Opis choroby *

Definicja

MGAT2-CDG is a form of congenital disorders of N-linked glycosylation characterized by facial dysmorphism (large, posteriorly rotated ears with prominent antihelices, convex nasal ridge, open mouth, large and crowded teeth), stereotypic hand movements, seizures, and varying degrees of developmental delay. A bleeding tendency is also observed and this results from diminished platelet aggregation. The disease is caused by loss-of-function mutations in the gene MGAT2 (14q21).

Dane

Klasyfikacja
Choroba

Synonimy
CDG syndrome type IIa
CDG2A
CDG-IIa
Niedobór N-acetyloglukozoaminylotransferazy 2
Wrodzone zaburzenie glikozylacji typu 2a
Wrodzone zaburzenie glikozylacji typu IIa
Zespół CDG typu IIa
Zespół obniżonej glikozylacji glikoprotein typu IIa
CDG-IIa
CDG2A
Carbohydrate deficient glycoprotein syndrome type IIa
Congenital disorder of glycosylation type 2a
Congenital disorder of glycosylation type IIa
N-acetylglucosaminyltransferase 2 deficiency

Kod ORPHA
79329

Kod OMIM
212066

Kod ICD10
E77.8

Kod ICD11
5C54.0

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*Źródło

orphanet