Endoscopic third ventriculostomy in hydrocephalus associated with achondroplasia

Report of 3 cases

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Hydrocephalus in patients with achondroplasia is thought to be due to increased dural sinus venous pressure resulting from narrowing of the jugular foramen. In this setting, where hydrocephalus is presumed to be “vascular” in origin and therefore communicating, endoscopic third ventriculostomy (ETV) would seem contraindicated. The authors describe 3 patients in whom ETV was successfully performed, resulting in MR imaging–documented decreases in ventricle size. The patients were 11 months, 33 months, and 13 years at the time of surgery. All patients had serial preoperative MR images demonstrating progressive hydrocephalus in a “triventricular” pattern with a small fourth ventricle but an open aqueduct. All patients had undergone suboccipital decompression for foramen magnum stenosis prior to the treatment of hydrocephalus. Preoperative retrograde venography revealed variable pressure gradients across the jugular foramen. It is postulated that the increase in intracranial venous pressure resulting from jugular foramen stenosis may lead to disproportionate venous engorgement of the cerebellum and some degree of obstructive hydrocephalus amenable to ETV. The authors discuss the role of suboccipital decompression in the progression of hydrocephalus in patients with achondroplasia.

Key words • achondroplasia • hydrocephalus • endoscopic third ventriculostomy

Hydrocephalus has been recognized in patients with achondroplasia at least since the time of Dandy. Currently, indications for surgical intervention are controversial. In the absence of overt symptoms or signs of raised ICP (for example, headaches or papilledema), achondroplasia patients with large ventricles are usually considered “compensated”—a condition that is often viewed with some suspicion by pediatric neurosurgeons.

The first investigation of the possible etiology of hydrocephalus in achondroplasia was conducted in 1972 by James et al., who concluded that hydrocephalus was “communicating” in 2 patients studied with radionuclide cisternography. Since that time, a series of authors have reached the same conclusion by documenting jugular foramen stenosis, pressure gradients across the jugular foramen, and elevated dural venous sinus pressures. Thus, by all accounts the etiology of ventricular enlargement and hydrocephalus in achondroplasia is considered a result of impaired CSF absorption due to increased dural venous sinus pressure, which in turn is the result of jugular foramen stenosis.

In our experience ventricular enlargement in achondroplasia usually involves only the lateral and third ventricles. The fourth ventricle is usually small, and the posterior fossa appears “crowded.” This “triventricular” pattern of ventricular dilation is considered to be a good prognostic sign for the success of ETV in the treatment of hydrocephalus. Thus, we report on a series of patients in whom ETV was performed as treatment for hydrocephalus associated with achondroplasia.

Methods

After obtaining institutional review board approval, we identified in the Neurosurgeons for Children database all patients who had undergone neurosurgical evaluation for achondroplasia at the Children’s Medical Center in
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Dallas between January 1, 1992, and December 31, 2008. After identifying the patients, charts and radiographic studies were reviewed for data collection. Collected data included patient age at presentation, sex, symptoms at presentation, age at follow-up, the presence of any neurological comorbidities, and surgical procedures performed. All available neuroimaging studies (CT and MR imaging) were reviewed to calculate ventricular size using 2 methods, that is, frontooccipital horn ratios and ventricular volumes.

**Results**

Between January 1, 1992, and December 31, 2008, 57 patients with achondroplasia were evaluated by Neurosurgeons for Children in Dallas, Texas. The patients ranged in age from newborn to 18 years (mean 3.4 years). Twenty-six patients (45.6%) underwent suboccipital decompression, and 8 patients (14%) had surgical treatment for hydrocephalus (ETV or VP shunting). Interestingly, suboccipital decompression for foramen magnum stenosis had been performed in 7 of the 8 patients who subsequently had surgery for hydrocephalus. Thus, 31% of patients who underwent suboccipital decompression also underwent surgery for hydrocephalus. The average interval from suboccipital decompression to hydrocephalus surgery was 4.97 months.

