STAT3-related early-onset multisystem autoimmune disease

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. STAT3-related early-onset multisystem autoimmune disease. ORPHA:438159

A rare, genetic, lypmhoproliferative syndrome characterized by early onset recurrent infections, lymphadenopathy with hepatosplenomegaly and variabe autoimmune disorders, including hemolytic anemia, thrombocytopenia, neutropenia, enteropathy, type I diabetes, scleroderma, arthritis, atopic dermatitis, and inflammatory lung disease. Patients commonly have failure to thrive. Variable immunologic findings include decreased regulatory T-cells, hypogammaglobulinemia, and reduction in memory B cells.