

Magnetic Resonance Imaging Findings of the Skeletal Muscle of a Patient with Nemaline Myopathy

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This is the first magnetic resonance imaging (MRI) report of nemaline myopathy in which muscle atrophy was not apparent clinically in the lower extremities because of subcutaneous fat. The patient is a 38-year-old woman who was admitted to our hospital because of muscle weakness of the four extremities. Until the age of 17 years, she was asymptomatic except that her running speed was slow. The T₁-weighted image of muscle MRI at the mid-thigh level showed hyperintensity of the quadriceps femoris muscle and relatively spared hamstring muscle. The T₂-weighted image of muscle MRI at the maximum diameter of the lower leg showed hyperintensity of the tibialis anterior muscle and a relatively spared triceps surae muscle. The biopsy specimen of the right deltoid muscle showed nemaline bodies and type II fiber deficiency. (Internal Medicine 37: 776–779, 1998)

Key words: type II fiber deficiency, extensor muscle group, magnetic resonance imaging (MRI)

Introduction

Nemaline myopathy is a neuromuscular disorder characterized by muscle weakness and the presence of nemaline bodies (rods) in the muscle fibers, in the absence of other known conditions sometimes associated with rods (1), such as polymyositis, central core disease, chronic alcoholic myopathy, human immunodeficiency virus infection, congenital muscular dystrophy, multiple acylCoA dehydrogenase deficiency (2) and adult-onset rod disease (3). Reports of muscle magnetic resonance imaging (MRI) in nemaline myopathy are rare (4). This is the first MRI report of nemaline myopathy in which the muscle atrophy was not apparent clinically in the lower extremities due to subcutaneous fat. We report a case of nemaline myopathy in which muscle MRI was useful in evaluating the distribution and severity of myopathy.

Case Report

The patient is a 38-year-old woman who was admitted to our hospital because of muscle weakness of the four extremities. Until the age of 17 years, she was asymptomatic except that her running speed was slow. At the age of 18, she noticed mild weakness in her lower extremities. She has not been able to climb stairs since the age of 33. She noticed difficulty in walking at the age of 36. In addition to the weakness in the lower

extremities, she began to have weakness in the upper extremities at the age of 38. Her past history was unremarkable. Her family history showed that her healthy parents were cousins and that she had a sister with nemaline myopathy and a healthy brother. The sister was a 42-year-old woman with a clinical picture similar to the present patient and the diagnosis was based on muscle biopsy findings but muscle MRI of the sister had not been taken.

On admission, she was 150 cm tall with a body weight of 40.2 kg, blood pressure 126/82 mmHg, pulse rate 108 and regular. Physical examination of the chest and abdomen was normal. Oval face, pes cavus and pes equinovarus were noted but dysmorphic face was absent. Her consciousness was clear, speech normal, and mini-mental state examination score was 30 points; mild facial weakness and moderate atrophy and weakness of the sternocleidomastoid muscle were present. The muscle strength (right/left, manual muscle test) was as follows: neck flexor 2, neck extensor 3, pectoralis major 3/3, serratus anterior 3⁺/3⁺, deltoid 3/3⁺, biceps brachii 4⁺/4, triceps brachii 4⁺/4, wrist extensor 4⁺/4⁺, wrist flexor 4⁺/4⁺, opponens pollicis 4⁻/4⁻, iliopsoas 3⁻/3⁻, quadriceps femoris 2⁺/2⁺, hamstrings 2⁺/2, great adductor 2/2, biceps femoris 2⁺/2, tibialis anterior 2/2, triceps surae 4/4, tibialis posterior 4/4, flexor digitorum longus 4/4, extensor hallucis longus 2/2⁻, extensor digitorum longus 2/2⁻, peroneus longus and brevis 2/2. Muscle atrophy was moderate in the upper extremities but was not apparent in the lower

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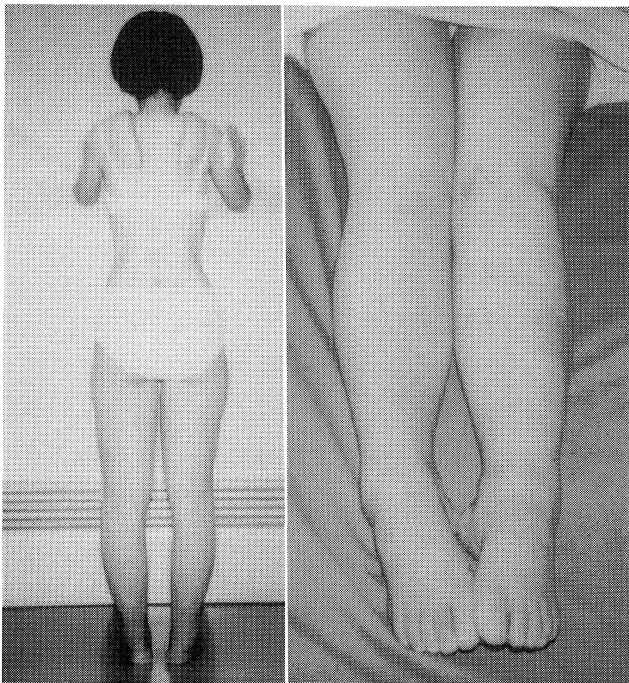


