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Case Report

Spinal fusion in a patient with Fukuyama congenital muscular dystrophy

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Abstract

Many studies have evaluated surgical treatments for spinal deformities in patients with neuromuscular disease. However, few reports have described patients with Fukuyama congenital muscular dystrophy (FCMD). A 13-year-old boy with FCMD was unable to sit for long periods or sleep in the supine position because of progressive scoliosis. His Cobb angle worsened from 27° to 41° in 5 months. He underwent standard posterior spinal fusion and pedicle-screw-alone fixation from T5 to S1. Postoperatively, his Cobb angle improved from 41° to 25° without exacerbation for 2 years. After the surgery, he was able to sit for longer periods without pain, and he and his family were satisfied with the efficacy of the spinal fusion. Some patients with mild FCMD can sit at the age of puberty, but progression to scoliosis is possible. Therefore, spinal fusion for progressive scoliosis in patients with FCMD should be considered.

Keywords: Scoliosis; Spinal fusion; Fukuyama congenital muscular dystrophy

1. Introduction

Scoliosis is a common deformity in many types of neuromuscular disease (NMD), and scoliosis management is an important but often challenging issue. For patients with severe scoliosis, sitting is often difficult and accompanied by pain and decreased quality of life [1].

Spinal fusion for progressive scoliosis in patients with NMDs such as spinal muscular atrophy, cerebral palsy, and particularly Duchenne muscular dystrophy (DMD) is becoming more widely performed. In one study of patients with DMD, respiratory function and the ability to sit comfortably and maintain balance were significantly better in the surgical than nonsurgical group [2]. Therefore, we should provide information about surgical options to patients and their parents and support them in choosing appropriate and adequate treatment.

Despite the establishment of care guidelines for congenital muscular dystrophies [3], few reports have described surgical correction in patients with Fukuyama congenital muscular dystrophy (FCMD). FCMD is the second most common form of muscular dystrophy among Japanese. FCMD is characterized by hypotonia, symmetric generalized muscle weakness, and central nervous system migration disturbances. A 3-kb retrotransposal insertion was detected as the ancestral founder

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mutation of the FCMD gene *FKTN*. In one analysis, 80% of Japanese patients with FCMD were homozygous for this founder mutation [4,5]. In another report, 20 of 36 patients with FCMD had scoliosis; however, the natural course and treatment of scoliosis in patients with FCMD is poorly understood [6]. We herein describe a patient with FCMD and the ancestral founder mutation of *FKTN* who underwent scoliosis surgery. We obtained informed consent from him and his family.

2. Case report

A 13-year-old boy with FCMD (homozygous for 3-kb insertion mutation of *FKTN*) had progressive scoliosis. He displayed a typical FCMD phenotype: he had moderate intellectual disability, could have simple conversations, could sit unassisted, and could move around in a motorized wheelchair. Gradually, he became uncomfortable in his wheelchair, felt pain after sitting for 1 h, was unable to sleep in the supine position, and needed postural changes every 1–2 h because of pain arising from progressive scoliosis.

His scoliosis rapidly worsened as follows: Cobb angle, 41° (increased from 27° 5 months previously); thoracic kyphosis, 47°; lumbar lordosis, −10°; and pelvic obliquity, 12° (Fig. 1). One week preoperatively, his percentage of forced vital capacity (%FVC) and percentage of vital capacity (%VC) were 59.0% and 56.2%, respectively, and he was not ventilator-dependent. His ejection fraction was 64%. His parents were informed about the risks, benefits, and prognosis with and without surgery. We also explained the surgery to the patient in plain words, and he roughly understood the procedure.

He underwent standard posterior spinal fusion and pedicle-screw-alone fixation from T5 to S1 under general anesthesia with no respiratory or infectious complications. The improvement with spinal fusion is shown in Fig. 1. His postoperative %FVC and %VC were 52.7% and 53.9%, respectively. After the surgery, his respiratory function gradually deteriorated; after 2 years, his %FVC and %VC were 44.1% and 48.6%, respectively. However, the spinal fusion prevented exacerbation of scoliosis. At the time of this writing, he was able to sit for longer periods without pain, allowing him to study in school comfortably and attend a graduation trip that involved sitting on a bus for 5 h. He also became able to sleep in the supine position without frequent postural changes. He and his family are fairly satisfied with the surgery according to a direct interview.

3. Discussion

We have described a patient with FCMD who had progressive scoliosis and underwent successful spinal fusion. The extent of scoliosis was improved, and he and his parents were satisfied with the efficacy of spinal fusion. These improvements were sustained for 2 years.

In patients with DMD, early surgical corrections may prevent the rapid decrease of lung function. In one study, the ratio of FCV deterioration was significantly decreased in the surgical group [2]. However, many studies have not demonstrated obvious benefits of scoliosis surgery in patients with DMD in terms of respiratory

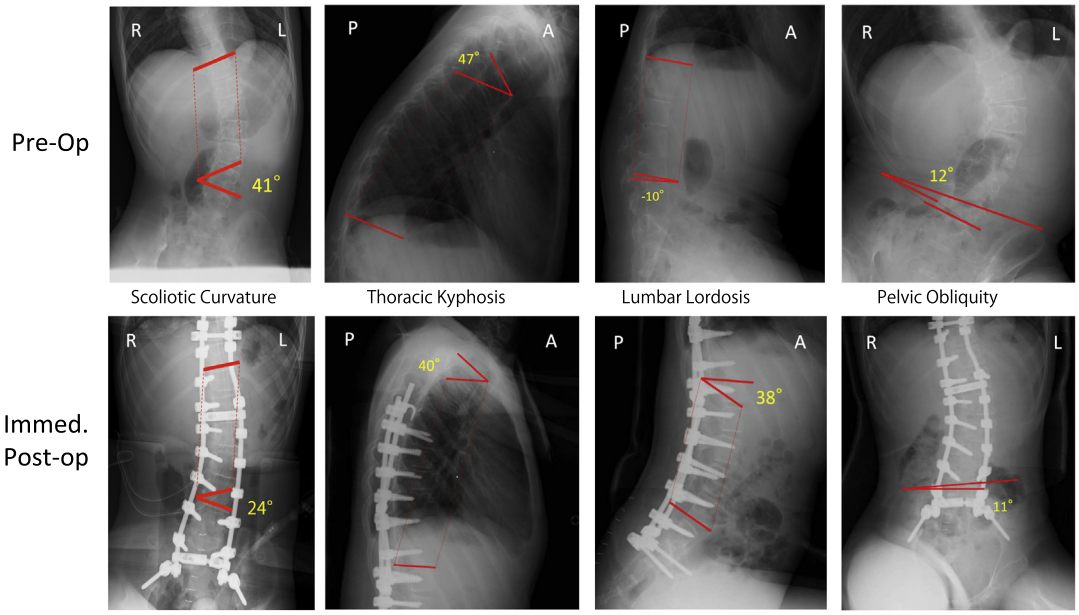

Fig. 1. Pre- and postoperative images of scoliosis. Changes in radiological findings after surgery were as follows: Cobb angle correction, from 41° to 24°; thoracic kyphosis, from 47° to 40°; lumbar lordosis, from −10° to 38°; and pelvic obliquity, from 12° to 11°.