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The knee in congenital femoral deficiency and its implication in limb lengthening: a systematic review

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- Congenital femoral deficiency (CFD) is a rare disorder with several limb anomalies including limb shortening and knee cruciate ligament dysplasia.
- Limb lengthening is usually performed to correct lower limb discrepancy. However, complications, such as knee subluxation/dislocation, can occur during this treatment.
- Here, we explore CFD knee abnormalities and knee dislocation during limb elongation, discussing when and whether knee ligament reconstruction prior to the lengthening would be necessary to reduce the risk of knee dislocation.
- There is not enough support in the literature for the routine reconstruction of cruciate ligaments in CFD patients.
- Of note, in cases of severe anteroposterior or posterolateral rotatory instability, cruciate ligament reconstruction might be considered to decrease the risk of knee subluxation/dislocation during the lengthening treatment.

Keywords: articular ligaments abnormalities; bone lengthening; joint instability

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Introduction

Congenital femoral deficiency (CFD) is a rare congenital disorder characterized by failure of normal development of the femur. It is estimated that one in 50,000–200,000 individuals in the population may present this malformation.[1] Clinical findings include shortening of the affected limb and flexion, external rotation and abduction of the hip.[2] Normally, CFD is accompanied by other proximal femur abnormalities, such as coxa vara, pseudoarthrosis and acetabular dysplasia. The disease spectrum varies from mild cases with minimal limb shortening and normal hip development to severe cases with complete absence of the femur and hip joint.[2]

Among the several classification systems created to cover the wide range of CFD presentations, Paley's classification is widely used to guide reconstructive lengthening operations. In this classification system, four different types are described based on the progressive clinical worsening of CFD.[3] Overall, in Type I the femur is intact, with normal knee and hip mobility. Type II describes mobile pseudarthrosis of the hip. Type III characterizes severe diaphyseal femoral deficiency, and Type IV presents deficiency of the distal femur.[3]

Other congenital anomalies are often associated with CFD,[4] such as dysplasia of the cruciate ligaments, leading to an anteroposterior knee instability.[5] Manner et al assessed the ligamentous dysplasia in congenital lower limb deformities, particularly in cases of CFD and fibular hemimelia, showing that the anterior cruciate ligament (ACL) was involved in all cases of ligamentous anomalies.[7]

Patients with CFD and important limb discrepancy may require multiple procedures to correct shortening and other bone deformities.[7] Different devices are used for limb lengthening, including monolateral fixators, circular external fixators or intramedullary nail. Regardless of the method, complications may occur,[7] and one of the major concern relates to knee subluxation or dislocation, which is usually associated with congenital ligament dysplasia.[8,9] Knee subluxation/dislocation may delay or interrupt the lengthening treatment, worsening the prognosis.[7,10]

Considering that most knee surgeons are not accustomed to or comfortable when facing these complex cases, a review is called for. Here, we aim to provide an overview of the congenital knee abnormalities associated with CFD, specifically cruciate ligament anomalies, as well as to describe the complication of knee subluxation/dislocation during limb lengthening. Moreover, we

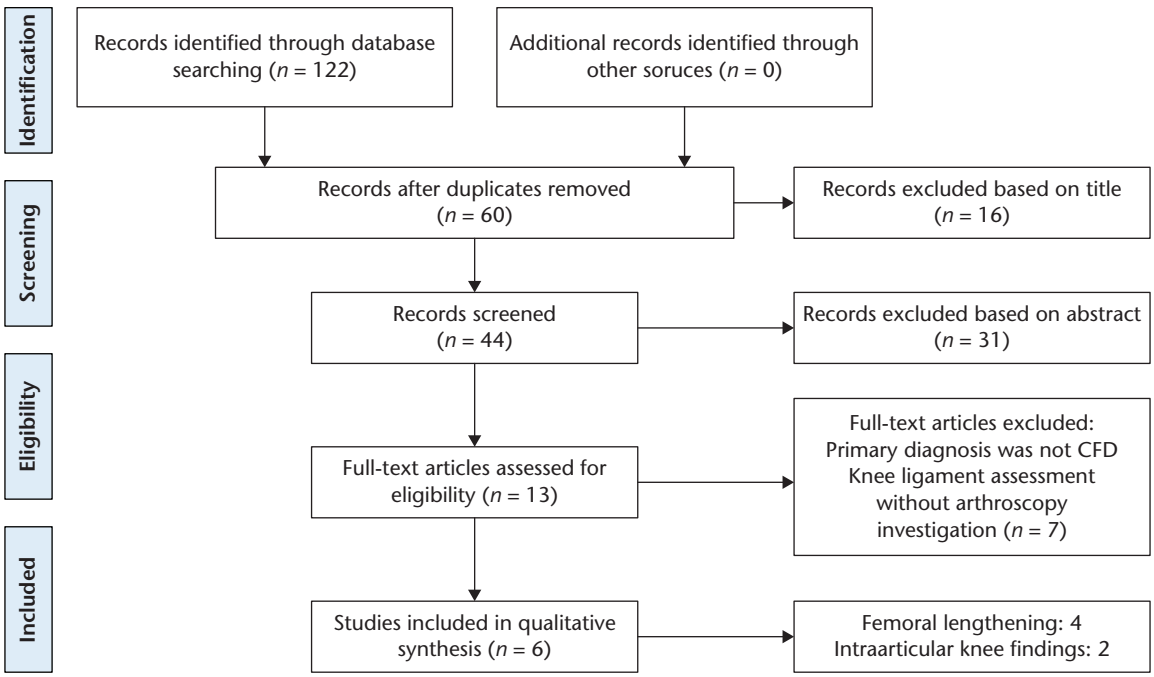

Fig. 1 Flowchart showing the selection criteria used to identify studies with the search strategy.
Note. CFD, congenital femoral deficiency.

discuss whether or when ligament reconstruction prior to the lengthening would be necessary to reduce the risk of subluxation/dislocation of the knee.

Methods

For the purpose of this review, a systematic literature search was performed by consulting the PubMed, Cochrane, and Google Scholar databases. Peer-reviewed and English-language studies related to (1) knee cruciate ligament dysplasia or (2) knee subluxations/dislocations during limb lengthening in patients with congenital femoral deficiency/proximal femoral focal deficiency/congenital short femur, were searched. The findings were summarized to ensure an understanding of the information obtained from the literature. In terms of cruciate ligament dysplasia, only studies that investigated both clinically and arthroscopically the status of the knee cruciate ligaments were included. In regard to knee subluxation/dislocation, studies that described limb lengthening in CFD patients were evaluated as long as they reported episodes of subluxation/dislocation during limb elongation; thus, studies involving limb elongation in CFD patients that did not report this related complication were not included. Because of the lack of literature regarding those topics, non-restriction of level of evidence was applied. Studies in which the primary diagnosis was not CFD, review articles and book chapters were excluded.

Literature search strategy

The following terms were screened: “Congenital Femoral Deficiency” OR “Proximal Focal Femoral Deficiency” OR “Congenital Short Femur” AND “Knee”; “Congenital Femoral Deficiency” OR “Proximal Focal Femoral Deficiency” OR “Congenital Short Femur” AND “Femoral Stretching”; “Congenital Femoral Deficiency” OR “Proximal Focal Femoral Deficiency” OR “Congenital Short Femur” AND “Knee Subluxation”; “Knee” AND “femoral stretching”; “Femoral stretching” AND “knee subluxation”.

After removing duplicates and grouping selected studies, the remaining articles were independently reviewed by two authors to verify their eligibility according to title and abstract. The articles chosen were then completely assessed to confirm the inclusion criteria. In addition, all references of the included studies were revised for posterior addition in case of absence in the initially selected articles. The systematic review was conducted between October and November 2019 and included studies dating from 1983 to 2015 (the latest study found in our search).

