Opis choroby *

Definicja

A rare mitochondrial disease characterized by prenatal or early infantile onset of severe cardiomyopathy, failure to thrive and global developmental delay, sensorineural hearing loss, and severe lactic acidosis. Hepatic involvement and adrenal insufficiency, as well as encephalopathy and anomalies of deep gray matter structures on brain MRI have also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
QRSL1-related COXPD
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Kod ORPHA
570491

Kod OMIM
618835

Kod ICD10
E88.8

Kod ICD11
5C53.23

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*Źródło

orphanet