

Opis choroby *

Definicja

A form of neutral lipid storage disease characterized by the accumulation of lipid vacuoles in leukocytes (so-called Jordan's anomaly seen in peripheral blood smears) and a variety of other cell types. The clinical picture consists of congenital ichthyosis of the congenital ichthyosiform erythroderma type together with variable multisystem involvement. Manifestations include hepatosplenomegaly, myopathy, intestinal disease, growth retardation, cataracts, sensorineural hearing loss, and intellectual disability, among others.

Dane

Klasyfikacja

Choroba

Synonimy

Dorfman-Chanarin disease

Choroba spichrzania neutralnych tłuszczów z rybią łuską

NLSDI

NLSDI

Kod ORPHA

98907

Kod OMIM

275630

Kod ICD10

E75.5

Kod ICD11

5C52.2

*Źródło

orphanet