THE 'SMALL-PATELLA' SYNDROME

HEREDITARY OSTEODYSPLASIA OF THE KNEE, PELVIS AND FOOT

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We describe a syndrome combining abnormalities of the pelvis, knee and foot in three related patients with a familial history of small dislocated patellae. The clinical and radiological appearance of the patella and pelvis is consistent with the 'small-patella' syndrome, a rare autosomal dominant disorder. There were also previously unreported deformities affecting the feet.

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The 'small-patella' syndrome, first described by Scott and Taor in 1979, is characterised by patellar aplasia or hypoplasia and abnormalities of the pelvic girdle. We describe a family in which the classic anomalies of this syndrome are associated with minor morphological variations of the hind-foot. We present the radiological features and discuss the aetiology.

CASE REPORTS

Case 1 (III3, Fig. 1). A 68-year-old woman was admitted with bilateral chronic knee and ankle pain. She was obese and had a personal and family history of recurrent dislocation of small patellae (Fig. 1).

Physical examination revealed small, laterally placed patellae, flat feet, and limited subtalar motion. The other peripheral joints were normal and there was no nail dystrophy or leg shortening. Radiographs (Fig. 2) showed multiple skeletal abnormalities, including small dislocated patellae, bilateral defective formation of the ischiopubic

Fig. 1

Genealogical table of the family reported.

junction, hypoplasia of the lesser trochanters, infra-acetabular notches, ball-and-socket ankle joints, secondary radiological signs of bilateral talocalcaneal coalition with narrowing of the posterior talocalcaneal joint space and a talar beak, hypertrophy of the neck of the talus and broad-based plantar calcaneal exostoses.

Three-dimensional CT of the pelvis showed thin ischiopubic junctions and emphasised the infra-acetabular notches (Fig. 3a). Coronal CT of the right hindfoot (Fig. 3b) identified talocalcaneal coalition at the level of the sustentaculum tali. No other skeletal abnormalities were seen.

Case 2 (IV1, Fig. 1). This 49-year-old woman, the daughter of the patient in case 1, had small patellae. Radiographs of the pelvis, knees and hindfeet showed small patellae, hypoplasia of the ischiopubic junction, hypertrophy of the neck

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Fig. 2a

Fig. 2b

Fig. 2c

Case 1. Figure 2a – AP radiograph of the right knee showing a small laterally placed patella. Figure 2b – AP radiograph of the right ankle showing the ball-and-socket joint. Figure 2c – Lateral radiograph of the left hindfoot showing talar beaking, narrowing of the posterior talocalcaneal joint space, hypertrophy of the talar neck and broad-based plantar calcaneal exostosis (arrow).

Fig. 3a

Fig. 3b

Case 1. Figure 3a – Three-dimensional CT of the pelvis showing thin ischiopubic junctions and infra-acetabular notches (black arrows). Figure 3b – Coronal CT of right ankle at the level of the sustentaculum tali showing talocalcaneal coalition.

of the talus and broad-based plantar calcaneal exostoses.

Case 3 (V2, Fig. 1). The 20-year-old daughter of the patient in case 2 also had hypoplasia of the patellae. Radiographs showed changes similar to those in her mother although defective formation of the ischiopubic junction was more severe (Fig. 4).

The radiological findings of the patients are summarised in Table I.

DISCUSSION

In the nail-patella syndrome (hereditary onycho-osteodysplasia) (Duncan and Souter 1963; Duthie and Hecht 1963) patellar abnormalities may be associated with iliac horns, nail dystrophy, elbow deformities and renal dysplasia. Other anomalies may also be present but a ball-and-socket ankle joint, talocalcaneal coalition, hypertrophy of the talar neck, calcaneal exostoses or a hypoplastic ischiopubic junction

Fig. 4

Case 3. AP radiograph of the pelvis showing absence of the ischiopubic junction, hypoplasia of the lesser trochanters and drum-stick appearance of the upper femoral epiphyses.

Table I. Details of the radiological findings in the three cases

	Case		
	1	2	3
Small patella	+	+	+
Defective formation of the ischiopubic junction	+	+	+
Hypoplasia of the lesser trochanter	+	+	+
Hypertrophy of the talar neck	+	+	+
Broad-based plantar calcaneal exostosis	+	+	+
Drum-stick appearance of the upper femoral epiphysis	-	+	+
Infra-acetabular notch	+	-	-
Ball-and-socket ankle joint	+	-	-
Tarsal coalition	+	-	-

tion have not been described (Hogh and Macnicol 1985). In 1979, Scott and Taor identified a new entity in 12 closely-related members of one family, the 'small-patella' syndrome, in which absent or small dislocated patellae were seen in association with coxa vara or valga with buttressing of the femoral head, apparent hypoplasia of the lesser trochanters, and prominent or defective ossification of the ischiopubic junction. Other skeletal abnormalities included flat feet and syndactyly of the toes. Vanek (1981) reported three familial cases with infra-acetabular notches while Burckhardt (1988) described three more sporadic cases. Deformities of the tarsal bones were not mentioned in any of the above patients. Scott and Taor (1979) and Vanek (1981) postulated an autosomal dominant mode of transmission with 100% penetrance for the patellar changes. This genetic pattern of inheritance is also suggested by the genealogical table of our family (Fig. 1).

