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Original Article

Achondroplasia and limb lengthening: Results in a UK cohort and review of the literature

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ABSTRACT

Aims: We aim to review the results, complications and outcomes of a single surgeon's series of lower limb lengthening in patients with achondroplasia.
Methods: Ten achondroplastic children underwent limb lengthening. The patients, medical records and radiographs were reviewed.
Results: The average age at the time of the index operation was 7.8 years. A single surgeon undertook all procedures. The average total length gain was 20.5 cm. The commonest complication was a fractured femur after removal of the frame.
Conclusion: Although complication rates were high (70%), none were left with any long-term sequelae and all were pleased with the results.

1. Introduction

Achondroplasia is the most common genetic skeletal dysplasia. It is characterised by a rhizomelic form of dwarfism, exaggerated lumbar lordosis, a prominent forehead and a low nasal bridge. The trunk is generally of near normal length.

Society places a premium on height and the short statured population is more likely to be disadvantaged.[1] In addition an increased number of achievement problems, social skills deficits, and behavioural problems have been shown in short children.[2] Everyday activities such as shopping, using public transport, and simple bathroom and toilet hygiene may be difficult.[3]

Limb lengthening remains controversial in patients with achondroplasia. Limb lengthening is associated with a high complication rate, particularly stiffness of adjacent joints and fractures leading to a poor outcome.[4] Many authors suggest a goal of lengthening a bone segment to 20% of its original length.[5]

The high risk of complications needs to be carefully balanced against any potential cosmetic gains. Improved techniques and understanding of distraction osteogenesis in limb lengthening has led to an increase in the number of limb lengthening procedures and more successful outcomes. In addition the rhizomelic pattern of dwarfism in achondroplasia lends itself favourably to limb lengthening to restore more normal body proportionality.

A growing number of patients request limb lengthening with the awareness that this is the only effective treatment to increase their height. The reasons behind it are numerous but share a common objective to alleviate a physical defect to improve quality of life. However there is no consensus

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Table 1 – Patient details.

Patient no.	Age at presentation	Current age	Index operation	Length gained	Age at 1st treatment	Sessions	Last f/u
1	6	18	Jul-03	20 cm	9	4	Jul-11
2	21 m	17	May-01	20 cm legs, 6 cm arms	6	4	Oct-11
3	10	23	Jan-00	22 cm	11	2	Sep-07
4	4	9	Mar-08	22 cm	5	2	Feb-12
5	7	27	Mar-95	20 cm	10	2	Feb-05
6	5	20	Sep-00	22 cm	8	2	Jun-04
7	5	18	Jan-01	23 cm	6	4	Oct-09
8	6	21	Mar-99	24 cm	7	4	Jan-06
9	8	27	Jul-95	20 cm	9	2	Sep-97
10	3	19	Jan-00	22.5 cm	7	4	May-08

amongst doctors who deal with the physical, psychological and social problems associated with a short stature.

We aim to review the results of a UK cohort of patients with achondroplasia who underwent lower limb lengthening and the complications encountered. We also review the current literature on the topic in an attempt to portray the potential risks and benefits.

2. Materials and methods

The results of a single surgeon's series of lower limb lengthening in patients with achondroplasia were reviewed. The patients were followed up to skeletal maturity. All were lengthened using modern distraction osteogenesis techniques with either an Ilizarov or Taylor spatial frame (TSF). The patients, medical records and radiographs were reviewed.

The complete treatment episode involved a number of stages. Initially a pre-operative workup visit was undertaken with radiographs followed by operative planning. The patients and their families were counselled in specialist multidisciplinary limb reconstruction clinics with each stage explained as well as the likely treatment time, the importance of physiotherapy and the frequency of follow up visits. The patient and parents were introduced to other patients with comparable circular frames, and in many cases were put in contact with other persons with achondroplasia who had, or were undergoing, a similar lengthening procedure.

2.1. *Operative technique*

All procedures were similar with a general anaesthetic, supine position and no tourniquet. The circular frame and metaphyseal corticotomy were performed in a similar manner as described by Ilizarov.[6,7] Patients were treated with crossed lengthening (simultaneous lengthening of one femur and contralateral tibia). This technique ensures that pelvic balance and gait is not altered significantly and that excessive load is not applied to the joints – especially the knee, and if the patient wishes to abandon further treatment their overall leg lengths are equal (despite having knees at different levels). Lengthening of the contralateral bones was commenced approximately six months after completion of the first stage.

