

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 *< i> SSR4</i>* (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

*[Źródło](#)

orphanet

