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NAIL PATELLA SYNDROME WITH TOE NAIL DYSPLASIA- A RARE CASE REPORT

[1]Dr. Supradeeptha C, [2]Masapu Azay, [2]Parnapalle Sharath Kumar, [2]Lalam Pavan Kumar and [3]Satyaprasad J.
[1]Assistant Professor, [3]Professor,
Department of Orthopaedics
G.S.L Medical College, Rajahmundry, Andhra Pradesh, India
2Residents, G.S.L Medical College, Rajahmundry, Andhra Pradesh, India
E-mail: supradeep_challa@yahoo.co.in

Abstract: Nail-patella syndrome is a hereditary osteo-onychodysplasia. It is an autosomal dominant disease with involvement of the LMX 1 gene. We report a case of Nail-patella syndrome with bilateral absence of patella, hypo plastic finger nails, dysplastic toe nails, and bilateral iliac horns. The toe nails dysplasia is rare in Nail- patella syndrome.
Keywords: Nail-Patella syndrome, Hereditary onycho-osteodystrophy.

INTRODUCTION

Nail-patella syndrome also known as Fong disease, Osterreicher-Turner syndrome, Turner-Kieser syndrome, or hereditary onycho-osteodystrophy is a rare autosomal dominant condition with incidence at birth is estimated at 1/45000 and prevalence [1] at 1/50000.

Nail-patella syndrome is due to mutations of the LMX1B gene [2] located at the distal end of long arm of chromosome 9q34 [3].

LMX1B gene is a transcription factor of the LIM-homeodomain type that plays an important role for dorsoventral patterning of the limb and renal development. More than 140 heterozygous mutations in LMX1B have been reported [4], including missense, splicing, deletions, and nonsense mutations. Most mutations are located in the LIM domains. Most mutations are located in the LIM domains [5]. Although there is a phenotypic variability within families, the nephropathy is more often associated with homeodomain mutations [6, 7].

Nail-patella syndrome is defined by three major features [8]:

Nail anomalies- in 80-90% of patients they are bilateral and symmetrical; nails can be absent, hypoplastic, or dystrophic (discolouration, triangular lunulae, splitting, ridging, thinning).

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Skeletal anomalies- the most frequent are: absent or hypoplastic patellae, iliac horns (pathognomic), dysplastic elbows. Rarely genu recurvatum, genu varum, genu valgum, hypoplasia of cruciate ligaments, pectus excavatum, scoliosis, increased lumbar lordosis, spondylolisthesis, spondylosis, spina bifida, etc.

Other features- glaucoma, depression, attention deficit disorder, lean habitus and difficulty to gain weight, gastrointestinal symptoms (constipation, irritable bowel syndrome).

Renal disease[9]- found in approximately 40% of patients: proteinuria, microscopic hematuria, hypertension progression to renal failure has been reported in 3-15% of Nail-patella syndrome patients.

CASE REPORT

A 7 year old male child presented to the outpatient clinic with flexion deformity of bilateral knee. He was functionally impaired and had difficulty participating in sporting activities. He also complained of pain while climbing up and down stairs.

The parents gave history of full term, normal delivered child, with mild deformity of knees since birth.

The flexion deformity of knees gradually worsened from the age of one and half years, when he started walking.

On examination of nails, dystrophic changes involving several finger nails were noted. Another rare feature like toe nail dystrophy is noted.

On examination of knees, there was complete bilateral absence of patellae. Femoral condyles are large and prominent with sulcus in between condyles. Fixed flexion deformity in knee measuring 50 degrees on the left side and 40 degrees on the right side was noted. The power of quadriceps, hamstrings and glutei are normal.

Radiographs of both knees showed absence of both patellae. Radiograph of pelvis showed bilateral posterior iliac horns. Ultrasound abdomen was normal. The hematological parameters are within normal limits.

