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A Case of Rimmed Vacuole Myopathy with Limb-girdle Atrophy and Boule Musculaire

Yoshitaka Takase, Miki Yamagata*, Kiyoshi Yamamoto*, Toshihiro Fukusako* Mitsunori Morimatsu* and Syuhei Kawazawa***

*Department of Neurology, Yamaguchi University School of Medicine, Ube, Yamaguchi 755, Japan.
**Division of Neurology, Shimonoseki Saiseikai Hospital, Shimonoseki, Yamaguchi 751, Japan.
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Abstract A case of 58-year-old man with rimmed vacoule myopathy was reported, who had limb-girdle distribution of muscle weakness and atrophy, and had a boule musculaire in the left quadriceps muscle. Muscle biopsy was done in the quadriceps muscle neighbouring boule musculaire, and the specimen showed rimmed vacuoles in 7.6% of the muscle fibers, which appeared mainly in the border between muscle fibers and degenerative fat. Almost all of the rimmed vacuoles were observed in type 1 fibers, and occupied with many membranous whorls and myelin debris.

Key Words: Rimmed vacuole, Myopathy, Proximal atrophy, Boule musculaire

Introduction

It is well known that rimmed vacuoles are well observed in a distal myopathy inherited as an autosomal recessive trait, and may also be seen in miscellaneous neuromuscular disorders such as oculopharyngeal dystrophy[1]. The present case had a muscle weakness and atrophy of limb-girdle distribution and had a boule musculaire, which did not conform to clinical criteria of rimmed vacuole myopathy hitherto reported. We described and discussed the clinical features of the case, especially about boule musculaire.

Case Report

A 58-year-old man was admitted to the Yamaguchi University Hospital to evaluate weakness of the extremities. At the age of 27 he noticed slight muscle weakness of the extremities. The weakness progressed very slowly, and at the age 53 he was not able to stand up from a squatting posture, followed by a great difficulty in walking and getting out of bed. At admission, he was 159 cm in height and 41.5 kg in body weight. On physical examination, the blood pressure was normal, and the liver and spleen were not palpable. Neurological examinations showed the patient to be alert with moderate to severe muscular atrophy and generalized weakness, particularly in the neck flexors and extensors, in the proximal muscles of both upper and lower limbs, and in the truncal muscles. The boule musculaire was seen in the lower part of the left thigh (Fig. 1). Neither abnormality in the cranial nerves nor any sensory disturbance was found. The muscle stretch reflexes revealed decreased responses. Computed tomography (CT) of the muscles showed that the neck extensors, paravertebral muscles and right quadriceps were severely replaced by fat. However, the part of the boule musculaire of the left quadriceps was not so affected as the right quadiceps (Fig. 2). Electromyographic (EMG) examination

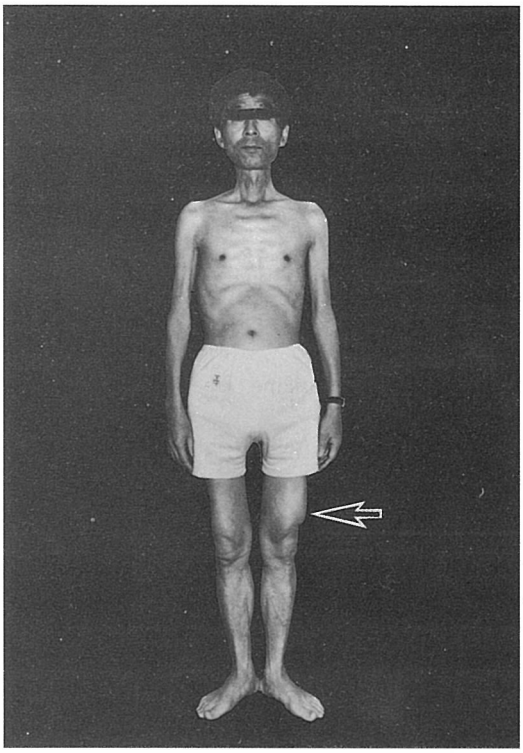

Fig. 1 Photograph of the patient showing muscle wasting of limb-girdle distribution, with a boule musculaire in the left thigh (arrow).

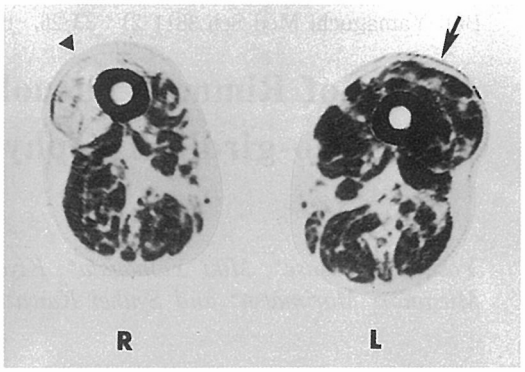

Fig. 2 Muscle CT showing severly affected thigh muscles. Note the boule musculaire (arrow), which is not so severely replaced by fat as the right quadriceps muscle.

