

Opis choroby *

Definicja

Congenital bile acid synthesis defect type 4 (BAS defect type 4) is an anomaly of bile acid synthesis (see this term) characterized by mild cholestatic liver disease, fat malabsorption and/or neurological disease.

Dane

Klasyfikacja

Choroba

Synonimy

2-methylacyl-CoA racemase deficiency

BASD4

Niedobór AMACR

Niedobór racemazy 2-metyloacylo-CoA

Niedobór racemazy alfa-metylo-acylo-CoA

Zespół choroby wątroby, barwnikowego
zwyrodnienia siatkówki, polineuropatii i
padaczka

AMACR deficiency

Alpha-methyl-acyl-CoA racemase deficiency

BASD4

Liver disease-retinitis pigmentosa-
polyneuropathy-epilepsy syndrome

Kod ORPHA

79095

Kod OMIM

614307

Kod ICD10

K76.8

Kod ICD11

5C52.11

*Źródło

orphanet