

Opis choroby *

Definicja

An extremely rare metabolic disorder described in fewer than 10 patients to date and characterized by variable signs and symptoms, mostly in infancy, including transient failure to thrive, slightly prolonged neonatal jaundice, equivocal or mild hepatomegaly, microcytic anemia, frequent upper respiratory infections, gastroenteritis, dehydration and flat and coarse facies. Learning difficulties and seizures may occur in childhood.

Dane

Klasyfikacja

Choroba

Synonimy

Sialuria, French type

Sialuria, typ francuski

Kod ORPHA

3166

Kod OMIM

269921

Kod ICD10

E77.8

Kod ICD11

5C56.4

*Źródło

orphanet