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 adenozyny 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