Case 1469

Eurorad

Nail-patella syndrome

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Section: Paediatric radiology
Imaging Technique: MR
Case Type: Clinical Cases
Authors: J. Yarr, D. Gracey, A. Paterson
Patient: 6 years, male

Clinical History:

The patient was referred for radiographic examination when it was noted on clinical examination that there was a reduced range of movement of the left elbow. Dysplastic nails had been noted in the patient soon after birth.
Imaging Findings:

The patient was referred for radiographic examination when it was noted on clinical examination that there was a reduced range of movement of the left elbow. Radiographs were taken of the patient's pelvis, both knees and both elbows.
Dysplastic nails had been noted in the patient soon after birth. The patient's older sibling and father were similarly affected.

Discussion:

Nail-patella syndrome (onycho-osteodysplasia, Fong's Syndrome, hereditary onycho-osteodysplasia (HOOD), Turner-Kieser syndrome) is a connective tissue disorder characterised by dysplasia of the nails, absence or hypoplasia of the patellae and nephropathy. Inheritance is autosomal dominant (100% penetrance) with a wide spectrum of severity.
Dysplasia of the nails is usually noted in infancy, but other clinical features may not be apparent until the second or third decades of life. However, the diagnosis may be established radiologically soon after birth by the demonstration of bilateral iliac horns on a radiograph of the pelvis.

Nail dysplasia (hypoplasia, absence, spooning, ridging, flatness) particularly involves the thumb and is present in 80-98% of cases. The nails from the index finger to the little finger are progressively less damaged.

The iliac horns arise from the central area of the outer surface of the iliac wing and are symmetrical. They are rarely absent and are clinically palpable in 75% of cases. Bilateral iliac horns are pathognomonic of nail-patella syndrome and have not been described in any other condition. Flaring of the iliac wings and a small iliac angle may also be present.

Characteristic abnormalities of the knee include absence or hypoplasia of the patellae (in 60% of cases), asymmetrical development of the femoral condyles (the lateral condyles being small with relative overgrowth of the medial condyles), sloping of the tibial plateaux and prominence of the tibial tuberosities. The patellae are best seen on a 'skyline' view, however the radiographic diagnosis of patellar dysplasia is not possible until ossification occurs, usually at around 4 years of age. Clinically these features may present as a delay in walking, waddling gait, patellar

instability, knee deformity or genua valga.

Involvement of the elbow joint occurs in 60-90% of patients. Elongation of the radius with hypoplasia of the radial head and dorsal subluxation or dislocation of the radio-capitellar joint is the most common finding. Hypoplasia of the capitellum and asymmetrical development of the humeral condyles is also seen. These features present as an increased carrying angle or an inability to extend, pronate or supinate the forearm.

There are many other reported skeletal abnormalities including: hypoplasia of the scapula with a shallow glenoid fossa and a long acromial process, clavicular horns, thickening and convexity of the lateral border of the scapula, hypoplasia of the first ribs, Madelung's deformity, a triangular configuration of the lunate, clinodactyly, camptodactyly, finger joint laxity, equinovalgus deformity of the hindfoot and supination of the forefoot, calcaneovalgus deformity of the hindfoot, a ball-and-socket ankle joint, forefoot supination and lateral subluxation of the tarsal-metatarsal joints, asymmetrical development of joints, degenerative arthritis, renal osteopathy and pectus carinatum.

Associated nephropathy presents as proteinuria and may progress to nephrotic or nephritic syndrome. End-stage renal failure occurs in up to one third of patients and has been described in the first decade of life. Electron microscopy has demonstrated a characteristic glomerular basement membrane abnormality.

Ocular abnormalities include ptosis, strabismus, glaucoma and micro-cornea. 50% of affected individuals have heterochromia of the iris with an inner 'clover-leaf' darker area (Lester iris).

