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Adult Pierre Robin Syndrome-Related Difficulties in the Treatment of Sleep Apnea: A Case Report

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Abstract

Most reports of sleep apnea in Pierre Robin Sequence (PRS) patients are for pediatric PRS, with few reports for adults. We report a case of obstructive sleep apnea in an adult patient with PRS that was difficult to diagnose and treat. A 46-year-old female, was referred to our hospital with a complaint of awakening because of respiratory disorder while sleeping. When she was 16 years old, she had undergone otoplasty, mandibulectomy, and bone extension surgery. She had no tonsillar hypertrophy but presented with typical bird-like facies and difficulty in opening the mouth. Imaging findings showed mandibular inferior growth, airway narrowing, bilateral mandibular condyle head deformity, and adhesions between the temporal bone and mandible condyle head. A mono-block type oral appliance was fabricated and patient was recommended to use it, but the airway was not widened with this therapy. We suggested other treatment options, such as surgery, nasal airway tube, and myofunctional training, but the patient refused surgical treatment because of the psychological burden. Therefore, we requested an all-night polysomnography (PSG) test, as a simple sleep test may not accurately capture a patient's breathing events. The PSG results showed apnea hypopnia index (AHI) of 23.1, indicating that the patient could not get deep sleep because of frequent awakenings caused by hypopnea. Continuous positive airway pressure, which was not indicated in the simple sleep test, was now indicated after the PSG. After three months of using continuous positive airway pressure, the patient's Epworth sleepiness scale score improved dramatically from 17 to 3 and AHI from 37 to 2.1. We thus concluded that treatment options vary depending on the patient's episode and background, and the evaluation by performing an all-night polysomnogram is necessary.

Keywords: Obstructive sleep apnea; Pierre Robin syndrome; Continuous positive airway pressure; Polysomnography; Oral appliance

Abbreviations: PRS: Pierre Robin Syndrome; OSA: Obstructive Sleep Apnea; CPAP: Continuous Positive Airway Pressure; RDI: Respiratory Disturbance Index; OA: Oral Appliance; PSG: Polysomnography; AHI: Apnea Hypopnea Index; AASM: The American Academy of Sleep Medicine; EEG: Electroencephalogram

Introduction

Pierre Robin syndrome (PRS) is characterized by micrognathia, glossoptosis, and airway obstruction. Although some cases of PRS in the pediatric population have been reported, few reports discuss the treatment of obstructive sleep apnea (OSA) in adults with PRS. Surgery has been widely reported as a treatment for PRS in children. The use of a continuous positive airway pressure (CPAP) is not covered by insurance in Japan if the apnea-(RDI) is less than 40 on a simple sleep test. An oral appliance (OA) is not indicated for patients who cannot move the mandible forward because of temporomandibular joint deformity. Surgery or other treatment options should be considered for such patients. Herein, we report the case of OSA in an adult woman with PRS who underwent jaw surgery in childhood, and her condition was difficult to diagnose and treat.

Case Report

A 46-year-old female visited our otolaryngology department because her family members informed her that she used to stop breathing while sleeping; general fatigue during the day was another concern. The patient was referred to our department after a simple home sleep test. Her medical history included diabetes mellitus (glycated hemoglobin level [HbA1c] 7.6), hypertension, hyperlipidemia, and PRS. Ear and jaw surgery, including otoplasty, mandibulectomy, and bone extension surgery,

had been performed by plastic and oral surgeons since the age of 16 years. Her physique was normal, with no tonsillar hypertrophy, and she had not reached menopause. She had bird-like facies with difficulty in opening the mouth (23mm Figures 1A & 1B). Oral findings included misaligned teeth, stenosis of the mandible, and five missing teeth (Figure 1C).