We identified 3 patients with achondroplasia and hydrocephalus who underwent ETV. These patients were considered for ETV due to MR imaging morphology demonstrating triventricular hydrocephalus with apparent fourth ventricle CSF outflow obstruction. Magnetic resonance imaging in these patients also revealed a patent aqueduct and outward bowing of the floor of the third ventricle. These cases are detailed below with representative MR images in Figs. 1–3. Frontooccipital horn ratios and ventricular volumes are listed in Table 1 and presented graphically in Fig. 4.

**Summary of Cases**

**Case 1**

**History and Examination.** This boy was born full term without complications, and a diagnosis of achondroplasia was made on the basis of physical findings. Magnetic resonance imaging performed when the boy was 2 days old showed small ventricles without stenosis of the foramen magnum (Fig. 1A). A subsequent MR image obtained at 4 months showed enlargement of the ventricles and stenosis of the foramen magnum (Fig. 1B). Due to the change in the dimensions of the foramen magnum, a suboccipital decompression (foramen magnum craniectomy only) was performed when the boy was 6 months old.

At 8 months, MR imaging revealed further enlargement of the ventricles, and although the foramen magnum was now decompressed, there appeared to be slight herniation of the cerebellar tonsils (Fig. 1C). Due to progressive ventricular enlargement, a decision was made to treat the hydrocephalus. Retrograde venography was performed, demonstrating a 2-mm Hg pressure gradient across the jugular foramen. Since the gradient was minimal, it was assumed that the hydrocephalus was more “obstructive” in character.

**Fig. 1.** Case 1.  
A: Sagittal and axial MR images obtained at 2 days of age, showing small ventricles without stenosis of the foramen magnum.  
B: Sagittal and axial MR images obtained at 4 months of age, revealing enlargement of the ventricles and stenosis of the foramen magnum.  
C: Sagittal and axial MR images obtained at 8 months of age, 8 weeks after suboccipital decompression, showing additional enlargement of the ventricles, decompression of the foramen magnum, and slight herniation of the cerebellar tonsils.  
D: Sagittal and axial MR images obtained at 16 months of age, 5 months after ETV, demonstrating significantly decreased ventricles.
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Operation. An ETV was performed when the boy was 11 months of age.

Postoperative Course. Postoperatively, the ventricles decreased significantly in size (Figs. 1D and 4A). The patient has had no further neurosurgical procedures and is now 7 years of age and attending an age-appropriate class at school.

Case 2

History and Examination. Achondroplasia was diagnosed based on physical findings in this girl. Magnetic resonance imaging performed when she was 5 months of age showed moderate foramen magnum narrowing and a small pineal region arachnoid cyst (Fig. 2A). Follow-up MR imaging at 22 months showed a moderate increase in ventricular size with the development of periventricular signal changes on FLAIR sequences. Foramen magnum stenosis appeared worsened, and C-1 appeared stenotic (Fig. 2B). When the patient was 26 months of age, suboccipital craniectomy and C-1 laminectomy were performed along with the placement of a Codman intraparenchymal ICP monitor. Intracranial pressure was measured for 5 days, revealing an average ICP of 22 mm Hg when the patient was calm and at rest. Due to this elevated ICP, a VP shunt was placed with an Orbis Sigma II valve.

Postshunt neuroimaging studies failed to show any decrease in ventricular size and actually demonstrated a slight increase. Shunt reservoir puncture showed a patent ventricular catheter with an opening pressure of 20 cm H$_2$O, indicating abnormally high resistance either in the valve or distal catheter. Endoscopic third ventriculostomy was considered given the distended appearance of the third ventricle (Fig. 2C). Retrograde venography demonstrated a 6-mm Hg pressure gradient across the jugular foramen.

Operation. An ETV was performed when the patient was 33 months of age, and the VP shunt was left in place.

Postoperative Course. Post-ETV MR imaging demonstrated a decrease in ventricular size (Figs. 2D and 4B), and repeat shunt reservoir puncture showed the opening pressure to now be 9 cm H$_2$O. The shunt was subsequently removed, and at that time, endoscopic inspection revealed that the third ventriculostomy stoma was patent. The patient is now 8 years of age and in an age-appropriate class at school.

Case 3

History and Examination. This girl was one of a set of identical twins with achondroplasia, which was diagnosed when she was 4 months of age. Magnetic resonance imaging studies at 1 year of age showed small ventricles (Fig. 3A). At 17 months of age the patient presented for neurosurgical evaluation for possible sleep apnea. She began walking at 18 months and was then lost to follow-up until the age of 5 years, when she presented with headaches predominantly in the occipital-cervical region. Magnetic resonance imaging showed enlargement of the ventricles and CTH (Fig. 3B). At the age of 6 years the patient underwent suboccipital decompression.
Six months postoperatively, a decline in the girl’s school performance was reported. Magnetic resonance imaging showed persistent distortion of the supratentorial ventricles, especially the third ventricle. Retrograde venography demonstrated a 15-mm Hg gradient across the jugular foramen, and it was therefore thought that ETV would not be successful. Thus, a VP shunt with an Orbis Sigma valve was implanted when the patient was 7 years of age. The patient fared well for 5 years until at the age of 12 she reported recurrent neck pain. Magnetic resonance imaging of the cervicomedullary junction again showed CTH, and she underwent suboccipital and C-1 bony decompression. One year later the patient reported chronic generalized headaches, and her school performance again declined. Magnetic resonance imaging showed persistent large ventricles (Fig. 3C), and a shunt tap revealed an opening pressure of 18 mm H2O.

**Operation.** An ETV was performed when the patient was 13 years old.

**Postoperative Course.** Postoperatively, her headaches and school performance improved. A follow-up shunt tap showed an opening pressure of 8 mm H2O, and MR imaging revealed a modest but measurable decrease in ventricle size (Figs. 3D and 4C). No further operations for hydrocephalus have been performed in 5 years of follow-up.

**Discussion**

Achondroplasia is a disorder of chondrocyte function occurring with a frequency of about 0.5 cases per 10,000 live births. Inheritance is autosomal dominant, but more than 75% of cases represent a fresh mutation of the paternal *FGFR3* gene. Clinical manifestations include a disproportionally short stature, macrocephaly with frontal bossing, and midface hypoplasia. In the skull the disorder affects the endochondral-derived bone of the skull base, resulting in shortening of the clivus and narrowing of the foramen magnum. Patients with achondroplasia typically present for neurosurgical evaluation in infancy for foramen magnum stenosis, in childhood for enlarged ventricles, and in adulthood for diffuse spinal stenosis.

**TABLE 1: Frontooccipital horn ratios and ventricular volumes before and after ETV**

<table>
<thead>
<tr>
<th>Case</th>
<th>Pre-ETV</th>
<th>Post-ETV</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>FOHR</td>
<td>0.54</td>
</tr>
<tr>
<td></td>
<td>ventricle vol (cm³)</td>
<td>141</td>
</tr>
<tr>
<td>2</td>
<td>FOHR</td>
<td>0.46</td>
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<tr>
<td></td>
<td>ventricle vol (cm³)</td>
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</tr>
<tr>
<td>3</td>
<td>FOHR</td>
<td>0.51</td>
</tr>
<tr>
<td></td>
<td>ventricle vol (cm³)</td>
<td>329</td>
</tr>
</tbody>
</table>

* FOHR = frontooccipital horn ratio.
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Hydrocephalus is known to occur in achondroplasia, although the significance of enlarged ventricles is debated. (Please note that for the purposes of this discussion, the terms “hydrocephalus” and “ventriculomegaly” both refer to enlarged ventricles and may be used interchangeably. Actual measured ventricle size, ICP, and the presence or absence of symptoms of elevated ICP are specifically mentioned.) In the past, the etiology of ventricular enlargement has been variously attributed to stenosis of the foramen magnum with fourth ventricle outflow obstruction, compression of the basal cisterns, aqueductal stenosis, and elevated dural venous sinus pressure. Currently, ventricular enlargement is almost universally considered to be due to impaired CSF absorption resulting from dural venous sinus hypertension, which is in turn the result of jugular foramen stenosis. In this model, CSF accumulation stabilizes with the development of venous collaterals, and the treatment of hydrocephalus in achondroplasia is rarely necessary.

Endoscopic third ventriculostomy is generally not considered in the treatment of hydrocephalus in achondroplasia due to the belief that the pathophysiology is “nonobstructive” or “communicating.” However, hydrocephalus in this setting may be triventricular—an anatomical pattern that is considered to be a good predictor of success in ETV. Etus and Ceylan recognized this triventricular pattern and performed ETV in 2 patients (ages 12 and 14 years) with achondroplasia and hydrocephalus. The procedures resulted in the relief of symptoms but failed to decrease ventricle size. These authors commented on the technical difficulties associated with the procedure but did not discuss the unique pathophysiology of hydrocephalus in this condition. They described the hydrocephalus as both communicating and noncommunicating and, despite MR imaging evidence to the contrary, attributed the obstructive component to narrowing of the cerebral aqueduct. More recently, successful ETV was anecdotally mentioned in a general discussion of the neurosurgical aspects of achondroplasia.

Is it possible that in some patients with achondroplasia and hydrocephalus, the pathophysiology is at least partially obstructive and therefore amenable to ETV? If so, how does this fit with known pathophysiological observations? We reviewed the literature pertaining to these observations.

**Etiology of Ventricular Enlargement and Hydrocephalus in Achondroplasia**

Before the advent of modern computerized neuroimaging techniques, James et al. performed radionuclide cisternography in 2 patients with achondroplasia and concluded that hydrocephalus was communicating.

In 1980 Pierre-Kahn et al. evaluated a series of 25 patients with achondroplasia by using a variety of tests, including CT (24 patients), skull tomography (13 patients), ventriculography/pneumoencephalography (5 patients), isotopic cisternography (5 patients), and cerebral angiography (1 patient). All patients had enlarged ventricles, but only 1 patient underwent surgery—posterior fossa decompression. Head circumference data in 4 patients showed a rapid increase during the first months of life followed by a slowing of the growth rate between the 4th and 24th months. Head size was up to 6 SDs above normal. Dilated scalp veins were observed in the majority of patients, and in the 1 patient who underwent angiography, severe stenosis of the sigmoid sinus was noted at the level of the jugular foramen. Based on these findings, the authors concluded that although hydrocephalus is frequent in achondroplasia, it “usually stabilizes” and that the observed ventricular enlargement is due to impaired CSF...
absorption resulting from increased dural venous sinus pressure. The authors also concluded that stabilization depends on venous collateral formation. Since the publication by Pierre-Kahn et al., a number of papers have contributed support to these conclusions.

Friedman and Mickle\textsuperscript{10} documented elevated superior sagittal sinus pressures in 2 patients with achondroplasia treated using ventricular shunts. In 1 patient, venography demonstrated venous sinus outflow obstruction at the skull base. In 1981 Yamada et al.,\textsuperscript{43} reported narrowing of the jugular foramen on retrograde venography with dilated transverse and sigmoid sinuses. Interestingly, in this study air and contrast ventriculography revealed fourth ventricle outflow obstruction in 2 of 8 patients with achondroplasia. Decompression of the foramen magnum, however, failed to improve hydrocephalus.\textsuperscript{43}

