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Management of children with congenital hypoplasia - defficient femur

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ABSTRACT

Congenital femoral hypoplasia is an extremely rare and severe dysplasia. The main characteristic is the severe leg length discrepancy. It covers a wide spectrum of femoral deformities ranging from hypoplasia to severe proximal deficiency. The aim of our study is to describe the management of the femoral deficiency. We used distraction histogenesis, with a circular Ilizarov frame, for the children that have prospective to equalize the discrepancy. A series of 6 children (5 girls and 1 boy) were treated over a period of 10 years. Patients were classified with the Pappas method. During the first 5-6 years of age, using appropriate orthotic devices, all of them had achieved a reasonable independence for their daily and school activities. In 3 of our patients treatment provided was distraction histogenesis using Ilizarov frame. For 1 patient, with untreatable discrepancy, we continue with special orthotic device. The increase in length was 5 cm to 6 cm, with an increase of 32% of the initial length (ranging from 33% up to 42%). The mean healing index was 17 days/cm. Severe complications presented during the procedures. Twice a child had fractures of the regenerated bone, soon after the removal of the device. They were treated with reapplication of the frame until union of the fractured bone. One child had knee subluxation that is partly corrected with realignment of the device at the knee joint. Treating children with deficient femur is a great challenge for the surgeon. It is almost impossible to correct in one procedure the whole discrepancy. The active life of the surgeon is not sufficient to correct all the deformities of the growing child.

KEY WORDS: congenital short femur; femoral deficiency in children; leg lengthening with Ilizarov

1. Introduction

Congenital hypoplasia of the femur is an extremely rare congenital disorder, with a reported incidence 1 per 100,000 newborns. The term femoral deficient child, similar with congenital hypoplasia, covers the spectrum of dysplastic femur, from simple hypoplasia to proximal femoral focal deficiency. In femoral hypoplasia, we refer in severe shortening of the femur up to

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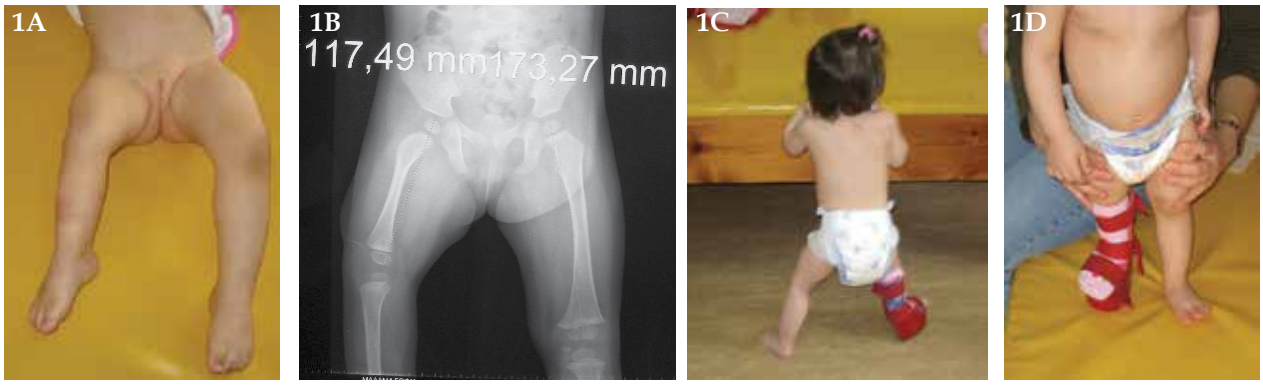

Fig. 1 a, b, c, d. *This girl, using appropriate orthotics and physiotherapy, achieved standing and walking without delay in her motor development*

10-40% of the length of the normal femur. Initial LLD is greater than 4 cm. All the femoral elements are present, but the femur is shorter and thinner than the normal side. The radiological examination reveals the hypoplastic femur, with the presence of the proximal and distal part of the femur [1, 2, 3].

In proximal femoral focal deficiency (PFFD), there is a spectrum of pathology of the proximal part of the femur, from partial shortening of the subtrochanteric area, to complete absence of the upper part, with severe shortening of the femur.

