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Case Report

Pseudotumor Associated with a Bioabsorbable Screw in a Patient with Reconstructed Congenital Absence of the Anterior Cruciate Ligament: a Case report

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Abstract

Congenital absence of the anterior cruciate ligament (ACL) is an extremely rare condition associated with a wide spectrum of malformation. Here we describe a rare complication in a patient with congenital absence of the ACL after ACL reconstruction using a bioabsorbable screw. A 35- year-old woman presented a right knee mass that had been slowly growing for several months. Five years previously, she experienced acute right knee pain, locking, and instability after hiking. Images and diagnostic arthroscopy at that time revealed an absence of the anterior cruciate ligament, a hypoplastic lateral distal femoral condyle, a stenotic intercondylar notch, and hypoplastic posterior cruciate ligament along with a bucket handle tear of the medial meniscus. A right anterior cruciate ligament reconstruction was performed, and she did well for the next five years without knee joint instability until she presented a mildly painful subcutaneous pretibial soft tissue mass. Imaging studies demonstrated a 2.4 cm subcutaneous lobulated soft tissue mass protruding from the expanded tibial tunnel. The mass was excised, and the histology showed a solid and cystic lesion composed of a histiocytic and foreign body giant cell reaction to the degraded polymer along with spheres of calcium phosphate particles. At a two-year follow-up after debridement, the patient reported an overall improvement without any knee instability or local recurrence. To the best of our knowledge, this is the first report of a pseudotumor developed after ACL reconstruction in a patient with a congenital absence of the ACL.

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Abbreviations: ACL, anterior cruciate ligament; ACLR, anterior cruciate ligament reconstruction; PCL, posterior cruciate ligament.

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Introduction

First described in a radiographic study by Giorgi in 1956,[1] congenital absence of the anterior cruciate ligament (ACL), also known as ACL aplasia or agenesis, is an extremely rare condition with the estimated prevalence of 17 per million live births.[2] It has been reported as an isolated defect but is more commonly associated with other lower limb defects, particularly around the knee. This includes the absence of the posterior cruciate ligament (PCL), fibular hemimelia, congenital femur shortening, and knee dislocation.[3–5] Congenital absence of the bilateral ACL/PCL has also been reported to be associated with multiple organ syndromes, such as thrombocytopenia and absent radius (TAR) syndrome.[6] Furthermore, there is little understanding of the etiology or underlying pathogenesis. The condition is generally considered to be a sporadic disorder with a few cases reported to have a familial disposition.*7* A novel copy number variation involving CEP57L1 gene deletion has been recently identified in both mother and daughter with a bilateral absence of ACL/PCL.[8]

Clinically, the impact on daily life in patients with congenital absence of ACL is not well understood. Based on a review of previous publications, 31% of patients start experiencing pain and instability due to mild trauma.[9] Isolated case reports have also indicated that patients with this disorder might progress to earlier knee osteoarthritis.[10,11] Given the rarity of the condition and wide spectrum of malformation, there is still no consensus regarding the optimal treatment.[9] While selecting treatment options is required on a case-by-case assessment, many studies have reported clinical improvement after anterior cruciate ligament reconstruction (ACLR) for symptomatic patients.[12–14] Here we describe a rare complication in a patient with leg length discrepancy and congenital absence of the ACL after ACL reconstruction using a bioabsorbable screw (part of the case was previously reported).[13]

Case report

A 35-year-old woman presented a painless right knee mass that had been growing slowly for several months. Five years previously (in 2013), she experienced acute right knee pain, locking, and instability after a hiking injury. A physical exam at that time demonstrated patellar hypermobility and apprehension. Past medical history was significant for a leg length

