NAIL-PATELLA SYNDROME: EVOLUTION OF PATTELAR INSTABILITY

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SUMMARY

The nail-patella syndrome is an autosomal dominant disease with some dermatological, musculoskeletal and, occasionally, renal, ocular and gastrointestinal classical characteristics. Its main clinical orthopaedic symptom is patellofemoral pain associated with patellar instability since early childhood. The best way to achieve good results in such cases is to establish an early treatment for knee instability, otherwise it may lead to early arthrosis and functional limitation of the knee joint. The present case describes a patient with such syndrome who underwent late surgical treatment and evolved with joint degeneration and functional limitation of the knee. The objective of this study is to consider the syndromic phenotypic features of the disease, correlate them with orthopaedic complaints commonly reported to the physician, such as pain and instability of the knee joint and maybe of the elbow joint and, finally, be able to provide an early treatment for symptoms in order to ensure a favorable evolution to the patient.

Keywords: *Nail-patella syndrome; Patella; Joint instability/therapy; Joint instability/surgery.*

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INTRODUCTION

The nail-patella syndrome is a rare disease of autosomal dominant nature[1,2,3]. It is associated to musculoskeletal, dermatologic (nail dystrophy), renal, eye and gastrointestinal changes[1,2].

The most frequent orthopaedic complaint of these patients is patellar instability associated with pain and functional restraint. In general, in this congenital patellar change, symptoms onset only occurs when gait starts[4]. Such deformity may course with a very painful picture and, if left untreated, may evolve to early patellofemoral arthrosis.

The authors present a patient carrying this syndrome and showing a number of the characteristics described, as well as her evolution 30 years after her first surgery, and 18 months after the last procedure. This pathology's rare nature, the difficult diagnosis and therapy approach are discussed here, so that surgeons can become aware of this pathology and its eventual limited results.

CASE REPORT

A Caucasian, 46 year-old, 1.59 tall, 50-kg woman sought the Knee Group of Hospital das Clinica's Orthopaedic Service in the year of 2002 because of bilateral knee pain, worse at the right side, as well as functional disability. She told that pain had strongly worsened in the previous 12 months, after a direct trauma on the right knee.

At that time, the patient has shown to be anxious and depressed. She reported generalized pain on wrists' joints, elbows, cervical spine and knees, especially at the right side, key reason of her complaints. She was wearing crutches for pain relief. As a major history item, she had been submitted to a bilateral procedure to realign the patella (at the age of 14) due to instability experienced since her childhood.

At physical examination, she presented strong pain at palpation of the anterior surface of the right patella with no local phogistic signs, no instability or patellar dislocation recurrence signs. The active range of motion on the knees was limited from 10 to 90 degrees, with bilateral quadriceps hypotrophy, reduced muscular strength in both lower limbs. She also showed some limitation of the active and passive elbow's range of motion (15 degrees of extension deficit, total bilateral pronation-supination of 80 degrees), feet and hand nails hypotrophy, especially on toes and thumbs (Figures 1-4). The Nail-Patella Syndrome was proven by clinical signs and X-ray images, evidencing dystrophy of the first fingers of the hands and feet, bilateral patellar hypoplasia with subtle bilateral arthrosis at femoropatellar compartment, hypotrophy of the lateral condyle at elbows and iliac horns (Figures 5-9).

As there was a disproportional knee pain upon reported trauma, sympathetic-reflex dystrophy (resulting from trauma) was also suspected, which was confirmed by bone scintiscanning. Therefore, she was referred to Physiatrics for follow-up.

Additionally to the Sympathetic-Reflex Dystrophy, she presented the following clinical co-morbidities: hyperthyroid-

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ism, depression, fibromyalgia and epigastralgia. She was using chlorpromazin Hcl, codeine, proptyltiouracyl, amitriptiline, atenolol, levomepromazin maleate, diazepam, and omeprazol.

