

# Choroba limfoproliferacyjna sprzężona z chromosomem X spowodowana niedoborem SH2D1A

## Kod Orpha: 538931 Kod OMIM: 308240

### 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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## **Rozszerzony opis choroby**

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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