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Congenital Absence of Patella Presenting as Chronic Knee Pain in Adulthood: A Rare Delayed Presentation in a 30-Year-Old Male

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Abstract: ***Background**: Congenital absence (aplasia) of the patella is an extremely rare developmental anomaly usually diagnosed during childhood. Delayed presentation in adulthood is exceptionally uncommon. **Case Presentation**: A 30-year-old male presented with chronic right knee pain and inability to perform daily activities for six months. Clinical examination revealed valgus deformity with prominent femoral condyles. Musculoskeletal ultrasound demonstrated direct visualization of the distal femur and proximal tibia with absence of the patella. Plain radiographs confirmed complete absence of the patella. The patient had no history of trauma, surgery, or syndromic manifestations. **Conclusion**: This case highlights a rare delayed adult presentation of congenital absent patella and emphasizes the role of ultrasound as an initial diagnostic modality before radiographic confirmation.*

Keywords: congenital absent patella, adult knee pain, patellar aplasia, musculoskeletal ultrasound, rare case report

1. Introduction

Congenital absence of the patella is a rare developmental anomaly that may occur either in isolation or as part of syndromic conditions such as nail-patella syndrome, Meier-Gorlin syndrome, or small patella syndrome. Most cases are diagnosed in childhood because of gait abnormalities, extensor mechanism dysfunction, or visible deformity. Adult presentation is exceedingly rare.

The patella plays an important biomechanical role in improving quadriceps efficiency and knee extension mechanics. Absence of the patella may lead to altered biomechanics resulting in chronic pain, deformity, and functional limitation. We present a rare case of isolated congenital absent patella diagnosed for the first time in adulthood.

2. Case Presentation

A 30-year-old male presented with complaints of chronic right knee pain for the past six months associated with difficulty performing daily activities including prolonged standing, walking, squatting, and stair climbing.

The patient was apparently well since childhood and denied any history of trauma, surgery, recurrent patellar dislocation, inflammatory arthritis, or systemic illness. There were no known congenital anomalies or family history of similar complaints. No significant comorbidities were present.

On clinical examination, valgus deformity of the right knee was noted with prominent distal femoral condyles. The patella could not be palpated clinically. Mild functional limitation due to pain was present. Distal neurovascular examination was normal.

Imaging Findings

Musculoskeletal ultrasound examination of the anterior knee demonstrated direct visualization of the distal femur and proximal tibia without the normal intervening patella. Absence of patellar bone and patellar shadow in the expected anatomical location was noted.

Subsequent radiographic evaluation with anteroposterior and lateral radiographs confirmed complete absence of the patella with associated valgus deformity and prominent distal femoral condyles.

3. Discussion

Congenital absence of the patella is exceptionally rare and is usually associated with syndromic disorders. Isolated absent patella diagnosed in adulthood is extremely uncommon. Most reported cases are identified during infancy or childhood because of gait abnormalities or extensor mechanism dysfunction.

Our case is unique because the diagnosis remained undetected until 30 years of age and ultrasound served as the initial imaging modality suggesting the diagnosis before radiographic confirmation. Very few similar cases of delayed adult presentation have been described in the literature.

Differential diagnoses include post-traumatic patellar loss, previous patellectomy, severely hypoplastic patella, and chronic patellar dislocation. However, the absence of trauma or surgical history along with imaging findings supported the diagnosis of congenital patellar aplasia.

4. Conclusion

Congenital absence of the patella presenting in adulthood is extremely rare. This case highlights that isolated patellar aplasia may remain undiagnosed until adulthood and present as chronic knee pain with deformity. Careful clinical examination combined with ultrasound and radiographic evaluation can establish the diagnosis. Awareness of this rare entity is

important to avoid misdiagnosis and guide appropriate management.

Patient Consent
Written informed consent was obtained from the patient for publication of clinical details and imaging findings.

Conflict of Interest
The authors declare no conflict of interest.

Funding
No funding was received for this study.

Figures

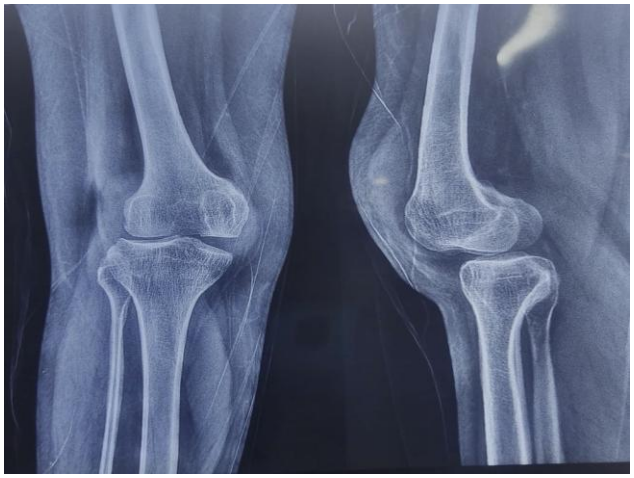

Figure 1: Plain radiographs demonstrating complete absence of the patella.

Figure 2: Clinical photograph demonstrating prominent femoral condyles and altered knee contour.

Figure 3: Ultrasound image demonstrating direct visualization of femur and tibia with absence of patella.

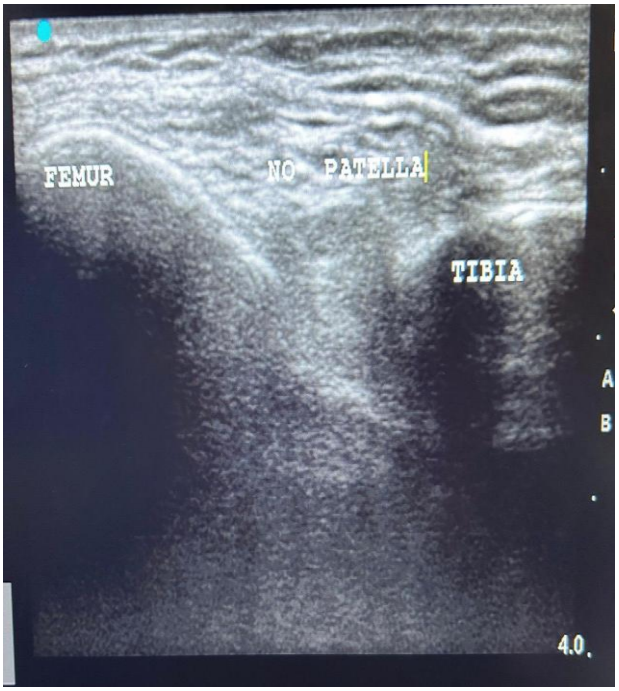

Figure 4: Standing clinical photograph showing valgus deformity of the lower limbs

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