

Opis choroby *

Definicja

Familial LCAT (lecithin-cholesterol acyltransferase) deficiency (FLD) is a form of lecithin-cholesterol acyltransferase deficiency (LCAT; see this term) characterized clinically by corneal opacities, hemolytic anemia, and renal failure, and biochemically by severely decreased HDL cholesterol and complete deficiency of the LCAT enzyme.

Dane

Klasyfikacja	Synonimy
Podtyp kliniczny	Complete LCAT deficiency Całkowity Niedobór LCAT Choroba Noruma FLD FLD Norum disease

Kod ORPHA 79293	Kod OMIM 245900	Kod ICD10 E78.6
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Kod ICD11
5C81.0

*Źródło

orphanet