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A Case Series on Nail Patella Syndrome in A Tertiary Care Hospital

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Conflict of interest: Nil

Abstract

Background: Nail–Patella Syndrome (NPS), also known as hereditary onycho-osteodysplasia, is a rare autosomal dominant disorder caused by mutations in the LMX1B gene. It is characterized by a classical tetrad of nail dysplasia, patellar abnormalities, elbow dysplasia, and iliac horns. Early diagnosis is essential due to the possibility of associated renal and ocular complications.

Case Series: We report three familial cases of Nail–Patella Syndrome presenting with varying degrees of nail and musculoskeletal involvement. A 64-year-old male presented with congenital nail dystrophy and recurrent knee instability associated with a strong family history. An 8-year-old female child presented with dystrophic nails, bilateral elbow dislocation, limb-length discrepancy, and a positive family history. A 27-year-old female exhibited nail dystrophy, digital deformities, and bilateral patellar hypoplasia. Radiological evaluation demonstrated characteristic findings including bilateral iliac horns, hypoplastic patellae, and radial head dislocation. Renal and ophthalmological evaluations were normal in all patients.

Conclusion: This case series highlights the variable clinical presentation of Nail–Patella Syndrome and emphasizes the importance of careful nail examination, pedigree analysis, and radiological assessment in establishing the diagnosis. Early recognition facilitates family screening, genetic counselling, and long-term surveillance for systemic complications.

Keywords: Nail–Patella Syndrome, Hereditary Onycho-osteodysplasia, LMX1B, Iliac Horns, Nail Dysplasia, Patellar Hypoplasia.

Introduction

Nail–Patella Syndrome (NPS), also known as Fong disease or hereditary onycho-osteodysplasia, is a rare autosomal dominant genetic disorder with an estimated

prevalence of approximately 1 in 50,000 individuals. The disorder results from mutations in the LMX1B gene located on chromosome 9q34, which plays an essential role in dorsoventral limb development, renal morphogenesis, and ocular development.

The syndrome is classically characterized by a tetrad of nail dysplasia, patellar hypoplasia or aplasia, elbow abnormalities, and iliac horns. Nail abnormalities are the most consistent clinical feature and are present in approximately 90–95% of affected individuals. Renal manifestations range from asymptomatic proteinuria to progressive renal failure, while ocular involvement may include ocular hypertension and primary open-angle glaucoma.

Because of its rarity and variable expressivity, the diagnosis is frequently delayed. We report three familial cases of Nail–Patella Syndrome demonstrating characteristic clinical and radiological features.[1,2,5,6,10]

Materials and Methods

This descriptive observational study was conducted in the Department of Dermatology, Venereology and Leprosy, Santhiram Medical College and General Hospital, Nandyal. Three affected individuals belonging to a single multigenerational family with features suggestive of Nail–Patella Syndrome were identified and evaluated.

A detailed history regarding age at onset, family history, musculoskeletal symptoms, renal complaints, and ocular manifestations was obtained. All individuals underwent complete dermatological examination with particular attention to nail abnormalities. Musculoskeletal examination was performed to assess involvement of the knees, elbows, and other skeletal structures.

Radiological evaluation including pelvic radiographs, knee radiographs, and elbow radiographs was performed wherever indicated. Renal function assessment and ophthalmological examination were carried out to identify associated systemic manifestations.

Pedigree analysis was performed to determine the inheritance pattern within the family. These representative cases demonstrating the characteristic and diverse manifestations of Nail–Patella Syndrome were selected for detailed clinical and radiological presentation.

Ethical Considerations

The study was approved by the Institutional Ethics Committee (IEC) of Santhiram Medical College and General Hospital, Nandyal. Written informed consent was obtained from all patients and/or their legal guardians prior to clinical examination, photography, radiological evaluation, and publication of anonymized clinical data. Patient confidentiality and privacy were strictly maintained throughout the study.

