Combined immunodeficiency with granulomatosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined immunodeficiency with granulomatosis. ORPHA:157949

A rare, genetic, non-severe combined immunodeficiency disease characterized by immunodeficiency (manifested by recurrent and/or severe bacterial and viral infections), destructive noninfectious granulomas involving skin, mucosa and internal organs, and various autoimmune manifestations (including cytopenias, vitiligo, psoriasis, myasthenia gravis, enteropathy). Immunophenotypically, T-cell and B-cell lymphopenia, hypogammaglobulinemia, abnormal specific antibody production and impaired T-cell function are observed.