Opis choroby *

Definicja

A rare, genetic, inborn error of metabolism disorder characterized by global developmental delay, hypotonia, choreoathetosis, hypo-/alacrimia, and liver dysfunction which manifests with elevated liver transanimases and hepatocyte cytoplasmic storage material or vacuolization on liver biopsy. Additional features reported include acquired microcephaly, hypo-/areflexia, seizures, peripheral neuropathy, intellectual and language/speech disability, additional ocular anomalies and EEG and brain imaging abnomalities.

Dane

Klasyfikacja
Choroba

Synonimy
NGLY1 deficiency
NGLY1-CDDG
Niedobór NGLY1
NGLY1-CDDG

Kod ORPHA
404454

Kod OMIM
615273

Kod ICD10
E77.8

Kod ICD11
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*Źródło

orphanet