Recurrence of axial malalignment after surgical correction in congenital femoral deficiency and fibular hemimelia

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Received: 28 March 2011 / Revised: 4 April 2011 / Accepted: 4 April 2011 / Published online: 3 May 2011
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Abstract

Purpose Recurrent genu valgum deformity complicates treatment of congenital femoral deficiencies (CFD) and fibular hemimelia (FH). We analysed factors influencing recurrence.

Methods Patients who underwent limb lengthening or deformity correction for CFD and/or FH were reviewed. Radiographs after surgery and after a minimum of a further six months were analysed. Change in parameters of mechanical axis deviation per month (Δ MAD/month) and of angle per month were calculated. These parameters were tested against cofactors patient age, baseline MAD, type of CFD and FH, severity of ball-and-socket joints, ankle-joint stiffness, absence of cruciate ligaments and resection of the fibular anlage.

Results Recurrent valgus deformity was found in 23 of the 42 limbs included with a mean change of MAD of 23.4 mm (5–60 mm). There was no significant difference between patients with Δ MAD/month <0.5 mm versus >1 mm regarding MAD in the first radiograph and patient age. CFD cases Pappas types VII and VIII showed a Δ MAD/month of 1.6 mm, whereas milder cases of Pappas IX showed a Δ MAD/month of 0.8. Mild FH (type Ia) showed a mean Δ MAD/month of 0.39 mm, whereas mean Δ MAD/month for FH type Ib/II was 0.72 mm. In FH type II cases, mean Δ MAD/month was 0.79 mm after resection of the fibular anlage compared with 1.98 mm in those without resection.

Conclusions Recurrence in FH and CFD was not dependent on patient age but partly on FH and CFD type. Limbs with more severe ball-and-socket knee joints showed more recurrence. Overcorrection depending deformity type should be performed.

Introduction

Congenital femoral deficiency (CFD) and fibular hemimelia (FH) are rare longitudinal defects of the lower limb with an incidence of approximately 1:40,000–50,000 live births [1]. CFD shows variable involvement of the femur from mild shortening to severe dysplasia [2, 3]. FH presents with hypoplasia or aplasia of the fibula, with shortening and malalignment of the tibia, deformities at the knee and ankle joint and ray deficiencies [4–7]. CFD and FH are found combined in the same leg in about 68% of cases [4]. Although amputation and prosthetic fitting has traditionally been recommended for most cases [8–10], reconstructive treatment with good functional results can be performed using external fixation [11–15]. Recurrent and progressive genu valgum deformity has been reported by different authors in reconstructive as well as in ablative treatment approaches [4, 5, 10, 16–19]. Initial observations suggest that genu valgum deformity was the result of tethering of the posterolateral structures, including the fibular anlage [19], or resulted from growth suppression due to abnormal loading of the knee joint in the presence of anteromedial bowing [17]. However, other studies have questioned those factors and suggested insult to the limb bud to be the primary cause of hypoplasia of the lateral femoral condyle and subsequent valgus deformity [10]. To account for the tendency to recurrence, overcorrection of valgus deformity by distraction towards

Investigation performed at the Orthopaedic Hospital Speising, Vienna, Austria

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varus alignment has been advocated [11]. However, no studies are available on factors that might influence recurrence, such as patient age or type of deformity. We postulated that recurrence is most evident in FH and is dependent on patient age and type of hemimelia and fibular anlage status. Furthermore, we hypothesised that recurrence is less evident in CFD, but still related to type of CFD and age.

Methods

We retrospectively reviewed our prospective database of patients undergoing limb lengthening or deformity correction from August 1999 to December 2009. Inclusion criteria were: (a) CFD and/or FH, (b) at least one operation with an external fixator aimed at correcting axial malalignment and/or lengthening and (c) availability of a long-standing radiograph after surgical intervention and a second long-standing radiograph after a minimum of a further six months. Patients older than 16 years, patients with bilateral involvement and patients in whom surgery was not performed with the aim of reconstruction and full leg-length equalisation were excluded. Demographic data and factors that might influence recurrence were analysed. All CFD cases were classified according to Pappas et al. [2], and Achterman and Kalamchi classification was used for FH cases [4]. We further classified the knee and ankle joints with respect to presence and severity (mild, moderate, severe) of a so-called ball-and-socket joint. The term ball-and-socket joint describes the distal articular surface, which is hemispherical and has a proximal congruent, concave, articular surface with which to articulate. At the ankle, the ball-and-socket joint configuration is well documented [4]. At the knee, a ball-and-socket joint configuration has been found mostly in combination with congenital absence of cruciate ligaments [20].

