Dynamic cervicomedullary cord compression and alterations in cerebrospinal fluid dynamics in children with achondroplasia

Report of four cases

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Achondroplasia is the most common of the heritable skeletal dysplasias. Compression at the cervicomedullary junction can result in myelopathy, hypotonia, sleep apnea, and even sudden death. However, most children with achondroplasia do not suffer from severe neurological symptoms and achieve normal motor and intellectual development without surgical intervention. At the authors’ institution, magnetic resonance (MR) imaging and cerebrospinal fluid (CSF) flow studies have been incorporated in the assessment of children with achondroplasia for cervicomedullary junction compression. The authors recently identified four children with achondroplasia who had normal findings on MR imaging and flow studies obtained in the neutral position. On flexion studies, however, three had complete blockage of CSF flow, and more dramatic posterior cervicomedullary compression was demonstrated on extension studies. Some of these patients had severe neurological abnormalities and sleep apnea, while others just developed headaches and/or had apnea episodes when sleeping or in a car seat. Three children underwent decompressive surgery with dramatic improvement or resolution of signs and symptoms. The fourth patient had increased CSF pressure on MR images obtained in the flexed position, possibly due to venous outflow obstruction. Her condition improved dramatically after placement of a ventriculoperitoneal shunt. The increased risk of dynamic cord compression and alterations in CSF dynamics in patients with achondroplasia constitute indications for surgical intervention.

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KEY WORDS • achondroplasia • decompression • dynamic cervicomedullary compression • foramen magnum stenosis • hydrocephalus • pediatric neurosurgery

Abbreviations used in this paper: CCJ = craniovascular junction; CSF = cerebrospinal fluid; CT = computed tomography; ICP = intracranial pressure; MR = magnetic resonance; VP = ventriculoperitoneal.

Deformed foramen magnum.5,12 Cord compression can result in cervical myelopathy manifested as clonus and hyperreflexia, hypotonia, sleep apnea, and sudden death. Due to the potentially lethal nature of cervicomedullary compression, neurosurgical suboccipital decompression has frequently been used to open the foramen magnum and relieve the pressure on the emerging cervical cord.2,9 However, there has been controversy over the degree of risk for sudden death in children with achondroplasia, how often to operate, the indications for decompression, and the surgical methods that should be used.13

Published mortality rate estimates in infants with achondroplasia have ranged from 2.7 to 7.5%.3,7 Over the past 35 years, however, we have seen hundreds of children with achondroplasia, only one of whom died in infancy (< 0.5% mortality rate) and one in early childhood. The rate of cervicomedullary decompression in children with achondroplasia has been reported to be from 4.5 to 42.2%.1,2,6,9,10 The
rate of cervicomedullary decompression reported in an international collaborative study was 6.8% in patients 4 years of age and 16.5% at the conclusion of the study. Criteria for decompression in a recent large surgical series included fulfillment of two of the three following criteria: 1) severe lower limb hyperreflexia or clonus on examination; 2) central apnea or hypopnea on polysomnography; and 3) foramen magnum measurements that fall significantly below the mean for children with achondroplasia with evidence of cervical spine compression or diminished CSF flow on MR imaging. Unfortunately, this study alone does not provide a fail-safe method for prospectively identifying patients who are likely to die or experience severe neurological complications if decompressive surgery is not performed, and this may result in surgeries undertaken in individuals who would have spontaneously gained normal neurological function with time.

At our center, we have followed up infants with achondroplasia using MR imaging that incorporates CSF flow studies. Most infants with achondroplasia have a decreased amount of space around the emerging cervical cord, and frequently the lip of the foramen magnum indents the posterior spinal cord. As these children grow older and gain normal motor development, the impingement on the cord decreases and may or may not eventually disappear, presumably because the size of the foramen is growing faster than the size of the cord. The narrowing of the cord itself, however, may persist or not become entirely normal despite the patient having gained normal neurological function. Indeed, infants with achondroplasia who have undergone surgical decompression and have gained normal neurological function may have a persistence of cord narrowing.

The existing literature has focused on the surgical treatment of static myelopathy attributed to fixed cervicomedullary compression due to foramen magnum stenosis. The present study calls attention to a related but previously unrecognized dynamic phenomenon. We identified four infants and young children with achondroplasia who developed symptoms of headache and/or nausea and vomiting or severe central sleep apnea after sitting in a car seat or sleeping. These children had adequate CSF flow at the foramen magnum when the images were taken at a neutral position, but CSF flow was dramatically diminished or completely blocked when the neck was flexed in three cases. Sagittal MR images demonstrated increased compression of the cervicomedullary spinal cord with the neck in the flexion position; the symptoms resolved after foramen magnum decompression. In a fourth patient we documented elevated ICP on neck flexion studies that resolved along with the symptoms after placement of a VP shunt. Thus, there is an increased risk of dynamic cord compression and alterations in CSF flow dynamics in patients with achondroplasia that also constitutes an indication for surgery.