The lesion can be unilateral or bilateral [4]. There is a characteristic clinical picture with the affected leg in flexion, abduction, and external rotation. There is a variety of other lesions of the affected leg, as valgus knee, knee instability because of the absence of cruciate ligaments, fibular hemimelia, foot dysplasia like club foot or ball joint of the ankle.

The main feature of these children is the severe leg length discrepancy (LLD).

The use of distraction osteogenesis (Ilizarov) has completely changed the natural history of children with congenital femoral hypoplasia. It requires series of lengthening procedures, as it is difficult to equalize severe LLD in one procedure. Lengthening for more than 20% of the initial length has a lot of severe complications, but the final result is very encouraging [5, 6]. When the initial LLD is over the 50% of the length of the non affected femur, lengthening is difficult to achieve the purpose of equalization. The Van Nes procedure, using the ankle joint as a knee joint and fitting an appropriate orthotic, is the most suitable solution.

The purpose of our paper is firstly to present a series of 6 children with congenital femoral hypoplasia and secondly describe their management. The use of distraction osteogenesis is a challenge for the pediatric orthopaedic surgeon.

2. Patients and method

This is a series of children with femoral deficiency that are under our supervision since they are born. They are classified according to Pappas criteria in type 9 two girls, type 7 two girls, type 3 one boy and one girl with bilateral lesion.

Early in life, using appropriate orthotics and physiotherapy, all our children achieved standing and walking, without delay in the motor development. (**Fig. 1a, b, c, and d)**

From the age of 5 yrs, we started treatment of severe shortening of the leg. In order to start the plan of lengthening, there must be the option to equalize the leg. Using the multiplier charts of Paley [7], we estimated the final LLD at 12, 15, 16 , 22 cm and at the child with Pappas 3 more than 30 cm.

We could not estimate the final difference in the child with the bilateral affected femurs.

In 3 girls (type 9, and 2 of those with 7 Pappas classification) we used the leg lengthening procedure with the Ilizarov method, while for the child with the type 3 Pappas, we decided to proceed with orthotics, since equalization was not possible. In that child the initial position of the foot was at the level of the knee of the non affected limb.

