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Case Report

A rare case of serologically diagnosed overlap syndrome presents as an idiopathic inflammatory myositis without any overlapping features

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ABSTRACT

Overlap syndrome is a rare inflammatory rheumatic condition that shares features suggestive of at least two distinct autoimmune diseases with a reported prevalence of less than 34/100,000 persons and an incidence of less than 20/million/year. One example of an overlap syndrome is the presence of dermatomyositis or polymyositis with other autoimmune afflictions such as systemic lupus erythematosus (SLE), rheumatoid arthritis (RA), or Sjogren's syndrome. We present a case report of a 22-year-old male presented with progressive weakness in both upper and lower limbs without any other significant complaints. On investigation, serology revealed antibodies suggestive of overlap syndrome, which on further investigations, categorized as idiopathic inflammatory myositis (IIM). This shows that overlap syndrome, as an IIM, is one of the differential diagnoses in patients presenting with progressive extremity weakness involving both extremities even if there is no involvement of any sensory function.

Keywords: Overlap syndrome, IIM, Autoimmune, Differential diagnoses

INTRODUCTION

Overlap syndrome is an uncommon medical condition characterized by the presence of clinical features of multiple different and widely recognized rheumatological disorders. The disorders commonly involved are SLE, RA, systemic sclerosis (SSc), Sjogren's syndrome or IIM.

IIM is a condition that typically presents with symmetrical progressive weakness in proximal muscles of the extremities, which mainly encompasses polymyositis (PM), dermatomyositis (DM), juvenile myositis (JM), overlap myositis (OM), myositis associated with cancers (MC), immune-mediated necrotizing myopathies and sporadic inclusion body myositis (sIBM).[1] IIM is extremely rare with an incidence ranging from 1.16 to 19 per million persons at risk per year and prevalence ranging from 2.4 to 33.8 per 100,000 persons.[2] Because it is common to find overlap features clinically, Troyanov et al. provided a new classification for IIM which centers around the overlap clinical features as well as utilizes myositis-associated autoantibodies (MAA), and myositis-specific autoantibodies (MSA), coining a new term "overlap myositis" to define IIM associated with clinical features of other connective tissue disorders (CTDs).[3] The diagnosis of IIM is based on clinical features, elevated creatine kinase, radiological imaging, and histopathological work-up of a skeletal muscle biopsy.[4]

We present a rare case presentation with findings suggestive of overlap syndrome upon work-up, i.e., autoantibodies, radiological imaging, and histopathological findings, but without any clinical overlap feature of a connective tissue disorder (CTD). A study has shown that only 15% of patients with OM present without overlapping clinical features, which typically develop at follow-up.[5]

CASE REPORT

A 22-year-old male came to OPD with a complaint of progressive weakness bilaterally in both upper and lower limbs for four years. He had no other significant complaints, including no difficulty in swallowing, breathlessness, fever, or bowel or bladder incontinence. When asked, past, family, and personal history were unremarkable. His vitals stable throughout admission.

On physical examination, tone was reduced in both upper and lower limbs as shown in Table 1.

Table 1: Clinical assessment of extremity power.

Power	Upper limbs	Lower limbs
Proximal	3/5	2/5
Distal	4/5	4/5

Deep tendon reflex (DTR) was also absent in this patient. Routine blood investigations showed hemoglobin of 14.8 gm/dl, total leukocyte count of 4,500/cmm, and platelet count of 81,000/cmm. Total creatine phosphokinase was 4920 on admission. Nerve conduction velocity measurement showed axonal pure motor neuropathy. Immunofluorescence testing showed a positive titer (1:160) for anti-nuclear antibody (ANA) with cytoplasmic pattern. Full ANA profile showed antinuclear ribonucleoprotein (anti-RNP), anti-smith (anti-Sm), anti-U1 ribonucleoprotein (anti-U1 RNP), anti-Ro/SSA and anti-La/SSB antibodies with significant titers. That is, both myositis-specific autoantibodies (MSA) and myositis-associated autoantibodies (MAA) were positive in this patient on auto-antibody testing.

