Isolated Congenital unilateral Aplasia of Patella with contralateral Hypoplasia of Patella-A Case Report.

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ABSTRACT

Congenital absence of patella(aplasia) or small size patella(hypoplasia) is very rare and usually associated with other anomalies like Nail Patella Syndrome, Ischiopatellar dysplasia and Turner-Kiesser syndrome. This condition can have diverse clinical manifestations. Congenitally absent patella is usually bilateral. The diagnosis of patellar aplasia and hypoplasia is often delayed because the patella is cartilaginous from birth till third and sixth years of age when it starts ossifying. We present a very unique and extremely rare case report of an isolated congenital absent patella on the right side and small sized patella on the left side in an adolescent and which was not associated with any syndrome.

Keywords: Nail patella syndrome, Patella aplasia, Patella hypoplasia

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INTRODUCTION

Congenital absence of patella(aplasia) can be unilateral or bilateral.[1] It is a rare anomaly and most cases occurs in association with Nail Patella syndrome, Small Patella Syndrome, Meirer-Gorlin Syndrome and Trisomy 8 Mosaicim.[1,2] Other abnormalities like recurrent knee dislocation, Genu Valgus, slip of medial tibial Plateau, hip and proximal femur anomalies can be found in patients with absent patella or hypoplastic patella.[1-3] The exact prevalence of aplasia or hypoplasia of the patella is unknown.[4] The most frequently described syndrome in the literature which is associated with absent patella is the Nail Patella syndrome.[5] This syndrome has an autosomal dominant inheritance with a frequency of 1 in 50000 live births.[1,5] It affects kidneys, bones nails and eyes.[2]

We reported a very rare case of Isolated congenital unilateral aplasia of Patella with contralateral hypoplasia of Patella in a 13 year old girl.

CASE REPORT

A 13 year old girl was brought to the Orthopaedic out patient department(OPD) of Punjab Rangers Teaching Hospital Lahore with the complaints of difficulty in walking and frequent falls for many years which had worsened for the last six months. History revealed that patient was a full term and delivered by caesarian section without any complication. All her other siblings were normal and healthy. Her developmental mile stones were normal and she started walking by the age of 1 year. Parental consanguinity was absent and there was no family history of any such problem. On physical examination the patient had multiple hyperpigmented patches around the knees and ankles which were asymptomatic and appeared suddenly two years back following fever. Local examination showed that the

inter condylar sulcus was empty on the right side with no patella while small hypo-plastic patella on the left side with normal medial and lateral mobility was noted. Patient had well developed musculature bilaterally with normal intact extensor mechanism on the left and and intact but week extensor mechanism(assisted) on the right.(Fig.I,Fig.II)

Gait analysis revealed varus thrust on right side. Patient was able to sit and squat but had hands on knee posture while standing. Valgus stress test was negative bilaterally while varus stress test was positive on right side.No flexion deformity was observed. No limb length discrepancy was noted. (Fig.III,Fig.IV)

The antero-posterior and lateral knee radiographs confirmed absent patella on right side and a small sized or hypo-plastic patella on the left side.(Fig.IV,Fig.V) MRI was normal except absent medial patellofemoral ligament on the right side.Complete blood count and other blood parameters were within the normal range. Abdominal ultrasound showed no renal anomalies.Radiographs of pelvis and proximal femur were normal. Nails of the patient were normal and no other syndromic associations were observed clinically.Based upon the clinical findings and patient expectations and willingness we decided to treat the patient conservatively.We applied brace to the right knee joint and supervised physical theraphy was started for the left knee joint.The patient was examined at 6 months follow up with improvement in knee pain and bipedal independent mobility.The patient was instructed for regular follow up at 3 months interval.

Fig. I & II: Photographs showing normal extensor mechanism on the left side and week extensor mechanism (assisted) on the right side.

Fig III, Fig. IV: Standing phtographs of patient indicating no obvious limb lengthening discrepancy and knee flexion contracture.

