Foramen magnum stenosis from overgrowth of the opisthion in a child with achondroplasia

Case report

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Achondroplasia has a known association with foramen magnum stenosis that can result in cervicomedullary compression, which is most often due to a hypertrophied posterior occipital rim and an undersized transverse diameter. The authors present a unique case of a child with achondroplasia with symptomatic craniovertebral compression from marked overgrowth of his opisthion anterior to the posterior arch of the atlas. This 22-month-old child with achondroplasia presented with severe respiratory and motor disabilities, including progressive quadriplegia and apneic episodes requiring continuous positive airway pressure. Magnetic resonance imaging and CT scans revealed marked foramen magnum stenosis from overgrowth of the opisthion, a hypoplastic C-1 ring, and spinal cord edema at the cervicomedullary junction. Foramen magnum decompression and a C-1 laminectomy were performed. Postoperatively, steady motor improvement has been observed and the patient no longer requires ventilatory support. To the authors’ knowledge, this is the first report of this unusual anatomical entity. (DOI: 10.3171/PED/2008/2/8/136)

KEY WORDS • achondroplasia • cervicomedullary compression • foramen magnum stenosis • opisthion

Abbreviation used in this paper: CSF = cerebrospinal fluid.
graphic studies using MR imaging and CT revealed marked foramen magnum stenosis (4 mm anteroposterior diameter) because of overgrowth of the opisthion and a hypoplastic C-1 ring with concomitant severe cervicomedullary compression and myelomalacia (Fig. 1). The vertebral arteries pierced the dura mater through the atlantooccipital membrane on either side of the anomalous opisthion.

**Operation.** The patient underwent foramen magnum decompression and a C-1 laminectomy. The laminectomy revealed the aberrant overgrown opisthion. The overgrown opisthion, including the posterior rim of the foramen magnum, was drilled down using a diamond-tipped drill until it was eggshell-thin. The final bone was then removed using small Kerrison rongeurs. The integrity of the dura was maintained, and the vertebral arteries were preserved. After the osseous decompression was accomplished, an intraoperative ultrasonography study, which others have used in patients with achondroplasia, showed good pulsations of the cerebellar tonsils, and it was believed that a duraplasty procedure was unnecessary.

**Postoperative Course.** Postoperatively, the patient no longer required ventilatory support and showed steady improvements in his motor function. At his 2-month follow-up visit, he was able to roll over and exhibited a marked increase in extremity movement and strength, although his left side remained comparatively weaker. There was minimal improvement in his muscle tone, and he continued to have significant developmental delay. His nasal oxygen requirement continued to decrease, and he has had fewer respiratory infections and apneic episodes. The MR images obtained after the surgical decompression revealed a 9-mm foramen magnum (significantly improved from 4 mm preoperatively) with CSF visualized anterior and posterior to the cord (Fig. 2). Subsequent MR imaging has shown persistent T2 signal changes within the cord that are consistent with myelomalacia.

**Discussion**

Achondroplasia is an autosomal dominant dwarfism syndrome that results from a point mutation in the *FGFR3* gene on chromosome 4. Occurring almost exclusively on the paternal allele, and increasing with advanced paternal age, de novo mutations account for 80% of these cases. The syndrome is characterized by dwarfism, rhizomelia, frontal bossing, midface hypoplasia, and a myriad of other systemic morbidities—especially those of a respiratory and neurological nature. Foramen magnum stenosis and resultant cervicomedullary compression is a frequently encountered neurological manifestation of achondroplasia that has been implicated, in infants, in apnea and sudden unexpected death from encroachment on the respiratory center in the medulla. Often, concomitant ligamentous hypertrophy causes an accompanying dense fibrotic epidural band. To our knowledge, our case is the first report of achondroplasia-associated overgrowth of the opisthion invaginating under the ring of C-1 as the cause of craniocervical compression.

Establishing the presence, severity, and degree of craniocervical compression is crucial to management. Somatosensory evoked responses have also proven useful in the identification of compressive syndromes because 43% of asymptomatic patients have somatosensory evoked response abnormalities. Clinical signs and symptoms indicating severe compression include hyperreflexia, clonus, paresis/paralysis of upper or lower extremities, apnea, or hypopnea.
Others suggest that Paul Klimo Jr., M.D., M.P.H., 88th J. Neurosurg.: Pediatrics / Volume 2 / August 2008 and others considering a constellation within this latter faction, however, there is disagreement concerning the signs, symptoms, or findings that warrant surgery, with some physicians basing the decision on objective findings such as CSF flow and intracord lesions and others considering a constellation of clinical symptoms in addition to radiographic findings.

Magnetic resonance imaging scans are invaluable in evaluating the size of the foramen magnum (sagittal and transverse diameters) to determine the degree of compression. Once the diagnosis and the degree and severity of the compression have been ascertained, the debate about treatment options ensues. As noted earlier, the management of cervicomedullary compression is controversial. Some physicians propose early intervention and even prophylactic surgery for asymptomatic patients or those without severe compression because of the potential for irreversible neurological damage and sudden death. Others suggest that the stenosis resolves spontaneously with time, and these advocates would thus reserve surgery for symptomatic patients. This group suggests using serial MR images to monitor asymptomatic children. Within this latter faction, however, there is disagreement concerning the signs, symptoms, or findings that warrant surgery, with some physicians basing the decision on objective findings such as CSF flow and intracord lesions and others considering a constellation of clinical symptoms in addition to radiographic findings.

Conclusions

We present a unique case of foramen magnum stenosis in the setting of achondroplasia. In this case, symptomatic cervicomedullary compression ensued from the inferiorly curved overgrowth of the opisthion ventral to the ring of C-1, presenting a therapeutic challenge. Multiple factors influence the prognosis of this disease, such as the time of diagnosis, duration of the compression, disease severity, comorbidities, access to neurosurgical care, treatment provided, and the time of intervention relative to disease progression. Foramen magnum stenosis and consequent cervicomedullary compression are not only common but also potentially lethal neurological manifestations of achondroplasia, thus rendering their prompt identification and appropriate management crucial.

References


Fig. 2. Postoperative T2-weighted sagittal MR imaging study obtained after the patient underwent suboccipital craniectomy and C-1 laminectomy showing that the foramen magnum dimensions have increased from 4 to 9.0 mm. Unlike in the preoperative images, CSF is now observed anterior to the proximal aspect of the spinal cord. The T2 signal prolongation within the proximal spinal cord likely represents myelomalacia.