ORIGINAL ARTICLE

Orofacial Deformities and Obstructive Sleep Apnoea Syndrome

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Introduction

Obstructive sleep apnoea syndrome (OSAS) is a disorder characterized by repetitive closure of the upper airway during sleep. OSAS shows a strong association with obesity. However, OSAS is also associated with orofacial deformities, particularly in the young,[1] irrespective of body weight. Over the last decade, there has been a growing recognition that craniofacial abnormalities occur commonly in OSA patients. These cases need evaluation of the upper airway anatomy. Cephalometry radiographs have been used for many years to evaluate facial growth and development.[2–4] Cephalometry enables the analysis of dental and skeletal anomalies as well as of soft tissue structures and form. Computed tomography (CT),[5] magnetic resonance imaging (MRI),[6] and other imaging techniques of the upper airway tract are helpful in obtaining a detailed anatomy prior to treatment. The recognition of the role of craniofacial abnormalities in the development of OSA has led to a number of treatment strategies aimed at correcting or improving the craniofacial structure, thereby preventing upper airway collapse during sleep. Surgery represents the best therapeutic option for OSAS patients with craniofacial deformities. Numerous options are available, and combinations of the various options are possible, depending on the type of deformity. Continuous positive airway pressure (CPAP) therapy and/or oral appliances may be used in cases where surgical treatment is not available or results in failure.

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Our Experience

We assessed 31 patients with symptoms of OSAS and orofacial deformities who attended our outpatient department or were referred for preoperative evaluation prior to corrective surgery.[7] Of the 31 cases included in this study, the age range was from 6 to 27 years, the mean age being 15.10 years; 17 (54.8%) were males and 14 (45.2%) were females. The mean body mass index (BMI) was 16.72kg/m^2. Snoring and excessive daytime sleepiness (EDS) were the predominant symptoms in 27 (87.1%) and 20 (64.5%) cases, respectively. The other symptoms included choking during sleep 16 (51.6%), scholastic backwardness 15 (48.4%), irritability 14 (45.2%), non-refreshing sleep 13 (41.9%), enuresis 12 (38.7%) and early morning headache 11 (35.5%) cases. In most cases, symptoms were present since early childhood. Late presentation at a mean age of 15.10 years despite early childhood symptoms indicates lack of awareness of OSAS related to orofacial deformities.

The predominant orofacial deformity associated with OSAS in this study group was bilateral temporomandibular joint (TMJ) ankylosis with micrognathia/retrognathia in 14 (41.9%) and unilateral TMJ ankylosis with micrognathia/retrognathia in8 (25.8%). Other deformities were retrognathia 4 (12.9%) and micrognathia 2 (6.5%). Rare syndromes such as Goldenhar syndrome, Klippel–Feil syndrome, Laurence–Moon–Biedl syndrome and Treacher–Collins syndrome, each contributed to 1 (3.2%) case. Surgery was performed in 25 (80.6%) cases, 5 (16.1%) refused

surgery and opted for CPAP and 1 (3.2%) case was treated with oral prosthesis (see Figure 2). Symptoms improved significantly in 96.8% of the patients following surgery. Similarly, postsurgery polysomnography (PSG) showed improvement, with the mean apnoea-hypopnoea index (AHI) dropping from 19.21 to 5.19 and the mean saturation values of 79.52 increasing to a mean value of 91.81, indicating significant improvement and response to therapy. Thus, OSAS in this study showed definite improvement to surgical correction. In 1 (4%) case, despite multiple surgery the patient had persistent OSAS, which improved with CPAP therapy. CPAP therapy was advised in five cases who refused surgery and where sleep study with these devices showed improvement. One case of retrognathia treated with oral prosthesis could not be reassessed with PSG but reported improvement in symptoms.

Common Orofacial Abnormalities Associated with OSAS

The association of OSAS with orofacial deformity seems to be due to compromised upper airway space. Compared with OSAS occurring with obesity, which commonly occurs in the middle-aged people, OSAS with orofacial deformity is observed mostly in early childhood as deformities are mostly congenital where growth of the facial bone is hampered. Bilateral ankylosis of TMJ is the most common craniofacial deformity causing OSAS in children. Unilateral TMJ ankylosis causes sleep apnoea rarely, and only if associated with micrognathia/ retrognathia. Syndromic associations of craniofacial deformities causing OSAS have also been reported.[7–9]

Investigations for OSAS with Orofacial Deformities

Patients with a suspected OSAS can be evaluated by PSG in a sleep laboratory. This involves continuous polygraphic monitoring the neuro (electroencephalography [EEG], electromyography [EMG] and electrooculography [EOG]) and cardiorespiratory (heart rate, pulse oximetry, snoring airflow and respiratory effort) channels. Some additional channels such as body positioning may also be included. The criterion for diagnosis of OSAS was AHI >5 with symptoms in those above 18 years. In children however, AHI >1 with symptoms is considered positive for sleep apnoea.[10] Lowest oxygen saturation (LSAT) of less than 80% predicts significant airway risk. Orofacial deformity requires evaluation of the upper airway anatomy so as to consider appropriate surgery. Cephalometry radiographs have been used for many years to evaluate facial growth and development. Cephalometry enables the analysis of dental and skeletal anomalies as well as of soft tissue structures and form. CT and MRI of the upper airway tract have recentlyproved to be more helpful in obtaining a detailed anatomy prior to surgery.

Therapeutic Options for OSAS with Orofacial Deformities

Various surgical methods[11] such as TMJ ankylosis release, mandibular distraction osteogenesis (DO) and maxillomandibular advancement (MMA), either alone or in combination, showed a good success rate. TM joint ankylosis, with or without retrognathia or micrognathia, can cause OSA due to anatomical defect, leading to difficult mouth opening and upper airway narrowing. TM joint arthroplasty can be performed to release the ankylosis.[12] MMA is very effective in patients with OSAS, both with and without orofacial anomalies.[13] Mandibular DO also is a viable option for young patients with upper airway obstruction due to mandibular deficiency.[14,15] DO allows facial skeleton expansion without requiring bone grafting and with less risk of relapse. For later-onset OSAS, distraction osteogenesis may represent an alternative when acute bone movement is expected to be difficult (scarring from previous surgery or radiation therapy).Cases that fail after surgery or refuse surgery may show improvement with CPAP.

Conclusion

There is a definite association of OSAS with orofacial deformity, and reason appears to be compromised upper airway space. Compared with OSAS observed with obesity, which commonly occurs in middle-aged people, OSAS with orofacial deformity is seen mostly in early childhood as deformities are mostly congenital or traumatic, where growth of the facial bone is hampered.Ankylosis of TMJ is the most common orofacial deformity causing sleep apnoea in children and is observed with bilateral TMJ ankylosis. Unilateral TMJ ankylosis causes sleep apnoea rarely, and only if associated with micrognathia/retrognathia. Syndromic associations need to be recognized to treat concomitant problems. Classic symptoms of sleep apnoea are not

commonly seen in children. They present with snoring, scholastic backwardness and irritability as measures symptoms. The gold standard for diagnosis of sleep apnoea is PSG. Radiological investigations such as CT of the face with the upper airway anatomy and MRI of the upper airway anatomy help in understanding and detecting the etiological/predisposing cause for sleep apnoea. In cases of OSAS caused by orofacial deformity, surgery is the definitive therapy. Untreated OSAS in the young are neurocognitive disorders. Cardiac consequences are seen later in life. Hence it is important to recognize and treat OSAS associated with orofacial deformities as early as possible to avoid these consequences.

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