Thoracolumbar spinal deformity in achondroplasia

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The authors review the management of thoracolumbar kyphotic deformity in cases of achondroplasia. The presence of angular thoracolumbar kyphosis in achondroplasia is well recognized. In children this is initially a nonfixed deformity that persists, however, in more than 10% of individuals and becomes a fixed thoracolumbar kyphotic deformity. Additionally, with the coexistent spinal canal stenosis, neurological damage can occur and manifest as spinal cord or cauda equina compression. The nature of this condition, the natural history, and management options are discussed. Anatomical and biomechanical factors relevant to the condition are specifically highlighted. Avoidance of pitfalls in the management of these patients is discussed for both pediatric and adult patients.

KEY WORDS • achondroplasia • kyphosis • spinal deformity • thoracolumbar spine • dysplasia

One of the chondrodysplasias, achondroplasia has an autosomal-dominant pattern of inheritance. The majority of cases arise as spontaneous genetic mutations. Point mutation on the long arm of chromosome 4 has been identified in this population. This is a dysplasia with predominantly metaphyseal involvement. At a cellular level there is an abnormality in the chondrocytes in the growth plate, and the process of their maturation and a defect in the Type 3 FGF receptor has been demonstrated.*1,5,16,21 It is known that unlike Types 1 and 2 FGF, which are expressed early in the fetal period on the periosteum and perichondrium, Type 3 FGF is found later in fetal development and occurs on maturing chondrocytes.5 This metabolic abnormality affects both limb growth (with the consequent rhizomelia) and intracartilaginous spine formation (with the associated spine abnormalities).1,2,3,13,15

As a result of this dysplasia the individual has a characteristic appearance. In infants the face is normal with a somewhat low nasal bridge accompanied by a frontal prominence of the cranial vault. As the infant grows, the abnormality in the biological growth of the skeletal system becomes more obvious. Short stature accompanied by shortened limbs is notable. Characteristically, the shortening is not uniform, the arms are shorter than the forearms and the thighs are shorter than the legs. With walking and maturation there is development of thoracolumbar kyphosis with compensatory lumbar hyperlordosis, pelvic tilt, and subsequent fixed flexion deformity of the hip joints.15

Thoracolumbar deformity in patients with achondroplasia is a recognized problem.2,3,6-13,17,18,20 The deformity first occurs in infancy when the child begins sitting, and the condition follows a predictable course over time until the child is walking.7,9,13,14,20 Management strategies for this thoracolumbar deformity must address two issues. Deformity-related neurological and structural deterioration both need to be prevented and, if already present, corrected. An understanding of the anatomy and natural history of the sagittal balance in these patients will help the surgeon avoid the pitfalls of later skeletal and neurological compromise. In this paper the authors address this management strategy in patients with achondroplasia in cases of reversible thoracolumbar kyphosis and those of a fixed kyphosis.

The overall prevalence of thoracolumbar kyphosis is estimated to be 94% in children younger than 1 year of age. The incidence decreases in older children to 11% by the age of 10 years. Subsequently the prevalence is greater than 30% in patients greater than 30 years of age.9

DEVELOPMENT OF THE SPINE

Intracartilaginous ossification commences in the developing fetus in the thoracolumbar region and progresses in a cranial and caudal direction from the thoracolumbar

Abbreviations used in this paper: AP = anteroposterior; CT = computerized tomography; FGF = fibroblast growth factor; MR = magnetic resonance; TLSO = thoracolumbosacral orthosis.
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 to 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 to 20 years. Factors interfering with the anterior longitudinal growth during the intervening period will therefore be accompanied by kyphosis.

Disrupted intracartilaginous ossification at the neurocentral synchondroses is thought to be the basis for the abnormal growth of the axial skeleton. 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 low, well below the iliac wings.

**CLINICAL PRESENTATION**

**Spinal Deformity in Childhood**

Thoracolumbar kyphosis is first noticed in infancy in children with achondroplasia. This is not a congenital fixed deformity but is due to mechanical factors, specifically the general muscular hypotonia of achondroplastic children. It has been suggested that the incidence is greater than 90% in children younger than 1 year of age. In some cases kyphosis is accompanied by apical wedging of the vertebral bodies, which begins as a result of the physiological anterior compression of the vertebral growth areas. The C-shaped configuration adopted by the child’s spine in the seated position is best appreciated on the lateral plain x-ray film (Fig. 1).

