Opis choroby *

Definicja

DPAGT1-CDG is a form of congenital disorders of N-linked glycosylation characterized by hypotonia, intractable seizures, developmental delay, microcephaly and severe fetal hypokinesia. Additional features that may be observed include apnea and respiratory deficiency, cataracts, joint contractures, vermian hypoplasia, dysmorphic features (esotropia, arched palate, micrognathia, finger clinodactyly, single flexion creases) and feeding difficulties. The disease is caused by loss-of-function mutations in the gene DPAGT1 (11q23.3).

Dane

Klasyfikacja
Choroba

Synonimy
CDG syndrome type Ij
CDG1J
CDG-Ij
Niedobór fosfotransferazy dolicholofosforanu N-acetylogalaktozoaminy
Wrodzone zaburzenie glikozylacji 2j
Wrodzone zaburzenie glikozylacji IIj
Zespół CDG typu Ij
Zespół obniżonej glikozylacji glikoprotein typu Ij
CDG-Ij
CDG1J
Carbohydrate deficient glycoprotein syndrome type Ij
Congenital disorder of glycosylation type 1j
Congenital disorder of glycosylation type Ij
Dolichyl-phosphate N-acetylgalactosamine phosphotransferase deficiency

Kod ORPHA
86309

Kod OMIM
608093

Kod ICD10
E77.8

Kod ICD11
5C54.0

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*Źródło

orphanet