

# Choroba limfoproliferacyjna sprzężona z chromosomem X spowodowana niedoborem XIAP

## Kod Orpha: 538934 Kod OMIM: 300635

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

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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