Opis choroby *

Definicja

A rare mitochondrial oxidative phosphorylation disorder characterized by a highly variable phenotype which may present as exercise intolerance with prominent exertional dyspnea, progressive muscle weakness, spasticity, and neuropathy, but without cognitive impairment or cardiac involvement, or as global developmental delay, growth retardation, hypotonia, and spasticity. Hypertrophic cardiomyopathy, optic atrophy, seizures, and dysmorphic facial features have also been reported in the more severe phenotype. Serum lactate may be elevated, and muscle biopsy shows myopathic features and variably decreased activity of mitochondrial respiratory chain complexes.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD26
COXPD26

Kod ORPHA
477684

Kod OMIM
616539

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet