Renal failure with skeletal abnormalities

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A 40-year-old man presented with complaints of persistent vomiting for six months, periorbital puffiness for four months, ankle swelling for one month and chest pain for three days. He was apparently asymptomatic prior to the onset of these symptoms. He had experienced persistent vomiting two to three times a day, which was non-projectile and non-bilious with no constant relation to meals. He had noticed increasing puffiness of face, maximally in the morning for the past four months. One month prior to admission he had noticed swelling around his ankles. Along with these complaints he also noticed a decrease in his urine output. The patient had been diagnosed as suffering from chronic renal failure due to chronic glomerulonephritis at another hospital and had received peritoneal dialysis. He had then been referred to this hospital for further management.

Examination revealed a pale man of average build with anasarca. He had a pulse rate of 112 beats/min, blood pressure of 180/115 mmHg, respiratory rate of 20 breaths/min and a temperature of 99°F. His jugular venous pressure was 11 cm above the sternal angle with normal wave patterns. The thumb and index nails were hypoplastic as were all the toe nails. Other findings included fixed flexion deformity of bilateral elbows, excessive lumbar lordosis with high iliac crests, genu varum and bilateral absent patellae. Abdominal examination revealed a firm non-tender spleen 6 cm in size. The rest of the systemic examination was unremarkable.

Investigations showed him to be in renal failure. The patient was subjected to peritoneal dialysis following which there was improvement in biochemical parameters. X-rays of the left knee, right knee and pelvis are shown in figures 1, 2 and 3, respectively. Ultrasound examination of the abdomen revealed bilateral contracted kidneys with loss of corticomedullary differentiation and small cortical cysts in both kidneys.

Figure 1 X-ray of left knee

Figure 2 X-ray of right knee

Figure 3 X-ray of pelvis

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Questions

- 1 What are the positive findings on the X-rays?
- 2 What is the most likely diagnosis?

Answers

QUESTION 1

The X-rays show a hypoplastic patella on the left side, an absent patella on the right side, and the presence of bilateral pelvic horns.

QUESTION 2

Nail patella syndrome. The radiological picture along with the nail changes, elbow and knee deformity and renal failure makes nail patella syndrome the diagnosis.

Discussion

Nail patella syndrome is a rare autosomal dominant inherited disorder involving usually the skeletal and renal systems but rarely also the eye and the skin.¹ The skeletal involvement can have multiple features. The patella can be hypoplastic or absent along with poorly developed tendons which cause the patella to dislocate sideways causing gait disturbances in some patients. Early onset of secondary osteoarthritis is common. The nail abnormalities may vary from triangular lunule of the nails to complete absence of nails or rudimentary nails, especially the thumb nails.² Another common feature is poor development of the radial head, along with the capitulum and lateral condyle of the humerus, causing subluxation at the elbow joint resulting in a fixed flexion deformity of elbows and a wide carrying angle. Bilateral iliac horns³ can be seen in plain films of the pelvis as pyramidal spurs projecting laterally from the centre of the iliac bones. Although the iliac horns are pathognomonic of nail-patella syndrome they may not be present in all cases. At times hypoplasia of the lateral condyle of the femur can result in genu varum. Scoliosis, prominent straight clavicles, hypoplasia of the first rib,⁴ calcaneovalgus and equinovarus are the other skeletal abnormalities described. Ocular abnormalities like microcornea and cataract and mental retardation have also been described.²

Another characteristic feature, the most important from the clinical viewpoint, is the renal involvement. This is seen in about 40 to 50% of patients with nail patella syndrome.⁵ It usually takes the form of asymptomatic proteinuria and, rarely, haematuria can occur. It can progress to end stage renal disease in about 20 to 30% of these patients.⁵ About 8% of nail patella syndrome patients die by the age of 40 years due to the renal involvement.⁶

The present patient showed most of the described features, namely, the nail changes, bilateral iliac horns, elbow deformity and knee deformity, as well as renal failure, for which he was already being managed. Even before the development of renal insufficiency, he had

Nail patella syndrome: skeletal features

- hypoplastic or absent patella
- nail abnormalities (rudimentary to absent nails)
- poor development of radial head
- bilateral iliac horns

Rare

- scoliosis
- prominent straight clavicle
- hypoplasia of first rib
- calcaneovalgus
- equinovarus

Nail patella syndrome: clinical features

- nail changes
- elbow and knee deformity
- renal failure
- ocular changes - microcornea, cataract
- mental retardation

Nail patella syndrome: renal involvement

- 40 - 50% patients have renal involvement
- usually present as asymptomatic proteinuria
- in 20 - 30% patients it can progress to end stage renal disease
- no renal histological pattern is pathognomonic of the disease

been seen by a number of physicians for his skeletal deformity but no definite diagnosis had been made. Had an early diagnosis of nail patella syndrome been made and knowing that a significant number of these patients develop renal insufficiency, serial renal function analysis would have allowed the early detection of renal failure, giving time to evaluate and work-up the patient for a possible renal transplant. The present case highlights the fact that nail patella syndrome is not a benign disease as was initially thought. Early recognition of this entity is important to allow early detection of renal involvement and serial follow-up since these patients will do well with a renal transplant once they reach end-stage renal failure.

Final diagnosis

Nail patella syndrome

Keywords: nail patella syndrome, renal failure

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