Case report:

Magnetic Resonance Image Findings in A Rare Case of Bilateral Knee Pain in A Small Patella Syndrome Patient

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Abstract:

Congenital aplasia or hypoplasia of the patella can manifest as an isolated disease or more commonly part of rare genetic disorders such as small patella syndrome (SPS). To our knowledge this is the first report of a patient with small patella syndrome who complained of bilateral knee pain with no preceding injury. Magnetic resonance imaging (MRI) revealed early degenerative changes of the knee joints which had not been mentioned in any literature. The MRI findings in our case are important as it would help in finding the cause of the pain and allow early treatment to be initiated thus delaying the progression of the arthritic changes.

Keywords: Small patella syndrome; patella; magnetic resonance imaging; osteoarthritis; congenital

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Introduction:

Small patella syndrome (SPS) is a rare autosomal dominant disorder[1,2]. It is characterized by small or absent of uni /bilateral patella with abnormalities seen at the pelvis and feet that include hypoplasia of the ischium, irregular ossification at the ischiopubic junction, infra-acetabular axe cut notches and flat feet [1,2]. The presentation of congenital partial or total absence of patella varies from recurrent patella dislocation in younger patients to knee pain due to severe arthritic changes in elderly[3]. We report a magnetic resonance imaging findings of a 12-year-old girl complained of having bilateral knee pain who has bilateral absent of patella with underlying small patella syndrome. To our knowledge this is the first case report of MRI findings in a SPS patient with missing patella in both knees.

Case Report:

We report a case of 12-year-old female who presented with gradual onset of bilateral knee pain associated with weakness of knee extension that was more severe on the right lower limb for a month duration. She denied having any preceding trauma and fever. However, she was able to participate in normal school physical education activities involving running and jumping. There was no significant antenatal history and the milestone is normal. On physical examination she does not exhibit any syndromic features and stands almost the same height as the parents. Her gait pattern revealed a shortened stance phase over right lower limb and excessive swinging of bilateral hip forward to aid in full extension of knee. She was unable to squat unsupported and stand from squatting position without assistance. With

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palpation we detected the absence of bilateral patella with prominent femoral condyles. There was no demonstrable localized bony tenderness. Both knee extension mechanism was impaired, more profound on the right knee. Systemic examination revealed short bilateral 4th and 5th toes, flat feet, bilateral sandal gap deformity and generalized ligamentous laxity evidenced by Beighton score of 6/9. Plain radiographs demonstrated complete absence of bilateral patella, with genu valgus, dysplastic fibula head and hypoplastic lateral femoral condyle over both knees. Pelvic radiograph revealed absent ossification at ischiopubic junction and presence of infra-acetabular axe cut notches bilaterally. Symptomatic treatment for the analgesics and regular physiotherapy were minimally effective thus we decided to proceed with MRI. The MRI revealed absent of bilateral patella with hypoplastic quadriceps muscles and tendons. There was absent of the normal configuration of vastus lateralis and intermedius with bulky vastus medialis seen at the anteromedial compartment. The tendons are continuous with a thin, fascial like structure that inserts at the anterior aspect of the proximal tibia. There was present of bilateral trochlear dysplasia, more severe on the left knee. Interestingly the patient had features of early arthritis changes manifested by chondral loss worst involving the lateral tibial plateau. The lateral supporting structures like iliotibial band, lateral collateral ligament and biceps femoris show chronic sprain and strain changes. The parents refused to undergo further genetic and chromosomal analysis tests. However, from our observation the features of this patient are consistent with small patella syndrome.

Discussion:

Small patella syndrome is a rare autosomal dominant disorder, first described in 1979 by Scott-Taor is also known as coxo-podo-patella syndrome, Scott-Taor syndrome, ischio-patella dysplasia or ischio-pubic-patella syndrome. It is characterized by total absent or hypoplastic patella and abnormalities seen at the pelvic bone and feet [1,2,4-6]. Scott-Taor revealed 12 members from a family with either absent of small patella were seen to have abnormalities at the pelvic girdle and proximal femur[9]. This skeletal dysplasia disorder resulted from mutations in the TBX4 gene that causes impairment in the early and later stages of the limb development leading to the skeletal deformities[2].

