Opis choroby *

Definicja

A rare, progressive metabolic liver disease due to marked to complete lysosomal acid lipase deficiency and characterized by dyslipidemia and massive lipid accumulation leading to hepatomegaly and liver dysfunction, splenomegaly, accelerated atherosclerosis.

Dane

Klasyfikacja
Choroba

Synonimy
LAL deficiency
Niedobór LAL

Kod ORPHA
275761

Kod OMIM
278000

Kod ICD10
E75.5

Kod ICD11
5C56.0Y

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*Źródło

orphanet