Case Series

Case 1

A 64-year-old male from Nandyal presented with deformity of fingernails and toenails since birth along with recurrent dislocation of the knee joints since childhood. He experienced difficulty rising from a sitting position and reported significant functional impairment.

Family history revealed a strong autosomal dominant inheritance pattern. Multiple members across three generations were affected, including his mother, siblings, and children.

Clinical examination revealed dystrophic fingernails and toenails, with maximal involvement of the thumbs.

Nail abnormalities included anonychia, onycholysis, koilonychia, triangular lunulae, onychoschizia, longitudinal ridging, and nail hypoplasia.

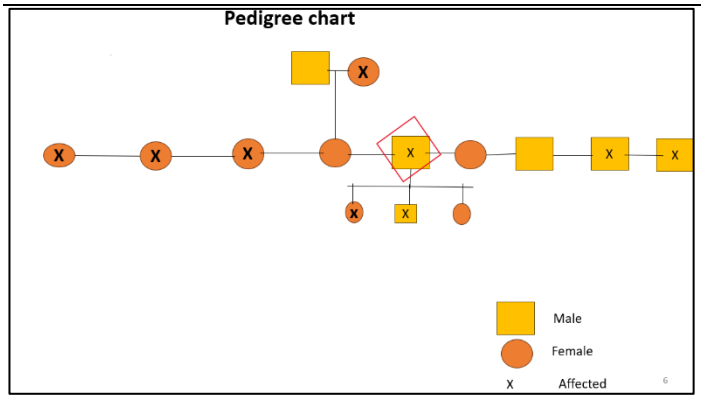

Figure 1.1: Pedigree chart of case 1

Musculoskeletal examination demonstrated bilateral patellar hypoplasia. Posterior iliac horns were palpable clinically. Laboratory investigations including complete blood count and renal function tests were within normal limits. Ophthalmological examination revealed no evidence of glaucoma.

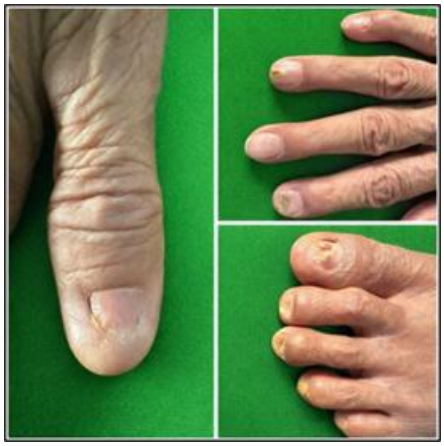

Figure 1.2- Clinical examination of Nails

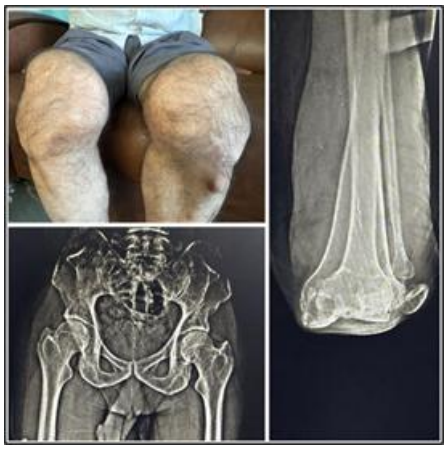

Figure 1.3- Radiological Findings

Radiographic examination showed bilateral iliac horns on pelvic radiographs and hypoplastic patellae on skyline knee radiographs. Based on the clinical and radiological findings, a diagnosis of Nail–Patella Syndrome was established.

Case 2

An 8-year-old female child presented with nail deformities, elbow abnormalities, and limb-length discrepancy since childhood. She experienced difficulty participating in physical activities and complained of pain while walking. A history of congenital clubfoot was present.

Family history revealed multiple affected relatives on the paternal side. Pedigree analysis demonstrated that her grandmother was related to Case 1, supporting familial inheritance.

