Achondroplasia: lower limb axial deviations

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Only those deformities that are associated with achondroplasia (ACH) in the strictest of senses will be discussed in this chapter, thus excluding other causes of short stature such as pseudo-achondroplasia, in which case lower limb deviations are more severe and more invalidating. Axial deviations in hypochondroplasia may respond to comparable approaches as achondroplasia.

Most patients with achondroplasia do not suffer from functional limitations other than short stature and have a sustained physical activity, as is evident by their regular participation in show business, the circus, and televised games (Fig 1).

The more invalidating functional limitations are the consequence of associated spinal deformities. The latter should be ruled out by a thorough physical exam. A full spine MRI should be obtained before any surgical intervention requiring general anesthesia is undertaken. However, coronal axial deviations of the lower limbs are frequently encountered and may sometimes lead to functional limitations, both in childhood, and later in life due to early osteoarthritis.

It is estimated that around 50% of these patients suffer from significant coronal axial deviations, around half of which would require treatment [1].

1. Description of the deformities

These deformities are almost always in varus. The primary deviations in valgus in patients with ACH are exceptionally rare.

1.1 Bony deformities

Similarly to all children, physiologic varus during gait acquisition is found in achondroplasia. This varus deformity can sometimes spontaneously correct itself or, contrarily, persist and progress.

The deviation is usually bilateral and symmetric. It is found mostly at the level of the tibia and is a complex, three-dimensional deformity. Along with varus, there is usually internal rotation and sometimes recurvatum [2,3]. Varus may be located at the level of the proximal metaphysis, distal metaphysis, or even at both metaphysis. Proximal localization of the varus deformity is found in younger children, while distal locations are later in onset. Varus deformity is usually not epiphyseal in origin [4] (Fig 2).

The fibula is often too long compared to the tibia. The role of the relative excess in fibular length on the development of tibial deformity has been explored since the times of Ponseti in 1970 [5]. It may seem logical for this excessive lateral growth to lead to varus constraints on the Tibia. Studies attempting to show this relationship are contradictory [4]. According to Ain in 2006, the amount of excessive fibular length is not proportional to the severity of the varus deformity [6]. In contrast, Lee in 2007, while studying the morphology of the entire lower limbs, found a positive correlation between fibular length and varus deformity, but only in patients with unfused physes [7].

This medial rotation may also be explained by the excess in fibular growth owing to the axis of the diaphysis, the distal half of which is slightly oriented anteriorly. This could prevent the physiological progressive lateral rotation expected at the level of the tibia.

1.2 Laxity

The other characteristic findings specific for achondroplasia is coronal laxity of the knees [8]. This could be induced by three morphological elements, as well as constitutional laxity:

- The femoral and tibial epiphysial length is insufficient which would limit collateral ligament tension.
- The excessive length of the fibula would elevate the distal insertion of the lateral collateral ligament.
- The femoral condyles are twisted thus giving a typical radiographic appearance: The proximal insertions of the collateral ligaments are lowered (Fig 3).

2. Short term consequences

The varus and internal rotation deformities can lead to discomfort during gait, sometimes leading to fatigability, falls, and secondary pain. Laxity, especially lateral, aggravates varus and vice versa. The femoro-tibial joint may lose its congruency, which would lead to painful subluxations during ambulation. The epiphyses, initially normal, may become secondarily deformed.

The consequence is painful functional limitation which may sometimes appear as early as the end of the first decade [9]. Although progression toward osteoarthritis may be presumed, it is actually rare. This may be due to a certain protection conferred to the cartilage by the FGFR3 gene mutation that itself is responsible for the ACH [10,11]. However, when arthritis does develop, it is particularly difficult to treat due to the small size of the bony elements and the associated laxity [12,13].

As a result, clinical gait analysis, assessment of fatigability and pain, clinical and radiographic evaluation of axial deviations and its progression are thus important to assess during infancy and when approaching adolescence (Fig 4).

3 Management

Over half of patients with achondroplasia would require only observation and would not require treatment.

For the rest, indications for treatment depend essentially on functional criteria. Angular criteria by themselves are not sufficient for preventive surgical intervention in the absence of symptoms [9,14].

3.1 Functional treatment

Both device-assisted prophylactic and preventive treatment strategies are contraindicated. Such treatment strategies would be difficult to implement, ineffective, and dangerous, since they may lead to worsening of the laxity.

Although rehabilitation may be effective to counteract hip flexion contracture, it is ineffective in preventing varus.

The only acceptable treatment strategies are surgical and include growth modulation and/or osteotomies [14].

3.2 Growth modulation

Despite some attempts, there is no consensus on treatment with classic epiphysiodeses techniques, such as staplers, screws, or curettage. The risks related to the alteration of growth that is already very limited, the unpredictability of the outcomes, and the understandable hesitation of the families, do not allow us to suggest these techniques.