One child is scheduled for lengthening during the

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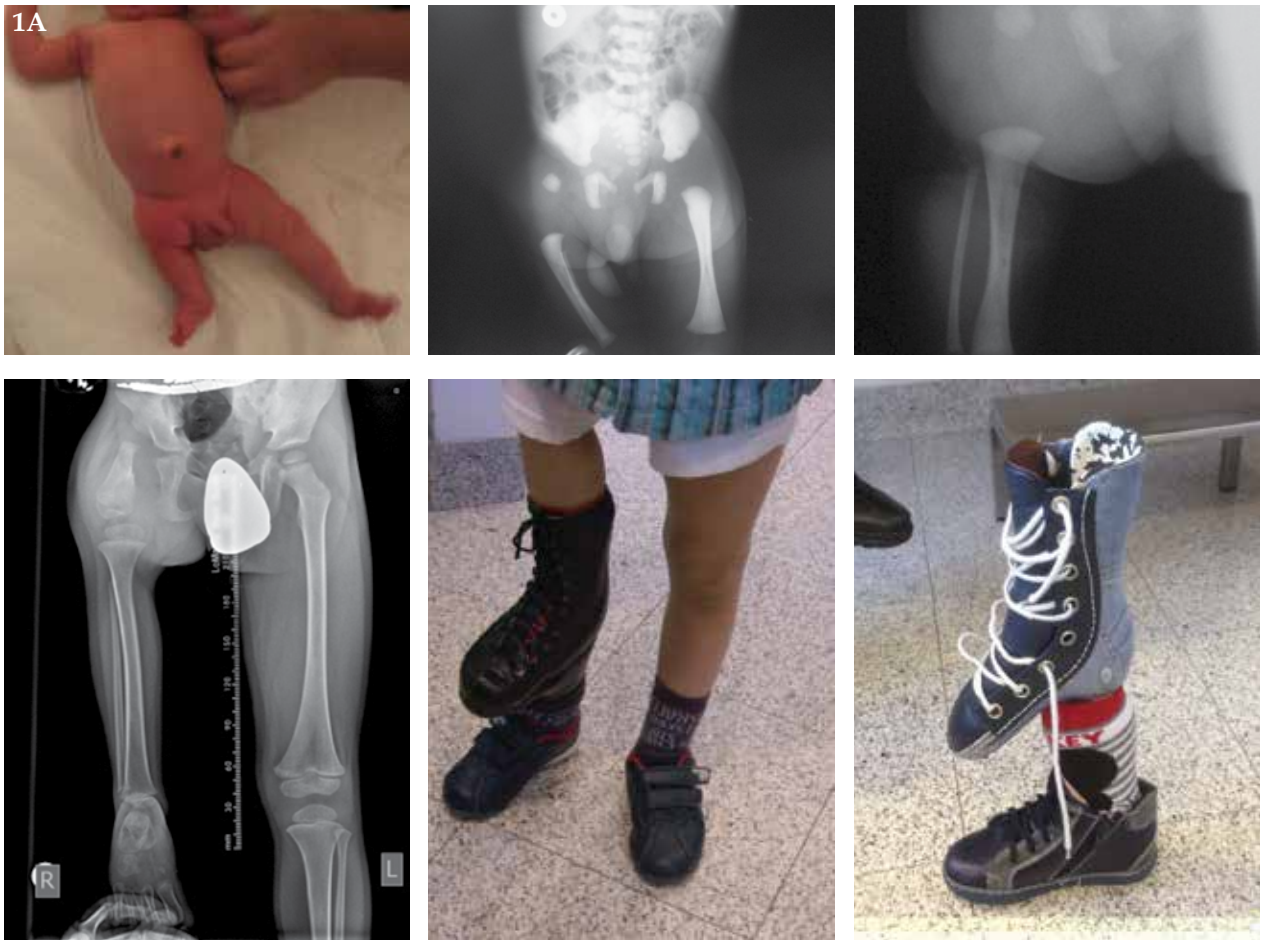

Fig. 2 a, b, c, d, e, f. *Clinical and radiographic features of a boy with untreatable discrepancy. He is planned to accept a Van Nes procedure in the future*

next year. The girl with bilateral affected femur, after the initial treatment and mobilization, had chosen to receive medical management elsewhere.

The boy with untreatable discrepancy had a special orthotic shoe, with double base, so he can walk independently. Further plan is to convert the ankle joint in knee joint (Van Nes procedure), once it will be required. (**Fig. 2a, b, c, Fig. 2d, e, and f)**

Three patients were treated with distraction histogenesis for the management of leg length discrepancy. One had initially treatment of the dislocated hip with closed management. We used a preassembly device, with circular frames that was constructed the previous days of the surgery. The system consisted from 3 rings. Stabilization of the knee was done during the progress of the lengthening. We did not use rings for the pelvis.

3. Results

One girl had a complete equalization of the limbs with an increase of the length of the femur for 5 cm, a 33% of the initial length of the affected femur, with a healing index of 17 days/cm. During the first days after the removal of the device, we noticed a small plastic deformation of the regenerate that was treated conservatively with non weight bearing and a functional brace of the femur. With 2 years follow up, the LLD is today 2cm, with a valgus knee (it was part of the initial dysplasia). Our plan is to perform a new procedure for leg lengthening at the age of 12, with epiphysiodesis with 8 plates for the valgus knee.

The 2 girls with initial LLD of 8 cm of the femur and 2 and 3 cm consecutively of the tibia, were treated with distraction histogenesis of the femur, gaining a lengthening of 35% and 42% of the length of the af-

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fected femur. They had a 6 cm increase of the femur.

All 3 girls had a nice consolidation of the regenerated femur. Our rate was increase of 0,1 cm per day in 4 daily intervals.