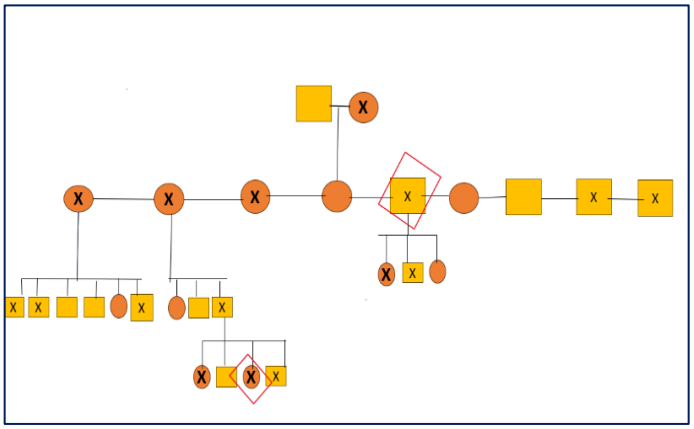

Figure 2.1: Pedigree chart of case 2

Examination showed hypoplastic thumb nails with nail plate thinning and splitting. The remaining fingernails showed variable involvement, while toenails were relatively spared. Limb-length discrepancy was evident on examination.

Figure 2.2- Clinical examination of Nails

Musculoskeletal examination revealed bilateral elbow dislocation. Ophthalmological evaluation was normal. Radiographs demonstrated radial head dislocation of the right elbow. The combination of characteristic nail abnormalities, positive family history, and radiological findings confirmed the diagnosis of Nail–Patella Syndrome.

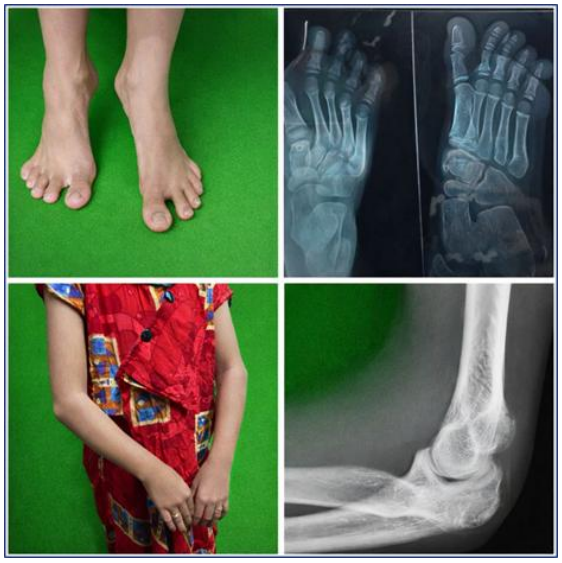

Figure 2.3 – Musculoskeletal and Radiological Findings

Case 3

A 27-year-old female presented with dystrophic fingernails and digital deformities characterized by hyperextension of the proximal interphalangeal joints with flexion of the distal interphalangeal joints, resembling swan-neck deformity. She also reported difficulty rising from a sitting position.

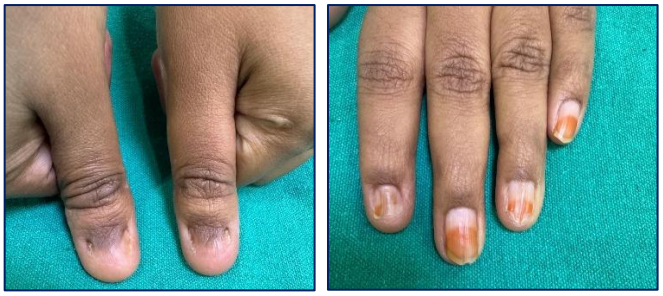

Figure 3.1 & 3.2- Clinical examination of Nails

Family history was positive for similar complaints among relatives. Clinical examination demonstrated dystrophic fingernails predominantly involving the thumbs. Bilateral patellar hypoplasia was detected on musculoskeletal examination.

