

## Opis choroby \*

### Definicja

Congenital bile acid synthesis defect type 1 (BAS defect type 1) is the most common anomaly of bile acid synthesis (see this term) characterized by variable manifestations of progressive cholestatic liver disease, and fat malabsorption.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

3-beta-hydroxy-delta-5-C27-steroid

oxidoreductase deficiency

BASD1

Niedobór oksydoreduktazy 3-beta-hydroksy-  
delta-5-C27-steroidowej

BASD1

#### Kod ORPHA

79301

#### Kod OMIM

607765

#### Kod ICD10

K76.8

#### Kod ICD11

5C52.11

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#### \*Źródło

orphanet