Opis choroby *

Definicja

A severe form of lysosomal acid lipase deficiency characterized by rapidly progressive lipid accumulation in organs and tissues that presents in the neonatal or infantile period with massive hepatosplenomegaly, liver failure, diarrhea/steatorrhea and vomiting.

Dane

Klasyfikacja
Podtyp kliniczny

Kod ORPHA
75233

Kod OMIM
278000

Kod ICD10
E75.5

Kod ICD11
5C56.0Y

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*Źródło

orphanet