When we removed the device in the second girl, in 3 days time she had fracture of the regenerate. We reapplied the Ilizarov device to stabilize the fracture. The fracture healed but that was hard for the emotions of the girl. We achieved to reduce the LLD at 2 cm (there was also another 2 cm difference in the tibia). The same girl had a new lengthening procedure 2 years later, with an increase of 4 cm, but again at the removal of the device she had a fracture of the regenerate. We reapplied the Ilizarov device and achieved the healing of the fracture. Today, 1 year after her 2nd elongation, she is scheduled for a new procedure, since there is a LLD of 5 cm in the femur (there is a continuous increase in the LLD).

The last girl with an enormous increase of the length of the affected femur (43% of the initial length), was complicated from knee subluxation, that was partially treated with changes in the Ilizarov device, with hinges acting in the knee joint. Our patient, 2 years after the initial procedure of the lengthening of the femur and lengthening of the tibia, had a LLD of 5 cm, with varus deformation of the hip joint. Our plan is to correct the hip, perform a new lengthening of the femur and treat her equinus of the ankle that partially covers today the severe LLD.

4. Discussion

There are few series in the literature for the management of an extremely rare condition, like the femoral deficient child. Various types of the hypoplasia have been described.

Pappas classification has 9 types, with type 1 the most severe with complete absence of the proximal part and type 9 the less severe, where there is LLD and mild deformity in the subtrochanteric area [8].

Classification of Lloyd Roberts and Fixsen in 3 types was based on the hip stability, as the result of the absent part of the proximal femur. Aitken classification has 4 types, depending on the presence of the anlage of the femoral head. It is a guide for the type of surgical management of the problem. Paley has proposed a classification based on the stability of the hip and knee joint, focusing to elements that influence the results of leg lengthening procedures [9, 10, 11, 12].

We have chosen the Pappas classification as it has a more detailed description of the elements of the hypoplasia, both for the upper part as well for the whole femur.

Management of these children starts from the period they are babies, for the proper motor development. We must offer early physio assistance and appropriate orthotics for the standing and walking progress of the babies.

Appropriate orthotics that equalize the length discrepancy, permit children to function well for their motor development.

It is not possible to achieve equalization in all cases of femoral deficiency. If the initial discrepancy is more than 50% of the length of the normal leg or if the foot of the affected limb is at the level of the knee joint of the other side, then the continuous developing discrepancy cannot be covered with continuous limb lengthening procedures. The need for continuous severe procedures until the adult life, makes the choice of using the ankle joint as a knee joint (Van Nes procedure) the most appropriate one [13,14].

Lengthening a hypoplastic femur is an extremely difficult procedure, since we treat both the hypoplastic bone and the stiff shortened muscles. It requires continuous monitoring during the lengthening [5, 6, 15].

In our series the regenerate had a good radiological consolidation and we had not cases to change the rate of lengthening. It is possible, when we lengthen already lengthened femurs, to have weak regenerate. Then we may delay the rate of lengthening.

The rate of complications when lengthening hypoplastic femur is extremely high. In the literature it is been referred that there is almost 100% fracture rate of the newly formed bone, after removal of the external device. Using an intramedullary pin at the beginning of the procedure is essential for the protection of the new bone. Using today intramedullary lengthening devices or submuscular plates, the incidence of fractures has been severely reduced. For our cases, with fractures of the new bone, we reapplied the Ilizarov device. In our next

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case, we plan to use, from the beginning, an intramedullary pin [16 -24].

A common complication when we lengthen the femur is the dislocation or subluxation of the knee joint. It is the result of the shortened posterior thigh muscles in combination with the absence of cruciate ligaments and hypoplasia of the femoral condyles. This complication is common when the lengthening is more than 20-25% of the initial length. In our case, this happened because we overestimated our abilities, performing lengthening of 42% of the initial length. Using a ring to stabilize the knee in extension or special joints for controlled knee motion, can protect the knee from subluxation. The same complication exists for the hip joint, a common complication also. It requires regular radiological evaluation, mainly when the lengthening is exceeding 5-6 cm [5, 6, 25, 26].

