

Opis choroby *

Definicja

A rare, genetic primary immunodeficiency characterized by recurrent respiratory and skin viral infections (Epstein-Barr virus, herpes simplex virus, human papillomavirus), deficient spontaneous cytotoxicity of natural killer cells, but preserved antibody-dependent cellular cytotoxicity. No other abnormalities are present on immunologic work-up.

Dane

Klasyfikacja

Choroba

Synonimy

Autosomal recessive primary immunodeficiency
with defective spontaneous NK cell cytotoxicity
Autosomalny recesywny pierwotny Niedobór
odporności ze spontanicznym defektem
cytotoksyczności komórek NK
CD16 deficiency

Kod ORPHA

437552

Kod OMIM

615707

Kod ICD10

D84.8

Kod ICD11

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*Źródło

orphanet