Opis choroby *

Definicja

A rare congenital disorder of glycosylation characterized by cerebral and portal vein thrombosis, portal hypertension, macrocephaly, and persistent absence seizures. Additional reported features include mild to moderate global developmental delay and intellectual disability, as well as thrombocytopenia. Brain imaging may show variable stages of infarction and cerebral and cerebellar atrophy.

Dane

Klasyfikacja
Choroba

Synonimy
Congenital disorder of glycosylation due to PIGM deficiency
PIGM-CDG
PIGM-CDG

Kod ORPHA
83639

Kod OMIM
610293

Kod ICD10
E88.8

Kod ICD11
3B61.0Y

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*Źródło

orphanet