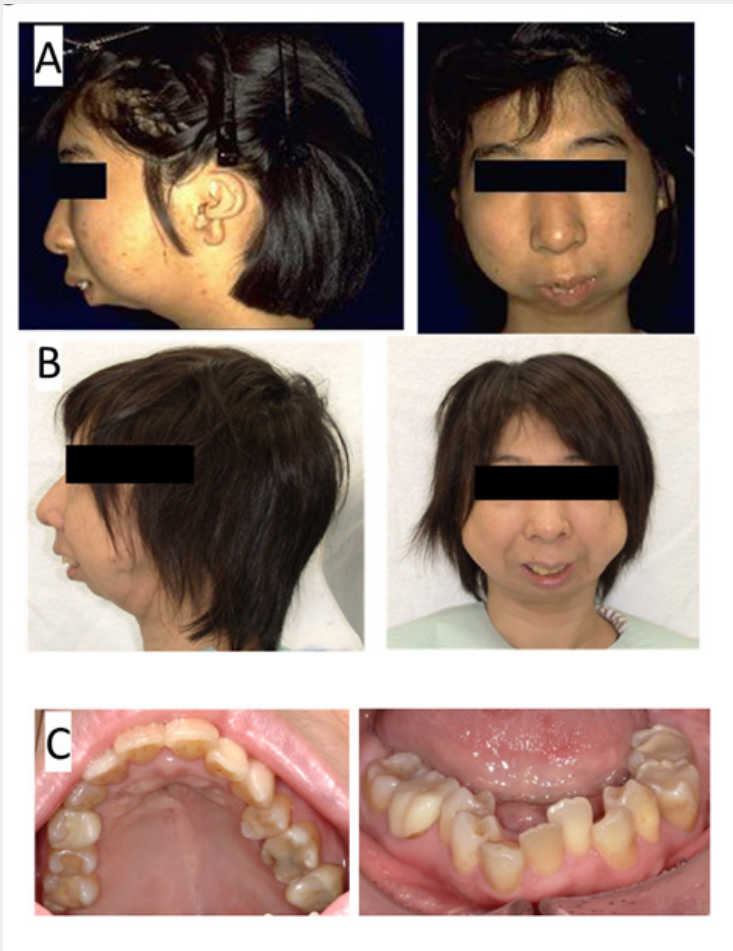

Figure 1: Facial appearance at ages 16 [A]and 46[B], respectively.
There is obvious inferior growth of the mandible.
When the patient was 16-years old, the pogonion moved anteriorly and inferiorly resulting from mandibular growth or temporary bone extension surgery.

Panoramic radiograph imaging exhibited deformities of bilateral mandibular condyle and their fusion with the temporal bone (Figure 2A). A cephalometric analysis showed with obvious mandibular undergrowth and a narrowed airway (Figure 2B). A simple sleep test was performed at the otolaryngology department; the AHI (apnea hypopnea index) and the Epworth sleepiness scale scores were 37 and 17, respectively.

An OA was used as the first-line treatment for the present case; however, the patient was unable to open her mouth (2 lateral finger opening), and adhesion of the mandible to the maxilla prevented the forward positioning of the mandible. No airway opening was observed using an endoscope (Figure 3). Thereafter, we explained the remaining treatment options of surgical therapy, nasal airway tube, and oral muscle functional training to the patient. However, the patient wanted to avoid surgical therapy owing to the multiple surgeries she had undergone previously. Therefore, we requested an all-night polysomnography (PSG) test, as a simple sleep test may not accurately capture a patient's breathing events. The PSG test results showed an AHI of 23.1, arousal index of 28.7, minimum oxygen saturation of 77%, and Oxygen Desaturation Index of 18.7, indicating that the patient could not get deep sleep because of frequent awakenings caused by hypopnea. CPAP, which was not indicated in the simple sleep test, was now indicated in the PSG; as such, CPAP with a nasal mask (Sleepmate, Teijin healthcare Co. Ltd., Tokyo, Japan) was used. The PSG test results with CPAP also demonstrated dramatic improvements after three months. Despite the small sized jaw, a positive pressure for CPAP was achieved by using the nasal mask. After three months, she saw a drastic

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decrease in her daytime sleepiness and increased physical energy in the morning. When using a CPAP, the Epworth sleepiness scale score decreased to 3, and a dramatic improvement in AHI score was observed (2.1).

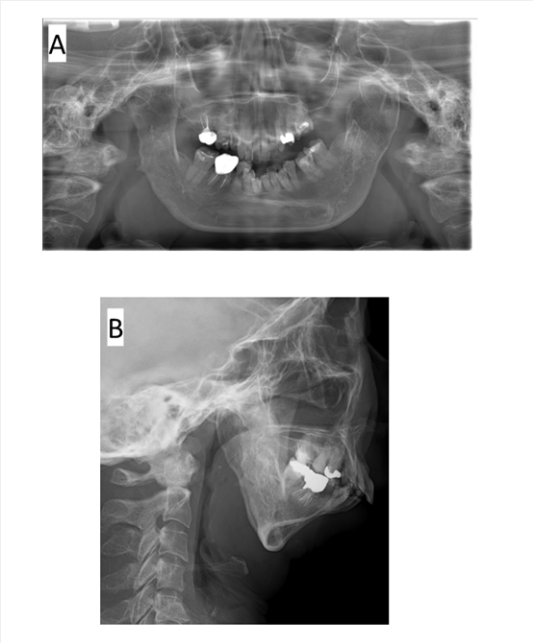

Figure 2: Panoramic and cephalometric radiographs.
Abnormal mandibular morphology and bony adhesions between the maxilla and the mandible are observed. Cephalometric X-P shows inferior growth of the mandible and an obvious narrowing of the airway.

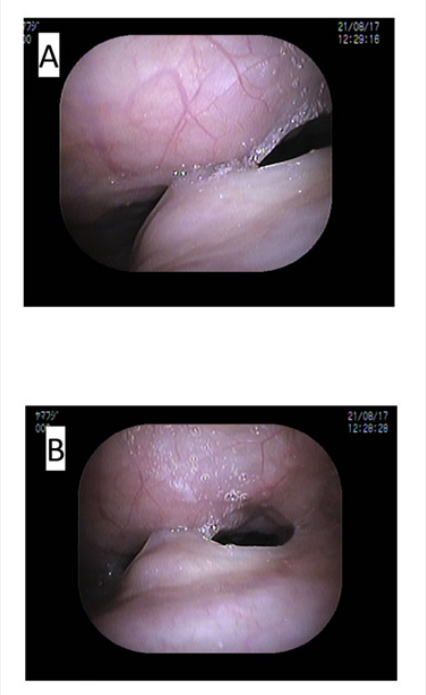

Figure 3: Endoscopy findings.
No difference between A: normal and B: with obstructive apnea (OA), no airway opening, no effect.

