Autoimmune Lymphoproliferative Syndrome: Overcoming Diagnostic Hurdles in a Tertiary Care Centre in Eastern India

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Autoimmune Lymphoproliferative Syndrome (ALPS) affects lymphocyte regulation due to defects in the FAS apoptotic pathway. It is marked by non-malignant lymphoproliferation, autoimmune disorders—especially multilineage cytopenias—and an elevated risk of B-cell lymphoma. Most patients harbour heterozygous germline mutations in the gene for the TNF receptor-family member Fas (CD 95, Apo-1) which are inherited in an autosomal dominant fashion. Somatic Fas mutations are the second most common genetic etiology of ALPS. We encountered three cases at our center with recurring skin issues, upper respiratory tract symptoms, and prolonged fever. Upon examination, these patients have multiple clusters of enlarged lymph nodes, splenomegaly, and persistent, unexplained cytopenias. Flow cytometry of peripheral blood samples revealed a substantial population of double-negative CD3+ T-lymphocytes. According to the updated diagnostic criteria, all the cases fulfilled the criteria for a "Probable Diagnosis" of ALPS. However due to the financial constraint further genetic workup could not be done to confirm the mutations. The children were started on immunosuppressants which led to symptomatic relief.