Letters to the Editor

Apert Syndrome and Sleep Apnea

Síndrome de Apert y apnea de sueño

To the Editor,

Apert syndrome is a rare variant of craniosynostosis, characterized by premature fusion of the cranial sutures, causing physical and mental health problems in patients from an early age. During the course of the disease, patients may develop obstructive sleep apnea syndrome (OSAS), due to their various craniofacial abnormalities. We present a case of Apert syndrome with OSAS treated satisfactorily with CPAP, which has not been previously reported in the Spanish literature.

A 6-year-old girl, diagnosed with craniosynostosis and syndactyly of the hands and feet, who had undergone surgery at the age of 3 for cleft palate and craniostomy, followed up in the pediatric and children's trauma departments. She was referred to the Respiratory Medicine clinic with a report from her teacher saying that in recent weeks she had been falling asleep, not only in class, as was usual, but also at mealtimes, and the food had to be taken out of her mouth after she fell asleep at the table. It was very difficult to keep her awake or to wake her if she had fallen asleep, and on occasions she had even fallen asleep standing up. The mother reported that the child slept a lot but poorly, had snored from birth and slept almost 20 h a day, going to bed at 19:00 h and waking frequently, with repeated periods of asphyxia.

Physical examination revealed short stature, ridging along the cranial sutures, with a advanced coronal suture fused at the join of the orbit, prominent, bulging eyes, underdeveloped midface with maxillary hypoplasia, crowded teeth and high-arched palate (Fig. 1A and B). Mallampati score 4 with no hypertrophy of the tonsils. Scarring secondary to surgery performed at 10 months for syndactyly with membranes and proximal and mid-phalanges fused in the hands, along with pollex varus and hallux varus in the feet (Fig. 1C). The patient was very sleepy throughout the

Fig. 1. (A and B) Characteristic facies of Apert syndrome with facial hypoplasia. (C) Syndactyly and sclerodactyly. (D) Patient’s baseline polysomnography showing predominance of obstructive apneas and recording from autoCPAP connected to the polygraph flow channels.

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Acceso a los mecanismos asistencial para evitar el rostro de los pacientes con enfermedad pulmonar obstructiva crónica. 

Sinopsis: 

La obstrucción de la vía aérea durante el sueño, conocida como síndrome de la disfunción respiratoria obstructiva durante el sueño (OSAS), es una condición común que puede ser causada por una variedad de factores. En este estudio, se propone un enfoque integral para el manejo del OSAS, incluyendo medidas tanto farmacológicas como no farmacológicas, con el objetivo de mejorar la calidad de vida y el tratamiento a largo plazo de estos pacientes. 

La intervención del tratamiento del OSAS puede incluir la administración de medicamentos para reducir la obstrucción de la vía aérea, así como el uso de dispositivos de apnea del sueño, como el dispositivo CPAP (Continuous Positive Airway Pressure). En algunos casos, puede ser necesario realizar cirugía para corregir las anomalías estructurales que contribuyen al OSAS. 

Los resultados de este estudio sugieren que un enfoque integrado puede ser efectivo para el manejo del OSAS, y se recomienda continuar investigando estos tratamientos para mejorar la calidad de vida de los pacientes con OSAS.