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Short communication

A case of adult-onset reducing body myopathy presenting a novel clinical feature, asymmetrical involvement of the sternocleidomastoid and trapezius muscles

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ABSTRACT

We herein report a 32-year-old woman with adult-onset reducing body myopathy (RBM) who had a mutation in the four-and-a-half LIM domain 1 gene (*FHL1*) and showed a marked asymmetrical involvement of sternocleidomastoid and trapezius muscles. At 30 years of age she noticed bilateral foot drop, and over the next two years developed difficulty raising her right arm. At 32 years of age she was admitted to our hospital for a diagnostic evaluation. Neurological examination showed moderate weakness and atrophy of her right sternocleidomastoid muscle, right trapezius muscle, and bilateral upper proximal muscles. There were severe weakness and atrophy of her bilateral tibialis anterior muscles. Her deep tendon reflexes were hypoactive in her upper extremities. Her serum creatine kinase level was mildly increased. Muscle biopsy specimens from the left tibialis anterior muscle revealed marked variation in fiber size, some necrotic or regenerating fibers, and reducing bodies. Gene analysis of *FHL1* demonstrated a mutation: a heterozygous missense mutation of c.377G > A (p. C126T) in *FHL1*. Compared with previous adult-onset RBM cases harboring mutations in *FHL1*, our case was characterized by asymmetrical atrophy of the sternocleidomastoid and trapezius muscles.

1. Introduction

Reducing body myopathy (RBM) is an extremely rare muscle disorder. It is characterized by the presence of intracytoplasmic inclusion bodies that reduce nitroblue tetrazolium in the absence of substrate α-glycerophosphate, in a menadione-linked α-glycerophosphate dehydrogenase (MAG) reaction [1]. RBM was first described as a congenital disorder in 1972 [1], and in 1999, the first adult-onset RBM patient was reported [2]. In 2008, Schessl et al. [3] identified the X-chromosomal four-and-a-half LIM domain 1 gene (*FHL1*) as a causative gene for RBM. *FHL1* encodes the FHL1 protein, composed of four LIM domains preceded by a single N-terminal zinc finger. FHL1 is highly expressed in skeletal and cardiac muscles and considered to regulate muscle mass and strength enhancement [4].

At present, detailed clinical features of adult-onset RBM remain to be delineated owing to its extreme rarity. Here, we report a case of a 32-year-old woman with adult-onset RBM harboring a mutation in the *FHL1* gene, and describe its clinical presentation. Together with a literature survey on adult onset RBM cases, this case study could provide new clinical findings of adult-onset RBM, and might give some insight on a question about what defines the clinical course of RBM.

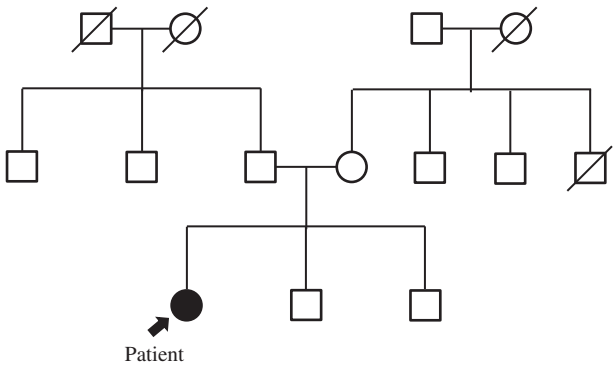

Fig. 1. A pedigree of the present case. An affected individual is indicated in black, and unaffected individuals in white. An arrow indicates the index patient.

