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

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KEYWORDS
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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 radiologically 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 centering of the quadriceps muscle. Considering the underlying pathology good clinical results can be expected.

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 [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 ABO 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 131,3 cm height (below the third percentile) and 25 kg weight (below the third percentile). Both thumbs 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.

![Figure 1](image1.jpg) **Figure 1** The preoperative picture shows an peculiar left knee anatomy with an ‘‘empty’’ trochlear groove.

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

![Figure 2](image2.jpg) **Figure 2** One of the characteristics of the nail-patella-syndrom are nail abnormalities: the finger- (A) and toenails (B) are paper-thin.
Malformative patellar instability in patients with nail-patella syndrome

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

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.

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 dunking 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 Kroglius-Lecz`ene 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 reefing 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 realignement 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].

Outcome

Postoperatively passive knee flexion was possible up to 130°.
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 (1983)</td>
<td>Case reports</td>
<td>4</td>
<td>3–10</td>
<td>3 female/1 male</td>
<td>Quadricepsplasty</td>
<td>10</td>
<td>Successful results</td>
</tr>
<tr>
<td>Guidera et al. (1991)</td>
<td>Review</td>
<td>10/44</td>
<td>3.5 mths at diagnosis</td>
<td>Female/male</td>
<td>Realignment of the patella</td>
<td>6,5</td>
<td>Poor results after proximal realignment; favourable outcome after combined proximal and distal realignment Improved gait, 30° extension deficit Excellent</td>
</tr>
<tr>
<td>Marumo et al. (1999)</td>
<td>Case report</td>
<td>1</td>
<td>26</td>
<td>Female</td>
<td>Stanisavljevic procedure</td>
<td>4</td>
<td>Improved gait, 30° extension deficit</td>
</tr>
<tr>
<td>Doughty et al. (2005)</td>
<td>Case report</td>
<td>1</td>
<td>11</td>
<td>Male</td>
<td>Arthroscopic retrograde lateral release with medial reefing</td>
<td>&lt;1</td>
<td>Good short-term-results</td>
</tr>
<tr>
<td>Niemeyer et al. (2006)</td>
<td>Case report</td>
<td>1</td>
<td>40</td>
<td>Male</td>
<td>Early resection of the plicae</td>
<td>&lt;1</td>
<td>Good short-term-results</td>
</tr>
<tr>
<td>Heckman et al. (2012)</td>
<td>Case report</td>
<td>2</td>
<td>18 47</td>
<td>Male</td>
<td>Resection of the septum</td>
<td>3,5</td>
<td>1 patient required two arthroscopies</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


