Opis choroby *

Definicja

A fast growing group of inborn errors of metabolism characterized by defective activity of enzymes that participate in glycosylation (modification of proteins and other macromolecules by adding and processing of oligosaccharide side chains). This group is comprised of phenotypically diverse disorders affecting multiple systems including the central nervous system, muscle function, immunity, endocrine system, and coagulation. The numerous entities in this group are subdivided, based on the synthetic pathway affected, into disorder of protein N-glycosylation, disorder of protein O-glycosylation, disorder of multiple glycosylation, and disorder of glycosphingolipid and glycosylphosphatidylinositol anchor glycosylation.

Dane

Klasyfikacja
Kategoria

Synonimy
CDG
CDG
Zespół niedoboru wodorowęglanu glikoproteiny
Carbohydrate deficient glycoprotein syndrome

Kod ORPHA
137

Kod OMIM
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Kod ICD10
E77.8

Kod ICD11
-

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*Źródło

orphanet