A Rare Presentation of Systemic Lupus Erythematosus with Juvenile Dermatomyositis

Ashish Varma[1], Sneh Kumar[2], Rupali Salve[3], Anjali Kher[4], Jayant Vagha[4], Sachin Damke[5]

[1]Associate Professor, [2]Junior Resident, Department of Pediatrics, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences, Sawangi[M], Wardha, [3]Assistant Professor, [4]Professor, [5]Professor & HOD, Department of Pediatrics, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences, Sawangi[M], Wardha

Abstract

The systemic lupus erythematosus (SLE) is a rare type of autoimmune disease, which is associated with the involvement of multiple systems. It is characterized by the production of autoantibodies. Usually, it has a relapsing and remitting course. Juvenile dermatomyositis is the most prevalent chronic inflammatory muscle disease among children and adolescents with SLE. This disease predominantly involves the skin and skeletal system. Its most common symptoms include distinctive skin rashes and inflamed muscles. The insufficient disposal of apoptotic cells may result in the stimulation of T cells and B cells by the antigens in the patients with SLE. At the surface of the dying cell, fragments of cellular material develop during the cycle of cell death. We presented the case of a 15-year-old female, who complained of erythema and generalized rash present on the face, markedly present over her cheek, bridge of the nose and the forehead since 7 months. She developed erythematous, scaly and crusted lesions on the scalp, back, trunk and upper limbs along with the vesiculobullous lesion of the oral mucosa. She also developed a progressive generalized muscle weakness for the past 3 months. To a large extent, intravenous glucocorticoids are helpful, but standardization of tests and treatment schemes are required to enhance the awareness of this rare case.

Keywords: *SLE, lupus erythematosus, juvenile dermatomyositis.*

Introduction

Lupus is a chronic autoimmune inflammatory condition, presenting with a vast range of clinical presentations, which arises due to the involvement of multiple systems[1,2]. The systemic lupus erythematosus (SLE) is the prevalent form of lupus, which is usually referred to as "lupus," but it is distinguished from other types of lupus due to its effects on the multiorgan systems. It is a chronic autoimmune disease, which has clinical and serological heterogeneity[2,3]. SLE is a rare autoimmune disease that involves multiple organ systems, characterized by inflammation and production of antibodies to nuclear and cytoplasmic antigens. It has a relapsing and remitting course[4]. The prevalence of SLE ranges from 0.03% in Caucasians to 0.2% in Afro Carribeans. Its incidence in females is 90% or more, and the most common age is 20-30 years[5]. SLE's most important feature is the development of auto-antibody, which is specific to a wide variety of targets but often attacks the antigens present inside the cell or nucleus [4]. SLE accompanies a sustained production of immune complexes and autoantibodies. The antigens cause the release of cytokines, vasoactive peptides, chemokines, proteolytic enzymes and oxidants due to the activation of complement. This causes activation and influx of multiple tissue cells into target tissues of T and B cells, monocytes or macrophages, and dendritic cells[4]. It is believed that SLE may arise as a result of programmed

Corresponding Author:
Dr. Ashish Varma
Associate Professor, Dept of Paediatrics, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences (Deemed to be University), Sawangi, (M), Wardha-442001, (Maharashtra) India
e-mail- avarma2055@gmail.com

cell death or inequitable clearance of these cells. It corresponds to inherited mutations in complements C1q, C2 and C4.

Juvenile dermatomyositis is the most common chronic inflammatory muscle disease in children and adolescents with SLE. It predominantly involves the skin and skeletal system[4,6]. Distinctive skin rashes, as well as inflamed muscles, are the most common symptoms. It can be monocyclic or polycyclic, with periods of remission and relapse. The findings include proximal muscle weakness and violaceous discolouration of eyelids, also called as heliotropic rashes [3)]. 10-20% of cases can be ulcerative, while 30% of cases can progress to calcinosis later on. In many cases, intravenous methylprednisolone and methotrexate can induce a rapid response. Cyclophosphamide can be used for the treatment of Lesional ulcerations. Cutaneous manifestations are mostly attributed to juvenile cases than adults. SLE commonly involves the skin, and many skin eruptions can be due to exposure to UV light.

The main types include the following[2,4]:

- The classical facial rash is elevated, erythematous and painful, and appears on the cheeks. It can be itchy. The nasolabial folds are usually spared.
- The discoid rash is hyperkeratosis with alopecia which is of scarring type especially on the scalp.
- Non-scarring alopecia
- Urticarial eruptions
- Livedo reticularis

Case report: A 15-years-old female born of secondary consanguineous marriage presented with the complaints of erythema and generalized rash on the face, markedly over her cheek, bridge of the nose and forehead for the past 7 months. She developed vesiculobullous lesion of the oral mucosa and erythematous, scaly and crusted lesions on the scalp, upper limbs and back, which healed with hypopigmentation. Similar lesions were found on the trunk and the extensor surface of the upper limbs. She also complained of painful oral ulcers, which resulted in significant difficulty in eating.

