

Opis choroby *

Definicja

Congenital bile acid synthesis defect type 3 (BAS defect type 3) is a severe anomaly of bile acid synthesis (see this term) characterized by severe neonatal cholestatic liver disease.

Dane

Klasyfikacja

Choroba

Synonimy

BASD3

BASD3

Niedobór 7-alfa-hydroksylazy oksysterolowej

Oxysterol 7-alpha-hydroxylase deficiency

Kod ORPHA

79302

Kod OMIM

613812

Kod ICD10

K76.8

Kod ICD11

5C52.11

*Źródło

orphanet