Growth modulation, which respects the physis, which is possible with 8-plates, is theoretically more acceptable. Very few surgeons have the required experience. McClure's small case series published in 2017 showed a tendency toward the improvement of varus with a low complication rate. Nevertheless, complete correction was only obtained when associating femoral, tibial, and fibular growth modulation before the age of 5 years old. The impact on the patient's final height is unknown [15]. Consequently, isolated asymmetrical epiphysiodesis does not seem to be an appropriate treatment strategy.

Some surgeons have suggested isolated early epiphysiodesis of the proximal fibular. Such interventions would make sense mechanically, but the evaluation of outcomes is still in progress. This intervention requires a direct approach with curettage of the physis while protecting the common fibular nerve. There is still no consensus on its effectiveness or the age at which it should be undertaken.

Weiner recently published a series on planned fibular pseudarthroses which have shown their effectiveness on the partial correction of varus. This furthers the idea that excessive fibular length may be an actor in the development of varus but poses the problem of early aggressive surgery [16].

3.3 Osteotomies

The gold standard in surgical treatment is tibial osteotomy.

The objective of such a treatment is to normalize the axis of the limbs, first to improve function, and secondarily to reduce the risk of arthritis.

Patients are not generally operated before the ages of 7 or 8 years, or even adolescence. Early surgery has a high risk of recurrence.

Osteotomy is often bilateral and undergone simultaneously.

The osteotomy site should be the location of the varus deformity:

- Most often proximal, sub-tuberosity
- If required, distal metaphyseal
- Middle of the diaphysis if the varus is both proximal and distal
- Bifocal

Correction of the internal rotation deformity is undergone simultaneously. Adjustment is done with a plumbline which requires the visualization of both lower limbs in their entirety in the surgical field to locate the hips.

The femoral head, patella, and middle of the tibio-talar joint should be aligned.

Owing to the smaller size of the bones, the angular correction required to correct the axial deviation is proportionately higher than in a typically developing child (Fig 5).

Contrary to other pathologies, ACH does not justify hypercorrecting the deformity, but a slight valgus of the lower limb should be the goal in order to neutralize lateral hyperlaxity. Fixation of the osteotomy is then achieved with a plate and screws.

A shortening osteotomy of the middle 1/3 of the fibula is generally required. Proximal fibular epiphysiodesis may also be discussed if there is remaining growth.

It is possible to undertake two osteotomies through a single antero-lateral approach and to place the plate on the lateral aspect of the tibia (Fig 6).

In the case of associated femoral varus, a femoral osteotomy may also be undertaken in order to avoid any obliquity of the joint line which would not be tolerated.

There is a risk of compartment syndrome which would justify preventive fasciotomy, at least of the anterior and antero-lateral compartments through the same approach as was used for the

tibial and fibular osteotomies, as well as control of compartment pressure according to the preferences of each surgeon.

It should be noted that spinal anesthesia is contraindicated in this case due to the high risk of associated lumbar stenosis [14].

Coronal laxity, especially medial, may render the surgery difficult and falsify the assessment of limb alignment, thus increasing the risk of hypercorrection. Provisional femorotibial epiphyseal pinning in extension before undertaking the osteotomy may be helpful, with the pin being left in place until after fixation has occurred [14] (Fig 7).

3.4 Limb lengthening

If the rare indication for limb lengthening were to be met, a simultaneous correction of the axial deviation may also be required.

This is frequently done with Ilizarov-type circular or hexapodal external fixators. Outcomes and complication rates should be weighed against gain in centimeters [17-19]. The use motorized lengthening nails would reduce the invasiveness of these types of surgeries while allowing a direct correction of the axis. These interventions are especially interesting in hypochondroplasia.

Conclusion

In our experience as well as in the literature, there is little consensus on the management of patients with achondroplasia and many questions which remain to be resolved.

Lower limb varus is inconsistently found, and its treatment, which is exclusively surgical, is only indicated in case of notable functional limitation.

The gold standard in treatment is tibial and fibular osteotomy, the techniques of which should be adjusted based on the patient’s age and location of varus.

The questions which remain to be answered include:

- What is the pathophysiological mechanism of varus? Specifically, what is the role of the relative excessive fibular length?
- To what degree is varus tolerated in the long term? To what extent can this varus be treated with growth modulation?
- What is the prevalence of knee osteoarthritis in adults with achondroplasia?

Fig 1: Two patients with achondroplasia without symptomatic varus.