Spontaneous resolution of this postural kyphosis occurs in the majority of children by the age of 3 years. Kyphosis may persist in approximately 30% of these children.

**Progressive Fixed Thoracolumbar Kyphosis in Adolescents**

Progressive fixed thoracolumbar kyphosis results from the progressive disruption of the vertebral epiphyseal ring, which begins in childhood. In the presence of an uncorrected thoracolumbar kyphotic deformity, an abnormal force is placed on the epiphyseal ring. The growth in the vertebrae is known to be sensitive to physiological forces. There is a resultant decrease in growth of the anterior column and subsequently the formation of a fixed thoracolumbar kyphotic deformity. If symptomatic, the deformity requires correction to reduce the chance of neurological injury. In the study of achondroplastic patients with neurological symptoms and neurological compromise, by Kahanovitz et al., six of the eight patients had begun experiencing symptoms prior to the age of 21 years and as early as 11 years.

**Symptomatic Deformity in Adults**

In adults with achondroplasia patients present with back pain, with or without neurological symptoms and deficit. The narrowing of the spinal canal and consequent thecal sac canal ratio decreases with the hypertrophy of the facet joints. According to a recent paper, this constriction is most marked at the L2–3 junction. The treatment of adults differs from that of adolescents in that the kyphotic deformity and canal stenosis is exacerbated by aging-related degenerative hypertrophic changes in the ligaments and facet joints of the spine. Standing renders obvious the degree of lumbar hyperlordosis that may exacerbate the lumbar stenosis symptoms because of reduced spinal canal volume.

Symptomatic progression of 80% has been reported in adults during a 10-year period by one group. These authors considered the spectrum of spinal disease to include five major groups (asymptomatic, lumbar pain, disc herniation, claudication, and paraparesis). Significant thoracolumbar kyphosis, although not emphasized by these authors, was notable in all of their patients in whom neurological symptoms developed. Thoracolumbar kyphosis may be a significant factor predictive of future onset of neurological symptoms.

**MANAGEMENT OF DEFORMITY**

As part of the initial assessment of patients with thora-
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Thoracolumbar kyphosis, a detailed history and neurological examination are required. Musculoskeletal disease–related symptoms or associated neurological symptoms must be noted. Physical examination should include determination of spinal cord or nerve compromise. Similarly, in both the history taking and examination, the physician must not neglect the coexistent abnormalities at the cranial and cervical levels. The presence of hydrocephalus and foramen magnum stenosis may cause impaired neurological status and require early intervention.

Preliminary neuroimaging studies are initially required to establish the degree of abnormality. They may then play a role in the monitoring of the deformity prior to or in response to therapy. More focused neuroimaging studies may be required if the thoracolumbar deformity is thought to require operative intervention.

**Neuroimaging Studies**

In the assessment of the achondroplastic child with kyphosis, neuroimaging should include cranial and whole-spine MR imaging. Plain radiography should include supine AP and lateral plain x-ray films of the thoracolumbar spine followed by a lateral x-ray film of the spine in the seated individual. The kyphotic angle is an objective value necessary when following the deformity and assessing its response to treatment. Comparison of the seated and supine radiographs gives a clear impression of the mobility of the deformity.

In children and adults lateral radiographs of the spine allow evaluation of the degree of thoracolumbar kyphosis and anterior vertebral wedging. Both are important pieces of information in the evaluation and long-term management of these patients (Fig. 2).

**Magnetic Resonance Imaging.** Standard MR imaging protocols for screening of the spine will provide relevant information in the management of these patients. In children the brain, craniovertebral junction, and entire spine should be examined. The normal restlessness of children will necessitate general anesthesia and airway protection for such an involved study (Figs. 3 and 4).

**Computerized Tomography Scanning.** Computerized tomography scanning provides information regarding the structural narrowing of the canal, nerve root exit, foramina, and the dimensions of the pedicles. It is important to record the diameter and trajectory of the pedicle in sagittal, coronal, and axial planes as well as those of the VBs. The CT study can be supplemented by a myelography with postmyelography CT images.

