

## Musculoskeletal case 22. Diagnosis

### Nail patella syndrome

**N**ail patella syndrome, or osteo-onychodysostosis, is characterized by a clinical tetrad of nail dysplasia (hypoplasia, pitting or ridging), hypoplastic or absent patellae, dislocation of the radial head and iliac “horns.”<sup>1</sup> It is also known as horn syndrome, hereditary onycho-osteodysplasia (HOOD), Fong’s disease and Turner–Kieser syndrome. Fig. 1 (see page 205), the anteroposterior view of the left knee, demonstrates a hypoplastic left patella. The anteroposterior view of the right knee (not shown) demonstrated identical changes. Fig. 2 (see page 205), the skyline view of both patellae, demonstrates small patellae bilaterally (arrow) subluxed anterior to hypoplastic lateral femoral condyles (curved arrow). Note how the medial femoral condyles appear relatively enlarged. On the frontal view of the pelvis (Fig. 3, see page 205) there are characteristic bony protuberances at the iliac wings (arrows). These are known as iliac horns or Fong’s lesions (“Fong’s prongs”).

The syndrome is inherited as an autosomal dominant condition and is linked to the ABO blood group locus.<sup>2</sup> Many cases, however, are secondary to spontaneous mutation. Nail patella syndrome occurs in ap-

proximately 2.2 of every 100 000 people. The clinical findings have been widely described, dating back to Chatelain in 1820 and may initially become manifest in childhood, although presentation is usually in the second or third decades.<sup>2</sup> Hypoplasia or even absence of the patella and lateral femoral condyle can lead to deformity, abnormal gait, patellar instability and genu valgum.

The patellar abnormalities often give the knees a square look. Because of malformations in the bones, muscles and ligaments the joint is often very unstable, causing it to dislocate. Associated abnormalities of the foot and hip, particularly talipes equinovarus, are not uncommon.<sup>2,3</sup> Deformity of the elbow due to condylar hypoplasia and radial head subluxation is usually obvious clinically, but significant disability is rare.<sup>2</sup> Skeletal deformity can also occur as a result of soft-tissue dysplasia and joint flexion contractures, which may require surgical release.<sup>3</sup>

The presence of posterior iliac (Fong’s) horns (Fig. 3) is considered pathognomic.<sup>1,4</sup> These arise at the attachment of the gluteus medius but do not cause functional impairment.

Renal dysplasia, which can progress to renal failure and renal osteodystrophy, may be seen in later life. The frequency of kidney involve-

ment in nail patella syndrome varies but seems to range from 30% to 50% by adulthood.<sup>2</sup> Most renal involvement presents as proteinuria, which is often mild and not life-threatening. Renal abnormalities are sometimes worsened by pregnancy, and women having no history of renal problems can also experience pregnancy-related kidney problems like toxemia and pre-eclampsia. In a relatively small number of cases these abnormalities can progress to full renal failure necessitating kidney transplantation.

Management consists of physiotherapy and, if necessary, surgery to maintain limb and joint function. Renal function should also be monitored.

### References

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