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 XIAP gene, resulting in B cell lymphoproliferation and manifestating with various phenotypes which include EBV-driven hemophagocytic lymphohistiocytosis, hypogammaglobulinemia, recurrent splenomegaly, hepatitis, colitis, and intestinal bowel disease with features of Crohn's disease. Additional manifestations include variable auto-inflammatory symptoms such as uveitis, arthritis, skin abscesses, erythema nodosum, and nephritis. Neurological involvement is rare and lymphoma is never observed. Laboratory findings include normal or increased activated T cells, low or normal iNKT cells, and normal or reduced memory B cells.

Dane

Klasyfikacja
Choroba

Synonimy
X-linked lymphoproliferative syndrome type 2
XIAP deficiency syndrome
XLP2
X-linked lymphoproliferative syndrome type 2
XIAP deficiency syndrome
XLP2

Kod ORPHA
538934

Kod OMIM
300635

Kod ICD10
D82.3

Kod ICD11
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*Źródło

orphanet