CLINICAL VIGNETTE

Management of a Child with Achondroplasia Presenting for Nephrectomy

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Introduction

Achondroplasia is the most common form of dwarfism with a prevalence of roughly 1 in 10,000 births.[1] It is caused by a gain of function mutation in the gene for fibroblast growth factor receptor 3 (FGFR3). The increase in receptor signaling results in impaired growth plate chondrocyte proliferation and bone growth. This manifests in the characteristic phenotype of disproportionate proximal limb shortening (rhizomelia), craniofacial abnormalities, and kyphoscoliosis. However, there are other, less apparent sequelae of achondroplasia that may complicate medical management, especially due to airway and respiratory abnormalities during anesthetic administration.

Case Report

A 14-year-old male with achondroplasia who recently emigrated from Honduras was found to be hypertensive on multiple clinic visits. He reported a few sporadic episodes of headaches, one of which was accompanied by blurry vision. There were no significant findings on review of systems, and he denied any previous treatments or interventions for achondroplasia in his previous country. He was performing well in school and was physically active. On examination, he had a blood pressure of nearly 160/100 on multiple visits. His height of 102 cm was approximately at the mean for his age of the achondroplasia-specific growth chart. He exhibited recognizable features of achondroplasia, including a long trunk, shortened limbs, midface hypoplasia, and frontal bossing. His neurologic exam was remarkable for increased patellar and achilles deep tendon reflexes.

As a result of his exam and complaints, an MRI showed mildly enlarged ventricles suggestive of mild communicating hydrocephalus. The imaging also demonstrated moderate stenosis of the subarachnoid space at the foramen magnum without significant cervicomedullary junction compression.

Renal ultrasound demonstrated a thinned, atrophic left kidney with severe hydronephrosis, and further investigation demonstrated near absent left kidney function on a MAG-3 lasix renal scan. There was no urinary reflux noted on voiding cystourethrogram. These findings, in conjunction with an elevated serum renin, prompted a multidisciplinary team to recommend nephrectomy for treatment of hypertension.

On the day of surgery, the patient was administered moderate sedation with intravenous propofol, midazolam, and fentanyl while endotracheal intubation was performed via a fiberoptic bronchoscope. General anesthesia was then induced with further propofol, and the halogenated inhaled anesthetic sevoflurane. His head, neck, and limbs were carefully padded and positioned into the lateral decubitus position for the start of the nephrectomy. At the end of successful surgery, he recovered overnight in the intensive care unit under continuous cardiorespiratory monitoring. A postoperative clinic visit three months later revealed that he had recovered well and was normotensive.

Discussion

Most instances of achondroplasia arise de novo from parents without the mutation, and in nearly all cases, the mutation in FGFR3 is the same substitution of arginine for glycine (Gly380Arg).[2] In a minority of cases, the mutation has been inherited in an autosomal dominant fashion.[2] Though some cases require genetic testing, most cases can be confidently diagnosed prenatally and at birth by characteristic radiographic and clinical appearances of skeletal abnormalities, such digit shortening, or brachydactyly, on hand x-rays. The identification and treatment of complications from this condition are important, as the mortality rate for these individuals is nearly doubled compared to the general population.[3] Sudden, unexpected death in infants and young children with achondroplasia is a well-recognized complication.[3,4,5] Furthermore, in adults, increased mortality and shorter life expectancy persists in older groups with a median survival that is 10 years shorter.[3] This is primarily attributed to higher rates of cardiovascular disease, the cause for which is unclear.

This pediatric patient demonstrates some of the neurologic complications of achondroplasia. Narrowing of the foramen magnum and resultant medullary compression may cause motor dysfunction, hyperreflexia, central apnea or hypopnea, and impaired consciousness. Though brainstem compression may result in disordered breathing, a study of 17 patients found the degree of stenosis on MRI did not correlate with the severity of central sleep apnea on polysomnogram.[6] The age of maximal narrowing is considered to occur during infancy, and in a study of patients presenting for decompressive craniectomy at a single institution, most surgeries were performed at an age less than 2 years.[7] If brainstem compression is suspected, care should be taken to avoid extreme neck movements or sporting activities that may acutely exacerbate the condition. Similarly, during airway manipulation for endotracheal intubation and

surgical positioning, the patient's head and neck was maintained in a neutral position.

Other neurologic complications of achondroplasia include hydrocephalus, kyphoscoliosis, and spinal stenosis. Hydrocephalus is theorized to result from jugular foramen stenosis, causing high intracranial venous pressure. Progressive macrocephaly is the most common indication for surgery, and studies estimate up to 10% of patients have intracranial shunts by the end of adolescence.[2,7] Spinal stenosis is common due to short pedicles and small neural foramina, though symptoms of neurogenic claudication and myelopathy generally present at older ages.[7]

Obstructive sleep apnea (OSA) and sleep-disordered breathing is another frequent complication of achondroplasia that is attributed primarily to narrowed airways, upper airway obstruction, and to a lesser degree, effects of cord compression on central respiratory control.[8] In a review of 95 children with achondroplasia, 38% of these patients had evidence of OSA, of which most were treated with adenoidectomy, tonsillectomy, or both.[9] Furthermore, in a study of 29 achondroplasia patients with a median age of 3 years, all but two had an abnormal polysomnogram.[10] In the case above, endotracheal intubation was performed using a fiberoptic endoscope as midface hypoplasia and anatomic abnormalities may decrease the success of more routine direct laryngoscopy. Also, this patient was maintained on a continuous pulse oximeter during the immediate recovery period after surgery to monitor for adequate oxygenation and ventilation.

The significant neurologic and respiratory effects of achondroplasia should be anticipated in the perioperative setting due to the rapidly changing clinical environment during surgery and anesthetic administration. Caregivers should focus on identifying those at risk of craniocervical compression and OSA and intervening appropriately, such as by modifying induction and intubation techniques and augmenting intra- and postoperative monitoring.

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