Myelography demonstrates the characteristic hourglass pattern of the intraspinal contrast, reflecting the degree of spinal canal abnormality (Fig. 5 upper left and right). The levels of most severe canal narrowing can be identified on these images (Fig. 5 lower left and right).

**DEFORMITY CORRECTION**

The decision to undertake brace or operative therapy to achieve deformity correction hinges on whether the curvature is fixed or mobile. Based on this distinction there are two groups: 1) children with a nonfixed deformity and 2) adolescents and adults with a fixed thoracolumbar kyphosis.

**Thoracolumbar Kyphosis in Infancy**

Precautions regarding posture in the care of these infants are necessary in the early phase of treatment. They include the avoidance of sitting in abnormally unsupported flexed posture. Routine upright lateral and AP radiographs should be obtained at 6-month intervals to assess the degree of thoracolumbar kyphosis until the child reaches 3 years of age.

Indications for a TLSO are a progression of the kyphosis to greater than or equal to 30°, the appearance of anterior vertebral wedging, or vertebral offset during the observation period. The TLSO must be tailored to the individual to ensure that an extension moment is created at the apex of the thoracolumbar kyphosis (Fig. 6).

As the correction proceeds, the moment will decrease and hence the TLSO will require modifications. Brace therapy–related results in infancy are encouraging. Pauli, et al. have studied infants younger than 3 years of age and found that their degree of truncal hypotonia has a positive predictive value in long-term projection for persistent thoracolumbar kyphosis. They also demonstrated control of the thoracolumbar kyphosis and a regrowth of the anterior portions of the wedged vertebrae when brace therapy was initiated prior to age 3 years.

It is important to note that when left untreated, the lesion’s natural history involves a flexible structural kyphosis becoming a fixed deformity. This is thought to result from vertebral wedging accompanying the damage to the growth plate and lack of longitudinal growth of the spine and ligamentous structures.

**Fixed Thoracolumbar Deformity**

Based on clinical and the neuroimaging findings, pre-

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**Fig. 2.** Left: Lateral thoracolumbar radiograph obtained in a symptomatic adolescent patient in whom the major symptoms were neurogenic claudication and back pain. The radiograph demonstrates a fixed thoracolumbar angular kyphosis, anterior wedging of the vertebral column, and scalloping of the posterior vertebral margin. Right: Lateral radiograph obtained in an adult patient with back pain, demonstrating the absence of anterior vertebral wedging.
operative planning determines the spinal levels to be decompressed and subsequently equipped with instrumentation for fusion. An anterior or posterior procedure alone or in combination can be considered. The approach must address the pathological entity in all planes at the affected levels. When choosing the construct, one should consider the natural history of anterior and posterior constructs, because it will avoid a major pitfall in the management of spinal disease—that is, that of delayed instability or deformation.

Stand-alone anterior constructs are known to be kyphosing in nature and are best avoided. If anterior decompressive corpectomy and reconstruction is required, this procedure should be supplemented by a posterior instrumentation and fusion. Progression of thoracolumbar kyphosis is probable following laminectomy without supplemental fusion, if the kyphotic angle is greater than 40°.20 Because of consequent loss of the posterior elements and ligamentous tension band, laminectomy alone is associated with an increased lever arm acting across the apex of the kyphotic deformity. Failure to address the kyphosis may result in ongoing pain with or without neurological sequelae.17

The angle of the kyphotic deformity is critical when considering the biomechanics of the planned instrumentation-augmented fusion. The end result is to achieve arthrodesis with or without deformity correction. The corollary to this is that the goal is to avoid pseudarthrosis, the risk of which may be greater involving the stand-alone posterior procedures if the kyphosis is greater than 50°. This risk may be as great as 55%, as reported in cases of congenital kyphosis.22 Therefore, in patients with this degree of kyphosis it is mandatory to perform combined anterior–posterior fusion. Additionally the fusion site must include the end vertebra of the kyphosis so as to avoid the shift of the kyphosing moment onto adjacent motion segments, consequently creating a junctional kyphosis. Care must be taken in the choosing of anchor points for the instrumentation. A congenitally narrow spinal canal precludes the fixation of sublaminar hooks. Pedicle screw placement is a better option. Further, it is advisable to halt the posterior fusion at L-4 to permit motion at the lumbosacral region, which combined with the pelvic motion is important for ambulation in people with rhizomelia. Fu-

