

## Opis choroby \*

### Definicja

A rare mitochondrial disease characterized by neonatal onset of severe cardiac and/or neurologic signs and symptoms mostly associated with a fatal outcome in the neonatal period or in infancy, although a milder phenotype with later onset and slowly progressive neurologic deterioration has also been reported. Clinical manifestations are variable and include respiratory insufficiency, hypotonia, cardiomyopathy, and seizures. Serum lactate is elevated in most cases. Brain imaging may show cerebellar atrophy or hypoplasia.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

COQ4-related neonatal encephalomyopathy

Encefalopatia noworodków związana z COQ4

#### Kod ORPHA

457185

#### Kod OMIM

616276

#### Kod ICD10

E88.8

#### Kod ICD11

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#### \*Źródło

orphanet