Acta Orthop. Belg., 2004, **70**, 373-376

CASE REPORT

Congenital patellar aplasia in conjunction with trisomy 8. A case report

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Congenital patellar aplasia and hypoplasia are quite rare. Bilateral flexion deformity of the fifth finger, patellar aplasia in the right knee and patellar hypoplasia in the left knee were noted in a 6-year-old boy brought in for treatment of curvature of both little fingers. Active and passive movements of the knees were within normal range, and the quadriceps muscle was of normal strength. Trisomy 8 was determined on chromosome analysis.

In contrast with the literature, the patellar abnormality was not concomitant with any other syndromes in our case, and most clinical findings of trisomy 8 were absent.

INTRODUCTION

Congenital patellar aplasia is quite rare. It is generally concomitant with other deformities and is a familial trait *(1,2)*. In the literature, it has been reported that patellar aplasia often occurs in conjunction with small patella syndrome and nail-patella syndrome (osteo-onychodysplasia, Turner-Fong syndrome), and rarely with Kuskokwin syndrome, trisomy 8, and Coffin-Sirris syndrome *(1-5)*.

In the present study, a case with patellar aplasia on the right side and patellar hypoplasia on the left with concomitant bilateral flexion deformity of the fifth finger distal interphalangeal joint and trisomy 8 is presented.

CASE REPORT

A six-year-old boy was brought in for treatment of curvature in the little fingers of both hands and deformity in the knees. The other members of the patient's family, including six siblings, were healthy.

Orthopaedic examination showed flexion deformity of the fifth finger in both hands. The flexion deformity was in the distal interphalangeal joints, and was of moderate degree. Finger extension increased during flexion of the wrist and metacarpophalangeal joint. There was also compensatory extension contracture in both metacarpophalangeal joints.

The patient's gait was normal. Physical examination of the lower extremities indicated that the anterior aspect of both knees was flatter than

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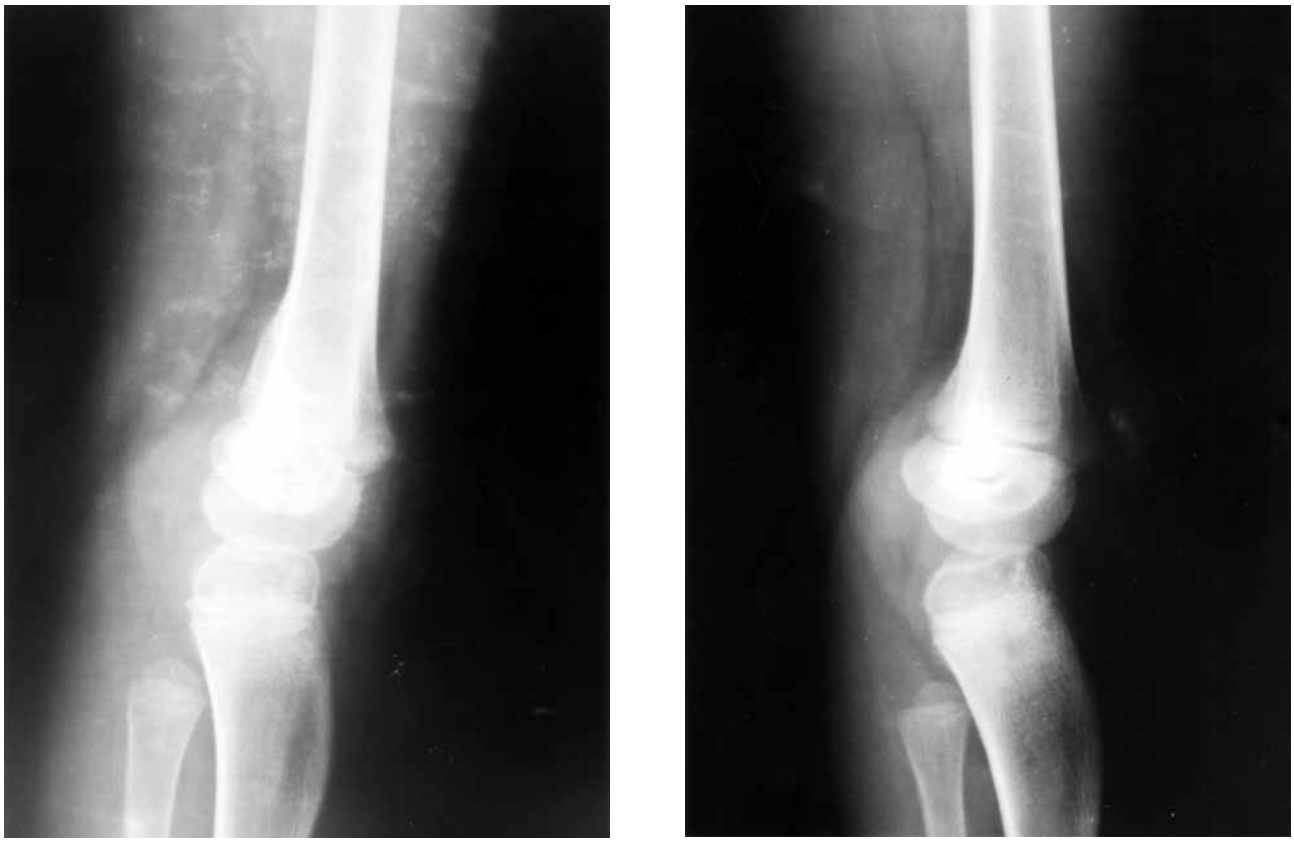

