Letters to the Editor

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Comment on: Clinical utility of anti-signal recognition particle antibody in the differential diagnosis of myopathies

Sirs, I read with great interest the article by Suzuki et al. [1] on clinical utility of anti-signal recognition particle (anti-SRP) antibody in the differential diagnosis of myopathies. Suzuki et al. [1] demonstrated that anti-SRP antibody is positive in 8.3% of the patients with polymyositis but in none of the patients with muscular dystrophy. About 60% of patients with polymyositis due to anti-SRP antibody depicted lymphocytic infiltration on their muscle biopsies [1]. The authors conclude that anti-SRP antibody is useful for discriminating polymyositis from muscular dystrophy [1].

It is known that muscle biopsy from patients with muscular dystrophy typically depicts necrotic fibres with a certain degree of inflammatory reaction; however, a number of subtypes of muscular dystrophy have a pathological mimicry to myositis and are potentially misdiagnosed as inflammatory myopathy. These myositis-mimicking muscular dystrophies include dysferlinopathy, calpainopathy and facioscapulohumeral muscular dystrophy (FSHD) [2–4]. Dysferlinopathy covers a variety of skeletal muscle disorders caused by DYSF gene mutations including limb-girdle muscular dystrophy type 2B (LGMD2B), Miyoshi myopathy and distal myopathy with anterior tibial onset (DMAT). About 70% of patients with dysferlinopathy have significant lymphocytic infiltration on the muscle biopsy samples [2] and 25% of the patients were initially diagnosed with polymyositis [5]. These findings highlight the clinical and pathological resemblances between dysferlinopathy and polymyositis. The recent report of an LGMD2B patient with Addison disease and sarcoidosis and the known susceptibility of SJL/J mice, the animal model of dysferlinopathy, to autoimmune disorders strengthen the link between dysferlin and the inflammatory mechanism [6, 7]. Despite the discovery of the role of dysferlin in membrane-repair machinery, it remains unclear how dysferlin mutations trigger these inflammatory changes [7].

It would be interesting if Suzuki et al. [1] could clarify whether antibody-negative LGMD patients included in their study carry dysferlin mutations. Although Selva-O’Callaghan et al. [6] reported the absence of myositis-specific autoantibodies in a single case of LGMD2B patient with Addison disease and sarcoidosis who initially was diagnosed with polymyositis, it needs a study in a larger scale before drawing any conclusions. Further study of myositis-specific autoantibodies in these particular myositis-mimicking muscular dystrophies would not only shed the light on the pathomechanism but also give the clue for the potential therapeutic strategies. Moreover, the screening for dysferlinopathy by either immunohistochemical study or western blot analysis in the steroid-unresponsive or autoantibody-negative polymyositis patients may identify a surprising number of patients with muscular dystrophies.

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Comment on: Clinical utility of anti-signal recognition particle antibody in the differential diagnosis of myopathies: reply

Sirs, We appreciate Dr Liewluck’s interest [1] in our work [2] and thank him for raising an important issue in the diagnosis of PM. As suggested, some types of myopathies including dysferlinopathy, calpainopathy and facioscapulohumeral muscular dystrophy (FSHD) show inflammatory findings on muscle biopsy. It is sometimes difficult to discriminate PM from these ‘myositis-mimicking muscular dystrophies’ based on clinical manifestations and routine staining methods for muscle biopsy specimens. In fact, patients with limb-girdle muscular dystrophy (LGMD) type 2B, the representative phenotype of dysferlinopathies, were misdiagnosed as having PM before dysferlin protein studies because of histological findings and limb weakness accompanied by rapid progression and/or pain [3]. Gallardo et al. [4] emphasized that a diagnosis of dysferlin myopathy should be considered in young patients with sporadic proximal weakness, high creatine kinase levels, necrotic fibres and inflammation on muscle biopsy. On the other hand, Dimitri et al. [5] reported a 31-yr-old man with anti-signal recognition particle (SRP) antibody-positive PM resembling chronic progression of LGMD.

In order to make a correct diagnosis in these clinical conditions, additional immunohistochemical examinations of sarcocemmal proteins including dystrophin and dysferlin and also inflammatory markers including MHC class I, CD3, CD4, CD8 and complement C5b-9 are useful [5]. Genetic analyses for each form of muscular dystrophy are also required. Unfortunately, these comprehensive diagnostic procedures with specialized techniques can be performed only in a few selected institutions and are very far from being routine examinations. Since clinical concepts have been expanding, physicians should realize that it is not easy to exclude various types of myopathies for the final diagnosis of PM. In this regard, detection of myositis-specific