Case Report

Severe pneumonia leading to death in an achondroplastic infant: a rare case report

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Abstract: This report concerns a 9-month-old male with achondroplasia presenting with severe respiratory distress having history of frequent respiratory infections, developmental delay and failure to thrive. Child had septicemia and ended up in multiorgan failure, ultimately leading to death due to cardiopulmonary arrest.

Keywords: Achondroplasia, Mortality, Multiorgan failure, Severe pneumonia

INTRODUCTION

Achondroplasia is the best described and most common form of the congenital short-limbed dwarfing. This disorder is characterized by frontal bossing, midface hypoplasia, otolaryngeal system dysfunction, and rhizomelic short stature. Achondroplasia is apparent at birth and has a birth prevalence of 1 in 20000-30000 live-born infants. It is inherited as an autosomal dominant condition, although 80% of cases occur sporadically as new events in their families. Achondroplasia is caused, in virtually all of the cases, by a G380R mutation in fibroblast growth factor receptor 3 (FGFR3). Being at risk for a variety of respiratory problems, increased frequency of airway malacia in infants and young children with achondroplasia is well known.

CASE REPORT

A 9-month-old boy with achondroplasia reported to the Paediatric Emergency, SGRDIMSR with complaints of fever and cough for 1 week and regurgitation of feeds following an apneic episode which was followed by respiratory distress requiring assisted ventilation. Child presented with history of frequent chest infections since birth. He was born at term with birth weight of 2 kg to gravida 2nd mother with both parents being achondroplastic, and elder 5-year-old normal female sibling. History of delayed milestones and failure to thrive was also present.

At presentation, on examination heart rate was 168/minute, respiratory rates of 58/minute, temperature of 105°F was recorded. Pallor was evident. All anthropometric parameters being less than 3rd percentile on growth charts for achondroplastics, rhizomelic shortening of limbs, saddle nose and prominent frontal bossing was present with large bulging anterior fontanel. Severe respiratory distress was present with tachypnea and marked retractions. Hepatomegaly with liver span of 7 cm was present.

Hemoglobin was 8.5 gm/dl on presentation, sequentially decreased to 5.8 gm/dl corrected to 12.5 with blood transfusions with leukocytosis of 18.200 and neutrophilia. Platelet count remained normal throughout. Liver function tests were deranged with SGOT of 88 and SGPT 28 on day 1, increased to 228 and 997 respectively and serum bilirubin rising to total of 3.8 and direct fraction 1.6 in a span of 2 days and ultimately TSB/DSB of
4.0/2.4 and SGOT/PT of 8248/4449 respectively. Renal functions initially normal increased to blood urea 57 and S. creatinine of 2.08 with dyselectrolytemia. Blood gas analysis was suggestive of hypoxia, hypercarbia and metabolic acidosis throughout the stay with bicarbonate levels ultimately falling to 9.1. Hypoglycemic readings were also documented. Chest X ray showed B/L infiltrates suggestive of bronchopneumonia. CSF examination done was positive with TLC of 10/HPF and 6 neutrophils and raised proteins. CT Head done was normal.

**Figure 1: Achondroplasic parents.**

Child was managed with immediate intubation, fluid resuscitation followed by inotropic support and IV antibiotics.

**Figure 2: Achondroplasic infant.**

Child was managed on lines of septicemia. Blood transfusions given in view of decreased hemoglobin. Ventilatory setting were increased as per clinical and blood gas deterioration. Despite adequate efforts child landed in cardiopulmonary arrest after 6 days of hospital stay and was declared dead.

**DISCUSSION**

With frequent respiratory infections being common, respiratory difficulties and breathing disorders in achondroplasia are thought to underlie the increased risk for sudden infant death and neuropsychological deficits seen in this condition. Respiratory failure due to small thoracic volumes is also reported. Mortality rate at all ages was 2.27 (CI: 1.7-3.0) with age-specific mortality increased at all ages. Despite earlier recognition and treatment of respiratory complications of achondroplasia, increased mortality rates and other complications remain high. Prenatal testing for pregnancies at increased risk is possible once the pathogenic variant has been identified in the family. In individuals in whom there is diagnostic uncertainty or atypical findings, molecular genetic testing can be used to detect a pathogenic variant in FGFR3, the only gene known to be associated with achondroplasia.

Future and ongoing evaluation of the prevalence and impact of respiratory disorders in achondroplasia are recommended.

**CONCLUSION**

Monitoring of height, weight, and head circumference in childhood using growth curves standardized for achondroplasia; evaluation of developmental milestones throughout infancy and childhood; baseline CT scan of the brain in infancy; monitor for signs and symptoms of sleep apnea; monitor for middle ear problems or evidence of hearing loss in childhood; clinical assessment for kyphosis and bowed legs, with radiographic evaluation is necessary. Since respiratory infections are common in these patients prevention and appropriate care should be taken to avoid these, as even early recognition may also have a poor prognosis.

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**REFERENCES**


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