ORIGINAL ARTICLE

Craniofacial deformities and obstructive sleep apnoea syndrome (OSAS) in the young

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Abstract

Background: Obstructive sleep apnoea syndrome (OSAS) shows a strong association with obesity. However, OSAS is also associated with craniofacial deformities irrespective of body weight, particularly in the young. Surgery represents the best therapeutic option for OSAS patient with craniofacial deformities. This study was undertaken to evaluate OSAS caused by craniofacial deformities and to study the effect of surgical correction in these cases.

Methods: Thirty one patients with symptoms of OSAS and craniofacial deformity who attended our outpatient department or were referred for pre-operative evaluation prior to corrective surgery were included in this retrospective study. Details of demography, history and clinical examination were obtained. Polysomnography (PSG) performed was level III (cardio-respiratory or limited channel study). The diagnostic criteria were symptoms suggestive of sleep apnoea and apnoea hypopnoea index (AHI) of > 1 in children and > 5 in adults. Surgical treatment consisted of release of the temporo-mandibular joint (TMJ) ankylosis, mandibular distraction osteosis (DO) and maxillomandibular advancement (MMA) either alone or a combination of surgeries. Patients were reassessed with repeat PSG to document improvement post intervention. Those patients who did not show improvement with surgery alone were treated with CPAP therapy. CPAP therapy was advised in cases that refused surgery. One case of retrognathia was treated with oral prosthesis.

Results: Of the 31 cases were included in this study, age range from 6-27 years, the mean age was 15.10 years and 17 (54.8%) were males while 14 (45.2%) were females. The mean body mass index (BMI) was 16.72 kg/m^2. Snoring and excessive day time sleepiness (EDS) were the predominant symptoms in 27 (87.1%) and 20 (64.5%) cases respectively. The other symptoms included choking during sleep in 16 (51.6%), scholastic backwardness in 15 (48.4%) irritability in 14 (45.2%), non refreshing sleep in 13 (41.9%), enuresis in 12 (38.7%) and early morning headaches in 11 (35.5%) cases. In most cases symptoms were present since early childhood. The predominant craniofacial deformity associated with OSAS in this study group was bilateral TMJ ankylosis with micrognathia/ retrognathia.in 14 (41.9%) and unilateral TMJ ankylosis

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with micrognathia/ retrognathia in 8 (25.8%). Other deformities were retrognathia 4 (12.9%) and micrognathia 2(6.5%). 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. Symptoms improved significantly in 96.8% of the patients following surgery.

Conclusion: OSAS with craniofacial deformity is caused by compromised upper airway space. Bilateral or unilateral ankylosis with retrognathia/ micrognathia of TMJ is the most common craniofacial deformity causing OSAS in the young. Syndromic associations of craniofacial deformities also cause OSAS. Surgical methods TMJ ankylosis release, mandibular DO and MMA, either alone or in combination show good success rate. The consequences of untreated OSAS in the young are neuro cognitive disorders and cardiac consequences seen later in life. Hence it is important to recognize and treat OSAS associated with craniofacial deformities as early as possible to avoid these consequences.

Introduction

Obstructive sleep apnoea syndrome (OSAS) shows a strong association with obesity. However, OSAS is also associated with craniofacial deformities particularly in the young[1] irrespective of body weight. These cases need evaluation of upper airway anatomy so as to consider appropriate surgery. Cephalometrie radiographs have been used for many years to evaluate facial growth and development.[2,3] Cephalometry enables analysis of dental and skeletal anomalies as well as soft tissue structures and form. Computed tomography (CT) and magnetic resonance imaging (MRI) and other imaging techniques of upper airway are helpful in knowing the detailed anatomy prior to surgery

Surgery represents the best therapeutic option for OSAS patients with craniofacial deformities. There are numerous options available and combinations of the various options are possible, depending upon the type of deformity. Continuous positive airway pressure (CPAP) therapy may be given in cases where surgical treatment is not available or fails. This study was undertaken to evaluate OSAS caused by craniofacial deformities and to study the effect of therapy that was surgical correction in these cases.