She also presented with progressive generalized muscle weakness for the past 3 months and complained of the inability to perform household chores. She had difficulty getting up from the bed and lifting the arm while changing clothes. She complained of easy fatiguability and tightening of limb, with more involvement of the lower limb in comparison to the upper limb.

She later developed low-grade intermittent fever associated with joint pain, particularly in the large joints like knee joints and hip joints. She complained of a reduction in vision in the eyes, with more involvement of the left eye, which gradually progressed. She also complained of dryness in the eyes. She complained of alopecia for the past 5 months, more on the temporoparietal regions, presenting in patches with sustained frontal margin. She showed a decrease in appetite and weight loss for the past 7 months. She complained of irregular menstrual bleeding with the decreased flow, which lasted for 1-2 days over a gap of 35-40 days. It was not associated with pain or blood clots.

On general examination, the patient was thin-built and malnourished. On cutaneous examination, there was a diffused loss of hair with patchy alopecia at some areas along with few hyperpigmented scaly lesions, erythematous rashes over the face including both cheeks, bridge of nose and forehead, and scaling and crusting accompanying the lesions. Local examination revealed oral ulcers in the buccal mucosa. Systemic examinations were found normal.

Laboratory investigations were performed. Complete blood count (CBC) revealed Haemoglobin (Hb) = 8.9 g/dl, White blood cells (WBC) = 2600 cells/ mm3 (Monocyte = 4, Granulocyte = 34, Lymphocyte = 60, Eosinophil = 02, Basophil = 00), platelets = 75,000/mm3, erythrocyte sedimentation rate (ESR) = 70 mm/hr, C Reactive Protein (CRP) = Positive, Serum creatinine = 0.73, direct Coomb's test = positive and serum creatine kinase = 400 unit/litre. The CBC showed a raised erythrocyte sedimentation rate (ESR), which suggested of thrombocytopenia, thereby pointing towards an infectious cause. Liver function tests (LFT), renal function tests (RFT) and Urine routine were found within the normal range. Specific tests were carried out, and anti-*double stranded DNA* (anti-*dsDNA*) was found positive (4.16) and anti- antinuclear antibody (anti-ANA) was positive (102.44). The ophthalmologic test was suggestive of anterior subcapsular cataract of the left eye. An autoimmune etiology was suspected as per the pattern of rashes and cachexia in the adolescent female. Further investigations were performed, and the skin biopsy reported of scleroderma. USG pelvis revealed hypoplastic uterus.

According to Bohan and Peter classification, this patient fit in the criteria of Juvenile Dermatomyositis. According to the American College of Rheumatology (ACR) criteria of SLE, the diagnosis was confirmed. The diagnosis was 'SLE with juvenile dermatomyositis'.

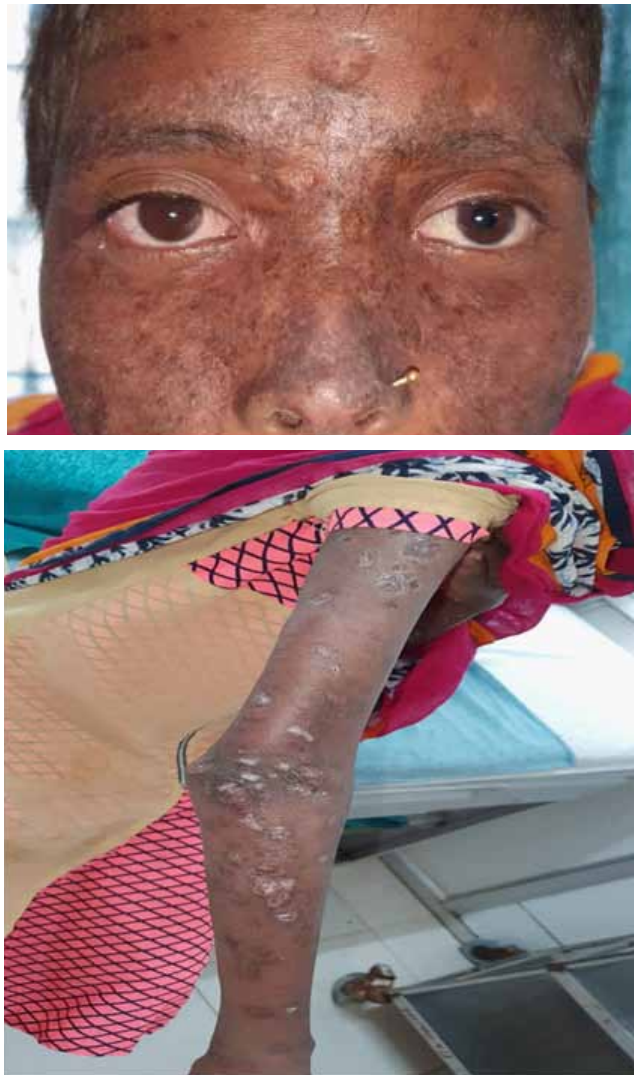
Discussion

SLE is a chronic disorder involving multiple organ systems, predominantly due to the development and deposition of immune complexes and autoantibodies, resulting in subsequent impairment in the organs[1,7].

