

CASE OF THE MONTH

History

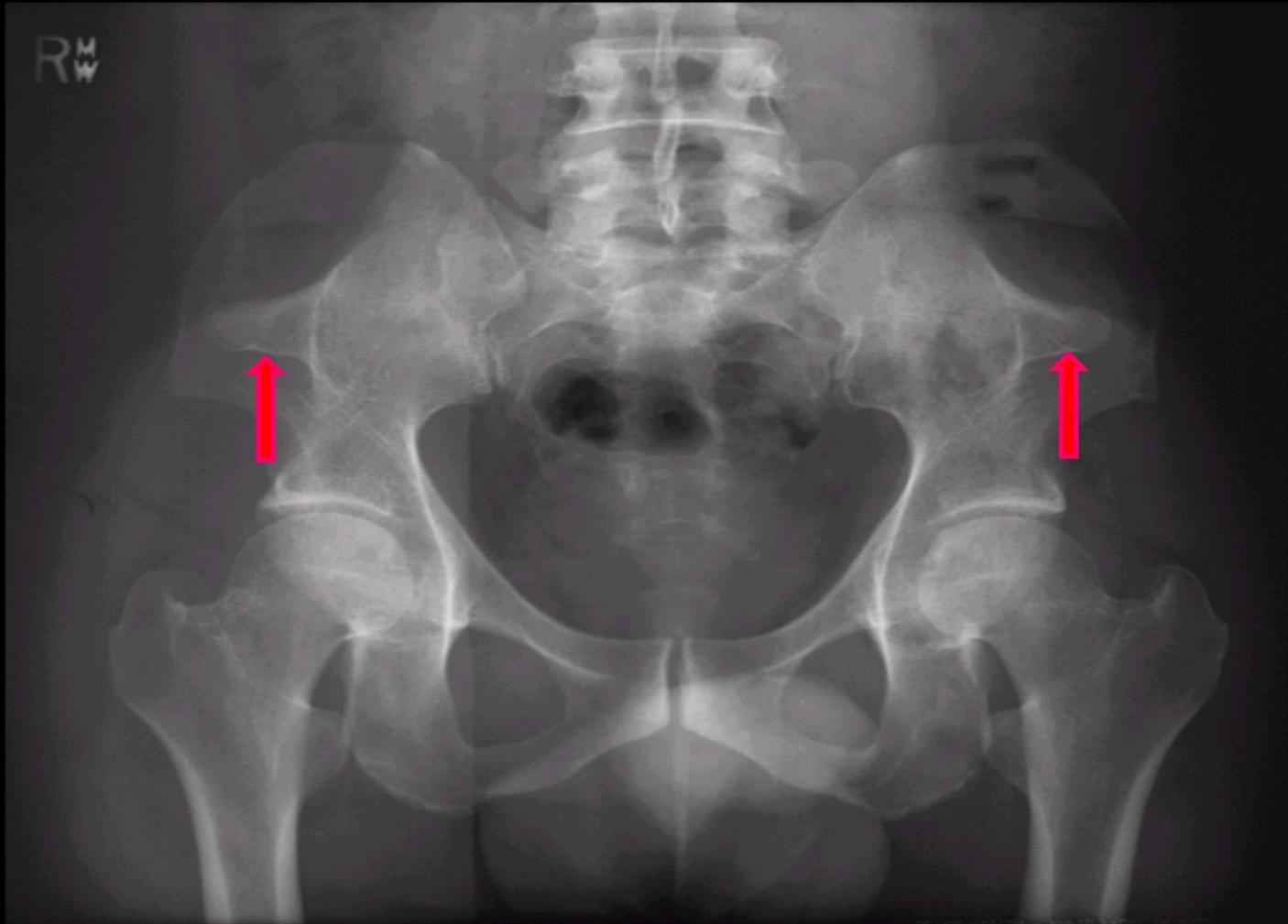
- A 22yrs male pt. presented with painful gait and proteinuria found on routine screening of the urine.
- Physical examination demonstrates malformed nails on index finger bilaterally and immobility of the both elbow



X Ray AP & Lateral Both Knee joint showing hypoplastic patella on the right side (Arrow) with absent patella on the left side (Patella is not seen in the expected region in circle)



X Ray Both Elbow showing deformity of radial head (arrow) and dislocation as well as degenerative changes in form of reduction in elbow joint space and marginal osteophytes.



X Ray Pelvis with both hip joints showing posterior iliac horns on either side.

Findings

- Hypoplastic patella on the right side with absent patella on the left side
- B/L radial head deformity and dislocation
- Posterior iliac horns

Diagnosis

Nail patella syndrome

Nail-patella syndrome

- It is a well known **autosomal dominant** condition characterized by nail dysplasia, patellar aplasia-hypoplasia, arthrodysplasia of the elbows, iliac horns, and nephropathy.
- The **LMX1B gene** plays a central role in the patterning of the nails, patella, long bones and kidney during development.
- **loss of function with one allele of this gene** is the most likely cause of pathology of the all manifestation in NPS.

Clinical features

- Patellar abnormalities
 - Skeletal deformities include patellar absence or hypoplasia, which may decrease flexion.
 - Osteoarthritis, osteoarthrosis, and knee effusions are associated complications; however, disability is not a major concern.
- Dysplastic nails
 - Absent or dysplastic nails are the most common nail findings and other nonspecific changes include discoloration, longitudinal ridging, and poorly formed lunulae.
 - Nails are progressively less affected toward the fifth digit.

Iliac horns

- These symmetrical, bilateral, central-posterior, iliac processes are asymptomatic and vary from a small dimple to a well-marked spur.

Elbow arthrodysplasia

- Clinical manifestations include an increased carrying angle and limited supination and pronation.
- On radiographs, the head of the radius is underdeveloped and displaced.

- **Renal symptoms** range from proteinuria to nephrotic syndrome. **Nephropathy (resembles glomerulonephritis)**-30-55%
- Patients present with proteinuria as the first clinical symptom.
- Some cases progress to ESRD, requiring transplantation & renal failure is the leading cause of death
- **Open-angle glaucoma**- due to pleiotropic effect of the *NPS1* gene.
- Age at diagnosis ranges from birth to late adulthood.
- Optic nerve and visual field damage can be prevented with early diagnosis & treatment of elevated intraocular pressure.

Imaging

- Radiography findings reveal **iliac horns** and **hypoplastic patella** in adults.
- Conduct MRI to **identify abnormal muscle insertions** (which cannot be observed by radiography).
- **Sonography** is indicated in young children for a better evaluation of the unossified patella.

Treatment

- Unless the patient presents with acute findings, evaluation should be done on an **outpatient basis**.
- **knee replacement** is suggested if the patient develops severe osteoarthritis in the knees.
- **Dialysis and transplantation** are necessary for patients who develop ESRD. Approximately 30-55% of patients have renal involvement.
- **Avoid excision of the radial head** to correct elbow arthrodysplasia. This excision forces the ulna into the wrist, creating a painful condition.

Authors

Dr Rajendra Solanki

Dr Chetan Mehta