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