In 1984 Sainte-Rose et al.,\textsuperscript{14} measured pressures in the lateral ventricles, superior sagittal sinus, and jugular veins of 20 patients with various forms of hydrocephalus. In 2 patients with achondroplasia, sagittal sinus pressures were found to be greater than simultaneously measured pressures within the jugular veins—confirming a hemodynamic effect of the jugular foramen stenosis that had been demonstrated by angiography. In addition, the withdrawal of CSF from the ventricle failed to decrease sagittal sinus pressure, indicating that raised sinus pressure was not the result of raised intraventricular pressure.\textsuperscript{38}

In 1989 Steinbok et al.,\textsuperscript{40} recorded ICP in 5 patients and performed jugular venography in 4 patients with achondroplasia. The authors reported elevated ICP in all patients, although the magnitude was severe in only 1 case. Pressure gradients across the jugular foramen measured 3–10 mm Hg, with control patients measured at 1–2 mm Hg. Interestingly, in 2 patients who had at least 1 parent with achondroplasia, an additional venous pressure gradient was recorded across the thoracic inlet. This observation may represent a more severe phenotype, as it is known that patients who are homozygous for achondroplasia have thoracic growth restriction, whose severity ultimately proves fatal.\textsuperscript{29}

In logical progression, Lundar et al.,\textsuperscript{25} in 1990, performed unilateral jugular foramen decompression in a 10-month-old boy with achondroplasia. These authors considered the procedure a success as evidenced by decreased fontanel tension and slowing of the rate of head growth. The CT appearance did not change, however, and perhaps more significantly, there have been no subsequent reports of such procedures in the literature to date.\textsuperscript{25} Unpublished observations involving further attempts at jugular foramen decompression indicate that the procedure is limited by the need to traverse and disturb large collateral venous structures (Cochrane, personal communication, 2010).

With advances in MR imaging technology, a number of reports have described venous anatomy at the jugular foramen in achondroplasia.\textsuperscript{16,28,37} Both Rollins et al.,\textsuperscript{37} and Moritani et al.,\textsuperscript{28} found that the severity of hydrocephalus correlated with the degree of jugular foramen stenosis. Moritani and colleagues further correlated the severity of hydrocephalus with various patterns of collateral venous drainage and found that patients who have demonstrated collaterals through the condylloid emissary vein were less likely to experience progressive hydrocephalus.

Thus, it appears that patients with achondroplasia frequently have obstructions to venous outflow at the jugular foramen, resulting in a pressure gradient and intracranial venous hypertension. The degree of ventricular enlargement correlates with the severity of jugular foramen stenosis and with collateral venous drainage patterns.

**Obstructive Hydrocephalus in Achondroplasia**

Despite the fact that many patients with achondroplasia clearly have raised dural venous sinus pressure due to jugular foramen stenosis, the etiology of hydrocephalus may not be purely “communicating” in all cases. Growth of the posterior fossa is restricted in achondroplasia.\textsuperscript{1,6,20,41,42} In our experience, MR imaging of the posterior fossa frequently shows “crowding” of the cerebellum, with a small fourth ventricle and cisterna magna. This anatomical configuration may lead to obstruction of CSF outflow from the fourth ventricle, resulting in an increase in the size of the third and lateral ventricles with a small fourth ventricle. Further “cerebrocerebral disproportion” in the posterior fossa may also lead to herniation of the cerebellar tonsils, which may result in obstructive hydrocephalus. Indeed, in the featured cases, all 3 patients had the MR imaging appearance of a “tight” posterior fossa and 2 had CTH. All patients demonstrated outward bowing of the floor of the third ventricle.

