Bilateral Clubfoot in Nail-Patella Syndrome: A Rare Syndromic Case Successfully Treated with the Ponseti Method

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Learning Point of the Article:

Syndromic clubfoot associated with Nail-Patella Syndrome should not be presumed rigid or resistant. With early diagnosis and timely intervention, excellent outcomes can be achieved through conservative treatment using the Ponseti method.

Abstract

Introduction: Nail-patella syndrome (NPS) is a rare autosomal dominant disorder characterized by nail dysplasia, hypoplastic or absent patellae, elbow anomalies, and iliac horns. Its association with congenital talipes equinovarus, or clubfoot, is extremely uncommon and rarely documented.

Case Report: We describe a 1.5-year-old male child, born of a 2° consanguineous marriage, who presented with bilateral clubfoot and complete absence of fingernails. Great toenails were absent bilaterally, whereas the remaining toenails were preserved. The patellae were non-palpable clinically, and ultrasound confirmed the absence of ossification centers bilaterally. Radiographs revealed bilateral iliac horns. Ponseti casting was initiated at 2 months of age, and full correction was achieved after five casts, followed by bilateral percutaneous Achilles tenotomy. At 6-month follow-up, the patient exhibited plantigrade, pain-free feet with good dorsiflexion and no recurrence.

Conclusion: NPS can occasionally be associated with flexible clubfoot, which may respond well to conservative management. The Ponseti method, when initiated early, remains an effective option in such rare syndromic presentations.

Keywords: Nail-patella syndrome, clubfoot, Ponseti method, congenital talipes equinovarus, syndromic clubfoot, LMX1B mutation.

Introduction

Nail-patella syndrome (NPS), also known as hereditary onycho-osteodysplasia, is a rare autosomal dominant condition caused by mutations in the LMX1B gene on chromosome 9. This gene plays a crucial role in dorsal limb patterning, nephron development, and anterior segment formation in the eye [1]. NPS is characterized by classical musculoskeletal features, including nail dysplasia, hypoplastic or aplastic patellae, elbow anomalies, and pathognomonic iliac horns on pelvic radiographs [2]. Renal involvement and open-angle glaucoma may also be seen in some patients.

Congenital talipes equinovarus (CTEV), or clubfoot, is a congenital foot deformity characterized by hindfoot equinus and varus, forefoot adduction, and cavus. It occurs in approximately 1–7/1000 live births [3]. While most cases are idiopathic, about 20% are syndromic or associated with underlying neuromuscular conditions [4]. The association between NPS and CTEV is rare, with only a few cases described. The Ponseti method has become the gold standard in the management of CTEV, including many syndromic variants.

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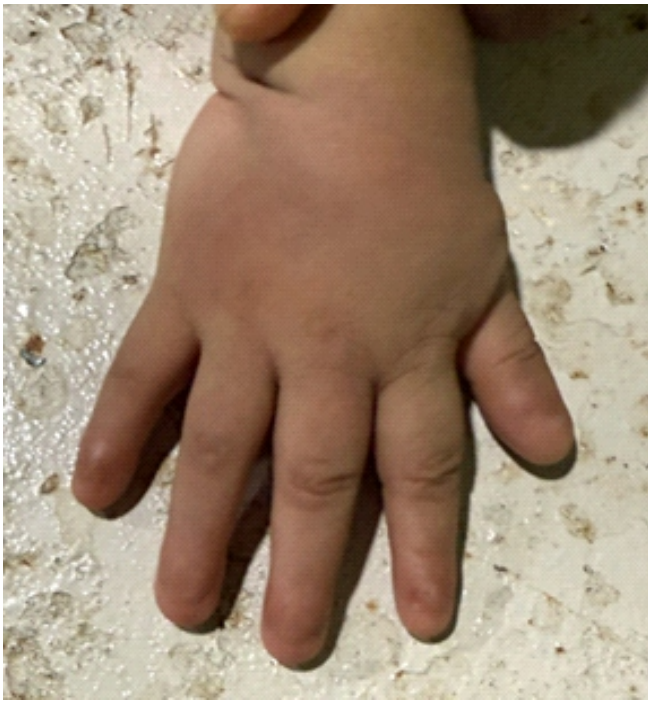

Figure 1: Right hand dorsal aspect.

