Progressive anterior knee pain associated with patellar instability in a 57-year-old father and his daughter

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Diagnosis

Patellar instability in a family with small patella syndrome

Discussion

Small patella syndrome is a generalized bone dysplasia with diagnostic features in the pelvis and knees and subtle changes in other parts of the skeleton. Scott and Taor [1] reported 12 individuals from an autosomal dominant pedigree with small or absent patellae. Seven individuals also had abnormalities of the pelvis or upper femora consisting of coxa vara, hypoplasia of the lesser trochanter, and abnormal ossification of the ischio-pubic junction. Bernhang and Levine [2] reported bilateral absence of the patella detected in a 14-year-old child because of a cystic swelling over one knee. This revealed a clear fluid and the

cyst had to be surgically excised. Radiologically, other than the absent patellae, there were no osseous lesions. The patient's brother was similarly affected. Bongers et al. [3] provided the first evidence for the crucial role of human *TBX4* gene in the skeletal development of the patella, pelvis, and feet. They identified the heterozygosity of *TBX4* mutations in SPS patients, together with the similar skeletal phenotype of animals lacking *Tbx4*.

Patellar stability relies on the limb alignment, the osseous architecture of the patella and the trochlear, the integrity of the soft tissue constraints, and the interplay of the surrounding muscles [4]. Previous reports described trochlear dysplasia as an abnormality of the shape and depth of the trochlear groove, mainly in its proximal extent. This was defined radiologically by Dejour et al. [5] on the basis of the crossing sign (*croisement*). If the line of the trochlear floor crosses the anterior border of one or both condyles, the trochlea is said to be flat at that level. The same feature was described by Grelsamer and Tedder [6] and called the lateral trochlear sign. Trochlear dysplasia was further classified by Dejour and Locatelli [7] and Tavernier and Dejour [8] on the basis of the CT morphology of the index scan and the direct lateral radiograph.

Small patella syndrome should be differentiated from the more common and better-known nail patella syndrome. Nail patella syndrome is characterized by bilateral dysplastic patellae, glaucoma, and nephropathy. Iliac horns are present in only approximately 70–80% of cases; they represent a pathognomonic radiographic sign [9]. Additional skeletal abnormalities include talipes equinovarus, a malformed capitellum of the radius, and fibular aplasia. The nails are abnormal from birth and are either absent or small and longitudinally ridged.

The case presentation can be found at doi: 10.1007/s00256-010-0880-x.

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The family described here presented with distinctive radiographic features. Figure 1a in the case presentation, a lateral knee radiograph, showed trochlear dysplasia, including the crossing sign, supratrochlear spur, and double contour (a hypoplastic medial facet). Figure 1b, a skyline view, showed lateral tracking of the patella with trochlear dysplasia and lateral patellar compression syndrome of the patello-femoral joint, especially of the left knee with subchondral degenerative changes (this is a common feature in patients with lateral tilted patella, when there is more pressure at the lateral facet). This is typical of osteoarthritis of the patello-femoral joint. Strikingly, there is hypoplasia of the medial condyle and the failure of formation of a femoral notch, more marked on the right side. In Fig. 2a, the skyline view showed dysplasia and subluxation of the patellae and aplasia of the trochlear groove bilaterally and in Fig. 2b the transverse 3D double echo steady-state gradient-echo MRI showed a flat trochlear groove (arrows) and a hypoplastic/subluxated patella.

Finally, we wish to stress that patello-femoral dysplasia and patello-femoral osteoarthritis are typical manifestations in association with a small patella.

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