CASE REPORT

Patellar Dislocation in a Patient with Kabuki Syndrome with Severe Mental Retardation: A Case Report

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Background: Kabuki syndrome is a rare congenital syndrome. Individuals with Kabuki syndrome have intellectual disabilities, often combined with skeletal anomalies and joint laxity. We herein report the first case of rehabilitation after reconstruction of the medial patellofemoral ligament in a patient with Kabuki syndrome. Case: A 27-year-old woman with Kabuki syndrome and severe intellectual disability fell during an epileptic seizure. The right patella dislocated and then spontaneously reduced; similar episodes occurred repeatedly. Reconstruction of the medial patellofemoral ligament and lateral retinacular release were performed. Despite an intensive rehabilitation protocol, the patient’s activities of daily living (ADL) did not quickly improve postoperatively because of her severe intellectual impairment and unwillingness to participate in rehabilitation exercises. About 3 months postoperatively, staff encouraged the patient to transfer from a wheelchair to a car, and she was able to get into the car with a little assistance. Subsequently, the patient’s ADL gradually improved. By approximately 1 year postoperatively, the patient was able to ambulate independently for a few meters. Discussion: The patient was thought to be interested in cars and in going for drives. Rehabilitation training for ADL improvement in patients with severe developmental disorders should include activities that the patients consider interesting.

Key Words: Kabuki syndrome; mental retardation; patellar dislocation

INTRODUCTION

Kabuki syndrome is a rare congenital syndrome that involves distinctive facial features and intellectual disability.1–3) The degree of intellectual disability differs substantially among affected individuals1,2); those with serious intellectual disability have profound impairments in adaptive behaviors and communication, whereas those with mild disability whose intelligence is within the normal range do not have these impairments. Kabuki syndrome often involves skeletal anomalies of the vertebrae, hands, hips, and ankles.2,3) Furthermore, joint laxity can cause severe problems in patients with Kabuki syndrome: affected individuals have a relatively high incidence of joint dislocation compared with the general population. Patellar dislocation has been reported in patients with Kabuki syndrome.2,3)

In such patients with patellar dislocation, severe intellectual disability is an impediment to the improvement of their activities of daily living (ADL).4) Intellectual impairment causes difficulties in communication, limiting the ability of clinicians to understand the mindset and feelings of their patients and hindering evaluation of the disease state. Herein, we report a patient with Kabuki syndrome with severe intellectual disability and communication difficulty who underwent reconstruction of the medial patellofemoral ligament (MPFL). Informed consent was obtained from the patient’s parental guardian for the reporting of this case, including the publication of photographs of the patient.

CASE

The patient was born to healthy parents and her intel-
Intellectual disability gradually became evident. She had the characteristic facial features of a wide forehead, arched eyebrows, almond-shaped eyes, depressed tip of the nose, and downward slanting corners of the mouth. She was diagnosed with Kabuki syndrome at 3 years of age. The patient experienced epileptic seizures approximately once per week. She was not able to articulate meaningful words and had serious communication difficulties. She was only able to indicate her intentions with very simple hand signals. The patient was sometimes able to walk and jog, although she had poor balance during gait; she often used a wheelchair.

At 27 years of age, the patient fell during an epileptic seizure. Her right patella dislocated and then spontaneously reduced. Similar episodes of patellar dislocation recurred. However, on one occasion the right patella dislocated and could not be reduced. Consequently, the patient’s mother brought her to the Orthopedic Center of our hospital for treatment.

Radiographs of the right knee revealed complete lateral dislocation of the patella, with hypoplasia of the femoral trochlear groove (Fig. 1). Preoperative evaluation of patellar height and Q angle were difficult because of patellar dislocation. The patient’s lower limb alignment was not clearly varus or valgus; however, no full-length anteroposterior radiographs of the lower limbs were taken. Axial radiography showed the patient’s Dejour classification was grade A. The tibial tubercle–trochlear groove (TT-TG) distance measured on computed tomography was 15 mm (Fig. 2). Manual reduction was tried when the patient was awake. The patella reduced but dislocated immediately following reduction. Treatment using a brace to stabilize the patella in the normal position was tried for 2 weeks. The patella consistently dislocated as soon as the brace was removed. The state of the patella was thought to be almost permanent dislocation rather than a recurrent or habitual dislocation. The patient did not attempt to walk, presumably because of right knee pain. A surgical procedure was considered necessary. Manual reduction was attempted, but the patella could not be stabilized in the normal position even under anesthesia. The contralateral knee showed hyperextension of approximately 20°. A large lateral retinacular release was performed initially under direct vision to reduce the lateral force on the patella because reconstruction of the MPFL alone would not be sufficient to stabilize the patella. However, the patella still tended to dislocate laterally, even during moderate knee flexion. Part of the iliotibial band was released, which finally decreased the tendency of patellar dislocation (Fig. 3). MPFL reconstruction was performed to enable reduction and stabilization of the patella. The gracilis and semitendinous tendons were harvested. These tendons were thin; as a result,

