CASE REPORT

A Curious Case of Diffuse Systemic Sclerosis with Discoid Lupus Erythematosus-Like Lesions: Enigma of an Overlap

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Summary

Overlap syndrome is a term used to describe a condition wherein a patient has features of more than one classic inflammatory rheumatic disease like systemic lupus erythematosus, polymyositis, scleroderma and rheumatoid arthritis. Individuals with an overlap syndrome may, but need not meet, complete diagnostic criteria for one or more than one classic rheumatic disease. Mixed connective tissue disease is a specific subset of overlap syndrome wherein patients have antibodies to the U1 small nuclear ribonuclear protein (anti- U1RNP) and clinical features like hand edema, synovitis, Raynaud phenomenon, acrosclerosis and biologically or histologically proven myositis. We came across an interesting case showing clinical features of both Systemic Sclerosis and Discoid Lupus erythematosus (DLE). On complete evaluation, a final diagnosis of Diffuse Systemic Sclerosis - DLE overlap was made on the basis of histopathopathological and serological findings. Patient was started accordingly on systemic and topical medications and responded well.

Key words: *Systemic sclerosis, discoid lupus rrythematosus, antinuclear antibodies*

Introduction

Overlap syndrome describes the clinical situation wherein symptoms of various connective tissue diseases co-exist. It is more common in females (female to male ratio of 3.3:1). Systemic sclerosis overlaps with other autoimmune disorders in about 10-20% cases. Here we highlight a patient with clinical, serological and histopathologic findings consistent with diffuse systemic sclerosis (dSSc) and discoid lupus erythematosus overlap.

Case Report

A 36-year-old married multipara Hindu female farmer presented with itchy lesions over back, hands and legs since 2 months, with intermittent fever, breathlessness, brownish skin discoloration, patchy hair loss, skin tightness and photosensitivity. Patient denied history of difficulty in swallowing, squatting or combing hair, bowel disturbances, joint pain, oral lesions, Raynaud's phenomenon, dyspnea, abdominal pain or burning micturition. She was not on any treatment. Dermatological examination showed taut shiny skin on face and scarring alopecia over scalp. Face showed

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erythematous - hyperpigmented non-confluent malar rash. Erythematous scaly plaques with central depigmentation and atrophy [DLE- like] were present on scalp, upper and lower back and upper arms (photo-exposed areas). Salt and pepper pigmentation were seen over neck, trunk, upper and lower limbs. Tightness of skin was noted on digits of hands and feet extending beyond metacarpo and metatarso-phalyngeal joints upto mid-forearm and mid-shin respectively and over face, neck, chest and upper back. Oral, genital and ocular mucosae were normal. Clinical differential diagnosis considered was dSSc-DLE overlap syndrome and mixed connective tissue disease (MCTD).

Hemogram and renal function tests were normal. Aspartate aminotransferase, Alanine aminotransferase levels and Erythrocyte sedimentation rate were raised. C-reactive Protein, Rheumatoid factor, HIV- Elisa and HBsAg were negative. Creatinine Kinase (MB), 24-hour urine protein were normal. Serum ANA was positive (2.92). ANA blot indicated high levels of anti Ro/ SSA (17AU) and anti Scl-70 (60KD) with normal anti ds DNA and anti-Smith antibodies. Ultrasonography (Abdomen and pelvis) showed no abnormality except right ectopic kidney. Chest X-ray (PA view) showed few homogenous opacities in bilateral middle lobes. HRCT chest confirmed interstitial lung disease (ILD). Pulmonary function tests showed restrictive pattern. Barium swallow and nail fold capillaroscopy were unremarkable. Histopathology of DLE-like lesion (over neck) showed focal basal vacuolar degeneration and increased dermal collagen with periadnexal mononuclear infiltrate.

Biopsy from hidebound skin over left thumb demonstrated increased dermal fibrocollagenous tissue. Direct Immunofluorescence could not be done due to non-availability. Final diagnosis of diffuse Systemic sclerosis-DLE overlap was made.

