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Case Report

Antisynthetase Syndrome: An under recognized rare disease in a developing country

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Abstract

Anti-synthetase syndrome (ASS) is an autoimmune disease characterized by the presence of auto antibodies against one of many aminoacyl transfer RNA synthetases and spectrum of clinical manifestations. Here we report a case of a 52 year old lady who had been extensively evaluated for progressive muscle weakness in a tertiary care center, diagnosed as Antisynthetase syndrome which was evidenced by the presence of antisynthetase antibody, dermatomyositis, Raynaud's phenomenon, arthritis and mechanics hand. Despite aggressive management with intravenous immunoglobulins, steroids and immune suppressors she died because of underlying respiratory failure causing difficulty in weaning off ventilator.

Keywords

Antisynthetase Syndrome, developing country, Muscle weakness.

Introduction

Anti-synthetase syndrome is a rare systemic autoimmune syndrome, characterized by the presence of anti-aminoacyl-tRNA antibodies accompanied by a constellation of clinical findings including myositis, interstitial lung disease (ILD), mechanics hand, fever and Raynaud's phenomenon. This occurs mainly in adults and more common in females and etiology is not known. There are several anti-synthetase antibodies and anti-Jo-1 is the commonest and occurs in 80 % of patients with anti-synthetase syndrome.(1) Immunosuppressive agents, such as azathioprine, mycophenolate mofetil, cyclophosphamide and/or tacrolimus, are usually required in addition to corticosteroids for management of the myositis and pulmonary manifestations of anti-synthetase syndrome.(2)

Case report

A 53 year old lady who was independent of her day to day activities presented with progressive muscle weakness associated with pain, inflammatory polyarthralgia, Raynaud's phenomenon and severe constitutional symptoms for one month duration. On examination, she had facial rash, inflamed tender small joints, mechanics hands and evidence of proximal myopathy with severe muscle tenderness. Her other systemic examinations revealed no abnormalities.

Baseline hematological and biochemical investigations were normal except elevated liver enzymes. Inflammatory markers were high (ESR 79mm1st hour; CRP 59.2mg/l). The muscle biomarkers including AST, LDH and Creatinine phosphokinase were found to be elevated in the values of 1218U/L, 5463U/L and 6351U/L respectively. Serial blood and urine cultures were negative. The electrocardiogram showed T inversions in inferolateral leads, Troponin I was positive and, the echocardiogram revealed moderate left ventricular dysfunction. Chest radiograph showed bilateral pleural effusion. HRCT chest showed bilateral mild pleural effusion. The electromyogram showed a generalized myopathy with active denervation features and muscle biopsy revealed focal muscle fiber atrophy in perifascicular area. Her autoimmune profile showed positive Anti-Nuclear Antibody with the titer of 1: 160, negative dsDNA, normal complement levels, positive rheumatoid factor and positive anti synthetase antibody.

Considering her clinical presentation and investigations she was diagnosed as Antisynthetase syndrome which was fulfilling the Soloman et al 2011 criteria complicating myositis induced cardiac involvement. During the evaluation of possible antisynthetase syndrome she developed respiratory

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arrest, intubated and ventilated. She had been started on 2g/kg of intravenous immunoglobulin, intravenous methylprednisolone pulse therapy followed by high dose of oral steroids (1mg/kg/day) after excluding all the possible infective causes. Although she had significant clinical improvement to the treatment modalities which was evidenced by normal muscle biomarkers within couple of weeks of treatment, it was difficult to wean her off from the ventilator which led to her death.

Discussion

Antisynthetase syndrome is a rare clinical entity which is less reported in Asian population, particularly few case reports are available in Sri Lanka. Soloman et al criteria 2011 is used to diagnose antisynthetase syndrome which includes the required criteria of presence of antisynthetase antibodies, one of the two major criteria which includes Dermatomyositis / Polymyositis and ILD, and two of three minor criteria including arthritis, Raynaud's phenomenon and mechanics hand.(2)

There are few case reports available in Sri Lanka regarding ASS associated with ILD, but the association with Dermatomyositis as in this case is less.(3) ILD mostly occurs in association with ASS with significant poor outcome. Although the absence of ILD had been a good prognostic indicator in this patient, the myositis causing respiratory muscle paralysis in this patient was associated with adverse outcome.

The myositis induced cardiac involvement is observed in this patient, while other cardiac manifestations of ASS include congestive heart failure, pulmonary hypertension, pericarditis and myocarditis.(4)

When a critically ill patient develops cluster of symptoms including muscle and lung involvement the possibility of ASS should be raised and sophisticated investigations to be done to diagnose it early which leads to prompt, timely management with aggressive immunosuppressive treatment.

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