

Opis choroby *

Definicja

A rare, genetic neurometabolic disease characterized biochemically by an almost complete absence of plasma high-density lipoproteins (HDL), and clinically by liver, spleen, lymph node and tonsil enlargement along with multifocal peripheral neuropathy, corneal, skin and nail and, occasionally, cardiovascular disease.

Dane

Klasyfikacja

Choroba

Synonimy

ATP-binding cassette transporter A1 deficiency
Analfalipoproteinemia
Defekt kasety wiążącej trójfosforan adenozyiny
transportera A1
Niedobór kasety wiążącej ATP transportera A1
Analphalipoproteinemia

Kod ORPHA

31150

Kod OMIM

205400

Kod ICD10

E78.6

Kod ICD11

5C81.0

*Źródło

orphanet