

# Nail-Patella Syndrome in Saudi Arabia With New Features and Surgical Procedures: The First Described Study

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## Abstract

The purpose of this study was to reveal the occurrence of nail-patella syndrome (NPS) in Saudi Arabia together with the detection of abnormal attachment of lateral meniscus in the left knee and new surgical procedures applied to the right and left knee, reported for the first time in this study. This was a case study of a 23-year-old young man presenting with bilateral knee pain, giving way and locking since the age of 15 years. Clinically, most of the NPS features were noted, including ocular problems. The complex features affected both knees, especially the previous attempted surgeries for recurrent dislocation of patellae. Deficient ligaments were reconstructed using the Leeds-Keio ligament, starting with the right knee and continuing with the left knee 6 months later.

Early and late follow-up showed favorable outcome of surgery revealed as independent ambulation and stable right and left knees. In conclusion, NPS, although rare, presents a complex problem and unexpected surgical outcome, and we recommend this procedure with close follow-up.

## Introduction

NPS is a pleiotropic disorder exhibiting autosomal dominant inheritance.[1] It is a result of mutations in the LIM-homeodomain gene *LMX1B*. Haploinsufficiency of *LMX1B* underlies this disorder, with a high degree of variability in phenotypes.[2] NPS is characterized by absence or hypoplasia of nails and patella, posterior iliac horns in more than 80% of cases, congenital nephropathy, cervical ribs, and eye problems.[3] It is also known as hereditary onycho-osteodysplasia (HOOD), with thumb and index fingers mostly affected.[4] Other abnormal features have been reported, such as antecubital pterygium with poor functional results.[5–7] Additional features include clavicular horn,[8] foot abnormalities,[9] spondylolisthesis,[10] skull defect,[11] absence of fibula,[12] and shoulder and first rib dysplasia.[13,14] Nephropathies contribute significantly in NPS[15] in addition to the classical tetrad of the syndrome.[16] Further relationships with other clinical conditions such as pregnancy and their complications have also been noted.[17] These wide variable clinical features have enabled the development of a scoring

system to quantify them.[18] The autosomal dominant expression of NPS has been noted sporadically and in families at different locations.[6,7,10,19–22] The diagnosis of NPS is based on clinical and radiologic parameters that have allowed further evaluation of the iliac horns by computed tomography and magnetic resonance imaging.[16,23] In addition, prenatal diagnosis was possible through ultrasonography.[3,24] Of note, another clinical entity has been described as small-patella syndrome.[25] It resembles NPS in patellar abnormalities but includes other abnormalities such as coxa vara or valga with buttressing of femoral head, hypoplasia of lesser trochanter, bilateral defective ischiopubic junctions, and foot abnormalities.[26,27]

This study describes for the first time new clinical features and the surgical procedures applied in NPS.

## Methods

A 23-year-old young man presented to the Orthopedic Clinic under the care of the author in October 1988, complaining of bilateral knee pain but more on the right side. He reported inability to walk or perform normal functional duties with history of giving way and locking, especially the right knee, since the age of 15 years. He had multiple surgical procedures at other centers for both knees due to recurrent dislocation of patellae since 1981, starting with tibial tubercle transposition in the left knee. The same procedure was carried out on the right knee in 1982. The last operation before presenting to us was right patellectomy in 1986.

On examination, the patient appeared to be thin and was ambulating on the wheel chair. Nail dysplasia was noticed in all fingers and thumbs as well as restriction of elbow movements and involvement of both eyes, but no abnormalities were noted in the feet. Knee examination revealed multiple surgical scars on both knees with a small patella in the left knee and absent patella on the right side. Diffuse tenderness, anterolateral instability in both sides, and marked lateral collateral laxity on the right side were noticed as well. Lachman's test was grade IV in the right knee and grade I in the left knee. Of note, marked clicking was found at the end of extension in the left knee, which had an explanation as noted during surgery from lateral meniscus. Extension lag of 20 degrees was noted in the right knee. Radiologic investigations showed a small patella in the left knee, absent patella in the right knee, hypoplasia of the radial heads, and iliac horns on both sides.

