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A Woman with Mechanic's Hands

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A 31-year-old woman presented with shortness of breath on exertion for the past six months. The symptoms had progressively worsened during the past month. She had an episode of chest infection before her visit to the clinic. She also complained of intermittent lower limb weakness with bilateral large joint pain leading to difficulty in ambulation. In addition, she experienced intermittent episodes of non-specific joint pain of the hands and wrist associated with skin rash. She had no known medical illness.

On examination, she was afebrile, mildly tachypnoeic, with normal vital signs. Examination of the hands showed hyperpigmentation and hyperkeratosis of the palms, as shown in Figure 1. There was no joint deformity or rash. Chest examination revealed bilateral fine inspiratory crepitations. The rest of the examination was unremarkable. Radiographic images are shown in Figures 2 and 3.

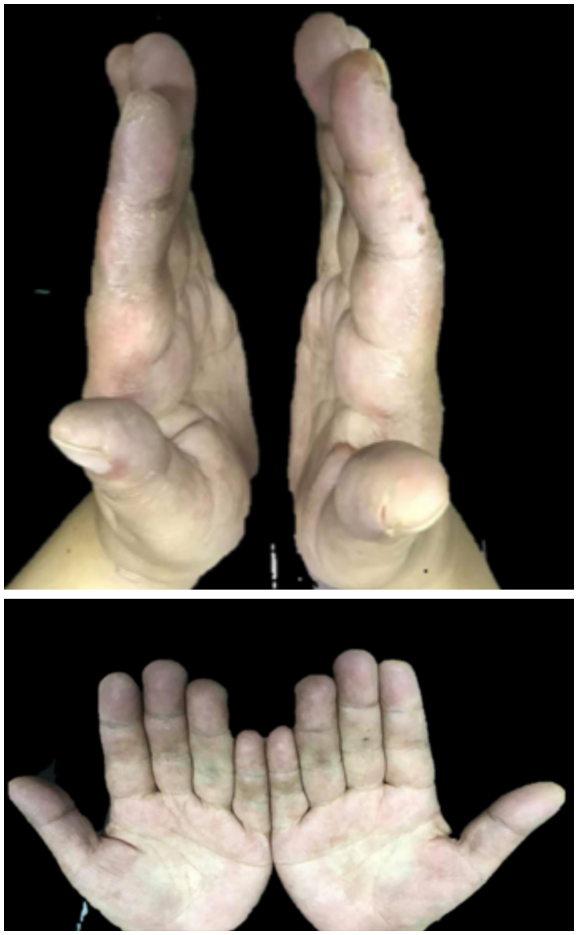

Figure 1: Hyperpigmentation and hyperkeratosis over the palmar aspect.

Question

1. What is the most likely diagnosis?
   a) Anti-synthetase syndrome.
   b) Systemic sclerosis.
   c) Rheumatoid arthritis.
   d) Systemic lupus erythematosus.

Answer

a) Anti-synthetase syndrome.

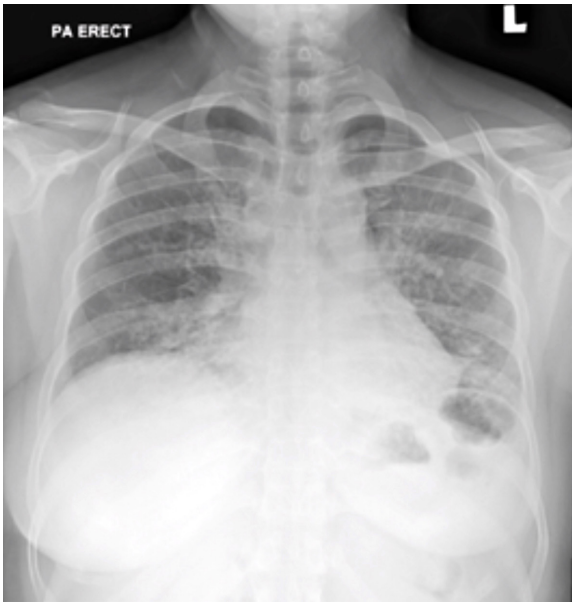

Figure 2: Chest X-ray of a 31-year-old woman.