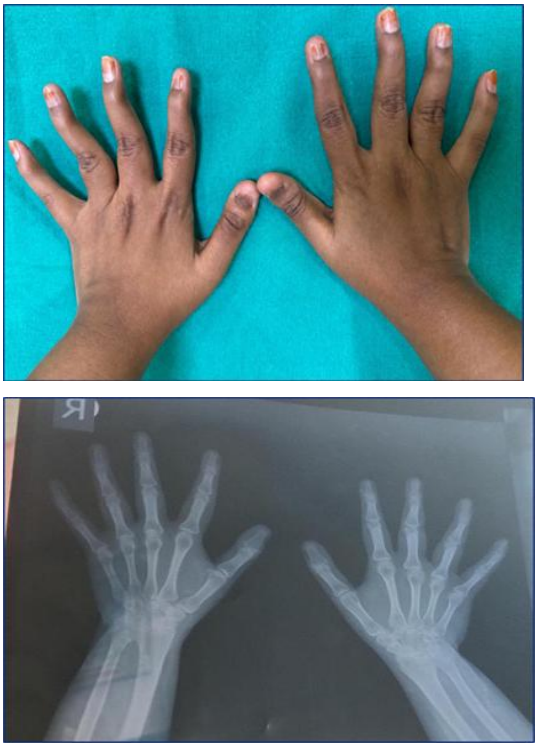

Figure 3.3 & 3.4 Clinical and Radiological images showing Swan Neck Deformity

Pelvic radiographs revealed bilateral iliac horns, while skyline radiographs of both knees demonstrated hypoplastic patellae. These findings were diagnostic of Nail–Patella Syndrome.

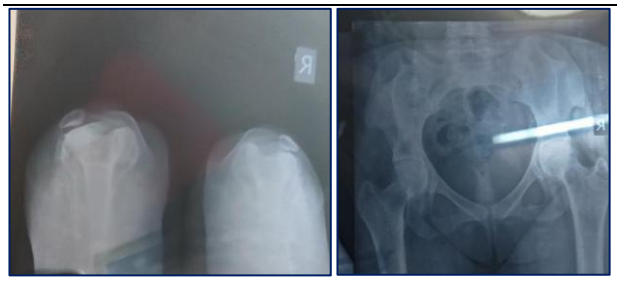

Figure 3.5 & 3.6 Radiological images showing Bilateral Patellar Hypoplasia and Bilateral Iliac Horns

Table 1: Clinical and Radiological Characteristics of the Three Cases of Nail–Patella Syndrome

Feature	Case 1	Case 2	Case 3
Age/Sex	64/M	8/F	27/F
Family history	Present	Present	Present
Nail dystrophy	Yes	Yes	Yes
Triangular lunula	Present	Absent	Not documented
Patellar hypoplasia	Present	Not documented	Present
Iliac horns	Present	Not documented	Present
Elbow abnormality	Absent clinically	Bilateral dislocation	Absent
Radial head dislocation	No	Present	No
Renal involvement	No	No	No
Ocular involvement	No	No	No

Discussion

Nail–Patella Syndrome is an uncommon autosomal dominant disorder caused by mutations in the LMX1B gene. [2,8,9] The syndrome demonstrates marked variable expressivity, even among members of the same family. The three cases presented here illustrate this phenotypic variability while sharing the characteristic features of the disorder.

Nail abnormalities are considered the hallmark of the disease and are frequently the earliest manifestation. The thumbnails are typically most severely affected, with decreasing severity toward the fifth digit. Findings may include hypoplasia, longitudinal ridging, splitting, triangular lunulae, onycholysis, and complete absence of nails. All three patients in our series demonstrated characteristic nail involvement, with the thumbs being predominantly affected.[5,6]

Skeletal abnormalities are another major component of the syndrome. Patellar hypoplasia or aplasia may result in recurrent dislocations, instability, and difficulty with ambulation. Case 1 and Case 3 demonstrated bilateral patellar hypoplasia, while Case 1 had a history of recurrent knee dislocation.