Surgery was planned for October 16, 1988 to reconstruct the deficient ligaments on the right knee. Examination under general anesthesia revealed the same preoperative clinical findings, and

attempted arthroscopy failed due to severe adhesions. Subsequently, the right knee was approached through the previous scar without exploring the joint cavity due to severe adhesions. Reconstruction of deficient lateral collateral ligament and popliteus tendon was done using a Leeds-Keio (L/K) ligament. This was a synthetic material with an open weave polyester mesh having rectangular holes (1.5 mm<sup>2</sup>) and a tensile strength of 2.2 KN. It acted as a scaffold for tissue ingrowths and neoligament formation. Drilling and bone grafts were taken through the lateral tibial and lateral femoral condyles using L/K ligament, which was applied in a U shape, based laterally, and fixed with staples. The slack iliotibial tract was advanced medially and distally, fixing it with a screw.

The patient had uneventful recovery with early ambulation and was discharged home on the 10th day. The cast was removed 12 weeks postoperatively followed by physiotherapy and gait training. Consequently, the left knee was operated 6 months later on April 3, 1989. The approach was through the previous medial parapatellar surgical scar. The anterior cruciate ligament (ACL) was found to be fibrotic, hypoplastic, and attenuated more proximally. The other significant finding was a discoid lateral meniscus with abnormal attachment to the lateral femoral condyle; it was excised and histopathologic study was carried out. Reconstruction of the ACL was done using the L/K ligament with extra-articular reinforcement laterally. The left knee had uneventful recovery as well, with good healing of wounds and response to physiotherapy.

## Results

The diagnosis of NPS in this study was clinically evident from nail changes, a small patella left knee, and radiologic findings ([Figures 1a](#) and [and1b](#)),[1b](#)), whereas the right knee had patellectomy from previous surgery ([Figures 2a](#) and [and2b](#)),[2b](#)). Additional diagnostic radiology revealed hypoplasia of radial heads ([Figures 3a](#) and [and3b](#)),[3b](#)) and bilateral iliac horns in the pelvis ([Figure 4](#)). These clinical abnormalities differed from the right to the left knee because of previous exposures to surgery ([Table 1](#)). The right knee was operated first to reconstruct the deficient lateral collateral ligament using the L/K ligament followed by advancement of slack iliotibial band distally, as seen in [Figure 5](#). Postoperative x-rays showed the right knee with staples fixing the ends of the L/K ligament and a single screw fixing the advanced end of the iliotibial band ([Figures 6a](#) and [and6b](#)),[6b](#)). The left knee was approached surgically differently from the right knee as shown in [Figure 7](#). This was basically to reconstruct the deficient ACL using, similarly, the L/K ligament of double size. However, during exploration of the left knee, a

discoid lateral meniscus was noticed running along the intercondylar groove and attaching to the lateral femoral condyle ([Figure 8](#)).

**Figure 1a**

X-rays of left knee, anteroposterior view.



**Figure 1a**

X-rays of left knee, anteroposterior view.

**Figure 1b**

X-rays of left knee, lateral view.



**Figure 1b**

X-rays of left knee, lateral view.

**Figure 2a**

X-rays of right knee, anteroposterior view.



**Figure 2a**

X-rays of right knee, anteroposterior view.



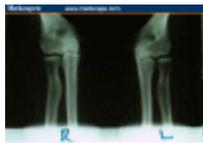
**Figure 2b**

X-rays of right knee, lateral view.



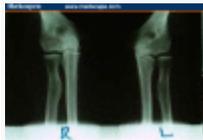
**Figure 2b**

X-rays of right knee, lateral view.



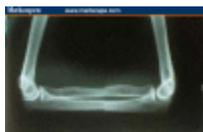
**Figure 3a**

X-rays of elbows, anteroposterior view.



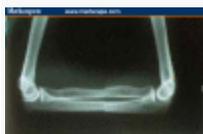
**Figure 3a**

X-rays of elbows, anteroposterior view.



**Figure 3b**

X-rays of elbows, lateral view.



**Figure 3b**  
X-rays of elbows, lateral view.



**Figure 4**  
X-rays of pelvis, anteroposterior view.



**Figure 4**  
X-rays of pelvis, anteroposterior view.

A table with a header "Subjective and Objective Disabilities in Both Knees" and several rows of text. The text is mostly illegible but appears to be a list of items with corresponding values or descriptions.

