DISEASE:
X-linked lymphoproliferative disease due to SH2D1A deficiency

NAME:	X-linked lymphoproliferative disease due to SH2D1A deficiency
DESCRIPTION:	A rare, genetic, primary immunodeficiency disorder characterized by an abnormal immune response to Epstein-Barr virus (EBV) infection, caused by hemizygous mutations in the X-linked SH2D1A gene, resulting in B cell lymphoproliferation and manifesting with various phenotypes which include EBV-driven severe or fulminant mononucleosis, hemophagocytic lymphohistiocytosis (presenting with fulminant hepatitis, hepatic necrosis, bone marrow hypoplasia, and neurological involvement), hypogammaglobulinemia, and B-cell lymphoma. Additional variable manifestations include vasculitis, lymphomatoid granulomatosis, aplastic anemia, and chronic gastritis. Occasionally, T-cell lymphoma may be observed. Laboratory findings include normal or increased activated T cells and reduced memory B cells.
ORPHACODE:	538931
SYNONYMS:	SAP deficiency SH2D1A/SLAM-associated protein deficiency X-linked lymphoproliferative syndrome type 1 XLP1
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SH2D1A

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RELATED CONTENT

Related Genetic Tests

- Lymphoproliferative syndrome, X-linked (SH2D1A gene) / Duncan's disease
- Primary immune deficiencies (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- SH2 domain containing 1A

Related Gene Panels

- Immunogenetics (21 genes)
- Primary immune deficiencies (444 genes) - KUL