ORIGINAL ARTICLE

Crouzon Syndrome and Obstructive Sleep Apnea

Preeti Devnani[1], Raju Khubchandani[2], Rahul Motwani[3] Ankit Bajpai[4], Shrinivas Desai[4]

[1]Department of Neurology and Neurophysiology, Jaslok Hospital and Research Centre, Mumbai, India.
[2]Departmentof Pediatrics, Jaslok Hospital and Research Centre, Mumbai, India.
[3]Sleep Disorders Clinic, Mumbai, India
[4]Department of Radiology, Jaslok Hospital and Research Centre, Mumbai, India.

DOI No:10.5958/0974-0155.2014.01105.X *Indian J Sleep Med 2014; 9.2, 59-61*

Abstract

In children with Crouzon syndrome, craniofacial dysmorphism predisposes them to sleep-related breathing disorders. In this review, we have discussed the clinical manifestation, consequences, and need for management with both temporary and definitive measures of Obstructive sleep apnea syndrome (OSAS).

Introduction

Crouzon syndrome is a rare genetic disorder with autosomal dominant inheritance and has a prevalence of 1 in 25,000 live births[1]. It is caused by a mutation in the fibroblast growth factor receptor 2 (FGFR2)[2] gene. This syndrome is characterized by premature closing of calvarial and cranial base sutures as well as the orbit and maxillary complex[3]. Premature cranial suture closure is the most common skull abnormality[4.] Optic disc edema and proptosis are among the most common ocular findings. Orofacial manifestations include maxillary hypoplasia, external nasal deformity, and prognathism[3]. Resultant airway compromise of both upper and lower airways can manifest as sleep apnea (sleep-related breathing disorder).

Sleep nasendoscopy and magnetic resonance imaging have shown choanal stenosis, maxillary hypoplasia, posteriorly displaced tongue, lengthened soft palate, and adenoid tissues contributing to upper airways obstruction[5].

Address for correspondence
Dr. Preeti Devnani
Department of Neurology and Neurophysiology, Jaslok Hospital and Research Centre, Mumbai.
Email:add-drdevnani@gmail.com

Obstructive sleep apnea syndrome (OSAS) can present with snoring, disturbed and fragmented sleep patterns, and excessive daytime sleepiness. Consequences of sleep apnea include neurologic manifestations such as psychomotor vigilance, impairment of memory and executive functioning, daytime inattention, and behavioral difficulties. In addition, sleep apnea is linked with increased inflammation and impaired immunological response[6].

Radiological findings

There is an abnormal development of the first branchial arch. Radiologically, the presentation can be varied and can be classified as craniofacial and noncraniofacial manifestations.

The most notable characteristic of Crouzon syndrome is cranial synostosis[7]. Brachycephaly is the most common among the various craniosynostoses, which results from premature fusion of coronal sutures. Other anomalies such as shallow orbits and widely spaced orbital axes resulting in proptosis and hypertelorism are noted. Low set ears and external auditory canal defects may lead to conductive hearing loss.

Midfacial anomalies including maxillary hypoplasia[8], which may result from non-pneumatization or premature

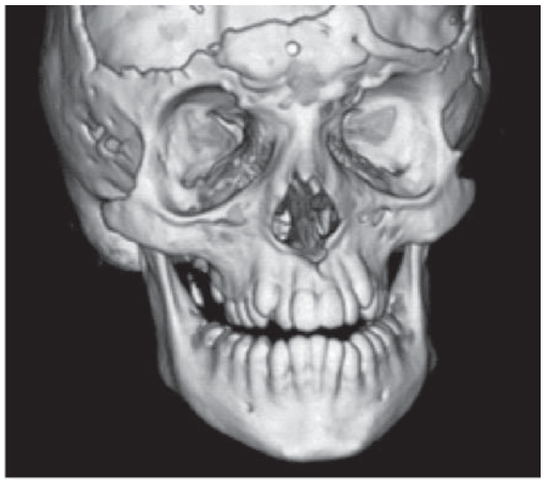

Figure 1: Three-dimension alreconstruction image of hypo plastic maxilla and crowding of teeth

