Clinical Case Report

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Benign monomelic amyotrophy with lower limb involvement in an adult

A case report

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Abstract

Rationale: Monomelic amyotrophy (MMA) is a benign motor neuron disease with bilateral muscular atrophy in asymmetry and abnormal in the electromyography (EMG). However, we report a case by the muscle biopsy which shows symptoms of slowly progressive amyotrophy despite having a normal EMG.

Patient concerns: A 51-year-old male was diagnosed with a lower limb amyotrophy, insidious at the onset and located in the distal thigh and the proximal crus near the knee, slowly progressive weakness, and wasting of his right gastrocnemius muscle for the last 20 years.

Diagnoses: He was diagnosed with MMA by the clinical profile, natural history, examinations, and the biopsy.

Interventions: We perform dynamic physical therapy for the patient in this case.

Outcomes: The positive effects of dynamic physical therapy in this case with MMA were shown in this report.

Lessons: The outcome of physical therapy is satisfactory.

Abbreviations: CT = computed tomography, EMG = electromyography, MMA = monomelic amyotrophy, MRC = Medical Research Council, MRI = magnetic resonance imaging.

Keywords: Biopsy, Hirayama disease, lower limb involvement, lower motor neuron, monomelic amyotrophy, spinal muscular atrophy

1. Introduction

Monomelic amyotrophy (MMA) is a benign motor neuron disease in which neurogenic amyotrophy is restricted to either the upper or the lower extremities.[1–5] Abnormalities are present in a large proportion of the cases with electromyographic findings. However, in our study, we report a case of 51-year-old male who showed symptoms of slowly progressive amyotrophy despite having a normal EMG. Additionally, we analyzed cervical magnetic resonance imaging (MRI) in detail with the intention of uncovering any possible spinal cord abnormality, but no such abnormality was found. The majority of the auxiliary laboratory examinations, with the exception of EMG and pathology, display no abnormalities. In comparison with numerous studies in the past, in which distal muscle groups are predominantly involved in this disorder,[6] our cases have patient's limb amyotrophy predominately located at the distal thigh and the proximal crus nearing the knee. This rare location of amyotrophy is what compelled us to investigate further and propose some theories according to previous literature.

2. Case report

A 51-year-old male was diagnosed with lower limb amyotrophy located at the distal thigh and proximal crus near the knee (Fig. 1A), which was insidious at the onset and has been slowly weakening and progressively wasting his right gastrocnemius muscle for the last 20 years. Initially, the weakness was detected during certain physical activities such as squatting and standing up. The muscle weakness had been stable for the past 18 years. However, the weakness has worsened during the last 2 years and is combined with occurrences of cramps at night. Additionally, he denied history of trauma and knee pain.

A careful examination revealed mild atrophy of his right distal thigh muscle groups and significant atrophy of his right proximal crus muscle groups without apparent fasciculations. The diameter of the right crus is 34 cm (Fig. 1B) and 40 cm in the contralateral side (Fig. 1C). The diameter of the thigh is measured to be 44 cm on the left side (Fig. 1D) and 42 cm on the right side (Fig. 1E). According to Medical Research Council (MRC), muscle

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Consent: The current study was approved by the patient for publication of this case report and any accompanying images and ethics committee of the Dalian Municipal Friendship Hospital of Dalian Medical University and Wuxi Ninth People's Hospital affiliated to Soochow University.

The authors report no conflicts of interest.

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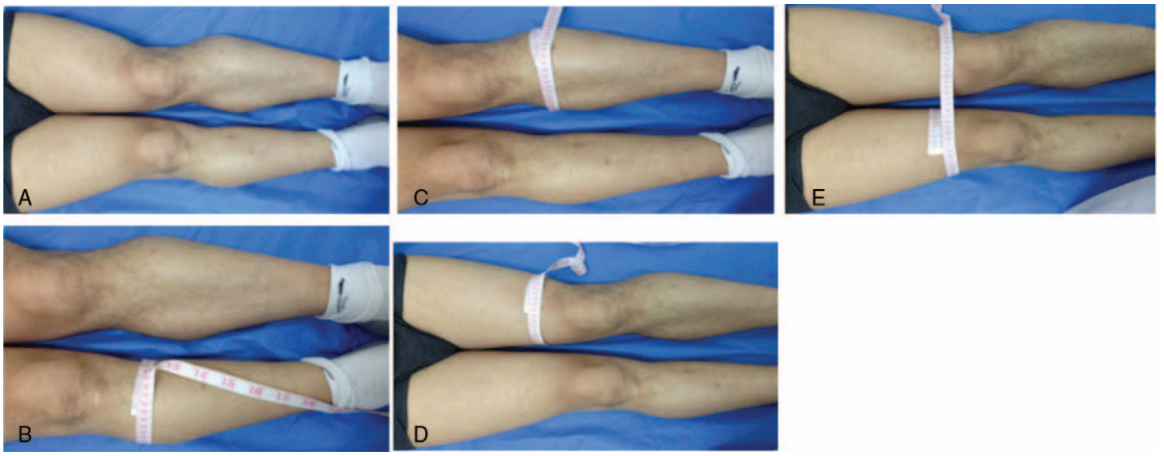