Fig. 3. Sagittal MR imaging studies. Left: Cranial image obtained in a 12-month-old child with achondroplasia, revealing significant foramen magnum stenosis warranting operative intervention. Center: Spinal image obtained at age 7 months. Right: Spinal image obtained at age 12 months. Subtle changes in the spinal axis are present with mild thoracolumbar kyphosis.

Fig. 4. Sagittal MR images of the thoracolumbar spine obtained in a 24-year-old patient with symptomatic thoracolumbar kyphosis. The major symptom was debilitating neurogenic claudication.
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Positioning. In view of the short stature and compensatory hip contractures, the patient is best positioned on a Jackson table, with careful attention to pressure areas to accommodate the hip flexion contractures. This is particularly important for posterior procedures requiring the prone position.

Exposure. The planned posterior skin incision should incorporate adjacent to those levels being surgically treated. The exposure of the lumbosacral region is difficult because of the exaggerated lumbar lordosis and the relative high position of the iliac crests.

The thoracolumbar fascia is incised in the midline and the paravertebral musculature is dissected laterally to expose the facet joints bilaterally. Care must be taken not to interfere with the facet joint capsules above and below the levels to be instrumented because damaging these structures may predispose the patient to progressive deformity at the adjacent, uninstrumented level. The transverse processes, which are often dysmorphic, are exposed to facilitate the subsequent instrumentation-augmented fusion.

Anterior access to the thoracolumbar junction is obtained via a left thoracotomy. If performed through the bed of the 10th rib, this bone can be later used as autograft material. The parietal pleura is divided, and the peritoneum dissected away from the anterior and posterior abdominal walls. The peritoneum is dissected from the hemidiaphragm, which is divided circumferentially. A cuff of the hemidiaphragm is left on the thoracic wall for later repair. The hemidiaphragm and cuff are tagged with sutures for accurate reapproximation at the time of repair. The dissection is commenced at the level of the intervertebral discs.
bral disc after intraoperative radiography confirms the level of the spine. Segmental vessels are ligated and the psoas muscle is detached from the spinal column and mobilized posteriorly to expose the anterior margin of the intervertebral foramen. This exposure permits partial vertebrectomy, metal cage–assisted reconstruction (packed with allograft), and placement of vertebral screws and rod construct.

**Laminectomy and Foraminotomy.** Meticulous dissection of the lamina is required to minimize the risk of dural injury. Because the spinal canal is significantly narrowed, the first step is to drill the lamina down to expose the yellow ligament. The yellow ligament is preserved because this offers some protection to the dura from the implanted hardware. The thinned lamina is then removed piecemeal using Kerrison rongeurs and elevated off the dura. In younger individuals the entire lamina may be removed in one piece (Fig. 7).

Older individuals, however, have coexistent degenerative disease affecting the facet joints and spinal ligaments with adherence to the dura, which does not permit this maneuver. All bone removed during the laminectomy is set aside to be used as the substrate for the bone fusion as an onlay graft. Bilateral foraminotomies are an integral part of a satisfactory decompression in achondroplasia (Fig. 8).

**Pedicle Screw Insertion.** Landmarks used when placing pedicle screws are identical to those in nonachondroplastic spines. Care must be taken, however, to palpate the pedicle by using a ball-tipped probe as the intended path of the screw is tapped. Because of the dysmorphic nature of the vertebrae and the presence of the kyphosis, the trajectory of the pedicle changes both superior and inferior to the apex of the curvature. One must therefore adjust the trajectory in the superoinferior direction to reduce the chance of breaching the pedicle cortex. The entry point is chosen at the midlevel of the vertebral transverse process into the site of the superior articular facet of the vertebra below. Care must be taken at the superior end of the planned construct so as to prevent interference with the facet joint not included in the arthrodesis.

