**CASE REPORT**

**Correction of malformative patellar instability in patients with nail-patella syndrome: A case report and review of the literature**

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**KEYWORDS**
Nail-patella syndrome; Malformative patellar instability; Operative stabilization; Myotome

**Summary**
Nail-patella syndrome (NPS) or hereditary onycho-osteodysplasia is a relatively rare autosomal dominant disorder with the classic tetrad of fingernail abnormalities, hypoplastic patellae, radial head dislocation and iliac horns. The anatomic abnormalities in NPS often lead to subluxation or dislocation of the patellae causing knee instability and pain. Although most existing literature regarding the knee manifestation of this syndrome has focused on the clinically and radiological changes, only a few articles discussed the surgical treatment. This study reports the clinical, radiological and arthroscopical findings and a 24-month follow-up after operative stabilisation considering the underlying pathomorphology of malformative patellar instability in an 11-year-old girl. The findings of this study confirm the unique pathology of NPS with a synovial band preventing the engagement of the patella into the trochlear groove. NPS is a rare disorder and has to be considered in cases with untypical patella dislocation. The underlying pathology differs completely from patients with patellofemoral instability. The aim of orthopaedic surgery should be correction of the underlying pathology with resection of the synovial band and an additional realignment of the patella by recentering of the quadriceps muscle. Considering the underlying pathology good clinical results can be expected.

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**Background**

Nail-patella syndrome (NPS) or hereditary onycho-osteodysplasia is a relatively rare autosomal dominant disorder with the classic tetrad of fingernail abnormalities, hypoplastic patellae, radial head dislocation and iliac horns.
horns [1]. It has been first described in 1820 by Chatelain
[2].

The gene involved is the distal portion of the long arm of chromosome 9 in the 9q34.1 locus, close to the AB0 blood group locus. The main cause of the disease is the LMX1B mutation [3,4].

The anatomic abnormalities in NPS often lead to subluxation or dislocation of the patellae causing knee instability and pain. Most existing literature regarding the knee manifestation of this syndrome has focused on the clinical and radiological changes, however, only a few articles discussed the surgical treatment [1,2,5—7]. There is still no consensus concerning the most effective operative strategy.

This study reports the clinical, radiological and arthroscopical findings and a 24-month follow-up after operative stabilisation considering the underlying pathomorphology of a malformative patellar instability as proposed by Bensahel et al. [8] in an 11-year-old girl.

Case report

An 11-year-old girl presented with bilateral patella dislocations with left knee pain. She had no complaints of pain in her elbows or hands. Her birth history was not significant and she had no other medical or surgical history.

Her mother described that many family members have problems with their nails or with their peculiar knees (Fig. 1). Furthermore, the girl’s grandfather had an elbow surgery due to extension deficit.

The older brother has dysmorphic fingernails, two sisters (siblings) show no noticeable features.

Physical examination showed an 11-year old girl of 131,3cm height (below the third percentile) and 25kg weight (below the third percentile). Both thumbnails were flat and paper-thin but only the left thumbnail was grooved. All finger- and toenails were paper-thin (Fig. 2). An extension deficit of 30° of the right elbow and 10° of the left elbow was present. Bilaterally, the calf muscles were not well developed and flat feet revealed.

Her spine and hip examinations showed no remarkable features. The thigh foot angle was normal. Leg lengths were equal, no valgus or varus deformity occurred.

Both knees exhibited a range of motion from 0° to 130°. Bilaterally, the patella dislocated laterally at knee flexion of 30° and showed progressive lateral dislocation with increasing flexion. No pain occurred during flexion and extension examination.

Internal and neurological examinations were unremarkable.

Radiographs of the knees (Fig. 3A), the pelvis, the elbows and the left hand were taken. Both patellae were subluxed on the 30° axial views (Fig. 3C). On the lateral radiographs, no sign for trochlear dysplasia could be detected. The radiograph of the left hand revealed a retarded bone age of 10 years. The elbows and the pelvic radiographs showed no typical abnormalities of NPS.

The MRI of both knees confirmed a thick soft tissue band dividing the joint in medial and lateral compartments (Fig. 4).

Surgical treatment

Surgery took place in the supine position under general anesthesia. Arthroscopic examination through a standard anterolateral incision revealed a midline synovial septum...
dividing the trochlea into a medial and a lateral part. The septum extended from the anterior to the superior recessus (Fig. 5). The medial and lateral tibio-femoral compartments showed no pathologies.