Figure 1. Patient. Right lower limb amyotrophy, located in the distal thigh and the proximal crus near the knee (A); 34 cm in the right crus (5 cm below the tubercle tibia) (B); 40 cm in diameter of the left crus (5 cm below the tubercle tibia) (C); 44 cm in diameter of the left thigh (5 cm above the patella) (D); 42 cm in the right thigh (5 cm above the patella) (E).

strength in his right gastrocnemius muscle was 3/5, which differed from his left side significantly. His muscle strength was 4+/5 in his biceps femoris, and 5/5 in the remaining lower limb muscles. There was no significant abnormality regarding deep tendon flexor reflex and sensory functions in the right lower limb muscles. Furthermore, there were no lower motor neuron signs, and sensory testing showed no obvious abnormality. There was no apparent change for gait and muscle power grades along joint movers.

Routine accessory examinations, including electrocardiogram, abdomen ultrasound, blood biochemical index, erythrocyte sedimentation rate, liver, renal function, and cervical, lumbar MRI revealed no obvious abnormality (Fig. 2E and F). Instead, crus and thigh MRI revealed that fibro adipose tissue lacks muscular tissue (Fig. 2A–D). The motor and sensory conduction velocity examinations show extreme similarity to that of normal limbs. For the sake of clarifying the diagnosis, we examined the atrophic gastrocnemius muscle with local infiltration anesthesia using 1% lidocaine. During the operation, we observed partially light pink appearance of medial head of gastrocnemius muscle and the apparent filling of adipose tissue. We sent the muscle tissue sample to the pathology laboratory. The most clinically significant examination is the muscle biopsy that revealed the coexistence of muscle atrophy and muscle hypertrophy, marked by degeneration and necrosis of muscle fiber, increased nucleus, nuclear chain and solidification nuclear clumping, and the filling of fat between the muscle fiber (Fig. 2G–J). Finally, we considered MMA based on local pathology findings and lack of evidence of spinal cord compression by MRI. An exercise program was planned to strengthen the muscles around the knee joint and to increase the active range of motion. This exercise program was prescribed to the patient in the form of daily physical activities. After a comprehensive assessment of the physique of the patient, he was instructed to perform straight leg raises in repetitions of 20 second leg raising and 10 second rest. The patient was asked to perform the routine daily for 30 minutes. In addition, swimming was advised and patient started to swim 2 days per week. The patient's physical endurance has improved and his swimming distance had increased substantially after the implementation of the exercise program and the outcome of physical therapy is satisfactory.

