Opis choroby *

Definicja

A rare mitochondrial disease characterized by prenatal complications including oligohydramnios, fetal growth restriction, hydrops, and anemia, followed by severe lactic acidosis, hyaline membrane disease, pulmonary hypertension, cardiac anomalies, liver dysfunction, urogenital abnormalities and progressive renal disease, seizures, thrombocytopenia, and sideroblastic anemia resulting in multisystem organ failure and death shortly after birth. Less severely affected patients surviving the neonatal period and showing sensorineural hearing loss and developmental delay have been reported.

Dane

Klasyfikacja
Choroba

Kod ORPHA
528091

Kod OMIM
617021

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet