Case Report

Mixed Connective Tissue Disease- A Rare and Distinct Rheumatological Case Study

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Abstract

In this article we present this 35 year old lady with history of fever, multiple joints pain with swelling and skin conditions such as tightening, glistening and bluish discoloration while exposure to cold. After taking detailed history and clinical examinations, she was found to have features consistent with multiple rheumatological conditions such as systemic lupus erythematosus and systemic sclerosis. With directed and specific investigation she was diagnosed to have Mixed Connective Tissue disorder.

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Case report

Mrs. Beauty Begum, 35 years old housewife, mother of two kids, hailing from Tangail presented with the complaints of intermittent fever, generalized bodyache, multiple joints pain with swelling involving hands, knees and ankles for the 3 months duration. She did not have any morning stiffness, skin nodules or shortness of breathing. She also noticed that her skin of face, forehead, arms and fore-arms has become tightened for 1 month. On further query, she added that she is having pain and numbness of hands while handling water (especially cold). There is change in skin colour and experienced pain under the skin which is not related to the joint pain. She does not give any history of leg swelling, chest pain, dryness of eyes or mouth, difficulty in swallowing or hair loss.

On clinical examination, she had expressionless face, tight and glistening skin of face with loss of wrinkles on forehead. The perioral skin area was tightened and she could not open her mouth fully as she could have previously. The skin of arm, fore-arm was tightened and fingers examination revealed sclerodactyly with superficial ulcers in metacarpophalangeal and interphalangeal joints of both hands. There is pain, swelling and tenderness of joints of hands, ankles and knees but no deformity. There was no cervical spine or joints lower back involved. Her BP was 110/70 mmHg, pulse rate 96, Temp-101°F.

In clinical summary she had fever, polyarthritis, sclerodactyly, Raynaud's phenomenon, sclerosis and calcinosis of skin which are suggestive of combined features of SLE and systemic sclerosis. There was no indication of other rheumatological conditions such as rheumatoid arthritis or dermatomyositis or polymyositis etc. She did not have any shortness of breath, dysphagia, peripheral oedema that could suggest pleuropulmonary, gastrosophageal or renal involvement. The provisional diagnosis was Mixed connective tissue disease. Overlap syndrome of rheumatological diseases and undifferentiated connective tissue disease (UCTD) were included in differential diagnoses.

Fig1: Expressionless face, tight and glistening skin

The relevant investigations were done and reports are given below.

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The characteristic lesions in the involved organs are intensive obliteratorive, proliferative vascular lesions in large, medium and small vessels with less inflammatory infiltrates.

In the early phases of the MCTD many patients complain of easy fatigability poorly defined myalgias, arthralgias and Raynaud’s phenomenon; systemic involvements are as follows-

- Fever of unknown origin may be the presenting feature of MCTD. The most common skin change is the Raynaud’s phenomenon, others are malar rash, generalized erythematous rash.
- Raynaud’s phenomenon is seen in 75%-100% of patients. Approximately 60 percent of patients develop an obvious arthritis. Myalgias and myositis can be seen in 50%-60% of patients.
- The gastrointestinal symptoms may range from heartburn, dysphagia, diarrhea and symptoms of malabsorption. Disordered motility in the upper gastrointestinal tract is the commonest problem.
- Pleuropulmonary involvement is common, it may be asymptomatic or the patient may present with pleurisy and effusion, interstitial lung disease, pulmonary arterial hypertension (PAH).
- All three layers of the heart may be involved in MCTD with abnormal ECG findings.
- Neurological manifestations are less common, presenting as aseptic meningitis, trigeminal neuralgia, demyelination, transverse myelitis and peripheral neuropathy.
- The absence of severe renal disease is a hallmark of MCTD; it is possible that high titers of anti-RNP antibodies, which are characteristic of MCTD, may protect against the development of diffuse proliferative glomerulonephritis. But patients may have glomerulonephritis, nephritic syndrome or renal crisis of scleroderma.

**Discussion**

Diagnosis is clinical along with supporting laboratory investigations. There criteria are-

- **A. Serological criteria**: positive Anti RNP antibody.
- **B. Clinical Criteria**: (1) Swollen hands (2) Synovitis (3) Myositis (4) Raynaud’s phenomenon (5) Acrosclerosis. MCTD is present if A is associated with three or more clinical criteria.
The laboratory findings are anemia, leucopenia, elevated erythrocyte sedimentation rate, hypergammaglobulinemia (100%), positive combs test, and Rheumatoid factor positive in 50 to 70% of patients. Antinuclear antibody positivity is seen in 100% of patients in high titre with coarse speckled pattern. Anti U1RNP antibodies by haemagglutination test is highly characteristic feature of MCTD. Many patients also make antibodies directed against hnRNP-A2, fibrillin-1, and nucleosomes, but not to RNA polymerases. The absence of anti-Sm antibodies and anti-DNA antibodies in a seropositive for anti U1RNP is an important discriminating finding for MCTD from SLE. Also scleroderma-specific antibodies, including anticentromere and anti-Scl-70 (topoisomerase) are absent. Antiphospholipid antibodies occur, but are less common than in those with SLE.

No controlled trials have been performed to guide therapy. Instead, the management of patients with MCTD generally rests upon the effectiveness of specific therapies for similar problems seen in SLE, scleroderma, or polymyositis. By comparison, scleroderma-like features (eg, Raynaud phenomenon, pulmonary hypertension) are usually less responsive to therapy. NSAIDs are given for pain, calcium channel blockers for PAH. Immunosuppression (steroid and other agents) is particularly required in cases with severe arthritis, pulmonary hypertension and serositis involving pericardium or pleura.

MCTD has relatively good prognosis in view of low prevalence of serious renal disease and life-threatening neurologic problems. Morbidity is quite high in patients with MCTD due to multiple factors including recurrent musculoskeletal pain, fibromyalgia, gastroesophageal acid reflux etc. Mortality associated with MCTD ranges, in different studies, from 16 to 28 percent at 10 to 12 years. Those patients with more features of scleroderma and polymyositis had a worse prognosis. The major causes of death include progressive pulmonary hypertension and its cardiac complications.

**References**