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Muscle Disorders: A Profile of Patients Undergoing Biopsy in a Tertiary Care Teaching Hospital in South India

Saumya H Mittal*, Shreejit Chakrobarty, Shivanand Pai, Rakshith KC, Misri ZK, Salony Mittal and Nisha Shenoy

Department of Neurology, KMC Hospital, Mangalore

***Corresponding Author**: Saumya H Mittal, Department of Neurology, KMC Hospital, Mangalore.

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Abstract

Objective: To correlate the muscle biopsy reports with the clinical profile of the patients.

Materials and Methods: Demographic data, presenting symptoms, neurological features, and investigations in 33 patients who underwent muscle biopsy over a period of 3 years in our tertiary care centre were evaluated.

Results: Over a period of three years, 33 patients underwent muscle biopsy in our tertiary care hospital. Of these there were 16 females and 17 males. The mean age of the patients was found to be 36.81 years. Most common symptom (28 of the 33) was proximal weakness followed by myalgia (15 of 33 patients). The CPK levels were raised in 27 of the 33 patients and 6 patients had normal levels. The EMG done in all the patients showed myopathic pattern. Of the biopsy reports, 6 were found to have inflammatory myopathy (5 of whom were found to have polymyositis) while 15 were found to have muscle dystrophies. 3 patients had a myopathic biopsy and only 1 showed neurogenic changes. The remaining 4 were normal.

Conclusion: Muscle biopsy is considered the gold standard in diagnosis of inflammatory myopathies and muscular dystrophies.

Keywords: *Muscle Biopsy; Inflammatory myopathy; Muscle Dystrophies*

Introduction

Muscle dystrophies can occur at any age. There is a profound loss of muscle function that affects the posture and cardiorespiratory functions as well. The symptoms may be less prominent in adult onset dystrophies [1,2].

Inflammatory myopathies include polymyositis, dermatomyositis and inclusion body myositis. Patients with polymyositis develop and present with symmetrical proximal muscle weakness with or without rise of creatine kinase (CPK). Electromyography reveals polyphasic motor units, positive sharp waves and fibrillations. There is a characteristic early recruitment pattern [3].

Muscle biopsy is considered the gold standard in diagnosis of inflammatory myopathies and muscular dystrophies. Early detection and confirmation may help in initiating early treatment in the patients [4].

Material and Methods

The data was gleaned off the files of the patients who underwent muscle biopsy over the last 3 years in our tertiary care centre in South India for analysis, from January 2011 to December 2013 for our retrospective and prospective study. The clinical data included age, gender, presenting symptoms and neurological examination. Investigations included creatine phosphokinase, electromyoneurographic findings and muscle biopsy reports.

The muscle biopsy was done from the moderately affected muscles after a written consent and fixed in formalin. Masson's trichrome staining was done for all cases.

None of the patients had molecular genetic studies. Electron microscopy was not available at out centre and therefore not performed.

The retrospective study was performed to correlate between the clinical features, CPK levels, electrophysiological details and the biopsy data. The data was analyzed using the standard means of statistics.

Results

Over a period of three years, 33 patients underwent muscle biopsy in our tertiary care hospital. 7 biopsies were performed in the 1st year of the study. 13 studies were performed in the subsequent 2 years each. Of these there were 16 females and 17 males. The mean age of the patients was found to be 36.81 years (range from 7 years to 66 years of age). The disease had a progressive course in all the patients. The mean delay between the onset of symptoms and presentation to the hospital was 4.9 years.

Almost all patients (28 of the 33) presented with complaints of proximal weakness. The other 5 presented with complaints of myalgia. A total of 15 patients had myalgia. Arthralgia was a complaint in 4 of the patients while weight loss was a complaint in 1 patient. None of them had fever, respiratory involvement.