Fig. 1. Preoperative radiographs of the right knee. (A) Anteroposterior radiography showed that the patella was shifted to the lateral side, despite manual reduction. (B) Lateral and (C) axial radiography showed that the patella was completely dislocated laterally. Axial radiography also revealed hypoplasia of the trochlear groove. The sulcus angle was 152° and the Dejour type was grade A.
they were overlapped. The harvested overlapped tendon was secured at both ends with Krackow sutures and then folded in the middle using a suspensory system with one button. Transverse tunnels (4.5 mm and 5 mm) were drilled in the proximal border of the patella under an image intensifier. Also under the image intensifier, a 6.5-mm femoral tunnel was drilled from an anatomical point slightly distal to the medial femoral epicondyle and aimed toward the proximal lateral aspect. The side of the tendon with the suspensory system button was then passed through the femoral tunnel from the medial to the lateral side. Both sutured ends of the tendon were passed between layers 2 and 3 and then passed through the two patella tunnels from the medial to the lateral side. The graft tendon was fixed with suture wires tied at the lateral side of the patella; graft tension was confirmed at about 20° of knee flexion. The tendency of the patella to dislocate was reduced during passive movement under anesthesia; however, passive movement tended to be difficult at more than 90° of knee flexion.

Postoperatively, the right leg was immobilized in an extended position with a splint from the proximal right femur to the right foot because of the tendency for the patella to be easily dislocated intraoperatively. The knee was fixed in an extended position until postoperative day 10, then a brace was used to stabilize the right patella. The patient’s intellectual impairment prevented her from performing ROM exercises with continuous passive motion. The patient did not perform aggressive ROM exercises of the right knee because intraoperative findings indicated the possibility of patellar dislocation. She could not get in her wheelchair independently and always needed complete assistance, especially in transfer and movement. Communication between the patient and medical staff was difficult because we were unable to make her understand our intentions. The communication difficulties and the patient’s intellectual disability prevented us from performing muscle training and from instructing her on voluntary training. We tried straightforward methods of training to improve the patient’s ADL. The patient’s mother used hand signals to encourage her to transfer to the wheelchair; however, the patient did not move independently other than to eat meals. Standing practice using a tilting table was also attempted. Three weeks postoperatively, several rehabilitation staff tried to help the patient walk with a walker, but she just sat on the floor. About 2 months postoperatively, the patient eventually began to transfer to the wheelchair independently and began to walk using a walker with assistance. However, walking training remained difficult because the patient tended to sit on the floor and did not show signs of trying to walk (Fig. 4A). The reasons for this refusal to walk were unclear because of communication challenges.
About 3 months postoperatively, staff encouraged the patient to transfer from the wheelchair to a car, and she was able to get into the car with a little assistance. The patient’s mother told the staff that the patient liked to go for car rides and would remember pleasant memories. Subsequently, the patient’s ADL gradually improved. She became able to walk a few meters using a walker, the wall, and handrails. About 4 months postoperatively, the patient was discharged from hospital. She was able to walk at home using the wall and the back of a chair as support. Walking training continued in the outpatient clinic once weekly, although the patient used hand signals to convey her desire to go home during her initial outpatient sessions. About 6 months postoperatively, the patient was able to walk for 30 to 50 m using a walker without assistance. About 1 year postoperatively, the brace was worn only when the patient left home. The patient was able to walk without a walker and brace for a few meters and to slowly go up and down stairs using handrails at about the same time (Fig. 4B).

The patient’s knee ROM about 3 months after surgery was 60° in passive flexion and −10° in extension. About 7 months after surgery her passive flexion was 80° and extension was −10°. About 2 years after surgery her passive flexion was 90° and extension was −10°. The right patella maintained an almost normal position (Fig. 5).

**DISCUSSION**

Kabuki syndrome was first reported in 1988 by Niikawa and Kuroki and was so named because the facial features of affected individuals resemble those of Kabuki actors. The causal gene mutations have been reported. Individuals with Kabuki syndrome often have skeletal abnormalities as well as ligamentous laxity, joint hypermobility, and joint dislocation. Reports of patellar dislocation in Kabuki syndrome are relatively rare, and surgical treatment has been reported to be sufficiently effective.