Figure 3  The a.p. radiograph before the operation shows the slightly lateralized patella in knee extension (A). On the 30° axial view, the patella showed a lateral subluxation (C). After surgery, the patella was centered (B).

The septum was arthroscopically removed. The band had cut a deep groove into the trochlea with visible cartilage damage (Fig. 6). Although the septum could be totally removed the realignment of the patella even under manual medialization failed. For this reason an open surgery followed.

Figure 4  The axial MRI view of the left knee confirmed a thick soft tissue band (white arrow) dividing the joint in a medial and a lateral compartment.

Figure 5  Arthroscopic examination of the left knee revealed a midline synovial septum (white arrow) extending from the intercondylar notch to the superior recessus.
On follow-up 2 years postoperatively, the knee was stable in each position, passive and active range of motion was 130°–0°–0° for flexion-extension. The patient has returned to full activities. The radiograph of the left knee confirmed the recentered patella after surgery (Fig. 3B). In the right knee the nonoperative management was continued as the girl was pain free.

**Discussion**

NPS is a rare disorder typically leading to patella dislocation. The clinical aspect is different to patients with conventional patellofemoral instability. Whereas patients with patellofemoral instability show positive apprehension sign and relocation of the patella between full extension and 30° of flexion, patients with NPS show progressive lateral dislocation with increasing flexion. The reason can be seen in the different morphology of the femoral trochlea in patients with NPS. First the trochlea is not dysplastic but well formed. The intra-articular reason for the dislocation of the patella is the midline synovial septum, which divides the knee into a medial and a lateral part. This septum cuts a deep groove into the trochlea and prevents the patella from sinking into the trochlear groove (Figs. 5 and 6).

Secondly failure of internal rotation of the myotome, which contains the quadriceps muscle and the patella, is considered to be a major reason for dislocation [6,10,11].

In literature there are only few reports with different reported approaches, thus operative correction of patella dislocation in NPS still is a controversial issue (Table 1).

Guidera et al. published a review of 44 orthopaedic patients with NPS. Ten of the 44 patients underwent knee surgery to realign the patella. They report poor results after proximal realignment and favourable outcome after combined proximal and distal realignment [2].

Beguiristain et al. performed the Krogius-Lécène procedure (proximal soft-tissue procedure) in four patients aged between three and five years with NPS with good long-term results [3].

Yakish and Fu have reported good results after medial reconfiguring and lateral release in children with NPS [7].

Marumo et al. performed a Stanisavljevic procedure (medial rotation of the quadriceps, Z-lengthening of the rectus femoris and medialization of the tibial tuberosity) in a 26-year-old woman, which resulted in an improved gait, but a 30°-extension deficit remained [6]. Niemeyer et al. suggested that the occurrence of synovial plicae might account for at least part of the characteristic symptoms of NPS and recommend an early resection of these plicae [12].

Doughty et al. suggested an early intervention whenever the syndrome is recognized and reported about good short-term-results on an 11-year-old boy treated with arthroscopic retrograde lateral release with medial reefing [1].

In contrast to Doughty et al. in this case isolated resection of the midline synovial septum was not sufficient to relocate the patella. Therefore an extensive soft-tissue realignment was necessary. This can be explained by the fact, that additionally failure of internal rotation of the myotome with consecutive lateralization of the quadriceps muscle is present in patients with NPS and patella dislocation [6,10,11].

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**Figure 6** After the resection of the midline synovial septum in the left knee the deep trochlear groove (red arrow) with cartilage damage (white arrow) is visible.

The first step involved a distinctive soft-tissue release of the lateral knee capsule and the lateral retinaculum. Then a release of the quadriceps tendon followed to allow recentering of the lateralized quadriceps muscle. For distal realignment the patellar tendon was split vertically. The lateral half was pulled under the medial half and attached to the pes anserinus (Goldthwait procedure). Because of continuous patella dislocation the vastus medialis was extended distally and laterally. Furthermore, a reconstruction of the medial patellofemoral ligament sparing the distal femoral physis according to the technique described by Nelitz et al. [9] and a lengthening of the lateral retinaculum followed. The medial border of the patella was exposed subperiosteally. Two converging V-shaped tunnels were drilled to the depth of about 1.5 cm at the superomedial half of the patella. The gracilis tendon was harvested proximally with a tendon stripper. Distally the tendon was sharply detached from the tibia and then inserted into the tunnel, forming a loop through the patella. After verification of the femoral insertion, a guide pin was drilled to the lateral condyle distal to the physis. A medial blind tunnel was drilled along the guide pin. The graft was then pulled between the second and third capsular layer to the femoral insertion point.

