

Acute neurological deficit after minor trauma in an infant with achondroplasia and cervicomedullary compression

Case report and review of the literature

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✓Cervicomedullary compression at the foramen magnum in patients with achondroplasia can be associated with apnea, neurological deficits, and sudden death. Decompressive operations are often performed in symptomatic patients. In asymptomatic patients, the indications for prophylactic decompression are controversial. The authors present the case of a previously neurologically intact 4-month-old girl with achondroplasia who presented with severe hemiparesis after a low-velocity motor vehicle accident. Imaging studies demonstrated osseous compression of the medulla and upper cervical spinal cord with associated parenchymal signal changes. To the authors' knowledge this is the first reported case of a new neurological deficit after a minor trauma in this patient population. The authors review the relevant literature, focusing on the indications for cervicomedullary decompression in infants with achondroplasia. They propose that asymptomatic patients with achondroplasia and osseous compression at the foramen magnum should be offered prophylactic surgery if T2-weighted magnetic resonance imaging signal changes in the spinal cord are observed. Prophylactic surgery can be considered an option in patients whose imaging studies do not show signal changes in the spinal cord but demonstrate significant osseous compression and absence of visible subarachnoid spaces.
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KEY WORDS • achondroplasia • cervicomedullary compression • pediatric neurosurgery

ACHONDROPLASIA is an autosomal-dominant disorder of skeletal maturation resulting from a gene mutation in the fibroblast growth factor receptor located on chromosome 4.⁹ The gene mutation results in abnormal endochondral bone formation,¹⁷ and its incidence ranges from 0.5 to 1.5 in 10,000 live births.¹¹

Abnormal bone formation in patients with achondroplasia can result in foramen magnum stenosis along with stenosis involving other parts of the neuraxis.⁶ Stenosis at the foramen magnum, when symptomatic, can cause apnea, neurological deficits, and sudden death in patients with achondroplasia.^{2,12,13} Mortality rates can approach 7.5% in the 1st year of life and 2.5% in patients between 1 and 4 years of age.⁵

The rate of documented cervicomedullary compression from foramen magnum stenosis in patients with achondroplasia is variable in previous studies. Of 186 patients with achondroplasia evaluated at the University of Iowa Hospitals and Clinics over a 13-year period, Ryken and Menezes¹⁶ found six that required surgery for symptoms of cervico-

medullary compression. In another retrospective study, performed at The Johns Hopkins Hospital, of the 58 patients with achondroplasia whom they screened, Aryanpur et al.¹ performed surgery in 15 symptomatic patients with cervicomedullary compression. In a prospective study of infants with achondroplasia, Reid et al.¹⁴ found imaging evidence of cervicomedullary compression in 35% of those studied and found smaller than average foramen magnum diameters in all.

The authors of these previous studies described patients who underwent surgery for symptomatic compression at the foramen magnum and/or severe imaging-documented compression. To our knowledge there are no reports on patients who presented with new neurological deficits after minor trauma. In the current report we present the case of a patient who suffered a new neurological deficit after a minor trauma and recovered after decompressive surgery. We also review the relevant literature regarding indications for decompressive surgery.

Case Report

History and Examination. This 4-month-old girl with

Abbreviations used in this paper: CCJ = craniocervical junction; CT = computed tomography; MR = magnetic resonance.

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achondroplasia was involved in a low-velocity minor motor vehicle accident. Her mother reported that her car, while stationary, was hit from behind by another vehicle that was driving approximately 5 mph. The child was cleared by paramedics at the scene and did not present directly to a medical facility.

Later that evening, the patient's parents noticed that she was moving her left side less than her right side. In the emergency room, the patient had no movement of the left upper extremity and trace movement of the left lower extremity to deep stimulation. Cranial, spinal, and brachial plexus imaging studies were performed. A CT scan of the CCJ demonstrated osseous overgrowth of the posterior margin of the foramen magnum (Fig. 1). A CT scan of the cervical spine demonstrated no fracture or dislocation. Magnetic resonance imaging demonstrated marked signal abnormality in the upper cervical spinal cord and caudal medulla with no visible subarachnoid space posteriorly (Fig. 2). There was no evidence of swelling or signal abnormality in the prevertebral soft tissue. A CT scan of the brain and an MR image of the brachial plexus did not reveal remarkable findings.

Operation. We performed a suboccipital craniectomy and C-1 laminectomy. Intraoperatively, we noted obvious indentation of the dura mater at the foramen magnum posteriorly due to osseous and fibrotic tissue. Bone and epidural tissue were removed to decompress the CCJ. The dura mater was not opened. A postoperative MR image demonstrated increased subarachnoid space at the CCJ both anteriorly and posteriorly (Fig. 3).

Postoperative Course. The patient was kept in a hard cervical collar for 3 months postoperatively. She exhibited remarkable improvement in neurological status throughout her postoperative course. A few days after surgery, she was



FIG. 1. Sagittal bone window CT scan obtained at presentation following the accident, demonstrating osseous overgrowth of the posterior foramen magnum.



