

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	Synonimy
Choroba	2-methylacyl-CoA racemase deficiency BASD4 Niedobór AMACR Niedobór racemazy 2-metyloacylo-CoA Niedobór racemazy alfa-metylo-acylo-CoA Zespół choroby wątroby, barwnikowego zwydrodnienia 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