

DISEASE:

X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia

NAME:	X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia
DESCRIPTION:	X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia is a rare combined T and B cell immunodeficiency characterized by recurrent sinopulmonary and viral infections, persistent elevated Epstein-Barr virus (EBV) viremia and increased susceptibility to EBV-associated B-cell lymphoproliferative disorders. Immunological analyses show normal lymphocyte count or mild to moderate lymphopenia, inverted CD4:CD8 T-cell ratio and hypogammaglobulinemias.
ORPHACODE:	317476
SYNONYMS:	CID due to MAGT1 deficiency Combined immunodeficiency due to MAGT1 deficiency XMEN
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	MAGT1
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