

Juvenile systemic lupus erythematosus: A diagnostic dilemma

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Abstract

Juvenile systemic lupus erythematosus is an autoimmune disorder characterized by inflammatory damage to joints, kidney, central nervous system, and hematopoietic system in the form of fever, cutaneous lesion including skin rash, arthritis, anemia, and fatigue. We report a case in which the patient had features mimicking idiopathic thrombocytopenic purpura and juvenile dermatomyositis, but on a detailed Hematological investigation and kidney biopsy patient was diagnosed as juvenile SLE.

Key words: Juvenile systemic lupus erythematosus, lupus nephritis

INTRODUCTION

Juvenile systemic lupus erythematosus is an autoimmune disorder with multisystem involvement, leading to inflammatory damage to the joints, kidney, central nervous system, and hematopoietic system. Although the prevalence rate of juvenile systemic lupus erythematosus in a developing country is not known, as per literature the female-to-male ratio rises from 4.5 : 1 in adolescence to 8--12 : 1 in adult-onset patients.^[1] The mean onset age of lupus nephritis in Indian children is 9.6 ± 2.6 years.^[2] Presentation of systemic lupus erythematosus is highly variable, which usually shows prostration in the form of fever, anemia, rash, arthritis, and fatigue.

Here we report a case in which the patient presented with cutaneous manifestations in the form of erythematous rash, multiple erosive ulcers with hemorrhagic crust in the oral cavity, periorbital swelling, and telangiectasias. The patient also had perivalvular edema without any erosions and bilateral isolated quadriceps myositis.

Although these manifestations are commonly seen in juvenile dermatomyositis, we investigated and confirmed the diagnosis of juvenile systemic lupus erythematosus.

CASE REPORT

A 7-year-old, female, first child of non-consanguineous marriage presented with cutaneous manifestations in the form of an erythematous, nonblanching, macular ill-defined hyperpigmented rash over the body, mainly involving malar, upper eyelid, and periorbital region; multiple erosive ulcers with hemorrhagic crust in the oral cavity; and telangiectasias involving face and upper right hand [Figures 1 and 2]. The patient also had perivulvar edema without any erosions and bilateral isolated quadriceps muscle myositis in the form of severe tenderness and swelling of both thigh; there was no involvement of conjunctiva.

The patient had past history of two episodes of skin rash that was erythematous, nonblanching presented at an interval of 8 months. During both episodes, the treating consultant found persistent thrombocytopenia and did bone marrow biopsy, which showed the presence of large megakaryocytes; hence, the patient was diagnosed with idiopathic thrombocytopenic purpura. The first episode was treated with intravenous gamma globulin, and during the second episode, the patient was started on oral prednisolone 2 mg/(kg day), which continued till the age of 6 years. After this, the patient had an uneventful

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period of 1 year; again after 1 year, the patient presented with similar complaints described above. So investigations were done that showed thrombocytopenia (white blood cell count 23,000/cmm), muscle creatine kinase marginally raised, normal electromyogram, normal liver function test, and serum aldolase within the normal range.

On further investigation, to our surprise, we found antinuclear antibody positive with titer of 1 : 280 (>1 : 80 significant), urine routine and microscopy suggestive of proteinuria of 3+, and 24-h urinary protein to creatinine ratio of 8.38 (>3 significant). So renal biopsy was done, which showed WHO stage IV histological-type diffuse glomerulonephritis with mesangial and subendothelial deposits [Figures 3 and 4]. Systemic evaluation showed minimal ascites on ultrasonography; two-dimensional echocardiogram (2D ECHO) showed pericarditis and myocarditis with left ventricular ejection fraction of 35%, which dropped to 10% in 1 month after admission; the patient succumbed to death due to multisystemic involvement.

DISCUSSION

Systemic lupus erythematosus is an autoimmune disorder characterized by the production of autoantibodies and polyclonal activation of B lymphocytes. The child with lupus nephritis presented with systemic and often severe manifestations in the form of arthritis (60-80%), skin rash (60-78%), malar rash (20-70%), central nervous system manifestations (5-30%), and cardiopulmonary manifestations (10-30%) at the time of diagnosis.^[3] It shows immunogenetic association with HLA-A1, B8, DR3, DR2, C_{4a} null and inherited defects of the complement component C₂, C₄, C₁ esterase inhibitors.^[4] A familial presentation involving chromosome 1q23^[4] has been noted.

In our case, the 7-year-old girl initially presented with features such as heliotrope rash and myositis similar to the presentation of juvenile dermatomyositis. Later on, a detailed investigation such as urine analysis, renal biopsy, and anti-dsDNA antibody confirmed the diagnosis



Figure 1: Multiple erosive ulcers with hemorrhagic crust in the oral cavity.



