Opis choroby *

Definicja

Combined oxidative phosphorylation defect type 8 is a mitochondrial disease due to a defect in mitochondrial protein synthesis resulting in deficiency of respiratory chain complexes I, III and IV in the cardiac and skeletal muscle and brain characterized by severe hypertrophic cardiomyopathy, pulmonary hypoplasia, generalized muscle weakness and neurological involvement.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD8
COXPD8

Kod ORPHA
319504

Kod OMIM
614096

Kod ICD10
E88.8

Kod ICD11
5C53.23

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*Źródło

orphanet