

DISEASE:
MOGS-CDG

NAME:	MOGS-CDG
DESCRIPTION:	MOGS-CDG is a form of congenital disorders of N-linked glycosylation characterized by generalized hypotonia, craniofacial dysmorphism (prominent occiput, short palpebral fissures, long eyelashes, broad nose, high arched palate , retrognathia), hypoplastic genitalia, seizures, feeding difficulties, hypoventilation, severe hypogammaglobulinemia with generalized edema, and increased resistance to particular viral infections (particularly to enveloped viruses). The disease is caused by loss-of-function mutations in the gene MOGS (2p13.1).
ORPHACODE:	79330
SYNONYMS:	CDG syndrome type IIb CDG-IIb CDG2B Carbohydrate deficient glycoprotein syndrome type IIb Congenital disorder of glycosylation type 2b Congenital disorder of glycosylation type IIb Glucosidase 1 deficiency
XREF(S):	Orphanet OMIM ICD-10

ANALYTE(S):	<u>MOGS</u>
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