Opis choroby *

Definicja

A rare, genetic, combined T and B cell immunodeficiency characterized by T- and B-cell lymphopenia, hypergammaglobulinemia and intermittent neutropenia. It presents with recurrent opportunistic viral, bacterial and fungal infections involving skin (cutaneous papillomatosis, molluscum contagiosum, skin abscesses, mucocutaneous candidiasis), upper and lower respiratory tract or septicemia. Other clinical features include autoimmune manifestations (autoimmune hemolytic anemia) and congenital heart defects (atrial septal defects, patent foramen ovale, mitral, triscupid and pulmonary valve insufficiency).

Dane

Klasyfikacja
Choroba

Synonimy
CID due to STK4 deficiency
CID z powodu niedoboru STK4

Kod ORPHA
314689

Kod OMIM
614868

Kod ICD10
D81.8

Kod ICD11
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*Źródło

orphanet