Achondroplasia is the most common of the skeletal dysplasias. It is an autosomal dominant condition, with most cases due to new activating mutations in the FGFR3 gene. The molecular defect in achondroplasia causes a quantitative decrease in the rate of endochondral bone formation, resulting in short and wide long bones, short stature, a large calvaria, midface hypoplasia, and a short basicranium with narrowed foramen magnum and vascular channels (Fig. 1). Compression at the foramen magnum can result in cervical myelopathy manifested as clonus and hyperreflexia, hypotonia, sleep apnea, and even sudden death. Due to the potentially lethal complications associated with symptomatic disease, neurosurgical decompression has been used to widen the foramen magnum and relieve the pressure on the emerging cervical cord. Fortunately, most children with achondroplasia do not develop neurologic symptoms until adulthood or late childhood. Patients with achondroplasia is cervicomedullary junction (CMJ) compression caused by a tight deformed foramen magnum. The most serious neurological complication in patients with achondroplasia is cervicomedullary junction (CMJ) compression caused by a tight deformed foramen magnum.
Dynamic cervicomedullary cord compression in achondroplasia

not suffer neurological symptoms and achieve normal motor and intellectual development without surgical intervention.8,12,19

Criteria for decompression have been previously described in large surgical series.17,20 Unfortunately, there has never been a study that provided a fail-safe method for prospectively identifying patients who are likely to die or experience severe neurological complications if decompression surgery was not performed. This left clinicians with the possibility of performing operations on individuals who would have spontaneously gained normal neurological function with time.

Past surgical series have focused on the surgical treatment of static myelopathy attributed to fixed cervicomedullary compression at the level of the foramen magnum.2,3,10,19,20 We previously identified 4 infants and children with achondroplasia who developed symptoms of cervicomedullary compression with their necks in flexed position. In 3 of these children, we demonstrated significantly diminished or blocked CSF flow anterior to the spinal cord with the neck in flexed position.6 It remained unclear 1) what percentage of symptomatic patients have a normal MRI scan in neutral position but have significant cervicomedullary compression and obstruction of flow on dynamic studies, 2) whether patients undergoing a bony foramen magnum decompression without placement of a patch graft have an adequate decompression which establishes CSF flow, and, finally, 3) whether there is a group of patients who are minimally symptomatic but have a higher risk of progression that can be predicted based on dynamic studies. We attempted to gain some insight into these questions with an 11-year retrospective review of a single-institution, single-surgeon surgical series of 35 symptomatic patients with achondroplasia, a large proportion whom were evaluated for cervicomedullary compression using dynamic cervical flexion/extension MRI and CSF flow studies, and underwent cervicomedullary decompression.

Methods

All patients with achondroplasia and symptomatic foramen magnum stenosis who were referred for neurosurgical evaluation between the years 2000 and 2010 at Cedars-Sinai Medical Center and subsequently underwent cervicomedullary decompression were identified. Presenting signs/symptoms including sleep apnea, nausea, vomiting, incontinence, headaches, full fontanel, clonus, excessive sweating, dilated facial veins, hyperreflexia, abnormal tone, gait disturbance, paresthesias of the extremities, weakness, distrextreflexia, or other signs of myelopathy were documented. Additionally, head circumference was measured on all presenting patients and documented on an achondroplasia head circumference growth chart. Criteria for symptomatic patients with cervicomedullary compression to undergo decompression were based on previously published clinical and radiographic measures.6

All patients underwent preoperative and postoperative polysomnography studies as well as MRI of the brain and cervical spine. On brain imaging, a ventricular ratio was calculated for all patients to assist in cataloging the extent of ventriculomegaly Additionally, 29 of the 34 patients underwent preoperative dynamic imaging studies (T1-weighted, T2-weighted, cine MRI) of the cervical spine in neutral, flexion, and extension positions. Thirteen of these 29 patients had postoperative dynamic MRI studies performed between 1 and 3 months following decompression. Primary measurements in these positions included the narrowest portion of the CMJ, the spinal cord diameter at the CMJ, C1–2 anterior ligamentous complex thickness, and the diameter of the foramen magnum (Figs. 2–4).