Most of the patients would complain of having

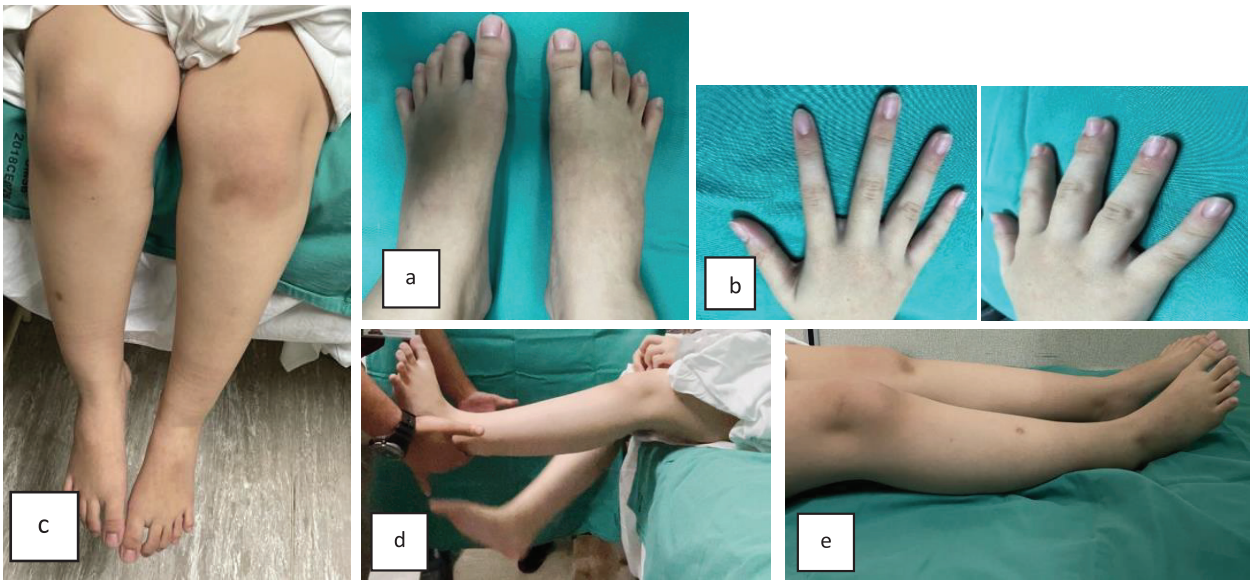

Figure 1 (a) Both feet exhibit widening of the first web space (sandal gap deformity) and relative short fourth and fifth toe. (b) There is no nail dysplasia at the hands which is the salient features in a nail patella syndrome (c) Active flexion of the knees are normal. Clinically there is prominent lateral parapatellar guttering possibly due to the hypoplastic lateral condyle of the femur and vastus lateralis muscle (d) Weak extensor mechanism was present bilaterally but more profound over the right knee (e)30 degrees extension lag were also observed over bilateral knees.

