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

Kod ORPHA
468699

Kod OMIM
616721

Kod ICD10
E77.8

Kod ICD11
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*Źródło

orphanet