and IV. Brain imaging may reveal variable abnormal signal intensities in the thalamus, basal ganglia, and/or brain stem.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD23
COXPD23

Kod ORPHA
444013

Kod OMIM
616198

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet