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CASE REPORT

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A case of achondroplasia with severe pulmonary hypertension due to obstructive sleep apnea

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Abstract Achondroplasia is the most common skeletal dysplasia in children. Achondroplasic patients have a short cranial face and midface hypoplasia. They often have sleep-related respiratory disturbances that lead to hypoxemia caused by midfacial hypoplasia, a small upper airway, hypotonia of airway muscles, or brain stem compression. It has been well described that obstructive sleep apnea can cause pulmonary hypertension (PH) through the mechanism of chronic hypoxemia. However, severe PH due to obstructive-type sleep disorder is rare in patients with achondroplasia. In this report, we describe a 5-year-old girl with achondroplasia whose severe PH was caused by upper-airway obstruction and was resolved gradually after adenotonsillectomy.

Keywords Achondroplasia · Pulmonary hypertension · Upper airway obstruction

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Introduction

Achondroplasia, the most common skeletal dysplasia in children [1], is characterized by dwarfism, macrocephaly, and short extremities. Children with achondroplasia have a short cranial face and midface hypoplasia [1, 2], and often have sleep-related respiratory disturbances that lead to hypoxemia [3, 4]. Such respiratory disorders in patients with achondroplasia are associated with upper-airway obstruction or brain stem compression resulting from the anatomic features of achondroplasia [3] Airway obstruction may be caused by midfacial hypoplasia, a small upper airway, and hypotonia of airway muscles. Maxillary hypoplasia may cause reducing the nasopharyngeal airway [5, 11]. It has also been well documented that obstructive sleep apnea (OSA) can cause pulmonary hypertension (PH) through the mechanism of chronic hypoxemia [6–8]. However, severe PH due to obstructive-type sleep disorder is very rare in patients with achondroplasia [9].

This report describes a child with achondroplasia whose severe PH was caused by upper-airway obstruction and was resolved gradually after adenotonsillectomy.

Case

A 5-year-old girl with respiratory distress was referred to our clinic. Her parents complained of recurrent respiratory disturbances, snoring, sleeping with open mouth, and orthopnea for the past 3 years. She also had fatigue and failure to thrive. She had been born at term with macrocephaly and short extremities, and as an infant was diagnosed with achondroplasia. The parents were not consanguineous. Her weight was 14 kg, and height 84 cm. They were both in the lowest 3% for children of her age. Her head circumference was 45 cm. Physical examination revealed that she had tachypnea, tachycardia, and cyanosis. Chest examination showed that she had crackles and intercostal retractions, and the heart sound of S2 was hard. The liver was palpable. Ear–nose

and throat examination showed that she had nasooropharyngeal narrowness and soft palate prolapse with grade 2 adenotonsillar hypertrophy (Fig. 1). Lateral roentgenogram of the nasopharynx demonstrated the narrowness of the upper airway (Fig. 2).

When she was asleep, her oxygen levels with pulse oxymetry were in the range between 87 and 90%, and when she was awake, 92 and 96% at room air. On chest radiography, she had cardiomegaly (Fig. 3), and echocardiography showed right atrial and ventricular enlargement with pulmonary artery dilation. Tricuspid regurgitation flow velocity was 3.28 m/s. We performed cardiac catheterization after the heart failure symptoms improved. In the catheterization, the pulmonary arterial pressure was 108/77 mmHg (mean 91), when simultaneous aortic pressure was 114/53 mmHg (mean 75). Cranial tomography findings were normal and did not show brain stem compression. An adenotonsillectomy was performed. Peri- and postoperative complications did not occur. Respiratory disturbances improved dramatically. Pulmonary hypertension decreased gradually. Six weeks after the operation, she had normal echocardiographic and clinical findings.

Discussion

Children with achondroplasia often have respiratory complications related to the anatomic features of the disease [3–5, 10, 11]. The most common of these complications is sleep-disordered breathing, which is associated with either obstructive-type sleep apnea or central apnea [3, 4]. Mogayzel et al. [4] reported that a minority of achondroplasic children have severe sleep-disordered breathing. Zucconi et al. [5] reported that in children with achondroplasia, upper-airway obstruction is a more frequent cause of sleep-disordered breathing than central apnea.