Since muscle biopsy is one of the criteria for diagnosis of IIM overlapping with systemic disease, an MRI was done of the thigh region, to suggest sites of biopsy. It showed patchy areas of hyperintensity in muscles of the pelvic girdle and anterior thigh bilaterally upon T2-weighted short-tau inversion recovery (T2w-STIR) imaging indicating the presence of edema in muscles, Figure 1.

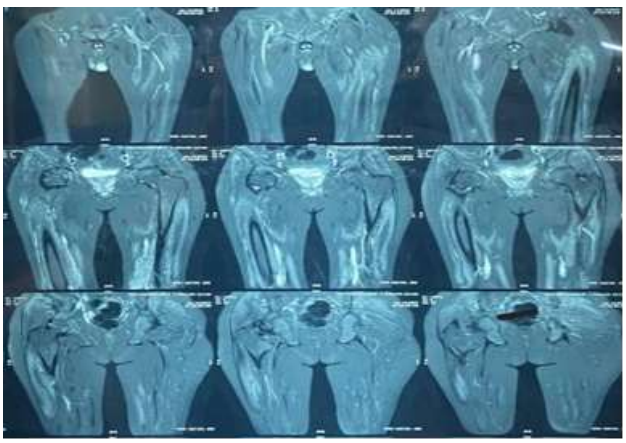

Figure 1: MRI (Coronal-section) of thigh region showing patchy areas bilaterally.

Biopsy was taken from vastus lateralis and the histopathological investigation was suggestive of polymyositis, shown in Figure 2 (low-power field) and Figure 3 (high-power field).

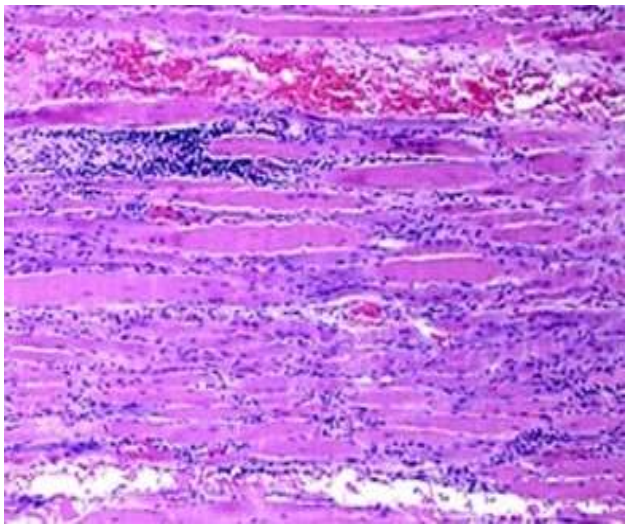

Figure 2: Histopathological slide of muscle biopsy from vastus lateralis (low-power field) showing lymphocytic infiltration between the muscle fibers.

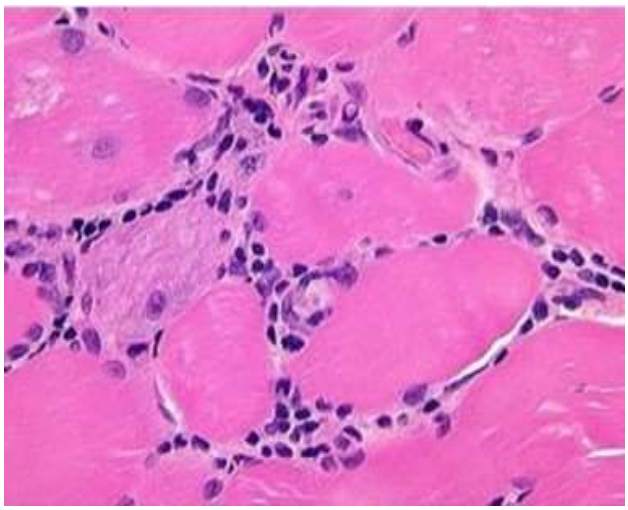

Figure 3: Histopathological slide of muscle biopsy from vastus lateralis (high-power field) showing endomysial lymphocyte infiltration, suggesting features of polymyositis.

As the patient didn't have any significant signs and symptoms other than weakness in extremities, further investigations were done to exclude scleroderma which included, barium swallow (Figure 4) and high-resolution computed tomography (HRCT), and both didn't show any significant findings (Figure 4).

