



Case report

Total knee arthroplasty in a patient with nail-patella syndrome (NPS)



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ABSTRACT

Nail-patella syndrome (NPS) or hereditary onycho-osteodysplasia is a rare genetic condition involving a mutation in the LMX1B gene affecting nails, elbows, knees, and pelvis. Due to the regulatory functions of the gene in many developmental processes through the body, patients with NPS experience wide-ranging musculoskeletal problems including patellar instability, fingernail anomalies, iliac exostoses/horns, and elbow abnormalities. The patellar changes often involve aplasia, hypoplasia, and chronic dislocation. Due to these musculoskeletal involvement, arthritis of joints can occur in patients with NPS causing severe pain and disability. This is a case report of a patient with NPS who underwent a total knee arthroplasty for symptomatic knee arthritis.

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1. Introduction

Nail-patella syndrome (NPS), also known as hereditary onychoosteodysplasia, Turner–Keizer, or Fong's disease is a rare genetic condition affecting 1/50,000 people [1]. NPS causes a classic four characteristic features of patellar abnormalities, fingernail dysplasia (triangular lunula), bilateral iliac horns, and elbow anomalies. Other findings may include scoliosis, scapular hypoplasia, scleral discoloration, and forehead abnormalities.

The severity of NPS can vary, and it can also affect other organ systems including renal, eye, and endocrine. While there is no cure available for NPS, several treatments are available to target specific symptoms.

Patellar hypoplasia is most common (86%), followed by patellar aplasia (four to 20%), in patients with NPS [2]. Ligament laxity and knee instability may also occur, and this can lead to early degenerative arthritis, eventually requiring a total knee arthroplasty [1]. To our knowledge, there has been only one other case report that reported a NPS patient undergoing TKA [3]. Here, we report a case with a two-year follow-up of a NPS patient with severe osteoarthritis of the knee who underwent a current total knee arthroplasty.

2. Case report

The patient is a 46-year-old male who complained of chronic left knee pain that worsened with stairs and with increased activity. He had a long history of left knee pain since his childhood and had seen multiple physicians. The patient was limited in his

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activities of daily living due to pain and had exhausted non-operative management including injections, Nonsteroidal anti-inflammatory drugs (NSAIDs), and physical therapy.

Past medical history included scoliosis, and anxiety. Past surgical history included right wrist surgery and a pain stimulator placement for chronic low back radiculopathy pain.

On physical exam, the patient had a limited range of motion of his left knee with 0 degree of extension and 15 degrees of flexion. He reported that his range of motion of his left knee has been worsening over the years. Close inspection revealed a very small, laterally dislocated patella. In addition, quadriceps were atrophic and weak. The patient had good anterior–posterior stability of the knee joint but there was eight to 10 mm of varus/valgus laxity. The patient's right knee had excellent range of motion with 115 degrees of flexion to full extension, and he denied any complaints in his right knee.

Radiologic exam showed significant moderate to severe arthritic changes to the medial and lateral tibiofemoral compartments with joint space narrowing, joint subluxation, and osteophyte formation (Figures 1 and 2). There was evidence of femoral condylar hypoplasia and a dysplastic trochlea in the left knee along with the chronic lateral patellar dislocation, which can be seen on the sunrise view (Figure 2c). On AP pelvic radiograph, the patient had bilateral iliac wing horns (exostoses), although the left iliac horn was obscured by the patient's pain stimulator (Figure 3). Elbow radiographs were not obtained.

The patient underwent non-operative management of osteoarthritis of the knee for six months with minimal pain relief, and his poor range of motion and arthritic pain continued to affect his quality of life. The risks and the benefits of the surgery were explained, and the informed consent for total knee arthroplasty was obtained. He underwent a cemented total knee arthroplasty (Evolution, Microport, Inc. Arlington, TN) using a standard medial parapatellar approach. The senior author uses standard gap-balancing technique using flexion and extension block, and, at the completion of the procedure, the patient had excellent stability and alignment without requiring excessive external rotation of the femoral component. Due to the patient's small (approximately 2.5 cm in width) patella, it could not be resurfaced at the completion of the procedure. A lateral and proximal release was performed, and medial imbrication sutures were placed to bring the medial retinaculum over the lateral retinaculum holding the patella in the trochlear groove. Unfortunately, the patient developed arthrofibrosis of the operative knee and underwent a manipulation under anesthesia at eight weeks after the initial surgery. The patient recently followed up with the senior author for his two-year follow-up and was doing well with excellent range of motion (0–110°) and satisfactory radiographs (Figure 4). His preoperative Knee Society Score of 49 improved to 91 at his latest follow-up.

