Opis choroby *

Definicja

Hyper-IgM syndrome without susceptibility to opportunistic infections is a rare, genetic, primary immunodeficiency due to a defect in adaptive immunity disorder characterized by normal or elevated IgM serum levels with low or absent IgG, IgA and IgE serum concentrations, which manifests with recurrent bacterial sinopulmonary and gastrointestinal infections, with frequent lymphoid hyperplasia (peripheral lymphadenopathy, tonsillar hypertrophy), with no increased susceptibility to opportunistic infections. Autoimmune manifestations (including immune cytopenias, arthritis and hepatitis) are occasionally associated. Immunologic findings reveal absent immunoglobulin class switch recombination and lack of defect of immunoglobulin somatic hypermutations in the presence of normal numbers of CD27+ memory B cells.

Dane

Klasyfikacja
Choroba

Synonimy
HIGM without susceptibility to opportunistic infections
HIGM bez podatności na zakażenia oportunistyczne

Kod ORPHA
183666

Kod OMIM
608184

Kod ICD10
D80.5

Kod ICD11
4A01.1Y

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*Źródło

orphanet