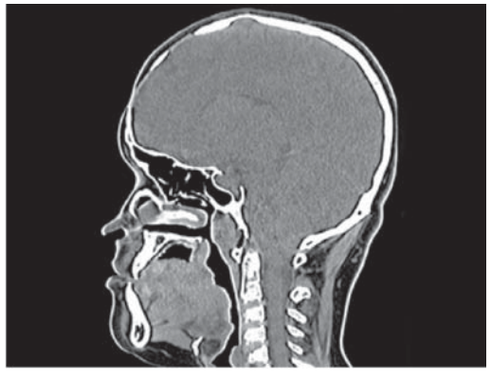

Figure 2: Compromise of the nasopharyngeal air way. Note the prominent mandibular protuberance and concave midfacial region

sutural fusion, give a concave appearance to the face and beaklike nose (*psittichorhina*) (Fig. 1). High-arched and cleft palates are also known to occur.

Hypoplastic maxilla in turn causes dental defects in the form of crowding of teeth, hypodontia, and bilateral posterior crossbite. Mandibular prognathism occurs because of relatively normal growth of the mandible over hypoplastic maxilla (Fig 2).

Other noncraniofacial anomalies include stylohyoid ligament calcification, cervical spine abnormalities, elbow malformations, hand deformities, and skin abnormalities such as *Acanthosis nigricans*[8]

Case reports

We present two cases. The first is of a 7-year-old child with Crouzon syndrome, snoring with excessive daytime somnolence, h/o of tonsillectomy, and a body mass index (BMI) of 26.5. On the polysomnogram, the total respiratory disturbance index (RDI) was 18.8 per hour with a non-rapid eye moment (NREM) RDI of 20.0 per hour and rapid eye moment (REM) RDI of 7.3 per hour. The lowest SaO_2 recorded during the study was 63%. These findings are consistent with those of moderate OSAS with significant oxygen desaturation.

The second case is of a 10-year-old child with a medical history of Crouzon syndrome after adenotonsillectomy with a BMI of 17.3. On the polysomnogram, the total RDI was 6.1 per hour with an NREM RDI of 7.9 per hour and REM RDI of 0.6 per hour. The lowest SaO_2 recorded during the study was 88%. These findings are consistent with those of mild OSAS, which was predominantly position related and was worse in the supine position.

Management of OSAS

Management of OSAS can be addressed in a temporary and definitive stepwise approach as given in the following sections.

Temporary management: Adenotonsillectomy is the first line approach followed by nasal continuous positive airway pressure (CPAP) to improve symptoms of OSAS. Adenotonsillectomy reduces symptoms and improves secondary outcomes of behavior, quality of life, and polysomnographic findings in children (N = 464), thus providing evidence of beneficial effects of early surgery. In a study, a family (father and two sons) with Crouzon syndrome, offspring aged 1 and 3 years were found to have significant OSAS with failure to thrive. Nasal CPAP was used as a treatment modality[3]. This treatment improved quality of life of these children.

Definitive treatment: Treatment of children with Crouzon syndrome is complex and is aimed at correcting the skull and midfacial abnormalities and therefore helps in treating OSAS. A stepwise surgical approach is recommended. Serial skull surgeries are frequently performed in the first 18 months of life. LeFort III operation addresses the midfacial region and is typically performed around 6–8 years of age with a goal to ensure an optimum airway. Tracheostomy, if performed earlier, can then be reversed.

Conclusion

OSAS adversely affects the quality of life of patients with Crouzon syndrome. Earlier complication identification and a stepwise temporary and definitive approach can help limit the consequences of sleep apnea.

The second case child underwent craniectomy to restore sutures at the age of 12 months and LeFort III osteotomy at the age of 10 years for better resolution in sleep apnea.

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