RFT1-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. RFT1-CDG. ORPHA:244310

RFT1-CDG is a form of congenital disorders of N-linked glycosylation characterized by poorly coordinated suck resulting in difficulty feeding and failure to thrive; myoclonic jerks with hypotonia and brisk reflexes progressing to a seizure disorder; roving eyes; developmental delay; poor to absent visual contact; and sensorineural hearing loss. Additional features that may be observed include coagulation factor abnormalities, inverted nipples and microcephaly. The disease is caused by mutations in the gene RFT1 (3p21.1).