The antigens, which cause the stimulation of T cells and B cells in patients with SLE may be due to the insufficient disposal of apoptotic cells[4,5]. The fragments of cellular material develop at the surface of the dying cell during the cycle of cell death. A higher concordance is seen in the monozygotic twins, and the disease is associated with polymorphic variants at the HLA locus. In a few instances, SLE is linked with inherited mutations. [8]. Genome-wide association studies have identified polymorphisms near other genes, which can lead to SLE; these genes are involved in regulating immune cell function.[9] The fact that environmental factors, which can cause flares of lupus such as UV light and infections, can increase the oxidative stress and cell damage supports the apoptotic theory.[4] Autoantibodies are prevalent in Juvenile Dermatomyositis. They can be categorised into two groups, as follows: myositis specific antibodies (MSA), which target the synthesis of proteins, cytoplasmic or nuclear components, and myositis-associated antibodies (MAA), which are usually present in many autoimmune disorders and syndromes that overlap. Approximately 70 per cent of children with Juvenile Dermatomyositis have one or more detectable MSA or MAA[11]. Balwani et-al reported progressive renal failure in Lupus patient.[12]

Conclusion

SLE with juvenile dermatomyositis is a rare disorder. Since the international consensus regarding diagnosis and treatment is currently unavailable, the management is thus variable. So, precise recommendations would help physicians in the proper management of patients with juvenile dermatomyositis. Specific recommendations at present involve examination of muscle disease, skin disease, calcinosis, biomarkers, and autoantibodies. Management with intravenous glucocorticoids is helpful to some extent. It is suggested that the standardization of tests and treatment plans with collaborative research studies would acknowledge the profundity of rare disorders like this.



Erythematous, scaly and crusted lesions on the scalp, upper limbs and back, which healed with hypopigmentation

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References

1. Maidhof W, Hilas O. Lupus: An Overview of the Disease and Management Options. :8.
2. Furie R, Cervera R. Systemic lupus erythematosus.

Best Practice & Research Clinical Rheumatology. 2017 Jun;31(3):289–90.

3. Battista MD, Marcucci E, Elefante E, Tripoli A, Governato G, Zucchi D, et al. One year in review 2018: systemic lupus erythematosus. Clinical and Experimental Rheumatology. 2018;15.
4. Ralston S, Penman I, Strachan Ma, Hobson R. Davidson's Principles and Practice of Medicine [Internet]. 23rd Edition. Elsevier; 2018. 1440 p. Available from: https://www.elsevier.com/books/davidsons-principles-and-practice-of-medicine/ralston/978-0-7020-7028-0
5. Jameson JL, Fauci AS, Kasper DL, Hauser SL, Longo, DL, Loscalzo J. Harrison's Principles of Internal Medicine. 20th ed. McGraw-Hill Professional; 4048 p.
6. Wu Q, Wedderburn LR, McCann LJ. Juvenile dermatomyositis: Latest advances. Best Practice & Research Clinical Rheumatology. 2017 Aug;31(4):535–57.
7. Aikawa NE, Jesus AA, Liphaus BL, Liphaus BL, Silva CA, Sampaio MC, et al. Organ-specific autoantibodies and autoimmune diseases in juvenile systemic lupus erythematosus and juvenile dermatomyositis patients. Pediatr Rheumatol. 2012 Jul;10(S1):A14, 1546-0096-10-S1-A14.
8. Papadopoulou C, Wedderburn LR. Treatment of Juvenile Dermatomyositis: An Update. Pediatr Drugs. 2017 Oct;19(5):423–34.
9. Bellutti Enders F, Bader-Meunier B, Baildam E, Constantin T, Dolezalova P, Feldman BM, et al. Consensus-based recommendations for the management of juvenile dermatomyositis. Ann Rheum Dis. 2017 Feb;76(2):329–40.
10. Varnier GC, Pilkington CA, Wedderburn LR. Juvenile dermatomyositis: novel treatment approaches and outcomes. Current Opinion in Rheumatology. 2018 Nov;30(6):650–4.
11. Ghirardello A, Borella E, Beggio M, Franceschini F, Fredi M, Doria A. Myositis autoantibodies and clinical phenotypes. Autoimmun Highlights. 2014 Dec;5(3):69–75.
12. Balwani, M et al. "An Uncommon Cause of Rapidly Progressive Renal Failure in a Lupus Patient: Pauci-Immune Crescentic Glomerulonephritis." Saudi Journal of Kidney Diseases and Transplantation: An Official Publication of the Saudi Center for Organ Transplantation, Saudi Arabia 29, no. 4 (2018): 989–92.