Differential Diagnosis List: Nail-patella syndrome

Final Diagnosis: Nail-patella syndrome

References:

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Radiographic diagnosis of osteo-onychodysostosis in infancy.
Radiology. 1973 Oct;109(1):151-4. (PMID: 4783116)
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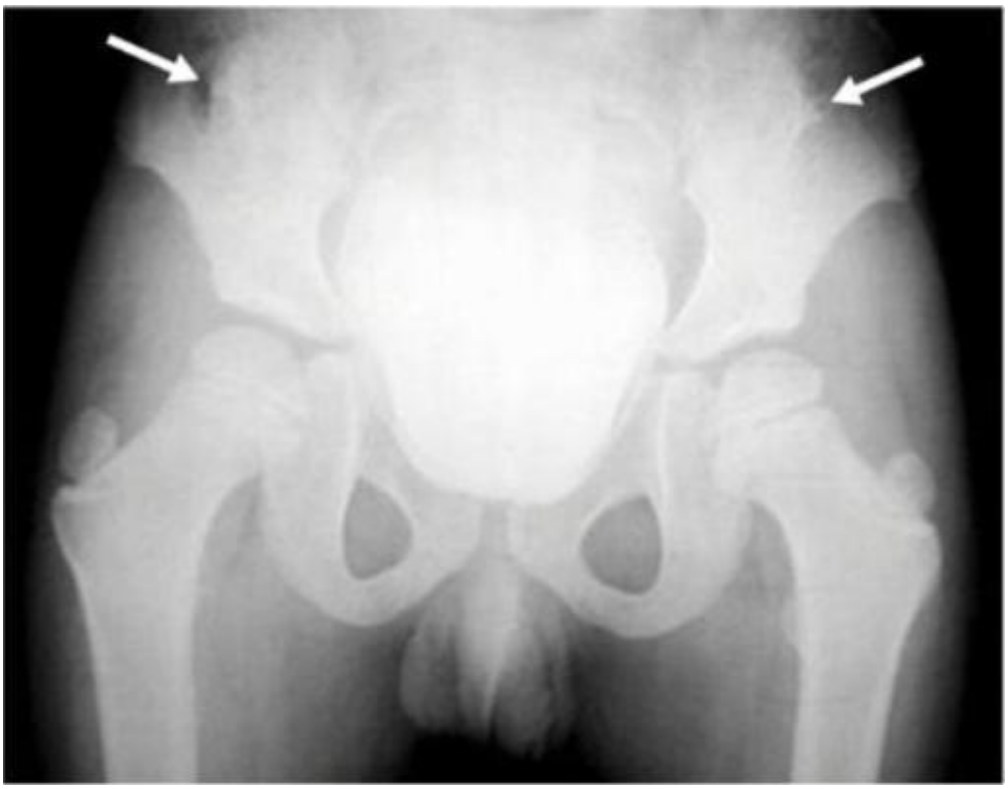
In Wynne-Davies R, Hall CM, Apley AG. (eds). Atlas of skeletal dysplasias. Churchill Livingstone, London, pp 614-16 (1985).

