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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.[1] 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 sclerodactyly 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

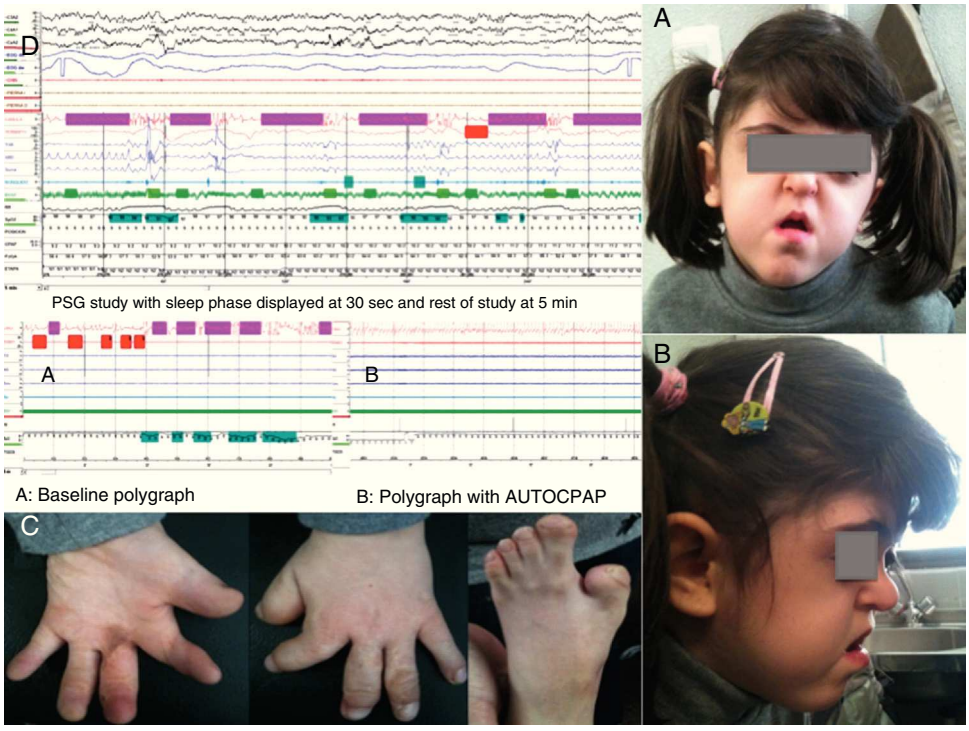

Fig. 1. (A and B) Characteristic facies of Apert syndrome with facial hypoplasia. (C) Syndactyly and sclerodactyly. (D) Patient's baseline polysomnography showing predominance of obstructive apneas and recording from autoCPAP connected to the polygraph flow channels.

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examination and even fell asleep on the chair in the consulting room. A diagnostic polysomnography showed: recording time 534 min (m), total sleep time 458.5 m, sleep latency 0.5 m, sleep efficiency 85.9%, N1 21.2%, N2 73%, N3 5.9%, REM 0%, arousal index 73.2 h^{-1}, 669 respiratory events recorded, with 331 predominantly obstructive apneas, apnea and hypopnea index 87.5 h^{-1}, 500 snores (10.4%), mean SaO_2 86%, minimum SaO_2 64% and desaturation index 96.1 h^{-1}. The following day CPAP was initiated at 5 cmH_2O with an oronasal mask, and an appointment was made one week later for adaptation and titration with the hospital auto-CPAP (REMstar Auto Intl Respironics®), connected to the polygraph flow channels, obtaining an apnea and hypopnea index of 5 h^{-1} with pressures between 10 and 15 cmH_2O, with complete resolution of snoring, and a 90th percentile of 14 cmH_2O (Fig. 1D).

She commenced treatment with an auto-CPAP, with an oronasal mask due to the high pressures required. Adaptation and compliance was very good. Three months later, the patient showed great clinical improvement: she no longer had daytime sleepiness and was active, able to talk and attend school practically normally, changing the CPAP interface to a nasal mask.

