

Opis choroby *

Definicja

A rare congenital disorder of glycosylation characterized by infantile onset of global developmental delay, severe intellectual disability, hypotonia, and variable additional features including short stature, cranial asymmetry, seizures, strabismus, recurrent infections, and osteopenia, among others. Laboratory analysis reveals decreased blood levels of zinc and manganese, as well as an abnormal serum transferrin glycosylation pattern with decreased tetrasialo- and increased asialo-, monosialo-, disialo, and trisialo-transferrin, consistent with a type II congenital disorder of glycosylation. Brain imaging shows cerebellar and/or cerebral atrophy.

Dane

Klasyfikacja

Choroba

Synonimy

CDG syndrome type II n
CDG2N
CDG-II n
Niedobór SLC39A8
Zespół CDG typu II n
Wrodzone zaburzenie glikozylacji typu 2n
Wrodzone zaburzenie glikozylacji typu II n
Zespół obniżonej glikozylacji glikoprotein typ II n
CDG-II n
CDG2N
Carbohydrate deficient glycoprotein syndrome type II n
Congenital disorder of glycosylation type 2n
Congenital disorder of glycosylation type II n
SLC39A8 deficiency

Kod ORPHA

468699

Kod OMIM

616721

Kod ICD10

E77.8

Kod ICD11

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[*Źródło](#)

orphonet