Progress in Orthopedic Science

Case Report

DOI: doi.org/10.47363/POS/2015(1)104

The surgical treatment of patellar subluxation in a case of kabuki make-up syndrome

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Received: April 01, 2015
Accepted: April 27, 2015
Published: May 11, 2015

ABSTRACT

Kabuki make-up syndrome (KS) or Niikawa-Kuroki syndrome is a rare autosomal dominant disorder with an incidence of 1 per 30,000-40,000 that is characterized by multiple congenital abnormalities. The diagnosis is based on the clinical findings. Pathogenic variants in the KMT2D gene have been identified as the most common cause of KS. Our patient underwent right knee surgery for patellar subluxation. At surgery, the patellar tendon was removed from its attachment on the tibial tuberosity, medialized, and anchored. The patellar retinaculum was loosened, and medial plication was performed (the Insall Technique). Post-operatively, the patient was treated in our rehabilitation clinic. KS is a rare syndrome and surgery for patellar subluxation can be very important to obtain clinical improvement. The rehabilitation program should be continued post-operatively.

KEY WORDS: Kabuki syndrome, patellar subluxation, surgery

INTRODUCTION

Kabuki syndrome (KS) or Niikawa-Kuroki syndrome is a rare autosomal dominant disorder involving pathogenic mutations in the *KMT2D* and *KDM6A* genes [1]. *KMT2D* has 54 coding exons and encodes a 5537-amino-acid histone-lysine N-methyltransferase that belongs to the trithorax group of proteins.

The diagnosis is based on clinical findings. KS has five cardinal features: Facial dysmorphism, development delay or mental retardation, postnatal short stature, skeletal anomalies, and dermatoglyphic anomalies. Short stature and cardiac anomalies differ in frequency between patients with and without *KMT2D* mutations. High arched eyebrows, a short fifth finger, and hypotonia in infancy are more frequent with the *KMT2D* mutation than the *KDM6A* mutation. Short stature and postnatal growth retardation are observed in all individuals with *KDM6A* mutations, but in only half of those with *KMT2D* mutations. Several chromosomal abnormalities have been found in a large number of affected individuals, but none is specific to KS [2].

The most frequent skeletal abnormalities in KS are brachydactyly, clinodactyly affecting the fifth finger, pseudarthrosis of the clavicle, clubfoot, ligament laxity, and joint dislocation [1-3]. Ligament laxity and joint hyper-extensibility have been reported in 74~96% of the patients with KS. Joint dislocation occurs in up to 50% of KS patients [4]. Patellar dislocation can lead to significant disability and require surgical treatment. A quick motion involving the patellar joint in KS patients with lax knee joints can increase the risk of patellar dislocation.

This paper presents a case of KS, who had many abnormalities. Her surgical intervention is described and discussed in light of the literature.

CASE REPORT

A patient with KS who underwent patellar surgery on her right knee was referred to our clinic. She was born by spontaneous vaginal delivery at the 7th month of pregnancy as a 750 g premature baby. At birth, she was purple-colored and cried with a thin sound, but a high-pitched tone. Her mother had taken medications for an ovarian cyst in the 7th month of her pregnancy. The patient's three siblings had no disease. The patient started walking and talking at the ages of 1 and 5 years, respectively. She complained of getting tired easily and feeling prostrated until she was five. In 2009, she underwent right knee surgery for patellar subluxation. The same operation is planned for the left knee, which has similar patellar problems.

The patient was 115 cm tall and weighed 30 kg. She had bad oral hygiene and a high-arched palate. Other clinical findings included arched eyebrows, long palpebral fissures, ectropion on the lateral side of the lower eyelids, thin nose-wings, and a short nasal septum. She had increased joint laxity in the upper and lower extremities, large ears, finger pillows in the hands, short fifth fingers bilaterally, brachydactyly and hammer-toe deformity of the toes, and thoracic scoliosis concave to the left. Mentally, the patient was 2 years behind children of the same age. Growth retardation was present relative to her age. No other abnormality was observed.

In the laboratory examination, the hemogram, routine biochemistry, and hormone parameters were within the normal ranges. A knee X-ray was obtained postoperatively [Figures 1 and 2]. Her abdominal ultrasonography (USG) was normal. USG of the left kidney showed focal parenchymal thinning in the upper part, the parenchyma was 3 mm thick, and the renal pelvis was divided into two by a hypoechoic parenchymal band (a bifid renal pelvis-duplex-collecting system). Her echocardiogram was normal. Genetic testing identified the *KMT2D* gene mutation.

