Foramen magnum decompression in an infant with homozygous achondroplasia

Case report

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Homozygous achondroplasia is a rare yet distinct clinical entity. Most infants succumb to an early death as a result of respiratory compromise due to upper airway obstruction, thoracic cage deformity, and/or cervicomedullary compression. The successful cervicomedullary decompression of a 16-week-old infant with homozygous achondroplasia is described. This report suggests that homozygous achondroplasia is not universally fatal and that these infants are potentially viable if managed by aggressive respiratory and surgical measures.

KEY WORDS • achondroplasia • homozygosity • foramen magnum • hydrocephalus • infant

ACHONDROPLASIA is an autosomal-dominant skeletal dysplasia affecting approximately one in every 26,000 live-born infants. Individuals with homozygous achondroplasia (AA) are the progeny of matings between two heterozygote achondroplastic parents (Aa x Aa). Homozygous achondroplasia forms a distinct clinical entity presenting with more severe characteristics than those expressed in the heterozygous individual. Compared to heterozygous infants, homozygous infants have a disproportionately larger neurocranium and a smaller foramen magnum. In addition, their limbs are shorter, their chest is smaller, and their trident hand configuration and skeletal abnormalities are more striking.

Although achondroplastic couples theoretically have a 25% chance of having a child homozygous for the achondroplasia gene, there are few reports of such individuals. It has been assumed that homozygous achondroplasia is uniformly fatal because most reports document the death of these infants within the first 11 weeks of life. Recently, however, Pauli, et al., described the first three homozygous achondroplastic infants to survive 29 to 37 months due to the implementation of an aggressive respiratory management system and in one case due to foramen magnum decompression.

This report offers insights into the surgical management of the rare clinical entity of homozygous achondroplasia and suggests the potential viability of these infants. Based on our experience, aggressive respiratory and surgical measures are indicated in infants with homozygous achondroplasia.

Case Report

This 2.145-kg homozygous achondroplastic baby boy (AA) was referred at 16 weeks of age. His parents were both spontaneous heterozygous achondroplasts (Aa). The child was delivered via elective Caesarean section at 36 weeks of gestation. Homozygosity was suggested in utero by an ultrasound study that revealed a very narrow chest. The patient was intubated immediately following delivery, and remained dependent on mechanical ventilation throughout his life with the exception of several very brief respites. He had many apneic episodes and left focal motor seizures. Early computerized tomography (CT) of the head revealed moderate hydrocephalus (Fig. 1). Placement of a ventriculoperitoneal shunt at the age of 10 weeks led to a moderate decrease in the number of apneic events and seizures. At birth, the head circumference was 34 cm and the chest circumference was 24 cm (normal 34 cm). On axial and coronal CT scans the foramen magnum had a transverse diameter of 8 mm and a sagittal diameter of 10 mm (normal 30 mm) with a constricted cervical canal (Fig. 2). The infant was referred to our institution for foramen magnum decompression.
Foramen magnum decompression in homozygous achondroplasia

Examination. The general examination was notable for pronounced achondroplastic features including a prominent forehead, maxillary hypoplasia, mandibular prognathism, a depressed nasal ridge, extremely short limbs, trident hands, thoracic hypoplasia with a narrow dystrophic rib cage, and cardiomegaly. The patient had marked respiratory insufficiency. He was intubated and was responding well to diuretic and bronchodilating agents. The neurological examination was notable for an alert child with full yet soft anterior fontanel, a palpable working left parietal shunt bulb, intact cranial nerves, increased tone in all four extremities with symmetrical withdrawal to physical stimulation, and bilaterally upgoing toe reflexes.

Operation. Attempts at prone positioning of the patient resulted in immediate oxygen desaturation as detected by transcutaneous pulse oximetry. Presumably this was secondary to further thoracic and abdominal compression and hence caused aggravation of his severe restrictive and obstructive lung disease. Experimentation with different head and body placement in the prone position revealed that mild flexion of the neck with longitudinal traction and prevention of thoracic compression by the abdominal viscera could be achieved without compromising respiratory function. This was evaluated by transcutaneous oxygen saturation and arterial blood gas analysis. To stabilize this position for the duration of the surgery, a halo ring and a body cast extending from the upper sternum to the groin were applied. Brackets and the halo frame were applied to the cast, and attached achieving 30° of head flexion and simultaneous longitudinal traction (Fig. 3). Turning the patient prone in this apparatus maintained respiratory function. A midline skin incision was made from the inion to the C-2 spinous process. A posterior fossa decompression and C-1 laminectomy were then performed using a Surg-airtome, curettes, and Rosen knives. It was apparent that the cervicomedullary junction was markedly compressed by the foramen magnum rim. After decompression, the cervicomedullary junction appeared healthy and pulsatile. Intradural exploration revealed that the medulla and cord appeared intact and healthy. The dura was closed primarily.