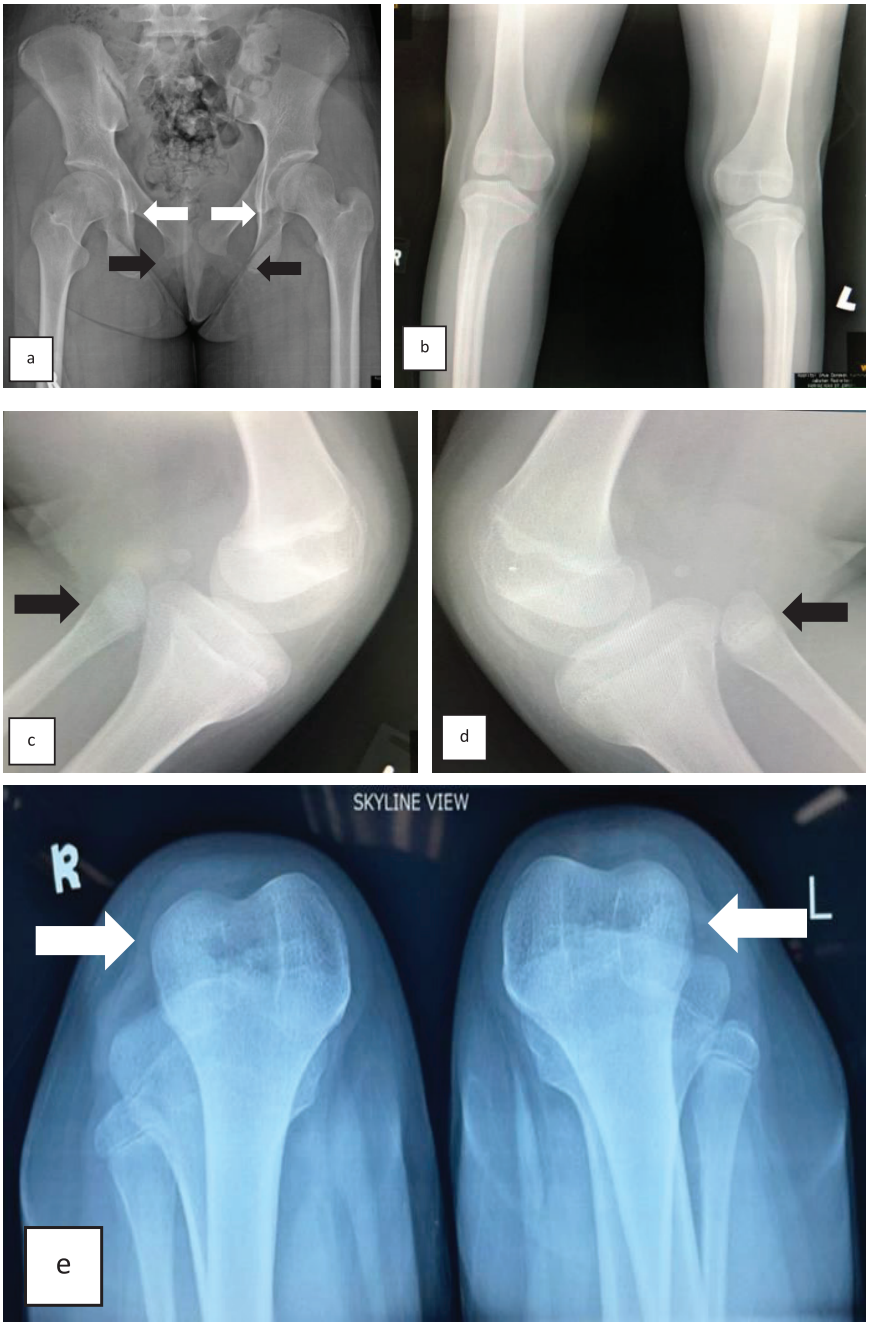

Figure 2 (a) Anteroposterior plain radiograph of the pelvis demontrates absence of the ossification of bilateral ischiopubic junction (black arrow) and presence of infra-acetabular axe cut notches (white arrow) (b) Bilateral genu valgus is shown from the anteroposterior view of the knee radiograph (c), (d) lateral views of of right and left knee show missing patella, different sizes of the condyles of the femur and dysplastic epiphysis of the proximal fibula (black arrow) (d) skyline view of the knee demostrate absent of patella and smaller lateral condyle of the femur(white arrow).

recurrent or habitual patella dislocation, muscle weakness of the lower limbs, inability to run and stand up from squatting position[6]. Late presenters tend to have knee pain secondary to recurrent patella dislocation since childhood while majority of the cases were asymptomatic[6].