3. Discussion

NPS is caused by an autosomal dominant mutation in the *LMX1B* gene located at 9q34. The *LMXB1* gene encodes the LMX1B protein, a LIM-homeobox transcription factor, which regulates gene expression in the kidney and other organs [1]. The LMX1B protein is also active during development, specifically in the dorsal-ventral patterning of the anterior eye and the lower extremity [4]. In addition to these classical findings, patients with NPS are commonly of increased height and decreased body weight [5]. Patients display decreased muscle mass in the extremities, contributing to the tall and slender phenotype [1]. In addition, some patients report vasomotor sensations and paresthesia, as well as neurological symptoms including hypoalgesia and loss of temperature sensation [5]. Elbow anomalies include decreased range of motion, and cubitus valgus and dysplasia of the radial head also

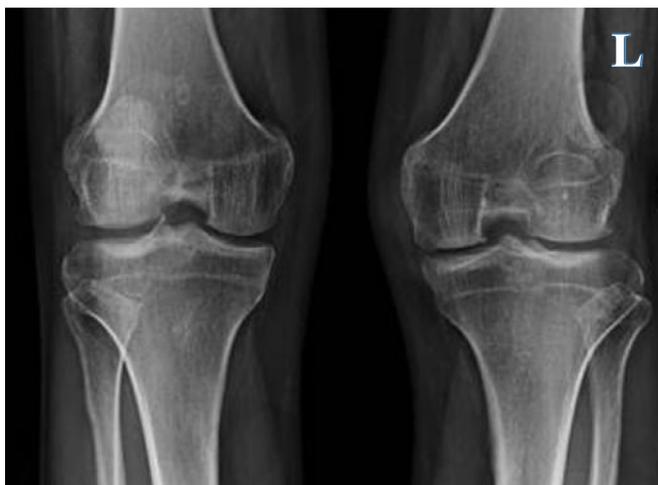


Figure 1. Weight-bearing AP radiograph of the patient with nail-patella syndrome. There is an evidence of femoral condylar hypoplasia and moderate to severe arthritic changes to medial and lateral tibiofemoral compartments of left knee.

