Eur Spine J
DOI 10.1007/s00586-016-4861-1

GRAND ROUNDS

The natural history of thoracolumbar kyphosis in achondroplasia

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Received: 30 April 2016 / Accepted: 4 November 2016
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Abstract

There are little published data on the natural history and evolution of thoracolumbar kyphotic deformity in achondroplasia. Furthermore, there are few published reports indicating the probability of symptoms and at what kyphotic angle progressive deformity might occur. The lack of knowledge of the fundamental natural history does not allow firm guidelines for the management of this problem. In this respect the role of bracing remains uncertain both in terms of altering the prognosis for further vertebral wedging and overall sagittal profile. This report describes three illustrative cases of thoracolumbar kyphosis (TLK) that occurred in achondroplastic infants. The long-term outcomes are presented to illustrate the diverse clinical behaviour of TLK in this patient group.

Keywords Achondroplasia · Thoracolumbar kyphosis · Bone dysplasias · Gibbus

Case presentations

Case 1

The first case is a 12 years old female who has been under follow-up for 6 years since she was initially seen in the outpatient clinic.

This infant female at first presentation aged 6 years, had a TLK angle of 57°. She was asymptomatic, and walking independently without aids. She had a normal neurological examination.

Diagnostic Imaging

On her regular annual follow-up, she had whole spine X-rays that shows increased TLK progressively rising to 70°, 97°, 98° and 104° at age of 10, respectively (Figs. 1, 2).

Whole spine MRI (Fig. 3) showed stretching and compression of the cauda equina over a 114° gibbus.

Outcome

Although at aged 11 years she had normal activities of daily living she developed increasing bilateral sciatic pain and neurogenic claudication symptoms.

At the time of writing the parents preferred a watchful wait policy.

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Published online: 22 December 2016