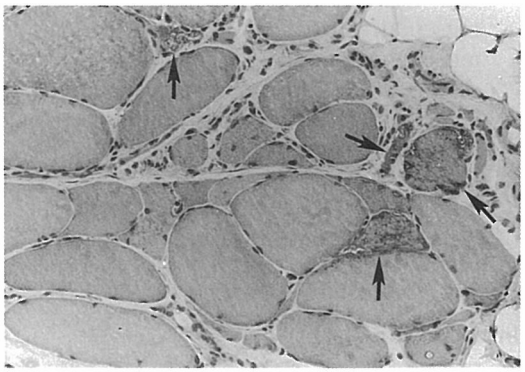

Fig. 3 Muscle biopsy specimen stained with hematoxylin eosin showing an increase of atrophic fiber and connective tussue. Note the fibers with rimmed vacuole (arrow) well observed at the border of fatty replacement. (×230)

of the right deltoid, biceps brachii and bilateral quadriceps muscles showed mixed patterns of neurogenic and myogenic origin. Fibrillation, positive sharp waves, and fasciculation were not observed. High amplitude potentials of short duration, some of which were polyphasic, were observed. There were no definitively different findings in the EMG between the boule musculaire and neighbouring atrophy. Biopsy specimens revealed a slight increase in atrophic fibers, internally nucleated fibers, and necrotic fibers. Neither group atrophy nor pyknotic nuclear clumps was seen, also without extensive fatty replacement (Fig. 3). Rimmed vacuoles were seen in 7.6% of the muscle fibers and they appeared mainly in the border between muscle fibers and degenerative fat (Fig. 3). Serial sections from boule musculaire to neighbouring atrophy showed an increase in the ratio of fatty replacement. However, no additional examination of the individual muscle fibers was done morphologically or histochemically. The electron microscope showed many membranous whorls and myelin debris in a vacuole, and showed no inclusion bodies (Fig. 4). Biochemical laboratory examinations demonstrated only a slight increase in serum creatine kinase. Peripheral blood, serum and urine electrolytes, and thyroid function were normal.

Discussion

Dubowitz and Brooke first stated that rimmed vacuoles were noted in 86% of cases

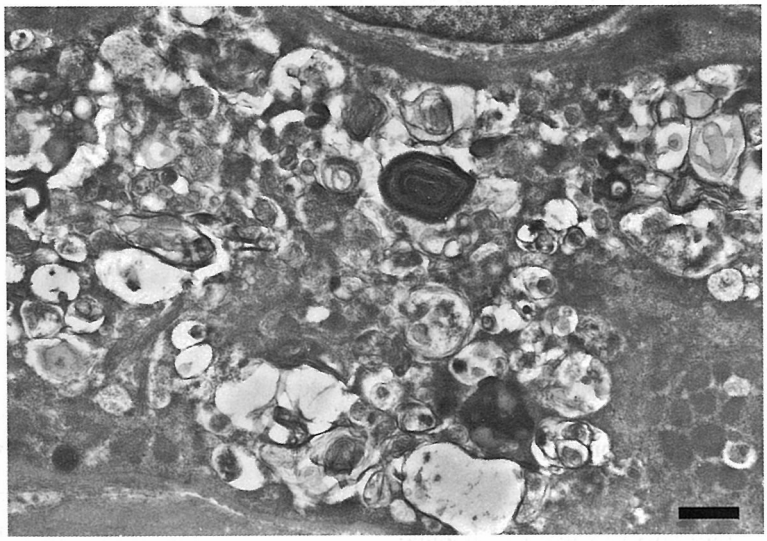

Fig. 4 Electron micrograph showing many membranous whorls in a vacuole. bar=1μm

with oculopharyngeal dystrophy and that therefore such findings might be favorable to this diagnosis[2], although they did not study these vacuoles systematically. Fukuhara et al.[1] studied rimmed vacuoles histochemically in 12 cases, and they showed that rimmed vacuoles were observed in both neurogenic and atrophic muscular diseases other than oculopharyngeal dystrophy. However, rimmed vacuoles were never described in limb-girdle myopathies except for dystrophy or glycogen storage disease[3]. This patient had rimmed vacuole myopathy showing proximal dominant atrophy. This distribution of affected muscle is rarely seen in rimmed vacuole myopathy, and of course boule musculaire in this myopathy has not been reported previously.

Hirayama and Fukuda[4] in referring to " boule musculaire" said that there was no report describing it in detail, and it was named by several scientists as follows: localized pseudohypertrophic areas (Hough)[5], bunching (Sandler)[6], boule musculaire (Lapresle)[7], Wulst (Görres[8], Stähli[9]), Wultung (Görres)[8], Wölbung (Görres[8]). Moreover, Hirayama and Fukuda[4] insisted that boule musculaire was not observed in patients with neurogenic muscular atrophies. As they said, the reported cases were all progressive muscular dystrophies. However, several cases of neurogenic atrophy with this change were reported thereafter: Kugelberg-Welander[10] and amyotrophic lateral sclerosis[11].

Hirayama and Fukuda[4] first studied the nature of boule muscualire and neibouring atrophic muscles from the point of view of EMG, muscle biopsy, and muscle chemistry. They suggested that the differences in clinical findings and laboratory data between the two might depend on the degree of disturbance of the muscles[4]. Muscle chemistry between the two sites was not examined in the present case. However, the findings of few frequencies of electrical silence in the EMG, high opacity in muscle CT, and a small amount of fatty replacement in the muscle biopsy also suggests that boule musculaire may be only an initial stage or slight degree of muscular degneration.

It will be necessary in the future for us to examine differences biochemically between boule musculaire and neighbouring abrophic muscle, which will give us an important clue to clarify a process or mechanism of muscular degneration.

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