Elbow involvement usually results from dysplasia of the radial head and capitellum, leading to restricted movements and radial head dislocation. Case 2 demonstrated bilateral elbow abnormalities with radiological evidence of radial head dislocation.

Iliac horns are considered pathognomonic for Nail–Patella Syndrome and are reported in approximately 30–70% of affected individuals. These bilateral conical bony projections arising from the posterior iliac bones are usually asymptomatic but provide an important diagnostic clue. Iliac horns were demonstrated radiologically in Cases 1 and 3.

Renal disease is one of the most clinically significant complications of Nail–Patella Syndrome and may manifest as proteinuria, hematuria, nephrotic syndrome, or progressive renal insufficiency.[7] Ocular abnormalities such as ocular hypertension and primary open-angle glaucoma have also been reported. None of our patients demonstrated renal or ocular involvement at the time of evaluation. Nevertheless, lifelong surveillance remains necessary.

Genetic testing was not performed in our patients because the diagnosis was established based on classical clinical findings, characteristic radiological features, and a clear familial pattern of inheritance. In resource-limited settings, recognition of these hallmark manifestations remains essential for diagnosis.[3,5,6,9]

Conclusion

Nail–Patella Syndrome is a rare hereditary disorder with characteristic nail and skeletal manifestations. Recognition of nail abnormalities, particularly thumb nail dysplasia and triangular lunulae, together with radiological identification of iliac horns and patellar hypoplasia, can facilitate early diagnosis. Family screening, genetic counseling, and periodic renal and ophthalmological evaluation are important to reduce long-term morbidity and improve patient outcomes.[1,2]

Limitations

- Genetic testing for LMX1B mutation was not performed due to resource constraints.
- Phenotypic variability among affected family members could not be fully assessed as only three representative cases were included.
- Findings were based primarily on clinical and radiological evaluation without molecular confirmation.

References

1. Barsoum Z. Nail-patella syndrome. Sudan J Paediatr. 2023;23(2):252-254.
2. Lovelace PD, May LA. Nail-Patella Syndrome. [Updated 2023 May 23]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan.
3. Dharmshaktu GS, Dharmshaktu IS. Nail-Patella Syndrome: A Case Series From Northern India. Cureus. 2023 Oct 27;15(10):e47792.
4. Annie Hui Nee Law, *MB BCh BAO, MRCP(UK), MMED,* Li Ching Chew, *BMed Sci, BMBS, MRCP* Department of Rheumatology and Immunology, Singapore General Hospital**.**
5. Sweeney E, Fryer A, Mountford R, Green A, McIntosh I. Nail patella syndrome: a review of the phenotype aided by developmental biology. J Med Genet. 2003;40(3):153-162.
6. Bongers EMHF, Gubler MC, Knoers NVAM. Nail-patella syndrome. Overview on clinical and molecular findings. Pediatr Nephrol. 2002;17(9):703-712.
7. Lemley KV. Kidney disease in nail-patella syndrome. Pediatr Nephrol. 2009;24(12):2345-2354.
8. McIntosh I, Dreyer SD, Clough MV, Dunston JA, Eyaid W, Roig CM, et al. Mutation analysis of LMX1B gene in nail-patella syndrome patients. Am J Hum Genet. 1998;63(6):1651-1658.

9. Bongers EMHF, Huysmans FTM, Levtchenko EN, de Rooy JWJ, Blickman JG, Admiraal RJC, et al. Genotype-phenotype studies in nail-patella syndrome show associations between LMX1B mutations and clinical manifestations. Eur J Hum Genet. 2005;13(8):935-946.
10. Sweeney E, Hoover-Fong J, McIntosh I. Nail-Patella Syndrome. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2025.