Open Peer Review on Qeios

STT3B-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>STT3B-</u> <u>CDG</u>. ORPHA:370924

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).