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# OSAS in craniofacial syndromes: an unsolved problem

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**Abstract**

Although the incidence of obstructive sleep apnea syndrome (OSAS) in craniofacial syndromes is high, it is often not recognized and, thus, not treated. In order to study the diagnostics and treatment options for these patients, we studied a group of 72 patients treated in our hospital for Apert, Crouzon or Pfeiffer syndrome, and compared our findings with the literature. There appears to be an agreement on polysomnography (PSG) and airway endoscopy as the main diagnostic options, but therapies are very diverse. Early diagnostics and prompt therapy will prevent serious complications.


*Keywords:* Childhood OSAS; Craniofacial syndrome; Polysomnography

## 1. Introduction

Obstructive sleep apnea syndrome (OSAS) is a complex of symptoms related to episodic, partial or complete obstruction of the upper airway during sleep. OSAS is often part of craniofacial syndromes because the abnormal anatomy has an effect on the potency of the upper airway. It may result in considerable morbidity or even mortality.

The incidence of OSAS in various syndromes is one in eight in Pierre Robin syndrome [1], about 40% in Apert, Crouzon and Pfeiffer syndromes [2–4], 35% in achondroplasia [5], 45% in Down syndrome [6] and 89% in mucopolysaccharidosis [7].

The high incidence of OSAS in these patients, and its serious consequences, prompted us to study the diagnostics and various therapeutic options for our own patients and in the

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literature. Therefore, we studied in a selected group of our patients with Apert, Crouzon or Pfeiffer syndrome if airway obstruction was mentioned in their records, if and how OSAS was diagnosed and which therapies were applied. Furthermore, we compared our findings with the data from other studies in the literature, with a view to evaluate diagnostic methods and the various therapies.

## 2. Methods

We studied retrospectively the records of all patients born between 1984 and 2001, diagnosed with Apert, Crouzon or Pfeiffer syndrome and under medical treatment in Sophia Children's Hospital.

Their reports were screened for a history or signs of airway obstruction, such as stridor, snoring or apnea. The use of polysomnography (PSG) in these patients was recorded, as well as the various treatments applied. The Medline database was searched for OSAS in Apert, Crouzon and Pfeiffer syndromes, more specifically its diagnosis and therapy.

## 3. Results

### *3.1. Study group*

The study group consisted of 72 patients (42 males and 30 females), with a mean age of 9.3 years (0–17). Their diagnoses were Apert syndrome (28), Crouzon (30) and Pfeiffer (14).

Symptoms of airway obstruction were recorded in 19 patients (26%), nearly all with Apert or Crouzon syndrome.

Polysomnography was introduced in our hospital in 1993. Since then, 11 children from the study group underwent this investigation. In five of them, OSAS was the diagnostic outcome. The studied patients were treated in the following ways: adenotonsillectomy in 22, tracheostomy in 4, nocturnal oxygen in 4, continuous positive airway pressure (CPAP) in 2, nasal washout in 5 and craniofacial surgery in 11 children. More than one treatment was possible in a single child.

### *3.2. Literature*

In studies on childhood OSAS, nocturnal polysomnography is considered the gold standard in the diagnosis of OSAS [8].

Endoscopic evaluation of the airway appears to be useful in children with craniofacial syndrome and OSAS; the same is true for cephalometry. [8] Surgical therapies which have proven their value in relieving symptoms are adenotonsillectomy [8] and tracheotomy [8], contrary to other surgical approaches such as mandibular [9], maxillar or hyoid advancement [10], partial glossectomy [8], etc., of which only case studies have been published. CPAP [11] and a nasopharyngeal tube [12] are very effective treatment modalities, although most children do not easily accept the latter. Nocturnal administration of oxygen

is somewhat controversial, as it may worsen hyperventilation. However, if monitored carefully, it may be beneficial and safe for the patient in which a first-choice therapy was not successful [13].

## 4. Discussion

OSAS in children with craniofacial disorders deserves more attention than has been given in the past. In 26% of our patients, symptoms of airway obstruction were recorded, while 40% or more would be expected according to the literature. Polysomnography was applied in only 11 patients. This low figure may be explained by several factors: the fairly recent introduction of PSG in our institution, the diagnosis was obvious or because OSAS was treated earlier with success. Treatment of our patients and of patients reported in the literature appeared very diverse, reflecting the diverse underlying pathology or absence of a single universally applicable therapy.

Because the incidence of OSAS in patients with craniofacial disorders is so high, it is advisable to pay attention to symptoms such as snoring, apnea during sleep, aspiratory stridor, chest retractions during sleep, disturbed sleep or arousals. Presently in our institution, polysomnography is indicated in all patients with craniofacial disorders and a history or signs of upper airway obstruction. The importance of PSG lies in the confirmation of the diagnosis of OSAS, establishing its seriousness and in comparing the effects of therapy. The first PSG is usually combined with endoscopy of the airway.

The best therapy of childhood OSAS, in general, is adenotonsillectomy. This procedure is also the first treatment of choice in children with craniofacial disorders and OSAS, unless the tonsils or adenoid are small. In patients with craniofacial syndrome, however, the operation is less successful, as the adenoid or tonsils are not the main contributors to the obstruction. Nocturnal oxygen can relieve symptoms in patients with moderate OSAS if an adenotonsillectomy was not successful. Many patients with severe OSAS benefit from noninvasive mechanical ventilation, CPAP or BiPAP (bilevel positive airway pressure). Alternatives are a nasopharyngeal tube and the treatment of last resort, a tracheotomy. The value of more causal therapies, such as tongue reduction in cases with macroglossia, maxillar or mandibular reconstruction, still has to be proven.

In conclusion, early recognition of the symptoms of OSAS in craniofacial disorders is mandatory for all physicians involved in the care of these children. Untreated childhood OSAS may lead to complications, such as right heart failure or sudden death.

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