DISEASE:
PGM3-CDG

NAME:	PGM3-CDG
DESCRIPTION:	PGM3-CDG is a rare congenital disorder of glycosylation caused by mutations in the PGM3 gene and characterized by neonatal to childhood onset of recurrent bacterial and viral infections, inflammatory skin diseases, atopic dermatitis and atopic diatheses, and marked serum IgE elevation. Early neurologic impairment is evident including developmental delay, intellectual disability, ataxia, dysarthria, sensorineural hearing loss, myoclonus and seizures.
ORPHACODE:	443811
SYNONYMS:	CID due to PGM3 deficiency Combined immunodeficiency due to PGM3 deficiency PGM3-related congenital disorder of glycosylation
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PGM3
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