In five out of the ten patients (50%) lengthening was performed in 2 stages, usually of 10 cm each. In the other 50% four stages were performed of roughly 5 cm each time.

2.2. *Postoperative rehabilitation*

Patients and parents were educated in the care of the fixator and pins. Physiotherapy was commenced on the day after surgery and discharge was generally 5–7 days after surgery. One week was allowed before distraction was commenced and thereafter the usual rate was 1 mm/day. Outpatient review fortnightly with radiographs was routine. The regenerate appearance was monitored and the rate of distraction was adjusted accordingly. The regenerate column should ideally be of the same width as the bone above and below.

When the desired length had been achieved, distraction was ceased and patients were seen on a monthly basis during this consolidation phase. The fixator was removed when three cortices were evident on the column of regenerate on the anteroposterior and lateral radiographs. A Sarmiento cylinder cast was routinely applied for 6 weeks after tibial frame removal.

3. Results

Ten children with achondroplasia underwent limb lengthening procedures between 1995 and 2010. The average age at presentation was 5.6 years. The average age at the time of the index operation was 7.8 years (see Table 1 for details). The average total length gain was 20.5 cm. A single surgeon undertook the procedures in two different specialist tertiary referral paediatric orthopaedic centres. All patients except one have been followed up to skeletal maturity and all were happy with the results achieved.

3.1. *Complications*

The commonest complication in our cohort was a fractured femur after the frame was removed (see Table 2). This was seen in four patients (40%). All were openly reduced and fixed internally and went on to unite uneventfully. No significant difference was seen between the groups lengthened in 2 or 4 stages.

One patient suffered a common peroneal nerve palsy, which resolved - the nerve was explored and decompressed. Ankle equinus was observed in two, one of which required tendo Achilles lengthening. One of the tibial lengthening's required a concertina manoeuvre with alternate compression and distraction of the regenerate to achieve full union.

Two of the ten patients underwent humeral lengthening at their request using a monolateral fixator. Six centimetres on

Table 2 – Further patient details including complications.

Patient no.	Length gained	Complications	Stages	Comments
1	20 cm	CPN palsy. Decompressed and resolved. R femur fracture post r/o frame (conservative)	4	b/l humerei lengthening 2010
2	20 cm legs, 6 cm arms	Premature consolidation and fracture. Settled	4	Hypertrophic regenerate and lengthening increased to 1.5 mm/day for 1 week
3	22 cm		2	
4	22 cm		2	
5	20 cm	FFD ankle (TA lengthened) and premature fibula consolidation (re-osteotomised) 1st treatment. Delayed tibial union 2nd op – concertina manoeuvre	2	
6	22 cm	Fractured R femur through regenerate – ORIF March 01	2	
7	23 cm	Fractured L femur – ORIF July 06	4	Wants further tibial lengthening. Denied.
8	24 cm	Fractured L femur – ORIF Sep 00	4	
9	20 cm	Extension of frame for foot equinus	2	
10	22.5 cm	Fractured R femur – ORIF July 03, Bone graft L tibia Sep 03	4	

each side was achieved without complication. One patient requested further leg lengthening after achieving an initial gain of 23 cm, but was denied.

4. Discussion

Achondroplasia is the most frequently encountered form of non-lethal skeletal dysplasia[8] and is characterised by defective enchondral ossification owing to a defective gene encoding for fibroblast growth factor receptor 3. It is a type of rhizomelic dwarfism with an incidence of approximately 1 in 10,000 live births.

Achondroplasia is associated with both physical and psychological handicaps owing to the disproportionate short stature and difficulty in performing routine activities of daily living that others take for granted. These individuals often feel different from their family and peers. In addition to short stature angular limb deformities, spinal stenosis and craniofacial abnormalities are commonplace.

Improved techniques of distraction osteogenesis have renewed interest in limb lengthening for achondroplasia and Ilizarov's method has been used for some decades, with varying success rates.[9–13]

The concept of leg lengthening is a contentious issue. Although total height can be increased, it is unclear whether lengthening influences patient function or quality of life. Furthermore the high complication rate may not outweigh any of the potential benefits. Kim et al[14] assessed whether patients were satisfied with leg lengthening using the AAOS lower limb, SF-36, and Rosenberg self esteem scores. The minimum follow up was 4.5 years. They concluded that even with numerous complications, serial limb lengthening is a good option in terms of improvement in quality of life scores. They found patients improved in the mental components of the SF-36 and the Rosenberg self-esteem questionnaire. There was no real difference between the operated and non-operated groups in terms of the physical and functional components of the AAOS and SF-36 scores. All scores were reduced if the complication rate rose to more than four.

Aldegheri and Dall'Oca[9] reported 85–95% of patients were satisfied with the surgery. They reported no major complications and the patients had resumed a normal social life after limb lengthening. Similarly Vargas Barreto et al[15] showed that all patients who had no or only minor complications were satisfied with the results.

The rhizomelic pattern of dwarfism lends itself favourably to limb lengthening to restore a more normal body proportionality. Achondroplastic dwarfs usually have normal joint structure, so the height gained from limb lengthening should improve function as well as cosmesis. Achondroplastic patients show increased ligament and joint laxity, their muscle length exceeding bone length before lengthening[4] thereby facilitating the lengthening process.

Some authors have noted a relationship between growth rate and limb lengthening. Shapiro[16] found that patients with a congenitally short tibia showed marked inhibition of growth after lengthening, and postulated that the soft tissues adapt poorly to lengthening and inhibit the physis – an increase in pressure is transferred to the physis inhibiting longitudinal growth[17]: the Hueter–Volkmann principle. Song et al[18] demonstrated similar findings of early physeal closure, mainly affecting the proximal tibial physis – which has a relatively poorer soft tissue envelope compared with the distal femoral physis. They found a significant decrease in mean growth rate relative and early physeal closure compared to a control group after extensive lengthening of more than 50% of the bone segment. This is in contrast to Paley[4] and Aldegheri,[19] who both reported the capacity for extensive lengthening in achondroplasia patients with few complications was due to the proportionally longer muscles and vessels compared to the bones.

Venkatesh et al[20] reviewed 20 achondroplastic patients and in their series of 40 lengthening's had a fracture rate of 15%. Others had a rate of 20%[5] and 27%.[21]

The question of age is a difficult one. In order to keep the child within a reasonable height of his, or her, peers lengthening should be started around 6–8 years of age. This is however before the age of when a child can decide for him or herself what they really want. Furthermore children's bones are smaller in both width and length. The proportion of increased length becomes much larger, leading to increased complications.

Ilizarov believed and has documented less cranial and facial disproportion than expected, and even an improvement of existing disproportion following limb lengthening.[22] He

believes this is due to the effect of growth stimulation from extensive limb lengthening. After the child has reached adulthood the body image is well established. On the other hand lengthening may inhibit growth and have adverse effects on the physis as shown by Song[18] and Ganel.[23] Both have suggested it may be better to defer lengthening until skeletal maturity to allow for maximal length gain.

The deformities seen in achondroplasia can be corrected simultaneously: lumbar hyperlordosis with an extension osteotomy of the femur; varus deformity of the leg and the disproportionately long fibula may be reduced to normal length during the lengthening process.

The soft tissues in achondroplasia are usually redundantly long. Agostini[24] performed angiograms before and after lengthening and found that the vessels were serpentine beforehand and straight afterwards. Schurov[25] found that the muscle lengths were proportionally longer relative to the bones in achondroplasia. These combined make lengthening easier in achondroplastic dwarfs with fewer soft tissue complications.

Distraction osteogenesis using an external fixator is now the standard technique for limb lengthening. Humeral lengthening is less frequently performed than tibial or femoral lengthening and there are fewer reports in the literature.

Finally a pre-operative psychological assessment, a team approach, and assessment and counselling for specific problems are important factors in the selection process.

5. Conclusion

We present a UK cohort of patients with achondroplasia that underwent leg lengthening using a circular frame and modern distraction osteogenesis techniques. The average length gained was 20.5 cm. This was enough to allow functional gains and quality of life improvements. Although complication rates were high, none were left with any long-term sequelae.

Surgical lengthening of the limbs is invasive, dangerous, complex and long-term. It is associated with a high degree of risk. Moreover it requires a special psychological approach on the part of the patient and their families. However, it can be a reasonable option for patients with achondroplasia, not only for improving height but also improving self-esteem and quality of life. Patients need to be carefully selected, counselled and monitored frequently with any complications promptly addressed when they occur.

Conflicts of interest

All authors have none to declare.

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