FIG. 2. Sagittal T2-weighted MR image obtained at presentation after the accident, demonstrating obliteration of subarachnoid spaces posteriorly and signal hyperintensity in the spinal cord.

moving her leg against gravity. Her arm moved minimally when stimulated but remained significantly weak. One month postoperatively, the patient had recovered full function of her left lower extremity and had Grade 4+/5 strength in her left upper extremity. Three months postoperatively, she had made a full recovery with normal (Grade 5/5) strength bilaterally in her upper and lower extremities.

Discussion

In this report, we have presented the first published case of a new neurological deficit in an infant with achondroplasia immediately following minor trauma attributed to foramen magnum stenosis. Although all infants have a certain degree of ligamentous laxity in the cervical spine, infants with achondroplasia are at a particular disadvantage. They tend to have relative hypotonia and a disproportionately large cranial vault. These factors contribute to poor head control and increased susceptibility to injury in the setting of cervicomedullary stenosis.¹³

The more common presentation of foramen magnum stenosis in these patients is apnea or sudden death.^{2,13} Hecht et al.⁵ studied a cohort of 733 individuals with achondroplasia. They found an increased incidence of sudden death in children younger than 4 years of age with this condition compared with patients without achondroplasia. These authors maintained that brainstem compression was the likely cause of many of these deaths.



FIG. 3. Sagittal T2-weighted MR image obtained on postoperative Day 2, demonstrating increased posterior subarachnoid spaces after decompression.

Previous authors have recommended a multidisciplinary prospective approach to treatment of patients with achondroplasia.^{4,14} Of 90 patients referred for evaluation to clinics at The Johns Hopkins Hospital, 38 underwent foramen magnum decompression after clinical screening conducted by an orthopedic surgeon, pulmonologist, geneticist, and neurosurgeon. The patients who underwent surgery presented with signs of myelopathy or central sleep apnea. Preoperative evaluation included imaging studies of the brain and cervical spine, somatosensory evoked potential monitoring, polysomnography, and echocardiography.⁴

Another retrospective review of 43 patients with cervicomedullary compression from the same institution was recently reported. All but one of these were symptomatic.² The single asymptomatic patient had severe compression observed on images, which the authors believed warranted a prophylactic surgery. The authors reported improvement or resolution of symptoms in all symptomatic patients. Five patients required a second operation due to restenosis and worsening of symptoms. There were no deaths in that series, and the most common complication was cerebrospinal fluid leakage that decreased when the authors stopped performing duraplasties in these cases.

A prospective study of 11 young infants with achondro-

plasia who had imaging-documented evidence of cervicomedullary compression but were asymptomatic at presentation was performed by Keiper et al.¹⁰ Two patients underwent prophylactic craniocervical decompression because of signs of severe compression on images including effacement of subarachnoid spaces and/or abnormal spinal cord signal change on T2-weighted MR images. Two became symptomatic with apnea and opisthotonic posturing within 3 months of the initial evaluation, and both underwent a foramen magnum decompression. The remaining seven of these patients were followed conservatively. Two of the seven had mild posterior spinal cord compression on MR images, and all had small foramen magnum diameters. The nonoperative group remained asymptomatic after approximately 5 years of follow-up.¹⁰

Quantification by CT scan of the degree of foramen magnum stenosis was reported by Hecht et al.^{6,7} These authors showed that the foramen magnum of individuals with achondroplasia was significantly smaller than that in healthy individuals at all ages. The diameter of the foramen magnum in patients with neurological dysfunction was smaller than that in asymptomatic patients.

Other authors have developed criteria to describe the degree of cervicomedullary compression based on a comparison of the diameter of the foramen magnum with the diameter of the pontomedullary junction and the spinal canal at C-3. These ratios can be considered along with clinical symptoms and age of the patients in the decision of whether to offer foramen magnum decompression. Operations for severe cervicomedullary compression, according to the ratios of the dimensions described earlier, are indicated in asymptomatic patients younger than 4 years of age due to the high risk of sudden death.²¹ On the other hand, Wassman and Rimoin^{15,19} have proposed that because the foramen magnum enlarges with age, the risks associated with prophylactic decompression in asymptomatic patients may outweigh those associated with conservative treatment. Thus, although decompressive surgery has been shown to clearly benefit symptomatic patients, controversy surrounds the topic of whether patients with achondroplasia and imaging-documented foramen magnum stenosis should be offered prophylactic operations.^{2,3,8,16,20}

We add the present case as another factor to include in the decision of whether to perform foramen magnum decompression on an asymptomatic patient with achondroplasia and foramen magnum stenosis. We agree with the recommendations of previous authors that all newborns with achondroplasia should undergo CT scanning or MR imaging of the CCJ.^{10,18} We hypothesize that the patient we have described would have been at much higher risk for sudden death in a higher speed accident than a child without achondroplasia. We believe that a prophylactic decompressive operation should be strongly considered even in asymptomatic patients in the presence of signal changes in the spinal cord or medulla. The risks of prophylactic surgery in these cases are likely smaller than the risks of progressive apnea, sudden death, or neurological injury following trauma if surgery is not performed. Further study is needed in this patient population to determine if asymptomatic patients with significant osseous compression and absence of visible subarachnoid spaces without signal changes in the spinal cord face greater risks from prophylactic decompression or from the condition's natural history. Until such studies are performed, we

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propose that prophylactic decompression can be considered as an option in such patients.

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