Opis choroby *

Definicja

A form of congenital disorders of N-linked glycosylation characterized by distal arthrogryposis (mild flexion contractures of the fingers, deviation of the distal phalanges, swan-neck deformity), retromicrognathia, general muscle hypotonia, delayed psychomotor development, autism spectrum disorder (speech delay, abnormal use of speech, difficulties in initiating, understanding and maintaining social interaction, limited non-verbal communication and repetitive behavior), seizures, microcephaly and mild to moderate intellectual disability that becomes apparent with age.

Dane

Klasyfikacja
Choroba

Synonimy
SLC35A3-CDG
SLC35A3-CDG

Kod ORPHA
370943

Kod OMIM
615553

Kod ICD10
Q87.8

Kod ICD11
5C54.0

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*Źródło

orphanet