Ankle-joint stiffness in FH was recorded, and the absence of the anterior and/or posterior cruciate ligament was estimated using the radiographic criteria recommended by Manner et al. [20]. Fibular anlage resection was another factor evaluated. All radiographs were analysed using the malalignment test proposed by Paley and Tetsworth [21, 22] and mechanical axis deviation (MAD), anatomical (aLDFA) and mechanical (mLDFA) lateral distal femoral angle and medial proximal tibial angle (MPTA) were measured. Additionally, the anatomical lateral distal femoral metaphyseal angle (aLDFMA) and the medial proximal tibial metaphyseal angle (MPTMA) were measured [6]. The change of the axial alignment was categorised in each patient who presented an MAD change of ≥ 5 . Cases with MAD change < 5 mm were not included in order to minimise bias of minor positioning errors, especially with

respect to the rotational position of the limb during the radiograph [23] and the intraobserver variability [24].

For the same subgroup, the amount of mechanical axis and measured angle change was related to factor time and parameter change in MAD in millimetres per month (Δ MAD/month), and angle degree change per month (Δ aLDFA/month, Δ mLDFA/month, Δ aLDFMA/month, Δ MPTA/month, Δ MPTMA/month) were calculated. These parameters were tested against cofactors patient age, deformity type, ball-and-socket joint presence, fibular anlage resection and cruciate ligament status. Subgroups according CFD and FH type were statistically compared regarding tendency of recurrence. Statistical analysis was conducted with SPSS 11.0 for Windows (SPSS Inc., Chicago, IL, USA).

Results

Eighty-six patients with CFD and/or FH underwent limb lengthening and lower-limb malalignment correction from August 1999 to December 2009. A postoperative long-standing radiograph and a second long-standing radiograph after a minimum of a further six months were available for review in 35 patients. In seven patients, alignment was followed up after two consecutive operations, resulting in 42 lower limbs meeting the inclusion criteria. Fourteen female and 21 male patients were evaluated. Mean patient age at the time of frame removal was 9.1 ($\pm$ 3.8; range 3–16) years. The right limb was affected in 22 patients and the left in 20. CFD was combined with FH in 36 patients, whereas three patients presented CFD without any signs of FH and three presented isolated FH. Characteristics of the included limbs are summarised in Table 1.

A change in MAD < 5 mm was seen 13 limbs, whereas 29 presented a change ≥ 5 mm. In one patient, there was no MAD change after 54 months. In this patient with a mild CFD (Pappas 9) combined with FH (type 1a), the frame was removed at an age of 3.7 years. In one patient, there was an MAD change from valgus to varus. This patient had the frame removed at 16 years of age and presented CFD and FH after fibular anlage resection and with a severe ball-and-socket knee joint. Seven patients showed varus MAD initially, with four patients presenting a mechanical axis change ≥ 5 mm. One of those four patients presented varus improvement towards a more normal alignment, with a MAD change of 6 mm. Three patients progressed from varus to valgus, with a mean MAD change of 33 mm (range 20–40 mm), resulting in mild valgus in two cases and severe recurrence with a MAD of 34 mm lateral in one case. As these cases represented recurrence of valgus deformity, they were included in the subgroup of valgus recurrence in further analyses. In four patients, valgus

Table 1 Characteristics of all included limbs

	No.	CFD type	FH type	BS-KJ	BS-AJ	Stiff ankle	ACL-A	PCL-A
CFD + FH	36	VII 3 VIII 14 IX 9	Ia 19 Ib 5 II 12	Mild 14 Moderate 13 Severe 5	Mild 13 Moderate 3 Severe 7	10	27	7
CFD	3	VII 0 VIII 1 IX 2		Mild 2 Moderate 0 Severe 0			2	0
FH	3		Ia 2 Ib 0 II 1	Mild 3 Moderate 0 Severe 0	Mild 0 Moderate 2 Severe 0	1	3	0
Total	42	VII 3 VIII 15 IX 21	Ia 21 Ib 5 II 13	Mild 19 Moderate 13 Severe 5	Mild 13 Moderate 5 Severe 7	11	32	7

BS-KJ ball-and-socket formation knee joint, *BS-AJ* ball-and-socket formation ankle joint, *ACL-A* presumed absence of anterior cruciate ligament, *PCL-A* presumed absence of posterior cruciate ligament

malalignment improved MAD >5 mm, mean change of 12.5 mm (range 6–16 mm) was observed. Two of those had an isolated FH, one an isolated mild CFD (type IX) and the fourth a very mild FH type Ia (Table 2).