Figure 2: Erythematous, nonblanching rash over eyelid, malar, periorbital region, and dorsum of hand

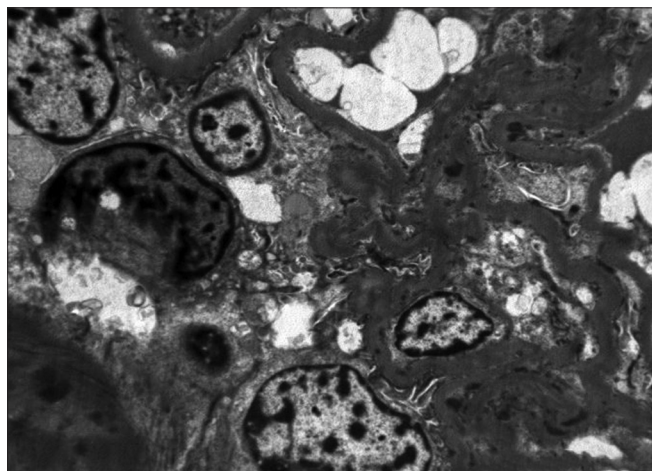


Figure 3: Electron microscopy showing thickened mesangium

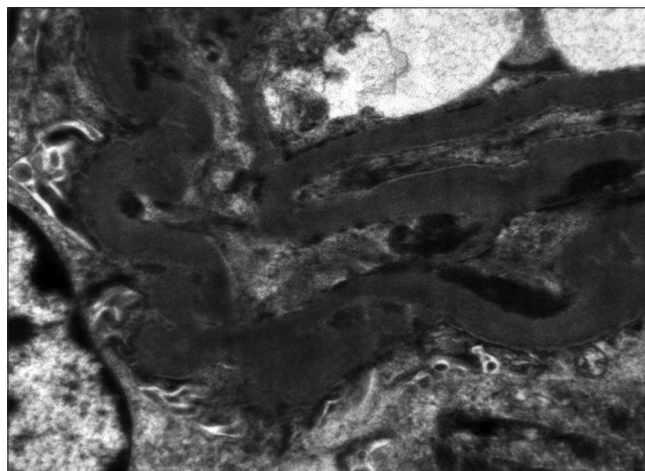


Figure 4: Subendothelial deposits in glomeruli

of lupus nephritis. Urine analysis was suggestive of proteinuria; renal biopsy was suggestive of WHO stage IV histological-type diffuse glomerulonephritis with mesangial and subendothelial deposits; and 2D ECHO was suggestive of pericarditis and myocarditis, which led to hypertrophy of left ventricle with left ventricular ejection fraction decreasing from 35% to 10% at the end of 1 month of admission, finally leading to heart failure. Diagnosis of systemic lupus erythematosus was confirmed by a combination of clinical and laboratory manifestations. As per American Rheumatism Association's 1997 revised classification criteria for systemic lupus erythematosus, 4 of 11 criteria are required^[5]; in our case, 6 of 11 criteria were fulfilled, and so diagnosis of lupus nephritis was confirmed. In contrast, although a heliotrope rash was present in our case, other criteria such as symmetrical weakness of proximal muscle and elevation of serum muscle enzymes (serum aldolase) were not present; hence, as per the criteria of Bohan and Peter,^[6] diagnosis of juvenile dermatomyositis is ruled out.

The patient was started on oral prednisolone 1.5 mg/(kg day) in divided doses and given intravenous methylprednisolone 30 mg/(kg day) over 60 min for 3 days as a short course. In severe cases, cytotoxic drugs such as intravenous cyclophosphamide are used to prevent progression of lupus nephritis, but in some studies it was found that the new regimen using lower doses of the same along with a reduced duration of intravenous cyclophosphamide administration has been found to reduce the toxicity. Mycophenolate mofetil is a variable alternative to cyclophosphamide for induction therapy of both proliferative and membranous lupus nephritis.^[7]

Other therapies include the administration of rituximab (anti-CD-20 monoclonal antibody), the T-cell costimulatory molecule BLyS, and a B-cell activating factor. Recently,

belimumab, a fully human monoclonal antibody (B lymphocyte stimulator-specific inhibitor), has been found to contribute to B-cell therapy with potential benefit for the patient with active systemic lupus erythematosus.^[8] Currently, the 5-year survival rate is >90% and the 10-year survival rate is 86%.^[9] A proportion of patients die of complications such as sepsis and renal failure. Patients with diffuse proliferative (WHO class IV) lupus nephritis exhibit the highest risk for progression to end-stage renal disease.

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