Opis choroby *

Definicja

A rare mitochondrial disease characterized by failure to thrive, infantile encephalopathy, muscular hypotonia, global developmental delay and regression, pulmonary arterial hypertension, episodes of apnea and bradycardia, respiratory failure, hyperglycinemia, and lactic acidosis. Hypertrophic or dilated cardiomyopathy have also been reported. Brain imaging may show leukoencephalopathy involving variable regions. The disease is typically fatal in early infancy.

Dane

Klasyfikacja
Choroba

Synonimy
MMDS1
Niedobór NFU1
NFU1 deficiency

Kod ORPHA
401869

Kod OMIM
605711

Kod ICD10
E88.8

Kod ICD11
5C53.21

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*Źródło

orphanet