Head circumference centiles were determined using a standardized head circumference chart for patients with achondroplasia. Additionally, the Evans ratio, as defined in the axial plane as the ratio of the diameter of the largest bifrontal measure of the ventricles over the corresponding bifrontal measure of the intracranial cavity, was documented and used as a way to compare ventricles over time within the same patient. Although an Evans ratio of at least 0.3 is consistent with a diagnosis of hydrocephalus in patients of average height, this ratio has not been directly validated in the population of patients with achondroplasia and was not used in our patients to assess for the presence or absence of hydrocephalus (Fig. 5).

Postoperative primary outcome measurements included clinical results of postoperative polysomnography

Fig. 1. Activating mutations in FGFR3 within chondrocytes can lead to spinal stenosis, central sleep apnea, or hydrocephalus by decreasing the relative rate of endochondral bone formation.
studies, changes in presenting signs and/or symptoms, and radiographic outcomes, such as change in ventricular size, presence or absence of CSF flow, and the diameter of the spinal cord in neutral, flexion, and extension.

The Pearson chi-square test was used to assess bivariate outcomes between radiographic changes in spinal cord diameter and clinical outcomes such as a full fontanel.

**Surgical Technique**

All patients diagnosed with symptomatic cervico-medullary compression and foramen magnum stenosis underwent foramen magnum decompression and removal of the posterior ring of C-1. Suboccipital craniectomy for decompression of the foramen magnum has been previously reported.1,4 We used a modified procedure whereby the dura was not opened surgically. Following induction of general anesthesia and placement of appropriate lines, the patients were positioned prone on the operating room table with the head and neck supported in a pediatric horseshoe in slight flexion. Upper-extremity and lower-extremity somatosensory evoked potentials were routinely assessed before and after positioning as well as throughout the surgical procedure. A midline suboccipital incision was marked and indurated with local anesthetic followed by an incision from the inion down to the spinous process of C-2. Incision was extended in the midline plane down to the suboccipital area and lamina of C-1. A subperiosteal dissection was

![Fig. 2. Primary measurements made using sagittal MR images of the cervical spine in neutral (A and B) and flexed (C) positions included the narrowest portion of the cervical spinal canal (green line), the spinal cord diameter at the cervicomedullary junction (red line), and thickness of the anterior ligamentous complex (blue line). These measurements regularly changed in the flexed versus neutral positions.](image)

![Fig. 3. Dynamic T2-weighted (A and C) and cine (B and D) sagittal MR images of the cervical spine demonstrating cervical stenosis greater in flexion (A and B) than in extension (C and D).](image)

![Fig. 4. Preoperative (A and B) and postoperative (C and D) T2-weighted (A and C) and cine (B and D) sagittal MR images obtained in a single patient in the neutral position illustrating decompression (C) and improved flow (D) postoperatively.](image)
performed to expose the occiput and the posterior ring of C-1. The operating microscope was then brought into the field, and a high-speed drill and curettes were used to remove the posterior ring of C-1. To ensure an adequate lateral decompression, care was taken to extend the craniectomy out laterally and anteriorly to the point where the dura of the cervicomedullary junction would curve anteriorly. As has previously been described, the posterior rim of the foramen magnum was thickened and in many cases was found to curve in the cephalad direction. To minimize the risk of a dural tear, we first would thin out the suboccipital bone to a thin shelf posterior to the rim of the foramen magnum while leaving the lip of foramen magnum intact. We would then continue to use a high-speed drill, microcurettes, and 1-mm Kerrison rongeurs to resect the remaining posterior ring of the foramen magnum. An abnormally thick fibrous ligamentous band was frequently encountered; it was incised without opening the dura itself. The outer leaf of the dura was then scored with a No. 15 scalp as needed to allow for better expansion of the dura. Ultrasound was frequently used to confirm adequate decompression in addition to observing improved dural pulsations. The wound was irrigated with antibiotic solution and then closed in multilayer anatomical fashion. Most of the patients were extubated at the end of the operation and sent to the pediatric intensive care unit. Extubation was delayed in patients with either a history of severe obstructive sleep apnea or the presence of facial and laryngeal edema.