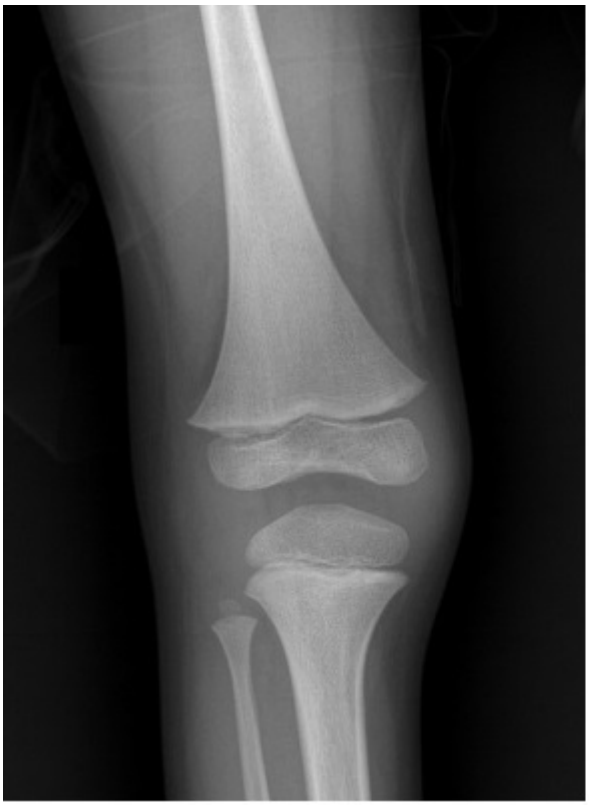

FIGURE 1 - Anteroposterior radiological view of right knee joint showing absence of patella

