Concurrent Presentation of Systemic Lupus Erythematosus in Mother and Daughter

Abstract

Systemic lupus erythematosus (SLE) is a connective tissue disorder with a high prevalence in certain races like Africans. It also has a female predilection. The pathogenesis of SLE is a complex interplay of genetic and environmental factors. Familial presentation of SLE has been well documented. We herein illustrate a scenario of SLE presenting in related females (mother and daughter) concurrently with similar clinical features and fulfilling the Systemic Lupus Collaborating Clinics criteria. Stress was identified as a common possible triggering factor.

Keywords: *Concurrent, familial, Systemic lupus erythematosus*

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Introduction

Systemic lupus erythematosus (SLE) is a multisystem disorder. The susceptibility to SLE has been attributed to complex interactions between multiple genetic and environmental factors. The genetic predisposition on susceptibility to SLE is supported by observations of various familial aggregations.[1] One of twin studies have confirmed concordance for SLE in 20 to 30% of monozygotic twins, as opposed to less than 2% concordance for SLE in dizygotic twins.[1] Herein we present a case of mother and daughter presenting with SLE for the same duration (10 years).

Case Reports

Case 1

A 45-year-old female presented with red-raised, scaly lesions over hands, feet, forearms, and lips gradually increasing in size and number for last 10 years, which resolve leaving behind hyperpigmentation. There was history of recurrent painless oral ulcers over buccal mucosa and hard palate for the same duration. She also had multiple joint pains without any redness or swelling, which started in the knee joint. History of photosensitivity was present. Patient also reported Raynaud's phenomenon in winter, diffuse hair fall, and breathlessness for last 5 to 6 years. She was a known case of hypothyroidism and is on regular treatment.

On examination, she had multiple well-defined, erythematous-to-depigmented atrophic plaques, varying in size from 1.5 × 1cm to 2 × 1.5 cm present over dorsa of hands and feet, left forearm, and lower lip with surface showing adherent whitish scale with tin tack sign [Figure 1a and b]. Telogen effluvium was present and hair pull test was positive. Mucosal and systemic examination was normal.

Hemoglobin was 9.4 gm/dl, total leukocyte count was 3000 cu/mm, and platelet count was 3 lakh/µl.Reticulocyte count was 1%. Liver function test, kidney function test, and 24-hr urine protein test werenormal. Antinuclear antibody (ANA) and double-stranded DNA (dsDNA) were positive. Anti-Ro antibody was negative. Skin biopsy from hand lesion revealed marked acanthosis, hyperkeratosis, hypergranulosis with vacuolar degeneration of the basal layer, and dermis showed perivascular lymphohistiocytic infiltrate. Direct immunofluorescence was negative. Direct Coombs' test was positive; complements levels were normal and systemic evaluation was within normal limits.

Case 2

Quite interestingly, her daughter who was 30 years old also had similar complaints of

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red-raised, scaly lesions over hands, feet, forearms, and lips gradually increasing in size and number for last 10 years, which resolved leaving behind hyperpigmentation. She had history of recurrent painless oral ulcers over buccal mucosa and hard palate for same duration and multiple joint pains including knee joint without any redness or swelling. She had history of photosensitivity. The patient also reported pulmonary tuberculosis 5 months back and was on antitubercular therapy (category 1) for last 4 months. She had history of three spontaneous abortions at 16, 6, and 10 weeks of gestation, respectively. Subsequently, she had two live births, both of which are healthy. On examination, she had multiple well-defined, erythematous-to-depigmented atrophic plaques, varying in size from 1 × 0.75 cm to 2 × 1.5 cm present over dorsum of hands, feet, and lips with surface showing adherent whitish scaling [Figure 1c and d]. Oral mucosal examination revealed single, nontender, irregular shaped ulcer over right buccal mucosa. Systemic examination was normal.

Hemoglobin was 10.7 gm/dl, total leukocyte count was 3.6 × 10^3 cu/mm, and platelet count was 1.82 lakh/µl. Reticulocyte count was 3%. Liver function test, kidney function test, and 24-hr urine protein test were normal. ANA, dsDNA, and anti-Ro were positive. Skin biopsy revealed acanthosis, lymphocytic exocytosis, vacuolar degeneration of the basal layer, focal basement membrane thickening, and dermis showed lymphohistiocytic infiltrate. DIF is negative. Direct Coombs' test was negative. Systemic evaluation was normal. Antiphospholipid antibody was negative. Thyroid function is normal.

