

Opis choroby *

Definicja

A form of neutral lipid storage disease characterized by adult onset of slowly progressive, typically proximal, muscular weakness of the upper and lower limbs, associated with elevated serum creatine kinase. Many patients develop cardiomyopathy later in the disease course. Additional, variable manifestations include hepatomegaly, diabetes mellitus, and hypertriglyceridemia, among others. Diagnostic hallmarks are triacylglycerol-containing lipid vacuoles in leukocytes in peripheral blood smears (so-called Jordans' anomaly), as well as massive accumulation of lipid droplets in muscle tissue.

Dane

Klasyfikacja

Choroba

Synonimy

NLSDM

Choroba spichrzania neutralnych tłuszczów z miopatią bez rybiej łuski

Kardiomiowaskulopatia z odkładaniem się trójglicerydów

NLSDM

Neutral lipid storage disease with myopathy without ichthyosis

Kod ORPHA

98908

Kod OMIM

610717

Kod ICD10

E75.5

Kod ICD11

5C52.2

*Źródło

orphanet