The surgical procedure used to treat patellar dislocation in previous reports is mainly medial and distal tibial tubercle transfer. The MPFL is an important soft tissue component for stability of the patella; MPFL reconstruction is an accepted procedure for patellar instability and dislocation.
of 15 mm was thought to be the upper limit of normal; distal tibial tubercle transfer was not considered essential in this case preoperatively.6,7) Therefore, MPFL reconstruction was performed, although this procedure had not been previously reported in Kabuki syndrome patients. In general, rehabilitation after MPFL reconstruction involves no restrictions in ROM exercises or weight-bearing from the first postoperative day5); however, femoral trochlear groove hypoplasia and joint laxity may cause redislocation of the patella.12) The preoperative radiographic findings and intraoperative observations of groove hypoplasia and joint laxity caused clinicians to carefully consider the appropriate postoperative treatment in the present case.

In the 2 years since the surgery, there has been no recurrent patellar dislocation even when the patient experienced epileptic seizures. This indicated that MPFL reconstruction with lateral retinacular release was effective, despite the patient having restricted ROM of the right knee. Intraoperative findings indicated tenderness in the right knee and that patellar dislocation and instability could recur if the patient performed a large ROM. On the basis of the results of clinical and imaging examinations and intraoperative findings, we considered that the state of the patella was almost permanent dislocation. We were not able to perform a more detailed observation preoperatively to determine whether the patella was always dislocated or was reduced between knee movements, even temporarily. Consequently, we were unable to determine the exact type of dislocation. Communication was difficult because of the patient’s intellectual disability; the level of right knee pain likely inhibited our clinical examination, resulting in cautious palpation. Judging from the patellar instability with the patient under anesthesia, if the appropriate surgical procedure was not performed, the patella would remain dislocated regardless of the knee flexion angle. It was not clear why patellar reduction and stabilization in the normal position was not achievable.

The extent of developmental disorders in Kabuki syndrome varies.1,2) There are reports of patients with Kabuki syndrome getting jobs after treatment of patellar dislocation.2) The extent of the developmental disorder in the present patient was major, as evidenced by the patient’s extremely poor communication ability and language difficulties, although precise evaluation was not performed. The severity of the patient’s developmental disorder impeded postoperative improvement in her ADL.4) In many situations, medical staff were unable to assess the patient’s postoperative pain or to determine her intentions. When staff attempted to improve the patient’s ADL by encouraging her to walk, she sat on the floor. The
relatively long duration of fixation of the right knee in the extended position, the postoperative loss of strength, and the patient’s stubborn nature may have temporarily prevented her from performing walking exercises. The patient’s ADL gradually improved after staff attempted to transfer her from a wheelchair to a car. The patient’s mother indicated that the patient was interested in cars and in going for drives. The patient initially wanted to go home during rehabilitation sessions in the outpatient clinic after her discharge from hospital. This suggests that she may have been more dissatisfied with her quality of life during her stay in hospital (including during rehabilitation) than clinicians had realized.

Children with developmental problems reportedly have very wide variations in difficulties with ADL and tend to participate less frequently in those ADL that can be avoided. It is important that medical staff understand each patient’s likes, hobbies, and interests and which activities patients find challenging, especially for those with severe intellectual disability. Obtaining this information is considered fundamental to the success of rehabilitation. Rehabilitation training for ADL improvement in patients with severe developmental disorders should include activities that the patients consider interesting. Medical staff should also relieve stress in patients with communication difficulties; when such patients are attempting to perform activities during rehabilitation that they are not able to do well, their psychological burden is thought to be heavy. To decrease the present patient’s level of anxiety, her mother almost always attended her rehabilitation training sessions. The present patient’s ADL would not have improved without the concerted efforts of her mother.

The present study had some limitations. First, the duration of follow-up was very short. It is possible that the right patella will dislocate again, especially if the patient falls during an epileptic seizure. A brace is now worn when the patient leaves home. Clearly, epilepsy control is important in the present patient, and longer follow-up is needed. Second, it is difficult to perform clinical evaluation via scores in a patient with a developmental disorder; however, the patient’s mother was satisfied with the improvement in the patient’s ADL. We were able to evaluate passive ROM of the right knee. The evaluation of extension strength, including extension lag, was not possible because of communication difficulties resulting from the patient’s intellectual impairment. Third, genetic diagnosis was not performed in the present patient; the patient’s mother did not want this testing done because a definitive genetic diagnosis would not positively change the patient’s situation, including the developmental disorder and ADL.

CONCLUSIONS

The present case of patellar dislocation in a patient with Kabuki syndrome with severe intellectual disability required MPFL reconstruction and lateral retinacular release. Postop-
ervative improvement in the patient’s ADL was difficult because the patient’s severe intellectual disability made communication very difficult. In patients with severe intellectual disability, training for ADL improvement should include activities that the patient considers interesting.

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CONFLICTS OF INTEREST

The authors certify that there is no conflict of interest with any financial organization regarding the material discussed in the manuscript.

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