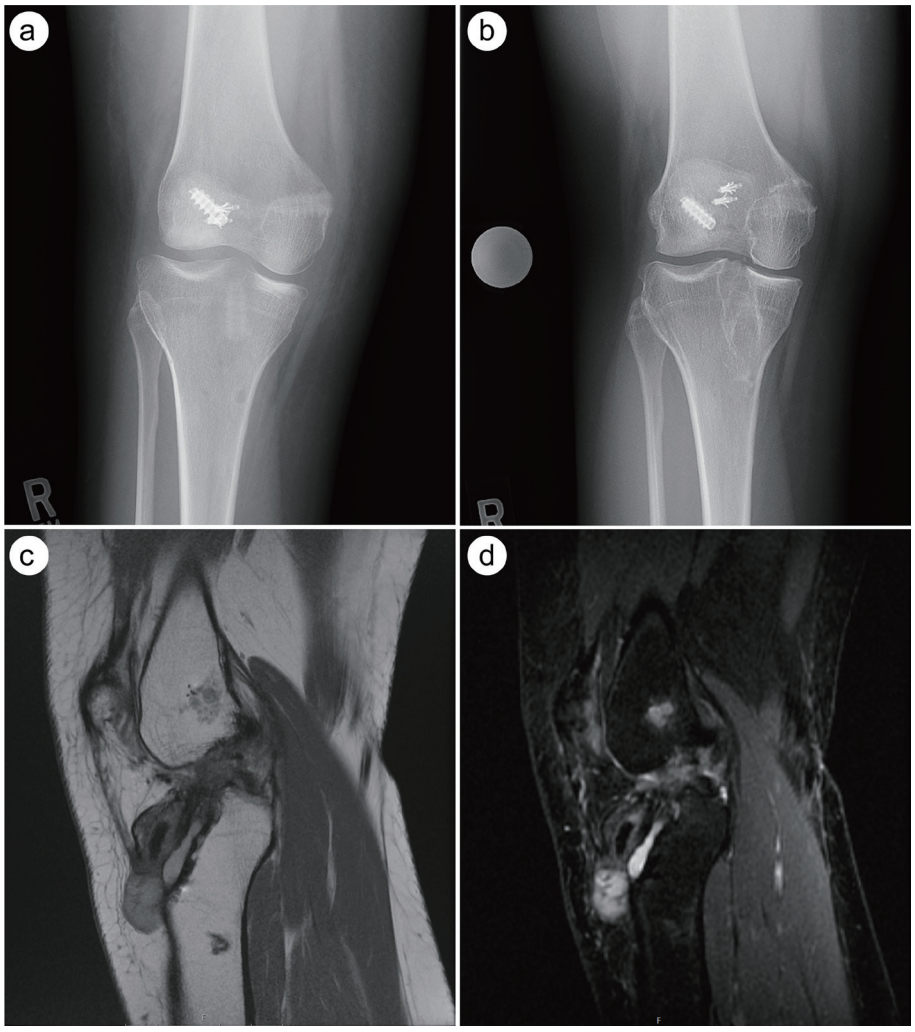

Fig. 1. Radiographic features. (a) ACL reconstruction with metallic femoral interference screw and bioabsorbable tibial screw in 2013. (b) Five years later (in 2018), the AP X-ray showed an expansile lytic intraosseous lesion within the tibial tunnel. Notice the larger tibial plateau and hypoplastic lateral femoral condyle, which are associated with the congenital absence of the ACL. (c, d) The MRI demonstrated a subcutaneous lobulated soft tissue mass protruding from the caudal margin of the expanded tibial tunnel measuring 2.4 cm × 1.8 cm × 1.4 cm.

discrepancy treated at the age of 9 years with epiphysiodesis. As previously reported,[13] imaging and diagnostic arthroscopy at that time revealed an absence of the ACL, a hypoplastic lateral distal femoral condyle, a stenotic intercondylar notch, and hypoplastic PCL along with a bucket handle tear of the medial meniscus. Subsequently (within two months), she underwent knee surgery at our hospital that included ACL reconstruction using an Achilles tendon allograft through an 11-mm bone tunnel with proximal interference screw fixation and a distal, 10 × 25mm polymer screw composed of 70% of Poly-L/D-lactide (PLDL) and 30% of biphasic calcium phosphate ("BioComposite", Arthrex, Naples, Florida, USA). Simultaneously, she underwent medial patellofemoral ligament reconstruction using a tibial tendon allograft fixed on the patellar side with suture anchors and on the femoral side with a bioabsorbable interference screw in a 10-mm diameter bone tunnel. Postoperatively she did well, and at one year had a full range of motion and had returned to recreational activities (Fig. 1a). She did well until several months prior to her current admission when she developed a mildly painful mass over her pretibial area. There were no clinical signs of infection, and the erythrocyte sedimentation rate (ESR) and C-reactive protein (CRP) were within the reference ranges. An anteroposterior radiograph (Fig. 1b) demonstrated a slightly expanded tibial tunnel with a minimal effusion. Notably, the right knee joint showed an apparent larger lateral tibial plateau and a hypoplastic lateral femoral condyle. Moreover, magnetic resonance imaging of the right knee showed an intact intra-articular anterior cruciate ligament graft. There was a 2.4 cm (craniocaudal) × 1.8 cm (transverse) × 1.4 cm (anteroposterior) extraosseous extension of a lobulated septate complex solid and partially cystic soft tissue mass extending out of the caudal margin of the tibial tunnel into the anterior subcutaneous soft tissue (Figs. 1c, d). The mass was centered at the level of the patellar tendon insertion at the tibial tubercle. The tibial tunnel was expanded measuring 1.4 cm (transverse) × 1.4 cm (anteroposterior). After discussion with the patient, the mass was carefully dissected around its edges down to its stalk that was found to extend into the tunnel. The reconstructed ACL was intact. The mass was removed, and the contents of the tunnel were debrided down to bleeding bone. An allograft bone dowel was soaked

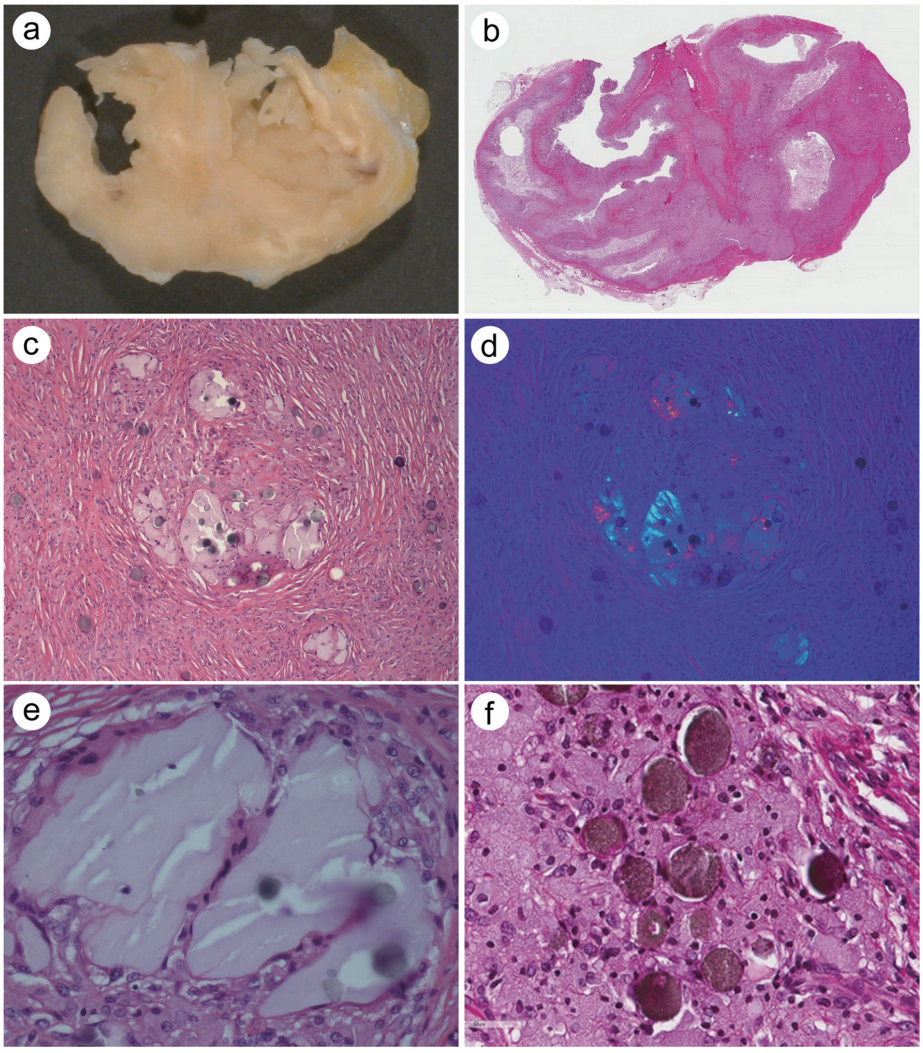

Fig. 2. Histologic characteristics. (a) Gross image of the well circumscribed tan partially cystic lesion. (b) Low-power view of a lobulated, solid, and cystic mass. (c-f) The mass was composed of a histiocytic and foreign body giant cell aggregation separated by fibrous tissue. These histiocytes had foamy cytoplasm and degraded polymers (refractile in d) and calcium phosphate particles.

in the previously prepared bone marrow aspirate concentrate and impacted into the tibial tunnel.

