

## 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

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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