Opis choroby *

Definicja

A rare autosomal recessive primary immunodeficiency characterized by Epstein-Barr virus (EBV)-triggered lymphoprolipherative disorders such as malignant B-cell proliferation, Hodgkin lymphoma, B-cell lymphoma and EBV-driven hemophagocytic lymphohistiocytosis (HLH). Aplastic anemia and inflammatory disorders such as uveitis and oral ulcers are also observed.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive lymphoproliferative disease due to CD27 deficiency
CD27 deficiency
Autosomal recessive lymphoproliferative disease due to CD27 deficiency
CD27 deficiency

Kod ORPHA
238505

Kod OMIM
615122

Kod ICD10
D47.9

Kod ICD11
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*Źródło

orphanet