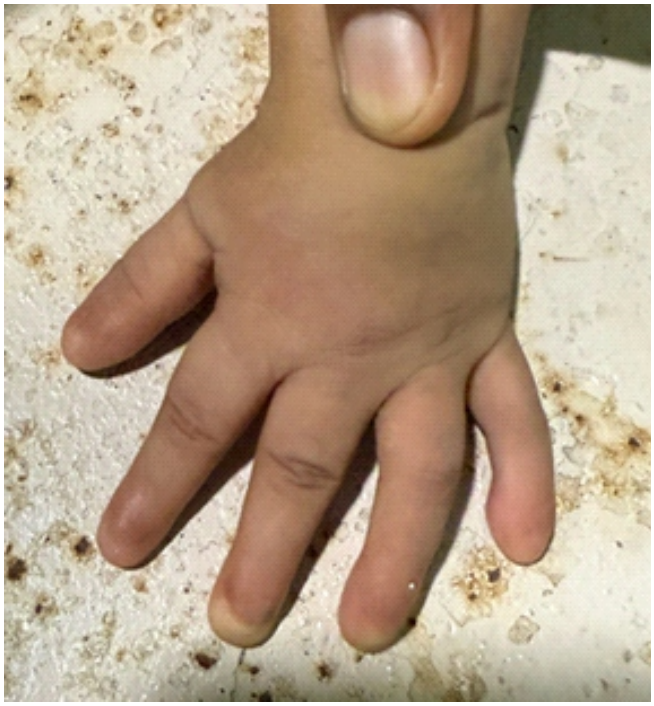

Figure 2: Left hand dorsal aspect.

Case Report

A 1.5-year-old male child was brought to our clinic with bilateral clubfoot noted since birth. He was the third child of healthy parents in a 2° consanguineous marriage. The elder siblings were developmentally normal. The antenatal and birth history were uneventful.

On examination, the child had bilateral clubfoot with equinus, hindfoot varus, forefoot adduction, and mild cavus deformity. The pre-casting Pirani Scoring was 5 in the right leg and 5.5 in the left leg. All fingernails were absent on both hands (Fig. 1 and 2). Toenails were present on all toes except the great toes bilaterally, which were anonychic (Fig. 3 and 4). The patellae

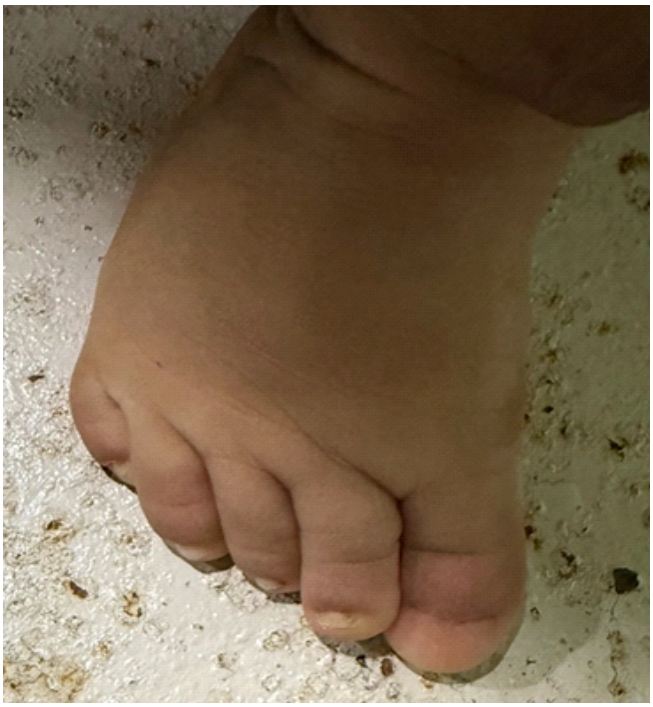

Figure 3: Right foot dorsal aspect.

