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Congenital Absence of Patella: A Case Report

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Abstract | **Case Report**

Congenital absence of the patella is a rare orthopedic pathology characterized by underdevelopment or complete absence of the patella. This condition is very rare when isolated and is usually accompanied by other anomalies. ***Case Presentation***: We report here a case of bilateral congenital absence of the patella in a 6-and-a-half-year-old boy who presented with walking difficulties and genu valgum, with the incidental discovery of patellar agenesis without any deficit in joint mobility. Radiological and biological assessments confirmed patellar aplasia without evidence of an associated syndrome. Management: We have planned growth modulation for his genu valgum and a transfer of the anterior tibialis to the third cuneiform for his supinated foot, but no treatment is currently planned for the patellar agenesis due to the absence of symptomatic issues related to this agenesis.

Keywords: Congenital patellar agenesis, Bilateral patellar absence, Genu valgum, Supinated foot, Orthopedic management.

INTRODUCTION

The congenital absence of the patella is defined by the absence of a visible patellar ossification center on radiographs beyond the age of 4 years in girls and 6 years in boys. The diagnosis is based on standard radiography when this condition is suspected. However, it can be easily overlooked if one is not familiar with the various stages of bone growth and maturation in children.

The patella plays a crucial role in enhancing the efficiency of the quadriceps' extension capability, which is essential for maintaining normal gait and mobility [1]. Although the exact cause is generally unclear, genetic predisposition has been identified as a major contributing factor in many cases [2].

This anomaly can be unilateral or bilateral, isolated or associated with other orthopedic malformations, but the most common is congenital dislocation of the knee or hip, and anomalies of the femur and fibula, genu recurvatum, genu valgum, cubitus valgus, and pelvic anomalies [3].

CASE PRESENTATION

A 6-and-a-half-year-old boy presents with walking difficulties and frequent falls without any psychomotor delay. Examination reveals bilateral genu valgum, more pronounced on the left side, with full extension and flexion, and a left foot in supination (Figures 1 & 2).

Figure 1

Figure 2

On palpation, the patella is absent and the trochlea and femoral condyles are hypertrophied; however, the strength and tone of the quadriceps, gluteus maximus, and hamstrings were appropriate for the child's age (Figures 3 & 4).

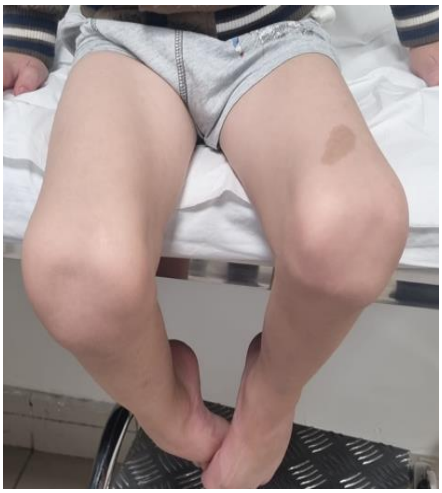

Figure 3

Figure 4

Standard radiography confirms the absence of patellae on both views (Figures 5 and 6). The biological assessment shows no particular abnormalities, and the abdominal ultrasound, conducted to search for associated visceral malformations, is normal.

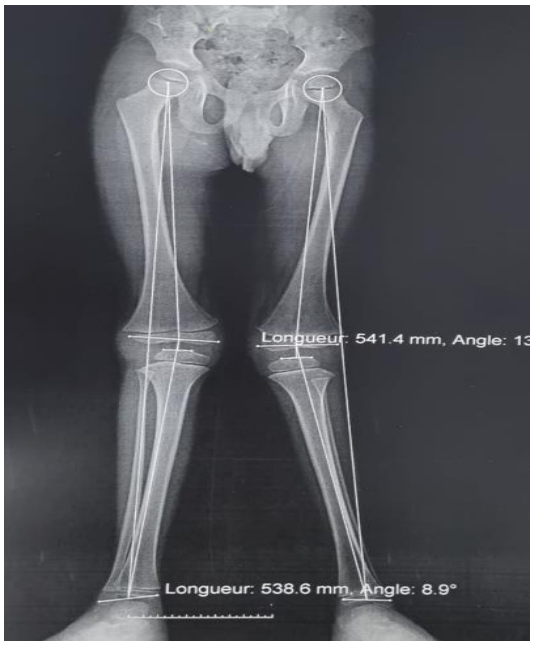

Figure 5

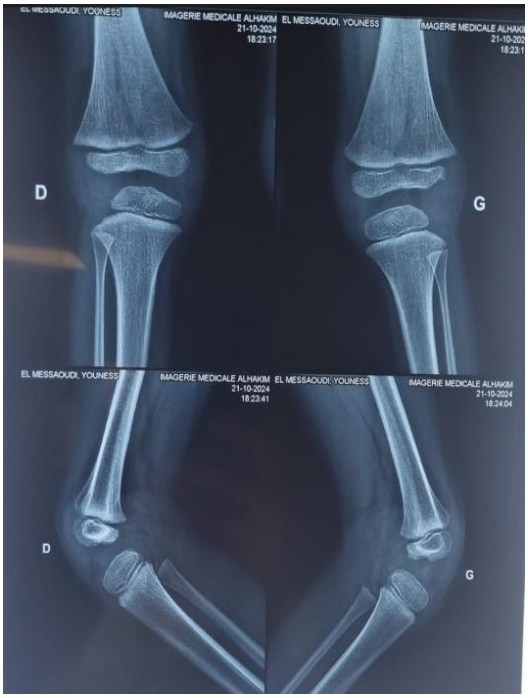

Figure 6

DISCUSSION

Congenital absence of the patella is a rare pathology that can be isolated or associated with a spectrum of other malformations.

Isolated patellar absence has been reported without the presence of other anomalies in only a few cases. The first study to report this pathology was conducted by Kutz [4] in 1949, who described a 9-year-old girl in whom the absence of the patella was discovered only after a routine physical examination.

The absence of the patella, particularly within the context of a syndrome, can be observed in association with renal insufficiency. Senguttuvan *et al*., [5] reported renal insufficiency in a 22-year-old patient with nail-patella syndrome. In the presented case, renal function

tests and renal ultrasound were negative. Nail-patella syndrome can also be accompanied by thyrotoxicosis [6]. Therefore, it is important to exercise caution regarding systemic issues.

In the skeletal system, radial subluxation may be observed [7]. Therefore, radiographs should be taken focusing on the elbow and other orthopedic issues; as in our case, where we found a genu valgum and an associated supinated foot.

The treatment of congenital absence of the patella depends on the degree of aplasia, the involvement of the extensor mechanism, and the impact on leg functionality. Non-surgical interventions include physiotherapy, the use of braces or an orthopedic device for the affected knee. Jerome *et al*., (2008) also agree that regular physiotherapy and stretching exercises can alleviate the deformity and improve the patients' gait [8].

In such cases, or when conservative treatments are ineffective, successful positioning of the mechanism in the groove between the femoral condyles, medialization of the tibial tuberosity, and the transfer of one or more medial hamstring tendons to the extensor mechanism are the preferred treatments, resulting in satisfactory functioning [9].

CONCLUSION

Congenital patellar aplasia is a rare pathology in the pediatric population, clinically diagnosed and confirmed by imaging.

It is frequently associated with other orthopedic anomalies and rarely isolated. The prognosis is generally good with conservative treatment in the majority of cases.

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