

## 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