Data collection

Relevant data extracted from the articles were recorded in the Microsoft Excel program (version 16.27, Microsoft Corporation, Seattle, Washington, USA). We collected information related to the publication (author, year of publication and original country), sample size, CFD

classification system, associated diagnoses, evaluation methods (clinical, radiographic and surgical), knee findings and occurrence of knee subluxation/dislocation during femoral lengthening.

Results

Figure 1 summarizes the study selection process. The primary search identified 122 studies. After removal of duplicates, 60 studies were screened. Of these, six met our criteria and were included in this review. Among those six included studies, two studies investigated the cruciate ligament abnormalities in CFD patients and four investigated the knee subluxation/dislocation during femoral lengthening. The number of patients listed in the articles was variable, ranging from six to 37.

Knee ligament abnormalities

As mentioned, two studies assessed the cruciate ligament abnormalities in CFD patients.[5,11] When combined, a total of 27 patients were evaluated, all of them presenting anterior knee instability represented by a positive anterior drawer test during clinical examination. In two of those 27 patients the ACL deficiency was not confirmed on arthroscopic examination, and, therefore, 25 patients (92.6%) with ACL abnormalities were reported. Posterior instability, assessed by posterior drawer test, was evident in 12 cases (44.4%). Considering the arthroscopic findings, 16 cases (59.3%) demonstrated posterior cruciate ligament (PCL) deficiency, of whom 12 had a PCL deficiency in association with ACL abnormality; that is, 12 patients (44.4%) had abnormalities in both anterior and posterior cruciate ligaments. One patient (3.7%) had isolated PCL dysplasia and one patient (3.7%) had no ligament anomalies verified in the arthroscopic investigation, despite the positive clinical test (Table 1). Importantly, no association was found between the severity of the ligament deficiency and the amount of femoral shortening. Similarly, there was no relationship between the type of dysplasia and other meniscal anomalies (e.g. discoid meniscus).

Knee subluxation during femoral lengthening

Considering the four studies included in this section,[13–16] 120 patients were assessed, with 54 of them showing clinical knee anteroposterior instability. Arthroscopic evaluation was conducted in only four of those patients. Different techniques were used to perform the limb lengthening, including the Wagner technique (38 cases), Ilizarov technique (38 cases) and monolateral external fixator (44 cases).

Regarding knee subluxation, 31 patients (57.4% of the 54 unstable knee patients or 25.8% of the 120 total patients) presented this complication during femoral lengthening, all of them presenting knee anteroposterior

Table 1. Articles related to clinical and arthroscopic correlation of knee ligament abnormalities in patients with congenital femoral deficiency

Author	Country	Journal	Number of cases *n*	Classification system	Associated diagnostics *n* (%)	Evaluation method	Normal ACL *n* (%)	ACL dysplasia *n* (%)	Normal PCL *n* (%)	PCL dysplasia *n* (%)	Complete ACL + PCL absence *n* (%)	Positive anterior drawer test *n* (%)	Positive posterior drawer test *n* (%)	Other findings
Chomiak et al (2012)[11]	Czech Republic	*J Pediatr Orthop*	21	Pappas	–	Clinical exam Radiographic Arthroscopic	2 (9.5)	19 (90.5) Hypoplasia 3 (14.3) Aplasia 16 (76.2)	8 (38)	13 (61.9) Hypoplasia 3 (14.3) Aplasia 10 (47.6) Obs: Isolated PCL aplasia 1 (4.7)	9 (42.8)	21 (100)	9 (42.8)	Meniscal hypoplasia 3 cases (14.3%) Lateral femoral condyle hypoplasia 21 cases (100%) Chondral lesions 3 cases (14.3%): 2 (9.5%) – medial and lateral compartment 1 (4.8%) – isolated medial compartment Valgus: 21 cases (100%)
Johansson and Aparisi (1983)[5]	Sweden	*J Bone Joint Surg Am*	6	Amstutz	Fibular hemimelia: 5 (83.3) Fifth digital ray agenesis: 1 (16.7)	Clinical exam Radiographic Arthroscopic	–	6 (100)	3 (50)	3 (50)	3 (50)	6 (100)	3 (50)	Hypoplasia of the medial and lateral tibial spines 3 cases (50) Lateral tibial spine hypoplasia 3 cases (50) Chondral lesion 1 case (16.7)

Note. ACL, anterior cruciate ligament; PCL, posterior cruciate ligament.

