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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*Źródło

orphanet