ScientificScholar®
Knowledge is power
Publisher of Scientific Journals

https://iap-kpj.org

Karnataka Paediatric Journal

Letter to the Editor

Management of obstructive sleep apnoea in Apert syndrome with non-invasive ventilation

N. Nagarjun[1]

[1]Department of Pediatric Pulmonology and Sleep, Shishuka Children's Specialty Hospital, Bengaluru, Karnataka, India.

***Corresponding author:**
Nagarjun N,
Department of Pediatric Pulmonology and Sleep, Shishuka Children's Specialty Hospital, Bengaluru, Karnataka, India.

drnagarjun.n@gmail.com

Received: 20 March 2024
Accepted: 01 December 2024
Published: 30 January 2025

DOI
10.25259/KPJ_8_2024

Quick Response Code:

Dear Editor,

Apert syndrome is an uncommon form of craniosynostosis marked by early fusion of the skull sutures. Patients with this condition can develop obstructive sleep apnoea syndrome (OSAS) due to their craniofacial anomalies. Here, we describe a case of Apert syndrome complicated by OSAS, which responded well to continuous positive airway pressure (CPAP) treatment.

A 7-year-old boy presented with characteristic features of Apert syndrome, including craniosynostosis, sclerodactyly, short stature, ridging along the cranial sutures, prominent bulging eyes, underdeveloped midface with maxillary hypoplasia, crowded teeth, and a high-arched palate.

The referral to the Paediatric Sleep Medicine clinic was prompted by reports from the boy's teacher indicating increased daytime sleepiness and instances of falling asleep during meals. This worsening in alertness was noted to be more severe than usual, with the child even falling asleep while standing. In addition, the mother reported that the boy slept excessively, averaging around 20 h/day, and had a history of snoring since birth, along with frequent awakenings at night and episodes of Apnoea. Child did not have choanal atresia. Drug-induced sleep endoscopy (DISE) ruled out any anatomical abnormalities. Computed tomography (CT) scan of head and neck was not performed.

A comprehensive diagnostic polysomnography (PSG) was conducted, providing valuable insights into the boy's sleep patterns and respiratory events during the night. The PSG revealed a total sleep time of 458.5 min, with a sleep efficiency of 81.9%. The distribution of sleep stages showed 21.2% in N1 sleep, 63% in N2 sleep, 5.9% in N3 sleep and 10% in rapid eye movement (REM) sleep.

Importantly, the PSG recorded 76 respiratory events, including 35 predominantly obstructive apnoeas. The apnoea and hypopnea index were calculated at 10.9 events/h, indicating significant respiratory disturbances during sleep, there were no central apnoeas. The central apnea-hypopnea index (AHI) was less than 0.1. In addition, there were 373 recorded snores, with a mean oxygen saturation (SaO_2) of 98% and a minimum SaO_2 of 72%, leading to a desaturation index of 10.8 events/h.

Based on these PSG findings and the clinical presentation, the decision was made to initiate CPAP therapy at 6 cm H_2O using an oronasal mask. Mask fitting and sensitisation were performed in the hospital for 48 hours. pressures were started at physiological pressures and gradually escalated. Oronasal mask was well tolerated. The boy responded well to CPAP treatment, demonstrating good adaptation and compliance. CPAP data extracted showed usage for more than 5 hours per night for more than 30 days, with minimal leak. Three months after starting CPAP therapy, the

boy showed remarkable improvement, with the resolution of daytime sleepiness, increased activity levels, improved ability to engage in conversations, and nearly normal school attendance, good school time behavior and better focus during tasks and activities.

Apert syndrome, the most severe form of syndromic craniosynostosis, is caused by mutations in the fibroblast growth factor receptor 2 (FGFR2) gene and inherited in an autosomal dominant manner. It is characterised by various craniofacial abnormalities such as symmetrical complex syndactyly of the hands and feet, bicoronal synostosis, exorbitism, hypertelorism and midface hypoplasia.[1] About 40% of Apert syndrome cases may develop OSAS, primarily due to midface hypoplasia. However, OSAS can also arise from changes in the changes in the tone of the pharynx, larynx and/or tracheal lumen.[2]

Untreated OSAS in Apert syndrome can lead to complications such as sleep disturbances, frequent infections, growth and developmental delays, cognitive impairment, cor pulmonale or even sudden death. Therefore, a PSG study is essential as it is the standard test for diagnosing OSA.

Studies indicate that CPAP therapy can be an effective treatment for severe OSAS in children with syndromic craniosynostosis.[3] However, recent experiments have shown that a significant number of children may still experience severe OSAS even after undergoing midfacial advancement surgery.[4] DISE ruled out the need for surgery as the collapse was mainly a pharyngeal collapse, and surgery would not be beneficial.

CPAP therapy significantly improved OSAS symptoms in Apert syndrome, emphasising its crucial role in comprehensive management.

Ethical approval

Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

REFERENCES

1. Ko JM. Genetic syndromes associated with craniosynostosis. J Korean Neurosurg Soc 2016;59:187-91.
2. Landete P, Pérez-Ferrer P, Chiner E. Síndrome de Apert y apnea de sueño. Arch Bronconeumol 2013;49:364-5.
3. Chang Y, Yu Y, Zhang W, Gao Y, Feng J, Li M, *et al*. The effect of continuous positive airway pressure on obstructive sleep apnea in children with syndromic craniosynostosis. Sleep Breath 2024;28:1439-48.
4. Saxby C, Stephenson KA, Steele K, Ifeacho S, Wyatt ME, Samuels M. The efect of midface advancement surgery on obstructive sleep apnoea in syndromic craniosynostosis. J Craniofac Surg 2018;29:92-5.

How to cite this article: Nagarjun N. Management of obstructive sleep apnoea in Apert syndrome with non-invasive ventilation. Karnataka Paediatr J. 2024;39:164-5. doi: 10.25259/KPJ_8_2024