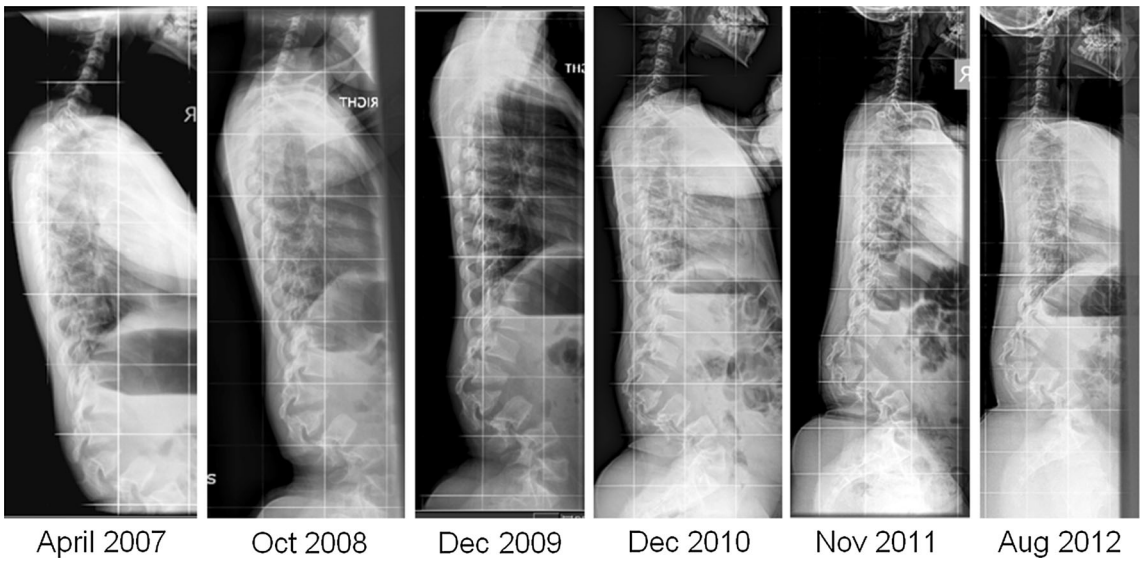
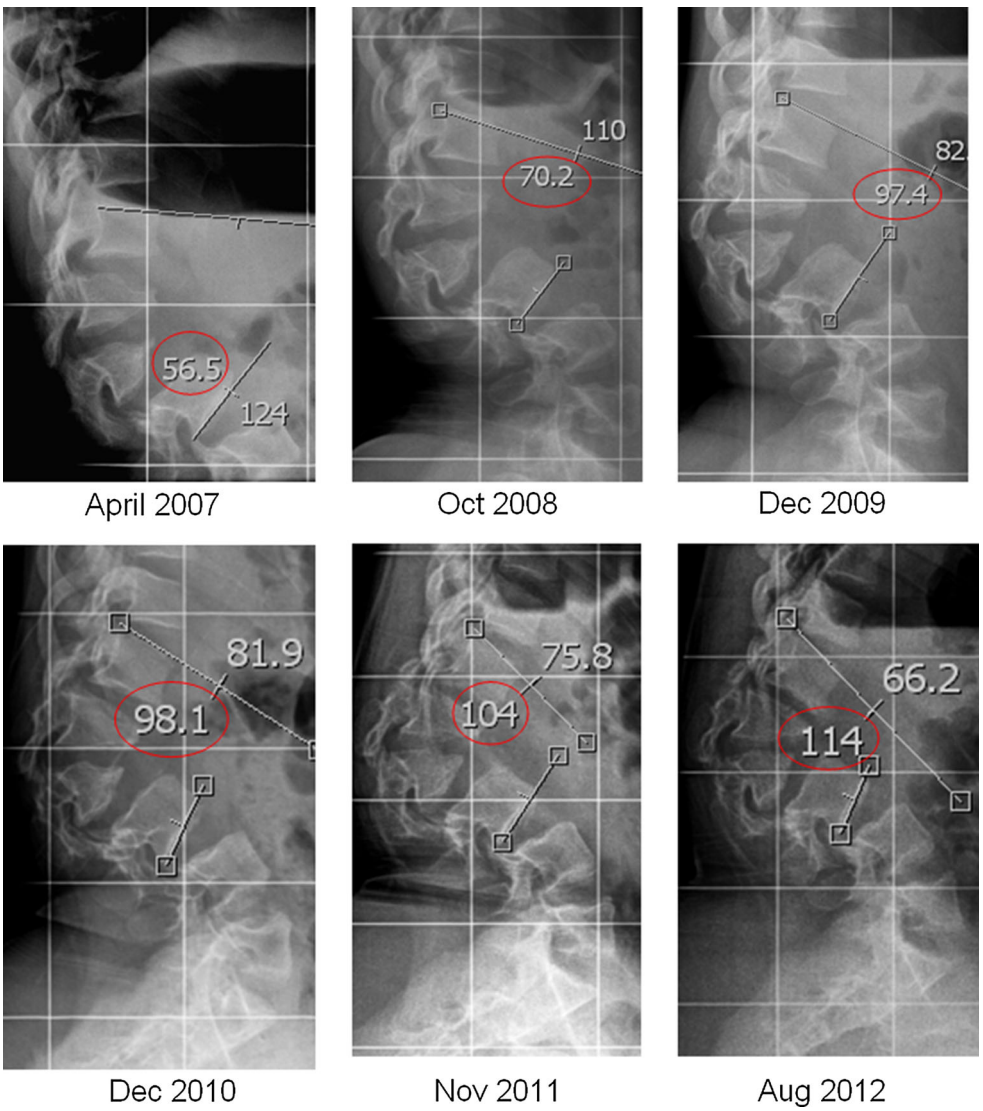


Fig. 1 Case 1: whole spine lateral X-rays showing progression of TLK and sagittal profile