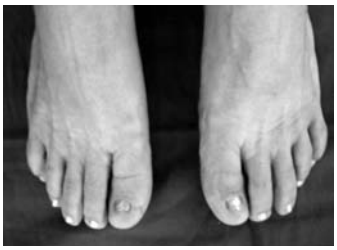

Figure 1 - *Hallux nail dystrophy*

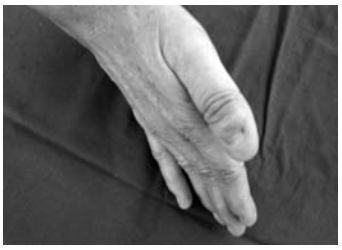

Figure 2 - *Thumb nail dystrophy*

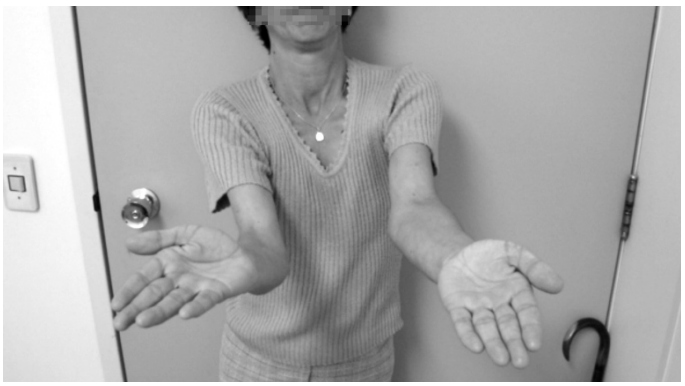

Figure 3 - *Elbow supination limit.*

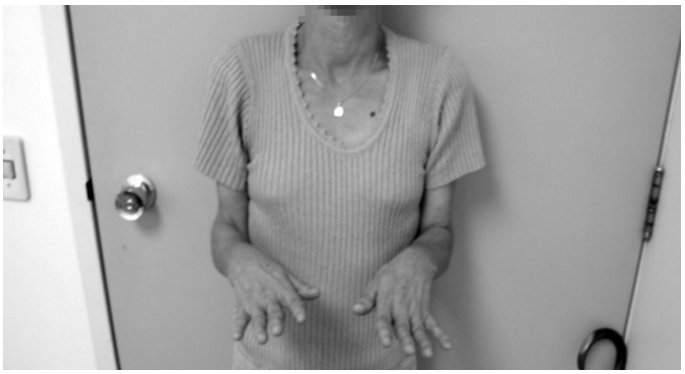

Figure 4 - *Elbow pronation limit.*

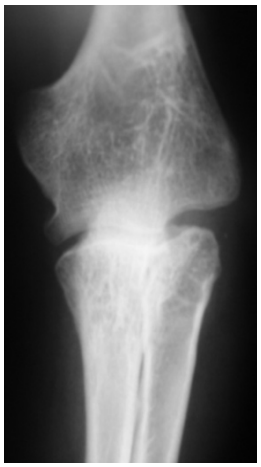

Figure 5 - *Elbow AP plane: Lateral condyle and radius head hypoplasia.*

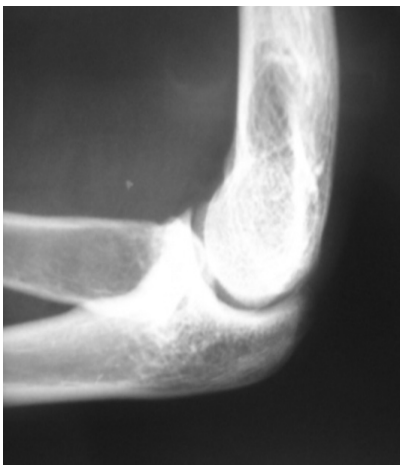

Figure 6 - *Lateral plane - radius head hypoplasia.*

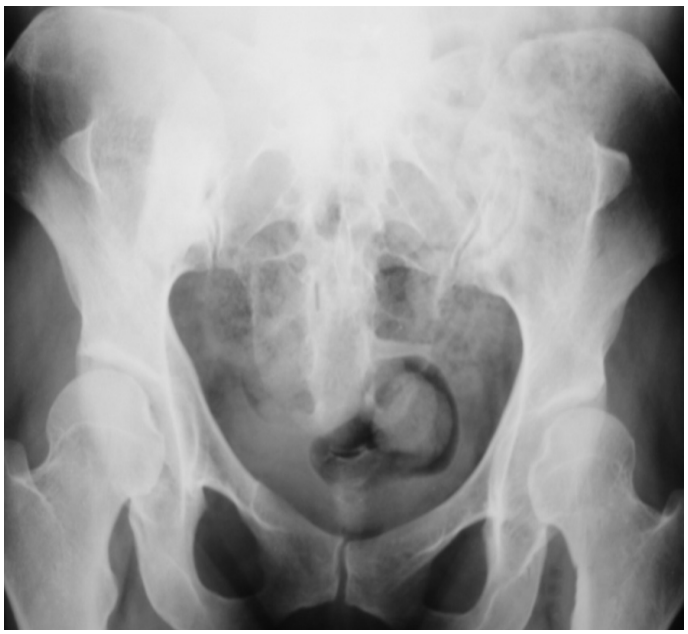

Figure 7 - *AP plane of the hip: iliac horns.*

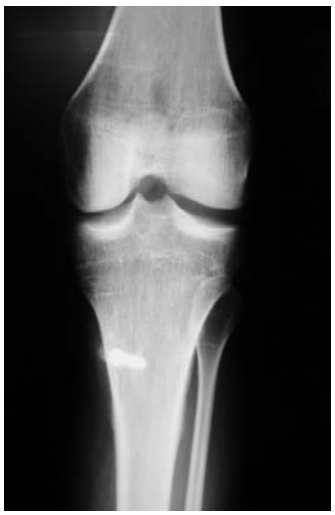

Figure 8 - *Left Knee AP plane: Left patella hypoplasia.*

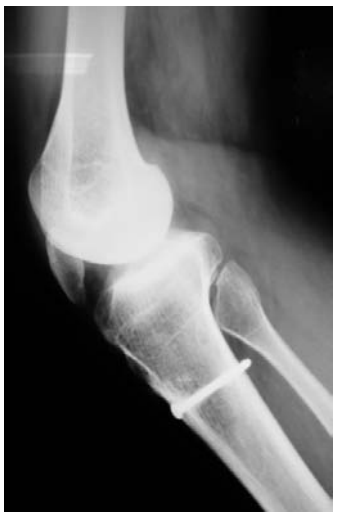

Figure 9 - *Left knee lateral plane: Patellar hypoplasia.*

As the patient continued feeling strong pain at the right knee throughout the whole treatment at Physiatrics, a new assessment was requested to the Knee Group in 2004. With the physical examination showing slow and very painful flexion-extension movement of the knee associated to subdislocation of the right patella and X-ray images evidencing strong femoro-patellar arthrosis at the right side, patellectomy was indicated to relieve the pain at the right knee. The procedure was performed in April 28th, 2004.

Now, 18 months after surgery (Figures 10-12), this patient is not working yet and fells pain on both knees, with no renal or ophthalmic changes. She keeps using drugs for depression, hyperthyroidism, fibromyalgia and arthrosis.

DISCUSSION

The nail-patella syndrome is a rare disease of dominant autosomal nature, resultant from a change of the LMX1B gene[1,2,3]. It is associated to skin changes, such as: nails