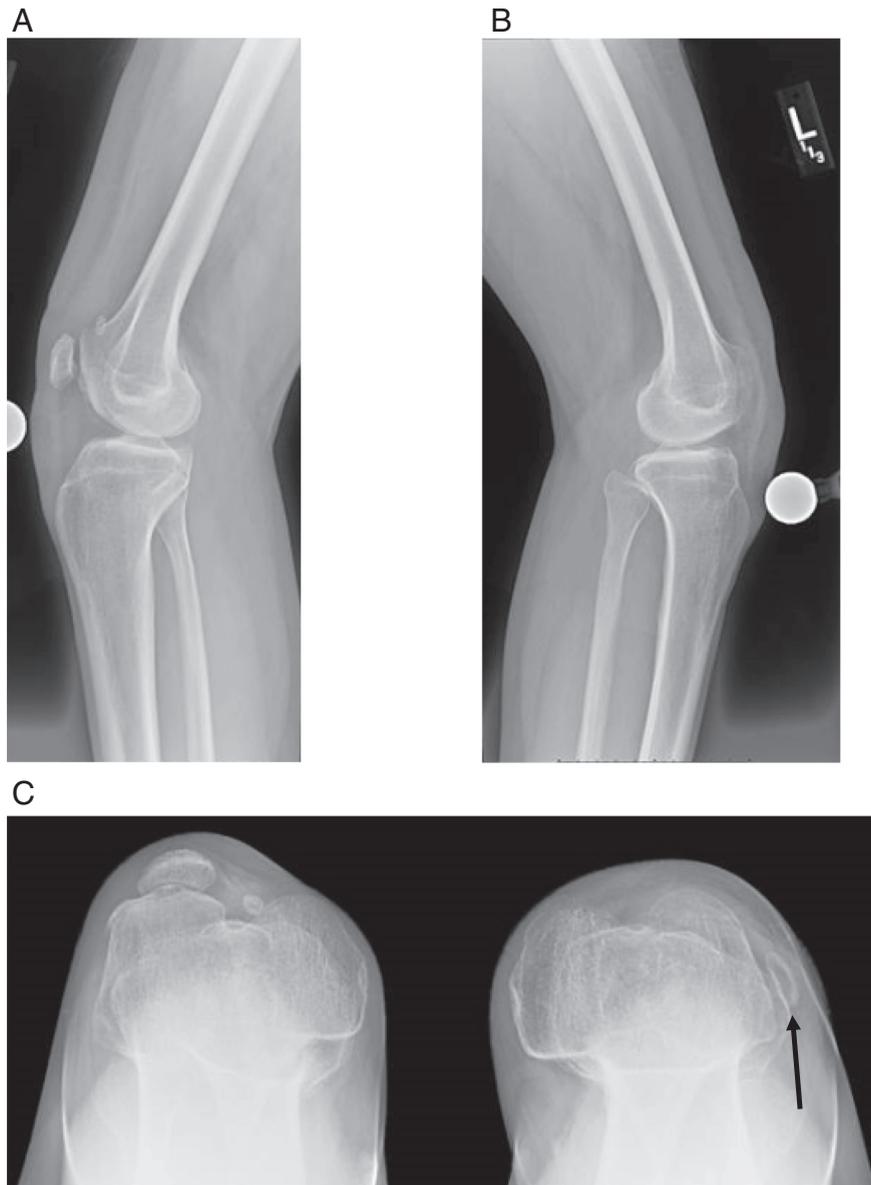


Figure 2. a, b, c. Bilateral standing lateral radiographs and sunrise view of the patient with nail-patella syndrome. There is obvious left chronic lateral patellar dislocation. The patella is marked by a black arrow.

occurs in nail-patella patients. Often, the radial head is dislocated posteriorly due to hypoplasia of the lateral epicondyle [1] (Figures 5 and 6).

Due to *LMX1B* gene activity in podocyte functioning and in the anatomy of the glomerulofiltration slits, patients with NPS can also develop glomerulonephritis with hematuria and proteinuria [6]. Often, the proteinuria and hematuria becomes more prevalent during pregnancy [7]. Glomerulonephritis occurs in 30–50% of nail-patella patients and ESRD occurs in five to 10% of patients [5]. Due to the mutated *LMXB1* gene in nail-patella patients, they are at higher risk for open-angle glaucoma and intra-ocular hypertension [8]. Lastly, there is an increased risk of attention deficit-hyperactivity disorder (ADHD) and major depressive disorder in nail-patella patients. These neurologic deficits have been attributed to the *LMX1B* protein functions in the development of mesencephalic dopaminergic signaling, as well as the serotonergic signaling system [9].

While total knee arthroplasty is a relatively frequent procedure for the general population, a total knee arthroplasty done on a NPS patient is quite unique due to the rarity of the condition. Because of the anatomic abnormalities of the knee in the syndrome, nail-patella patients experience arthritic changes that are severely limiting and painful, a strong indication for a total knee arthroplasty. The only other case report in literature is by Lachiewicz et al. who reported an excellent outcome using a

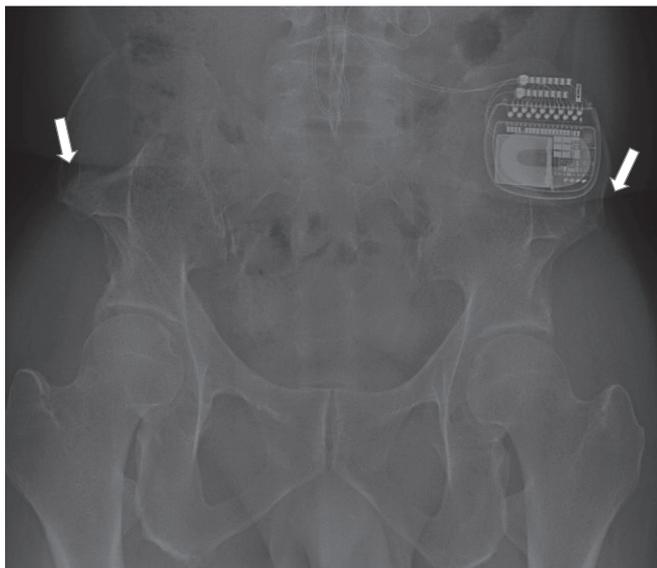


Figure 3. AP pelvis radiograph of the patient who has bilateral iliac wing exostoses/horns (marked by white arrows). Left iliac horn is obscured by the patient's pain stimulator.

posterior-stabilized cemented total knee arthroplasty [3]. Similar to our case, the authors reported that they were not able to re-surface the patella due to hypoplasia. They also reported that partial release of medial collateral ligament was required due to the fixed varus deformity of the knee, which was not required in our case.



Figure 4. The patient's left iliac horn is indicated by a black arrow.