Results

Overall Cohort

A total of 34 patients were identified who presented with symptomatic foramen magnum stenosis and underwent cervicomedullary decompression (Table 1) based on previously described criteria. Their mean age at presentation was 6.6 years old, and there was a slight female predominance (23 female patients). The mean duration of follow-up was 3.7 years (range 1–10 years). Among all patients, spinal cord diameter increased by 5.2 mm on postoperative MRI of the cervical spine in neutral position.

Subset With Flexion-Extension Imaging

A total of 29 of the 34 patients had preoperative flexion-extension dynamic MRI cine flow studies, with 13 of these patients having postoperative follow-up dynamic MRI cine flow studies as well. All 29 patients were found to have central sleep apnea at presentation, while 5 patients were hydrocephalic, 5 had a history of nausea/emesis, 4 were incontinent, and 2 presented with headaches. Additionally, 18 presented with a full fontanel, 17 with abnormal tone, 12 with clonus, 10 with excessive sweating, 11 with dilated facial veins, 6 with paresis, 6 with hyperreflexia, 3 with gait disturbance, and 2 with paresthesias in the extremities.

Preoperatively, the average change in spinal cord diameter from extension to flexion was \(-1.9\) mm (range \(-0.7\) to \(-3.9\) mm). Preoperatively, the average change in spinal cord diameter from neutral to flexion was \(-1.7\) mm (range \(-0.2\) to \(-2.9\) mm). Postoperatively, the average change in spinal cord diameter from extension to flexion was \(-4.2\) mm (range \(-1.7\) to \(-5.2\) mm). Postoperatively, the average change in spinal cord diameter from neutral to flexion was

![Fig. 5. The Evans ratio is defined as the maximum ventricular width (red line) divided by the largest biparietal distance between the inner tables of the skull (green line). An Evans ratio of at least 0.3 may be consistent with a diagnosis of hydrocephalus.](image)
the VP shunt procedure predated the cervicomedullary de-
centile postoperatively over time, despite several having
tients demonstrated any increase in head circumference
worsening in the postoperative period. None of these pa-
Eighteen patients presented with a full fontanel, with none
using the achondroplasia head circumference centile chart.
preoperative head circumference in the 46.9th centile to
cumference remained relatively unchanged from a mean
transfusions intraoperatively or perioperatively. Head cir-
cal complications without surgical decompression. Addition-
how often to operate, the indications for decompression,
of risk for sudden death in children with achondroplasia,
entially because the size of the foramen magnum is growing
cord decreases and may eventually disappear, presum-
function. We have used complete obliteration of CSF flow
posterior to the spinal cord as an indication for decompres-
s in symptomatic patients.

Previously at our center we identified 4 otherwise
healthy infants and children with achondroplasia who
developed symptoms of cervicomedullary compression
with their necks in flexed position. In 3 of these children,
we found evidence of adequate CSF flow at the foramen
magnum when the MR images were taken at a neutral po-
sion, but dramatically diminished or completely blocked
CSF flow when the neck was flexed. These 3 symptomatic
patients would not have undergone a decompression
procedure without the benefit of flexion-extension studies
demonstrating cervicomedullary compression and ob-
struction of CSF flow with the cervical spine in the flexed
position. All 3 of these patients had dramatic improve-
ment in central sleep apnea and headaches after under-
going a decompression procedure. We therefore decided
to review an 11-year single surgeon surgical experience
in our institution to determine the incidence of dynamic
cervicomedullary compression that is only demonstrable
with the cervical spine in the flexed position.

In the current review of our surgical series, we found
that 14% of our symptomatic patients who underwent a dy-
amic preoperative flexion and extension CSF flow study
would not have otherwise been candidates for decompres-
sion procedures. In other words, 14% of the patients who
underwent foramen magnum decompression met our cri-
teria for decompression based on obliteration of CSF flow
anterior to the CMJ on MRI studies of the cervical spine
in a flexed position. While the majority of patients under-
going a decompression procedure had significant stenosis
on neutral studies that was worse on flexion, these patients
did not have significant constriction of the spinal cord in
neutral position that would have led us to recommend a
cervicomedullary decompression.

