Opis choroby *

Definicja

A rare autosomal recessive primary immunodeficiency characterized by susceptibility to Epstein-Barr virus (EBV)-associated lymphoproliferative disorders such as malignant B-cell proliferation, Hodgkin lymphoma, B-cell lymphoma, lymphoid granulomatosis, hemophagocytic lymphohistiocytosis, and smooth muscle tumor. Patients present persistent symptoms of infectious mononucleosis including recurrent febrile episodes, lymphadenopathies, and hepatosplenomegaly, accompanied by high EBV viral load in the blood. Additional manifestations are autoimmune diseases like hemolytic anemia or renal disease.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive lymphoproliferative disease due to ITK deficiency
Autosomal recessive lymphoproliferative disease due to ITK deficiency
ITK deficiency

Kod ORPHA
538963

Kod OMIM
613011

Kod ICD10
D82.3

Kod ICD11
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*Źródło

orphanet