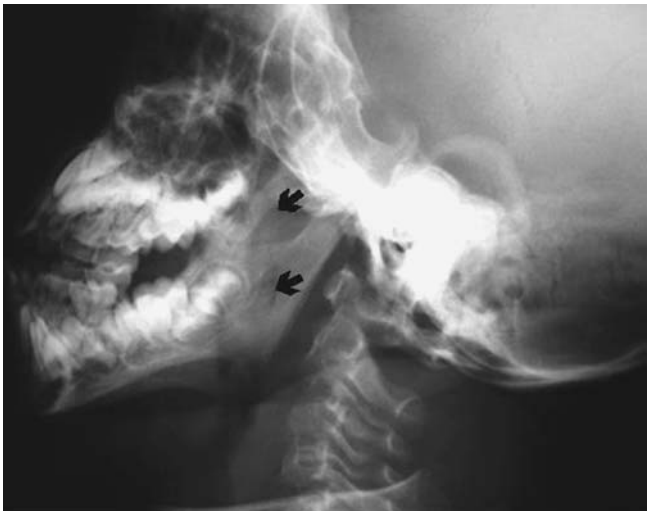

Fig. 2 Lateral roentgenogram of the nasopharynx demonstrates narrowness of the upper airway (*arrows*)

In these patients, respiratory disturbances cause marked hypoxemia. Chronic hypoxemia is the main reason that pulmonary vascular resistance increases, thereby inducing PH [6–8].

Although chronic hypoxemia due to sleep-disordered breathing can occur in achondroplasia, it is very rare for severe PH to follow as a complication such these patients. Ito et al. [9] reported one patient with achondroplasia who had severe PH due to narrowness of the foramen magnum and cervical canal. This case was diagnosed as central apnea. Gunthard et al. [12] also reported a case of achondroplasia with PH. In that case, the PH was secondary to marked pulmonary hypoplasia.

In our case, obstructive-type sleep-disordered breathing was the main cause of severe PH due to midface hypoplasia, adenotonsillar hyperplasia, lingual, and soft palate prolapse, and PH was more severe than the other reported cases caused by upper-airway

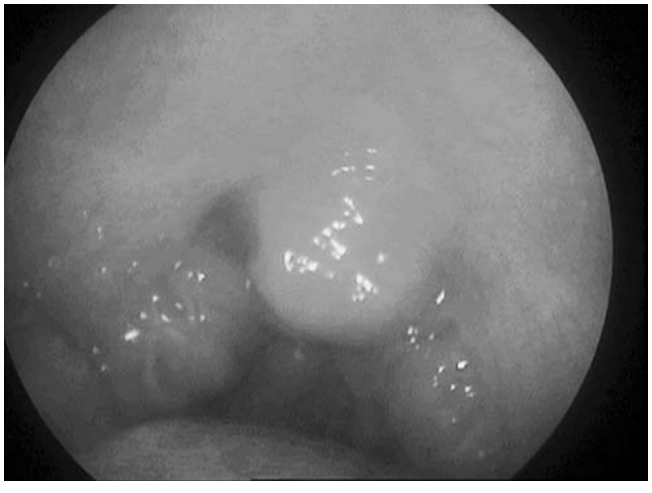

Fig. 1 Laryngoscopic view of grade 2 adenotonsillar hypertrophy with soft palate prolapse

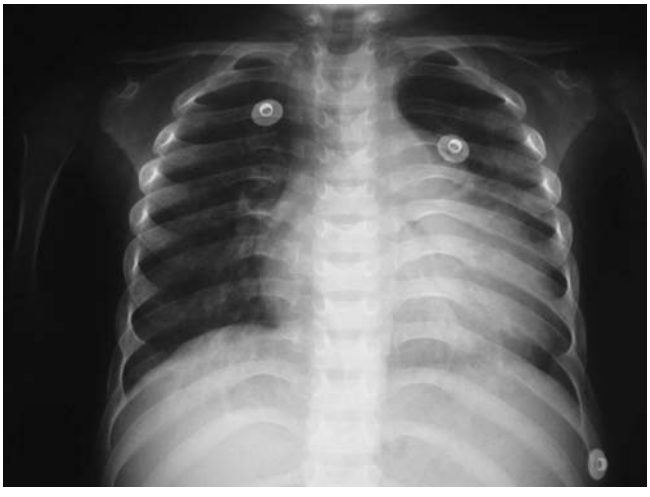

Fig. 3 Cardiomegaly on chest X-ray

obstruction in the literature. Unfortunately, we could not perform polysomnography because there is not laboratory in our region.

Although adenotonsillectomy is an effective treatment in general population with OSA, studies about the effectiveness of adenotonsillectomy in achondroplasic patients have been limited [11]. In our patient, we observed that severe PH resolved immediately after the adenotonsillectomy operation.

In conclusion, we considered that the skeletal and anatomic abnormalities associated with achondroplasia predispose patients to chronic hypoxemia and reversible severe PH could develop as a result. Surgery is indicated: decompression of the narrowness of brain stem in the case of central apnea, and adenotonsillectomy in the case of obstructive-type sleep disorder.

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