

## Opis choroby \*

### Definicja

STT3B-CDG is a form of congenital disorders of N-linked glycosylation characterized by intrauterine growth retardation, microcephaly, failure to thrive, developmental delay, intellectual disability, hypotonia, seizures, optic nerve atrophy and respiratory difficulties. Genital abnormalities (micropenis, hypoplastic scrotum, undescended testes) have also been reported. STT3B-CDG is caused by mutations in the gene *STT3B* (3p24.1).

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

CDG syndrome type Ix  
Carbohydrate deficient glycoprotein syndrome type Ix  
CDG1X  
CDG-Ix  
Wrodzone zaburzenie glikozylacji typu 1x  
Wrodzone zaburzenie glikozylacji typu Ix  
Zespół CDG typu Ix  
CDG-Ix  
CDG1X  
Carbohydrate deficient glycoprotein syndrome type Ix  
Congenital disorder of glycosylation type 1x  
Congenital disorder of glycosylation type Ix

#### Kod ORPHA

370924

#### Kod OMIM

615597

#### Kod ICD10

E77.8

#### Kod ICD11

5C54.0

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#### \*Źródło

orphanet