Clinical Case Report

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Five-year serial follow-up of muscle MRI in adult onset myotonic dystrophy type 1

A case report

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Abstract

Rationale: Although several studies have described the involvement pattern of myotonic dystrophy type 1 (DM1) using muscle MRI, most of these studies have limitations as cross-sectional studies. To the best of our knowledge, there have been no reports of longitudinal studies describing muscle involvement patterns in patients with DM1 via serial MRI.

Patient concerns: Progressive weakness of both lower extremities.

Diagnosis: Two patients with DM1.

Intervention: The serial muscle MRI performed in the 2 patients with DM1.

Outcomes: The serial muscle MRI showed early involvement of proximal (tensor fascia latae) and truncal muscles (spine extensor muscles), and these longitudinal imaging may be helpful to reveal the pattern of muscle involvement in patients with DM1.

Lessons: Since most previous studies on muscle involvement patterns in DM1 patients were cross-sectional studies, this case series of studying muscle involvement patterns through serial MRI in patients with DM1 may have significant clinical significance.

Abbreviations: 6 MWT = 6 minute walking test, DM1 = myotonic dystrophy type 1.

Keywords: 6 MWT, CTG, DM1, muscle MRI, myotonic dystrophy, serial MRI

1. Introduction

Myotonic dystrophy (OMIM 160900) is the most common form of adult onset muscular dystrophy. Myotonic dystrophy type 1 (DM1) shows an autosomal dominant trait and is caused by pathological cytosine-thymine-guanine (CTG) repeats expansion in the 3′-untranslated region of the DMPK gene in chromosome 19q13.3.[1] Although several studies have described the involvement pattern of myotonic dystrophy type 1 (DM1) using muscle MRI, most of these studies have limitations as cross-sectional studies.[2–4] To the best of our knowledge, there have been no reports of longitudinal studies describing muscle involvement patterns in patients with DM1 via serial MRI. The families of patients were informed and they provided their consent and this case series was approved by Institutional Review Board of Kyungpook National University Chilgok Hospital.

2. Case reports

Patient A is a 55-year-old woman diagnosed with DM1 5 years previously, with an increased CTG repeat length of 650. She had distal muscle weakness, which began in her early 40s. At the time of the first muscle MRI (Fig. 1A), the patient showed weakness only in ankle dorsiflexion and plantar flexion (grade 3) as well as finger flexion (grade 4), and her initial 6-minute walking test (6 MWT) result was 244.2 m. Five years later, a follow-up muscle MRI was performed (Fig. 1B), and the patient showed a deterioration in motor power, prominent ankle dorsiflexor weakness (grade 2), and a decrease in the 6 MWT result to 198.8 m.

Patient B is a 56-year-old woman diagnosed with DM1 5 years previously, with an increased CTG repeat length of 150. The patient only had finger flexor weakness (grade 3), and her 6 MWT result was 516 m at the time of the initial muscle MRI (Fig. 1C). After 5 years, a follow-up muscle MRI was performed (Fig. 1D), and the patient showed aggravated muscle weakness, ankle dorsiflexor weakness (grade 4), and a decreased 6 MWT result of 332.5 m.

Editor: N/A.

Author Contribution

J-SP: acquisition of data, revision of manuscript.

