Opis choroby *

Definicja

Mitochondrial DNA depletion syndrome, hepatocerebrorenal form is a rare, genetic, mitochondrial DNA depletion syndrome characterized by neonatal or early-infantile onset hepatopathy (manifesting with hepatomegaly, cholestasis, increased transaminases, coagulopathy, hypoalbuminemia, ascites, and/or liver failure), associated with renal tubulopathy and progressive neurodegenerative manifestations, which include muscular atrophy, hyporeﬂexia, ataxia, sensory neuropathy, epilepsy, sensorineural hearing impairment, psychomotor regression, athetosis, nystagmus, and/or ophthalmoplegia. Patients typically present with recurrent vomiting, severe failure to thrive, feeding difficulties, and fasting hypoglycemia.

Dane

Klasyfikacja
Choroba

Synonimy
mtDNA depletion syndrome, hepatocerebrorenal form
Zespół deplecji mtDNA, postać wątrobowo-mózgowo-nerkowa

Kod ORPHA
363534

Kod OMIM
271245

Kod ICD10
E88.8

Kod ICD11
5C53.20

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*Źródło

orphanet