Additionally, many patients with preoperative cen-
tral sleep apnea do not have complete resolution but do
have significant improvement of their sleep studies. In as-
sessing the adequacy of decompression procedure in this
subgroup of patients, we found postoperative dynamic
studies to be very helpful, as they were the most effec-
tive in demonstrating any significant residual or recurrent
compression.

One unexpected observation in our study regarded
the improvement in the fullness of the anterior fontanel
and the apparent lack of progressive ventriculomegaly or
jump in head circumference centiles in any of the patients
who underwent a foramen magnum decompression. Our
observation is very difficult to interpret as ventriculomeg-
aly is well documented in achondroplasia, but the clinical
significance of ventriculomegaly and defini-
tion of hydrocephalus can be elusive and controversial.

Discussion
The existing literature has focused on the surgical
treatment and outcomes of fixed cervicomedullary com-
pression due to foramen magnum stenosis. Unfortunately, there continues to be controversy over the degree of
risk for sudden death in children with achondroplasia,
how often to operate, the indications for decompression,
and surgical methods that should be used. Criteria for de-
compression in large surgical series7 have not provided a
fail-safe method for prospectively identifying patients
who are likely to die or experience severe neurological
complications without surgical decompression. Addition-
ally, there appears to be a group of patients who can be
symptomatic, with signs of elevated intracranial pressure
or compression of the cervicomedullary junction due to
dynamic phenomenon. Unfortunately, this may result
in surgeries undertaken in individuals who would have
spontaneously gained normal neurological function with
time or in needed surgery being withheld from patients
with normal imaging findings of the cervical spine in
neutral position.

At our center, we follow infants with achondropla-
sia using MRI that incorporates CSF flow studies with
the cervical spine in neutral, flexed, and extended posi-
tions. 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 eventually disappear, presum-
ably because the size of the foramen magnum is growing
faster than the size of the spinal cord. In fact, the chang-
es present in the spinal cord and the indentation or the
emerging cord may persist despite normal neurological
function. We use dynamic phenomenon.6 Unfortunately, this may result

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-3.2 mm (range -1.8 to -4.2 mm). Relative to preoperative
values, the average increase in spinal cord diameter post-
operatively was +3.1 mm in neutral position, +5.4 mm in
flexion position, and +0.2 mm in extension position. The
ventricular ratio remained relatively unchanged, from a
value of 0.37 preoperatively to 0.33 postoperatively. Post-
operative cine MRI studies demonstrated improved CSF
flow in all patients (13 of 13 patients).

Four of the 29 patients with flexion-extension imag-
ing had significant stenosis on flexion imaging without
obliteration of CSF flow anterior to the cord on neutral
images. This was equal to 13.7% of symptomatic achon-
droplastic patients in whom spinal cord compression with
obliteration of CSF flow anterior to the cord was only evi-
dent on flexion-extension imaging, and not with the neck
in neutral position.

Postoperative polysomnography demonstrated mod-
erate to complete resolution of central sleep apnea in all
patients. None of our patients required red blood cell
transfusions intraoperatively or perioperatively. Head cir-
cumference remained relatively unchanged from a mean
preoperative head circumference in the 46.9th centile to
a postoperative head circumference in the 45.7th centile,
using the achondroplasia head circumference centile chart.
Eighteen patients presented with a full fontanel, with none
worsening in the postoperative period. None of these pa-
tients demonstrated any increase in head circumference
centile postoperatively over time, despite several having
accelerated head circumference growth or a full fontanel
preoperatively. One patient in the series required a ven-
triculoperitoneal (VP) shunt for hydrocephalus, although
the VP shunt procedure predated the cervicomedullary de-
compression operation.

