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