Table 2. Articles presenting episodes of knee subluxation/dislocation during Congenital femoral deficiency after limb lengthening

Author	Country	Journal	Number of cases *n*	Classification system	Associated diagnostics *n* (%)	Evaluation method	Limb lengthening *n* (%)	Subluxation during lengthening *n* (%)	Treatment for subluxation	Other findings *n* (%)	Comments
Jeong et al (2006)[13]	Turkey	*Clin Orthop Relat Res*	23	Kalamchi	Fibular hemimelia: 11 (47.8)	Radiographic Clinical exam	23 (100)	10 (43.5)	Physiotherapy	Previous instability: 10 (43.5) Hypoplasia of lateral femoral condyle: 8 (34.8) Hypoplasia of the tibial spine: 16 (69.5)	Presence of knee arthrosis Total: 18 cases (78.3%) 9 mild cases (39.2%) 9 Severe cases (39.1%)
Grill and Dungl (1991)[14]	Austria Czech Republic	*J Bone Joint Surg Br*	37	Pappas	–	Radiographic Clinical exam	37 (100)	8 (21.6) of the 14 patients with cruciate ligament dysplasia	Release of soft tissues (iliotibial tract) Knee extension with Ilizarov Simple distraction with external fixator	Previous instability: 14 (37.8)	Post-operative ROM restriction: 34 cases (91.9%) All cases treated after physical therapy
Prince et al (2015)[15]	United States	*Clin Orthop Relat Res*	30	Paley	Fibular Hemimelia: 4 (13.3) Tibial Hemimelia: 2 (6.7) Multiple hereditary exostosis: 1 (3.3) Syndactyly: 1 (3.3)	Radiographic Clinical exam	30 (100)	4 (13.3)	–		Superknee procedure performed in 15 cases (50.0%)
Aston et al (2009)[16]	England	*J Bone Joint Surg Br*	30	Paley Pappas		Radiographic Clinical exam	30 (100)	9 (30.0)	Adjustments to the external fixator Reduction of distraction speed	Previous instability: 30 (100)	Post-operative ROM restriction: 9 cases (30.0%) Treatment: Manipulation under anaesthesia (2 cases) Quadricepsplasty (7 cases)

Note. ROM, range of motion.

instability. The exact direction of the subluxation is not clearly defined in these studies. The Wagner technique and Ilizarov technique were responsible for 13 cases of subluxation each (41.9% for each method), while the monolateral external fixator accounted for five cases of knee subluxation (16.1%). Subluxation episodes were treated by slowing the distraction rate, physiotherapy, surgical release of iliotibial band and/or frame adjustment of the external fixator (Table 2). All but one patient (30 out of 31 – 96.8%) were successfully treated using these measures. This one patient required PCL reconstruction to stabilize the knee.

Discussion

The most relevant findings of this study are that CFD patients usually present cruciate ligament dysplasia, and, in fact, cases with unstable knees have increased risk of subluxation/dislocation during limb lengthening. The following sections will present a literature review of the congenital knee anomalies associated with CFD. In addition, we present a critical view of the relationship among knee instability, knee subluxation/dislocation and femoral lengthening, and discuss the need for ligament reconstruction in this specific group of patients.

