Opis choroby *

Definicja

SSR4-CDG is a form of congenital disorders of N-linked glycosylation characterized by neurologic abnormalities (global developmental delay in language, social skills and fine and gross motor development, intellectual disability, hypotonia, microcephaly, seizures/epilepsy), facial dysmorphism (deep set eyes, large ears, hypoplastic vermillion of upper lip, large mouth with widely spaced teeth), feeding problems often due to chewing difficulties and aversion to food with certain textures, failure to thrive, gastrointestinal abnormalities (reflux or vomiting) and strabismus. The disease is caused by mutations in the gene SSR4 (Xq28).

Dane

Klasyfikacja
Choroba

Synonimy
CDG syndrome type ly
CDG1Y
CDG-ly
Wrodzone zaburzenie glikozylacji typu 1y
Wrodzone zaburzenie glikozylacji typu ly
Zespół CDG typu ly
Zespół obniżonej glikozylacji glikoprotein typu ly
CDG-ly
CDG1Y
Carbohydrate deficient glycoprotein syndrome type ly
Congenital disorder of glycosylation type 1y
Congenital disorder of glycosylation type ly

Kod ORPHA
370927

Kod OMIM
300934

Kod ICD10
E77.8

Kod ICD11
5C54.0

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*Źródło

orphanet