Opis choroby *

Definicja

A rare congenital disorder of glycosylation characterized by neonatal onset of global developmental delay, hypotonia, failure to thrive, hematological/immunological abnormalities, recurrent infections, liver involvement (with hepatosplenomegaly, cholestasis, fibrosis, or cirrhosis), and enteropathy. Additional reported manifestations include dysmorphic craniofacial features (such as microcephaly, broad palpebral fissures, and retrognathia), hypohidrosis, hyperkeratosis, and cardiac and musculoskeletal anomalies. Brain imaging may show hypoplastic corpus callosum, cerebral and cerebellar atrophy, and enlarged ventricles.

Dane

Klasyfikacja
Choroba

Synonimy
CDG syndrome type IIL
Wrodzone zaburzenia glikozylacji typu 2l
Wrodzone zaburzenia glikozylacji typu Ill
CDG-IIL
CDG2L
Congenital disorder of glycosylation type 2l
Congenital disorder of glycosylation type IIL

Kod ORPHA
464443

Kod OMIM
614576

Kod ICD10
E77.8

Kod ICD11
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*Źródło

orphanet