

PERMANENT CONGENITAL DISLOCATION OF THE PATELLA IN NIJKAWA-KUROKI (KABUKI MAKE-UP) SYNDROME

L. AULISA, F. SERRA, F. TAMBURRELLI, S. LUPPARELLI, R. PADUA

The Kabuki make-up syndrome (KMS) is a complex malformation syndrome characterized by peculiar facies, mental retardation, dermatoglyphic abnormalities, postnatal growth deficiency and skeletal anomalies. One case of permanent congenital bilateral dislocation of the patella in an Italian girl affected by KMS is reported. The condition of the knee in KMS seems to be coincidental and is the only one reported in the literature to date.

Keywords : Kabuki syndrome ; permanent patella dislocation.

Mots-clés : syndrome de Kabuki ; luxation permanente de la rotule.

INTRODUCTION

Permanent congenital dislocation of the patella may exist either as an isolated abnormality or in association with other skeletal deformities, although it has not yet been assessed whether this association is statistically significant or coincidental.

Kabuki make-up syndrome (KMS) is a complex syndrome characterized by peculiar facies, mental retardation, dermatoglyphic abnormalities, postnatal growth deficiency and skeletal anomalies. We describe one case of permanent congenital bilateral dislocation of the patella in an Italian girl affected by KMS. The clinical and radiographic features are discussed, along with the surgical procedure used for correction.

CASE REPORT

A 7-year-old Italian girl affected by the Kabuki make-up syndrome (fig. 1) was admitted to the



Fig. 1. — R. D., a 7-year-old Italian girl affected by Kabuki make-up syndrome. Note the typical facies with large palpebral fissures, everted eyelids, thinning hair at the lateral third of the eyebrow and depressedinion. (The patient's face was photographed for publication with parents' permission).

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Orthopedic Department because of bilateral genu flexum severely hindering her ambulation.

Physical examination showed marked hypotrophy of her thigh and calf muscles. The patellae were not palpable in the intercondylar fossa of both knees. Only by careful examination could two hypoplastic patellae, irreducibly dislocated on the lateral aspect of the lateral femoral condyle, be detected. Hamstring contracture limited active extension of the knees and full extension could not be passively obtained.

Hamstring tenoplasty and posterior capsulotomy was then performed; this resulted in complete passive extension of both knees.

Radiographic examination performed six months later showed two hypoplastic patellae, lying close to the lateral femur. Marked hypoplasia of both femoral condyles and a shallow trochlear groove were evident on sunrise views at 45° of flexion (fig. 2). The joint surfaces of the dislocated patellae were flat without a median ridge.

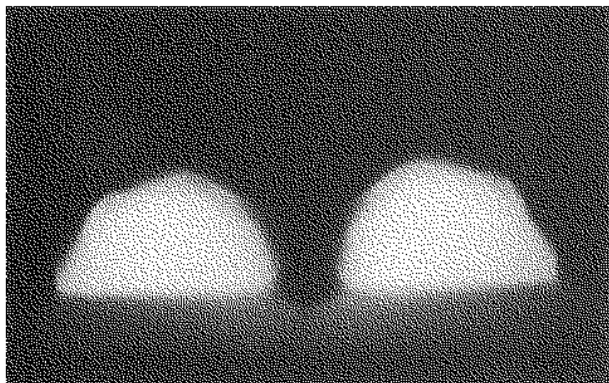


Fig. 2. — Comparative sunrise view of the knees at 45° of flexion. The lateral dislocation of both patellae is evident. The lateral femoral condyles are flat, and the trochlear groove is shallow.

Bilateral valgus deformity was demonstrated, as well as increased outward rotation and lateral subluxation of the tibias. Marked external tibial torsion was evident.

Open relocation of the dislocated patellae was then performed, initially on the right knee and, 1 year later, on the contralateral knee, using Krogus' capsuloplasty technique and a modification of the Goldthwait patellar tendon transfer technique. Some particular pathologic findings

were noted at surgery, and noteworthy technical aspects of the surgical proceeding are reported below.

After blunt exposure of the vastus medialis and a hypertrophic and stretched medial alar ligament, the patella was found to adhere to the lateral epicondyle and resisted all efforts at manual relocation (fig. 3).

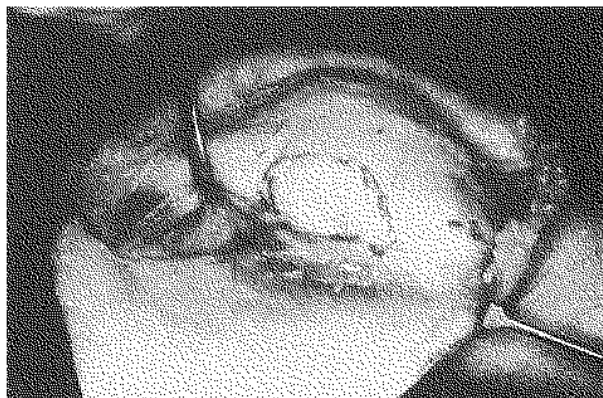


Fig. 3. — The surgical field shows that the whole extensor apparatus of the knee is laterally displaced. The patella adheres to the lateral femoral condyle, and the vastus lateralis inserts onto the superolateral margin of the patella. The medial portion of the capsule and the medial retinaculum are distended. Note the marked lateral displacement of the patellar tendon.

The rectus femoris and the vastus lateralis were found to be completely dislocated laterally and inserted onto the superolateral patellar margins. The iliotibial band and the patellar tendon were attached to the lateral aspect of the tibial metaphysis.

