Nail patella syndrome
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Section: Musculoskeletal system
Case Type: Clinical Cases
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Patient: 18 years, female

Clinical History:
Iliac horns and dysplastic fingernails.

Imaging Findings:
An otherwise healthy patient was admitted to hospital with abdominal discomfort. Physical examination and laboratory studies were unremarkable, with the exception of nail dystrophy of the ulnar digits. An abdominal plain film was performed revealing bilateral posterior iliac horns, pathognomonic of a rare syndrome called nail patella syndrome or osteo-onychodysplasia.

Additional radiographs showed hypoplastic patellae.

Discussion:
Nail patella syndrome is a rare, hereditary, autosomal dominant disorder, also called hereditary osteo-onychodysplasia and Fong's syndrome. It is characterised by dysplastic fingernails; hypoplastic or absent patellae; additional bony deformities, particularly about the pelvis and elbows; bony processes along the posterior surfaces of the iliac bones, also called iliac horns; and renal dysplasia.

Clinical manifestations are seen most frequently in the second and third decades of life. In infancy, this syndrome is identified by the iliac horns, absence or hypoplasia of the fibula and patella, asymmetric development of the femoral condyles or proteinuria.

Iliac horns are seen in 80% of cases. They are bilateral accessory outgrowths, consisting of cortex and medulla, continuous with the iliac bone. They are located at the site of attachment of the gluteus medius muscles and project posterolaterally. These smooth bony outgrowths are asymptomatic, frequently palpable, and, because they have no effect on gait, they need not be treated. Iliac horns are the pathognomonic feature of nail patella syndrome; that is, they occur in approximately 80% of cases and are observed only in this condition. The iliac crests are often flared with prominent anterior superior spines.

The patellae are absent or hypoplastic. The femoral condyles may be unusually prominent and there may be a valgus deformity. In the elbow, the head of the radius is poorly formed. In some cases the radius is abnormally long. Less frequently other bones show deformity in size and shape.

The disorder affecting the nails varies from unusual thinning and small size to a complete absence of one or more of the nails. The thumb is involved most frequently and severely.
Nail patella syndrome is also characterised by nephropathy. Skeletal defects and renal involvement might occur separately. The usual clinical presenting syndromes of the nephropathy are asymptomatic proteinuria, microscopic haematuria and sometimes nephrotic syndrome. In a considerable proportion of patients renal failure develops.

Other manifestations of this syndrome include ocular involvement, such as glaucoma and microcornea.

**Differential Diagnosis List:** Nail patella syndrome

**Final Diagnosis:** Nail patella syndrome

**References:**


Description: Plain film radiogram of the pelvis. There are characteristic bony horns (arrows) arising from the posterior ilia. The pelvis is abnormally flared and the anterior superior spines are unusually prominent. Origin:
**Description:** Plain film of the knees. The patella is hypoplastic and small for an 18-year-old female.

**Origin:**
Description: Radiogram of elbow. The head of the radius is poorly formed. Origin: