

# **Choroba Niemann i Picka typu C**

## **Kod Orpha: 646 Kod OMIM: 607625**

### **Opis choroby \***

#### **Definicja**

A rare lysosomal lipid storage disease characterized by variable clinical signs, depending on the age of onset, such as prolonged unexplained neonatal jaundice or cholestasis, isolated unexplained splenomegaly, and progressive, often severe neurological symptoms such as cognitive decline, cerebellar ataxia, vertical supranuclear gaze palsy (VSPG), dysarthria, dysphagia, dystonia, seizures, gelastic cataplexy, and psychiatric disorders.

#### **Dane**

#### **Klasyfikacja**

Choroba

**Kod ORPHA**  
646

**Kod OMIM**  
607625

**Kod ICD10**  
E75.2

**Kod ICD11**  
5C56.0Y

---

\*Źródło

orphanet

### **Rozszerzony opis choroby**

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.