Methods

A retrospective study of OSAS and craniofacial deformities was undertaken in the department of Pulmonary Medicine, BYL Nair Hospital. Institution ethics committee approval was obtained. Thirty one patients with symptoms of OSAS and craniofacial deformity who attended our out patient department or were referred for pre-operative evaluation prior to corrective surgery were included. Demographic profile was obtained. History included following details: excessive daytime sleepiness (EDS), snoring, choking, scholastic backwardness, irritability, early morning headache, nocturnal enuresis and non refreshing sleep. Clinical examination included body mass index (BMI), neck circumference and upper airway examination including nasal polyps and adenotonsillar enlargement. All patients were subjected to the following investigations hemoglobin (Hb), complete blood counts (CBC), haematocrit (PCV), thyroid function test, arterial blood gases (ABG), spirometry with flow volume loop, electrocardiogram (ECG), 2-dimensional echocardiography (2D ECHO), X ray chest, cephalometry and computed tomography (CT) of face and neck. Polysomnography (PSG) performed was level III (cardio-respiratory or limited channel study) using "Compumedics, P-Series Plus" sleep system. The diagnostic criteria for sleep apnoea were symptoms suggestive of sleep apnoea and apnoea hypopnoea index (AHI) of > 1 in children and > 5 in adults. The interventions were either surgery and / or CPAP. Surgical treatment consisted of release of the temporo-mandibular joint (TMJ) ankylosis, mandibular distraction osteosis (DO) and maxillomandibular advancement (MMA) either alone or a combination of surgeries. Patients were reassessed with repeat PSG to document improvement post intervention. Those patients who did not show improvement with surgery alone were treated additionally with CPAP therapy. CPAP therapy was advised in cases who refused surgery and where sleep study with these devices showed improvement in AHI. One case of retrognathia was treated with oral prosthesis.

Results

Of the 31 cases included in this study, age rangeed from 6-27 years, the mean age was 15.10 years and 17 (54.8%) were males while 14 (45.2%) were females. The mean body mass index (BMI) was 16.72 kg/m^2. Snoring and excessive day time 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.

The predominant craniofacial deformities associated with OSAS in this study group were bilateral TMJ ankylosis with micrognathia/ retrognathia.in 14 (41.9%) and unilateral TMJ ankylosis with micrognathia/ retrognathia in 8 (25.8%). Other deformities were retrognathia 4 (12.9%) and micrognathia 2(6.5%). Rare syndromes like Goldenhar syndrome, Klieppel Feil syndrome, Laurence Moon Biedl syndrome and Treacher Collins syndrome contributed to 1 (3.2%) cases each (table 1). The extrapolation of predominance of any craniofacial deformity in any particular sex could not be done due to small sample size and passive case findings.

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 (figure 1). Symptoms improved significantly in 96.8% of the patients following surgery. Similarly post surgery PSG showed improvement with mean AHI dropping from 19.21 to 5.19 (figure 2) and mean saturation values of 79.52 increasing to mean value of 91.81 (figure 3) indicating significant improvement and response

Table 1: Type of Craniofacial Deformity Associated with Obstructive Sleep Apnoea Syndrome (OSAS)

Diagnosis	Male (17)	Female (14)	Total (31) No (%)
Bilateral TMJ ankylosis with micrognathis/retrognathia	9	4	13 (41.9)
Unilateral TMJ ankylosis with micrognathis/retrognathia	4	4	8 (25.8)
Micrognathia	0	2	2 (6.5)
Retrognathia	2	2	4 (12.9)
Goldenhar syndrome	0	1	1 (3.2)
Klippel Feil Syndrome	1	0	1(3.2)
Laurence Moon Biedl Syndrome	1	0	1(3.2)
Treacher Collins Syndrome	0	1	1 (3.2)

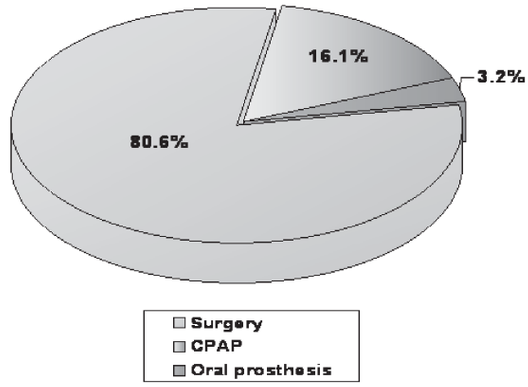

Figure 1: Therapeutic Options in the 31 Cases of Craniofacial Deformity Associated with Obstructive Sleep Apnoea syndrome (OSAS)