Fig. 2 Case 1: coned lateral thoraco-lumbar X-rays



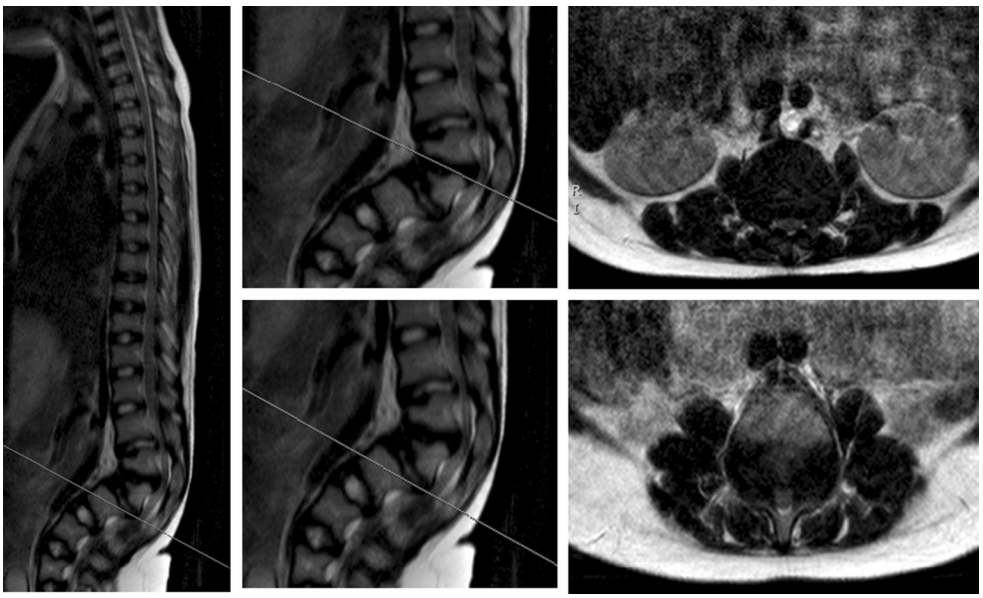


Fig. 3 Case 1: sagittal and axial MRI sections demonstrating spinal canal stenosis and thoracolumbar gibbus

Case 2

An infant with known achondroplasia was noted at the age of 2 years with a thoracolumbar kyphosis. With increased walking this deformity increased over the next 4 years. At 5 years of age he presented with increasing lower back pain and stiffness in the legs. Physical examination showed a large thoracolumbar kyphosis and an exaggerated lumbar lordosis beneath it. There was bilateral clonus with extensor plantar responses.

Imaging

MRI scan showed conus terminating at T12/L1 with severe L2 vertebral wedging and stenosis at that level.

Procedure

In 2003 he underwent first-stage L1 and L2 vertebrectomies. One week later he underwent posterior instrumented correction of the deformity and insertion of fibular allograft strut graft.

Procedure rationale

In surgical planning the approach should be carefully considered. Posterior alone, stand-alone and combined are the available approaches for surgical interventions. Spinal surgeons should respect the stability and the biomechanics of the spine when operating on this pathology. Not only the neurological decompression is the target but also the deformity correction and prevention of subsequent deformity and adjacent levels affection to be considered.

In rare cases of low (<30°) symptomatic thoracolumbar kyphosis, a posterior decompression (laminectomies or multiple foraminotomies) with instrumented fusion is an option. The classic presentation in this group of patients is due to foraminal canal narrowing.

In kyphotic angles more than 30° or compromise of the spinal cord, anterior vertebrectomy and instrumented posterior fusion is mandatory [14, 15, 16].

Spinal cord and cauda equina compression are considered surgical emergencies, which need urgent surgical intervention. These usually occur with fixed angular deformity. In these situations more complex, staged operations will be necessary, depending on the particular deformity characteristics and patient's features.

In 2010, Auregan et al. reported a case of 180° achondroplastic kyphotic deformity which illustrates some of these principles. Their case was first seen at the age of 16 years old because of cosmetic concerns and was neurologically normal. At the age of 18, he developed severe neurogenic claudication with abnormal central spinal cord signal in front of the apex of the kyphosis associated with the narrow spinal canal. The authors performed decompression of the spinal cord with five-level hemilaminectomies, decancellation osteotomy of three apical vertebrae with disc excision and translation of the spinal cord anteriorly and posterolateral fusion. A peroneal strut graft was inlayed anterolaterally. Patient was immobilized in a cast for 3 months, then, 6 months in a moulded orthosis. There was improved clinical status after 3 years [17].

Pedicle screws have been recently considered as the implant of choice in management of the achondroplastic deformities because of the abnormally narrowed spinal canal, which is likely to be compromised if, hooks or wires are used.