Craniosynostosis can be classified as isolated or syndromic. Within the syndromic presentations, the most common and well-known are Crouzon, Saethre-Chotzen, Pfeiffer and Muenke syndrome, and Crouzon syndrome with ancanthosis nigricans.[1] They are caused by a mutation in fibroblast growth factors during the formation of the gametes and alterations in the FGRFR1 and FGRF42 genes in patients with Crouzon, Apert and Pfeiffer syndromes, the TWIST gene in Saethre-Chotzen syndrome and the FGFR gene in Muenke syndrome.[1] Transmission is autosomal dominant, but sporadic mutations exist in non-affected parents.[1] The incidence is 1.2 per 100 000 live births. Apert syndrome is characterized by premature closure of the cranial sutures in a pointed shape, deforming the facial architecture and subsequently producing functional changes with a wide spectrum of clinical variability.[1,4] Although it has not been studied in depth, approximately 40% of cases develop OSAS, mainly due to midface hypoplasia,[2,3] but it may also be associated with changes in the laryngopharynx or larynx, tracheobronchomalacia and other abnormalities which contribute to OSAS.[5] If left untreated, OSAS may lead to sleep fragmentation, recurrent infections, growth and developmental delay, altered cognitive functions, cor pulmonale or sudden death,[3] so a polysomnography study[4] must be carried out, as must endoscopy of the airways, since obstruction has been observed at several levels.[2,3,5,6] Treatment of moderate to severe OSAS in patients with craniosynostosis is complicated and difficult, because CPAP not only must be administered at very high pressures, as in our case, but must also be initiated at an early age, and will probably have to be for life. In addition, adenotonsillectomy may be required, along with orthodontics and maxillary surgery,[1,2,3,6] all of which must be adapted in line with the patient's growth.

References

1. Jong T, Bannink N, Bredero-Boelhouwer H, van Veelen M, Bartels N, Hoeve L, et al. Long-term functional outcome in 167 patients with syndromic craneosynostosis; defining a syndrome-specific risk profile. J Plast Reconstr Aesthet Surg. 2010;63:1635–41.
2. Hein A, Schweitzer T, Strabburg HM, Wurm M. Diagnosis and therapy of obstructive sleep apnea syndrome in children with premature craniosynostosis syndromes. Klin Padiatr. 2011;223:424–9.
3. Hans L, Pijpers M, Joosten K. OSAS in craniofacial syndromes: an unsolved problem. Int J Ped Otorrhinolatyngol. 2003;6751:S111–3.
4. Bannink N, Nout E, Wolvius E, Hoeve L, Joosten K, Mathijssen I. Obstructive sleep apnea in children with syndromic craniosynostosis: long-term respiratory outcome of midface advancement. IntJ Oral Maxillofac Surg. 2010;39:115–21.
5. Lyons M, Vlastarakos PV, Nikolopoulos TP. Congenital and acquired developmental problems of the upper airway in newborns and infants. Early Hum Dev. 2012;88:951–5.
6. Randhawa PS, Ahmed J, Nouraei SR, Wyatt ME. Impact of long-term nasopharyngeal airway on health-related quality of life of children with obstructive sleep apnea caused by syndromic craniosynostosis. J Craneofac Surg. 2011;22:125–8.

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Care Mechanisms to Avoid Readmission of Patients With Chronic Obstructive Pulmonar Disease☆

Acerca de los mecanismos asistenciales para evitar el reingreso de los pacientes con enfermedad pulmonar obstructiva crónica

Dear Editor,

I read with interest the article by Jurado Gámez et al.[1] in which the authors suggest that early home monitoring does not decrease the readmission rate during the first month of patients discharged from hospital after COPD exacerbation. These results may be very discouraging, since this is a highly prevalent disease that uses up a large proportion of hospital resources.

However, I would like to highlight that the small size of the sample analyzed by the authors does not rule out a beta error, i.e. stating that are no differences between the groups when in reality there are. Thus, according to the data provided by the authors, the difference between a readmission rate of 16% in the intervention group and 20% in the control group gives an odds ratio (OR) for readmission in patients with intervention of 0.74, although the 95% confidence interval (0.21–2.62) is too wide to be considered statistically significant. In addition, the multivariate model developed by the authors to determine the profile of patients with a greater risk of readmission cannot be considered consistent, since 12 events (hospitalizations) are clearly insufficient for making an approximation with this model. In any case, taking into account these observations and the severity of the patients included (over 50% with GOLD stages 3 and 4 and a mean pO_2 of 51 mmHg on discharge), this is an avenue that must not be considered closed based on the results of this study.

In this respect, I think it would be very interesting to explore ways of avoiding not only hospital admission in these patients, but also emergency room visits. This is an aspect which is not examined in the report by Jurado Gámez et al. The emergency departments in Spain are frequently overstretched,[2] and patients who come in are faced with long waits, both before being seen and before being given a hospital bed, if needed. All this unquestionably raises issues regarding clinical safety.[3,4] In this situation, decisions taken in the emergency room, especially if they are to discharge patients, are not always correct and involve risks that should be avoided. It would clearly be of interest to study whether post-emergency room dis-

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