Case Reports

Case 1

This 5-year-old boy had a history of obstructive sleep apnea and had previously undergone tonsillectomy. He developed increasing headaches described as a 10 on a pain scale of 1 to 10, associated with nausea and vomiting that would arise about 1 to 2 hours after he went to sleep. The headaches would frequently wake him up, leaving him screaming and crying out in pain. The symptoms improved with sitting upright, but could last for several hours. On questioning, the parents recalled that the child frequently suffered the same symptoms while strapped in a car seat. The child denied any lower extremity pain or sensory changes.

Polysomnography demonstrated a mixed picture of severe central and obstructive sleep apnea. A prior cine MR image of the cervical spine had demonstrated adequate flow of CSF anterior to the spinal cord in the region of the foramen magnum. Repeated cine MR imaging of the cervical spine with the child’s neck in a flexed position demonstrated a dramatic reduction in CSF flow anterior and posterior to the spinal cord compared with the same studies in the neutral or extended positions. The cervical canal was significantly narrowed in flexion (Fig. 1 right) compared with extension (Fig. 1 left) and neural studies. There was no evidence of cervical instability at the cervicomedullary junction. The child underwent decompression of the foramen magnum and posterior ring of first cervical vertebra, and duroplasty without placement of a patch graft. The ligamentum flavum was sharply dissected and removed. The excised outer leaf of the dura mater was then scored with a No. 15 blade knife in multiple longitudinal lines from the foramen magnum to the lamina of C-2. Immediately after surgery, the child demonstrated both subjective and objective improvement in his symptoms. He no longer complained of any headaches nor did he have any episodes of sleep apnea or desaturation during the remainder of his hospital stay. A follow-up sleep study conducted 3 months after surgery demonstrated a dramatic decrease in the number and severity of desaturations during sleep. The follow-up MR images demonstrated excellent CSF flow at the foramen magnum in both flexion and extension positions. The child’s parents continue to report significant benefit 19 months after surgery, and there have been no episodes of significant desaturation, apnea, or nighttime headaches.

Case 2

This 5-year-old girl complained of increasing frequency and severity of headaches which had frequently awakened her from sleep, and lead on two separate occasions to evaluation at the local emergency department. The headaches were described as “very bad” and were aggravated by awakening from sleep or lying flat with her neck in flexion. A pain scale rating was not documented. The CT scanning of the brain did not reveal any increase in the size of her ven-

![Fig. 1. Case 1. Preoperative sagittal MR images with the cervical spine in extension (left) and flexion (right).](image-url)
tricular system compared with prior studies. Her neurological examination was significant for decreased muscle tone in her extremities. Sleep studies demonstrated severe mixed sleep apnea with a predominant central component. Cervical spine flexion/extension cine flow studies demonstrated minimal flow anterior to the spinal cord when in the neutral position, with near complete cessation of flow with the neck in the flexed position. The cervical canal was significantly narrowed in flexion (Fig. 2 right) compared with extension (Fig. 2 left) and neural studies. Once again there was no evidence of cervical instability on these studies. The child underwent a foramen magnum and first cervical vertebral bone decompression and duroplasty. Postoperatively, the child had immediate and complete cessation of headaches and noisy breathing during sleep. A postoperative sleep study demonstrated moderate improvement of preoperative obstructive and central sleep apnea. A repeated cine MR imaging study showed re-establishment of CSF flow anterior and posterior to the spinal cord both in flexion and extension. The child continues to do well 11 months postoperatively, without any severe headaches, nausea, or vomiting.

Case 3

This 5-month-old boy presented with an apparent life threatening apneic event. The child had received a diagnosis of achondroplasia shortly after birth and had undergone a CT scan which did not demonstrate any significant foramen magnum stenosis. The child began to gasp for air for approximately 10 to 15 seconds, became blue, and shortly thereafter turned red. Cardiopulmonary resuscitation was not administered. The child was brought to the emergency department and admitted to the pediatric service. Cardiac and apnea monitoring documented numerous episodes of apnea. A cervical spine flexion/extension cine MR imaging study demonstrated complete obstruction of CSF flow anterior to the spinal cord when the child was in the flexed position, and adequate flow in neutral and extended positions. The cervical canal was significantly narrowed in flexion (Fig. 3 right) compared with its appearance on extension (Fig. 3 left) and neural studies. Because we do not have polysomnography available for inpatients at our institution, we were unable to document episodes of central and obstructive sleep apnea preoperatively. The boy underwent a foramen magnum decompression and C-1 laminectomy with duroplasty in the same fashion as our first patient. The child showed immediate postoperative cessation of desaturations and apnea. Polysomnography conducted within 1 month postoperatively demonstrated moderate mixed sleep apnea. A follow-up polysomnography study obtained 12 months postoperatively demonstrated mild obstructive apnea without any episodes of central sleep apnea.

