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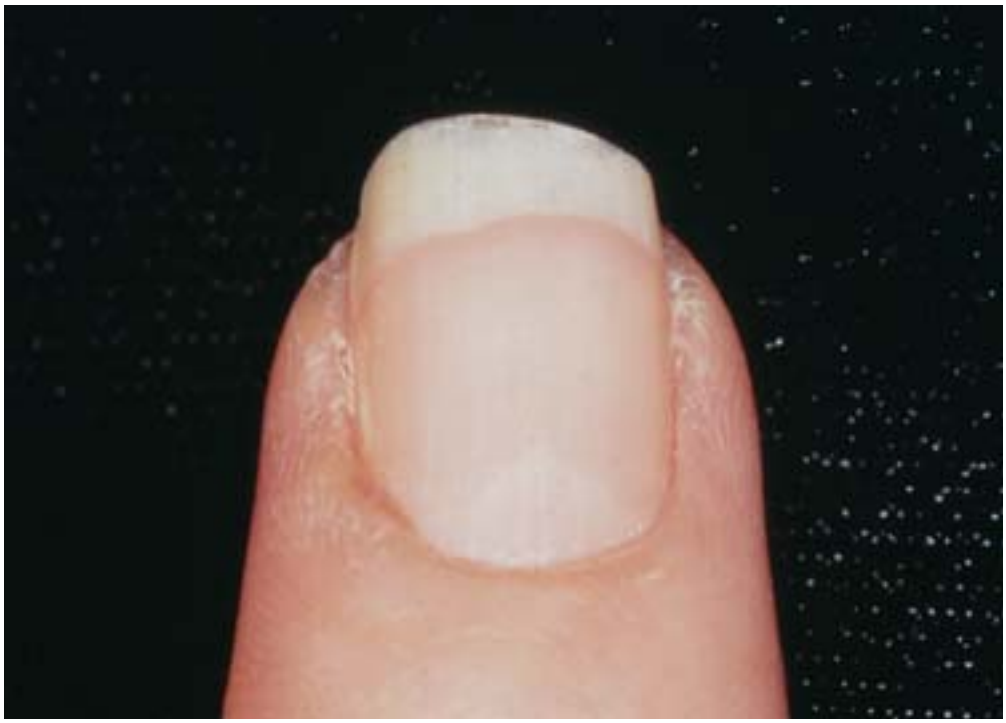

This patient relates a family history of kidney disease.

What is your diagnosis?

PLEASE TURN TO PAGE 75 FOR DISCUSSION

The Diagnosis

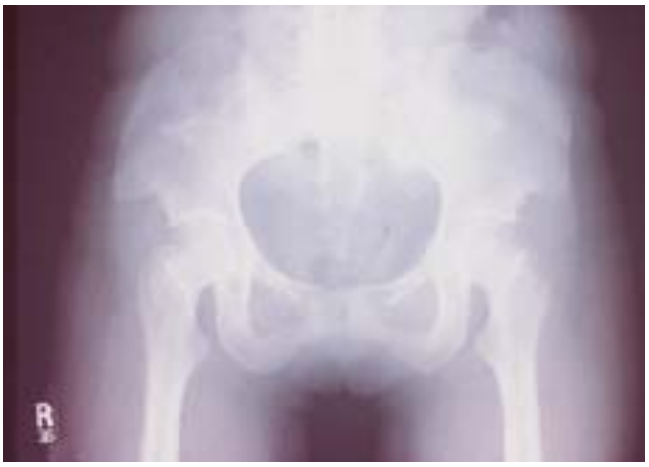

FIGURE 1. A frontal view of the pelvis shows bilateral iliac wing horns.

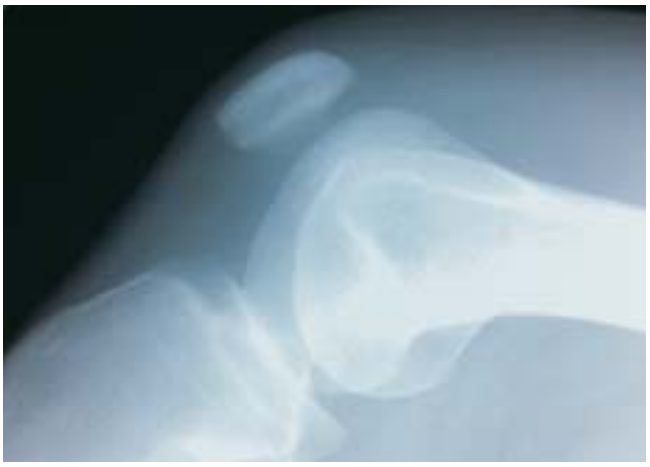

FIGURE 2. Patellas are hypoplastic.

Discussion

The nail-patella syndrome, or osteo-onychodysplasia, is an autosomal dominant disorder characterized by orthopedic changes, renal disease, glaucoma, and characteristic nail changes, especially the presence of triangular lunulae.[1] Other nail features include hypoplasia, most prominent on the thumb and index fingers. Orthopedic features of the syndrome include absent or hypoplastic patellae, elbow deformities, cervical ribs, and iliac horns. Abnormalities of the humerus, radius, ulna, and finger bones can result in early and painful degenerative disease.[2] The abnormal carrying angle of the elbow can be associated with contracture.[3] Renal involvement includes proteinuria, and may present as the nephrotic syndrome. Hypertension and renal failure may occur. Because of the risk of glaucoma, all family members should be screened by an ophthalmologist.[4]

The radiographic studies pictured show characteristic changes. A frontal view of the pelvis demonstrates bilateral iliac wing horns (Figure 1). Deformity of the anterior iliac crest is also noted bilaterally. Radiographs of the knees demonstrate slight hypoplasia of the patellas bilaterally (Figure 2). Both frontal and lateral views of the elbows demonstrate bilateral developmental radial head dislocations, with associated dysplasia of the radiocapitellar

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FIGURE 3. Bilateral developmental radial head dislocations are present.

articular surfaces (Figure 3). The radial heads are dislocated posteriorly relative to the capitella. The findings of bilateral iliac wing horns and bilateral radial head dislocations are characteristic of the nail-patella syndrome.

Studies have linked the syndrome to chromosome 9q34 and identified point mutations in the LMX1B gene.[5] Other kindreds have been linked to chromosome 17q21-22.[6] Prenatal diagnosis is possible, including non-invasive prenatal diagnosis using ultrasonography.[7]

Proper management of these patients requires a multispecialty approach. Orthopedic surgeons, nephrologists, and ophthalmologists have a role in management. The dermatologist may establish the diagnosis and should guide the patient and family members to the appropriate specialists for care. Treatment of kidney disease has included the use of angiotensin-converting enzyme inhibitors.[8]

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