The patients underwent biopsy at different sites. In our study, the most common site of biopsy was the vastus lateralis. 18 of the 33 patients underwent biopsy from the vastus lateralis, only 1 of them being from the right sided and the remaining from the left side. 8 patients underwent biopsy from the left biceps brachii, 5 from the deltoid (4 from the left side and 1 from the right side). The remaining 2 were performed from the right gastrocnemius.

The CPK levels were done in all the patients. 6 of them had normal levels of CPK. The remaining 27 were high. Of these 27, 16 patients had CPK levels of more than 1000 U/L, and the remaining 11 had CPK levels less than 1000 U/L.

The EMG done in all the patients showed myopathic pattern.

Of the biopsy reports, 6 were found to have inflammatory myopathy (5 of whom were found to have polymyositis) while 15 were found to have muscle dystrophies. 3 patients had a myopathic biopsy and only 1 showed neurogenic changes. The remaining 4 were normal (these patients were found to have hypothyroidism in 3 cases and hyperparathyroidism in 1 case).

Based on the clinical features and examination findings, the patients of muscular dystrophies could be identified as 5 patients with Becker muscular dystrophy. Another 5 were found to have limb girdle dystrophy. Amongst the remaining 5, 2 patients had facioscapulohumeral dystrophy and 3 patients had myotonic dystrophy.

22 patients who underwent biopsy showed a preserved fascicular structure. Other 11 showed altered fascicular structure. Masson's trichrome stain showed normal endomysial collagen pattern in 15 patients. It was found to be mildly increase in 9 patients, moderately increased in 4 and markedly increased in 5 patients. Myofiber size was preserved in 7 patients. It was found to have mild variation in 12, moderate variation in 3 and marked variation in 11 patients. 12 patients showed lymphocytic infiltration and a similar number of patients were found to have myophagocytosis.

Discussion

Over time, the diagnosis and diagnostic tests available have seen changes. Starting from routine histology, 1960s came up with enzyme histochemistry and 1990s brought up immunohistochemistry. Use of MHCs antigen immunostaining has been used now recently [4].

Inflammatory myopathies were considered common on India. They are potentially reversible and therefore need to be diagnosed. However, they may not be that frequent in India. Muscular dystrophies are more common. Duchenne muscular dystrophy in childhood and limb girdle muscle dystrophy in adults are known to be the commonest muscular dystrophies [5]. In our study too, most of the patients (15 of 33) were found to have muscular dystrophy. This was higher than that found in a larger study by Das where 27.4% of all myopathies were muscular dystrophies. Whereas in comparison only 6 of our 33 patients had inflammatory myopathies [6].

Polymyositis biopsy shows inflammatory cells (that include CD8+ T cells and macrophages) which can destroy the normal appearing myofibers. In contrast, dermatomyositis shows atrophic, degenerating and regenerating myofibers in a perifascicular distribution. This results due to the destruction of the capillaries of the area causing hypoxia and subsequent myofibril damage. Neovascularisation may be seen [3]. In our study 5 patients showed the changes suggestive of polymyositis and 1 patient had dermatomyositis.

Muscular dystrophies are characterized by 'dystrophic' changes that specifically include variation in size of the fibre, increased internal nuclei and presence of regeneration and degeneration of fibres. Inflammatory cellular infiltrate may be seen along with fibre splitting. These features occur due to recurrent muscle fibre necrosis and regeneration [1,2]. 15 of our patients showed such changes.

Conclusion

It is clear that wide varieties of myopathies exist. And few neurology consultants that India can boast of will usually have their hands full. Few centres have laboratories equipped to evaluate myopathies [5]. Most of the time, the patients of India are unable to afford extensive investigations which can run up the costs. Muscle biopsy is considered the gold standard in diagnosis of inflammatory myopathies and muscular dystrophies [3].

Proximal weakness remains the commonest symptom with which a patient would present to the hospital. Muscular dystrophy was found to be more common than inflammatory myopathies. Amongst inflammatory myopathies, polymyositis was found to be the commonest.

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