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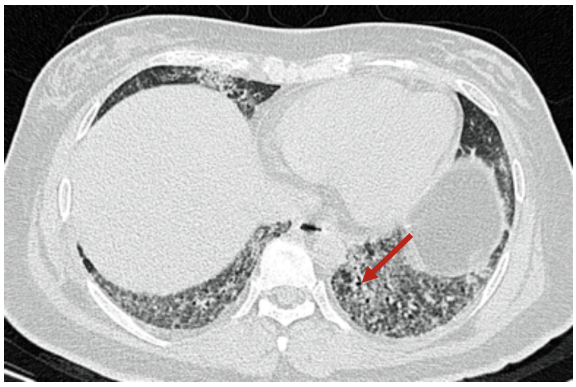

Figure 3: High-resolution CT scan of the thorax shows lower lobe predominant ground-glass opacity with peri-bronchovascular predominant, traction bronchiectasis (arrow), and reticulation.

DISCUSSION

Anti-synthetase syndrome is an autoimmune disorder characterized by the presence of autoantibodies to aminoacyl tRNA synthetases with clinical manifestations that may include interstitial lung disease (ILD), myositis, non-erosive arthritis, Raynaud's phenomenon, and/or mechanic's hands.[1,2] It is also known as one of the idiopathic inflammatory myopathies apart from dermatomyositis and polymyositis, with a higher rate of ILD. The ILD which occurs in anti-synthetase syndrome is far more severe and has a higher mortality rate. Anti-synthetase syndrome is a rare disorder with female predominance. The age of onset ranges from late adolescents to old age with a mean average age in the 50s.[2,3]

According to the revised criteria in 2011, the presence of anti-aminoacryl tRNA synthetase, two major, or one major with two minor criteria, is needed to make the diagnosis of anti-synthetase syndrome.[2] Major criteria include ILD, polymyositis, or dermatomyositis. The minor criteria include arthritis, Raynaud's phenomenon, and mechanic's hand.[2] Nevertheless, the absence of anti-synthetase antibody may not exclude the diagnosis since the antibody levels fluctuate with disease activity and must be interpreted with caution.

Patients with anti-synthetase syndrome may present with various clinical presentations and might mimic other connective tissue diseases. Chronic cough and progressive shortness of breath indicating ILD are reported to predominate during the initial presentation (86%), followed by myositis (73%) and arthritis (63%).[3] In patients with ILD, mechanic's hand, which is hyperkeratosis and hyperpigmentation characterized by usually a subtle sign that could be missed.[1] Depending on the clinical presentation, additional investigations may be warranted. However, any patient who presents with unexplained fever, progressive lung disease, and arthritis should be investigated for this condition.

The presence of anti-synthetase antibodies is the hallmark of this condition. The most commonly identified is anti-Jo-1, an anti-histidyl-tRNA synthetase.[4] Other myositis-specific antibodies are sometimes sent for excluding other diagnoses. High-resolution CT (HRCT) scan is required as a routine work up for patients presenting with respiratory signs and symptoms in anti-synthetase syndrome. The most common clinical findings are traction bronchiectasis, ground-glass opacities, and reticulation.[1,3,4]

Lung biopsy is infrequently performed in patients with anti-synthetase syndrome as diagnosis is typically made by analyzing high-resolution computed tomography findings, serological testing, pulmonary function testing, physical examination, and patient symptoms.[1] Skin and muscle biopsy may help understand the disease severity and the likelihood of response to therapy.[1,2]

Other differentials for this diagnosis are rheumatoid arthritis, inflammatory myopathies, systemic sclerosis, and systemic lupus erythematosus (SLE) since some features may overlap.[3,4] The age of onset may be similar in these diagnoses; however, each of the systemic rheumatic diseases has their own characteristics and pathognomonic features that differ from each other such as cutaneous manifestations in dermatomyositis, scleroderma, and SLE, as well as distribution of arthralgia. In general, patients with anti-synthetase syndrome have more pronounced muscle and lung disease.

As for the management of this condition, a multidisciplinary approach including both a rheumatologist and a respiratory physician is recommended. The first-line treatment is a corticosteroid which acts as an anti-inflammatory and immunosuppressive agent. However, most patients with this disease require immunosuppressive therapy such as azathioprine, tacrolimus, and cyclophosphamide.[1–4]

Patients with anti-synthetase syndrome should be followed up closely for monitoring progression of the ILD, myositis, and other associated

comorbidities such as pulmonary hypertension and risk of malignancy.[1] The five-year cumulative survival rate is reported to be 90% for patients with Jo-1 antibody and 75% for non-Jo-1 patients.[5] The most common cause of death is pulmonary fibrosis and pulmonary hypertension.[1,5] Hence, physicians should have a high index of suspicion since anti-synthetase syndrome has a diverse presentation.

Disclosure

The authors declared no conflicts of interest. Written and verbal informed consent was obtained from the patient.

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