DISEASE:
MAN1B1-CDG

NAME:	MAN1B1-CDG
DESCRIPTION:	MAN1B1-CDG is a form of congenital disorders of N-linked glycosylation characterized by intellectual disability, delayed motor development, hypotonia and truncal obesity. Additional features include slight facial dysmorphism (hypertelorism, downslanting palpebral fissures, large, low-set ears, hypoplastic nasolabial fold, thin upper lip), hypermobility of the joints and skin laxity. The disease is caused by mutations in the gene MAN1B1 (9q34.3).
ORPHACODE:	397941
SYNONYMS:	Carbohydrate deficient glycoprotein syndrome type II due to MAN1B1 deficiency Congenital disorder of glycosylation type 2 due to MAN1B1 deficiency Congenital disorder of glycosylation type II due to MAN1B1 deficiency Intellectual disability-truncal obesity syndrome
XREF(S):	Orphanet ICD-10
ANALYTE(S):	MAN1B1
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