Patient was started Tab Methotrexate 7.5mg weekly, Tab Hydroxychloroquine 200 mg bd and Tab Prednisolone (1mg/kg/day) in slow tapering doses along with topical corticosteroids and sunscreen. Currently, patient is under follow up with improvement in skin lesions and respiratory complaints.

Figure 1. (a) Hyperpigmented malar rash, (b) scarring alopecia, (c) DLE-like lesions over photo exposed parts. (d) Histopathology (DLE-like lesion) showing combination of DLE (hyperkeratosis with atrophy and flattening of rete ridges, focal basal vacuolar degeneration, peri-adnexal chronic mononuclear inflammatory infiltrate) and scleroderma (increased collagen in papillary and reticular dermis)

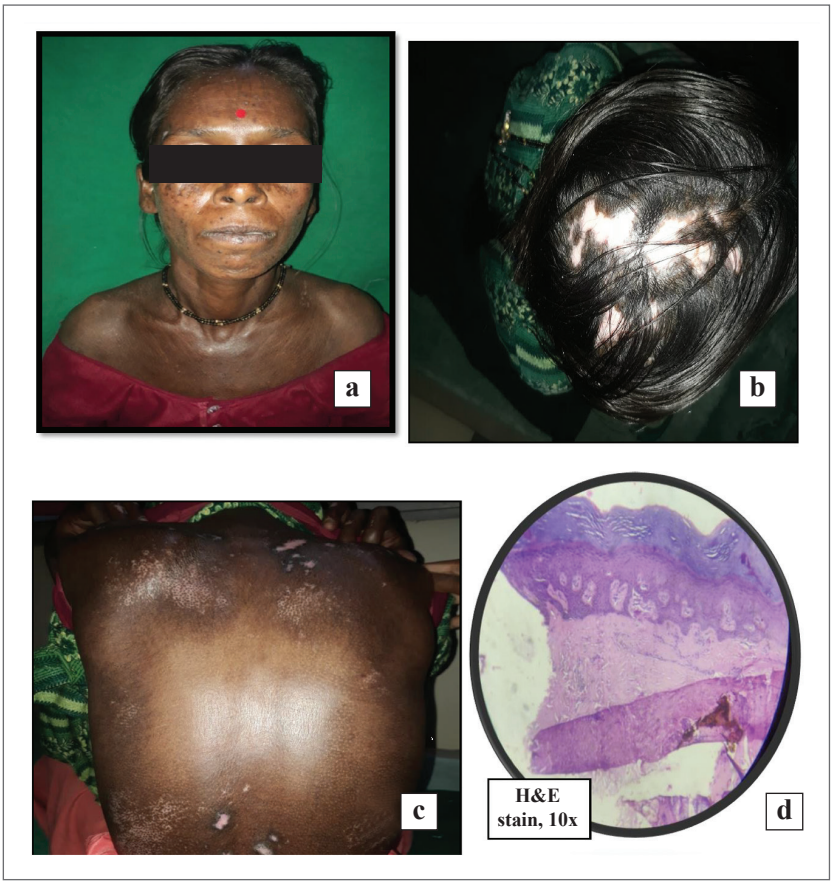
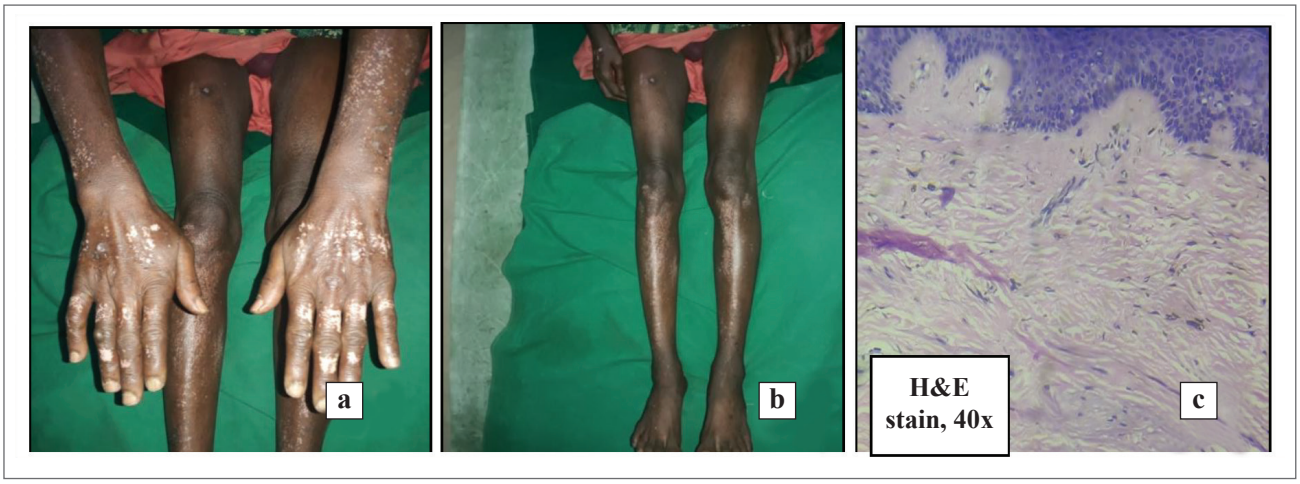