**DISCUSSION**

The presence of thoracolumbar kyphosis in childhood is common.9,13,14 This is not a fixed deformity and is best treated by brace therapy. Fixed angular thoracolumbar curvative is a characteristic of the spinal deformity that develops in achondroplastic individuals over time. This is coexistent with spinal stenosis, which becomes symptomatic in adolescents and adults and must be addressed concurrently. The presence of compensatory lumbar hyperlordosis exacerbates the situation.9

Immediate benefits in children such as reduction in the flexible kyphosis and long-term structural improvements in the vertebrae have been recognized.1,14,18 Remodeling of the spine due to uncorrected kyphosis may result in a progressive deformity. Knowledge of this concept of vertebral remodeling is pivotal to successful management. The musculoskeletal hypotonia in these children is overcome by avoiding unsupported seating and by instituting brace therapy.14 The TLSO is recommended if greater than 30° of thoracolumbar kyphosis, anterior vertebral wedging, or spondylolisthesis is demonstrated on the follow-up images. Using these guidelines, bracing effectively corrects the kyphotic angle to a residuum of 8° with repair of the wedged vertebrae if initiated before the patient is 3 years of age.14 Symptomatic improvement in thoracolumbar pain has also been documented when using brace therapy in adult patients with lumbar pain.18

The presence of a symptomatic fixed thoracolumbar kyphotic deformity warrants operative correction. When greater than 30° of kyphosis persists in a child beyond 5 years of age, despite aggressive brace therapy, further
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Fig. 9. Intraoperative photograph of probes placed in the pedicles prior to intraoperative radiography to confirm satisfactory position and trajectory of the planned screw insertion.

treatment may warrant posterior in situ arthrodesis. Laminctomy to decompress the lumbar stenosis can exacerbate the progressive deformity at the thoracolumbar junction. Serious consideration must be given to the prophylactic in situ instrumentation-assisted fusion of the spine if certain features are present; these include anterior vertebral wedging or recognized intraoperative disruption of facet joints during decompression. Authors differ in their recommendations regarding the optimal procedure in these patients. It is accepted that the entire spine is dysplastic. In association with the thoracolumbar kyphosis, the spinal canal is narrowed. Foraminal narrowing is a particularly prominent feature of the symptomatic achondroplastic spine because degenerative changes of the facet joints further compromise the already congenitally narrow spinal canal. Foraminal stenosis is believed by some to be clinically more relevant than the central canal narrowing that is present.19

When treating the achondroplastic patient in whom there are a symptomatic fixed angular thoracolumbar kyphosis and neurological symptoms, both the neurological compromise and the skeletal deformity must be treated. Failure to address the latter facet adequately is a pitfall one should avoid.

In cases of thoracolumbar kyphosis of 30°, a posterior decompressive laminectomy, foraminotomies, and in situ instrumentation-assisted fusion may be performed. A kyphotic angle of greater than 30° or compromise of the spinal cord anteriorly at the level of the deformity requires partial anterior vertebrectomy/instrumentation/fusion supplemented with posterior instrumentation-assisted fusion.11,17,20 A TLSO reduces postoperative discomfort, because these patients are encouraged to ambulate by postoperative Day 2. This orthosis is generally worn for 6 to 8 weeks as required by the patients. After the first postoperative visit, clinical evaluation is undertaken at 3, 6, 12, and 24 months. Standing AP and lateral x-ray films are obtained at these times to assess the success of fusion and construct positioning. Long-term follow up thereafter is recommended, because the recrudescence of musculoskeletal and/or neurological symptoms may signal the presence of significant disease in additional levels of the patients spine.

CONCLUSIONS

Thoracolumbar kyphosis is a common finding in achondroplasia. In the early stages of this process, which is observed in infancy, this is a nonfixed deformity. If there is progression of the kyphosis or anterior vertebral wedging in these children, it is best managed with a tailor-made TLSO. In a proportion of cases a fixed angular thoracolumbar kyphosis may develop. Such a fixed deformity is associated with a natural history characterized by neurological and musculoskeletal symptoms. Operative correction of the deformity and instrumentation-assisted fusion are required.

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