Opis choroby *

Definicja

A rare, genetic disorder of urea cycle metabolism characterized by either a neonatal-onset with manifestations of lethargy, poor feeding, vomiting and tachypnea or, more commonly, presentations in infancy, childhood or adulthood with chronic neurocognitive deficits, acute encephalopathy and/or coagulation defects or other chronic liver dysfunction.

Dane

Klasyfikacja
Choroba

Synonimy
HHH syndrome
Niedobór nośnika ornityny
Niedobór ORNT1
Zespół HHH
Zespół potrójnego H
ORNT1 deficiency
Ornithine carrier deficiency
Ornithine translocase deficiency
Triple H syndrome

Kod ORPHA
415

Kod OMIM
238970

Kod ICD10
E72.4

Kod ICD11
5C50.AY

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*Źródło

orphanet