**Table 1**  
Subjective and Objective Disabilities in Both Knees



**Figure 5**  
Surgical detail of right knee.



**Figure 5**  
Surgical detail of right knee.

**Figure 6a**  
X-ray of right knee postoperatively, anteroposterior view.



**Figure 6a**  
X-ray of right knee postoperatively, anteroposterior view.



**Figure 6b**  
X-ray of right knee postoperatively, lateral view.



**Figure 6b**  
X-ray of right knee postoperatively, lateral view.



**Figure 7**  
Surgical detail of left knee.



**Figure 7**  
Surgical detail of left knee.



**Figure 8**  
Discoid lateral meniscus with abnormal attachment, left knee.



**Figure 8**  
Discoid lateral meniscus with abnormal attachment, left knee.

Histopathology revealed crescent-shaped lateral meniscus of 8-cm length by 1.5-cm width and 1.0-cm thickness with myxoid and degenerative changes. The operative clinical findings were significant in both knees and some were considered important contributory factors to the patient's complaints ([Table 2](#)). The surgical procedures carried out on the same patient to reconstruct both knees were technically different from the right to the left side ([Table 3](#)). This allowed us to consider this study to be the first description of NPS with these new features as seen in ([Table 4](#)).

Abnormal Structures Noticed Operatively in Both Knees	
Structure	Location
Discoid lateral meniscus	Lateral compartment
Abnormal attachment	Anterior horn
Myxoid degeneration	Meniscus
Degenerative changes	Meniscus
...	...

**Table 2**  
Abnormal Structures Noticed Operatively in Both Knees

Surgical Procedures in Both Knees	
Procedure	Side
Arthroscopy	Right
Arthroscopy	Left
...	...

**Table 3**  
Surgical Procedures in Both Knees

**Table 4**  
Reasons This Is the First Report in NPS

## Discussion

Surgically treated patients with NPS have been reported due to different conditions. Some circumstances have been unrelated, such as being a donor for renal transplantation,[\[28\]](#) whereas a total knee replacement for severe deformity due to osteoarthritis in NPS was related to the abnormal structures in the knee.[\[29\]](#) The presence of a small or absent patella predisposes them to instability, recurrent dislocation, and further destruction and deformity. These conditions frequently require surgical intervention of soft tissue reconstruction such as quadricepsplasties and bony operations like tibial tubercle transposition.[\[30,31\]](#) Although knee problems are common in NPS, other sites like the foot and ankle have revealed a higher incidence for no explainable reasons.[\[9\]](#)

The complex nature of NPS clearly affects the outcome of surgery, which has varied from good[\[32\]](#) to bad.[\[5\]](#) Both the complex nature of deficiency and the type of surgery, using new methods, have been applied in our study. The right knee in this patient showed severe instability with deficiency of lateral collateral ligament, absent popliteus tendon, and 2 previous surgeries of tibial tubercle transposition and patellectomy. These, of course, were the prerequisite indications for further surgical intervention to solve these problems and allow the patient to ambulate. It was a major challenge that appeared clearly during surgery when attempted arthroscopy failed due to extensive adhesions and altered anatomy.

The L/K ligament was a good choice for reconstruction of deficient structures, having enough length and durability to provide stability and better function as thought of after all previous surgeries failed to improve the patient's knee conditions. This was the first attempted case to use this implant in NPS. The technique was unique in passing L/K ligament after preparation of drilling and bone grafting through the lateral tibial condyle and then the lateral femoral condyle without exploring the joint cavity due to extensive adhesions and no available space to dissect around. This technique provided excellent stability, especially after advancement of iliotibial tract. Although the use of the L/K ligament in other studies was reported for the reconstruction of ACL in a normal knee,[\[33\]](#) this study proved that it can be used in other complex disorders and, of course, in NPS.

This smooth satisfactory outcome of surgery and good wound healing encouraged the author to proceed with the next surgery in the left knee after 6 months. It is notable that the favorable results of mechanical and functional outcome for the reconstruction of the extensor mechanism of the knee using the L/K ligament were reported in the hands of others but, of course, not in NPS.[34,35] The left knee in this study presented a similar challenge with important findings preoperatively, especially painful clicking due to the abnormal lateral meniscus and its attachment. However, lateral menisectomy eased off the approach to the ACL, which was found to be abnormal as well. This unique abnormal finding of lateral meniscus was not described before in NPS as far as we know. The ACL was reconstructed with extra-articular reinforcement using the L/K ligament, which resulted in excellent stability in all directions. This reinforced the favorable outcome of using this technique and the implant, which was tested to be a success twice in the same patient with very complex knee problems. The patient tolerated the procedures nicely, with uneventful recovery and wound healing.

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