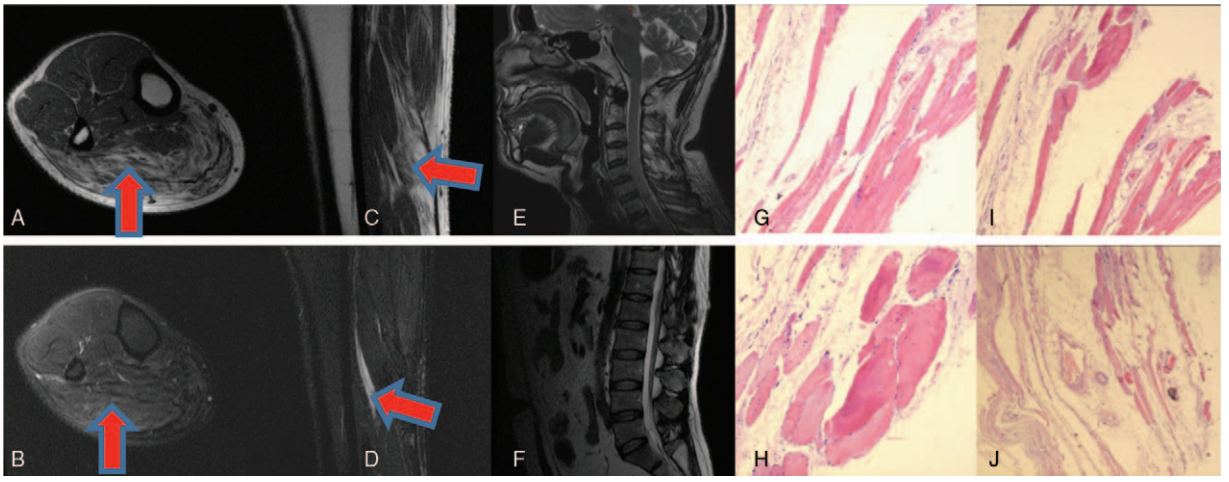

Figure 2. Magnetic resonance imaging (MRI) and muscle biopsy. Muscle atrophy in gastrocnemius muscle and soleus with the fill of adipose tissue between the muscular tissue (A, B, C, and D). Cervical spine and lumbar MRI, demonstrating no obvious abnormality (E and F). The coexistence of muscle atrophy and muscle hypertrophy, the degeneration and necrosis of muscle fiber, and the filling of fat between the muscle fibers (G, H, I, and J).

3. Discussion

MMA is a benign disease, initially described in 1959 by Hirayama et al,[7] features sporadic occurrence, insidious onset, unilateral, bilateral muscular atrophy in asymmetry,[7,8] and slowly progressive course. MMA is an extremely rare condition that has geographical imparity[9,10] with unknown incidence and no distinct familial history with higher occurrence in juvenile males.[11] The majority of the cases were reported in India and Japan.[9,10] However, the minority of cases have been delineated in Western countries. In 1978, Sobue et al[12] had a description of 71 cases of muscular atrophy in upper limb, which found that merely a father and a son suffered from the disorder and only 12 of the 71 patients were female. In comparison, males are more inclined to be diagnosed with the disease as shown by Robberecht et al[13] who reported 375 cases of typical focal amyotrophy of upper limb in 1997, in which only 10.7% of the 375 patients were female.

What is noteworthy is that the limb amyotrophy in our patient focuses on mild atrophy of his right distal thigh muscle groups and significantly obvious atrophy of his right proximal crus muscle groups. Ten cases reported by Gurie-Devie et al[14] found that the amyotrophy was only developed in the thigh and in no cases, were amyotrophy present in the proximal lower limb. However, the patient in our case has amyotrophy around the knee joint, which differentiated from the previous reported cases.

In regard to our patient, we did not find anything abnormal during routine EMG studies, especially in motor and sensory conduction velocity and fasciculation potentials. However, most reported cases indicate that almost all neurogenic changes were present in EMG studies and even in the contralateral limb, as described by Marcos.R.G et al.[15] Computed tomography (CT), MRI, and biopsy of the muscles involved can all contribute to making accurate judgement on muscle involvement. Nonetheless, when compared to CT scan, MRI scan and muscle biopsy yield more convincing results and offer more insights. Past studies[16] reported patients diagnosed with Benign monomelic amyotrophy (BMA) of the distal lower limb showed intensified signals in the MRI scan, particularly in the semimembranous, semitendinous, vastus intermedius muscles, as well as the soleus and medial gastrocnemius muscles on the right side of the affected limb. MRI of patient's muscles demonstrates that fibro adipose tissue is filled with nonmuscular tissue, which was completely in agreement with the results of muscle biopsy of muscles involved. Ultimately, MRI examination integrated with muscle biopsy contributed greatly to the detection of such affected muscles.

4. Conclusion

We should take the diagnosis of BMA into account if our patients show the symptoms, namely the slowly progressive amyotrophy constrained to one limb and clinical stabilization with neurogenic changes in the EMG. However, our 51-year-old male patient, who was diagnosed with distal thigh and the proximal crus involvement 20 years ago, showed no abnormality in the EMG performance to support the diagnosis of BMA. Meanwhile, an exercise program was planned to strengthen the muscles around the knee joint and to increase the active range of motion. The positive effects of dynamic physical therapy in this case with MMA are evident in this report.

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Author contributions

Supervision: Feng Han.
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