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Discussion

In the present case we reported the effectiveness of using CPAP in adult patients with PRS related OSA and also indicated that PSG tests must be performed again when simple sleep tests do not meet the key diagnostic criteria.

Patients with PRS present with micrognathia, tongue depressions, and consequent airway obstruction, and neonatal respiratory urgency at birth is dangerous. The prognosis is relatively good if airway management and other interventions are provided at the appropriate time, and if the patient receives appropriate patient education and parent counseling during infancy.

Treatments are tailored to the characteristics and features of each patient. Surgery is indicated to improve breathing and feeding and to prevent choking in neonates and infants. In addition, surgery is performed for esthetics, mastication, and sleep disorders in childhood. Previously, Goudy et al. [1] found that pediatric patients with PRS with cardiac or neurological disease and children who use CPAP or biphasic positive airway pressure require surgical treatment [1]. Adults with PRS who are free of intellectual developmental difficulties are often treated in childhood, and it has also been reported that the mandible develops and grows with age, resulting in a wider airway [2]. For this reason, reports of adults remain rare, and the oldest report of PRS that we have been able to identify is that of a 20-year-old woman by Miller et al. [3] Treatment of sleep apnea in adults with PRS is similar to that for OSA patients in general, and primarily consists of using CPAP, which can also be used in patients with micrognathia using a nasal mask. If these criteria are not met, OA, surgery, or other treatment options should be considered. OA may be more effective if the mandible can be forwardly positioned. However, if, as in the present case, mandibular advancement is not possible at all due to adhesions between the jawbone and temporal bone, surgical therapy, or other options represent the only options. The indications for surgery for sleep apnea in adults have been guided by Stanford University, USA, and are indicated for patients who have dropped out of other treatment modalities and patients for whom OA and CPAP are not indicated [4]. Nonetheless, patients with PRS are often operated in childhood and tend to avoid surgical therapy. As such, patients for whom neither OA nor CPAP is indicated and who do not wish to undergo surgical therapy may use a CPAP at their own expense.

Numerous systemic effects of apnea have been reported. Intermittent hypoxia due to sleep apnea induces inflammation and promotes the pathogenesis of atherosclerotic thrombosis due to metabolic syndromes. In addition, a correlation between sleep apnea and hypertension, cardiovascular disorders, and diabetes has been reported [5-7], indicating that sleep fragmentation due to apnea is closely associated with these diseases. Our patient did not get deep sleep, woke up frequently, and suffered from hypertension, diabetes mellitus, and hyperlipidemia, which may have been caused by her sleep apnea.

Sleep test devices are classified into four types, with the American Academy of Sleep Medicine (AASM) defining Type 1 as PSG, Type 2-4 as portable; Type 2 is capable of Electroencephalogram (EEG) recording and can be performed at home without an attendant, Type 3 has a minimum of four channels, and Type 4 has a minimum of one or two channels. In Japan, simple sleep tests are often carried out by non-specialists and false-negative results are common. As such, an algorithm has been developed by the AASM to ensure that sleep disorders requiring treatment are not overlooked by the simple sleep test [8]. In addition to clinical judgment based on a simple sleep test, PSG should also be carried out in patients with sleep disorder-related symptoms or in light of the presence of complications or pre-existing conditions. It has been reported that there was no correlation between the simple sleep test and PSG for mild-to-moderate AHI and mild cases tend to be less accurate in a diagnosis [9]. This is due to the fact that PSG can determine sleep duration based on EEG data, electro-oculography, and mentalis electromyography recordings. However, in a simple sleep test, the time from sleep onset to awakening is considered sleep time, and the time spent awake during the process is also included in sleep time. Therefore, the AHI tends to be lower in the simplified test given that the sleep time is longer. Therefore, portable devices without EEG are used in combination with actigraphs (small motor volume sensors), and the effectiveness of peripheral arterial blood flow measurement devices (peripheral artery tonometry) has also been reported [10].

Another reason for which AHI is calculated to be lower with the simple sleep test is the difference in the criteria for determining hypopnea. In Japan, diagnostic criteria are not defined for respiratory events using a simple sleep test. Therefore, instead of diagnosing hypopnea solely on the basis of decreased respiratory airflow, only events accompanied by decreased oxygen saturation should be considered, and the diagnostic criteria for hypopnea should be clearly tailored to the Japanese population. In summary, we conclude that the simple sleep test should not be trusted solely and that patients with sleep-related symptoms and complicated or pre-existing diseases should be diagnosed with PSG.

Conclusion

We report a case of sleep apnea in a patient with PRS who was diagnosed with OSA but was not indicated for CPAP on the basis of a simple sleep test. However, she was subsequently retested with PSG and her condition was found to be favorable for the use of CPAP.

Informed Consent

This case report has the patient's consent.

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