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Small patella syndrome - A case report with review of literature

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Abstract

Small patella syndrome (SPS) or Hypoplastic patella syndrome (HPS) is a rare autosomal dominant disorder due to mutations in the TBX4 gene, characterized by the absence or hypoplasia of the patella with various anomalies of the pelvis and feet. We describe a case of a patient with hypoplastic patella of both knees presenting with pain and difficulty in walking; he was managed conservatively with non-steroidal anti-inflammatory drugs and an extension brace for three weeks followed by physiotherapy focusing on range of motion and quadriceps strengthening exercise.

Keywords: Small patella syndrome, Hypoplasia, Nail-patella syndrome, Patella aplasia-hypoplasia, Ischiopatella dysplasia, Ischio-pubic-patella syndrome or Scott-Taor syndrome, Hypoplastic patella syndrome, Ischiopubic synchondrosis

Introduction

Small patella syndrome (SPS) or hypoplastic patella syndrome (HPS) is a rare autosomal dominant disorder due to mutations in the TBX4 gene [1], characterized by the absence or hypoplasia of the patella with various anomalies of the pelvis and feet; these may include hypoplasia of the ischium, irregular ischial ossification at the ischiopubic synchondrosis, flat-foot or club-foot deformity, and brachydactyly. The syndrome was first described by Scott and Taor [2] in 1979. SPS should be clinically differentiated from disorders with ahypoplastic patellae, in particular the autosomal dominant disorders isolated familial patella aplasia-hypoplasia (PTLAH) syndrome [3,5,6,7] and the more severe nail-patella syndrome (NPS) [4]. The latter is caused by mutations of the LMX1B gene on chromosome 9q34 [4]. Recently, a locus for PTLAH has been identified on chromosome 17q21-22 [5]. Clinically patients of SPS may have bilateral small patella with lateral subluxation of patella.

Case report

A 28 -year-old male patient presented with left knee pain with limping after a trivial injury due to slipping on the stairs. On physical examination, mild swelling and tenderness were identified in the medial aspect of the left knee. The power of quadriceps muscles was intact but he complained of pain during flexion-extension. The flexion was restricted up to 900 and there was extension lag of 100. Patellofemoral mal-tracking was not seen but the patellae in both knees were much smaller than normal.

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Plain radiographs showed small patellae with abnormal morphology in both knees. Further investigation was done with magnetic resonance imaging (MRI).

MRI finding of an axial fat-suppressed T2-weighted image showed small patellae (2.0×0.6×1.5 cm in size) in both knees with slight lateral displacement, thick eccentric non-ossified patellar cartilage with central high signal portion, and elongated medial patellofemoral ligament. Also, the trochlear groove was shallow (trochlear depth, 0.2 cm), suggestive of trochlear dysplasia. Normal morphology of the medial and lateral facets on the posterior surface of the patella was not seen.

Patient was managed conservatively with non-steroidal anti-inflammatory drugs and an extension brace for the symptomatic left knee for three weeks followed by physiotherapy focusing on range of motion and quadriceps strengthening. During this period, the patients were started on isometric quadriceps exercises, as well as analgesia and electro-stimulation. Passive mobilization of the knee during the patients' fortnightly visit. Weight bearing was allowed after three weeks with the help of a walker. The exercises for gain of range of motion were increased in the third week, with the introduction of the stationary bicycle without load. Initial proprioception and closed kinetic chain exercises were executed and gradually evolved into open chain exercises.

Discussion

SPS or HPS should be differentiated from disorders with a/hypoplastic patellae, including NPS and isolated familial PTLAH. In NPS, patellar aplasia or hypoplasia is associated with nail anomalies, deformation or luxation of the head of the radius resulting in impaired mobility of the elbow, iliac horns and often nephropathy. Nailhypoplasia or dysplasia and absent or hypoplastic patellae are essential features for the diagnosis. Posterior iliac horns are pathognomonic for NPS, but reported to be present in only 70% of cases. Various other skeletal anomalies, including pes equinovarus, dislocated hips, and contractures of major joints have been described

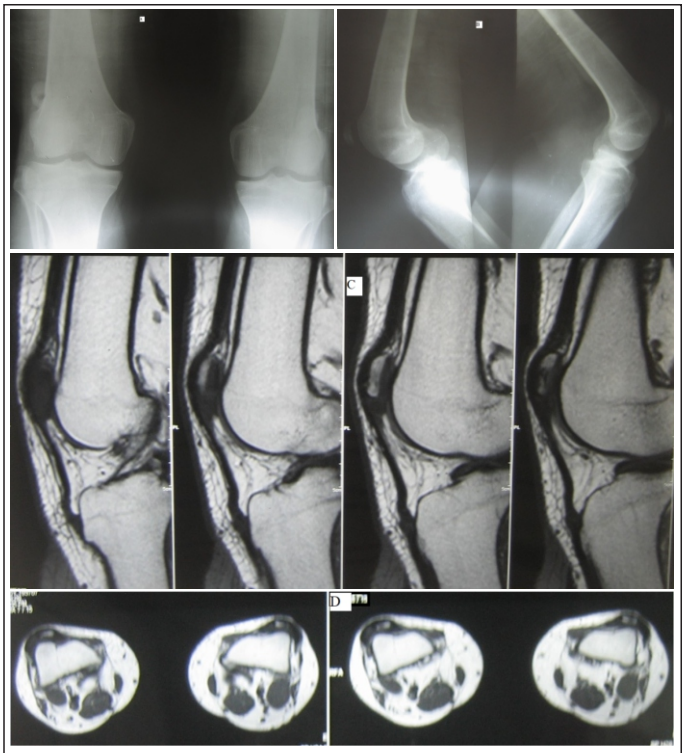

Figure 1: [A] X-ray Anterior posterior view of knee joint, [B] X-ray lateral view of knee joint, [C] MRI of knee joint sagittal section, [D] MRI of knee joint transverse section, [E] Clinical photograph

in NPS but do not contribute to the diagnosis. This skeletal dysplasia results from mutations in the LMX1B gene[8]. In patients with PTLAH, patellar aplasia or hypoplasia is an isolated anomaly without additional radiological or clinical features infra-acetabular notches have never been mentioned in PTLAH [9-14]. Pelvic anomalies, including anomalies of the ischiopubic joint, narrowed femoral necks, coxa valga and coxa vara, and hypoplastic lesser trochanters are features of PTLAH. On the other hand, foot anomalies seen in SPS include widened first web space between toes, flat-foot deformity, brachydactyly, and tarsal coalition. Some specific bony changes consistent with the diagnostic features of SPS were identified in the patient's knee, pelvis and foot. Increases in the web space between the first and second toes were observed along with pes planus and short fourth and fifth rays on the foot radiographs [3]. In the present case we observed only bilateral small patella and lateral subluxation of patella along with pes planus. In a series reported by Guidara KJ et al. [15], approximately 50 percent of children with nail-patella syndrome underwent for knee surgery to treat instability. SP Shaving hypoplastic patella is of minor importance and requires no specific treatment [19–22]. The major decision-making lies in the treatment of any disability about the knee and it depends primarily on its chief cause, for example, genu recurvatum, instability, lateral dislocation, discontinuity of the extensor mechanism [15–18] needs surgical intervention and correction. But in this case, we managed the patient with conservative approach using medication, rest along with physiotherapy and the final outcome was satisfactory.

Small patella syndrome is rare condition, but should be considered as a differential diagnosis in patients with knee pain without any obvious etiology. Imaging studies become necessary to diagnose this condition and non-operative treatment can be considered an option before embarking on to surgical interventions.

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