Hyper-IgM syndrome without susceptibility to opportunistic infections

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hyper-IgM syndrome without susceptibility to opportunistic infections. ORPHA:183666

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.