Anogenital ulceration; an unusual presentation of mixed connective disease

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Abstract

Mixed connective-tissue disease (MCTD) is an autoimmune disorder with overlapping clinical features of systemic lupus erythematosus (SLE), scleroderma, and myositis, with the presence of antiU1-ribonucleoprotein (RNP) antibody. We report a case of a male patient with unusual clinical presentation of the condition.

Key words

Mixed connective-tissue disease, systemic lupus erythematosus, scleroderma, antiU1-ribonucleoprotein antibody.

Introduction

Mixed connective-tissue disease is an autoimmune disorder with overlapping clinical features of SLE, scleroderma, and myositis, with the presence of antiU1-RNP antibody. Manifestations of MCTD can be protean. Most patients experience Raynaud phenomenon, arthralgia/arthritis, swollen hands, sclerodactyly or acrosclerosis, and mild myositis. We present a case of a 36-year-old male with very unusual presentation of the condition resulting in misdiagnosis and wrong management for many years.

Case Report

A 36-year-old male presented to us with complaints of multiple small painful persistent ulcerations in the anogenital area (Figure 1 and 2) involving the natal cleft, the groins, perineal area and the penis (shaft and glans penis) for the last 9 years. Patient initially went to a surgeon when he started developing ulcerations in the natal cleft. He was diagnosed as a case of pilonidal sinus. Patient underwent surgery for the problem five times in the subsequent three years. In the meantime, patient also developed similar ulcerations bilaterally in his axillae for which he again received surgical treatment. Those lesions, however, had not recurred till date.

The patient sought dermatological opinion due to failure of response of his anogenital ulceration to surgery. Patient received multiple oral, as well as, injectable antibiotics; even antituberculous therapy was given for 1 month and then abruptly held. Eventually, patient was provisionally diagnosed to have pyoderma gangrenosum and was started on systemic steroids. Patient kept on taking high dose oral, injectable and even intralesional steroid therapy intermittently for 3-4 years. Patient showed partial response such that the ulcers would heal at the start of therapy only to recur within a month even with the continuation of steroids. Adversely, patient started having pain, first in his right hip joint and in a year also in his left hip. He started to have difficulty in walking, sitting and getting up from sitting position. His subsequent work up showed that he had developed avascular necrosis bilaterally of femoral necks due to prolonged use of steroids. Now for the last 3 years, patient has not taken any form of systemic steroid.
When the patient presented to us, there was not only history of aforementioned ulcers and but on questioning, patient also mentioned history of Raynaud’s phenomenon. When we examined the patient, we found two hyperpigmented sclerotic plaques, one on each lower leg (Figure 3). They had started to develop around 9 years back, first on the right leg and within a month on the left leg, as localized non-itchy, non-scaly erythematous plaques gradually becoming sclerotic over a period of 2 to 3 months. Since then these plaques had increased neither in size nor in number. Patient had never sought any medical advice for these lesions. All of his nails showed subungual hyperkeratosis, few of the finger nails showed ragged cuticles and dilated capillary loops. Mat-like telangiectasias were present on face and upper chest. Due to the repeated and prolonged use of systemic steroids, patient had also developed bilateral avascular necrosis of the femoral heads. There was no history of photosensitivity, oral ulcers, arthritis/ arthralgias, dysphagia or muscle weakness.

The only significant finding on his previous labs was a persistently high ESR. We advised serological tests for connective tissue disease.

His current labs showed an ESR of 70mm/1st hour, positive (+++) speckled antinuclear antibodies (ANA), raised titres of anti-RNP antibodies (41U/ml), Anti-Ku antibodies (40U/ml), anti-Sm RNP antibodies (76U/ml) and anti-Sm antibodies (15U/ml), (normal cut
off for all these antibodies was 12U/ml). Anti-ds DNA antibodies were negative.

Biopsy samples were taken both from the margin of one of the ulcers and from the sclerotic plaque.

Biopsy from the ulcer showed acanthosis in the epidermis while the dermis showed a tract lined by inflamed granulation tissue. Perivascular and periadnexal mixed inflammatory cell infiltrate comprising of neutrophils, plasma cells and lymphocytes was also seen (Figure 4). Biopsy from the sclerotic plaque on the legs showed irregular acanthosis and increased pigmentation of basal layer. Dermis showed mild perivascular and periadnexal infiltrate (Figure 5).

On the basis of clinical features including Raynaud’s phenomenon, fingernails ulceration, mat-like telangiectasia, cutaneous ulcerations, localized plaques of morphea and the lab findings, we diagnosed him to be a case of mixed connective disease. Treatment was started with mycophenolate mofetil 500 mg twice daily and hydroxychloroquine 200 mg twice daily along with topical fusidic acid/betamethasone for the ulcers and topical calcipotriol/ betamethasone for the sclerotic plaques. On a two weeks’ follow-up, the patient has started to show improvement with progressive healing of the ulcers (Figure 6).

Discussion

Mixed connective tissue disease is a rare connective tissue disorder. The condition has features of three more common connective tissue diseases, systemic lupus erythematosus, systemic sclerosis, polymyositis. It consists of the following core clinical and laboratory features. Raynaud's phenomenon, swollen hands, arthritis/arthritis, acrosclerosis, esophageal dysmotility, myositis, pulmonary hypertension, high level of anti-U1-RNP antibodies.

In a recent population based study, Raynaud's phenomenon was the most common initial symptoms (50%) followed by arthralgia (30%) and swollen hands (16%). The diagnosis was frequently delayed with the median time from first symptom to fulfillment of criteria of 3.6 years. At fulfillment of criteria, arthralgia was the most prevalent manifestation (86%) followed by Raynaud's phenomenon (80%), swollen hands (64%), leukopenia/lymphopenia (44%) and heartburn (38%).

In our patient, the major presenting complaint was that of the recurrent painful ulcers and sinuses in the crural area. However the morphology of ulcers, presence of nail fold telangiectasia, telangiectasia on chest and plaques of morphea on the legs raised the suspicion and hence autoimmune workup was advised. The positive ANA, anti-Sm antibodies and anti RNP antibodies confirmed the diagnosis of mixed connective disease (SLE plus systemic sclerosis).

We have reported this case to highlight two things. First, unusual presentations of a disease should always be kept in mind. Second, the importance of fine details of dermatological examination should not be underestimated. A thorough history, detailed examination and relevant investigations can save a patient from misdiagnosis, mismanagement and subsequent agony and can reduce the morbidity immensely.

References