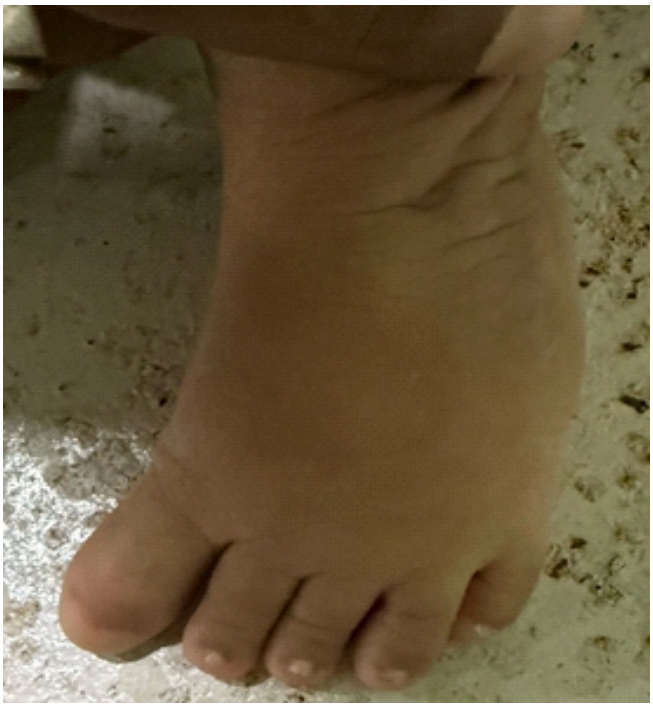

Figure 4: Left foot dorsal aspect.

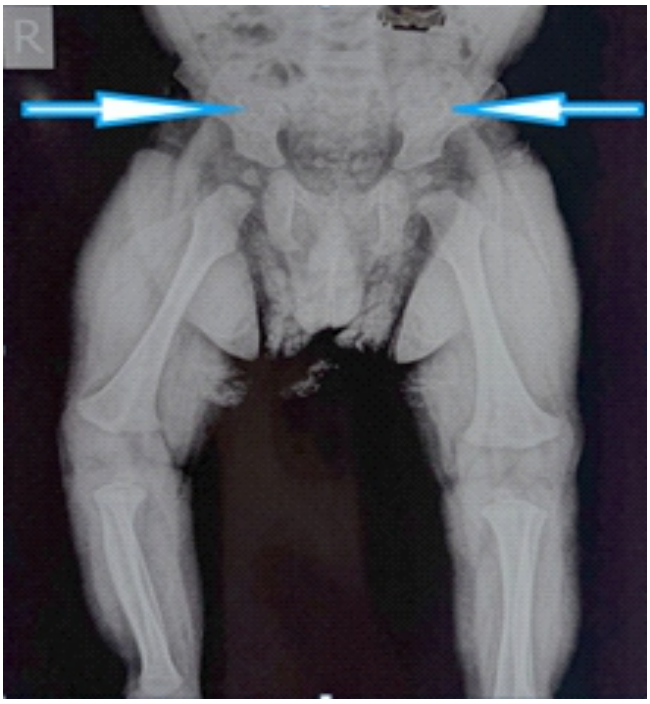

Figure 5: X-ray pelvis with both hips – anteroposterior with iliac horns. Marked with an arrow.

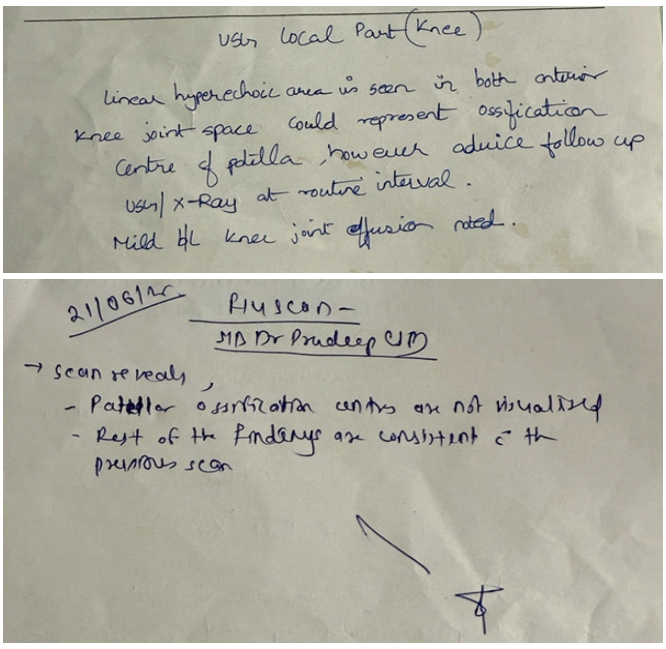

Figure 6: Ultrasound of the knee report.

were not palpable clinically. The elbow range of motion was full, and the rest of the musculoskeletal and systemic examination was unremarkable.

