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