

Opis choroby *

Definicja

Congenital bile acid synthesis defect type 2 (BAS defect type 2) is an anomaly of bile acid synthesis (see this term) characterized by severe and rapidly progressive cholestatic liver disease, and malabsorption of fat and fat-soluble vitamins.

Dane

Klasyfikacja

Choroba

Synonimy

BASD2

BASD2

Cholestaza z niedoborem delta(4)-3-oksosteroido 5-beta-reduktazy

Cholestasis with delta(4)-3-oxosteroid 5-beta-reductase deficiency

Kod ORPHA

79303

Kod OMIM

235555

Kod ICD10

K76.8

Kod ICD11

5C52.11

*Źródło

orphanet