



Type 2 Muscle Fiber Predominance: A Case Report

By Kirill Alekseyev, Shruti Amin, Malcolm Lakdawala, Nada Farooqui & Marc K. Ross

Introduction- Skeletal muscles are a heterogeneous group of tissues categorized into different fiber types.¹ These fibers are divided into slow-twitch type 1 and fast-twitch type 2 fibers, which are identified by their expression of specific myosin heavy chain isoforms.^{5,7,9} The predominance of type 2 muscle fibers has been observed in various diseases involving skeletal muscle such as Duchenne muscular dystrophy, hyperthyroidism, and type 2 muscle fiber predominance.^{2,4} Type 2 muscle fiber predominance is a rare muscular disease whose primary manifestations include proximal muscle weakness, exertional myalgia, fasciculations, and episodes of prolonged painful muscle cramping.^{3,4} These symptoms eventually led our patient to be wheel-chair bound with subsequent disuse muscle atrophy. In this case we describe a patient with type 2 muscle fiber predominance, the progression of his disease, and his experience in our inpatient rehabilitation facility.

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Type 2 Muscle Fiber Predominance: A Case Report

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I. INTRODUCTION

Skeletal muscles are a heterogeneous group of tissues categorized into different fiber types.¹ These fibers are divided into slow-twitch type 1 and fast-twitch type 2 fibers, which are identified by their expression of specific myosin heavy chain isoforms.^{5,7,9} The predominance of type 2 muscle fibers has been observed in various diseases involving skeletal muscle such as Duchenne muscular dystrophy, hyperthyroidism, and type 2 muscle fiber predominance.^{2,4} Type 2 muscle fiber predominance is a rare muscular disease whose primary manifestations include proximal muscle weakness, exertional myalgia, fasciculations, and episodes of prolonged painful muscle cramping.^{3,4} These symptoms eventually led our patient to be wheelchair bound with subsequent disuse muscle atrophy. In this case we describe a patient with type 2 muscle fiber predominance, the progression of his disease, and his experience in our inpatient rehabilitation facility.

II. CASE

A 68-year old Caucasian male with a past medical history of congenital type 2 muscle fiber predominance formally diagnosed in 1980 via multiple muscle biopsies presented to the inpatient rehabilitation facility (IRF). The patient was evaluated and treated for proximal muscle weakness in his thighs and calves, as well as less pronounced weakness in the upper extremities, exertional myalgia, and chronic dysphagia. The patient also reported episodes of severe spastic muscle cramping that involved all extremities, sparing his face, with exacerbations occurring 2 to 12 times per year for the past 48 years.

The disease first affected the patient with exertional myalgia at age five, which led to frequent falls. The patient has since then avoided running and exercise. The patient's muscular weakness progressively worsened, and he eventually became wheelchair bound at age 38. At age 30, he was admitted to the hospital for substernal chest pain and diaphoresis that the patient states "felt like having a heart attack". Work-up was negative for a myocardial infarction, and multiple subsequent episodes of substernal chest pain have also have also been negative for myocardial infarct.

Prior to his admission, the patient was unable to transfer from his bed to wheelchair. At the time of admission, his musculoskeletal exam demonstrated bilateral calf fasciculations at rest. Sensation and proprioception were intact in all four limbs. Muscle strength was 4/5 in shoulder girdle secondary to pain. In his lower extremities he had 1/5 strength proximally and 4+/5 strength distally. The patient was noted to hold a cup in either hand for approximately 10-15 seconds without dropping it. The patient's deep tendon reflexes were diminished to 1/4 in biceps brachii, triceps, and brachioradialis reflexes, and 0/4 in patellar and Achilles reflexes bilaterally.

After rehabilitation, the patient was able to perform bed to chair transfers independently with a transfer score of 6. The patient was also able to perform bicep curls and shoulder press-ups with four pounds of weight for 10 repetitions and the use of a thera-band. With balance, endurance, and neuromuscular facilitation training the patient advanced from 0 feet of ambulation at the beginning of his stay to 16 feet with parallel bar assistance. During the hospital course, the patient's diet was advanced from full liquids to solid food. Moist heat packs and bio freeze gel were applied to his shoulder and neck area to decrease pain and stiffness.

Nerve conduction studies were conducted as shown in Table 1. The patient had no response in the right peroneal motor nerve at the ankle with reduced amplitude (B Fib 1.7 mV and Popliteal 1.6 mV) and decreased nerve velocity (Pop-fib 36 m/s). The left tibial motor showed reduced amplitude (Ankle 3.5 mV) and reduced amplitude (Popliteal 1.7 mV). The right tibial motor nerve showed reduced amplitude (Popliteal, 0.8 mV). The left and right sural sensory nerves showed no response. F-wave studies indicated that the right tibia response had prolonged latency (59.11 ms).

On repeat nerve conduction studies, evaluation of the left tibial motor nerve showed reduced amplitude (Ankle 3.5 mV) and reduced amplitude (Popliteal 1.7 mV). On electromyography studies, the patient was found to have delayed recruitment and the poor activation in right and left vastus medialis, right medial gastrocnemius, and left anterior tibialis. Increased duration was seen in bilateral vastus medialis and left anterior tibialis muscles. The overall objective impression of the study showed primarily axonal and demyelinating sensorimotor peripheral polyneuropathy affecting the bilateral lower extremities.

