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CASE REPORTS

Severe Obstructive Sleep Apnea in a Child With Goldenhar Syndrome and Nasal Obstruction

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A 4-year-old boy with Goldenhar syndrome and severe obstructive sleep apnea does not tolerate nasal continuous positive airway pressure. Evaluation with imaging showed nasal obstruction with an inverted tooth. This case illustrates the importance of personalized evaluation and treatment plan in a child with Goldenhar syndrome.

Keywords: CPAP, Goldenhar, OSA, pediatrics

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REPORT OF CASE

A 4-year-old boy with Goldenhar syndrome was referred for management of severe obstructive sleep apnea (OSA). The child was initially referred for a sleep study due to loud snoring. The diagnostic sleep study showed an obstructive apnea-hypopnea index (AHI) of 27.6 events/h, central apnea index of zero, and a saturation nadir of 87%. Rapid eye movement (REM) sleep was not seen. REM sleep was achieved during the titration with continuous positive airway pressure (CPAP). The child had multiple obstructive events during REM with an oxygen saturation nadir of 77% while using CPAP at 4–5 cm H_2O. CPAP was increased to a pressure of 8 cm H_2O with a residual AHI of 6.9 and an improvement in saturation to a nadir of 93% (**Table 1**). There was no evidence of hypoventilation. CPAP trial with a nasal mask was recommended, but resulted in poor tolerability and frequent nocturnal awakenings with the patient pulling the mask off. A full-face mask worked better for the child. His medical history included complications from Goldenhar syndrome (cleft lip and palate, microtia with hearing loss), and gastrostomy tube feeding. Surgical history was significant for cleft lip and palate repair. Review of systems was positive for significant nasal congestion.

Physical Examination

The boy was alert and cooperative. The vital signs were within normal limits. He had facial asymmetry (**Figure 1**): left microtia with evidence of a surgically repaired cleft lip and palate. Nasal examination showed a deviated septum with bilateral nasal obstruction. Airway examination showed Mallampati score of class II with tonsil size of 2+. The remainder of the examination was within normal limits.

Radiologic Findings

Computed tomography of the head and neck (**Figure 2**) showed bilateral maxillary clefting yielding a central pre-maxilla, which contained multiple teeth and tooth follicles. At least one tooth was completely inverted and growing into the anterior aspect of the right nasal cavity. The nasal septum was deviated to the left by 5 mm from midline. Adenoid and tonsillar hypertrophy were seen (**Figure 3**).

Outcome

The patient underwent adenotonsillectomy with removal of the obstructing tooth. A follow-up sleep study showed only mild residual obstructive sleep apnea without gas exchange abnormality. The residual obstructive AHI was 1.3 events/h and saturation nadir 91%. Central apnea index was 1.5. The family opted for watchful waiting at this point.

DISCUSSION

Goldenhar syndrome is a complex congenital disorder involving the first and second branchial arches and characterized by oculoauriculovertebral dysplasia and hemifacial microsomia. In 85% of cases, the condition manifests with underdevelopment of one side of the face, including eye (microphthalmia), ear (microtia, hearing loss), and jaw (micrognathia) abnormalities (**Figure 1**). Findings are bilateral in 15% of cases. Spinal abnormalities (scoliosis and cervical fusion) are commonly seen. A variety of comorbid conditions are associated with this disorder, including cardiac and neurologic abnormalities. Prevalence is estimated at 1 out of every 5,600 live births, with 2% familial occurrence and a 3:2 male to female ratio.[1]

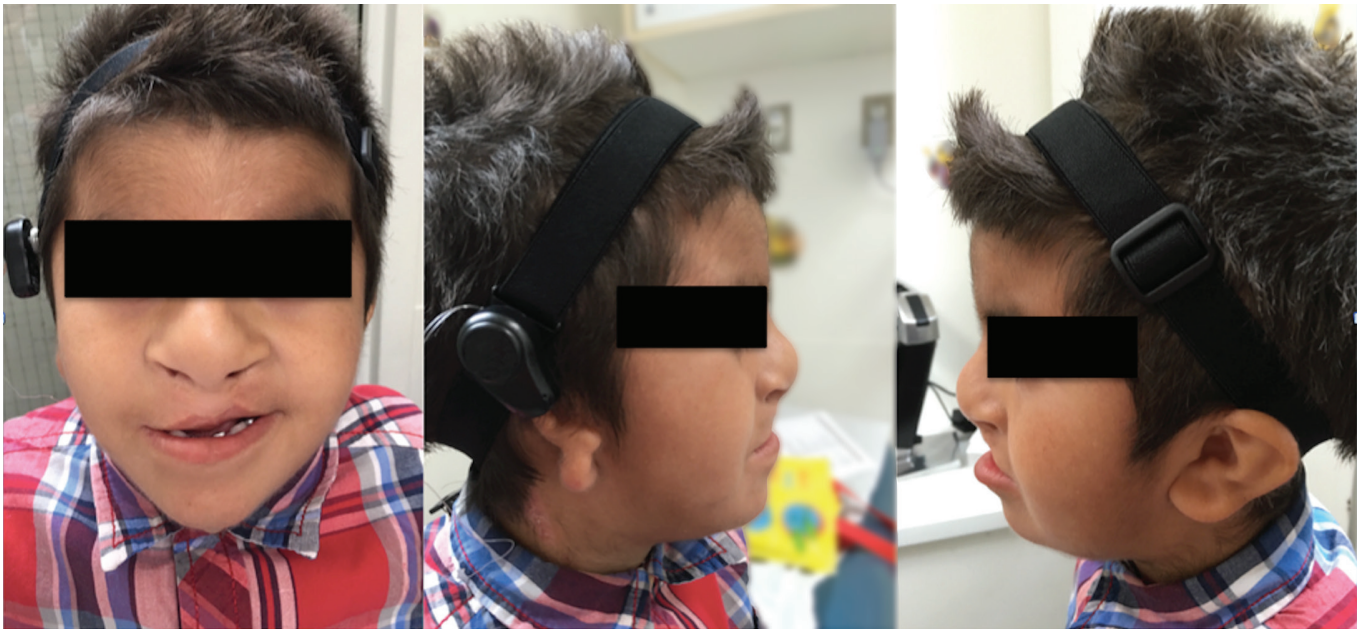
OSA has been reported in patients with Goldenhar syndrome. A retrospective review of 62 patients with unilateral craniofacial macrosomia revealed a prevalence of 11.2%; this is double the incidence of OSA in the general pediatric population.[2] A systematic review of the literature on children with craniofacial microsomia showed prevalence of OSA ranging from 7% to 67%.[3]