The conservative treatment has been offered to our cases as long as there is no incapacitating spinal deformity nor any neurological deficit was encountered. They are also kept under yearly basis follow-up for clinical and radiological evaluation. Whole spine standing PA and lateral X-rays are routinely performed for assessment of the whole spine sagittal balance as well as thoracolumbar kyphosis. Surgical treatment was offered to the case which neurologically deteriorated.

Postoperative imaging

Outcome

His symptoms resolved and his walking ability improved. Four years later he was walking normally without symptoms.

Case 3

This male child was noted at birth to have a TLK gibbus and initial X-rays showed an angulation of 25°.

Diagnostic imaging

Outcome

One year later, additional wedging of L2 occurred and increased to 34° till the age of three at which point progression ceased (Fig. 4). Thereafter there was regression of TLK and correction of the gibbus; the TLK had reduced to 2.5° at the last follow-up 5 years after initial presentation. This has remained stable in the intervening years (Fig. 5).

Historical review

Achondroplasia ("without cartilage formation") was first described in 1878. It is an autosomal dominant inherited mutation in the fibroblast growth factor receptor-3 (FGFR3) gene on the short arm of chromosome 4. This affects the maturation of chondrocytes in the growth plate [1]. However, 80–90 % of the patients have new mutations [2]. It is the most common form of human skeletal dysplasia, with frequency of between 1 in 15,000 and 1 in 40,000 live births [3].

Intracartilaginous ossification commences in the developing fetus in the thoracolumbar region and progresses in a cranial and caudal direction from the thoracolumbar junction. The primary ossification centers are located in the vertebral centrum and one on each side in the posterior elements, located anterior to the pedicle. The junction of these is the neurocentral synchondrosis. During maturation, there is increasing vertebral size and progressive expansion of the spinal canal. Fusion of these synchondroses at 6–8 years of age signals the cessation of spinal canal widening. This is also the period in which longitudinal growth of the posterior elements of the vertebrae ceases.

Anterior longitudinal growth, which occurs at the epiphyseal plates, continues in individuals to the age of 18–20 years. Factors interfering with the anterior longitudinal growth during the intervening period will, therefore, be accompanied by kyphosis [9].

Radiologically, the achondroplastic spine has short pedicles, particularly in the thoracolumbar region and progressive decrease in interpedicular distance in lumbar

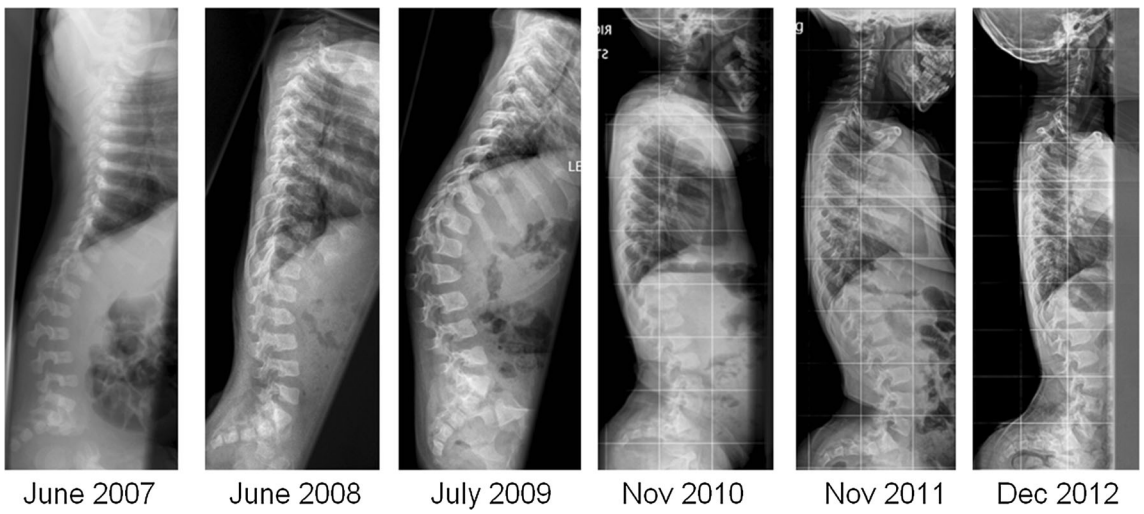


Fig. 4 Case 3: whole spine lateral follow-up X-rays showing spontaneous correction of TLK