In conditions in which posterior fossa growth is restricted (for example, multiple suture craniosynostosis), venous engorgement of the cerebellum has been postulated to contribute to CTH.\textsuperscript{9} It is difficult, however, to directly measure cerebellar venous blood volume within the posterior fossa and thus to be conclusive regarding this relationship. Even without overt growth restriction of the posterior fossa, however, intradural venous hypertension appears to be able to cause CTH. This is demonstrated in reports of CTH reduced following embolization in patients with arteriovenous malformations involving the vein of Galen.\textsuperscript{31,34} Thus, it is likely that elevated dural venous sinus pressures are transmitted “upstream,” resulting in increased cerebellar blood volume and “turgor.” This in itself may cause fourth ventricle CSF outflow obstruction but may also lead to CTH, which then worsens the degree of CSF obstruction.

Cerebellar tonsillar herniation in achondroplasia is rare, presumably due to the fact that the foramen magnum is generally stenotic, thus disallowing herniation of the hindbrain.\textsuperscript{15,37} To our knowledge, CTH in achondroplasia has been reported in only 3 papers.\textsuperscript{1,30,37} The report by Rollins et al.,\textsuperscript{37} includes patients in the current study. In the other 2 reports, the CTH is unusual and not applicable to the current discussion in that it appears to be caused by caudal displacement of the hindbrain by large posterior fossa arachnoid cysts.\textsuperscript{1,30} The finding of CTH in 2 of our patients was intriguing. In the oldest patient (Case 3) there was minimal herniation evident on the first MR imaging studies at 1 year of age. This herniation worsened after suboccipital decompression. In the patient in Case 1, mild CTH also occurred after suboccipital decompression. The patient described in Case 2 did not have CTH, but there was complete obliteration of the cisterna magna. These observations prompted us to review all other pa-
tients with achondroplasia evaluated in our practice to see if suboccipital decompression preceded hydrocephalus treatment in other cases. Of 57 patients with achondroplasia referred for evaluation, 8 (14%) were treated for hydrocephalus. Suboccipital decompression preceded treatment for hydrocephalus in 7 of these cases (87%). The relationship remains speculative and may simply represent greater disease involvement in these patients. Or it could be that by releasing the bony constriction of the foramen magnum, herniation of the cerebellar tonsils is possible and impedes CSF outflow from the fourth ventricle.

**Ventricular Shunts in the Treatment of Hydrocephalus in Achondroplasia**

Indications for the treatment of hydrocephalus in achondroplasia are controversial. Shunt placement rates are probably highly variable but have been estimated to be 10%,\(^9\) which is comparable to our treatment rate of 14%. Over the past 2 decades, there appears to be a trend away from shunt placement in this population.\(^2\) Pediatric neurosurgeons may accept large ventricles and macrocephaly with the assumption that relative elevated ICP will be tolerated and venous collaterals will develop, normalizing ICP.

While the majority of patients most likely do not require operation, there are indications for intervention, and patients in such cases should be promptly recognized. The difficulty in monitoring and detecting these patients (who invariably have macrocephaly) is attested to by the report of a 17 month old with achondroplasia who was found to have hydrocephalus and significantly elevated ICP after presenting with blindness.\(^7\) Although specialized head circumference growth curves for achondroplasia have been developed,\(^18\) measurements within this range, even in infants, do not rule out significant intracranial hypertension.\(^19\) The patients in the current study all had head circumference measurements within these standardized values. We agree with Steinbok et al.\(^40\) that reliance on these growth curves—which likely were constructed with the inclusion of patients with hydrocephalus—may be misleading and that attention should instead be paid to the individual ventricular anatomy of each patient.

As mentioned previously, there is a general reluctance to treat hydrocephalus in patients with achondroplasia, as the condition is believed to be compensated. Authors often reserve treatment for patients in whom “progressive” hydrocephalus is demonstrated.\(^28\) However, progression is delineated by the initial observation, and the cases featured in the present study clearly show that if neuroimaging is performed early enough, patients with achondroplasia begin life with normal-sized ventricles. The literature contains few reports of early neuroimaging in patients who are asymptomatic, and thus “progression” is rarely demonstrated. There is one report of increased ventricular size in a 9-year-old boy in whom ventricular size was normal on CT at 1 month of age. The child developed learning difficulties, and ICP monitoring revealed moderately elevated intraventricular pressure.\(^40\)