A locking suture was passed through the transepicondylar axis to pull the graft into the medial tunnel. A bioresorbable interference screw was then used to secure the graft within the medial condyle tunnel with the knee flexed to 30° [9].

Intraoperatively the patella remained centered in the trochlear groove up to full flexion.

Exercising started from the first postoperative day with passive and active mobilization of the left knee. A range of motion knee brace was prescribed with limitation of knee flexion at 90° to avoid suture rupture for the first six weeks. Full weight bearing was allowed on extension. Wound healing was uneventful.

**Outcome**

Postoperatively passive knee flexion was possible up to 130°.

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### Table 1: Summary of studies reporting about NPS.

<table>
<thead>
<tr>
<th>Author (Year)</th>
<th>Study design</th>
<th>No. of cases</th>
<th>Age at surgery (y)</th>
<th>Gender</th>
<th>Surgery</th>
<th>Follow-up (y)</th>
<th>Results</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yakish and Fu (1993) [7]</td>
<td>Case report</td>
<td>1</td>
<td>3-10</td>
<td>Male</td>
<td>Quadricepsplasty</td>
<td>6,5</td>
<td>Poor results after proximal realignment; favourable outcome after combined proximal and distal realignment</td>
</tr>
<tr>
<td>Marumo et al. (1999) [6]</td>
<td>Case report</td>
<td>1</td>
<td>26</td>
<td>Female</td>
<td>Realignment of the patella</td>
<td>3,5</td>
<td>Excellent results after proximal realignment; favourable outcome after combined proximal and distal realignment</td>
</tr>
<tr>
<td>Buchler et al. (2003) [1]</td>
<td>Case report</td>
<td>1</td>
<td>11</td>
<td>Male</td>
<td>Kropp-Lecène procedure</td>
<td>3,5</td>
<td>Excellent results after quadricepsplasty</td>
</tr>
<tr>
<td>Niemeyer et al. (2006) [12]</td>
<td>Case report</td>
<td>1</td>
<td>40</td>
<td>Male</td>
<td>Arthroscopic retrogradelateral release with medial reforeeing</td>
<td>&lt;1</td>
<td>Good short-term results</td>
</tr>
<tr>
<td>Guidera et al. (1991) [2]</td>
<td>Review</td>
<td>10/44</td>
<td>3-10</td>
<td>Female/male</td>
<td>Realignment of the patella</td>
<td>3,5</td>
<td>Excellent results after proximal realignment; favourable outcome after combined proximal and distal realignment</td>
</tr>
<tr>
<td>Heidenhain et al. (2012) [13]</td>
<td>Case report</td>
<td>1</td>
<td>18</td>
<td>Male</td>
<td>Resection of the septum</td>
<td>3,5</td>
<td>Successful results after quadricepsplasty</td>
</tr>
<tr>
<td>Nagi et al. (1998) [3]</td>
<td>Case reports</td>
<td>4</td>
<td>26 (24-28)</td>
<td>Male</td>
<td>Resection of the plica</td>
<td>&lt;1</td>
<td>Poor results after early resection of the plica</td>
</tr>
</tbody>
</table>

y: years; mths: months.
In addition, Heckman et al. [13] described one of two cases with NPS with a midline synovial septum having been recurred even after it had been resected at the initial surgery. The findings of the present study confirm the unique pathology of NPS with a synovial band preventing the engagement of the patella into the trochlear groove. As NPS is a congenital syndrome concomitant deformity with contracture of the lateral soft tissues is secondary. In literature an early resection of the synovial band is recommended to avoid longterm morbidity [1,12].

Conclusion

NPS is a rare disorder and has to be considered in cases with untypical patella dislocation. Surgical treatment is recommended to avoid early arthritis. The aim of orthopaedic surgery should be the correction of the underlying pathology. Since resection of the synovial band is only a part of the deformity additional realignment of the patella by recentering of the quadriceps muscle is mandatory. Considering the underlying pathology good clinical results can be expected.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

References