Figure 1. The back and the lower extremities of the patient. Muscle atrophy is moderate in the upper extremities but is not apparent in the lower extremities due to subcutaneous fat.

extremities due to subcutaneous fat (Fig. 1). The muscle tone and deep tendon reflexes were decreased in the four extremities. Pathological reflexes were absent. The gait was waddling and steppage. Coordination was normal in the upper extremities and could not be examined in the lower extremities because of muscle weakness. The sensory system was normal.

Laboratory examinations were as follows: Complete blood count, urinalysis and cerebrospinal fluid examination were normal; creatine kinase 13 U//, aldolase 2.1 IU//, blood chemistry was normal. Antinuclear antibody was negative, anti-DNA antibody negative, thyroid function normal, and pituitary hormone normal. Chest X-ray was normal and electrocardiogram showed a negative T wave at V₁ lead. The motor nerve conduction velocity was 61.7 m/sec at the right median nerve and 43.9 m/sec at the right tibial nerve. The sensory nerve conduction velocity was 57.4 m/sec at the right median nerve and 40.9 m/sec at the right sural nerve. The needle electromyogram was normal except for decreased interference pattern in the right flexor carpi radialis muscle, tibialis anterior muscle and triceps surae muscle.

The T₁-weighted MRI of muscle (Fig. 2) at the mid-thigh level showed hyperintensity of the quadriceps femoris muscle and relatively spared hamstrings, great adductor and biceps femoris muscles. The T₂-weighted MRI of muscle at the maximum diameter of the lower leg showed hyperintensity of the tibialis anterior, extensor hallucis longus, extensor digitorum longus, and peroneus longus and brevis muscles and relatively spared soleus, lateral head of the gastrocnemius, tibialis poste-

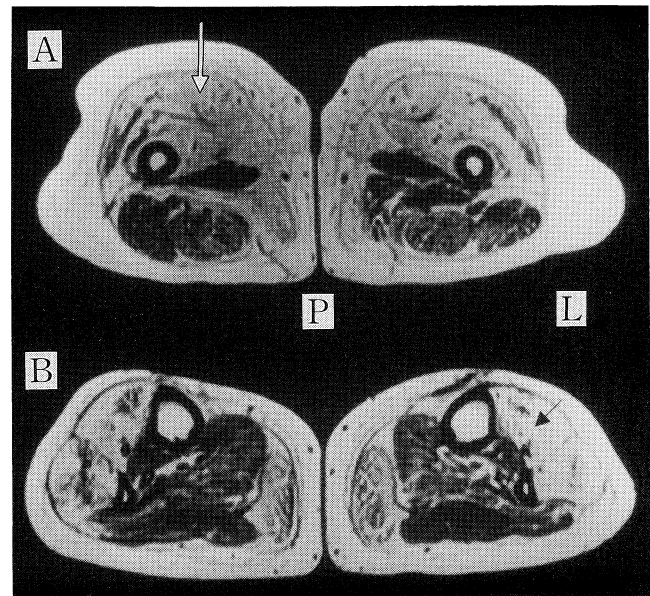


Figure 2. The T₁-weighted MRI of muscle (TR 600, TE 25) at the mid-thigh level (A) showed hyperintensity of the quadriceps femoris muscle (white arrow) and relatively spared hamstring muscle. The T₂-weighted MRI of muscle (TR 5945, TE 120) at the maximum diameter of the lower leg (B) showed hyperintensity of the tibialis anterior muscle (black arrow) and the relatively spared triceps surae muscle.

