

## 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