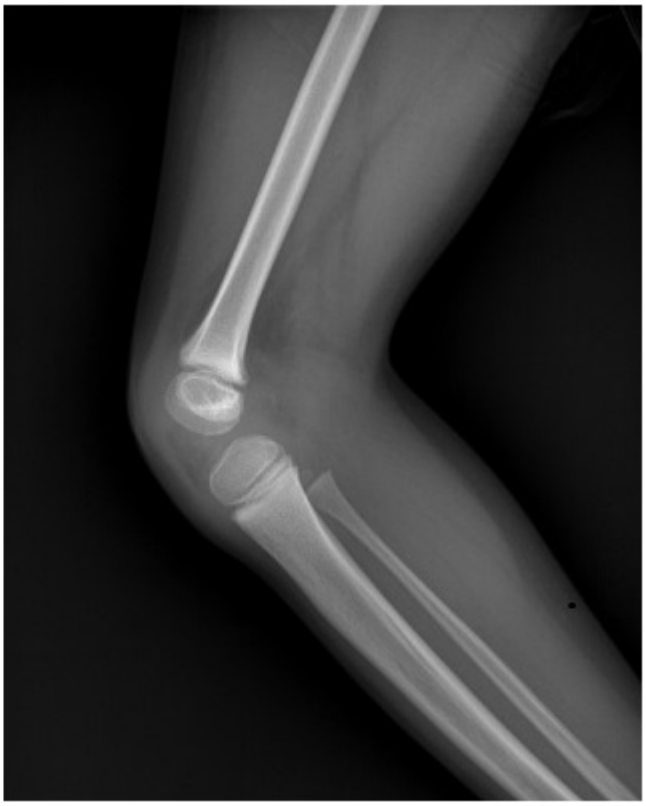

FIGURE 2- Lateral view of right knee showing absence of patella

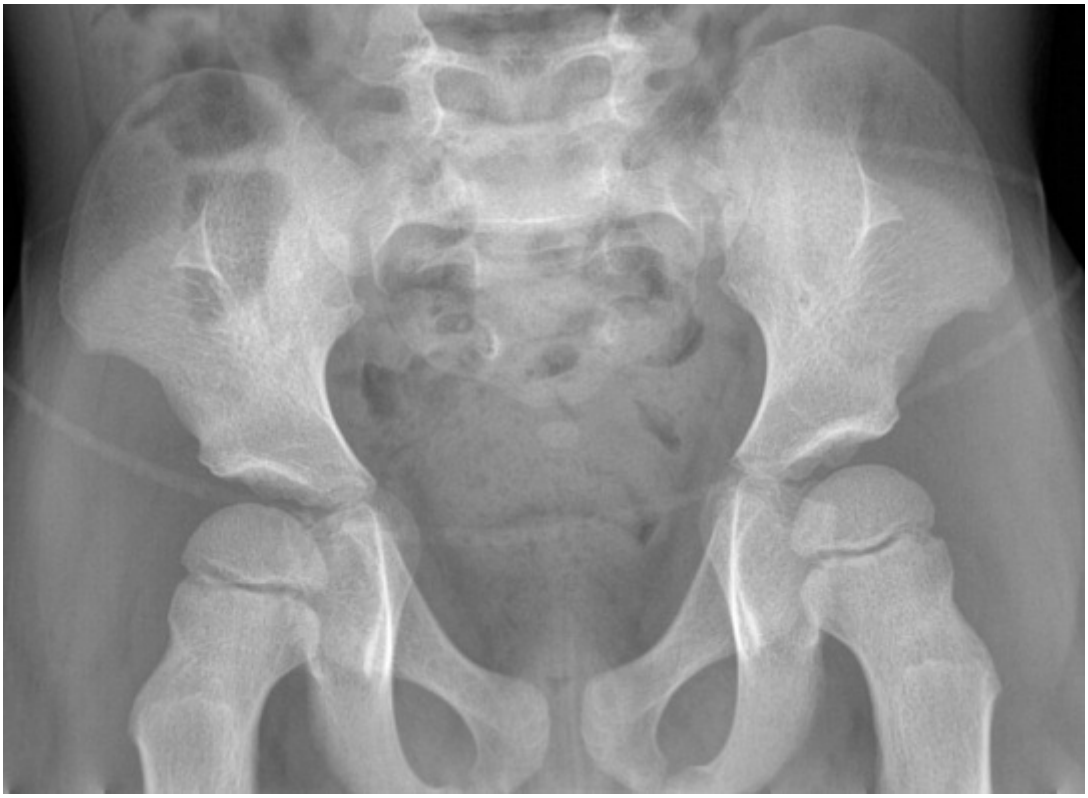

FIGURE 3- Radiography of pelvis showing iliac horns

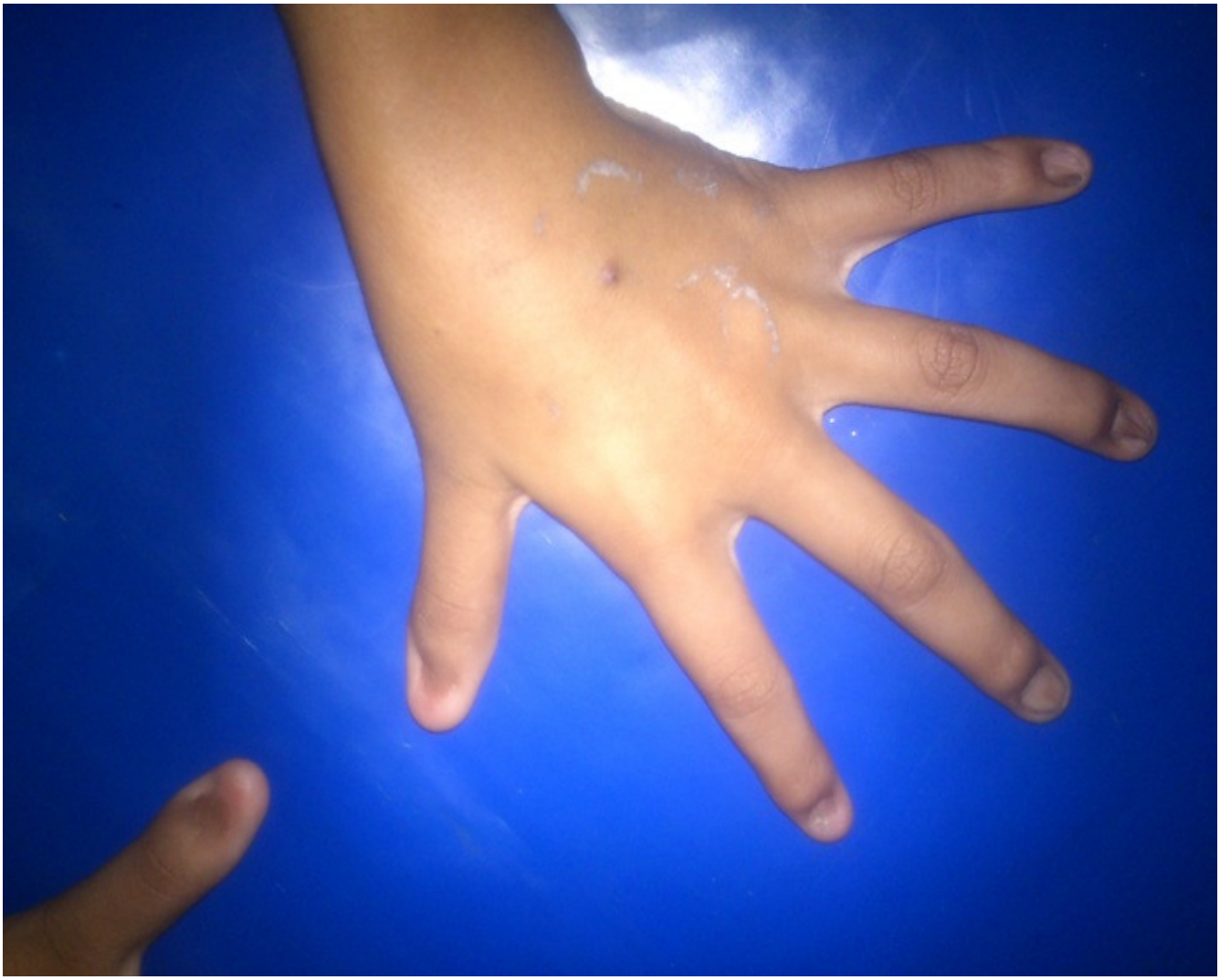

FIGURE 4- Dysplastic finger nails

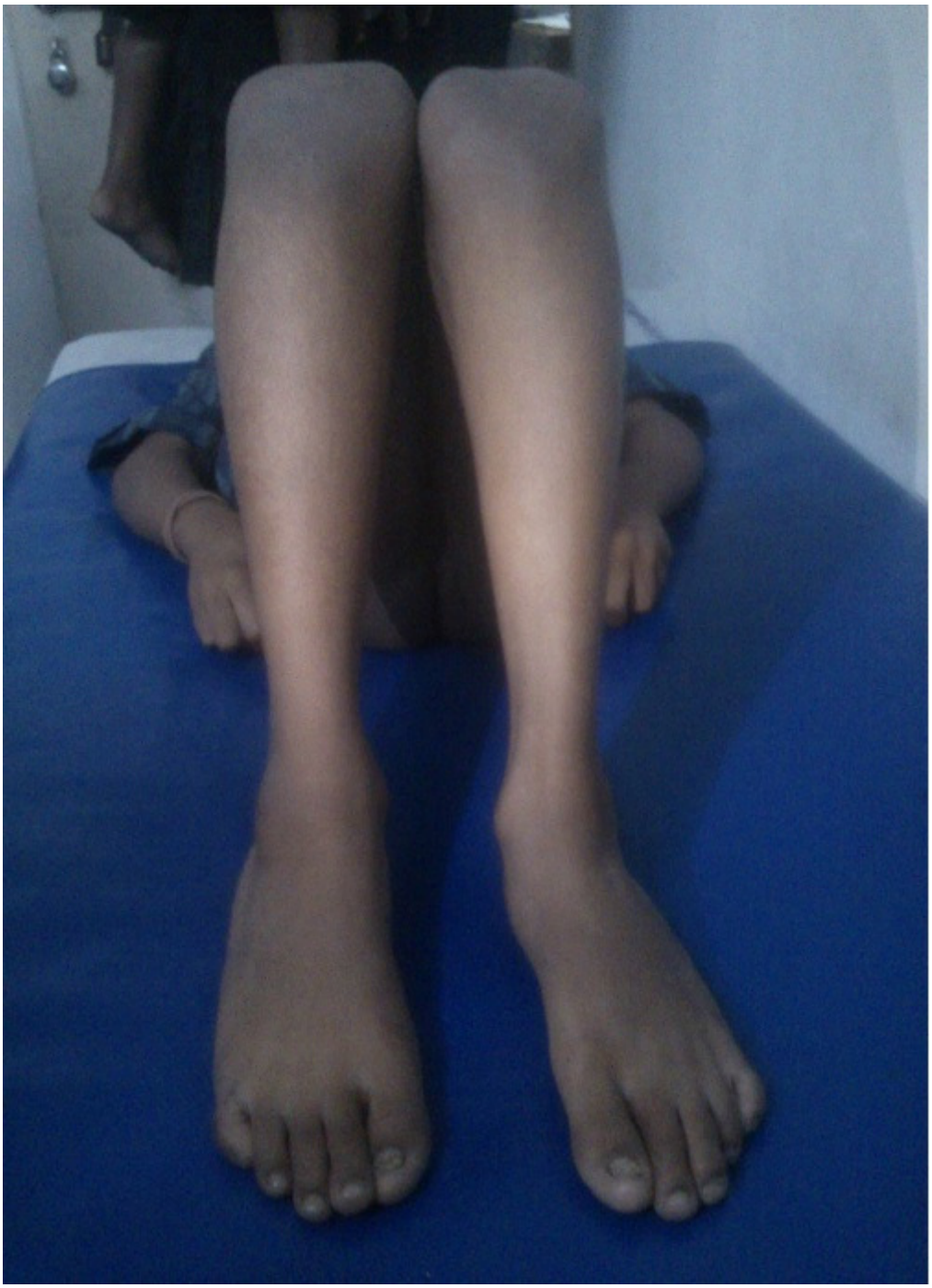

FIGURE 5- Sulcus of femoral condyles

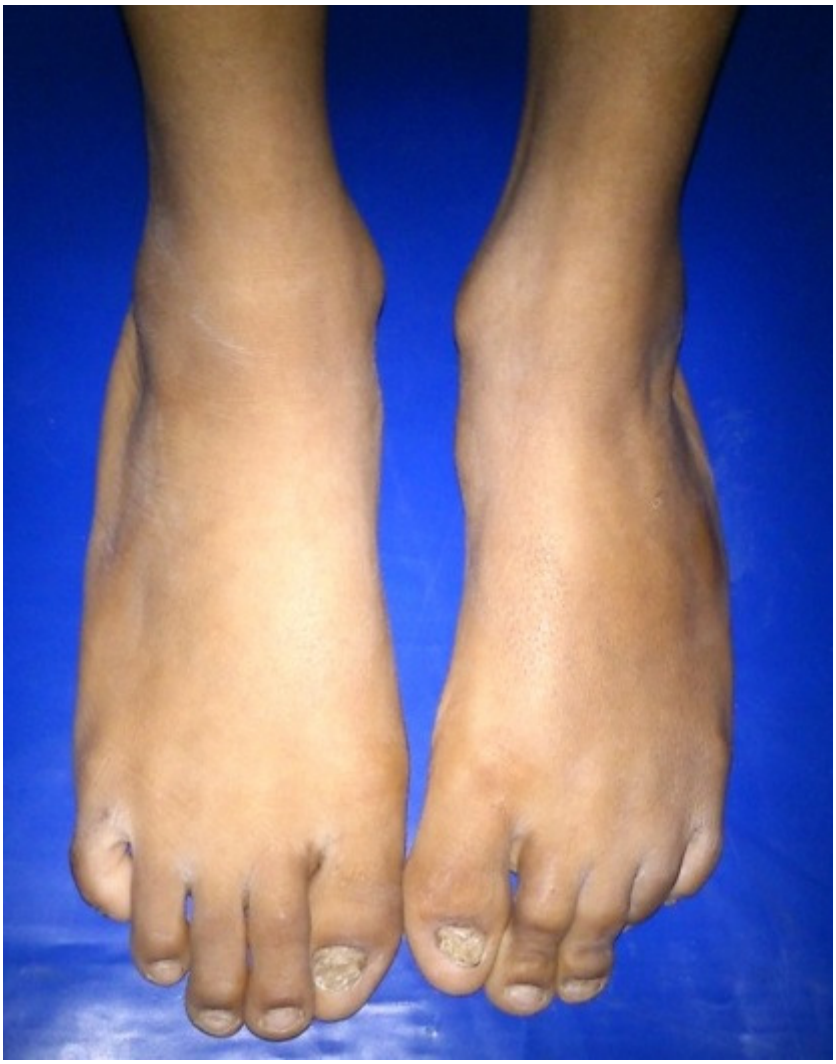

FIGURE 6- Dysplastic toe nails

DISCUSSION

Nail-patella syndrome is characterized by clinical triad involving the nails, skeletal system, and renal system. Although the diagnosis of nail patella syndrome can be established in early infancy through the radiographic demonstration of iliac horns, this