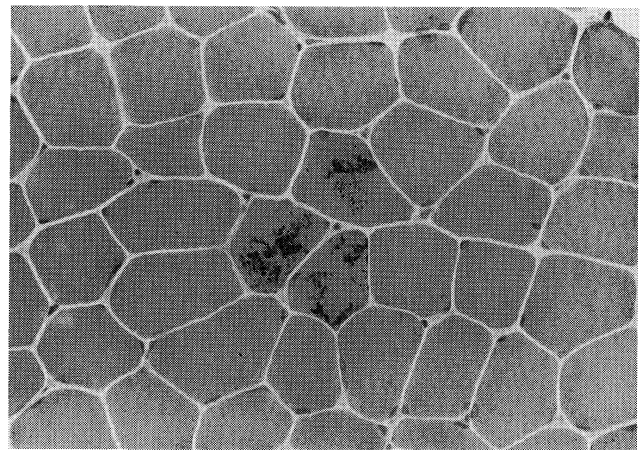


Figure 3. Biopsy specimen of the right deltoid muscle showed nemaline bodies (Gomori trichrome stain, ×80).

rior and flexor digitorum longus muscles. The medial head of the gastrocnemius muscle showed mild hyperintensity.

The biopsy specimen of the right deltoid muscle showed nemaline bodies (Figs. 3, 4) and type II fiber deficiency. Nemaline bodies were found in some of the muscle fibers.

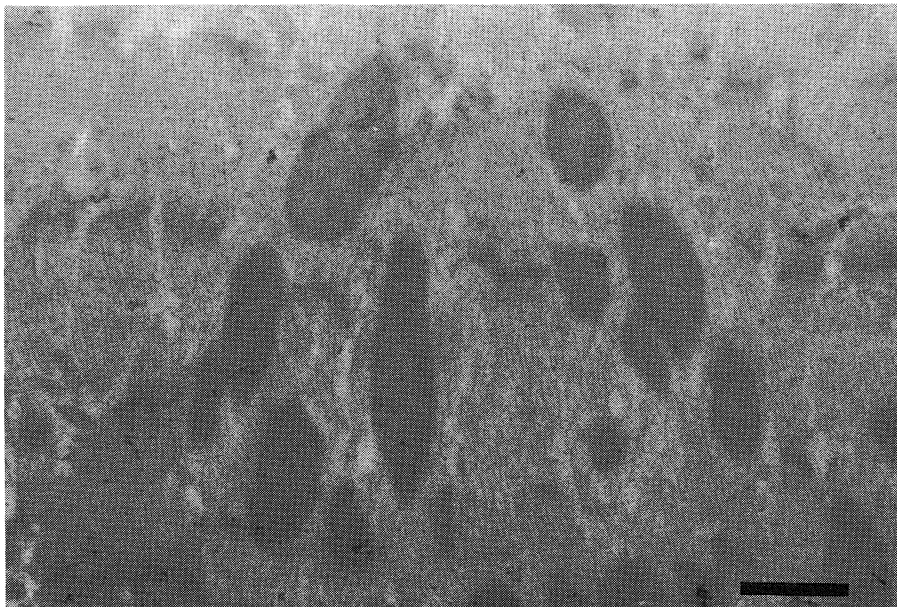


Figure 4. Electron microscopy showed nemaline bodies with the same electron density as Z-band (bar: 1,000 nm).

Discussion

We made the diagnosis of nemaline myopathy (1, 5–11) because of muscle weakness, the presence of nemaline bodies in the muscle fibers, muscle hypotonia, myopathic face and the slowly progressive course. The inheritance was considered to be autosomal recessive but gene analysis was not performed.

The present case is characterized by type II fiber deficiency. Nemaline myopathy tends to show type I fiber predominance (12–15) and the extreme case (16) of type I fiber predominance is considered to be similar to the present case. In the thigh of the present case, the quadriceps femoris, hamstrings, great adductor and biceps femoris muscles were weak, on MRI however, the quadriceps femoris was abnormal and the hamstrings, great adductor and biceps femoris muscles were relatively spared. In the lower leg of the present case, the tibialis anterior, extensor hallucis longus, extensor digitorum longus and peroneus longus and brevis muscles were weak and abnormal on MRI, but the triceps surae, tibialis posterior and flexor digitorum longus muscles were relatively strong and relatively spared on MRI. Although the muscle strength did not always correlate with the abnormality on MRI in the thigh, the muscle strength correlated with abnormality on MRI in the lower leg. The tibialis anterior muscle is a white muscle (type II fiber more than type I fiber) and the triceps surae muscle is a red muscle (type I fiber more than type II fiber). The influence of type II deficiency is considered to be greater in the tibialis anterior muscle than in the triceps surae muscle.

Muscle atrophy was moderate in the upper extremities where muscle weakness was relatively mild and muscle atrophy was not apparent in the lower extremities where muscle

weakness was relatively severe. The muscle MRI showed hyperintensity of the quadriceps femoris muscle and tibialis anterior muscle which suggests replacement of muscle fiber with fat (4, 17, 18). Distal muscular hypertrophy has been reported in a case of adult-onset nemaline myopathy and was considered to be compensative hypertrophy (19). Muscle MRI was useful in evaluating the muscle abnormality in the lower extremities in the present case.

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