

CASE REPORT

Congenital patellar syndrome

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Abstract

Congenital patellar syndrome is bilateral isolated absence of patella. Congenital patellar aplasia or hypoplasia associated with genetic disorders belongs to a clinically diverse and genetically heterogeneous group of lower limb malformations. Absence of patella as an isolated anomaly is extremely rare and we discuss such a case in a 9-year-old boy.

Keywords: congenital, patellar syndrome, rare, anomaly.

Introduction

The patella (knee cap) is the largest sesamoid bone of the skeleton that is formed and located within the tendon of the quadriceps femoris muscle. Its principal role is to facilitate the extensor function of the quadriceps muscle and to protect the ventral cartilage surfaces of the knee joint. Evolutionarily, the patella is considered as the last major osteological addition to the tetrapod knee. The importance of this unique sesamoid bone for the function of the knee is emphasized by its continued presence among a broad spectrum of tetrapod vertebrate species [1].

Congenital lower limb malformations without anomalies of the upper limb are estimated to occur in one of 10,000 human live births. Exact data on the prevalence rate of patellar aplasia or hypoplasia in live borns are not available and are difficult to obtain because the patellae are completely cartilaginous at birth and ossify from the end of the second year onwards [1]. They have an autosomal dominant mode of transmission with 100% penetrance and variable expressivity and linked on the chromosome with the ABO blood group gene. Surgical intervention should be timely and appropriate to ensure continued high levels of patient's activity, which again depends on the individual's need [2].

Patient, Methods and Results

Informed consent from the patient's father with proper ethical board clearance from the hospital was obtained in reporting this interesting case. A 9-year-old boy presented to our out patient department, with complaints of deformity in both knees, inability to straighten the knee, difficulty in standing, and walking. On further questioning, the parents gave the history of full term born, normal delivered child, with deformed knees since birth. Partially flexed knees noted at the time of birth, gradually worsened from the age of 1 1/2 years, when he started walking. Deformity was

more on the left knee, which made him difficult to walk. The patient's immediate family consisting of his parents and two brothers were then examined. To their knowledge no blood relative had any related knee problem or any abnormality of their fingernails. The patient was the youngest child. The parents were found to have normal patellae and fingernails.

On examination, there was bilateral absence of patella. Femoral condyles were prominent. Fixed flexion deformity in knee measuring 80 degrees on the left side and 30 degrees on the right side was noted. The power of quadriceps hamstrings and gluteus maximus were good. Nails were clinically normal (Figure 1).

No other deformity was present in the patient. Examination of upper limbs and other systems were clinically normal. His blood parameters were within normal limits. Ultrasound abdomen was normal. No renal abnormalities noted. Absence of both patellae and a hollow sulcus seen in between femoral condyles were the hallmark clinical features (Figure 2). Femoral condyles were large and prominent. Radiographs of both knees (Figure 3, a and b) showed absence of both patellae, and flexion deformity more on the left side.

Computerized Axial Tomography was done to measure the limb length discrepancy and to look for other associated disorders in both lower limbs. Limb lengths were equal on both sides. No femur or tibial anomalies were seen (Figure 4).

Left side fractional Hamstring lengthening was done with informed consent under spinal anesthesia. Suture removed after two weeks. Active and passive resisted physiotherapy were started after six weeks. Gait training was given. The patient was on regular follow up every two months a year. Patient is 13-year-old at the time of last follow up. He can now walk with a bipedal independent gait with minimal flexion at the left knee and mild equinus at ankle (Figure 5). He can also do his day today activities, play, squat, sit cross-legged and can lead a near normal life.



Figure 1 – Nail was normal.

Figure 2 – Clinical picture of the patient shows prominent femoral condyles with a hollow sulcus in between the condyles and bilateral absence of patella.

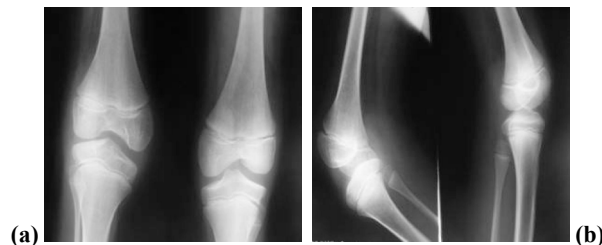


Figure 3 – (a), (b) Radiograph shows the absence of both patellae and large femoral condyles in antero-posterior and lateral views.



Figure 4 – CT-scanogram done to determine the limb length and other associated anomalies. There was no limb length discrepancy.



Figure 5 – Post-operative picture shows a minimal residual flexion deformity in the left knee and compensatory ankle equines.

Discussion

Isolated absence of patella is extremely rare [2]. It usually causes no disability to the patient. Congenital absence of patella [2, 3] is only one of several anomalies such as nail patella syndrome, dislocation of the knee, genu recurvatum, anomalies of the femur and fibula, clubfoot, or dislocation of the hip, dystrophy in thumb nails, bifid thumb nails, decrease in the length of nails, hypoplastic patella [3, 4], recurrent lateral dislocation of patella, genu valgum, slip of medial tibial

plateau, cubitus valgus, hypoplasia of elbow with decreased range of motion, iliac horns [4, 5], flaring of iliac crests with prominence of anterior superior iliac spines, pelvic abnormalities.

