Clinical heterogeneity of anti-PM/Scl positive conditions

Introduction

Although clinical phenotypes are usually linked to specific serological markers, sometimes the clinical appearance does not match the expected picture. This article introduces a case of anti-PM/Scl myositis which had a different clinical spectrum to that expected from the patient's serological markers. No interstitial lung disease or scleroderma features were seen. Magnetic resonance imaging and biopsy findings showed neither significant inflammation nor necrosis. After aggressive treatment with immunosuppressant drugs the patient achieved clinical remission and functional recovery.

Discussion

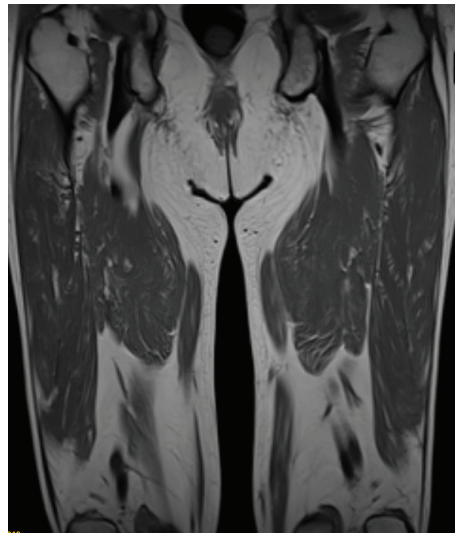
Anti-PM/Scl antibodies (anti-PM/Scl), also known as the human exosome complex, are a rarely found type of anti-nuclear antibodies included in the group of myositis-associated autoantibodies. They can be identified in up to 8–10% of cases of inflammatory myopathies. Usually, they have been reported to be linked to systemic sclerosis overlap syndrome, a clinical condition that combines Raynaud's phenomenon, myositis and arthritis, described as 'scleromyositis'. This overlap syndrome is also characterized by the presence of interstitial lung disease and has an overall good prognosis because of the absence of hidden tumours (Mahler and Reijmakers, 2007).

More recently, however, examples of other entities related to this serological hallmark have arisen. Reports of Sjögren's syndrome anti-PM/Scl positive (Vandergheynst et al, 2006), mixed connective tissue disease anti-PM/Scl positive (Muro et al, 2015), inclusion body myositis anti-PM/Scl positive (Selva-O'Callaghan et al, 2003) or even melanoma (Rajimakers et al, 2004) exist in the literature. This article introduces a case of isolated myositis which was anti-PM/Scl positive without scleroderma features or lung involvement in a patient with normal magnetic resonance imaging, electromyoneurogram and biopsy findings, who showed a good response to steroid and immunosuppressant drugs.

The authors believe that the relatively good prognosis of this autoantibody profile as well as the steroid treatment may explain the normal biopsy findings. According to classical accepted criteria (Bohan and Peter) for inflammatory myopathies, this case should be classified as probable polymyositis. The lack of any other plausible diagnosis and the response to treatment support this hypothesis.

This is a case of an anti-PM/Scl myositis with a diverse clinical pattern. This lack of homogeneity differs from the

Figure 1. Magnetic resonance imaging showing no oedema or necrosis.



CASE REPORT

A 60-year-old man came to the accident and emergency unit complaining of 10 days of continuous pain in both thighs and shoulders, and pelvic girdle weakness. He had alcoholic liver disease and alcoholic polyneuropathy so he was taking furosemide, spironolactone, folic acid and vitamins. He had recently being diagnosed with dyslipidaemia for which he was taking atorvastatin. He denied drinking alcohol for the last few months or taking any other drugs. Upon physical examination the patient was not feverish, he had no rashes or skin thickening, and cardiopulmonary auscultation was normal. Strength assessed by Medical Research Council scale was symmetrically and proximally diminished in both legs and arms. He was unable to comb his hair or lift his arms over his shoulder. He also had problems getting up from an armchair using his arms. No features of arthritis were found and temporal artery pulses were preserved.

As lab results showed negative blood alcohol test, high levels of inflammatory markers and a creatine phosphokinase level of 1446 U/litre (normal range <200 U/litre) the differential diagnosis included statin-induced side effects and an inflammatory myopathy. Creatine phosphokinase was elevated between five to ten times above normal in three different samples. Giant cell arteritis was also suspected because of the clinical picture of proximal weakness (pelvic and shoulder girdle claudication).

Temporal artery biopsy showed normal findings. He denied cranial symptoms and his erythrocyte sedimentation rate was below 55 mm/hr. An electromyogram was performed and magnetic resonance imaging showed normal findings (*Figure 1*).

Extended laboratory tests were performed and anti-PM/Scl antibodies were found by immunoprecipitation with negative antinuclear antibodies. Methotrexate and high dose prednisone were started and a clinical response achieved. Afterwards, biopsy of the left quadriceps muscle revealed no inflammatory infiltrates or necrosis but selective atrophy of type 2 fibres. A computed tomography body scan during follow-up was normal.

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classical concept of the myositis-associated autoantibodies causing a recognizable pattern of disease. Even so, the evident pathogenic role of these antibodies (between 43% and 88% of the patients positive for anti-PM/Scl antibodies are diagnosed as having a myositis/scleroderma overlap syndrome) (Rajimakers et al, 2004) and the absence of other concurrent serological findings may strengthen the diagnosis. Further studies in larger cohorts are needed to define the clinical significance of anti-PM/Scl antibodies in autoimmune diseases. **BJHM**

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LEARNING POINTS

- Anti-PM/Scl conditions, also called scleromyositis, usually combine features of myositis and scleroderma.
- Although clinical phenotypes are usually linked to specific serological markers, sometimes the clinical appearance does not match the expected picture.
- An inflammatory myopathy should be suspected and treated when the clinical picture and laboratory findings are consistent, even in the absence of biopsy findings.

Hydropneumothorax and intrapulmonary metastases secondary to breast cancer recurrence

A 48-year-old woman, who had previously been treated for stage IIB progesterone-positive breast cancer, presented with a 3-month history of cough and worsening breathlessness. Examination revealed percussion dullness and reduced air entry in the right lung. Plain chest radiograph showed right-sided hydropneumothorax and multiple left-sided pulmonary nodules (*Figure 1*). Despite chest drain insertion, the right lung failed to re-expand.

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Subsequent chest computed tomography revealed loculated hydropneumothorax and pleural thickening (*Figure 2a*). Additionally, the right lung was collapsed with ipsilateral areas of reduced enhancement, suggestive of a large metastasis occluding the right main bronchus and impinging the right pulmonary artery (*Figure 2b*).

Pleural aspirate analyses revealed an exudative effusion with no malignant cells or microorganisms. Biopsy of a distant metastasis and the lung pleura during subsequent video-assisted thoracoscopy both showed breast adenocarcinoma confirming disease relapse. The lung was 'trapped' and decortication was inappropriate. The patient was offered palliative chemotherapy and PleurX drain for symptomatic control. **BJHM**

Figure 1. Plain chest radiograph showing hydropneumothorax and multiple pulmonary nodules. Note artefacts from previous mastectomy visible in the right upper zone.

Figure 2. Chest computed tomography scans (chest drain in situ) showing (a) hydropneumothorax with multiple air–fluid levels and pulmonary nodules and (b) narrowed pulmonary artery and occluded right main bronchus as a result of a large metastasis (arrow).