Autoimmunity in Immunodeficiency

A 1-year-oldboy presented with recurrent attacks of severe life threatening infections since 5 months of age along with persistent diarrhea for last 3 months. He was visibly malnourished And had generalised erythroderma with scaling Immunophenotyping from peripheral blood showed very low CD4+ and CD8+ count (9/μL and 94/μL, respectively); CD4/CD8 ratio was 0.09. Immunoglobulin (IgM: 6 mg/100 mL;, IgA: 6 mg/100 mL, IgG:110 mg/100 mL: IgE :5.7I.U/mL) levels were below normal suggesting a diagnosis of severe combined immunodeficiency (SCID). This patient developed high fever, hepatosplenomegaly and pancytopenia (total leukocyte count 0.3 $\times 10^3$/μL, neutrophil 12 %, platelet count 78 $\times 10^3$/iL, hemoglobin 6.8 g/dL. Fasting triglycerides (342 mg/dL) and serum ferritin (9240 ng/mL) were raised, and bone marrow showed hemophagocytosis. Intravenous

immunoglobulins were started along with antibiotics. General condition of the patient deteriorated and the patient expired in next few days.

Although it sounds paradoxical, an autoimmune phenomenon can complicate a pre-existing primary immunodeficiency disorder, thereby creating a diagnostic and therapeutic challenge to the caring physician. Cases like Autoimmune thrombocytopenia and Autoimmune hemolytic anemia in Common variable immunodeficiency [1], and Omen syndrome and Autoimmune thrombocytopenia in SCID [2] have been reported. Impairment of both central and peripheral tolerance is responsible for autoimmunity observed in SCID [3]. Treatment with immunosuppressive agents such as corticosteroids can exacerbate the infections associated with immunodeficiency disorders. So non-immunosuppressive agents such as Intravenous immunoglobulins and targeted monoclonal antibodies are likely to be preferable [4].

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