In 2009, the patient underwent soft-tissue surgery on the right knee; the patellar tendon was removed from its attachment on the tibial tuberosity, medialized, and anchored. The patellar retinaculum was loosened, and medial plication was performed. The patient could walk independently, but she had pain and knee limitation. The patient was admitted to our clinic for strengthening exercises, stretching and therapeutic electrical stimulation. She could walk more comfortable and properly after the therapy.

DISCUSSION

Joint dislocations occur in 50% of KS patients, and protective measures should be taken. Patellar dislocation can cause disability and might require surgical therapy [5-17]. Older children, adolescents, obese girls, and patients with ligament laxity are at higher risk of patellar dislocation. There are few published reports of different surgical treatments in patients with KS for patellar dislocation [4,18].

The surgical techniques used for correction include the Elmslie Trillat procedure, Campbell procedure, patellar fragment resection and tibial tubercle transfer, and Insall Technique (tibial tubercle transfer and vastus medialis surgery) [5]. Seven patients operated on for patellar dislocation have been reported [8]. Niikawa and Burke reported two cases, but they did not explain the surgical process. Ikegawa *et al*. reported two adolescent patients with recurrent patellar dislocation who had general ligamentous laxity; the Elmslie Trillat and Campbell procedures were used and between these two techniques, the Elmslie Trillat procedure had the better result [17]. Kurosowa reported a patient who improved after resection of the patellar fragment and tibial tubercle transfer [18]. Rouffiange reported two cases who were operated on successfully with the Insall Technique [5]. The Insall Technique was used on our patient, who could walk independently after surgery. No complications occurred during rehabilitation.

The clinical parameters should be assessed regularly in the therapy and follow-up of patients with KS. Patients with joint dislocation should be treated primarily with conservative methods, with surgical treatment if required. Satisfactory post-surgical recovery and healing have been obtained with the Elmslie Trillat and Insall Techniques.

Patients with KS might require surgical treatment and the rehabilitation program should be continued post-operatively.

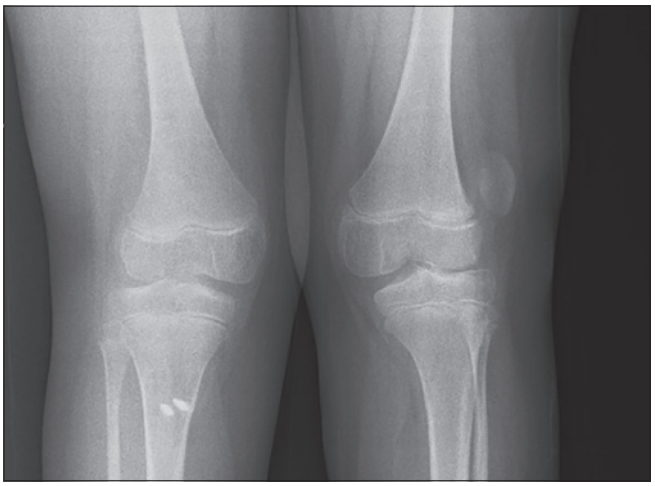

Figure 1: Posterior-anterior X-ray of the patient's knee

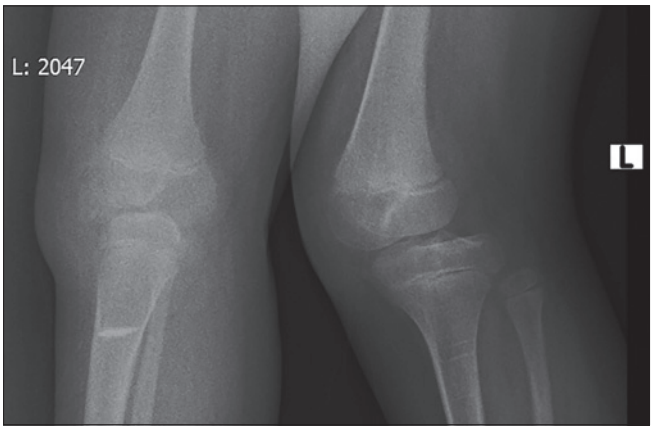

Figure 2: Lateral X-ray of the patient's knee

Strengthening exercises are advised for patients being treated conservatively. The rehabilitation therapy should be appropriate for the patient's situation. As these patients can have disabilities due to skeletal abnormalities, they should be examined carefully for such problems.

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Source of Support: Nil, Conflict of Interest: None declared.