Opis choroby *

Definicja

A rare, congenital disorder of glycosylation caused by mutations in the COG2 gene and characterized by normal presentation at birth, followed by progressive deterioration with postnatal microcephaly, developmental delay, intellectual disability, seizures, spastic quadriplegia, liver dysfunction, hypocupremia and hypoceruloplasminemia in the first year of life. Diffuse cerebral atrophy and thin corpus callosum may be observed on brain MRI.

Dane

Klasyfikacja
Choroba

Synonimy
COG2-related congenital disorder of glycosylation
Wrodzone zaburzenie glikozylacji związane z COG2

Kod ORPHA
435934

Kod OMIM
617395

Kod ICD10
E77.8

Kod ICD11
5C54.2

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*Źródło

orphanet