BILATERAL CONGENITAL ABSENCE OF THE PATELLA IN AN ADULT: CASE REPORT

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ABSTRACT

Bilaterally absent patella is a rare clinical entity. Most documented cases occur in association with Nail-Patella Syndrome. This patient presented at 60 years of age with severe bilateral knee arthritis and was found to have bilaterally absent patella which had previously been undiagnosed. The patient was managed with bilateral knee replacements. This presentation highlights the need for vigilance of the condition in patients presenting with knee arthritis.

Key words: Congenital absence patella knee arthroplasty

INTRODUCTION

The patella is a sesamoid bone found on the anterior aspect of the distal femur which contributes to the bones forming the knee joint (1). Congenital absence of the patella refers to a condition where an individual is born without the patella (2). It may be bilateral or unilateral (3). It is an extremely rare condition and when present mostly occurs as part of syndromes such as the nail patella syndrome, small patella syndrome, trisomy 8 mosaicism, and Meier-Gorlin syndrome (3,4). Limited data is available on congenital absence of the patella. Individual case reports highlight the rarity of the condition. No data on incidence and prevalence is available due to the small number of cases except where the patella was absent as part of a syndrome (1,5). Congenital absence of the patella was first reported by Scott and Taor *et al* (4) in 1979 where they described the congenital absence of the patella in a family of 12.

Congenital absence of the patella can occur unilaterally with a normal or small patella on the contra lateral side as described by Duygun *et al*(1) in their case presentation. Patients with an absent patella will also often present with other abnormalities in the same knee joint (4). These include recurrent knee dislocations, genu valgus, slip of the medial tibial plateau, hip and upper femur abnormalities (2,3,7). When occurring as part of a syndrome, congenital absence of the patella is also seen with systemic abnormalities such as thyrotoxicosis and renal insufficiency (6).

Nail patella syndrome which is an autosomal dominant hereditary disorder related to ABO blood group is the most described syndrome presenting with absence of the patella (6). It affects multiple systems with predominant involvement of kidney, bones, nails and eyes (4). It is estimated to roughly affect 1 in 50,000 newborns (3).

In the case presentation reviewed the patient underwent surgery to correct limb alignment together with physiotherapy and occupational therapy at a young age with favorable results (1, 6, 7). No case of bilateral absent patella has been reported before in Kenya. This could be attributed to low incidence as absence of the patella is a rare event (1).

CASE REPORT

A 60 year old female patient presented to the out-patient clinic complaining of severe pain in the knees for the past 10 years. The pain had been to this point successfully managed on over the counter pain medication including non-steroidal anti-inflammatory analgesics and acetaminophen. Pain had however increased recently to the point it had affected her activities of daily living. She revealed that she had always had difficulty extending the knees, a problem which had worsened over the preceding six months. There was no history of trauma. The patient reported that she had not been diagnosed with any knee problem previously. Her recollection of her early childhood was also non-revealing. Most recently she had visited an outpatient department where physiotherapy had been recommended together with X-ray imaging of the knees before referral to an orthopaedic surgeon.

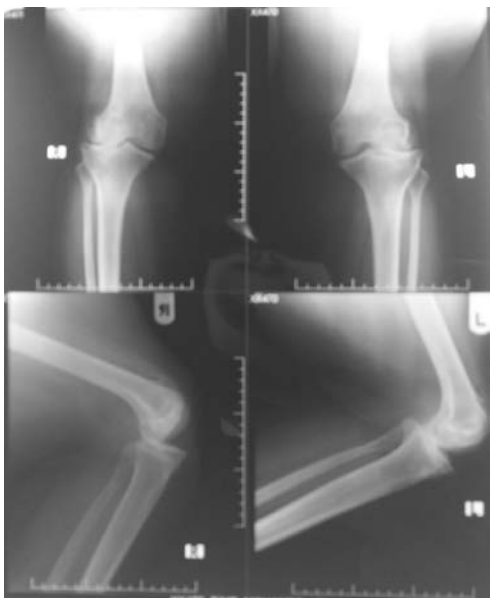
On examination we found a middle aged lady in good general condition. Her vital signs were within normal. She did not have any syndromic features. She had difficulty ambulating with minimal movement at the knee joints. Upon examination she was found to have bilaterally swollen knee joints which were warm and tender to touch. There were no overt skin changes. The patient had well developed musculature with an intact extensor mechanism. The patella in both knees could not be palpated. The knee alignment was normal with no varus or valgus deformity. She had a Q angle of 18 degrees. There was notable atrophy of the extensors bilaterally with reduced power. Extensor lag could not be effectively assessed due to pathology in both knees. Passive flexion was limited to 45 degrees with crepitus bilaterally. The patient had a negative anterior and posterior drawers test.

