Type 1a Autoimmune Lymphoproliferative Syndrome

National Cancer Institute

Source

National Cancer Institute. Type 1a Autoimmune Lymphoproliferative Syndrome. NCI Thesaurus. Code C39574.

A rare, primary immunodeficiency with an autosomal dominant pattern of inheritance but variable penetrance. It is the most common subtype of autoimmune lymphoproliferative syndrome (ALPS). It is usually caused by a germline mutation in the Fas gene that leads to defective Fas-induced apoptosis but in a minority of cases, it also may be attributed to a somatic Fas mutation. Disruption of Fas-induced apoptosis impairs lymphocyte homeostasis and immune tolerance. Characteristic laboratory findings include an increase in circulating, double-negative (CD4-/CD8-) T cells in the setting of immune-mediated anemia, thrombocytopenia and neutropenia. Clinical signs present in childhood include fatigue, pallor, bruising, hepatosplenomegaly and chronic, non-malignant, non-infectious lymphadenopathy. The clinical course is influenced by a strong association with other autoimmune disorders and an increased risk for developing Hodgkin and non-Hodgkin lymphoma.