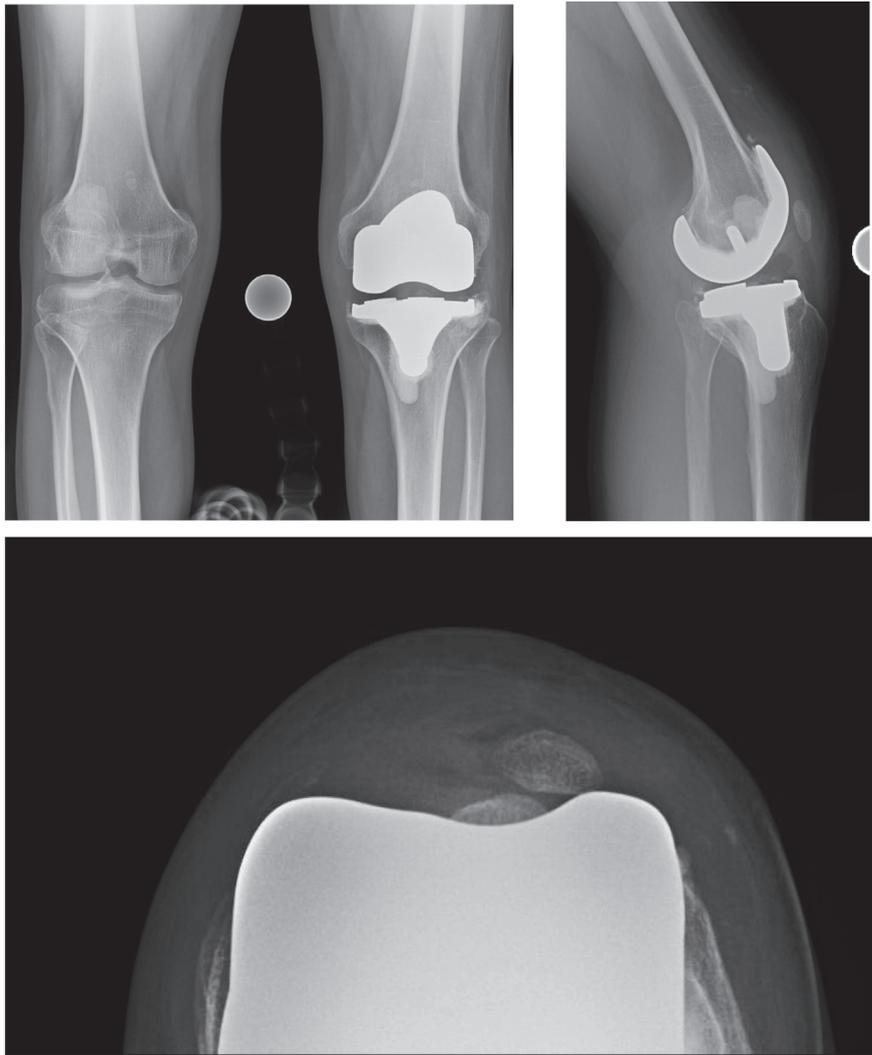


Figure 5. The patient is two years removed from left total knee arthroplasty and is doing well.

It is unclear whether or not NPS patients have genetic predisposition to develop arthrofibrosis after TKA, however, we recognize that the patient had several risk factors such as poor preoperative range of motion (0–15°) and history of smoking, which have been previously reported as risk factors [10]. In addition, the soft tissue release required to realign patella may have contributed to this complication as well.

In patients with trochlea hypoplasia and/or chronic dislocation of the patella, it may not be possible to resurface or completely realign the patella in the trochlear groove; however, the procedure can still improve the patient's quality of life. We recognize that patellar tracking is a challenging issue in these patients, and there is only a paucity of data regarding patellar management in patients with NPS. In our experience, we suggest that medial imbrication of retinaculum should be performed as well as a lateral release in order to optimize patellar tracking. We also recommend that the patella should be resurfaced if the surgeon can adequately resurface the patella with at least 12 mm of patellar thickness after resurfacing.

Declaration of interest

Maile E Curbo, Kwan J. Park, and Landon D. Brown have no declaration of interest
 Stephen J. Incavo has following declaration of interest:
 Stocks/Equity/Ownership Interests: Nimbic Systems
 Intellectual Property Rights: Innomed, MicroPort Orthopedics
 Consulting: Biomet, Kyocera Medical Corporation, Smith & Nephew
 Editorial Board: Journal of Arthroplasty, the Knee



Figure 6. Clinical photo of the patient's post-operative knee. The patella is just distal to the patient's hypoplastic left index finger nail, and it measures approximately 2.5 cm in width, indicated by a black arrow.

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