CASE REPORT

Isolated unilateral absent patella and contralateral small patella

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SUMMARY

Isolated bilateral absent patella is seen very rarely. It is generally encountered as a part of nail-patella syndrome. An 18-year-old male patient presented with left absent patella and right small patella.

BACKGROUND

Bilateral absent patella on its own rather than as a part of a syndrome has been previously presented. However, absence of the left patella together with right side small patella, not as part of a syndrome, is an extremely rare event that has not been previously reported.

CASE PRESENTATION

An 18-year-old male patient presented at our polyclinic with deformity in both knees, which was more extensive in the left knee, with pain and inability to actively raise the left knee. There was no family history of any such problem and it was revealed that he was the third of 4 children and

had been delivered by caesarean section at full term. The other siblings had no problems. The problems, particularly of pain and flexion contracture had increased after the age of 12 years.

Physical examination of the patient showed bilateral absent patella particularly evident on the medial femoral condyles of both sides. There was approximately 15° flexion contracture on the left side. On the right side, the quadriceps hamstring muscle strength was good but there was no knee extensor mechanism in the left knee. While the patient had active extension movement of the right knee, the left side could only be brought into extension passively. When the right knee extensor mechanism was in 80° flexion, there was lateral luxation. The results of full blood count and other blood parameters were normal. No kidney anomalies were determined by abdominal ultrasound. On radiographs, a small patella on the right and an absent patella on the left were observed (figure 1), bilateral medial meniscus tears were observed on MRI (figure 2) and flexion deformity of the left knee and

Figure 1 Small patella on the right side and absent patella on the left.

Figure 2 Bilateral medial meniscus tears.

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Figure 3 15° Flexion contracture in the left knee.

lateral luxation of the right side extensor mechanism were seen on photographs (figures 3–5).

TREATMENT

Treatment for these kinds of patients varies from analgesia and physical therapy to bracing and surgical choices.¹ As surgical treatment, the hamstring can be fractionally lengthened if there is excessive flexion contracture in the knee.² As the case presented here had neither excessive flexion contracture nor extensor mechanism in the left knee, bracing was applied to the left knee and physical therapy was applied to the right knee. Following the treatment, the patient did not feel the need for any support and was able to return to his normal life.

OUTCOME AND FOLLOW-UP

The patient received physical therapy for approximately 6 months. Following physical therapy, the pain in the left knee had decreased and the patient had reached a level where he was able to continue at school without any support.

DISCUSSION

Absent or small patella first examined by Scott and Taor,¹ 12 members of a family on 1979. It is generally encountered as part of a syndrome. Nail-patella syndrome, Meier-Gorlin syndrome, small patella syndrome and trisomy 8 mosaicism are just some of these.² Isolated bilateral absent patella was defined by Jerome *et al.*³ In a study of an 85-year-old cadaver, Humzah *et al.*⁴ determined absent patella on one side and patella infera and scoliosis on the other side. In the case presented, there was a small patella on the right side and complete absence of the patella and extensor mechanism on the left side.

Scott and Taor¹ reported a family with 12 members who had small patella and absent patella as part of a syndrome. These patients were seen to have abnormalities of the pelvis and upper femur.¹ There were no family members with such problems in the case presented.

Absent patella, particularly as part of a syndrome, may be encountered together with renal insufficiency. Senguttuvan *et al.*⁵ determined renal insufficiency in a 22-year-old male patient with nail-patella syndrome. In the case presented, kidney

Figure 4 Active extension in the right knee and only passive extension in the left knee.

Figure 5 Lateral luxation in the right knee at 80° extensor mechanism flexion.

function tests and renal ultrasound were performed was negative. Nail-patella syndrome may also be accompanied by thyrotoxicosis.⁶ Therefore, it is important for care to be taken with respect to systemic problems.

In relation to the skeletal system, radial subluxation may be seen.⁷ Therefore, radiographs should be taken oriented towards the elbow and other orthopaedic problems.

Learning points

- Patients with small patella and absent patella should be investigated as to whether they are part of a syndrome.
- Abdominal ultrasound and kidney function tests should be examined in respect of renal insufficiency in these patients.
- From an orthopaedic aspect, the stability of the extensor mechanism, flexion contracture and whether there are tears in the meniscus should be investigated.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Scott JE, Taor WS. The small patella syndrome. *J Bone Joint Surg Br* 1979;61-B:172–5.
- 2 Sweeney E, Hoover-Fong JE, McIntosh I. Nail-patella syndrome. In: Pagon RA, Bird TD, Dolan CR, Stephens K, Adam MP. eds. *GeneReviews™ [Internet]*. Seattle, WA: University of Washington, Seattle, 1993–2003 [updated 28 Jul 2009].
- 3 Jerome JT, Varghese M, Sankaran B, *et al.* Bilateral congenital absence of patella. *Indian J Orthop* 2008;42:228–30.
- 4 Humzah MD, Soames RW, Munn S, *et al.* Absent patella and contralateral patella alta observed with scoliosis. *Anat Anz* 1988;167:63–7.
- 5 Senguttuvan NB, Sivaraman A, Kandasamy D, *et al.* Nail patella syndrome: a rare cause of renal failure in a young adult. *Pan Afr Med J* 2011;9:31.
- 6 Haras B, Vulpoi F, Onose G. A case of nail-patella syndrome associated with thyrotoxicosis. *J Med Life* 2012;5:126–9.
- 7 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;87:1067–74.

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