Familial Absence of the Patella

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Bilateral absence of the patella as an isolated congenital abnormality is an extremely rare condition. The only previous report of a case we have been able to find was in a nine-year-old girl reported by Kutz in 1949. The family reported here is unique. Two members of the family have been found with absent patellae. Clinical examination failed to reveal any stigmata of hereditary osteo-onychodysplasia.

Case Report

A fourteen-year-old black boy was first seen in November 1972 because of a cystic mass in the anterior aspect of the right knee, which had been present for approximately one year and was reported to have disappeared on one occasion, but to have recurred. The patient was active in sports and the knee had begun to bother him in those activities.

His height was 153 centimeters and he weighed thirty-six kilograms. There was slight genu varum, and both patellae were absent (Fig. 1). Knee extension was 0 degrees bilaterally, and maximum voluntary flexion was 135 degrees. There was no lateral instability of the knee. There was a cystic mass lateral to the patellar tendon at the level of the knee joint, two centimeters in diameter, which was aspirated and yielded a clear, amber, gelatinous fluid. The elbows were normal as were the nails of the fingers and toes. The pelvis was palpated easily in this slender boy and there were no palpable iliac horns. The pupils were round and equal. The ankles were slender.

Roentgenographic examination of the knees confirmed the absence of the patellae (Fig. 2). Roentgenograms of the elbows were normal. Roentgenograms of the pelvis were negative for the presence of iliac horns.

The cyst refilled after each of two aspiration procedures and since the cyst was enlarging and interfering with the patient's activity, surgical excision was done. The hospital work-up

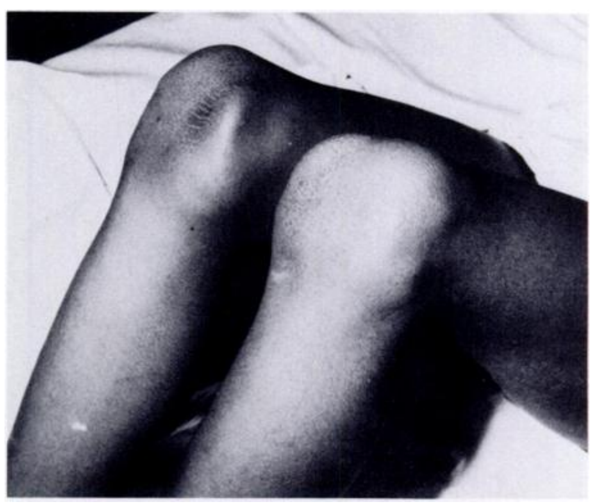

FIG. 1

Flexed knees. Note the prominence of femoral condyles due to absence of patellae.

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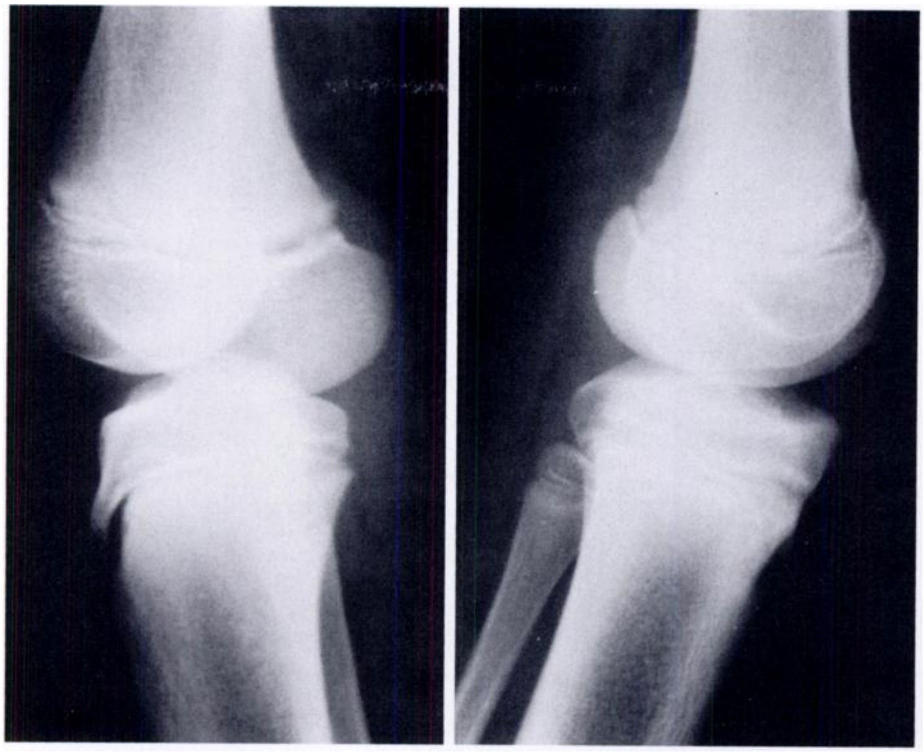

FIG. 2

Roentgenograms of patient's knees revealing absent patellae, but otherwise normal structures.

included urinalysis and hematological studies which were within normal limits. The serum calcium was 10.5 milligrams per 100 milliliters, and the serum phosphorus was 5.6 milligrams per 100 milliliters. The blood urea nitrogen was eleven milligrams per 100 milliliters and the serum creatinine, 0.7 milligrams per 100 milliliters. The liver function tests were normal. The alkaline phosphatase was markedly elevated at 424 milliunits per milliliter (normal, thirty to eighty-five). Postoperatively the value was 310 milliunits per milliliter. Roentgenograms were made of the hands and there was no evidence of hyperparathyroidism. No other manifestations of systemic disease were noted. No explanation of the hyperphosphatasemia was established. A chromosomal smear preparation was normal. The bone age was consistent with the chronologic age of the patient.

At surgical exploration the cyst was identified as a typical ganglion measuring 3.5 centimeters by two centimeters and filled with a straw-colored, clear fluid. The patellar tendon was found to be extremely wide and extended almost to the prominent edge of the lateral condyle. At its insertion into the tibial tubercle the tendon was four centimeters wide. The tendon was extrasynovial proximally to the level of the intercondylar femoral notch at which point the cyst originated. The stalk was excised at this point and the defect repaired. The postoperative recovery was uneventful. The pathological diagnosis was ganglion.

The patient's immediate family consisting of his parents and five brothers were then examined. To their knowledge no blood relative had any related knee problem nor any abnormality of their fingernails. The patient was next to the eldest child. The parents were found to have normal patellae and fingernails. Of the five brothers the second youngest, seven years old, was found to have absent patellae. He stood 117 centimeters tall and was well proportioned. His knees were stable and knee extension was complete and in flexion his heels reached his buttocks. The elbows were normal as were his fingernails and toenails. Palpation of the pelvis did not reveal any evidence of iliac horns. The patient had no clinical complaints. Roentgenograms of the knees confirmed the absence of the patellae.

Discussion

Hereditary osteo-onychodysplasia is reported to be inherited in an autosomal dominant pattern but this apparently is not the pattern of inheritance of the condition being described because in the family presented, both parents and three siblings were found to have normal patellae. Duncan, in his extensive review of families with hereditary osteo-onychodysplasia, found no case where the sole manifestation was the abnormality in the knee. In his corrected series of 120 cases of hereditary osteo-onychodysplasia, the incidence of patellar dysplasia was 93 per cent and total absence, 14 per cent. These findings were substantiated by Carbonara and associates.

It would appear unlikely that familial absent patellae is part of a hereditary osteo-onychodysplasia. It cannot be ascertained whether the pattern of inheritance is sex-linked or autosomal nor can it be predicted whether this mutation will be transmitted to other generations.

References

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