Unusual presentation of scleroderma

Nithyananda K. Chowta, Mukta N. Chowta
Departments of Medicine and Pharmacology, Kasturba Medical College, Mangalore, India

SUMMARY
Scleroderma is a group of rare and complex diseases with varied clinical manifestations. The most obvious manifestation of the diseases is skin hardening and sclerosis. A 39-year-old woman presented with complaints of swelling of bilateral lower limbs. The patient also complained of spotty pigmentation of chest, back, and forearm. The patient gave history of weakness of left leg since 3 months. Antinuclear antibody (ANA), anticentromere, and anti-Ds DNA antibodies were negative. Clinical manifestations were unusual, as the patient had only edema and skin rashes. Visceral organ involvement was only in the form of abnormal liver function test especially lactate dehydrogenase (LDH). The patient also had grossly elevated creatine phosphokinase (CPK) suggesting muscle involvement, though symptom of muscle weakness was only mild. The patient was negative for antinuclear as well as anticentromere antibodies. Diagnosis in this patient was mainly based on skin biopsy findings.

Key words: Antinuclear antibody, pigmentation, scleroderma

Introduction
Scleroderma is a rare disease affecting skin and internal organs such as gastrointestinal tract, lungs, heart, and kidneys. The etiology is largely unknown, the role of genetic factors is unclear, but environmental factors, such as, polyvinylchloride, silica, and coal, are thought to be of importance in inducing symptom and to have implications in the progression of disease. It is characterized by alterations of the microvasculature, disturbances of the immune system, and by massive deposition of collagen. The most obvious manifestation of the diseases is skin hardening and sclerosis. Liver is rarely involved in scleroderma. The condition was first described by Curzio in the 1753. Initially, it was seen as a condition affecting the skin but in 1945, Goetz proposed a description of progressive systemic sclerosis. In addition, a limited scleroderma was described in 1964 by Winterbauer. He referred to a syndrome with Calcinosis, Raynauds phenomenon, Esophageal dysmotility, Sclerodactyly, and Telangiectasia [CREST syndrome]. We present here a case of scleroderma associated with elevated levels of lactate dehydrogenase [LDH] and creatine phosphokinase [CPK] and negative antinuclear antibodies.

Case Report
A 39-year-old woman presented with complaints of swelling of bilateral lower limbs. The patient also complained of spotty pigmentation of chest, back, and forearm. The patient gave history of weakness of left leg since 3 months. The patient did not have any history of joint pain. There was no history of fever, decreased urine output, jaundice, abdominal pain, vomiting, diarrhea, chest pain, orthopnoea, hypertension, diabetes mellitus, or tuberculosis. Also, there was no history of oral ulcer, dysphagia, hair loss, and menstrual abnormalities.

On examination, pulse was 84/min and blood pressure was 120/70 mmHg. Systemic examination was normal. There was induration with infiltration of face and periorbital puffiness. Dermatological examination showed spotty pigmentation with induration of skin in extensor forearm, depigmentation over upper back and front of the chest. Oral cavity was normal. Nails showed pallor. Laboratory investigations showed mild anemia [Hb-10.4g %]. Total leukocyte count, differential count, platelet counts, and ESR were normal. Peripheral smear was normal. Renal function tests [Urea, serum creatinine], routine urine analysis, 24-hour urine protein and 24-hour urine volumes were normal. Stool for occult blood showed only traces. Blood sugar was normal. Thyroid function tests were normal. Serum proteins [total protein, albumin, globulin] and serum bilirubin [total and direct] also normal. Other liver function tests were abnormal with grossly elevated...
provides the major classification criteria for systemic disease, but more commonly, there is a patchy and often widespread hyperpigmentation that simulates Addison's disease frequently seen in scleroderma. Patients may develop tissue components. Pigmentary skin changes are also characteristic of excessive deposition of collagen and other connective tissue components. The characteristic induration of the skin develops as a result of changes of scleroderma last several months before the symptoms of morning stiffness, swelling of the hands and fingers. Initially, there is swelling and edema of the skin with tightening of skin evolving through several stages.

