

Case Report / Olgu Sunusu

Severe pulmonary hypertension due to sleep-disordered breathing in an achondroplastic child

Akondroplazik Bir Çocukta Uykuda Solunum Bozukluğuna Bağlı Gelişen Ciddi Pulmoner Hipertansiyon

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ÖZET

Akondroplazi çocuklarda en sık görülen iskelet displazisidir. Akondroplazik hastalarda üst solunum yollarının obstrüksiyonu ve kraniofasiyal dismorfoloji nedeniyle solunum yolu yakınmaları sık görülmektedir. Bu hastalarda özellikle geceleri gelişen kronik hipoksemi pulmoner hipertansiyona yol açabilmektedir. Bu makalede ciddi gün içinde de devam eden pulmoner hipertansiyon gelişen akondroplazik bir çocuk sunulmuştur.

Anahtar kelimeler: akondroplazi, uykuda solunum bozukluğu, pulmoner hipertansiyon, sürekli pozitif havayolu basıncı

ABSTRACT

Achondroplasia is the most common skeletal dysplasia in children. Achondroplastic patients often have respiratory problems associated with upper respiratory tract obstruction and craniofacial dysmorphology. Chronic hypoxemia in these patients can result in pulmonary hypertension. In this report an achondroplastic child with severe day-time pulmonary hypertension is presented.

Key words: achondroplasia, sleep-disordered breathing, pulmonary hypertension, continous positive airway pressure (CPAP)

INTRODUCTION

Achondroplasia is the most common skeletal dysplasia characterized by short stature with disproportionately short arms and legs, a large head, and distinctive facial features, including a prominent forehead and a flattened midface [1]. These patients often have respiratory problems associated with upper airway obstruction and craniofacial dysmorphism. Obstructive sleep apnea in achondroplasia has been linked to brain stem compression and poor mid-face development leading to a smaller upper airway [2-4]. The relationship between obstructive sleep apnea and pulmonary hypertension has been well documented that sleep apnea results in repetitive nocturnal arterial hypoxemia and hypercapnia, and acute increases in pulmonary artery pressure [5-8]. Severe day-time pulmonary hypertension has been rarely reported in patients with obstructive sleep apnea syndrome [6,9,10].

In this report, a 9-year-old achondroplastic male with severe day-time pulmonary hypertension and sleep-disordered breathing treated with continuous positive airway pressure is described.

CASE REPORT

The patient was referred to our pediatric cardiology department because of respiratory difficulty, fatigue and somnolence with increasing in intensity in last three months. He had been followed up with the diagnosis of achondroplasia and obstructive sleep apnea syndrome at another center. Physical examination revealed tachypnea, nasal flaring, intercostal retractions, tachycardia, cyanosis and somnolence in addition to dysmorphic features of achondroplasia. Cardiovascular examination also demonstrated a hard second heart sound with a grade II/VI systolic murmur best heard at the left sternal border. Ear-nose and throat examination showed grade II adenotonsillar hypertrophy. His oxygen saturation with pulse oxymetry were in the range between 70% at room air and 85% with nasal mask

oxygen supply. The pro-brain natriuretic peptide (pro-BNP) level was 831pg/ml.

Chest x-ray (Figure 1) demonstrated cardiomegaly with prominent pulmonary conus and echocardiography revealed enlarged right heart chambers with dilated main pulmonary artery, severe tricuspid and pulmonary regurgitation with a flow velocity of 4.3m/s and 3.2m/s respectively. Lateral roentgenogram of the nasopharynx demonstrated the narrowness of the upper airway (Figure 2).

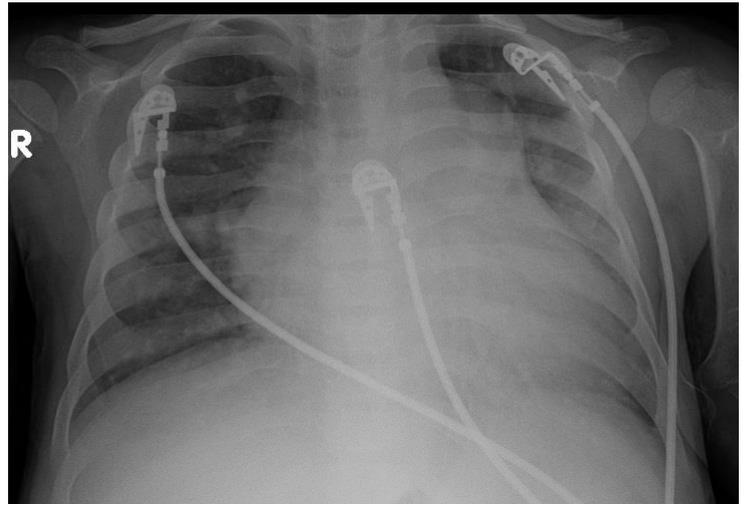


Figure 1. Chest x-ray of the patient demonstrating cardiomegaly and prominent pulmonary conus

He was operated 1 year ago for adenotonsillar hypertrophy at another center and polysomnography was done 5 months ago because of recurring snoring, sleeping with open mouth and sleeplessness, and demonstrated episodes of apnea. Nasal mask CPAP during sleep was recommended, but he did not use it properly. Prior echocardiographic examination, done at 6 months ago, was normal except mild tricuspid regurgitation with a flow velocity of 2.6m/s.

He was hospitalized and supportive medications (furosemide and angiotension converting enzyme inhibitor) and the use of nasal mask CPAP were started as initial treatment. Symptoms and echocardiography findings improved and patient was discharged with CPAP mask on the next week of hospitalisation. The pro-BNP level was 55pg/ml. On the follow up at 1 month, cardiothoracic ratio was decreased and

echocardiography demonstrated normal right chamber sizes and mild tricuspid regurgitation with a flow rate of 2.7m/s.



Figure 2. Lateral roentgenogram of the nasopharynx demonstrated the narrowness of the upper airway and also enlarged skull with relatively small skull base

DISCUSSION

Sleep-disordered breathing due to either obstructive type sleep apnea or central apnea is frequently seen in patients with achondroplasia. The upper airway obstruction accounts for the majority of the cases. These patients have decreased central respiratory response to both hypoxemia and hypercapnia occurring during sleep and also to decreased upper airway tone [2-4]. Recurrent episodes of alveolar hypoxemia especially during sleep leads to pulmonary vasoconstriction and pulmonary hypertension [5-8].

Day-time pulmonary hypertension, reported in approximately 20% of the patients with obstructive sleep apnea syndrome, had been attributed to continuing intimal hyperplasia and medial hypertrophy [5]. Although sleep-disordered breathing is common in achondroplasia (both obstructive and central apnea), severe pulmonary hypertension had been rarely reported [6,9,10].

Adenotonsillectomy is the treatment of choice in general population whereas its effectiveness in achondroplastic patients is limited [11]. As in our case, despite the prior surgical intervention the complaints of our

patient recurred and led to development of severe pulmonary hypertension. CPAP mask is another option for these patients [6,7]. Although it was recommended after surgery, he could not use the mask properly. After the use of mask ensured at hospital, symptoms and degree of pulmonary hypertension improved dramatically.

In conclusion, sleep-disordered breathing can lead to severe complications especially in patients with craniofacial dysmorphism as in achondroplasia. Although adenotonsillectomy is the treatment of choice in most of the cases, CPAP can be tried in patients not suitable for surgery or refractory to surgical intervention.

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