Knee in congenital femoral deficiency (CFD)

CFD is frequently associated with multiple knee abnormalities (Supplementary Table S1). Lateral tibial and lateral femoral condyle abnormalities are the most common findings, reported in up to 100% of the patients.[7,11] Besides that, valgus alignment, another extremely common deformity associated with CFD, is observed in 70–100% of cases.[6,11,17] In regard to cruciate ligament dysplasia in CFD patients, although the actual prevalence has not been determined, previous studies have reported rates as high as 90–100% of ACL anomalies and approximately 62% of PCL anomalies.[6,7,11] Yet, it is noteworthy that the amount of instability is widely variable, and neither the severity of limb shortening nor the presence of

valgus deformity have shown correlation with the intensity of AP instability.[17] In addition, regardless of instability, CFD patients usually do not complain of knee pain or giving way.[6,11,17] However, the stability of the knee plays an important role during femoral lengthening due to the potential risk of knee subluxation or dislocation during this procedure;[5,6,11,14] special care must be taken to avoid this complication.[17]

Knee subluxation/dislocation during limb lengthening procedures

Knee-related complications during limb lengthening procedures, including knee subluxation or dislocation, are relatively rare events. Indeed, Jones et al reported only eight cases of knee subluxation in a total of 329 related studies.[12] However, depending on the limb elongation technique this rate may increase, as observed after the monolateral external fixator technique (13%),[15] the transarticular Ilizarov technique (30%),[16] and the Wagner technique (33.0–43.5%).[12,13] Indeed, this current review showed a similar distribution of knee subluxation according to each referred technique. Additionally, knee dysplasia may increase the chance of luxation. Grill and Dungl have reported knee subluxation/dislocation during femoral lengthening in eight of 40 cases (20%); considering only cases with knee dysplasia, the percentage has reached 57% (eight out of 14 cases).[14] Once again, this review shows equivalent results as the rate of subluxation during limb elongation increases from 25.8% to 57.4% when considering only patients with unstable knees.

As referred above, pre-existing joint instability, usually due to congenital cruciate ligament alterations, has been pointed out as the most common predisposing factor for knee subluxation/dislocation during limb lengthening.[9] The force created by the hamstring muscles as a result of the limb elongation can pull the tibia posteriorly, in particular during knee flexion, leading to posterior subluxation/dislocation of the knee when joint instability is present. Unsurprisingly, this complication rarely occurs in full knee extension.[9] Besides ligament anomalies, lateral femoral condyle hypoplasia and lateral tibial plateau flattening also represent relevant risk factors.[12] Dysplasia of the lateral compartment tends to increase the posterolateral rotatory instability, enhancing the chance of knee subluxation during femoral lengthening.[12,15] The use of hinged transarticular external fixator including the tibia seems to be a successful strategy to reduce the risk of knee subluxation in these high-risk patients.[3,8]

In terms of treatment, knee subluxation/dislocation during femoral lengthening can be managed with physical therapy for muscle stretching (mild cases), surgical soft tissue release (particularly the iliotibial tract release),[14] or using the external fixator to reverse the dislocation, through knee extension, distraction and joint reduction (severe cases).[9] Moreover, if the subluxation is confirmed during the procedure, the lengthening must be slowed down or stopped.[15] As indicated before, these measures are highly successful, with an overall cure rate of approximately 96%. Cases in which the subluxation/dislocation is detected only after the external fixator removal might require a more aggressive approach, that includes tendon and capsular releases or external fixator replacement.[9] Therefore, close monitoring should be performed to prevent its misdiagnosis. Table 3 summarizes threatening clinical symptoms and signs for subluxation/dislocation during limb elongation.[12,16]

Table 3. Clinical signs and pre-operative risk factors for knee subluxation during femoral lengthening

Signs of subluxation during femoral lengthening	Risk factors for subluxation before femoral lengthening
Knee pain	Lateral femoral condyle hypoplasia
Hip pain	Lateral tibial plateau flattening
Knee oedema	
Shiny appearance of anterior knee skin	
Sudden discomfort during physiotherapy	
Inability to fully extend the knee	

Ligament reconstruction for cruciate ligament dysplasia

The need for ligament reconstruction in CFD patients with cruciate ligament dysplasia can be outlined in two distinct situations: before the lengthening, as a measure to prevent the potential knee subluxation/dislocation, or after the lengthening, to treat eventual instability that may impair the patient's quality of life.