Grossly, the mass was well-circumscribed, tan, partially solid and cystic (Fig. 2a). The tissue specimen was fixed in formalin, routinely processed, and was found to be composed of a histiocytic and foreign body giant cell reaction to the amorphous, refractile material consistent with the residual polymer. Spheres of mineral, consistent with the calcium phosphate additive to the polymer screw, were also present (Figs. 2b–f). There was no evidence of osteoconductive bone formation associated with the spheres of calcium phosphate.

At a two-year follow-up since the excision of the mass, the patient reported overall improvement without knee instability or local recurrence. She was also scheduled to have surgery to repair her leg length discrepancy (Fig. 3).

Discussion

The primary finding of this study was that the pretibial pseudotumor following ACLR with a tibial absorbable interference screw was due to a histiocytic and foreign body giant cell reaction. This appeared to have occurred in response to the polymeric material in this bioabsorbable screw. Even at five years following the initial ACL reconstruction, remnants of the polymeric material still remained. This led to the development of a pretibial pseudotumor at this relatively late time point after ACLR with this particular absorbable interference screw. Furthermore, there was no evidence of osteoconductive activity. Rather, there was tunnel expansion, which was likely due to osteoclastic bone resorption. The reason why the bioresorbable material was not integrated in this patient is unclear. Over the years, many different kinds of bioresorbable screw fixation materials have been tried with different efficacy and reliability. Some have exhibited better bone osseointegration and osteoconduction, which have assisted in the migration and proliferation of bone marrow derived mesenchymal stem cells. Thus, additional research would be needed.

The bioabsorbable screw materials included biodegradable polymers, such as poly (glycolic acid) PGA, poly-L-lactic acid (PLLA), and poly-D, L-lactic acid and an osteoconductive compound, such as calcium carbonate or trimethylene

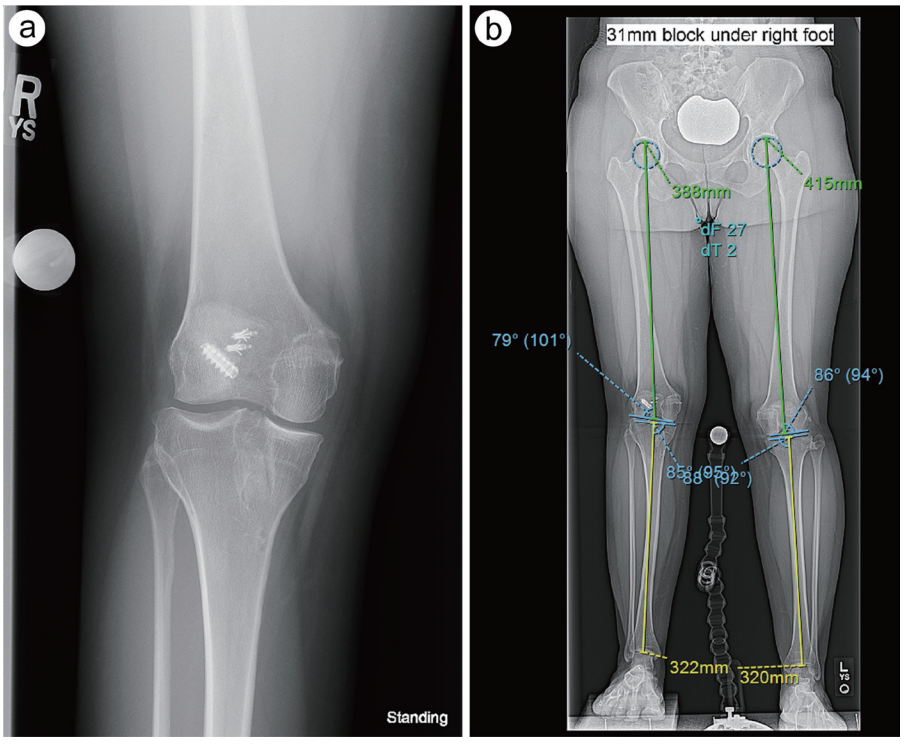

Fig. 3. Follow-up study. (a) Two years after the tumor excision, the AP radiograph showed prior ACL reconstruction. (b) Standing bilateral hip-knee-ankle film showed the valgus alignment of both lower extremities with leg length discrepancy with the right femur being 27 mm shorter than the left femur.