Twenty limbs showed valgus deformity deterioration. These patients were analysed together with the three patients that showed valgus recurrence from a varus alignment. Mean MAD change in these 23 limbs was 23.4 mm (range 5–60 mm), with a mean MAD in the first postoperative long-standing radiograph of 12.9 mm lateral [range 32 mm medial (varus) to 46 mm lateral (valgus)] and a mean MAD at the last long-standing radiograph of 36.3 mm lateral (range 2–74 mm lateral). These 23 cases

were analysed according to MAD change in millimetres per month (Δ MAD/month), and patients with a Δ MAD/month <0.5 mm ($n=12$) were compared with patients with a Δ MAD/month of >1 mm ($n=12$). Statistical analysis using *t* test after testing for normal distribution showed that there was no significant difference regarding baseline MAD in the first radiograph after frame removal. The group with less recurrence showed a postremoval MAD with more valgus compared with the second group (mean MAD 16.3 mm vs. 11 mm MAD; $p=0.56$). Age at the time of frame removal was identical in both groups (mean 9.1 years vs. 9.1 years; $p=0.957$). Regarding the parameters Δ aLDFA/month, Δ mLDFA/month, Δ aLDFMA/month,

Table 2 Characteristics of mechanical axis change (Δ MAD) between first radiograph after frame removal and follow-up radiograph (n number of cases, a patient age at frame removal)

Change	No. cases	Classification
Δ MAD $\geq$ 5 mm		
valgus to varus	1	CFD VIII, FH II fibular anlage resected; stiff ankle; severe ball-and-socket knee joint; 16a
varus to less varus	1	CFD IX, FH Ia mild ball-and-socket knee joint; 14a
varus to valgus recurrence	3	case 1: CFD VII, FH Ib; 12.4a case 2: CFD VII, FH II; 4.6a case 3: FH Ia; 9.4 a Ø age 8.8 a
valgus to less valgus	4	case 1: CFD IX; 12.5a case 2: CFD IX, FH Ia; 13,1a case 3: FH Ia; 6.5a case 4: FH II; 13,3 a Ø age 11.4 a
valgus to more valgus recurrence	20	Ø age 7.9 a (range 3–13 a)

MAD mechanical action deviation, *CFD* congenital femoral deficiencies, *FH* fibular hemimelia, *PCL-A* presumed absence of posterior cruciate ligament

Δ MPTA/month and Δ MPTMA/month, the only nonsignificant parameter between the two groups was the Δ aLDFMA/month ($p=0.6037$). This indicates that recurrence in the femur was mostly located in the epiphysis (Fig. 1a–d).

Other parameters, such as ankle-joint stiffness or FH or CFD type were not significantly different between the two groups. However, the severity of the ball-and-socket joint formation (graded from 1 to 3) at the knee joint (mean 1.3 vs. mean 1.7; $p=0.1753$) and the ankle (mean 1.3 vs. mean 1.6; $p=0.54$) was higher—but not significantly—in the group presenting more recurrence. CFD cases Pappas types VII ($n=2$) and VIII ($n=8$) showed a mean Δ MAD/month of 1.6 mm, whereas milder cases Pappas IX ($n=12$) showed a Δ MAD/month of 0.8. The Δ aLDFMA/month was 0.34 in Pappas types VII and VIII versus 0.11 in Pappas IX. The Δ aLDFMA/month was very similar and very low, with values of 0.07 (Pappas VII, VIII) and 0.06 (Pappas IX) in both groups. This indicates that there was more recurrence in higher CFD grades, coming nearly exclusively from the lateral epiphysis. In limbs with mild FH (type Ia, $n=24$,

mean follow-up 31.5 months), we found a mean Δ MAD/month of 0.39 mm, whereas the mean Δ MAD/month for FH type Ib or II ($n=18$, mean follow-up 24.9 months) was nearly double (0.72 mm). In patients without any sign of FH presenting isolated CFD ($n=3$, mean follow-up 18.9 months), the mean Δ MAD/month was lowest (0.11 mm). In limbs with FH type Ib or II ($n=18$), mean LDFA change per month was 0.17° compared with 0.03° for MPTA mean change per month, indicating that recurrence still primarily occurred in the femur. Mean aLDFMA change per month was only 0.02° , which would indicate that recurrence occurred mostly at the level of the epiphysis. The MPTMA changed 0.026° per month, indicating that all recurrence originated distal to the physis. Four of the eight patients with an FH type II had fibular anlage resection. In cases in which the anlage was resected, mean Δ MAD/month was 0.79 mm compared with 1.98 mm in patients with unresected anlage. Mean Δ aLDFMA/month was quite similar, with 0.36° in the group without resection versus 0.22° in the group after resection. The Δ MPTA/month was 0.47° in the group without

Fig. 1 Radiograph 6 weeks after frame removal showed normal alignment, with the mechanical axis running through the middle of the knee joint (a). The anatomical lateral distal femoral metaphyseal angle (aLDFMA) after frame removal was 87° (b). After only 10 months the mechanical axis deviation (MAD) changed, resulting in valgus deformity recurrence. Medial proximal tibial angle (MPTA) changed 1° , whereas lateral distal femoral angle (LDFA) decreased 4° (c). The aLDFMA changed only by 1° , indicating that the recurrence originated in the hypoplastic lateral epiphysis of the femur (d)

resection versus 0.12° in the group after resection. This would indicate that fibular anlage resection mostly influences valgus recurrence in the tibia. However, using a Wilcoxon signed ranks test, none of these differences was significant.