Figure 1

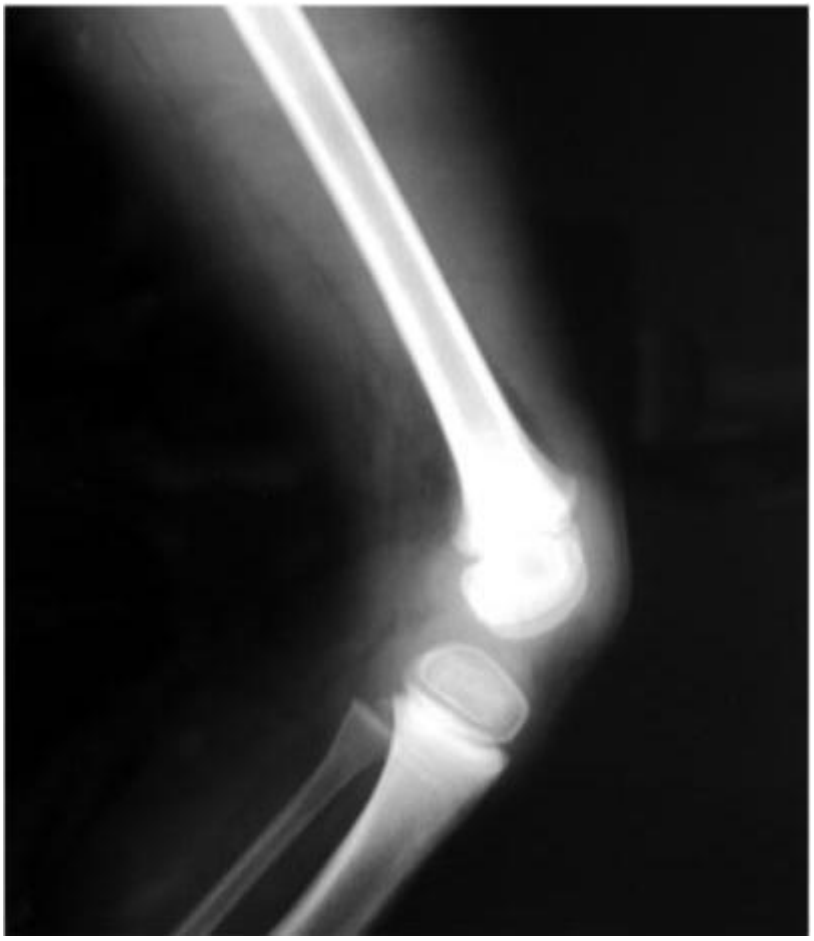
a



Description: Pelvic radiograph showing bilateral iliac horns. **Origin:**

Figure 2

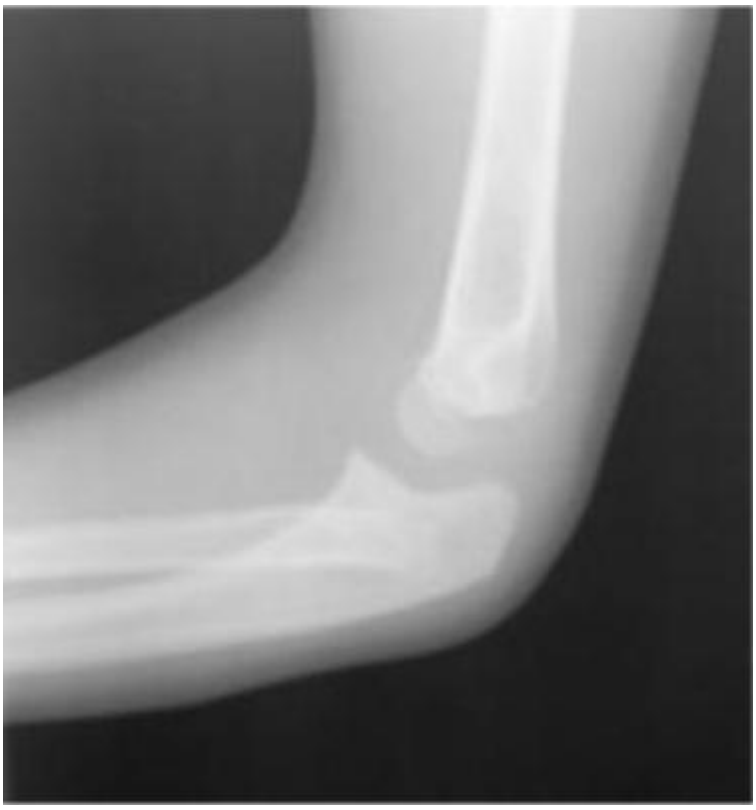
a



Description: Lateral radiograph of the left knee demonstrating absence of the patella. **Origin:**

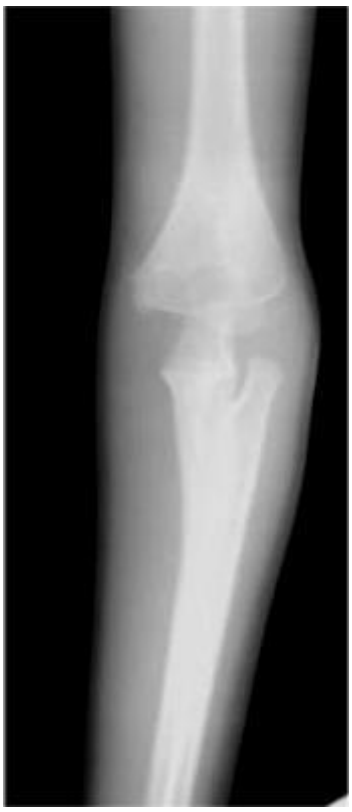
Figure 3

a



Description: Lateral view of the left elbow showing dislocation of the radio-capitellar joint. **Origin:**

b



Description: Anterior view of the left elbow showing dislocation of the radio-capitellar joint. **Origin:**