Clinicians need to recognize the clinical features and radiological abnormalities that can differentiate this disease from other syndromes such as nail patella syndrome or autosomal-dominant mode patella aplasia-hypoplasia (PTLAH) that can also present with absent or hypoplastic patella[2,4]. In 1995 Kozlowski K et al described two sporadic cases of SPS who had features of facial dysmorphism including low set and posteriorly angulated ears, low nasal bridges, anteverted nares with flat nostrils, long philtrum-high palate, prominent lower lip and micrognathia[7]. Other recognizing features are short fourth and fifth rays of the feet, flat feet and wide sandal gap between the first and second toes[1,2,4-9].

Radiologically, these patients found to have bilateral or unilateral hypoplastic or absent patella with dysplastic knee changes, pelvic and foot abnormalities. From a plain pelvic radiograph, the patients would exhibit salient features of SPS such as absent, delayed or irregular ossification of ischiopubic junction and / or infra-acetabular axe cut notches. Other findings that are present in some of the patients are coxa vara /valga, apparent hypoplasia of the lesser trochanter, elongated femoral necks and flattened and widened proximal femur epiphysis[1,2,4-9]. A plain knee radiograph would

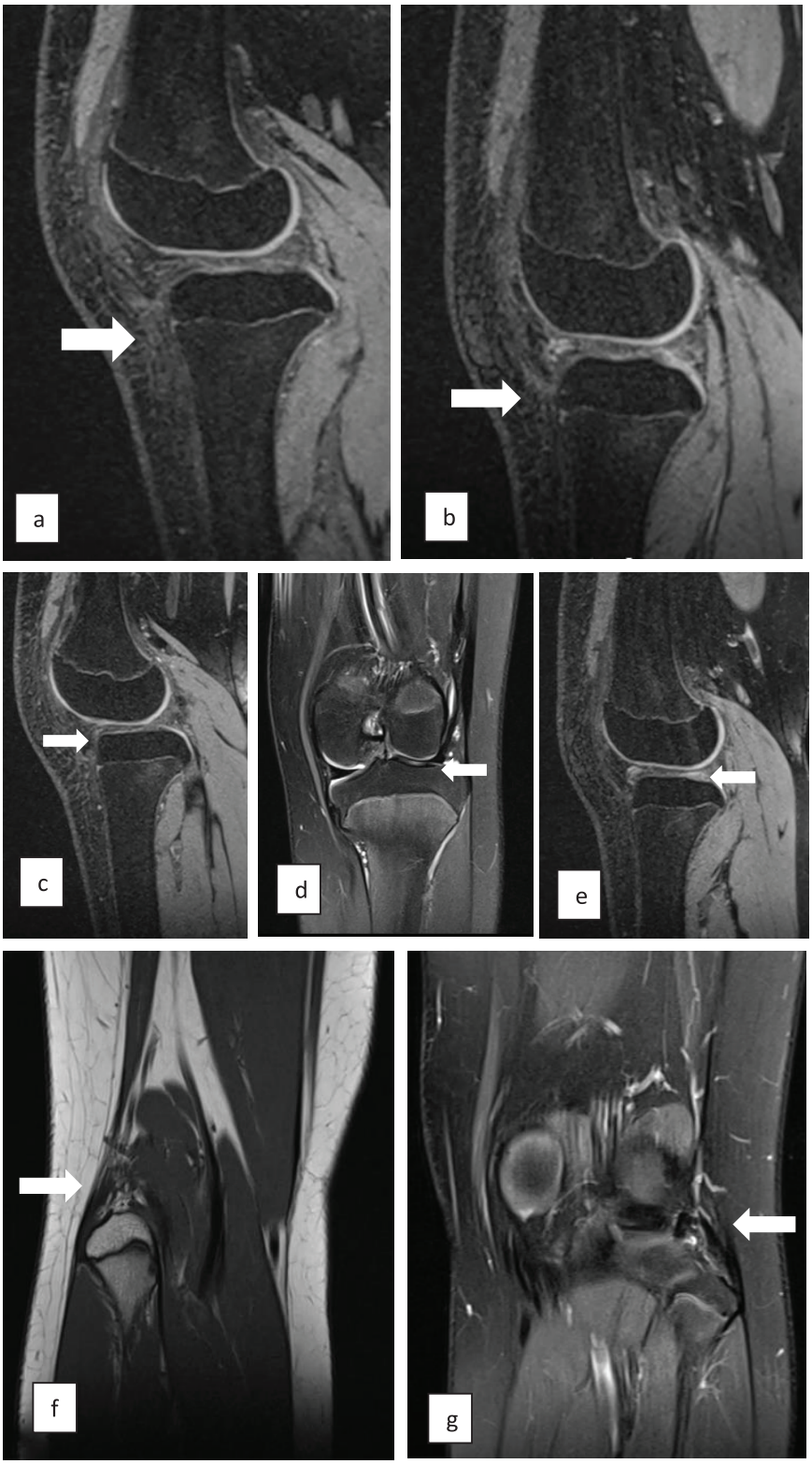

Figure 3 Sagital view of T2-weighted MRI image of the right knee (a) and left knee (b) demonstrate total absence of patella. The quadriceps tendon is seen. Distally it continues as a thin fascia like structure that is inserted at the anterior knee. Patella tendon is not visualized, most likely hpoplastic (white arrow). (c) and (d) Thining of the cartilage was observed at the lateral tibial plateu of the right knee at both sagittal and coronal view T2-weighted image of the right knee and sagittal view of the left knee (e) (white arrow). This may represent early degenerative changes of the knees. (f) Thickened lateral collateral ligament is visualized at it's fibular insertion seen at coronal view of T1-weighted image of the right and coronal view T2-weighted image of the left knee (g) (white arrow). Intermediate signal intensity is also observed indicating chronic sprain that might contribute to the knee pain.

reveal absent or small patella. Other abnormalities that possibly can be observed are subluxated or dislocated patella, small medial femoral condyle and lateral tibial plateu, dysplastic fibular head, flattening of the femoral intercondylar notch and trochlear dysplasia[4-6,8,9]. From a foot radiographs pathognomonic findings that are suggestive of SPS include widened first web space, short fourth