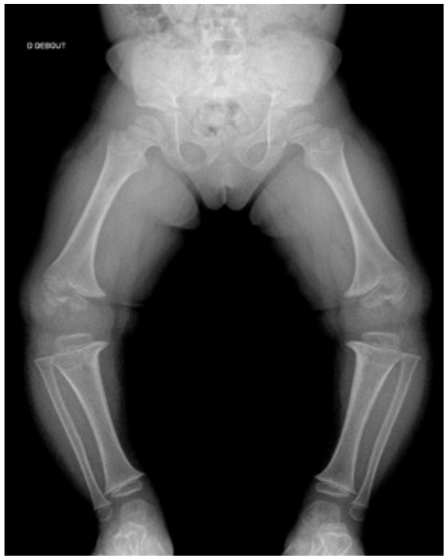

Fig 2: Genu varum worsened by lateral laxity.

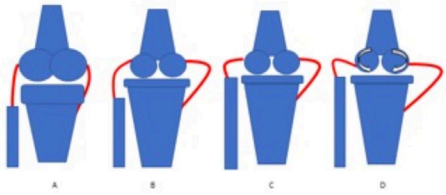

Fig 3: Elements contributing to laxity. A) Normal knee. B) Reduced epiphyseal width. C) Fibular head ascension. D) Condylar rotatory deformities

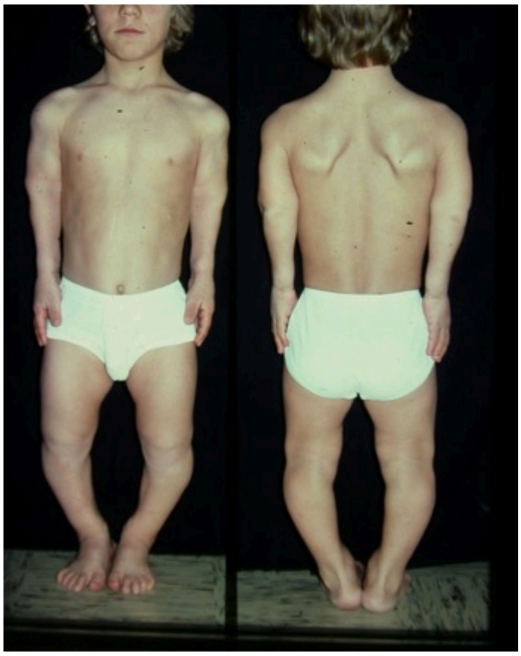

Fig 4: Symptomatic varus deformity.

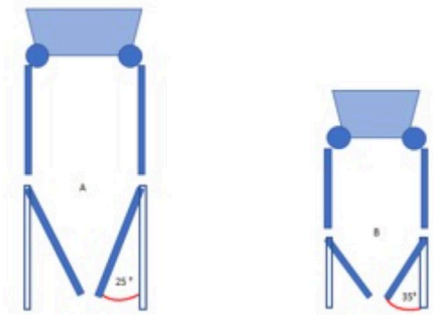

Fig 5: For a proportionally identical intercondylar width, more axial correction would be required in patients with achondroplasia (B) compared to patients with a normal height (A).

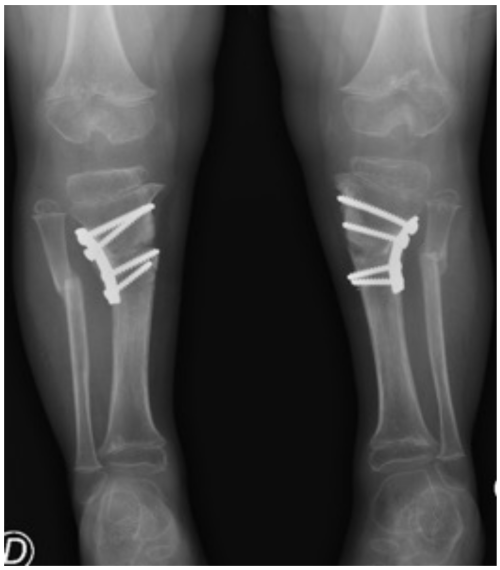

Fig 6: Dual bilateral osteotomies and fixation with lateral plates.

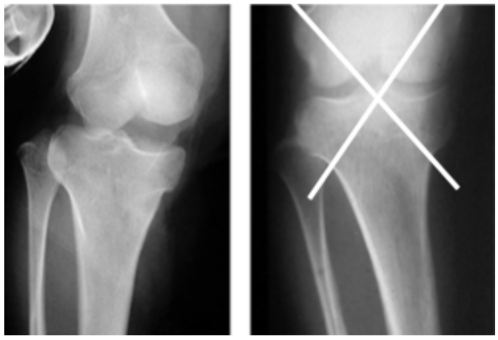

Fig 7: Medial laxity. Temporary intra-operative pinning.

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