Postoperative Course. The patient tolerated the procedure well without oxygen desaturation. On the 12th postoperative day he underwent tracheostomy. Two weeks postoperatively he had a cardiac arrest secondary to an apneic spell, necessitating resuscitation; there were no further cardiovascular or respiratory in-
may become less severe as skeletal growth occurs. Four weeks postoperatively there was no significant change in his neurological findings or in his ventilatory settings.

**Discussion**

Although homozygous achondroplasia is a recognizable clinical entity, it is extremely rare. Most cases are fatal. Respiratory compromise is the single most important factor determining infant mortality in these patients. In achondroplasia, respiratory compromise may be due to one or more of four previously described mechanisms. These are: 1) upper airway obstruction and sleep-disordered breathing secondary to facial hypoplasia and hypotonia of airway maintaining muscles; 2) thoracic cage deformity and hypoplastic lungs; 3) coincident chronic pulmonary conditions including asthma, cystic fibrosis, and cor pulmonale; and 4) cervicomedullary compression secondary to a small foramen magnum and cervical vertebral canal.

Medullary compression causes dysfunction of fibers emerging from the respiratory center and, hence, interference with involuntary ventilation. Cervical compression causes dysfunction of the lateral corticospinal tracts which innervate the diaphragm and intercostal muscles and impairs vital capacity, tidal volume, and cough and diminishes functional residual capacity. Hydrocephalus secondary to obstruction of cerebrospinal fluid pathways at the level of the foramen magnum or of venous outflow at the skull base exacerbate symptoms of cervicomedullary compression.

The four mechanisms mentioned above are even more serious in a homozygous achondroplastic infant, as illustrated by this case. Our patient experienced respiratory compromise secondary to all of the above four mechanisms. He had extreme facial hypoplasia, a very small thoracic cage, concomitant pulmonary abnormalities, and an extremely small foramen magnum.

All mechanisms of respiratory compromise in our patient were addressed medically and surgically. Upper airway obstruction and thoracic hypoplasia were so severe that constant mechanical ventilation was required. Coincident pulmonary problems responded to diuretic and bronchodilating agents and to aggressive pulmonary toilet. Venticuloperitoneal shunting to manage the patient's hydrocephalus markedly decreased the number of apneic events and led to improvement in oxygenation. Only by the application of a special halo device and constant oxygen saturation monitoring could we safely undertake cervicomedullary decompression surgery. This procedure did not lead to an immediate improvement of neurological or respiratory symptoms; however, because of the extreme nature of the compression and the presence of all other simultaneous factors interfering with respiration, it is possible that improvement may take place over a long period of time. With continuous aggressive respiratory management the facial hypoplasia and thoracic cage deformity may become less severe as skeletal growth occurs.

This is the second report of a foramen magnum decompression procedure in a homozygous achondroplastic infant. In the previous report, placement of a ventriculoperitoneal shunt and the performance of foramen magnum decompression surgery led to improvement of neurological symptoms and allowed the progression of developmental milestones. This report confirms that homozygous achondroplasia is not universally fatal in infancy and that these infants are potentially viable if managed expeditiously and aggressively, addressing all possible etiologies of respiratory failure.

If these individuals manage to overcome the severe respiratory compromise of early infancy, they can make developmental strides although with persistent mild general and gross motor delays secondary to their skeletal abnormalities. Because there are no documented reports of homozygous achondroplastic infants surviving beyond 3 years of age, it is premature to comment on the utility of neurosurgical management to extend viability and improve the quality of life beyond this age. Nevertheless, this experience confirms that early aggressive medical and neurosurgical management of these infants plays a positive role throughout infancy in prolonging valuable life expectancy.

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**References**


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