In conclusion, management of children with deficient femur is a great challenge for the orthopaedic surgeon. Trying to restore as much as possible, severe leg length discrepancies, we overestimate our abilities for lengthening of a small hypoplastic limb. These procedures start from a baby and end up for the patient as an adult. There is a very strong emotional relation between the surgeon and the child and his family. We must plan with an accurate time table the whole management, including our procedures in the school activities of these children.

The active life of an orthopaedic surgeon is not sufficient enough to deal all these problems of the growing child, as underlined always G Pistevos, the pioneer of lengthening procedures in children in Greece.

Conflict of interest:

The authors declared no conflicts of interest.

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ΠΕΡΙΛΗΨΗ

Η συγγενής υποπλασία του μηριαίου αποτελεί μια βαρύτατη και ιδιαίτερα σπάνια διαμαρτία, με κύριο χαρακτηριστικό την μεγάλη ανισοσκελία. Καλύπτει μεγάλο εύρος διαμαρτιών του μηριαίου, από υποπλασία έως απουσία του εγγύς τμήματος. Σκοπός της εργασίας μας είναι να παρουσιάσουμε την αντιμετώπιση παιδιών με υπολειμματικό μηριαίο. Χρησιμοποιήσαμε την διατατική ιστογένεση, με την συσκευή Ilizarov, στα παιδιά που είχαν προοπτική να ισοσκελισθούν.

Παρουσιάζουμε μια σειρά από έξι 6 παιδιά που αντιμετωπίζουμε από την γέννησή τους. Πρόκειται για 5 κορίτσια και 1 αγόρι. Η κατάταξή τους έγινε με τα κριτήρια Pappas.

Με την χρήση ειδικής κατασκευής ορθωτικών για την κάλυψη της ανισοσκελίας, επιτεύχθηκε η αυτόνομη βάδιση, χωρίς να υπάρχει σημαντική κινητική υστέρηση. Από την ηλικία των 5 ετών, σε 3 παιδιά, έγινε η διατατική ιστογένεση με την μέθοδο Ilizarov, ενώ σε 1 παιδί κρίθηκε μη εφικτή και αντιμετωπίζεται συντηρητικά. Μια ασθενής θα αντιμετωπισθεί στο μέλλον ενώ μία ασθενής αντιμετωπίζεται αλλαχού.

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Η επιμήκυνση ήταν από 5-6 εκ, με επίτευξη αύξησης του μήκους από 33 έως 42% του αρχικού μήκους. Ο μέσος όρος του δείκτη ωρίμανσης ήταν 17 ημέρες ανά εκατοστό.

Εμφανίσθηκαν πολλαπλές επιπλοκές στην πορεία της επιμήκυνσης. Αντιμετωπίσαμε κάταγμα του νεοσχηματισθέντος οστού, αμέσως μετά την αφαίρεση της συσκευής. Εφαρμόσθηκε εκ νέου σύστημα Ilizarov για την πώρωση του κατάγματος. Μία ασθενής με υπεξάρθρημα του γόνατος αντιμετωπίσθηκε με ειδική αρθρούμενη εφαρμογή του συστήματος στο γόνατο, με μερική διόρθωση της βλάβης.

Παρουσιάζουμε τις ιδιαίτερες δυσκολίες στην εφαρμογή της διατατικής ιστογένεσης λόγω του μικρού αρχικού μήκους του μηρού και δράσης των ρικνών μυών.

Η αντιμετώπιση της υποπλασίας του μηρού αποτελεί μια πρόκληση για τον ορθοπαιδικό. Πρόκειται για ιδιαίτερα δύσκολο πρόβλημα και με πολλαπλές επιπλοκές στην πορεία αλλά με εντυπωσιακά αποτελέσματα στο τέλος της θεραπείας. Η ενεργός ζωή του Ορθοπαιδικού δεν είναι επαρκής για την αντιμετώπιση όλων των προβλημάτων του αναπτυσσόμενου παιδιού.

ΛΕΞΕΙΣ ΚΛΕΙΔΙΑ: συγγενής υποπλασία μηριαίου, ανεπάρκεια μηριαίου παιδιών, επιμήκυνση μηριαίου Ilizarov