Case 4

This 6-year-old girl presented with urinary incontinence, occasional leg pain, headaches, nausea, and vomiting. Her headaches would typically start when she was coloring at school or when she played videogames with her neck in a flexed position. Flexion/extension cine flow MR imaging did not demonstrate obstruction of CSF flow at the skull base. Her symptoms improved somewhat with acetazolamide (Diamox) treatment. Intracranial pressure monitoring confirmed elevated pressures of up to 50 mm Hg when her neck was in a flexed position; the ICP would return to baseline within 10 minutes of turning her head back to a neutral position. Given the results of the CSF flow studies, the elevation in ICP was believed to be secondary to kinking of central venous drainage at the skull base when the cervical spine was in the flexed position. A VP shunt was placed, and the patient experienced resolution of her symptoms within days of its placement.

Discussion

There continues to be some disagreement with regards to the indications for surgical intervention for a narrowed foramen magnum in achondroplasia. Most specialists agree that surgical decompression can be effective and relatively safe in expert hands, although there can be complications. At our institution, patients are evaluated and followed up by a multidisciplinary team of neurosurgeons, neurologists, geneticists, pediatric neurosurgeons, and pediatric orthopedic surgeons at the skeletal dysplasia clinic. At our center we have incorporated MR imaging with CSF flow studies in assessing children with cervicomedullary junction compression. The four cases we report on here illustrate that children with achondroplasia are also at risk for dynamic obstruction of CSF flow at the CCJ.
Cervicomedullary cord compression and achondroplasia

Although progressive hydrocephalus in children with achondroplasia is probably the primary result of venous stenosis at the skull base, there may also be a transient outlet obstruction of CSF flow at the foramina of Luschka and Magendie which may be either static or dynamic. Based on our findings in three children with recurrent and reproducible signs and symptoms who have had dramatic improvement after suboccipital craniectomy and C-1 laminectomy, one can argue that any child with symptomatic achondroplasia should be evaluated with dynamic cervical spine MR CSF flow studies in both the flexion and extension positions; children who are found to have obstruction of CSF flow at the skull base may benefit from foramen magnum decompression prior to committing them to a CSF diversion procedure. This is not to say that all children with achondroplasia who are symptomatic with headaches with their neck in the flexed position suffer from dynamic obstruction of CSF flow at the cervicomедullary junction. The fourth patient in our series was not found to have CSF outflow obstruction at the skull base, but rather thought to be symptomatic due to venous outflow obstruction when holding her neck in a flexed position. Decompression of the foramen magnum in this child would most likely have no positive impact on her symptoms.

The origin of dynamic CSF flow obstruction at the skull base in children with achondroplasia is unclear. None of the children thus far have been found to have blatant cervical instability at the CCJ. Some of these children appear to have a small posterior facing odontoid process on the MR image but we have not yet confirmed this finding on CT.

In conclusion, we propose that there is an increased risk for dynamic cord compression and obstruction of CSF flow at the skull base in children with achondroplasia that may constitute an indication for surgical decompression at the cervicomедullary junction in symptomatic children. Conversely, children with achondroplasia who present with symptoms of hydrocephalus, without any demonstrated obstruction of CSF flow at the skull base may be better served by placement of a VP shunt than by undergoing a foramen magnum decompression. Although the algorithm for evaluation of children newly diagnosed with achondroplasia proposed by the American Academy of Pediatrics recommends initial MR imaging of the foramen magnum, we have not routinely obtained images in asymptomatic children because they would have to be anesthetized for the procedure. We are prospectively evaluating several newly diagnosed patients with achondroplasia using flexion/extension MR imaging with CSF flow studies to determine the natural history of dynamic compression in these children.

We do not yet know if children prospectively ascertained with dynamic alterations of CSF flow at the skull base will become symptomatic, requiring a foramen magnum decompression. Such children, however, should be carefully followed up clinically. In the case of clinical symptoms, operative intervention is advisable; all the children whom we took to the operating room were symptomatic and experienced dramatic and long-lasting improvement following surgery.

References


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