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

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*Źródło

orphanet