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Case Report

Infantile systemic lupus erythematosus without internal organ involvement: A rare case report

Bhumesh Kumar Katakam, Narsimha Rao Netha Gurram, Sudharani Chintagunta, Arunima Dhabal

Department of Dermatology, Venereology and Leprosy, Gandhi Medical College and Hospital, Secunderabad, India.

Abstract Systemic lupus erythematosus (SLE) is a rarity in children, especially in infancy. Current literature has very few reports of infantile SLE, and most of the reported patients had severe disease with systemic involvement and poor outcomes. We describe a case of infantile SLE in a female baby presenting with fever and rash, without any internal organ involvement.

Key words
Systemic lupus erythematosus; Infant.

Introduction

Juvenile systemic lupus erythematosus (SLE) is a multisystemic, inflammatory, autoimmune disease characterized by variable cutaneous and systemic manifestations, occurring in children below 16 years of age. It represents 10-15% of all SLE cases and is extremely rare in infancy. Unlike neonatal lupus, infantile SLE occurs in infants born to healthy mothers, and usually presents with clinical manifestations and positive serologies similar to classical SLE.[1] Current literature has very few reports of infantile SLE, and majority of the reported patients had associated systemic involvement with a poor prognosis.[1-3] We describe a female infant presenting with fever and cutaneous rash, in the absence of systemic involvement, later confirmed as a case of infantile SLE by serology.

Address for correspondence
Dr. Arunima Dhabal
Pediatric Dermatology Trainee,
Department of Dermatology, Venereology and Leprosy, Gandhi Medical College and Hospital, Secunderabad, India.
Email: ardhabal@gmail.com

Case Report

A female baby, born at term after an uncomplicated pregnancy, presented at the age of 2 years with intermittent fever and an erythematous rash primarily involving the face, which was aggravated on sun exposure. The child had first developed a similar episode of fever with cutaneous rash, facial edema and oral erosions at the age of 9 months. She was admitted to the hospital, where she was diagnosed as recurrent bullous impetigo and facial cellulitis. Her condition improved on treatment with parenteral antibiotics, a short course of oral prednisolone (1 mg/kg/day) and supportive care. Over the next one year she had three similar episodes of fever accompanied by facial edema, erythematous rash with crusting, and oral erosions. On each episode she required hospital admission and improved with a similar line of management, but the lesions recurred after a few months. The child was born of a non-consanguineous marriage and family history was non-contributory.

On examination, there were multiple bilaterally symmetrical erythematous scaly plaques on the malar areas, forehead, nose and chin (**Figure 1**).

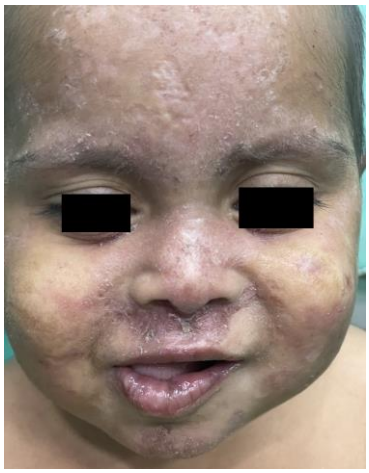

Figure 1 Bilaterally symmetrical erythematous scaly plaques on the cheeks, forehead, nose and chin, with involvement of nasolabial folds.

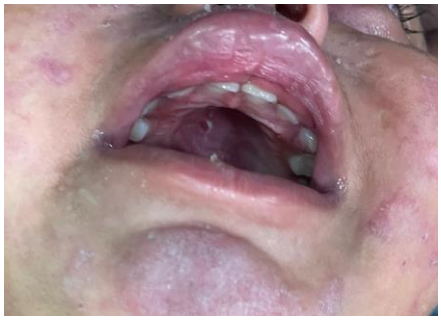

Figure 2 Erosion on hard palate.

Examination of the oral cavity revealed painless erosions on the inner surface of lips and hard palate (**Figure 2**). Hair and nail examination did not reveal any abnormality. The child weighed 11 kg and showed normal developmental milestones. She was febrile at the time of presentation, but systemic examination findings were within normal limits.

Complete hemogram revealed microcytic hypochromic anemia (hemoglobin 8.6 g/dl, anisocytosis, poikilocytosis, tear drop cells) and lymphopenia (absolute lymphocyte count 1782 cells/cmm, differential count 22%). Liver and renal functions were normal. Urine examination did not show any proteinuria, hematuria or porphyrin excretion. Among complement fractions, C1q, C3 and C4 levels were normal, but CH50 was low. Antinuclear antibody (ANA) titer was elevated at 1:64. ANA profile revealed a positive serology for Anti-Ro (SS-A), but was negative for Anti-dsDNA, Anti-La (SS-B), Anti-Sm, and Anti-U1RNP antibodies. These results prompted us to evaluate the patient's mother for lupus, but she had no clinical features or laboratory parameters suggestive of subacute cutaneous lupus erythematosus (normal ANA, anti-dsDNA, anti-Ro, and anti-La antibodies).