Owing to the severe external tibial torsion and the anomalous directions and insertions of the muscles, particular attention was given to the reattachment of the patellar tendon on the anteromedial cortex of the tibial metaphysis according to Goldthwait's procedure. The tendon inclination was set in such a fashion that patellar reduction was maintained throughout the whole range of motion.

Clinical follow-up six months after surgery revealed that the patient had attained a complete range of motion.

The control sunrise view of the patient's knees, one and two years postsurgery, showed a perma-

ment and satisfactory relocation of the patellae. Remodeling of the right femoral trochlea, when compared to the opposite side, was noted to have occurred in two years' time (fig. 4).

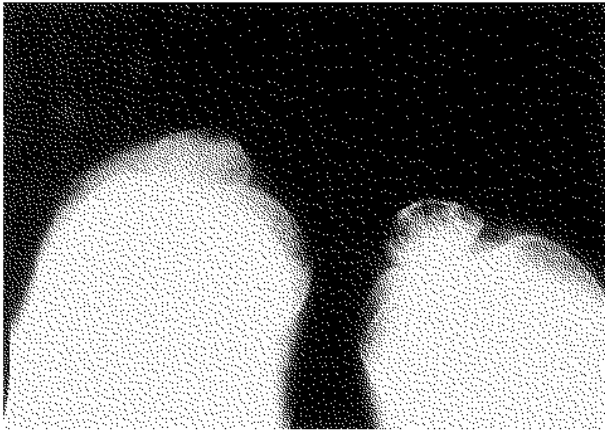


Fig. 4. — Comparative sunrise view of the knees at 45° of flexion two years after surgery. The xray film shows good patellofemoral congruence. Remarkably, remodeling of the right femoral trochlea occurred: the lateral femoral condyle is less hypoplastic when compared to its appearance in the radiograph taken before surgery (see fig. 2). The left trochlea, one year after surgery, has not yet shown any evidence of remodeling.

DISCUSSION

Kabuki make-up syndrome (KMS) was originally observed in Japanese patients in 1981 (3, 4). It has since been increasingly reported in the Japanese population as well as in other ethnic groups. Several skeletal anomalies were described in KMS, but only three cases of recurrent patellar dislocation in adolescent girls have been recorded so far (2). In our case permanent dislocation of the patella was found in association with the Kabuki make-up syndrome. The difference between the three cases just listed (2) and the present one is that in our case the dislocation was permanent, impairing the walking capability of the patient. To our knowledge this pathological condition associated with KMS has not been reported to date.

The basic anatomic features of permanent dislocation of the patella have been well delineated and can be summarized in the irreducibility of the dislocated patella throughout the whole range of motion. The absence of congruence between the patella and the femoral trochlea causes both joint surfaces to become dysplastic. On the contrary, a great deal of controversy regarding the etiopathogenesis of congenital dislocation of the patella still exists.

Some authors believe that the permanent dislocation may result from fibrosis of the vastus lateralis muscle. According to the extent and time of onset of the fibrotic processes, either type of dislocation, namely the permanent or the habitual one, would ensue (1).

A thorough discussion of the etiopathogenesis of permanent dislocation of the patella would go beyond the scope of this case report.

In this context our view is that any surgical treatment, by respecting the physiology of the growing skeleton, ought to aim at reducing the patella into the femoral trochlea, so that the reestablished femoropatellar congruence may remodel both joint surfaces (fig. 4).

This case report, by showing the presence of permanent dislocation of the patella in KMS, aims to make the orthopedic surgeon aware of such a pathological association.

REFERENCES

1. Bose K., Chong K. C. The clinical manifestations and pathomechanics of contracture of the extensor mechanism of the knee. *J. Bone Joint Surg.*, 1976, 58-B, 478-484.
2. Ikegawa S., Sagakuchi R., Kimizuka M., Yanagisako Y., Tokimura F. Recurrent dislocation of the patella in Kabuki make-up syndrome. *J. Pediatr. Orthop.*, 1993, 13, 265-267.
3. Kuroki Y., Suzuki Y., Chyo H., Hata A., Matsui I. A new malformation syndrome of long palpebral fissures, large ears, depressed nasal tip, and skeletal anomalies with postnatal dwarfism and mental retardation. *J. Pediatr.*, 1981, 99, 570-573.
4. Niikawa N., Matsuura N., Fukushima Y., Ohsawa T., Kajii T. Kabuki make-up syndrome: A syndrome of mental retardation, unusual facies, large and protruding ears and postnatal growth deficiency. *J. Pediatr.*, 1981, 99, 560-565.

SAMENVATTING

L. AULISA, F. SERRA, F. TAMBURRELLI, S. LUPPARELLI, R. PADUA. *Permanente luxatie van de patella in het syndroom van Niikawa-Kuroki (Kabuki make-up) syndroom.*

Het Kabuki make-up is een congenitaal malformatief syndroom met een specifieke facies, mentale retardatie, anomalieën van de huidlijsten van de vingertoppen, groeistoornissen en skeletafwijkingen. Wij beschrijven een geval met een permanente, bilaterale luxatie van de patellae bij een jonge Italiaanse met het Kabuki syndroom. Deze associatie werd voorheen niet beschreven.

RÉSUMÉ

L. AULISA, F. SERRA, F. TAMBURRELLI, S. LUPPARELLI, R. PADUA. *Luxation permanente de la rotule dans un syndrome de Niikawa-Kuroki (Kabuki make-up).*

Le Kabuki make-up syndrome (KMS) est un syndrome malformatif complexe caractérisé par un faciès particulier, un retard mental, des anomalies dermatoglyphiques, un déficit de croissance et des anomalies du squelette. Nous rapportons un cas de luxation permanente bilatérale de la rotule chez une jeune fille italienne présentant ce syndrome. Une telle association n'a jamais été décrite dans la littérature.