and fifth metatarsal bones and pes planus[1,4-6,8,9]. Dellestable F et al described several findings that were found in a foot radiograph in minority of the cases that comprise of talocalcaneal coalition, ball and socket ankle joint, hypertrophy of neck of talus and calcaneal exostoses[10].

Kim HS et al in 2016 reported magnetic resonance imaging findings in a 26-year-old small patella syndrome patient who complained of left knee pain after slipped and fell during walking. The MRI demonstrated a ruptured medial head of gastrocnemius tendon with presence of thick eccentric non-ossified patella cartilage[2]. Our patient had an unusual presentation of sudden and unprovoked bilateral knee pain that responded partially to conservative treatment. Surprisingly at a very young age the magnetic resonance imaging that was performed on her demonstrated early arthritic changes of the knee bilaterally. Early intervention for her is important as it can delay the degenerative process and prevent the likelihood of having premature arthroplasty surgery.

The outcome of patients with SPS was never been discussed previously. Usually, this syndrome would rarely lead into major disabilities. The symptoms generally would improve with regular physiotherapy and analgesics[10]. However surgical treatment was advocated in few cases with isolated congenital absent of patella. Varghese et al in 2007 reported a surgical procedure with good clinical outcome for a patient with aplasia of patella with defective extensor mechanism. The patient underwent femoral shortening and restoration of the knee extension by suturing the distal stump of the femoris to the semitendinosus tendon[11]. Hamstring lengthening procedure can be performed in patient with knee flexion contracture[10,12]. Khaimov M et al in 2018 reported two cases of brothers with congenital absence of patella due to underlying familial patellar absentia syndrome. 40 years after the first diagnosis both of them presented with severe degenerative osteoarthritis. The patient underwent bilateral total knee replacement and had excellent post-operative outcome[3]. As for our patient, we informed the patient and her parents regarding the possible severe degenerative changes that can happen to the knees. We also emphasize on the importance of being compliant to the physiotherapy and avoidance of strenuous activities that can propagate the degenerative process.