carbonate, or calcium phosphates, such as beta-tricalcium phosphate (β-TCP) and hydroxyapatite (HA) in order to promote bony ingrowth. In our case, a 10 × 25mm polymer screw composed of 70% of PLDL and 30% of biphasic calcium phosphate ("BioComposite", Arthrex) was used. The interference screw fixation using bioabsorbable implants had become the most common method of tibial graft fixation in a 2016 population-based epidemiologic study on 21,686 ACLRs performed from 2007 to 2014.[15] The outcomes based on the early revision rates have been similar. However, complications related to the bioabsorbable implant use have been found to be higher, especially in pediatric and adolescent athletes[16] with the most common surgical indication related with pain at the tibial screw site (4.9%). Up to a 5% incidence of tibial and pretibial cyst formation after ACLR with bioabsorbable interference screw fixation was reported, and the histologic findings were consistent with the foreign body/giant cell reaction as the pathogenesis of the cyst formation.[17] Such complication has not been reported in the ACLR treatment for patients with a congenital absence of ACL.

Congenital ACL absence is rare, and it can often be associated with other congenital abnormalities. In our case, the patient was found to have a leg length discrepancy with the ipsilateral femur shorter than the left femur. The ACL agenesis was also accompanied by a hypoplastic distal femoral condyle, a dysplastic intercondylar notch, and a dysplastic tibial eminence. The combination of these abnormalities further supported the hypothesis initially proposed by Giorgi that the development of the intercondylar notch and tibial eminence would depend on the fully developed ACL.[1]

In addition, the therapeutic approach in the patients with congenital ACL is not standardized. Some authors recommend conservative treatment as many patients may not have any symptoms, as they may have adapted to these developmental abnormalities.[9,18] Other studies have reported favorable outcomes with ACLR for symptomatic patients. One of the largest series was reported by Sachleben *et al*.[12] from the Boston Children's Hospital, Boston, MA, USA. Of all the 14 knees in 13 patients with congenital absence of the ACL treated with ACL reconstruction, only one patient required revision due to persistent knee instability related to the malformation of other ligaments. All others noted subjective improvement in knee stability. A surgical approach using an autogenous iliotibial band was used in young children with congenital absence of ACL.[19] Overall, the optimal treatment option should be determined on a case-by-case basis.

Conclusions

We reported a case of pretibial pseudotumor formation following ACLR using an absorbable interference screw that was due to a histiocytic and foreign body giant cell reaction to polymeric material in the interference screw. It is notable that this screw, which was meant to be absorbable and osteoconductive, still had remnants present at five years after surgery and was not associated with any obvious osteoconductive activity. Therefore, this report highlights a potential complication with polymeric absorbable materials used in orthopedic surgery.

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Conflict of interest

Dr. Zhang has been an editorial board member of *Journal of Clinical and Translational Pathology* since 2021. The authors declare that they have no other conflict of interest.

Author contributions

Study design and manuscript writing (YZ); Critical revision and technical support (TWB); Performance of the surgery, patient care, critical revision, and material support (SAR). All authors made a significant contribution to this study and approved the final manuscript.

Ethical statement

The patient gave consent for publication of this case report.

References

[1] Giorgi B. Morphologic variations of the intercondylar eminence of the knee. Clin Orthop 1956;8:209–217. PMID:13374913.

[2] Katz MP, Grogono BJ, Soper KC. The etiology and treatment of congenital dislocation of the knee. J Bone Joint Surg Br 1967;49(1):112–120. PMID:6019376.

[3] Manner HM, Radler C, Ganger R, Grill F. Dysplasia of the cruciate ligaments: radiographic assessment and classification. J Bone Joint Surg Am 2006;88(1):130–137. doi:10.2106/JBJS.E.00146, PMID:16391258.

[4] Benassi AB, Guerreiro JPF, de Oliveira Queiroz A, Gasparelli RG, Danieli MV. Anterior and posterior cruciate ligament agenesis. J Surg Case Rep 2018;2018(8):rjy216. doi:10.1093/jscr/rjy216, PMID:30151108.

[5] Kaelin A, Hulin PH, Carlioz H. Congenital aplasia of the cruciate ligaments. A report of six cases. J Bone Joint Surg Br 1986;68(5):827–828. doi:10.1302/0301-620X.68B5.3782255, PMID:3782255.

