Opis choroby *

Definicja

Combined oxidative phosphorylation defect type 25 is a rare mitochondrial oxidative phosphorylation disorder with decreased respiratory complex I and IV enzyme activities, characterized by hypotonia, global developmental delay, neonatal onset of progressive pectus carinatum without other skeletal abnormalities, poor growth, sensorineural hearing loss, dysmorphic features and brain abnormalities such as cerebral atrophy, quadriventricular dilatation and thin corpus callosum posteriorly.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD25
COXPD25

Kod ORPHA
447954

Kod OMIM
616430

Kod ICD10
E88.8

Kod ICD11
5C53.23

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*Źródło

orphanet