Tarsal coalition, an abnormal union of two or more tarsal bones (Cowell and Elener 1983; Carson et al 1991), may be either congenital or acquired, and has a reported prevalence of about 1% in the general population. The true frequency is probably higher, however, as many coalitions are asymptomatic (Sartoris and Resnick 1985). Secondary radiological signs of talocalcaneal coalition include talar beaking, narrowing of the posterior joint space and a ball-and-socket ankle joint (Conway and Cowell 1969; Sartoris and Resnick 1985); the last is characterised by a spherical articulation between the tibia and the talus (Lamb 1958; Channon and Brotherton 1979; Pistoia, Ozonoff and Wintz 1987). This rare deformity is more commonly congenital when it may be associated with shortening of the lower limbs, coalition of tarsal bones, absence or fusion of digital rays, and aplasia or hypoplasia of the fibula (Pappas and

Miller 1982; Pistoia et al 1987).

The clinical and radiological appearances of the patella and pelvis in our patients are consistent with the 'small-patella' syndrome. In case 1, the coexistence with this syndrome of two congenital conditions which commonly occur together, talocalcaneal coalition and a ball-and-socket ankle joint, suggests a common hereditary origin although, in view of the relatively high frequency of talocalcaneal coalition, this could also be a chance finding. Hypertrophy of the neck of the talus and plantar calcaneal exostoses were found in all our patients over three generations, as were the abnormality of the ischiopubic junction and the small patellae, indicating a clear hereditary pattern. The severity of the abnormality in each area is variable, but since the most clinically significant anomaly is patellar hypoplasia we believe that the disorder should continue to be called 'small-patella' syndrome, although abnormalities of the pelvis and foot are also important components. Every patient presenting with patellar aplasia or hypoplasia should have radiography of the pelvis and feet to help to establish an accurate diagnosis.

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REFERENCES

- Burckhardt A.** 'The small patella syndrome': a combination of knee and pelvic dysplasia. *Zeitschr Orthop* 1988;126:22-9.
- Carson CW, Ginsburg WW, Cohen MD, McLeod RA, Kitaoka HB.** Tarsal coalition: an unusual cause of foot pain - clinical spectrum and treatment in 129 patients. *Semin Arthritis Rheum* 1991;20:367-77.
- Channon GM, Brotherton BJ.** The ball and socket ankle joint. *J Bone Joint Surg [Br]* 1979;61-B:85-9.
- Conway JJ, Cowell HR.** Tarsal coalition: clinical significance and roentgenographic demonstration. *Radiology* 1969;92:799-811.
- Cowell HR, Elener V.** Rigid painful flatfoot secondary to tarsal coalition. *Clin Orthop* 1983;177:54-60.
- Duncan JG, Souter WA.** Hereditary onycho-osteodysplasia: the nail-patella syndrome. *J Bone Joint Surg [Br]* 1963;45-B:242-58.
- Duthie RB, Hecht F.** The inheritance and development of the nail-patella syndrome. *J Bone Joint Surg [Br]* 1963;45-B:259-67.
- Hogh J, Macnicol MF.** Foot deformities associated with onycho-osteodysplasia: a familial study and a review of associated features. *Int Orthop* 1985;9:135-8.
- Lamb D.** The ball and socket ankle joint: a congenital abnormality. *J Bone Joint Surg [Br]* 1958;40-B:240-3.
- Pappas AM, Miller JT.** Congenital ball-and-socket ankle joints and related lower-extremity malformations. *J Bone Joint Surg [Am]* 1982;64-A:672-9.
- Pistoia F, Ozonoff MB, Wintz P.** Ball-and-socket ankle joint. *Skeletal Radiol* 1987;16:447-51.
- Sartoris DJ, Resnick DL.** Tarsal coalition. *Arthritis Rheum* 1985;28:331-8.
- Scott JE, Taor WS.** The 'small patella' syndrome. *J Bone Joint Surg [Br]* 1979;61-B:172-5.
- Vanek J.** Ischiopatellar dysplasia (Scott and Taor's syndrome of the small patella). *Fortschr Röntgenstr* 1981;135:354-6.