DP: critical revision of manuscript for intellectual content

Author Disclosure

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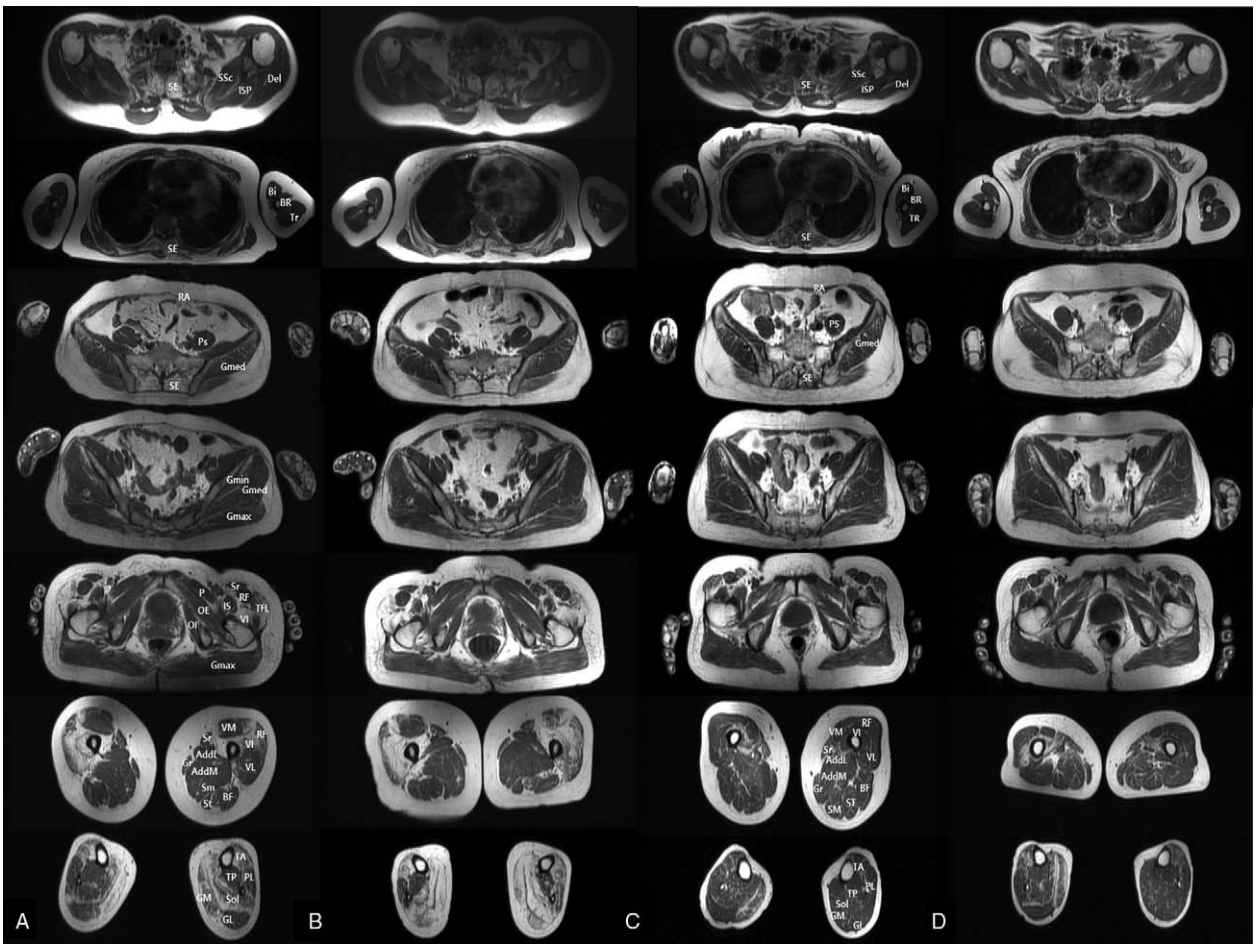

Figure 1. (A) Initial T1-weighted muscle MRI of patient A (51-year-old, CTG repeat 650). (B) Follow-up muscle MRI of patient A performed 5 years after presentation. (C) Initial T1-weighted muscle MRI of patient B (55-year-old, CTG repeat 150). (D) Follow-up muscle MRI of patient B performed 5 years after presentation. AddL = adductor longus, AddM = adductor magnus, BF = biceps femoris, Bi = biceps, Br = brachioradialis, CTG = cytosine-thymine-guanine, Del = deltoid, GL = lateral head of gastrocnemius, GM = medial head of gastrocnemius, Gmax = gluteus maximus, Gmed = gluteus medius, Gmin = gluteus minimus, Gr = gracilis, ISP = infraspinatus, PL = peroneus longus, RF = rectus femoris, Sm = semimembranosus, Sol = soleus, Sr = sartorius, SSc = Subscapularis, St = semitendinosus, TA = tibialis anterior, TFL = tensor fascia latae, TP = tibialis posterior, Tr = triceps, VI = vastus intermidius, VL = vastus lateralis, VM = vastus medialis.

3. Discussion

In the distal lower extremity of both DM1 patients, the medial gastrocnemius tended to be involved initially, followed by the soleus and tibialis anterior muscles. However, the tibialis posterior muscle seemed to be spared despite progression of the disease. Interestingly, in the proximal lower extremity, both the vastus intermedius and medial gastrocnemius muscles tended to show early involvement. In contrast, the vastus medialis and rectus femoris muscles were relatively unaffected. In addition, the tensor fascia latae and spine extensor muscles from the lower cervical to lumbosacral spine level showed an early involvement. It is well known that distal muscles of DM1 are severely affected and our study is of significance as the serial muscle MRI performed in the two patients with DM1 showed an additional early involvement of proximal (tensor fascia latae) and truncal muscles (spine extensor muscles). Although the power of explanation is reduced as we enrolled only two patients, these longitudinal imaging may be helpful to reveal the pattern of muscle involvement in patients with DM1. Furthermore, muscle involvement patterns through serial MRI in patients with DM1 may reveal the patho-mechanism in the progression of the disease and further well designed longitudinal studies are warranted to understand DM1.

References

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