disorder is rarely diagnosed in infancy. This is because the abnormalities of the nail and patella, which are essential for diagnosis, may not become apparent until later in life. The nails can be absent, hypoplastic, or dystrophic. These abnormalities present in 80-90% of patients. These abnormalities are often bilateral and symmetrical with more severely affecting thumb nails and least severity in little fingers. Dysplasia of toe nails is less frequent. Diminished skin creases over the distal inter-phalangeal joints of affected fingers are common.

Most common skeletal affection is hypoplastic or absent patella. Sometimes patella displaced or dislocated. Abnormalities of elbows may be asymmetric presenting with radial head and lateral epicondyle hypoplasia, causing subluxation of the radial head. Pathognomic radiological features of iliac horns are present in 30-70% of patients [10].

Extra-skeletal manifestations involving the kidneys [11] affects up to 40% of individuals, whereby 3-15% progress to end stage renal disease. Often, patients present with proteinuria with or without haematuria and may remain asymptomatic. Other manifestations are primary open-angle glaucoma and ocular hypertension, cloverleaf pigmentation of the iris (Lester's sign), attention-deficit hyperactivity disorder, sensory neuropathy, sensorineural hearing loss, irritable bowel syndrome and chronic constipation. Genetic studies were not performed in our patient as the diagnosis of Nail-patella syndrome was established based on the clinical and radiological manifestations.

CONCLUSION

It serves as a platform to understand and diagnose patients with presentation of abnormal nails and skeletal development. Future studies such as genetic therapy may have a role in reducing skeletal morbidity and reduce the risk.

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ADDRESS FOR CORRESPONDANCE
Dr. SUPRADEEPTHA CHALLA,
S/o C. JANAKIRAMA NAIDU
ASSISTANT PROFESSOR, DEPARTMENT OF ORTHOPAEDICS
G.S.L MEDICAL COLLEGE, RAJAHMUNDRY, ANDHRAPRADESH, INDIA
PIN-533296
PHONE 09941528076,
E-mail: supradeep_challa@yahoo.co.in