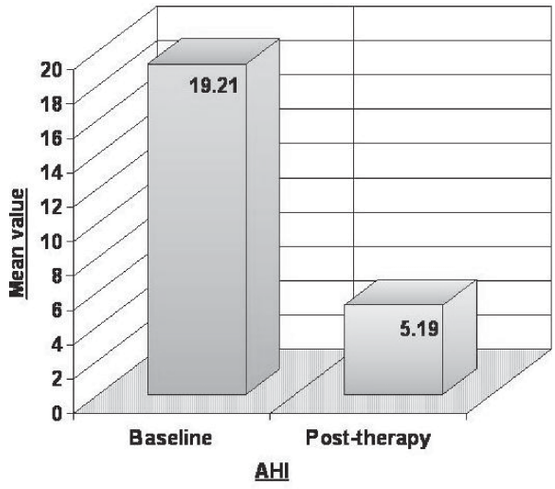

Figure 2: Baseline and Post Surgery mean AHI in Cases of Craniofacial Deformity Associated with Obstructive Sleep Apnoea syndrome (OSAS) Subjected to Surgery

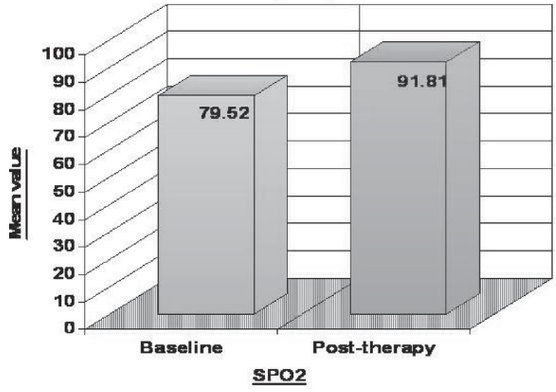

Figure 3: Baseline and Post Surgery Mean Oxygen Saturation in Cases of Craniofacial Deformity Associated with Obstructive Sleep Apnoea syndrome (OSAS)Subjected to Surgery

to therapy. Thus OSAS in this study showed definite improvement to surgical correction. One (4%) of the cases in spite of multiple surgeries had persistent OSAS, which however improved with CPAP therapy. CPAP therapy was advised in 5 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.

Discussion

There is definite association of OSAS with craniofacial deformity and the reason appears to be compromised upper airway space. As compared to OSAS seen with obesity which commonly occurs in middle aged person OSAS with craniofacial deformity is seen mostly in early childhood as deformities are mostly congenital where growth of facial bone is hampered. Bilateral ankylosis of TMJ is the most common craniofacial deformity causing OSAS in children. Unilateral TMJ ankylosis causes sleep apnea rarely only if associated with micrognathia/ retrognathia. Syndromic associations of craniofacial deformities causing OSAS have been reported[10, 11]. Late presentation at a mean age of 15.10 yrs in spite of early childhood symptoms in our study indicates lack of awareness of OSAS related to craniofacial deformities. It was also noted that the average BMI of our cases was below the normal range.

Our study population had snoring and EDS as predominant symptoms. Classic symptoms of OSAS are not commonly seen in the young. They have irritability, scholastic backwardness and nocturnal enuresis as major symptoms as also noted in our cases. Radiological investigations like cephalometry, CT face with upper airway imaging and MRI upper airway anatomy help us in understanding and detecting the nature, location and severity of upper airway obstruction predisposing to OSAS. The gold standard for diagnosis of OSAS is PSG. 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.[12]

In our study various surgical methods TMJ ankylosis release, mandibular DO and MMA, either alone or in combination showed good success rate. TM joint ankylosis, with or without retrognathia or micrognathia can cause OSA due to either anatomical defect leading to difficult mouth opening and upper airway narrowing. TM joint arthroplasty can be done to release the ankylosis. [13] MMA is very effective in patients with OSAS both with and without craniofacial anomalies[14]. Mandibular distraction osteogenesis (DO) also is a viable option for the young patient with upper airway obstruction due to mandibular deficiency.[15] DO allows facial skeleton expansion without the need for 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).[16] In the present study, cases who failed on surgery or refused surgery showed improvement with CPAP. The consequences of untreated OSAS in the young are neuro cognitive disorders. Cardiac consequences are seen later in life. Hence it is important to recognize and treat OSAS associated with craniofacial deformities as early as possible to avoid these consequences.

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