The patient's hip joints demonstrated normal range of movement and no arthritic changes could be detected. Her ankle joints were also normal. The rest of her musculoskeletal exam was also normal. Her cardiovascular, neurological and abdominal examinations were all normal.

The X-ray films of the knees were presented and they revealed osteoarthritic changes characterized by joint space narrowing and marginal osteophytes. Patellae in both knees were absent. In addition to the X-rays available, she was also sent for renal function tests, a full blood count and liver function tests. The results for these investigations were within normal parameters.

Figure 1

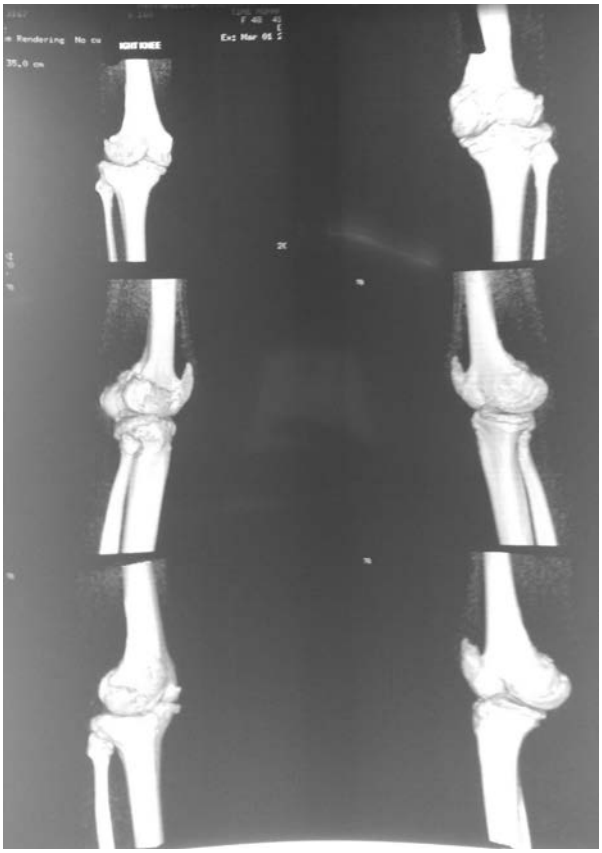
X-rays of both knees showing arthritic changes and absent patella bilaterally



An MRI was not immediately available therefore a CT scan was done to further assess the articular surfaces, collateral and cruciate ligaments as well as the menisci. In addition to the X- ray findings the 3D reconstructed CT scans revealed cartilage loss, meniscal changes but intact ligaments.