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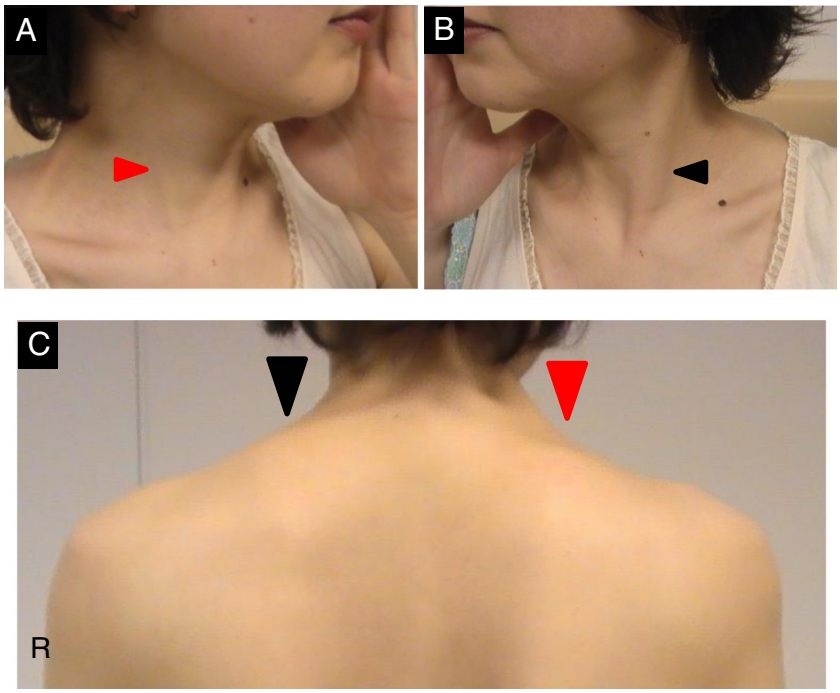

Fig. 2. Clinical picture. (A) The right sternocleidomastoid muscle shows severe atrophy (red arrowhead). (B) The left sternocleidomastoid muscle is intact (black arrowhead). (C) The right trapezius muscle is mildly atrophic (red arrowhead) compared with the contralateral muscle (black arrowhead).

2. Case report

A 32-year-old woman had been in good health and could participate regularly in sports activities until the age of 29. She had no family history of neuromuscular disease (Fig. 1). At 30 years of age, she experienced bilateral foot drop, and over the next two years developed difficulty raising her right arm. At the age of 32 years, she was admitted to our hospital for a diagnostic evaluation. A neurological examination showed moderate

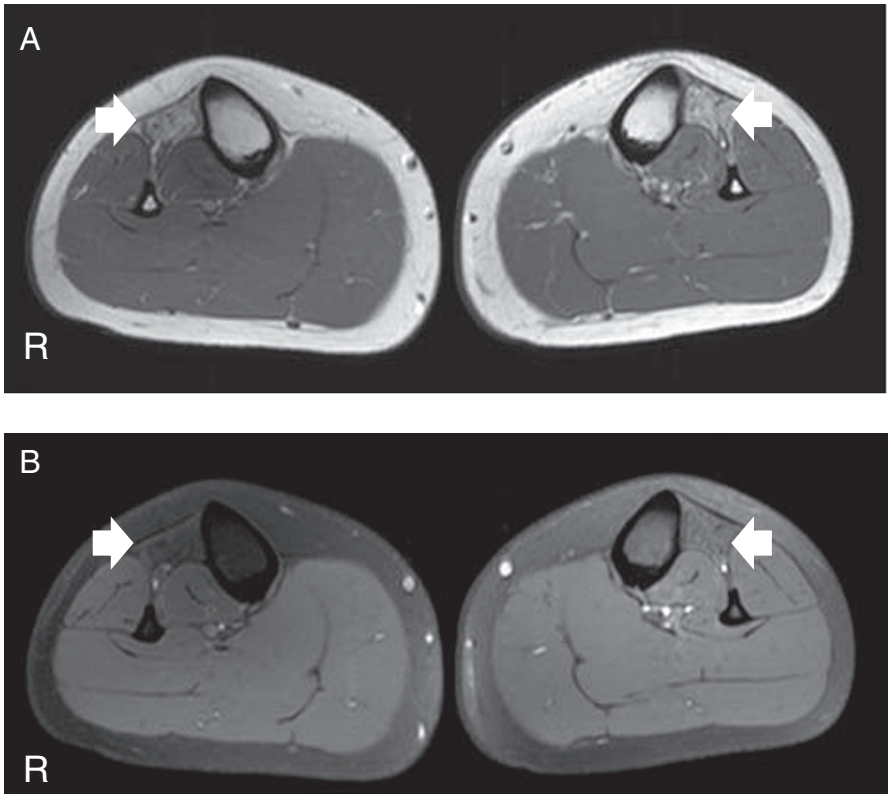

Fig. 3. Magnetic resonance imaging of calf muscles (A and B). (A) T1-weighted axial image at calf level (TR, 730.34 ms; TE, 10 ms) shows high-intensity areas and atrophy in the bilateral tibialis anterior muscles, predominantly on the right (arrows). (B) T1-weighted SPIR axial image at calf level (TR, 627.05 ms; TE, 10 ms) shows no high-intensity areas in the bilateral tibialis anterior muscles (arrows).