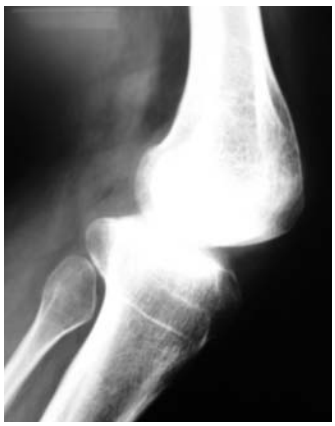

Figure 10 - *Right knee lateral plane: post-patellectomy*

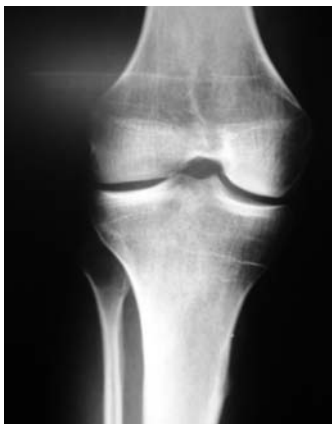

Figure 11 - *AP: Right knee after patellectomy*

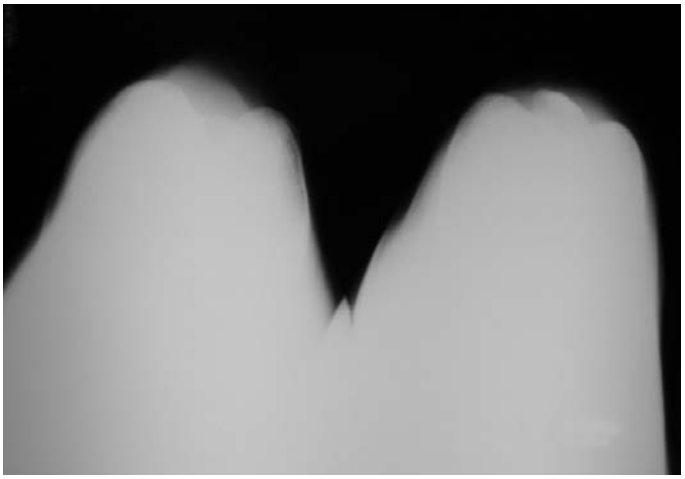

Figure 12 - *Left side: patellar hypoplasia.*

dystrophy; musculoskeletal changes, such as hypoplasia or absence of patella, radius head dislocation or hypoplasia of elbows' condyles, iliac horns evidenced on X-ray images[1-4]. Furthermore, this syndrome may present renal (focal glomerulosclerosis), eye (glaucoma) changes, as well as gastrointestinal symptoms[1,2] .

The most important orthopaedic complaint of these patients is patellar instability associated to pain and functional restraint. In general, in this congenital patellar change, the onset of symptoms only occurs when gait starts[4]. The fixed lateral instability is mainly due to patellar hypoplasia associated to shallow trochlear groove, lateral structures contraction, with thinner medial capsule and retinaculum. Patients can evolve with contraction in flexion, quadriccipital atrophy, genus valgus, and external rotation of the tibia[5]. Such deformity can course with a very painful picture and, if left untreated, it may evolve to early femoropatellar arthrosis.

Some authors recommend follow-up only in patients with congenital patellar dislocation. They believe that the natural history of this deformity is benign, the knee is often painless and surgical results are poor[5]. Other authors recommend patellar realignment during childhood (as early as eight years old) and suggest that surgical procedures in older children show worse results, evolving with progressive worsening of symptoms, quadriceps weakness, as well as functional restraints[5,6].

In patellar realignment surgeries, the principle consists of releasing lateral soft parts (vastus lateralis, retinaculum lateralis, contracted synovia) associated to a reinforcement folding on medial retinaculum and oblique vastus medialis, leaving an open lateral capsule and wrinkled medial capsule. Patellar ligament is thereby achieved, stimulating the development of a functional trochlear groove. Occasionally, a stretching on the myotendinous unit of the quadriceps is required, and, if severe contraction occurs at flexion, posterior release is also needed. In older patients, increasing medial vector strength is necessary, so a transfer of the semitendinous tendon by a tunnel in the patella can be made. Skeletically immature children are submitted to medial transfer of the patellar tendon. The older ones (those skeletically mature) are submitted to medial transfer of the tibial trabecule by means of osteotomy. Patellectomy is recommended in cases showing poor evolution in the long-term, with strong femoropatellar arthrosis, pain and functional disability[5,6].

In the present case report, the patient had always had a painful and functional disability picture on the knees - especially at right knee - despite of her psychic component. She carries the nail-patella syndrome, which, as previously noted, is strongly associated to congenital patellar dislocation and, since her childhood, presented instability symptoms. She was submitted to patellar realignment at a late phase (at the age of 14), which, as reported by literature, tend to provide poor outcomes due to the delayed intervention. The patient presented a poor evolution, with progressive functional restraint, severe pain, and, as expected, severe femoropatellar arthrosis. At 45 years old, as a salvage alternative, she was submitted to total patellectomy on her right knee. The patient has a reduced range of motion on the elbows (pronation-supination and flexion-extension deficit) by lateral condyle and radius head change. No complaints regarding nails and hips. No ocular or renal complaints, as reported by some studies addressing the syndrome.

The importance of the reported case is to drive focus to the early diagnosis of the nail-patella syndrome in children coursing with patellar instability, optimizing an effective treatment (either surgical or not). For arthrosis cases in adults, it is important to be aware of the unfavorable clinical picture evolution.

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