Conclusion:

Magnetic resonance should be considered as a part of investigating tools to evaluate the possible causes of knee pain in patients with underlying small patella syndrome. At the same time early treatment can be initiated and probable outcome can be predicted and informed to the patients who have early osteoarthritis changes in the knees. Although most patients respond well to conservative treatment, timely intervention can delay the arthritic changes. Total knee arthroplasty is largely considered in cases with late presentation and severe degenerative changes.

Reference:

1. Bongers EM, van Kampen A, van Bokhoven H, Knoers NV. Human syndromes with congenital patellar anomalies and the underlying gene defects. *Clin Genet*. 2005;**68**(4):302-319. doi:10.1111/j.1399-0004.2005.00508.x
2. Kim HS, Yoo JH, Park NH, Chang JH, Ban YS, Song SH. Magnetic Resonance Imaging Findings in Small Patella Syndrome. *Knee Surg Relat Res*. 2016;**28**(1):75-78. doi:10.5792/ksrr.2016.28.1.7
3. Khaimov M, Fakhoury J, Karim F, Bitterman A, Alpert S. Total Knee Arthroplasty in Patients with Familial Patellar Absentia Syndrome: A 40-Year Update: A Case Report. *JBJS Case Connect*. 2018;**8**(4): e100. doi:10.2106/JBJS.CC.17.00329
4. Bongers, E. M., Duijf, P. H., van Beersum, S. E., Schoots, J., Van Kampen, A., Burckhardt, A., Hamel, B. C., Losan, F., Hoefsloot, L. H., Yntema, H. G., Knoers, N. V., & van Bokhoven, H. (). Mutations in the human TBX4 gene cause small patella syndrome. *American journal of human genetics*, 2004;**74**(6):1239–1248. https://doi.org/10.1086/421331
5. Scott, J E, and W S Taor. "The "small patella" syndrome." *The Journal of bone and joint surgery*. 1979;**61**(2): 172-5. doi:10.1302/0301-620X.61B2.438269
6. Bongers, E M et al. "The small patella syndrome: description of five cases from three families and examination of possible allelism with familial patella aplasia-hypoplasia and nail-patella syndrome." *Journal of medical genetics* .2001; **38** (3): 209-14. doi:10.1136/jmg.38.3.209
7. Kozlowski, K, and J Nelson. "Small patella syndrome." *American journal of medical genetics* 1995;**57:** : 558-61. doi:10.1002/ajmg.1320570408
8. Bilgic, Serkan et al. "Bilateral absence of patella." *Congenital anomalies* vol. 2010;**50**: (1): 71-4. doi:10.1111/j.1741-4520.2009.00263.x
9. Dellestable, F. et al. "The 'small-patella' syndrome. Hereditary osteodysplasia of the knee, pelvis and foot." *The Journal of bone and joint surgery*. British volume 78 1 (1996): 63-5. https://doi.org/10.1302/0301-620X.78B1.1136
10. Jerome, J Terrence Jose et al. "Bilateral congenital absence of patella." *Indian journal of orthopaedics* 2008;**42**(2): 228-30. doi:10.4103/0019-5413.40264
11. Varghese, Renjit A, and Benjamin Joseph. "Congenital aplasia of the patella and the distal third of the quadriceps mechanism." *Journal of pediatric orthopedics*. 2007; **16** (5): 323-6. doi: 10.1097/01.bpb.0000243828.22552.74
12. Duygun, Fatih et al. "Isolated unilateral absent patella and contralateral small patella." *BMJ case reports* vol. 2013 bcr2013200353. 3 Sep. 2013, doi:10.1136/bcr-2013-200353