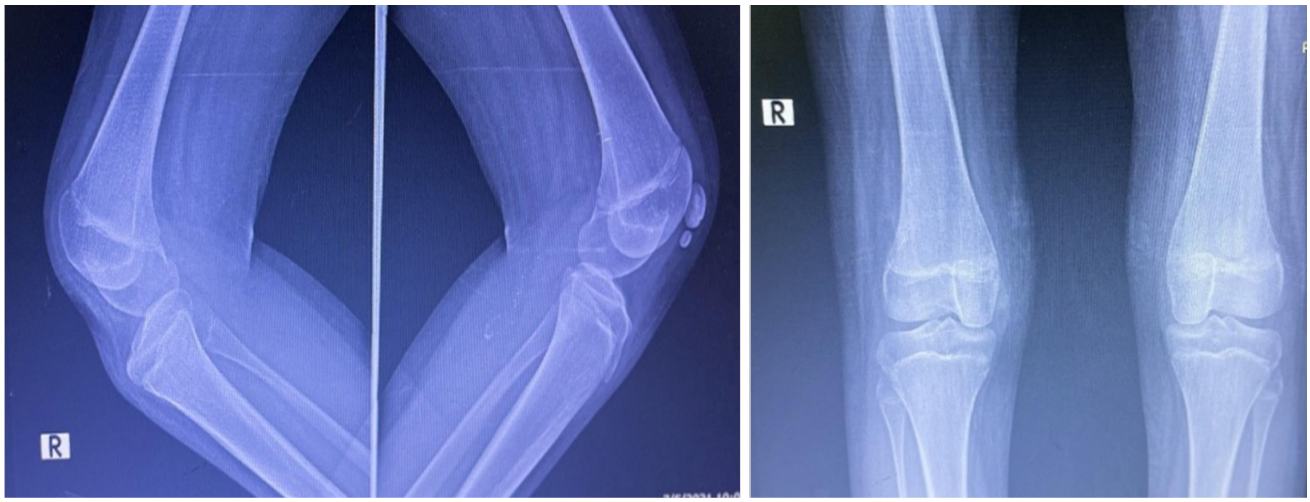

Fig. IV & V: Radiographs showing absent patella on right side and a small sized or hypo-plastic patella on left sideR

DISCUSSION

Aplastic or hypoplastic patella was first reported by Scott and Taor in 1979.[2] These authors noted that 12 members in one family had small and absent patella but the patients had anomalies of the pelvis and hip region. Humzah and Soames[6] reported aplasia of patella on one side and patella alta on other side with Scoliosis in 85 years old cadaver. Aplasia of the patella can be associated with renal anomalies as reported by Senguttuvan[5] in his case report of Nail Patella syndrome. Patients with these anomalies may have thyrotoxicosis and radial head subluxation.[7,8] Through Clinical examination, radiographs and biochemical investigations are therefore mandatory to exclude these anomalies in patients with aplasia or hypoplasia of patella.

We found only one case report in literature by Duygun and Sertkaya[9] which was similar to our case report but with only few minor differences. These authors reported hypoplastic patella on right side and absent patella on the left side in an 18 year old boy. There was no extensor mechanism on left side with 15 degree contracture on left side while quadriceps strength on right side was good. MRI revealed bilateral meniscus tear. These authors treated their patient with bracing on the left side and physical therapy for the right side. We had also treated our case report conservatively and achieved good outcome at 6th months follow up. We however recommend longer follow up of such cases to document any complication or disability.

In literature treatment of such cases vary from bracing, physical therapy and surgery in the form of fractional hamstring lengthening for excessive flexion contracture if present.[1,2]

The differential diagnosis of congenital aplasia and hypoplasia patella include Nail Patella syndrome(NPS), Ischiopatellar dysplasia(IPD) and isolated familial patellar Aplasia Hypoplasia(Patellar Aplasia-Hypoplasia- PTLAH).The differentiation among these can be done by careful clinical examination and with relevant biochemical and radiological investigations. IPD and NPS can also be differentiated by conducting genetic studies of alleles.[10]

Cases of congenital isolated absent patella can be accompanied by absent or dislocated distal quadriceps extensor mechanism.[11-17] In these cases the restoration of extensor mechanism function can be achieved by placing it in between the intercondylar groves, medial transfer of tibial tuberosity or hamstring transfer to extensor mechanism.[18]

CONCLUSION

We reported an isolated congenital unilateral aplasia of Patella with contralateral hypoplasia of patella which is an extremely rare anomaly. Although it had not caused any major disability to the patient, the need for thorough clinical examination and relevant investigations to rule out any association with syndrome was mandatory. The focus of attention in cases of isolated hypoplasia and aplasia of patella should be on the integrity of extensor mechanism, flexion contracture and knee stability. Early diagnosis and longer follow up is needed in all such cases.