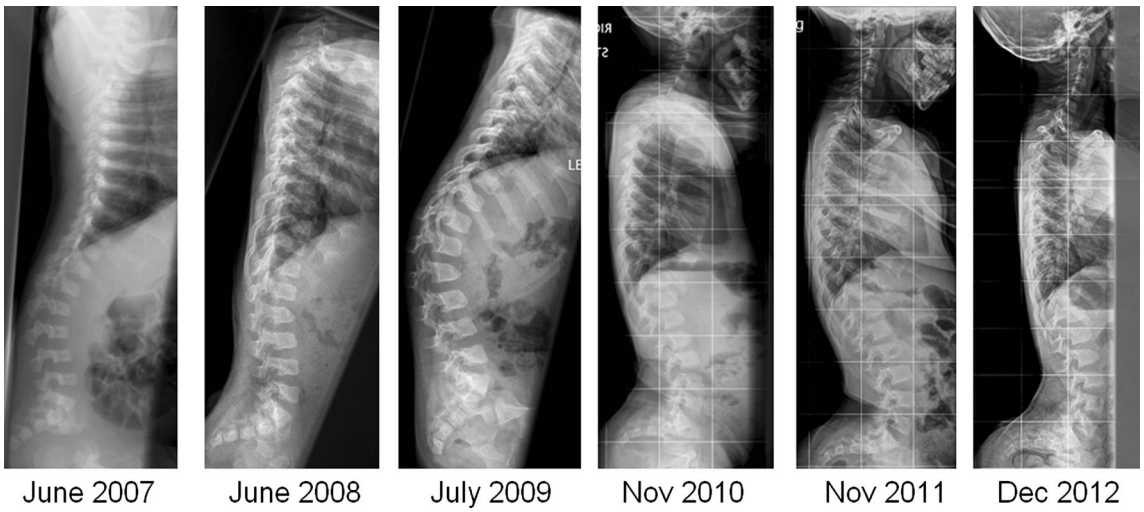


Fig. 5 Case 3: coned thoraco-lumbar lateral X-rays

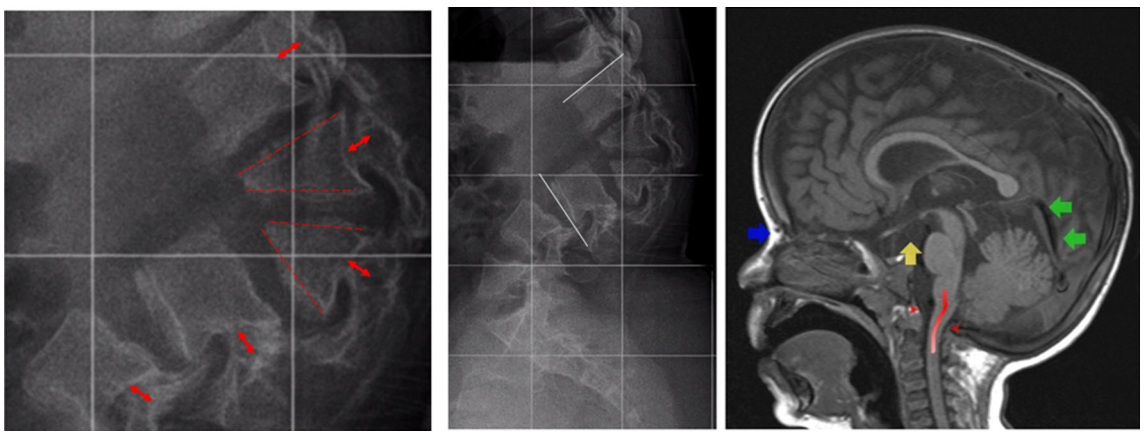


Fig. 6 (*Left*) Wedging of the vertebrae at the apex with short pedicles. (*Centre*) Thoraco-lumbar kyphotic deformity. (*Right*) Relatively large cranial vault with small skull base, prominent forehead with depressed nasal bridge (*blue arrow*), narrow foramen magnum is narrowed (*red arrowheads*), cervico-medullary kink (*red line*). Elevation of the brainstem gives rise to a large suprasellar cistern (*yellow arrow*) and a vertically oriented straight sinus (*green arrows*). Cranio-cervical junction constriction

