DIFFERENTIAL DIAGNOSIS BETWEEN JUVENILE SYSTEMIC LUPUS ERYTHEMATOSUS AND AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME CASE REPORT

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BACKGROUND

Juvenile systemic lupus erythematosus (JSLE) is a chronic inflammatory, multisystemic, and autoimmune disease with an incidence rate of 0.6/100,000 children under 15 years, with a higher prevalence within women, native Americans, Asian Americans, Latin Americans and African Americans. Some medical findings of this condition are similar to those found in autoimmune lymphoproliferative syndrome (ALPS), which is an inborn error of immunity that results in a defect in the lymphocyte apoptosis, with chronic nonmalignant lymphoproliferation, lymphadenomegaly, and/or splenomegaly. Its incidence and prevalence are unknown but the number of cases worldwide are estimated to exceed several hundred.

CASE REPORT

Female patient, 5 years old, first admitted at age of 3 with ecchymosis, disseminated lymph node enlargement, hepatosplenomegaly, recurrent fever and bicytopenia (thrombocytopenia and leukopenia). Tests for negative double T lymphocytes by flow cytometry and specific tests aimed at excluding infections, neoplasms and autoimmune diseases were executed. Positive cytomegalovirus IgG and indirect Coombs were found among the serologies performed. The main diagnostic hypothesis at the time was ALPS. At 5 years old, the patient underwent three more hospitalizations, mainly due to the persistence of thrombocytopenia and severe epistaxis. She used platelet concentrate, pulse therapy with methylprednisolone and immunoglobulin with an unsatisfactory response. Progress was made in the treatment with prednisolone, tranexamic acid and azathioprine and we observed good disease control. The NGS panel for ALPS with CNV did not find pathogenic variants that would define the molecular diagnosis of the syndrome, which made us contest the ALPS's diagnosis. In the latest hospitalization, intense epistaxis and severe thrombocytopenia were found, new laboratory tests were performed, anti-SM and anti-Ro were reactive, and proteinuria was observed in the urine test. Given this clinical and laboratory context, we observed that the patient had the classification criteria for JSLE and started a specific treatment protocol.

CONCLUSION

In the pediatric age group, JSLE has clinical and laboratory classes not only similar to ALPS, but also to other infectious conditions, which can make the diagnosis more complicated to achieve. Given the history, we believe that the careful evaluation of the pediatrician, as well as their attention to the differential diagnosis, are going to establish an early diagnosis in this condition and positively impact these patients' prognosis.

KEYWORDS

Juvenile systemic lupus erythematosus, Autoimmune lymphoproliferative syndrome, Differential diagnosis.