

Niedobór DOCK2

Kod Orpha: 447737 Kod OMIM: 616433

Opis choroby *

Definicja

A rare, primary combined T and B cell immunodeficiency characterized by early-onset of recurrent, invasive viral and bacterial infections associated with T and B cell lymphopenia, functional defects in T and B cells, poor antibody response and thrombocytopenia.

Depending on the type of infectious agent, variable clinical manifestations commonly include recurrent pneumonia, bronchiolitis, otitis media, meningoencephalitis, colitis, and diarrhea, leading to fatal multiorgan failure in severe cases.

Dane

Klasyfikacja

Choroba

Kod ORPHA

447737

Kod OMIM

616433

Kod ICD10

D81.8

Kod ICD11

4A01.1Y

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.