DISEASE:
STT3B-CDG

NAME:	STT3B-CDG
DESCRIPTION:	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).
ORPHACODE:	370924
SYNONYMS:	CDG syndrome type Ix CDG-Ix CDG1X Carbohydrate deficient glycoprotein syndrome type Ix Congenital disorder of glycosylation type 1x Congenital disorder of glycosylation type Ix
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	STT3B
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