Opis choroby *

Definicja

A rare, genetic, primary immunodeficiency disease characterized by increased susceptibility to recurrent and/or severe bacterial and viral infections (in particular, sinopulmonary bacterial and herpesvirus infections), chronic benign lymphoproliferation (manifesting as lymphadenopathy, hepatosplenomegaly and focal nodular lymphoid hyperplasia), and/or autoimmune disease (including immune cytopenias, juvenile arthritis, glomerulonephritis and sclerosing cholangitis). Immunophenotypically, variable degrees of agammaglobulinemia with increased IgM levels, increased circulating transitional B cells, decreased naïve CD4 and CD8 T-cells with increased CD8 effector/memory T cells are observed.

Dane

Klasyfikacja
Choroba

Synonimy
APDS
APDS
Zespół starzejących się komórek T, limfadenopatii i niedoboru odporności spowodowany mutacją aktywności p110delta
Senescent T-cells-lymphadenopathy-immunodeficiency syndrome due to p110delta-activating mutation

Kod ORPHA
397596

Kod OMIM
616005

Kod ICD10
D81.8

Kod ICD11
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*Źródło

orphanet