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REFERENCES

1. Sweeney, E., Hoover-Fong, J.E. and McIntosh, I. Nail-patella syndrome. In: Pagon RA, Bird TD, Dolan CR, Stephens K, Adam MP. eds. Gene Reviews™ [Internet]. Seattle, WA: University of Washington, Seattle, 1993–2003 [updated 28 Jul 2009].
2. Scott, JE, Taor WS. The small patella syndrome. J Bone Joint Surg Br. 1979; 61-B:172–175.
3. Varghese, R.A. and Joseph, B. Congenital aplasia of the patella and the distal third of the quadriceps mechanism. J Pediatr Orthop B. 2007;16(5):323–326.
4. Bongres EM, Van Kampen V, Van Bokhoven H, Knoers NV. Human syndromes with congenital patellar anomalies and the underlying gene defects, Clin Genet. 2005; 68(4):302–319.
5. Senguttuvan NB, Sivaraman A, Kandasamy D, Marimuthu K. Nail patella syndrome: a rare cause of renal failure in a young adult. Pan Afr Med J.2011;9:31-35.
6. Humzah MD, Soames RW, Munn S, et al. Absent patella and contralateral patella alta observed with scoliosis. Anat Anz. 1988;2013:63–67.
7. Haras B, Vulpoi F, Onose G. A case of nail-patella syndrome associated with thyrotoxicosis. J Med Life. 2012;2013:126–129.
8. Elliot KA, Elliot GB, Kindrachuk WH. The radial subluxation-fingernail defect-absent patella syndrome. Observations on its nature. Am J Roentgenol Radium Ther Nucl Med.1962;2013:1067–1074.
9. Duygun F, Sertkaya O, Aldemir C, Dogan A. Isolated unilateral absent patella and contralateral small patella. BMJ Case Rep. 2013; doi:10.1136/bcr-2013-200353.
10. Bongers EM, Van Bokhoven H, Van Thienen MN, Kooyman MA, Van Beersum SE, Boetes C, 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. J Med Genet 2001;38(3): 209-214.
11. Beguiristain JL, De-Pada PD, Barriga A. Nail-patella syndrome: long term evolution, J Pediatr Orthop B.2003;12(1):13-16.
12. Varghese R, Joseph B, Congenital aplasia of the patella and the distal third of the quadriceps mechanism, J Pediatr Orthop B;2007:16(5):323–326.
13. Dellestable F, Pere P,Blum A,Regent D, Gaucher A. The 'small patella' syndrome. Hereditary osteodysplasia of the knee, pelvis and foot, J Bone Joint Surg Br.1996; 78(1):63–65.
14. Letts M. Hereditary onycho-osteodysplasia (nail-patella syndrome). A three-generation familial study, Orthop Rev;1991:20(3):267–272.
15. Banskota K, Mayo-Smith W, Rajbandari S, Rosenthal DI. Case report 548: Nail-patella syndrome (hereditary onycho-osteodysplasia) with congenital absence of the fibulae, Skeletal Radiol. 1989;18(4):318–321.
16. Trinn C, Szoke B, Magyarlaki T, Turi S, Ormos J, Nagy J. Nail-patella syndrome: clinico-pathological characteristics. Orv Hetil.1996;137(41):2253–2256.
17. Duncan JG,Souter WA. Hereditary onchyoosteodysplasia: The nail patella syndrome.J Bone Joint Surg [Br];1963:45:242.
18. Jerome JT, Varghese M, Sankaran B. Congenital patellar syndrome. Rom J Morphol Embryol. 2009;50(2):291-233.