Opis choroby *

Definicja

A rare immune disease characterized by severely reduced mitochondrial DNA content due to DGUOK deficiency typically manifesting with early-onset liver dysfunction, psychomotor delay, hypotonia, rotary nystagmus that develops into opsoclonus, lactic acidosis and hypoglycemia.

Dane

Klasyfikacja
Choroba

Kod ORPHA
279934

Kod OMIM
251880

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet