

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, hyporexia, 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

[*Źródło](#)

orphanet