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III. DISCUSSION

Type 2 muscle fiber predominance is a rare congenital muscular disorder that primarily affects the strength of the proximal muscles. The main manifestations as seen in this patient include proximal muscle weakness, exertional myalgia, fasciculations, episodes of severe painful muscle cramping requiring muscle relaxants, and dysphagia. Normally, slow-twitch type 1 fibers are the first to contract during aerobic exercises such as running.⁶ Patients with this disease have shown to have a decrease in type 1/type 2 fiber ratio in proximal muscles³; thus, have difficulty with aerobic exercises. Most cases reported of this disease include patients ranging in age from the teenage years to our patient's age of 68.

In 1985, the first documented report of "type 2 fiber predominance" was studied.³ In this study, all 13 male patients demonstrated muscle cramping and exertional myalgia, but no abnormalities were found on physical neurologic or laboratory examinations.³ However, pathologic abnormalities of muscle biopsies of proximal muscles, such as the vastus medialis identified, a decrease in type 1/type 2 fiber ratio with 72.91% of type 2 fibers increased versus 59.9% in the control group.³

Similar findings were reported in a study in 2002, in which three cases of otherwise healthy men aged 18, 19 and 22 were identified with this disease despite being otherwise healthy.⁴ In all three cases, muscles of facial expression were spared and pseudohypertrophy or true hypertrophy of the calf muscle was absent. Concluded from these studies concluded that type 2 muscle fiber predominance is a diagnosis of exclusion. It is important to note that laboratory findings were normal in both studies. Normal findings in all patients of the Kim et al. study included serum creatinine kinase, aldolase, lactate dehydrogenase, electrolytes, and thyroid function tests. Findings of increased lactate levels during ischemic exercise rules out glycogen storage diseases. Thyroid function tests are important, as patients with hyperthyroidism have been shown to have an increased expression of type 2 muscle fibers.⁴

Pathologic studies in all three cases reported no significant abnormality observed on physical findings and normal expression of dystrophin along the sarcolemmal membrane on immunohistochemical studies. To note, all three cases demonstrated an increased percentage of type 2 fibers on enzyme histochemistry. To elaborate, in case 1, a 73% increase in type 2 fiber in the vastus medialis, 80.2% in vastus lateralis in case 2, and 75% in vastus lateralis in case 3. Furthermore, a marked decrease in type 1 fibers was shown in myofibrillar ATPase preincubated at pH 9.4.⁴

Hayat et al., reports one case of a 28-year-old male with proximal muscle weakness and motor neuron

dysfunction on electrophysiological studies. This patient had a 97% type 2 fiber predominance on muscle biopsy of vastus lateralis with type 2A being the largest compared to type 2B and 2C. The pathogenesis of type 2 fiber predominance remains unknown however, this study proposes a functional abnormality of motor neuron in utero leading to abnormal muscle fiber differentiation and growth.¹⁰ Another theory suggested in the literature is that the diminished type 1 fibers are directly affected from a depletion of an oxidative enzyme along with fiber switching of type 1 to type 2 fibers.⁴

Limited studies exist on the effect of rehabilitation and electrical stimulation on the transformation of type 2 to type 1 muscle fibers. One study states that long duration and high volume endurance type events promote the conversion of type 2 to type 1 fibers.⁸ High volume resistance training may also result in similar fiber changes as high volume endurance training.⁹ Another study suggests that at least two months of slow pattern stimulation is needed for transformation of original type 2B or 2X fibers into type 1 fibers.¹¹ Further investigation is needed to determine if endurance events or stimulation conversion of type 2 to type 1 fibers will therapeutically benefit patients with type 2 muscle fiber predominance.

IV. CONCLUSION

Patients affected by type 2 muscle fiber predominance exhibit proximal muscle weakness, exertional myalgia, and an additional finding unique to this case, chronic progressive dysphagia. As a result, patients with type 2 muscle fiber predominance have difficulty running by approximately age 5 and are often wheelchair bound in their late 20s to 30s. In this disease lower extremities seem to be affected more than upper extremities. However, our patient showed more improvement in the lower extremities than the upper extremities in his overall outcome. He improved in activities of daily living from moderate assistance to modified independent with wheelchair. The integration of physical and occupational therapy as well as the use of muscle relaxants are essential for patients with this disease. Few cases have been reported on type 2 muscle fiber predominance, but further studies are vital in order to improve the quality of life of current and future patients.

Table 1: Electromyography Study Results

Site	Onset (ms)	Norm Onset (ms)	O-P Amp (mV)	Norm O-P Amp	NegDur (ms)	Site 1	Site 2	Delta-0 (ms)	Dist (cm)	Vel (m/s)	Norm Vel (m/s)
Left Tibial Motor (Abd Hall Brev) Ankle Poplit	4.5 16.3	<6.0	3.5 1.7	>4.0 >4.0	4.69 5.0	Poplit	Ankle	11.8	48.0	41	>40
Right Peroneal Motor (Ext Dig Brev) B Fib Poplit	14.5 17.3	- -	1.7 1.6	>5.0 >5.0	6.09 5.78	Poplit	B Fib	2.8	10.0	36	>40
Right Tibial Motor (Abd Hall Brev) Ankle Poplit	5.5 16.4	<6.0	4.3 0.8	>4.0 >4.0	5.78 4.38	Poplit	Ankle	10.9			>40

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