Discussion

We found that recurrence was evident in both FH and CFD and that recurrence was not dependent on patient age but partly dependent on FH type. Whereas a stiff ankle joint showed no influence, fibular anlage resection clearly decreased recurrence. In CFD cases, recurrence was again not dependent on age but on type of CFD, with more severe cases showing twice as much recurrence as mild cases (Pappas IX). MAD at the time of frame removal seemed not to influence recurrence. However, limbs with more severe ball-and-socket knee-joint formation were more prone to recurrence. Another striking observation was that recurrence was found nearly exclusively in the femur and within the femur at the level of the epiphysis. In the tibia, recurrence originated primarily below the proximal epiphysis. Progressive valgus deformity has not only been indentified as a problem in reconstructive surgery [16] but also in ablative treatment of CFD and FH [10]. Fulp et al. analysed results of early amputation and prosthetic fitting in 25 children with 31 affected extremities [10]. They analysed the condylar height ratio of the lateral femoral condyle as described by Boakes et al. [17]. This ratio did not improve in patients who had fibular anlage resection, and the authors concluded that the tethering effect of the posterolateral structures was not the reason for valgus recurrence. In our cases of FH type II, fibular anlage resection reduced Δ MAD/months by half, with recurrence being found distal to the physis of the tibia. This does indicate that the fibular anlage has indeed a tethering effect. Nevertheless, there was hardly any influence on the LDFA, so we agree that resection does not improve the condylar height ratio of the lateral femoral condyle but seemed to decrease tibial bowing.

The fact that recurrence was mostly found in the distal femoral epiphysis accords with the findings of Fulp et al. [10] and Manner et al. [6]. They showed that the radiographic pathology of CFD and FH is mostly found in the distal lateral epiphysis in terms of epiphyseal height and width [6, 10], whereas in the tibia only, the epiphyseal width was affected [6]. Although the recurrence was mostly seen in the lateral epiphysis of the femur, cases of isolated CFD showed the lowest Δ MAD/month. We speculate that there are mild cases of CFD, comparable with mild terminal FH [7], with predominant proximal involvement of the femur where the lateral hypoplasia is less evident.

It seems that ball-and-socket joint configuration, especially in the knee joint, is a factor in recurrence not identified previously. The influence on recurrence might be due to a frontal-plane instability resulting from the ball-and-socket joint configuration and the absence of cruciate ligaments. We even found one case in which this instability and presumably the dynamic varus thrust resulted in an MAD change from mild valgus to varus (Fig 2a, b).

In the few cases showing valgus improvement, we found no similarities or patterns that could explain axis improvement. Our study shows multiple biases. To account for errors in position during radiography and angle measurement, we chose an MAD change of a minimum of 5 mm for defining recurrence [23, 24]. The number of cases included combined with multiple variables influencing recurrence

Fig. 2 Radiograph 3 months after frame removal showed residual genu valgum deformity, with the mechanical axis lateralised 39 mm (a). Within 21 months, mechanical axis deviation (MAD) increased to 62 mm lateral. Lateral distal femoral angle (LDFA) decreased only 1° and, medial proximal tibial angle (MPTA) improved by 2° . However, due to the severe ball-and-socket joint formation, an intra-articular deformity developed, increasing overall valgus alignment (b)

resulted in a low statistical significance. However, given the low incidence of CFD and FH, we were able to analyse a considerably high number of cases. A further bias in calculating the Δ MAD/month is the assumption that MAD rate of change is linear. A prospective study design with predefined radiographic follow-up intervals would be necessary to eliminate this bias. The presence or absence of cruciate ligaments was evaluated from the knee joint configuration in tunnel-view radiographs, as recommended [20], which were available in 36 limbs. However, in six cases only anteroposterior view radiographs were available which might have biased our classification. We did not assess LDTA or MPFA [21], as they hardly influence overall MAD. Additionally, the LDTA is hardly measurable in cases of severe ball-and-socket ankle joint. Despite these biases, we still think we sufficiently answered our hypotheses and defined clinical implications. Recurrence of valgus deformity was found in FH as well as in CFD, with more severe cases presenting more recurrence. Fibula anlage resection should be performed in all cases of FH type II, as this decreases recurrence in the tibia but without influencing recurrence in the distal femur. As a result of our findings, we now add temporary hemiepiphysiodesis at the medial distal femur in many cases to prevent recurrence or progressive valgus. Overcorrection should be performed depending on deformity type in corrections performed prior to skeletal maturity. At the end of growth or in cases with severe ball-and-socket joint formation, overcorrection should be avoided.

Conflict of Interest The authors declare that they have no conflict of interest.

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