[6] Héron D, Bonnard C, Moraine C, Toutain A. Agenesis of cruciate ligaments and menisci causing severe knee dysplasia in TAR syndrome. J Med Genet 2001;38(8):E27. doi:10.1136/jmg.38.8.e27, PMID:11483643.

[7] Frikha R, Dahmene J, Ben Hamida R, Chaieb Z, Janhaoui N, Laziz Ben Ayeche M. [Congenital absence of the anterior cruciate ligament: eight cases in the same family]. Rev Chir Orthop Reparatrice Appar Mot 2005;91(7):642–648. doi:10.1016/s0035-1040(05)84468-x, PMID:16327669.

[8] Liu Y, Li Y, March ME, Nguyen K, Xu K, Wang F, *et al*. Copy number variation in CEP57L1 predisposes to congenital absence of bilateral ACL and PCL ligaments. Hum Genomics 2015;9:31. doi:10.1186/s40246-015-0053-z, PMID:26561035.

[9] Lu R, Zhu DP, Chen N, Sun H, Li ZH, Cao XW. How should congenital absence of cruciate ligaments be treated? A case report and literature review. World J Clin Cases 2019;7(19):3082–3089. doi:10.12998/wjcc.v7.i19.3082, PMID:31624758.

[10] Stenqvist C, Sahlström A. Congenital aplasia of cruciate ligaments causing knee osteoarthritis (in Danish). Ugeskr Laeger 2014;176(33):V12130710. PMID:25293412.

[11] Kim KK, Kim TH, Kim DY, Choi JK. Bilateral congenital absence of the anterior cruciate ligament associated with bilateral knee and hip osteoarthritis: Case report. Int J Surg Case Rep 2020;72:313–317. doi:10.1016/j.ijscr.2020.05.099, PMID:32563093.

[12] Sachleben BC, Nasreddine AY, Nepple JJ, Tepolt FA, Kasser JR, Kocher MS. Reconstruction of Symptomatic Congenital Anterior Cruciate Ligament Insufficiency. J Pediatr Orthop 2019;39(2):59–64. doi:10.1097/BPO.0000000000000940, PMID:28178094.

[13] Chahla J, Pascual-Garrido C, Rodeo SA. Ligament Reconstruction in Congenital Absence of the Anterior Cruciate Ligament: A Report of Two Cases. HSS J 2015;11(2):177–181. doi:10.1007/s11420-015-9448-6, PMID:26140039.

[14] Lee JJ, Oh WT, Shin KY, Ko MS, Choi CH. Ligament reconstruction in congenital absence of the anterior cruciate ligament: a case report. Knee Surg Relat Res 2011;23(4):240–243. doi:10.5792/ksrr.2011.23.4.240, PMID:22570842.

[15] Tibor L, Chan PH, Funahashi TT, Wyatt R, Maletis GB, Inacio MC. Surgical Technique Trends in Primary ACL Reconstruction from 2007 to 2014. J Bone Joint Surg Am 2016;98(13):1079–1089. doi:10.2106/JBJS.15.00881, PMID:27385681.

[16] Kramer DE, Kalish LA, Kocher MS, Yen YM, Micheli LJ, Heyworth BE. Complications of Bioabsorbable Tibial Interference Screws After Anterior Cruciate Ligament Reconstruction in Pediatric and Adolescent Athletes. Orthop J Sports Med 2020;8(2):2325967120904010. doi:10.1177/2325967120904010, PMID:32154321.

[17] Gonzalez-Lomas G, Cassilly RT, Remotti F, Levine WN. Is the etiology of pretibial cyst formation after absorbable interference screw use related to a foreign body reaction? Clin Orthop Relat Res 2011;469(4):1082–1088. doi:10.1007/s11999-010-1580-5, PMID:20878286.

[18] Davanzo D, Fornaciari P, Barbier G, Maniglio M, Petek D. Review and Long-Term Outcomes of Cruciate Ligament Reconstruction versus Conservative Treatment in Siblings with Congenital Anterior Cruciate Ligament Aplasia. Case Rep Orthop 2017;2017:1636578. doi:10.1155/2017/1636578, PMID:28589051.

[19] Patwardhan S, Sodhai V, Sancheti P, Shyam A. Extra-articular Tenodesis Using Autogenous Iliotibial Band for Symptomatic Congenital Absence of Anterior Cruciate Ligament: A Report of 2 Cases. JBJS Case Connect 2020;10(2):e0240. doi:10.2106/JBJS.CC.19.00240, PMID:32649100.