Congenital absence of patella without any other osseous anomaly is accompanied by agenesis of the distal third of the quadriceps muscle [6] or severe lateral dislocation of the extensor mechanism [5, 7–11]. In these instances restoring the continuity or placing the mechanism in the groove between the femoral condyles, transplanting the tibial tuberosity medially, and transferring one or more of the medial hamstrings tendons to the extensor mechanism usually results in satisfactory function. In a series reported by Guidera KJ *et al.* [4], approximately 50 percent of children with nail-patella syndrome underwent knee surgery to treat instability. It is this anomaly that should be treated; the absence of patella is of minor importance and requires no specific treatment [8–11]. 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 [4–7].

The identification of the molecular basis of monogenic syndromes with patella dysplasia has contributed to illuminating the role of the underlying genes in limb patterning. More than 35 dysmorphic entities with absent or small patellae are distinguished in the Winter-Baraitser Dysmorphology Database (WBDD), version 1.0.4, London Medical Databases. The molecular basis has been identified in only few of these. Human developmental syndromes with patellar malformations may be caused by single gene defects or result in some cases from mosaic trisomy 8 [1].

The asymmetrical dorso-ventral identity of the limb is specified by reciprocal interactions between limb bud ectoderm and mesenchyme with *Wnt7a* expressed in the dorsal ectoderm, *Lmx1b* expressed in the dorsal mesenchyme, and the homeobox containing transcription factor *Engrailed-1* expressed in the ventral ectoderm [12, 13]. *Wnt7a* induces the expression of *Lmx1b* giving rise to the formation of proximal and distal dorsal limb structures such as the patellae and the nails. *Lmx1b*^{-/-} mice develop zeugopods and autopods with a double ventral phenotype, ulnar reduction, and absent patellae [12]. Interestingly, *Lmx1b* expression has been found in patellar mesenchyme, which ultimately gives rise to the patella and patellar tendon. These findings are emphasized by the limb phenotypes of human syndromes, such as a gradual decrease of the severity of nail malformations in the antero-posterior direction in individuals with *LMX1B* mutation, short fourth and fifth metatarsals in individuals with mutations in the *TBX4* gene and *RECQL4*, gene which may have additional functions in regulating limb patterning and/or endochondral ossification processes of the developing skeleton [14, 15].

The knee joint is the largest and possibly the most complex synovial joint in the body. The quadriceps, anterior cruciate ligament (ACL) and posterior cruciate ligament (PCL) help maintain normal arthokinematics of the knee through the four bar linkage system.

The joint compressive load and quadriceps force significantly decrease total anteroposterior translation by as much as 50–66% in ACL-intact knees. The patellofemoral joint is a sellar joint between the patella and the femur. Stability of the patellofemoral joint is dependent on the passive and dynamic restraints around the knee. The primary dynamic restraint is the quadriceps muscles. This quadriceps is usually strong, and the extensor mechanism is well developed and glides in the patellar groove between the femoral condyles in congenital patella hypoplasia or aplasia [7–11].

Soft tissue release (hamstring release, lateral retinaculum release, medial reefing, full posterior capsule release), proximal and distal alignment procedures, quadriceps plasty femoral corrective osteotomy are the different surgical procedures described in the literature which again is indicated depending upon individual's clinical findings [4]. The main goal is to release soft tissue contractures and achieve normal alignment in the knee to promote patient's knee functional. Satisfactory results were obtained after surgery if planned early to correct the deformity, instability and maintain adequate knee alignment. Successful rehabilitation requires the clinician to understand and apply these biomechanical concepts, which can maximize patient function while minimizing the risk for further symptoms or injury [2–11].

Our patient had severe flexion deformity in his left knee, which made him difficult to walk. Often at times, he required support to walk, and climb stairs. Conventional hamstring lengthening helped this patient in correcting the flexion deformity. Per-operatively the flexion deformity was almost corrected with few degrees of residual flexion left (Figure 5). Patient was put on regular physiotherapy, which gradually straightened the knee without compromising the other muscle functions. No surgery was done for right knee. Deformity became supple after regular stretching exercises. Patient now walks with bipedal independent gait and a minimal flexion deformity in his left knee. We feel it is interesting to report an isolated congenital bilateral absence of patella as "congenital patellar syndrome".

☐ Conclusions

Congenital patellar syndrome is an extremely rare anomaly. For a complete understanding of the molecular mechanisms, underlying limb anomalies with no associated organ system manifestations, knowledge of the molecular targets of the transcription factor genes and the complex interaction of genes involved is essential. In addition, genetic modifiers, current knowledge about molecular pathways in patella development

in combination with traditional linkage studies and environmental factors need to be elucidated for understanding the great clinical variability of these anomalies. Congenital patellar syndrome causes no major disability to the patient and the treatment is always directed towards the associated anomalies and problems around the knee.

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