

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Synonimy

SAP deficiency

SH2D1A/SLAM-associated protein deficiency

X-linked lymphoproliferative syndrome type 1

XLP1

SAP deficiency

SH2D1A/SLAM-associated protein deficiency

X-linked lymphoproliferative syndrome type 1

XLP1

Kod ORPHA

538931

Kod OMIM

308240

Kod ICD10

D82.3

Kod ICD11

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orphanet