Radiographs of the pelvis revealed bilateral iliac horns (Fig. 5). Ultrasound of the knees demonstrated absent ossification centers for both patellae (Fig. 6). There were no vertebral or renal anomalies on screening, and renal function tests were normal. Based on the combination of clinical and radiological findings – absent fingernails, great toenail dysplasia, iliac horns, and patellar aplasia – a diagnosis of NPS was made [1,2]. Genetic counseling was offered to the family.

Orthopedic management began at 2 months of age with serial weekly long leg casts according to the Ponseti protocol. Correction of the deformity was achieved in five casts. This was followed by bilateral percutaneous Achilles tenotomy and application of a final cast for 3 weeks. At the end of the tenotomy, the Pirani score was 0 for both legs. A foot abduction brace was prescribed, and bracing compliance was closely monitored.

At 6-month follow-up, the child had pain-free, plantigrade feet with 15° of dorsiflexion bilaterally. The Laaveg-Ponseti functional score was 95/100. No recurrence or brace intolerance was observed, and the child had begun to walk independently. The last follow-up images are given below (Fig. 1, 2, 3, 4).

Discussion

NPS is a multisystem disorder with a highly variable presentation. Its hallmark skeletal findings include nail dysplasia, hypoplastic or absent patellae, and iliac horns [1, 2]. These anomalies often go unrecognized unless specifically investigated, particularly in infants, where patellar ossification is delayed. [9]

In our case, the complete absence of fingernails on both hands and great toenail aplasia prompted evaluation for syndromic causes. The identification of iliac horns on pelvic radiographs and absent patellar ossification confirmed the diagnosis of NPS.[10] These findings highlight the importance of a detailed musculoskeletal and radiological assessment in children with bilateral clubfoot and nail anomalies.

Clubfoot in syndromic settings, particularly those involving neuromuscular conditions such as arthrogryposis or spina bifida, tends to be stiff, resistant to conservative methods, and associated with higher recurrence [4, 5]. However, limited data suggest that NPS-associated clubfoot behaves more like idiopathic cases. Ey Batlle et al. reported a small series of four children with NPS and clubfoot, all successfully treated with the Ponseti method without recurrence [6].

Our patient responded well to early Ponseti casting started at 2 months of age. Complete correction was achieved in five casts, and results were maintained following tenotomy and bracing. This reinforces the notion that not all syndromic clubfeet should be presumed resistant to conservative treatment. Joint laxity, commonly described in NPS, may contribute to the

favorable response and improved brace tolerance [7].

The presence of consanguinity in this case may explain the pronounced phenotypic features. While NPS is typically inherited in an autosomal dominant fashion, enhanced expression in consanguineous unions may reflect underlying homozygosity or more penetrant gene expression [8].

Overall, this case emphasizes the critical role of early recognition and comprehensive evaluation in syndromic clubfoot. When managed appropriately, children with NPS and associated CTEV can achieve excellent functional outcomes without the need for extensive surgical correction.

Conclusion

NPS should be considered in any child presenting with bilateral clubfoot and associated nail dysplasia, especially when patellae are not clinically palpable, and iliac horns are seen on radiographs. The Ponseti method remains a highly effective treatment, even in syndromic cases such as NPS, when initiated early and followed rigorously.

Clinical Message

Clubfoot associated with Nail-patella syndrome, though rare, is responsive to conservative treatment. Complete absence of fingernails and great toenail aplasia should alert clinicians to this diagnosis. Iliac horns and patellar aplasia, detectable radiologically, support the diagnosis. Early initiation of the Ponseti method can result in excellent long-term outcomes.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given the consent for his/ her images and other clinical information to be reported in the journal. The patient understands that his/ her names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil **Source of support:** None

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