spine, which is progressively smaller in the caudal direction [6]. In some cases there is narrowing of the foramen magnum and a short clivus. Most of the cases are characteristically accompanied by kyphosis at the thoracolumbar junction. When the kyphosis progresses there is wedging of the vertebral bodies at the apex of the deformity (bullet-shaped/hypoplastic vertebra) [7].

Other radiological features include posterior vertebral scalloping, laminar thickening, widening of intervertebral discs and increased angle between sacrum and lumbar spine [8] (Fig. 6).

Disrupted intracartilaginous ossification at the neurocentral synchondroses is thought to be the basis for the abnormal growth of the axial skeleton in achondroplasia. As the synchondroses have an oblique orientation, their normal growth results in an increase of the spinal canal in all dimensions, as well as growth of the pedicles. Abnormal maturation of these synchondroses results in short pedicles with a narrow spinal canal. A simultaneous occurrence is the underdeveloped and narrow sacrum, because this too forms from intracartilaginous ossification. The iliac wings undergo unimpeded growth and hence are located relatively higher in achondroplastic individuals. Consequently, the sacroiliac articulations are well below the iliac wings [10].

At birth, there is normal prominence of the mid-to-lower back with a small TLK, which usually resolves by the time of independent walking. Lumbar lordosis then evolves and increases till skeletal maturity.

In achondroplasia the exact factors associated with progression of TLK and the subsequent development of fixed kyphosis in adolescence or adulthood have not been fully elucidated but are thought to be connected to generalized hypotonia, a large head, hydrocephalus, and delayed walking [5, 13, 14].

Siebens et al. presented a hypothesis explaining the persistent curvature and resultant TLK in achondroplasia. The postulate was that occurrence of the deformity was related to the disproportionately enlarged head, lax ligaments with unusual vertical load on the anterior column of the spine. This is most apparent at the thoracolumbar junction where the transition from the relative rigidity of the thoracic cage to the upper lumbar spine creates an additional stress riser. Inhibited anterior vertebral growth reflects the Heuter-Volkmann principle on the vertebral apophyses and the subsequent development of vertebral wedging.

Neurologically they are liable to spinal compression because their spinal canal is abnormally narrowed by the short pedicles as well as by the compensatory hyperlordosis [11].

With walking and maturation there is exacerbation of thoracolumbar kyphosis with compensatory lumbar hyperlordosis, pelvic tilt and subsequent fixed flexion deformity of the hip joints [12].

The above hypotheses would accord with a reported prevalence of thoracolumbar kyphosis in achondroplastic children under 1 year of age of 94 % [4].

However, the biomechanical postulate would predict that with the initiation of wedging of the vertebral body and subsequent adoption of walking stance, there would be increasing differential growth between the anterior and posterior halves of the vertebrae. Thus, invariable progression of TLK should be the rule. This is illustrated in cases 1 and 2.

In contrast, the reported risk of progressive deformity and resultant spinal cord or cauda equina compression is only approximately 11 % [5]. In addition, the above theory cannot explain the regression of TLK seen in case 3. This anomaly cannot be explained by differential muscle tone as these reported cases were walking normally throughout the later follow-up period.

In summary, TLK is very common in achondroplastic infants. This deformity appears to defy the Heuter-Volkmann principle in that progression is not invariable and in certain instances may resolve. Generally the treatment options for treating the TLK deformity depend on the flexibility as well as the time of presentation. Early life diagnosis should be treated conservatively then bracing with a serial follow-up and curve assessment. If this line of treatment fails, then surgery will be an option. In adults or undiagnosed cases with a fixed deformity, there is no role for the conservative treatment. The progression of the deformity and the neurological impairments are the factors, which should be considered for decision-making.

Compliance with ethical standards

Conflict of interest None.

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