Fig. 1. — Lateral knee radiographs : (a) patellar aplasia on the right side ; (b) patellar hypoplasia on the left side.

normal ; this became more evident during knee flexion. The trochlear surface of the femur was detectable on palpation, more easily on the right. However, palpation demonstrated that both patellae were not in place. The extensor mechanism of the knee was not impaired, active and passive movements were within normal range, and quadriceps muscle strength was 5/5.

Lateral radiographs of both hands showed flexion deformity in the fifth finger (fig 1a, b) and lateral radiographs of the knees showed total congenital patellar aplasia on the right side and patella hypoplasia on the left (fig 2a, b).

The diagnosis of trisomy 8 was made, based on chromosome analysis.

Because both quadriceps had normal strength, no treatment was considered for the congenital patellar aplasia. Flexion deformity of the fifth fingers was not treated because contracture was of medium severity, and because surgery corrects only cosmetic appearance, having no effect on function. Only an orthosis and exercise were recommended. The patient was followed up because deformity may increase with growth.

DISCUSSION

Nail-patella syndrome (hereditary onycho-osteodysplasia) may be accompanied by patellar abnormalities, iliac horns, nail dystrophy, wrist deformity, and renal dystrophy *(4,5)*. Small patella syndrome was first diagnosed in 12 members of a single family and described in detail by Scott and Taor *(3)*. In addition to patellar aplasia or small, dislocated patellae, this syndrome also includes flattening of the femoral heads, coxa vara or valga,