Further reluctance to treat hydrocephalus in achondroplasia by using ventricular shunts may stem from the development of “slit ventricles” after shunting.\(^33\) Extreme ventricular collapse or slit ventricles may be more likely to occur in patients with achondroplasia due to the increased dural venous sinus pressure and resultant increased cerebrospinal venous pressure.\(^33\) In these cases the maintenance of a functioning shunt and control of ICP can prove frustratingly difficult.\(^24,33\)

**Endoscopic Third Ventriculostomy in the Treatment of Hydrocephalus in Achondroplasia**

As noted in the case histories, all 3 patients demonstrated a modest but measurable decrease in ventricular size after ETV (Table 1). It is well accepted that after successful ETV, the decrease in ventricular size can be subtle.\(^29\) In 2 patients in whom ICP was measured (Cases 2 and 3), the ICP was lower after ETV. In the oldest patient (Case 3) headaches improved after ETV. The patient who was youngest at the time of ETV (Case 1) demonstrated the most significant postoperative change in ventricle size (Fig. 4). No patient has undergone further surgery for hydrocephalus, with follow-ups averaging 5 years. Although the decrease in ventricle size supports the concept that in these cases hydrocephalus was at least partially obstructive, we must consider alternative explanations.

One possible explanation is that ventricle size in achondroplasia may decrease as collateral venous drainage develops. To date there has been no longitudinal survey of ventricular size in achondroplasia to support this concept. We are currently evaluating all available neuroimaging studies in this achondroplasia population to investigate this possibility.

A second alternative incorporates complex theories of cerebral and cranial compliance. Although a complete discussion of these concepts is beyond the scope of this article, there is a body of data supporting the efficacy of ETV in communicating hydrocephalus.\(^13,14\) In this model hydrocephalus is produced by decreased intracranial compliance and maintained by a subsequent increase in systolic pulse pressure in the brain. The creation of a communication between the ventricle and the subarachnoid space (via ETV) reduces the systolic pulse pressure within the brain and relieves hydrocephalus without increasing bulk CSF absorption.\(^13\) Should this model prove correct, the indications for ETV may be significantly widened. To date, however, we have not performed this procedure in patients with clearly open fourth ventricle outlets.

**Is Ventriculomegaly in Achondroplasia Concerning?**

The significance of ventriculomegaly in achondroplasia remains unclear. There is obvious concern that untreated hydrocephalus—if not severe enough to result in overt signs of elevated ICP—may result in cognitive impairment. Many authors broadly state that intelligence in achondroplasia is normal,\(^2,12,35\) whereas others have reported varying degrees of cognitive impairments in these patients.\(^3,17,36,42\) The etiology of these impairments is not always attributable to any disturbance of CSF circulation, however. No study has investigated measurements of ventricular size with neurodevelopment. Clearly, more data are needed in this area. Of possible interest is the observation that the youngest patients in the current study...
remain in age-appropriate classrooms, whereas the oldest patient experienced learning difficulties prior to treatment of her hydrocephalus. We are pursuing formal neuropsychological evaluations of these patients at this time.

Conclusions

The etiology of hydrocephalus in achondroplasia may not always be communicating. Cerebrospinal fluid outflow obstruction from the fourth ventricle may also be present, and therefore ETV may be indicated in a select group of these patients. This report documents decreased ventricular size following ETV in 3 patients with achondroplasia and progressive hydrocephalus.

Disclosure

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Author contributions to the study and manuscript preparation include the following. Conception and design: Swift. Acquisition of data: all authors. Analysis and interpretation of data: Swift. Drafting the article: all authors. Critically revising the article: all authors. Approved the final version of the manuscript on behalf of all authors: Swift. Statistical analysis: Nagy. Study supervision: Swift, Nagy.

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