Opis choroby *

Definicja

A rare congenital disorder of glycosylation characterized by early onset of hypotonia, severe global developmental delay, intellectual disability, and seizures. Ataxia, mild facial dysmorphism, and autistic behavior have also been reported. Brain MRI findings are variable and include cerebral atrophy, cerebellar hypoplasia/atrophy, and thin corpus callosum.

Dane

Klasyfikacja
Choroba

Synonimy
Congenital disorder of glycosylation due to PIGG deficiency
Wrodzone zaburzenie glikolizacji z powodu niedoboru PIGG
PIGG-CDG

Kod ORPHA
488635

Kod OMIM
616917

Kod ICD10
E77.8

Kod ICD11
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*Źródło

orphanet