Figure 2

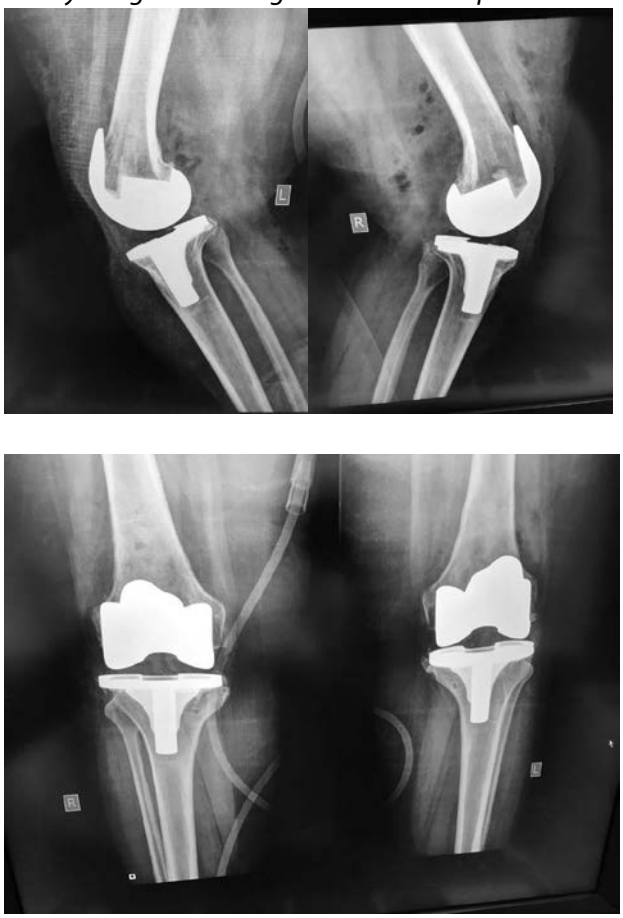
CT reconstruction of the knees



With the imaging and the patient's current quality of life in consideration, she was offered bilateral knee replacements which she consented to.

She underwent a total knee replacement with a bi-cruciate stabilized knee system distributed by smith and nephew®. Both knees were replaced in one sitting. The patient recovered well from the surgery. Flexion and extension post operatively was noted to be at 0 to 90 degrees with limited rotation. Both joints showed good stability withstanding significant distracting force. The patient was then taken for physiotherapy. Six weeks post-operatively, the patient developed adequate extensor strength and was able to resume near normal function and has currently been on follow up at the orthopaedic clinic for 3 years and is still asymptomatic.

Figure 3
X-ray images following bilateral knee replacement



DISCUSSION

Congenital absence of the patella is a rare disorder (1). Patients with the condition are usually noted to have other joint abnormalities (4) such as recurrent knee dislocations, genu valgus, slip of the medial tibial plateau, hip and upper femur abnormalities (2,4,7). A case of congenitally absent patella bilaterally discovered in late adulthood has not been documented in literature. Isolated bilateral absence of the patella is associated most commonly with a poorly developed extensor mechanism (8,9). This is evident in the patient presented who reported and exhibited difficulty in passive leg extension. In most cases, surgical intervention is required to replace the mechanism in the femoral groove (10-12). In the case presentation however the patient was able to function without surgery for a long duration. Late intervention however results in severe osteoarthritis of the joint (12, 13) as observed in this case.

The quadriceps muscle through the patella over the patella groove of the femur, gives both static and dynamic stability to the knee joint (10). In patients born without the patella, the quadriceps tendon glides unrestrained over the femoral groove (2), and as a result, the range of extension is limited and the rate of degeneration of the femoral condyles and contracture formation of the soft tissues is greatly accelerated (12,13). To limit this, different surgical approaches have been documented in literature to reduce deformity based on the findings clinically (2). The patient presented was however able to accomplish day to day activities without early surgical intervention as described in literature.

The patient underwent a bi-cruciate retaining knee replacement rather than traditional posterior cruciate retaining knee replacement. Twenty five percent of patients undergoing total knee replacement with the posterior stabilized knee report dissatisfaction with the results (14). This is presumed to be due to the altered knee kinematics with this implant (15). A bi-cruciate retaining implant has been shown to provide more natural knee kinematics (16) making it a suitable choice for a patient with pre-existing knee instability due to absence of patellae in-spite of the high failure rate of the bi-cruciate implant (17). Albeit with the attendant operational difficulties associated with the newer implant (17), the knee replacement was successfully done with the patient remaining asymptomatic on follow up.

CONCLUSION

Bilateral absence of the patella remains an extremely rare entry. Early identification of patients with bilateral absence of the patella can facilitate timely intervention which delays osteoarthritic changes in the knee. With late presentation the management of choice is total knee replacement.

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