Histopathology of lesional skin showed subepidermal separation with predominance of neutrophils and eosinophils in the bulla cavity, and mixed infiltrate in the papillary dermis. Direct immunofluorescence was negative for IgA, IgG, IgM, C3 and C1q. Based on the clinical and laboratory findings, we arrived at a diagnosis of infantile SLE. We advised chest X-ray, echocardiography, abdominal ultrasound and MRI of brain, but no abnormality was noted on any of the imaging studies, thereby ruling out internal organ involvement.

The parents were advised to avoid photo-exposure and oral prednisolone was initiated at a dose of 1 mg/kg/day along with oral hydroxychloroquine 6.5 mg/kg/day. Improvement of lesions was observed after 2 weeks of treatment. The baby is presently on the same treatment and is doing well on regular follow up.

Discussion

Infantile SLE is a very rare entity reported in only 15 patients so far, to the best of our knowledge.[1-3] In a review of 13 reports of infantile SLE, the patients were found to present with variable and non-specific symptoms such as fever, rash, irritability, edema and respiratory distress. While fever was the most frequent symptom, cutaneous rash was observed in three patients and oral ulcer in only two of the patients in the study.[1] The morphology of rash also

varies in reported cases. Malar rash was present in one case, while two other cases reported petechiae and purpuric rash indicating coagulation abnormalties.[1] Another patient reported by Garg *et al*. presented with pruritic, erythematous, atrophic and telangiectatic plaques over face, scalp and trunk, with few lesions progressing to form angiomatous papules, while others healed with pigmentation and scarring.[3] Our patient also presented with erythematous scaly plaques with crusting on face, although scarring was absent. Notably, unlike classical SLE, the rash did not spare the nasolabial folds. The variable clinical presentation often leads to erroneous diagnosis of cases at first presentation. This emphasizes the importance of thorough evaluation of patients presenting with recurrent non-specific but severe symptoms.

Infantile SLE should be differentiated from neonatal lupus, occurring in neonates born to mothers with circulating ANA, due to transplacental passage of maternal antibodies. It is characterized by discoid rash, cytopenia and often congenital heart block, but the disease is self-limiting with gradual recovery by 6 months, following clearance of maternal antibodies. Neonatal lupus was ruled out in our case by normal connective tissue profile of the mother. Unlike neonatal lupus, infantile SLE is associated with poor outcomes due to higher frequency of hematological abnormalities, seizures and renal involvement.[4] Among the 13 cases reviewed by Zulian *et al*., five infants died due to causes ranging from sepsis to pulmonary hemorrhage, while five of the survivors developed severe residual organ damage.[1] Severe lupus nephritis and concomitant Epstein Barr virus infection were reported in a 14-month-old Japanese boy with infantile SLE, while Garg *et al*. reported neurological complications in the form of delayed speech and development.[2,3] In contrast, our patient did not have any systemic symptoms and apart from hematological abnormalities, no organ involvement was detected on detailed systemic evaluation.

The laboratory investigations in our patient also revealed some interesting findings. While a majority of the previously reported cases had high levels of ANA and/or anti-ds DNA antibodies, our patient showed raised ANA along with positive anti-Ro antibodies.[1-3] Anti-Ro antibody is primarily a marker of neonatal lupus, but may be seen in up to 30-40% patients of SLE, who have an increased tendency to photosensitivity, secondary Sjögren syndrome, and pulmonary involvement. It is also found in SLE associated with genetic deficiencies of C1q, C2 or C4.[4] Although early complement levels were normal in our patient, CH50 was low, indicating deficient complement function. Thus, in this case a qualitative complement defect may be linked to the development of SLE. The conspicuous absence of internal organ involvement in this case may be explained by the absence of anti-dsDNA positivity, which is known to correlate with high disease activity and lupus nephritis.[4] Therefore, although a thorough evaluation for systemic involvement is mandatory in all cases of infantile SLE, not every case has a poor prognosis.

Conclusion

This report emphasizes the importance of considering SLE in the differential diagnosis of infants presenting with fever accompanied by photosensitive rash, and differentiation of such cases from neonatal lupus. Our patient not only represents a rare entity, but also had some unusual features like absence of systemic organ involvement, anti-Ro positivity, and anti-dsDNA negativity.

References

1. Zulian F, Pluchinotta F, Martini G, Da Dalt L, Zacchello G. Severe clinical course of systemic lupus erythematosus in the first year of life. *Lupus*. 2008;**17(9)**:780-6.
2. Kishi N, Suga K, Matsuura S, Kinoshita Y, Urushihara M, Kondo S, *et al*. A case of infantile systemic lupus erythematosus with severe lupus nephritis and EBV infection. *CEN Case Rep*. 2013;**2(2)**:190-3.
3. Ghosh JB. Infantile Systemic Lupus Erythematosus with Cerebral Calcification and Delayed Development: Correspondence. *Indian J Pediatr*. 2019;**86(2)**:202-3.
4. Goodfield M, Dutz J, McCourt C. Lupus erythematosus. In: Griffiths CEM, Barker J, Bleiker T, Chalmers R, Creamer D, eds. Rook's Textbook of Dermatology. 9th ed. West Sussex: John Wiley & Sons, Ltd; 2016:51.1-51.39.