For management, a rheumatologist's consultation was done, and based upon his recommendation patient was given five cycles of IVIG along with other immunologics. After that, the patient was kept on

maintenance steroid therapy along with azathioprine. Upon follow-up, the patient did not have significant improvement in muscle function which prompted another cycle of IVIG. But despite multiple immunologics, the patient has not yet shown improvement clinically and the prognosis is guarded.

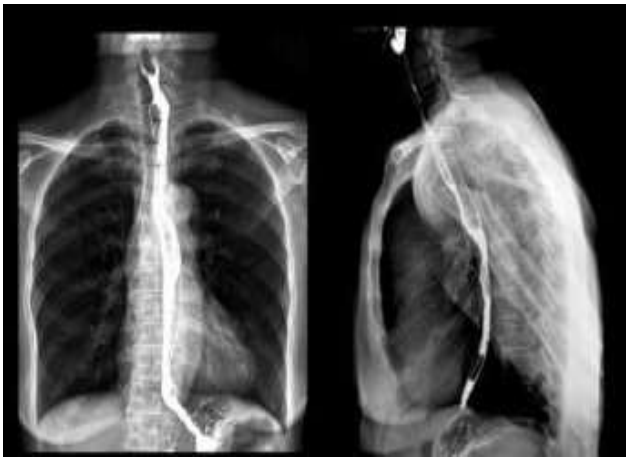

Figure 4: Barium swallow (Postero-anterior view and lateral view) showing no signs of dysmotility.

DISCUSSION

Idiopathic inflammatory myositis (IIM), an immune-mediated condition, consists of a heterogeneous group of acquired muscle diseases.[6] Patients with IIM typically present with sub-acute to chronic onset of proximal muscle weakness manifested by impaired getting up from the chair, climbing stairs, and lifting their arms. Extramuscular manifestations may also occur such as joint, skin, and lung involvement. Diagnosis may be confirmed with laboratory investigations, including raised serum creatine kinase (CK), myositis-specific autoantibodies (MSA), and myositis-associated autoantibodies (MAA), which may also help in differentiating clinical phenotype. Nonetheless, muscle biopsy continues to be the gold standard for diagnosis.[7,8]

Overlap syndrome with myositis (overlap myositis, OM) is defined as the presence of clinical features of myositis along with overlapping features suggestive of another connective tissue disease (CTD), like SSc, SLE, as well as Raynaud's phenomenon, arthritis, interstitial lung disease (ILD), and a presence of "overlap autoantibodies" including MSA and MAA.

However, approximately 15% of patients with OM do present with overlap autoantibody, but without any overlap clinical features, often with biopsy findings suggestive of myositis,[5] making this patient presentation even rarer, although patients with this presentation typically develop overlap features on follow-up. That is, a patient suffering from OM may initially present with clinical features suggestive of myositis only, i.e., without any overlap feature, confounding the original diagnosis. But broad analysis of auto-antibodies associated with myositis and histopathological examination may help define the diagnosis more distinctly, as seen in this patient whose histopathological examination was suggestive of polymyositis and tested positive for myositis-associated antibodies (MAA), like anti-Ro and anti-La antibody, and positive for myositis-specific autoantibodies (MSA), like anti-U1 RNP antibody.

This emphasis that OM should also be one of the differential diagnoses when a patient presents solely with clinical features of myositis, i.e., bilateral, progressive muscular weakness and no other significant symptoms because it is important to manage cases of OM in the early course of the disease, particularly at the onset of the disease, to closely monitor for development of possible complications, as at later stages of pathogenesis, patient end-up having involvement of many different organs and body parts, and the prognosis is usually worse.[9,10]

There is not enough literature available guiding how to organize patients' treatment because IIM is uncommon and heterogeneous. Corticosteroids are the first line treatment with prednisone given in adult-onset mild cases. Intravenous immunoglobulins (IVIG) can be used for refractory patients or as an add-on during relapses.[7,8]

CONCLUSION

In summary, overlap myositis (OM) should be in the list of differentials in patients presenting with clinical features suggestive of IIM like progressive and bilateral muscle weakness, even if there are no overlap features of CTD to be able to provide prompt and adequate treatment to get a better response to the provided therapy and to closely monitor for possible complications.

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