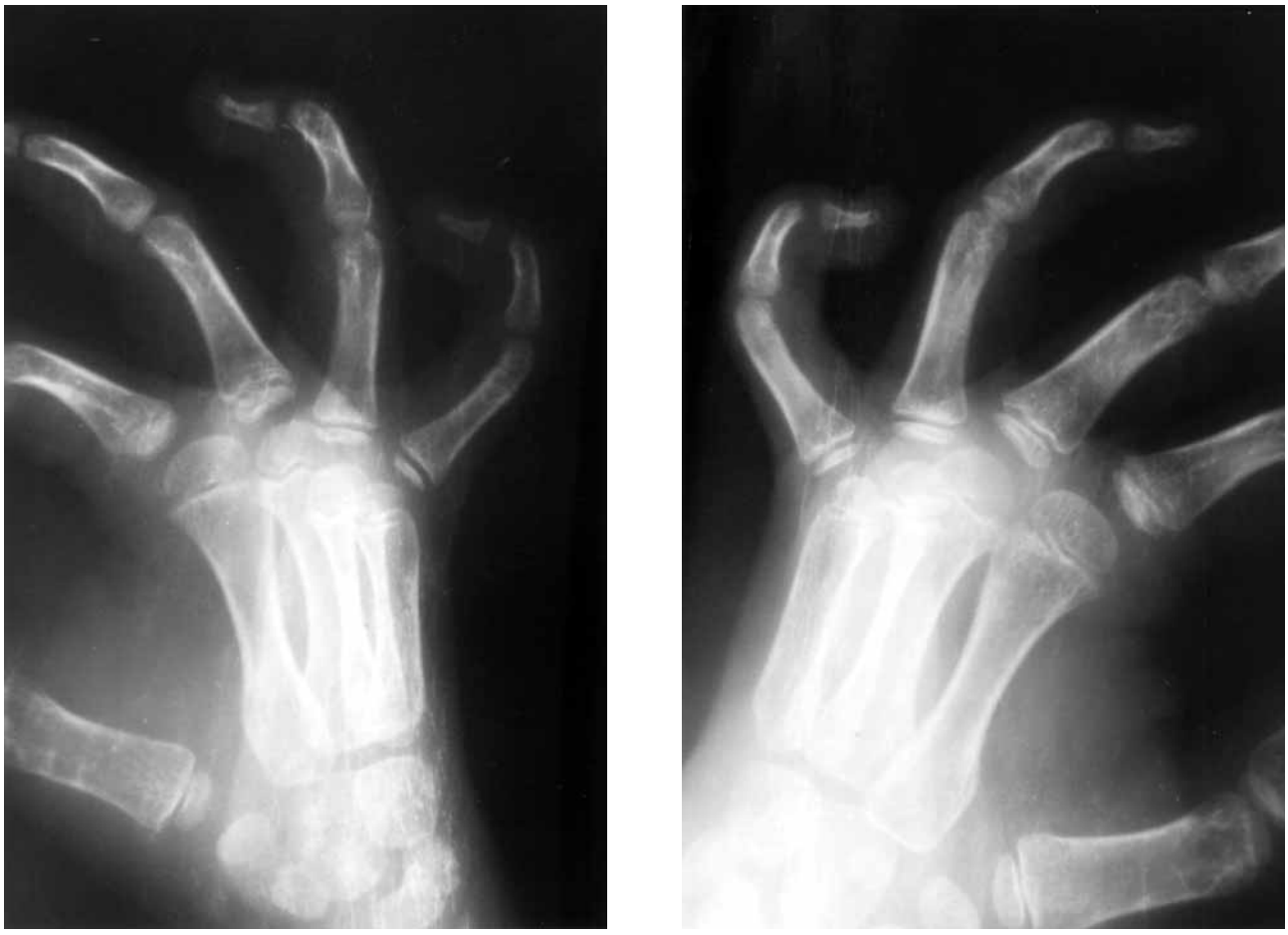

Fig. 2. — Oblique radiographs of the hands : flexion deformity in the right fifth finger(a) and left fifth finger (b).

hypoplasia in the trochanter minor and defective ossification at the ischiopubic junction. Furthermore, flat foot deformity and syndactyly in the feet may be present *(1,2,3)*. Our case was evaluated as distinct from these syndromes because it was not hereditary and the skeletal deformities were not typical.

Trisomy 8 is characterised clinically by a large head, speech delay, cleft palate, and patellar aplasia and hypoplasia *(1,5)*. In addition, it may be accompanied by psychomotor retardation, joint contractures, vertebral fusion, and congenital cardiovascular anomalies. While trisomy 8 was diagnosed by chromosome analysis, our case had none of the clinical findings mentioned above, other than patellar aplasia and hypoplasia, and, at variance with typical findings, had bilateral camptodactyly.

In Kuskokwin syndrome, one of the few known causes of patellar aplasia, there is contracture in a number of joints including the wrists, knees, and ankles, cyst formation in the clavicle and humerus, or congenital pseudoarthrosis, in addition to patellar aplasia or hypoplasia *(1,5)*.

Another syndrome that may include patellar hypoplasia is Coffin-Siris syndrome, in which nail aplasia and hypoplasia, radial head dislocation, mental retardation, coarse facial features, and sparse hair occur *(1)*. None of these other features were present in our case.

Although there is insufficient data available on treatment and outcomes in congenital patellar aplasia

and hypoplasia, it has been reported that no treatment is necessary in cases where the quadriceps muscle is sufficiently strong and there is no flexion contracture of the knee. In the light of these literature data, we carried out no treatment in our case *(1,2,3)*.

Although camptodactyly is a common anomaly, its pathological anatomy is still not fully known, and its treatment is disputed *(6,7)*. Furthermore, this deformity classically affects the proximal interphalangeal joint, and rarely occurs in the distal interphalangeal joint as in our case *(8)*. According to the literature, the initial treatment for camptodactyly should be conservative, and surgery should be resorted to only in cases that exhibit progression and do not respond to other treatment *(6,7)*. As in typical camptodactyly, our patient's finger flexion deformities were treated with exercise and splinting alone.

REFERENCES

1. **Azouz EM, Kozlowski K**. Small patella syndrome: a bone dysplasia to recognize and differentiate from the nail-patella syndrome. *Pediatr Radiol* 1997;27:432-435.
2. **Dellestable F, Pere P, Blum A, Gaucher A**. The 'small-patella' syndrome. *J Bone Joint Surg* 1996;78-B:63-65.
3. **Scott JE, Taor WS.** The "small patella" syndrome. *J Bone Joint Surg* 1979 ; 61-B : 172-175.
4. **Beals RK, Eckhardt AL**. Hereditary Onycho-Osteodysplasia (Nail-Patella Syndrome) : A report of nine kindreds. *J Bone Joint Surg* 1969 ; 51-A : 505-515.
5. **Bongers EM, Woods CG.** Evidence for genetic heterogeneity in familial ısolated patella aplasia-hypoplasia. *Am J Med Genet* 2002 ; 108 : 78-79.
6. **Hori M, Nakamura R, Inoue G.** Nonoperative treatment of camptodactyly. *J Hand Surg* 1987 ; 6 : 1061-1065.
7. **Siegert JJ, Cooney WP, Dobyns JH.** Management of simple camptodactyly. *J Hand Surg* 1990 ; 15-B : 181-189.
8. **Tachdjian MO.** *Tachdjian Pediatric Orthopedics.* Vol. 1. 2nd ed, WB Saunders Co, Philadelphia, pp 284-285.