As mentioned, although some patients with cruciate ligament dysplasia refer to the knee giving way or oedema,[18] joint instability is not a common complaint.[11,17,19,20] Therefore, ligament reconstruction has rarely been indicated for these patients, and the need for specific therapy is still unclear. Cases that require treatment for knee instability after limb lengthening may initially be approached with conservative measures, such as knee immobilizers and physical therapy.[19] Cruciate ligament reconstruction is only indicated for patients who maintain significant symptoms of instability after the limb correction.[8,19,21]

Regarding the ligament reconstruction before the limb elongation, no consensus exists on the necessity for a prior ligament reconstruction as a measure to prevent knee subluxation/dislocation during the lengthening. Indeed, the effectiveness of the treatment approaches in cases of knee subluxation (as shown to be higher than 96%) may discourage any 'prophylactic' procedures. However, selected cases might benefit from a preparatory surgery.[3] In this setting, patients between two and three years old,[3] who present severe knee instability marked by ACL absence along with explosive pivot shift, or an obvious posterolateral rotatory instability, may be ideal

candidates for a prior procedure. Cases that undergo iliotibial band removal due to hip surgery can take advantage of the excised fascia lata as a graft for knee ligament reconstruction.[15] Furthermore, if other knee disorders are noticed (e.g. patellar dislocation), all abnormalities should be addressed in a single surgical time by performing the so-called 'Superknee procedure'. This method is a combination of several procedures, including intra and extraarticular knee ligament reconstructions,[3,22] patellar realignment,[3,23–25] and posterior capsulotomy,[3] and is beyond the scope of this review. Main indications and observations for each step of the Superknee procedure are briefly demonstrated in Supplementary Table S2.

Limitations

The main limitation of this systematic review is the heterogeneity of the studies analysed in terms of sample size and study design. Despite the importance of pioneering in each selected study, the lack of standardization or consensus limits the overall conclusion about the topic, and therefore a meta-analysis was not possible. Even so, our initial aim of presenting CFD-related knee ligament anomalies and their relationship to knee subluxation/dislocation during limb lengthening was achieved. Moreover, as far as we know, this review is one of the only studies that provides a complete update on the topic, discussing different concepts in order to standardize and facilitate the clinical management of this specific group of patients.

Conclusion

Ligamentous dysplasia is commonly found in CFD patients, with more than 90% of ACL anomalies and approximately 60% of PCL anomalies observed in this current study. In addition, knee subluxation during limb lengthening is a frequent complication associated with these patients, as observed in 57.4% of those cases in this review. However, even considering these high frequencies, there is not enough evidence to support the routine indication of cruciate ligament reconstruction to prevent knee subluxation/dislocation during limb lengthening. Fortunately, cruciate ligament dysplasia is rarely associated with clinical symptoms, even after limb lengthening and deformity corrections, and the treatment of knee subluxation (when it occurs during the limb elongation) achieves satisfactory results. According to this review, patients should receive an intense and continuous monitoring during bone elongation to avoid misdiagnosis. Indications for prior ligament reconstruction (i.e. preparatory surgery) to decrease the risk of joint dislocation is reserved for cases with severe anteroposterior instability and vigorous pivot shift, or when severe posterolateral rotatory instability is present. In addition, in cases that undergo iliotibial tract removal by hip interventions the fascia lata can be used to reconstruct the ACL and PCL, serving as an additional mechanism to decrease the risk of knee subluxation/dislocation during limb lengthening.

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SUPPLEMENTAL MATERIAL

Supplemental material is available for this paper at https://online.boneandjoint.org.uk/doi/suppl/10.1302/2058-5241.6.200075

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