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In fact, standardized head circumference centile charts for children with achondroplasia were created without measurements of the size of the ventricles or measurements of intracranial pressures. Steinbok et al. pointed out that the enlarged head circumferences in patients with achondroplasia may in fact “reflect a significant incidence of underlying hydrocephalus in achondroplastic patients.” In their study they monitored intraventricular pressure followed by injection of radionucleotides in 5 children with achondroplasia. The intraventricular pressure was elevated and the reabsorption of CSF into the sagittal sinus was slow in all cases, but there was no demonstrable obstruction to CSF flow. Jugular venograms and pressure monitoring confirmed a narrowed jugular foramen in all patients, without any elevation of the venous pressures in a control group. The authors concluded that in patients with achondroplasia, intracranial venous sinus hypertension secondary to jugular foramen stenosis—and in some cases jugular vein stenosis at the thoracic inlet—is the cause of hydrocephalus. Interestingly, none of these patients were found to have any obstruction of CSF or delay in transit of contrast materials from the ventricular system to the basal cisterns, spinal canal, and into the subarachnoid space over the hemispheres. However, in a prior study by our group, and a case report by Miyamoto et al., several patients were noted to have decreased or obliterated CSF flow at the cervicomedullary junction as demonstrated using MR CSF flow studies, presenting the possibility that at least in some patients with foramen magnum stenosis there may be additional factors leading to ventriculomegaly or hydrocephalus. Additionally, in both the study by Steinbok et al. and Miyamoto et al., there was a compensatory increase in occipital and marginal sinus flow as well as condylar emissary vein dilation in the presence of jugular foramenal stenosis. In our case series we questioned whether one of the alternative pathways for venous outflow in these patients may be through the dural venous channels at the cervicomedullary junction, and whether performing a foramen magnum decompression in patients with both foramen magnum and jugular venous stenoses, these accessory venous outflow pathways, may play a significant role in providing alternative pathways for venous outflow and serve to lower intracranial venous pressure following foramen magnum decompression. Such alternative pathways between the cervicomedullary junction and cervical dural anastomoses were not directly measured in Steinbok et al.’s seminal work.

In this paper we review our clinical experience with the surgical treatment of symptomatic foramen magnum stenosis over the past 11 years for the presence or absence of dynamic cervicomedullary compression. We demonstrate that there is a risk of dynamic cord compression and obstruction of CSF flow at the skull base in children with achondroplasia. This phenomenon may be due to bowing of the Cl–2 anterior ligamentous complex. We postulate that this phenomenon may not only result in compression of the cervicomedullary junction, or obstruction of CSF flow, but may also result in exacerbating intracranial venous hypertension secondary to compression of outflow through the cervical dural venous plexus. The technique we used to perform a duroplasty was that of scoring the external leaf of the dura at the foramen magnum and removing thickened posterior cervicomedullary ligament following a suboccipital craniectomy. By doing so, we avoided sacrificing dural venous anastomosis between cranial dural venous channels and spinal venous channels. Our results suggest, but do not provide definitive evidence, that these anastomoses may serve an important function in patients with achondroplasia who have elevated intracranial venous pressures due to stenosis at the level of the jugular foramen. Most of our patients who presented with a full fontanel had subjective improvement in preoperative signs and symptoms without a need for placement of a VP shunt. This improvement may also be a result of improved CSF flow dynamics, but it is difficult to explain any improvement of intracranial pressure accompanying hydrocephalus without a mechanism that includes improvement in venous drainage through venous anastomoses. Finally, it remains to be determined whether isolated dynamic cervicomedullary compression in patients with achondroplasia is an appropriate indication for surgical decompression.

Conclusions

Dynamic cervicomedullary compression is a phenomenon that, although previously described in the literature, has been incompletely explored. This retrospective, single-surgeon, single-institution study represents the largest series to date examining the diagnostic utility of obtaining dynamic cervical imaging in symptomatic patients with achondroplasia. Although further study is needed to address the full diagnostic utility of this imaging modality, there is promise that such dynamic imaging may provide a useful adjuvant diagnostic tool of investigation for a subset of symptomatic patients with unremarkable neutral cervical spine MRI studies.

Disclosure

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

Author contributions to the study and manuscript preparation include the following. Conception and design: Mukherjee, Danielpour, Rimoin. Acquisition of data: Mukherjee, Danielpour. Analysis and interpretation of data: Mukherjee, Danielpour. Drafting the article: Mukherjee, Danielpour, Krakow, Rimoin. Critically revising the article: all authors. Reviewed submitted version of manuscript: all authors. Approved the final version of the manuscript on behalf of all authors: Danielpour. Statistical analysis: Mukherjee. Administrative/ technical/material support: Mukherjee, Danielpour, Pressman, Krakow. Study supervision: Danielpour, Pressman, Krakow, Rimoin.

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