

## Opis choroby \*

### Definicja

A form of congenital disorders of N-linked glycosylation characterized by distal arthrogyrosis (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

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#### Kod ORPHA

370943

#### Kod OMIM

615553

#### Kod ICD10

Q87.8

#### Kod ICD11

5C54.0

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#### \*Źródło

orphanet