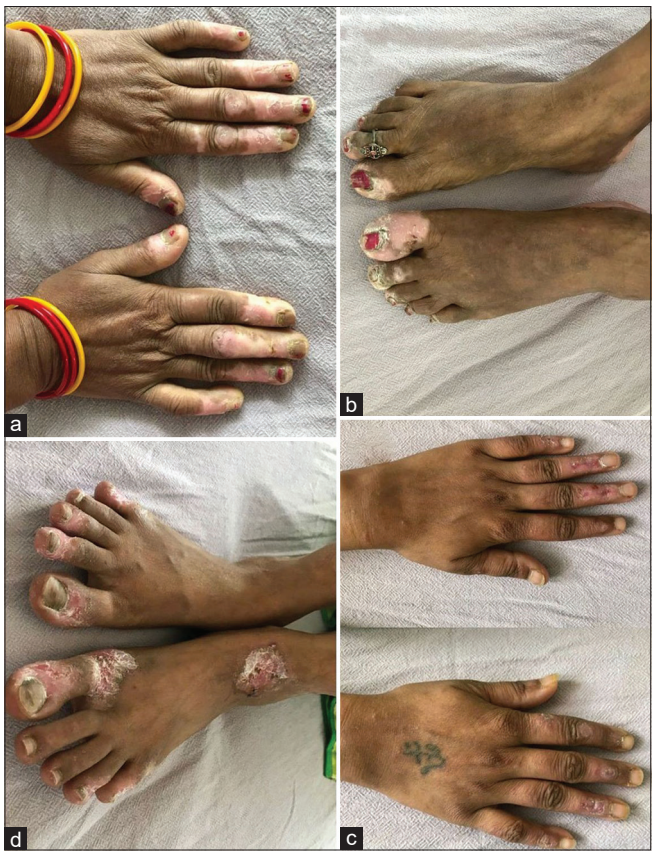

Figure 1: Well-defined erythematous–to-depigmented atrophic plaques presentover bilateral dorsum of hands (a) and feet (b), along with whitish adherent scaling in mother. Lesions with similar morphology and distribution as mother present in the daughter (c and d)

The Systemic Lupus Collaborating Clinics (SLICC) criteria[2] fulfilled in the mother and daughter have been tabulated in Table 1.

Discussion

SLE is a chronic multisystem disorder. The female predominance of SLE is well known. Higher susceptibility is noted in certain races and ethnic groups. The genetic predisposition of SLE is now well recognized, but appears to be polygenic (with more than 20 loci contributing).[3] Study by Michelle *et al.* indicated genetic heterogeneity of SLE patients with 100 multiplex families having two affected relatives, while 25 families having three or more affected individuals. The relationship was siblings (45%), parent offspring (35%), and second degree (24%).[4] An autosomal dominant mode of inheritance was predicted in one extended pedigree with a clinically affected member and a recessive pattern in five other families. In remaining families, no obvious mode of inheritance could be identified. Another study in Finnish population where they had taken 1,200 cases of SLE, incidence of familial SLE was found to be approximately 4 to 5%. In this study, 53 multiplex families were identified, 46 with 2 affected relatives, and 7 with 3 affected members.[5] There was no difference in clinical presentation between familial and sporadic cases of SLE; however, racial and ethnic differences in manifestations of SLE have been well documented by many others. A study of familial aggregation of SLE and other autoimmune diseases like Sjogrens syndrome, psoriasis, thyroid disease, ANA in an unusual multiplex pedigree has been done by Sestak *et al.*,[6] in which 8 females with SLE from a single family had 53 blood relatives— 15 of them had autoantibodies, 9 had autoimmune diseases (7 had SLE, 1 had psoriasis, and 1 had Sjogrens syndrome),

Table 1: SLICC criteria[2] fulfilled in the patients

Mother	Daughter
Clinical criteria	Clinical criteria
Discoid rash	Discoid rash
Recurrent oral ulcers	Recurrent oral ulcers
Arthralgia	Arthralgia
Diffuse hair fall	Diffuse hair fall
Serological criteria	Serological criteria
Leukopenia	Anemia, Leukopenia
ANA	ANA
dsDNA	dsDNA
Direct Coombs' test	

and 11 of the nonrelated spouses had autoantibodies, 1 had SLE, and 2 had thyroid disease. Among 68 spouses of SLE in other pedigree, only 9 had autoantibodies and none were symptomatic.[6] This study suggested a complex interaction of genetic and environmental factors.

Family studies have revealed a higher than expected prevalence among the relatives of patients with SLE.[1] Although the precise prevalence of familial SLE is not known, approximately 10% of patients with SLE have a first-degree relative with SLE, as compared with1% of patients in control families.[7-9] In familial SLE, the most frequent mode of familial intra-aggregation is affected sibling pairs,[9] and females predominate, with mother–daughter and sister–sister pairs being the most common and father–son pairs occurring relatively rarely.[9]

In the present case, it was a mother and daughter who presented with similar clinical features of SLE, and on top of that the symptoms appeared concurrently 10 years back, whereas in previously reported case mother and daughter had SLE but the daughter presented 6 to 7 years earlier than the mother.[3] The previously reported case had renal involvement in both the cases but no systemic involvement could be detected at present in our cases. In our cases, both the mother and the daughter were positive for dsDNA and ANA. However, the family screening did not reveal involvement of any other family member. A history of death of a close family member was present causing stress, which could be a possible triggering factor. In our scenario , presentation of disease simultaneously and with similar clinical features are interesting findings.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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