Opis choroby *

Definicja

A group of sterol metabolism disorders due to enzyme deficiencies of bile acid synthesis (BAS) in infants, children and adults, with variable manifestations that include cholestasis, neurological disease, and fat malabsorption. Nine inborn errors have been described, 7 of which lead to liver cholestasis.

Dane

Klasyfikacja
Kategoria

Kod ORPHA
79168

Kod OMIM
-

Kod ICD10
-

Kod ICD11
-

---

*Źródło

orphanet