DISEASE:
Neutral lipid storage disease with ichthyosis

NAME:	Neutral lipid storage disease with ichthyosis
DESCRIPTION:	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.
ORPHACODE:	98907
SYNONYMS:	Dorfman-Chanarin disease NLSDI
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ABHD5
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