The main clinical features center on the cutaneous manifestations, but the outcome depends on internal organ involvement. There is increase in the thickening and tightening of skin evolving through several stages. Initially, there is swelling and edema of the skin with swelling of the hands and fingers. Frequently, the initial complaint is of symptoms of morning stiffness, and pain in the joints of the hands. The initial skin changes of scleroderma last several months before the characteristic induration of the skin develops as a result of excessive deposition of collagen and other connective tissue components. Pigmentary skin changes are also frequently seen in scleroderma. Patients may develop widespread hyperpigmentation that simulates Addison's disease, but more commonly, there is a patchy and often follicular hypo- or hyperpigmentation. Involvement of the skin proximal to the metacarpophalangeal joints provides the major classification criteria for systemic sclerosis. Raynaud's phenomenon is an almost inevitable finding in the majority of these patients. Esophageal involvement in the form of esophageal dysmotility occurs in all types of scleroderma. As reported in the epidemiological study by Tamaki et al., the characteristic signs of systemic sclerosis were as follows: proximal scleroderma, 75%; sclerodactyly, 91%; pitting scars, 49%; short sublingual frenulum, 49%; pulmonary fibrosis, 45%; diffuse pigmentation, 45% and phalangeal contracture, 35%. Raynaud's phenomenon was present in 93% of patients and was the initial symptom in 59% of cases. Clinical manifestations in the present patient were unusual, as the patient had only edema and skin rashes. Visceral organ involvement was only in the form of adrenal gland, though symptom of muscle weakness was only mild. Muscle involvement in scleroderma is reported in the earlier literature also. There were no signs and symptoms of esophageal dysmotility, but endoscopy has revealed esophageal ulceration. There were no signs of Raynaud's phenomena also. The patient was negative for antinuclear as well as anticientromere antibodies. Diagnosis in this patient was mainly based on skin biopsy findings.

We initially considered mixed connective tissue disease (MCTD) as a differential diagnosis. MCTD was first described in 1972 as a disease syndrome with overlapping features of systemic sclerosis, systemic lupus erythematosus (SLE), and polymyositis associated with antibodies to RNAse-sensitive extractable nuclear antigen. When the antigen was subsequently characterized as polypeptides on the U1 ribonuclear protein component of the splicesosome (U1RNP), MCTD became the first rheumatic disease syndrome to be defined by a serologic test. Clinical features include a high frequency of Raynaud's syndrome, swollen hands, sclerodactyly, arthritis, polymyositis, and interstitial lung disease. As in our patient, the skin biopsy was convincing with the diagnosis of scleroderma and also patient did not have any history of joint pain, diagnosis was more in favor of scleroderma. There was no history of occupational exposure to PVE or perchlorethylene. Hence, this possibility was ruled out.

ANAs are a typical finding in 90% of patients. The presence of antiDNA antibody and anti-Sm antibody are not seen in contrast to that of SLE. The more typical antibodies seen in scleroderma are anticientromere antibodies. These are found in up to 90% of patients with limited systemic sclerosis. In contrast, they are found in only 10% of patients with diffuse scleroderma. Anti-Scl-70 antibodies seen in up to 40% of patients with diffuse scleroderma. It is not usually seen in limited
scleroderma. Patients with a negative antibody profile have fewer clinical and laboratory manifestations. The diagnosis is a clinical one, based on the history and the examination. The antibodies may be useful especially the anti-Scl-70 and the anticentromere antibody. However, a bedside technique of nailfold capillary examination reveals an abnormal nailfold capillary network. This is of predictive value in patients with Raynaud's phenomena. Other investigations are dependent on the clinical complaint.[8] The differential diagnosis of the skin changes of scleroderma includes linear scleroderma, with localized bands of skin thickening and erythema and with regional atrophy of the sub-cutaneous tissues. Morphea is seen as a plaque of erythematosus lesions with central hypopigmentation. Eosinophilic fasciitis is characterized by a shiny and erythematous rash usually involving the dermis of the forearms and legs, and occasionally the hands and trunk. There is inflammation of the dermis with thickening. Biopsy, however, shows an infiltrate of eosinophils.[2]

Chowta and Chowta: Unusual presentation of scleroderma

References


Source of Support: Nil, Conflict of Interest: None declare