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

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*Źródło

orphanet