

Case Presentation

1- **4 year old girl** presents with severe **anemia** and splenomegaly. Hb: 5 g/dL; Hct 16.6%; plt 82×10^9 ; WBC: 2.3×10^9 Reticulocytosis, high LDH, and indirect bilirubin.

2- Direct antiglobulin test (DAT) was negative. Osmotic gradient **ektacytometry** was compatible with decreased surface-to-volume ratio.

3- IVIG elicited a good response and a diagnoses of warm-ab **autoimmune** hemolytic anemia, was made.

4- Immunophenotype revealed increased **double-negative** T cells (DNTCs). c.755delA mutation was identified in FAS.

5- **Sirolimus** was targeted to a therapeutic dosage, with great response.

Autoimmune lymphoproliferative syndrome presenting as autoimmune hemolytic anemia

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Autoimmune Lymphoproliferative Syndrome: A Case Presentation

Autoimmune lymphoproliferative syndrome (ALPS) is a disorder of lymphocyte homeostasis caused by defects in the **FAS pathway**.

We present a case of a preschooler with warm-ab autoimmune hemolytic anemia. She had increased double-negative T cells (DNTCs) and a pathogenic FAS mutation.

The FAS gene is associated with **autosomal dominant** autoimmune lymphoproliferative syndrome (ALPS-FAS). Phenotype is highly variable. Worse presentations are present with biallelic mutations.

The risk for non **Hodgkin lymphoma** is 60 times higher compared to healthy individuals, and more than 150 times higher for Hodgkin lymphoma.

There is increased risk of relatives being carriers. Parental testing may clarify the inheritance of FAS mutations.

ALPS should be suspected in children with lymphadenopathy, splenomegaly and cytopenias. **Sirolimus** is well-tolerated and efficacious for these patients.