

Opis choroby *

Definicja

Hyper-IgM syndrome with susceptibility to opportunistic infections is a rare, genetic, non-severe combined immunodeficiency disorder characterized by normal or elevated IgM serum levels with low or absent IgG, IgA and IgE serum concentrations, which manifests with recurrent or severe bacterial infections and increased susceptibility to opportunistic infections (in particular, pneumonia due to *P. jirovecii*, but also chronic cryptosporidial, cryptococcal, cytomegalovirus and toxoplasma infections). Hematologic disorders (neutropenia, anemia, thrombocytopenia) are frequently associated. Immunologic findings reveal decreased numbers of CD27+ memory B cells and lack of germinal center formation.

Dane

Klasyfikacja

Choroba

Synonimy

HIGM with susceptibility to opportunistic infections
HIGM z podatnością na zakażenia oportunistyczne

Kod ORPHA

183663

Kod OMIM

606843

Kod ICD10

D80.5

Kod ICD11

4A01.1Y

*Źródło

orphanet