Figure 2. (a & b) Poikiloderma over extremities. (c) Biopsy (from hide-bound skin over left thumb) consistent with scleroderma {hyperkeratosis and acanthosis with focal elongation of rete ridges, increased fibro- collagenous tissue in dermis}



Discussion

Patients with one classic autoimmune connective tissue disease (CTD) are likely to possess multiple autoantibodies with a small proportion developing symptoms and/or signs of another CTD (overlap syndrome).[1] The commonest systemic sclerosis overlap is seen with Myositis (34.2%) and infrequently, Systemic lupus erythematosus (SLE) (8.4%).[2] Scleroderma- Lupus Erythematosus overlap can occur either in a setting of systemic disease or with isolated cutaneous involvement. Most previous cases have been examples of Type 1 (Table 1)[1] though few have been Type 2, with localized scleroderma coexisting with SLE or vice versa (like ours).[1,3] In our case, histopathology from DLE-like lesions showed a combination of interface changes with peri-adnexal inflammation (typical of DLE) and dermal sclerosis (characteristic of scleroderma) which supported the diagnosis of Overlap syndrome. Anti-Ro antibodies can be detected in SS (70-100%), SLE (40-90%), SS/SLE overlap, subacute cutaneous lupus erythematosus and neonatal lupus erythematosus.[4] They are also strongly associated with photosensitivity (present in our patient). Although anti-Scl 70 antibodies are considered to be more prevalent in SSc with ILD (as in our case) indicating poorer prognosis, they may occur in up to 25% of SLE[5] carrying higher risk of pulmonary hypertension and renal involvement.[6] Interestingly, Raynaud's phenomenon was absent in our patient. MCTD was ruled out due to lack of sufficient criteria.[7] In view of photosensitivity and Anti-Ro antibodies, she will require stringent monitoring for early detection of SLE.

Table 1. Dermatologic classification of overlap syndromes in connective tissue diseases

Dermatologic classification of overlap syndromes in connective tissue diseases	
Type 1	Systemic disease overlapping with systemic disease
Type 2	Cutaneous disease (e.g. localized scleroderma, cutaneous LE) overlapping with systemic disease (e.g. systemic LE)
Type 3	Cutaneous disease (e.g. localized scleroderma) overlapping with cutaneous disease (e.g. cutaneous LE); overlap may occur with distinctive lesions developing at separate sites or clinical and/or histological features of both diseases within the same site (coincident overlap)

Conclusion

Overlap syndromes pose diagnostic difficulty and impact management strategies. Meticulous interpretation of clinical, histopathological and serological clues is warranted in such scenarios.

Conflict of Interest

The authors have no conflict of interest to declare.

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