Opis choroby *

Definicja

A rare, genetic, mitochondrial oxidative phosphorylation disorder characterized by a highly variable phenotype which ranges from a fatal neonatal/infantile encephalomyopathy with lactic acidosis, hyporeflexia/areflexia, severe hypotonia and respiratory failure to less severe cases presenting with central hypotonia, global developmental delay, congenital sensorineural hearing loss, and renal disease. Additional, variably observed, clinical features include intellectual disability, seizures, and cardiomyopathy.

Dane

Klasyfikacja
Choroba

Synonimy
COXPD11
COXPD11

Kod ORPHA
324535

Kod OMIM
614922

Kod ICD10
E88.8

Kod ICD11
-

---

*Źródło

orphanet