Table 1—CPAP titration results.

Level (cm H_2O)	TIB (h:min:s)	REM (h:min:s)	Non-REM (h:min:s)	Obstructive Apnea	Central Apnea	Mixed Apnea	Total Hypopnea	AHI	RDI	Minimum SpO_2%	Mean SpO_2%
CPAP 4	0:26:04	0:03:48	0:20:30	1	0	0	17	44.5	44.5	88.0	96.0
CPAP 5	2:56:19	0:48:42	2:05:07	0	1	0	43	15.2	15.2	77.0	97.1
CPAP 6	0:51:19	0:07:26	0:43:53	0	0	0	18	21.0	21.0	90.0	97.2
CPAP 7	1:17:36	0:30:06	0:47:30	0	2	0	20	17.0	17.0	83.0	97.1
CPAP 8	1:44:54	0:51:58	0:52:57	0	2	0	10	6.9	6.9	93.0	97.8

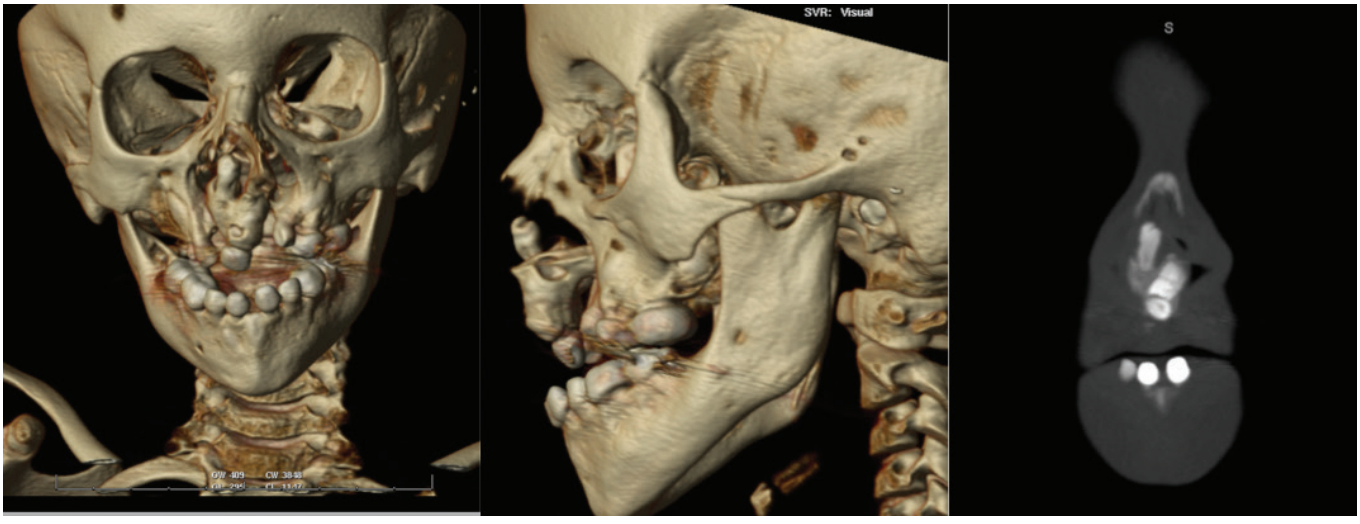
AHI = apnea-hypopnea index, CPAP = continuous positive airway pressure, RDI = respiratory disturbance index, REM = rapid eye movement, SpO_2 = peripheral oxygen saturation, TIB = time in bed.

Figure 1—Patient with Goldenhar syndrome.



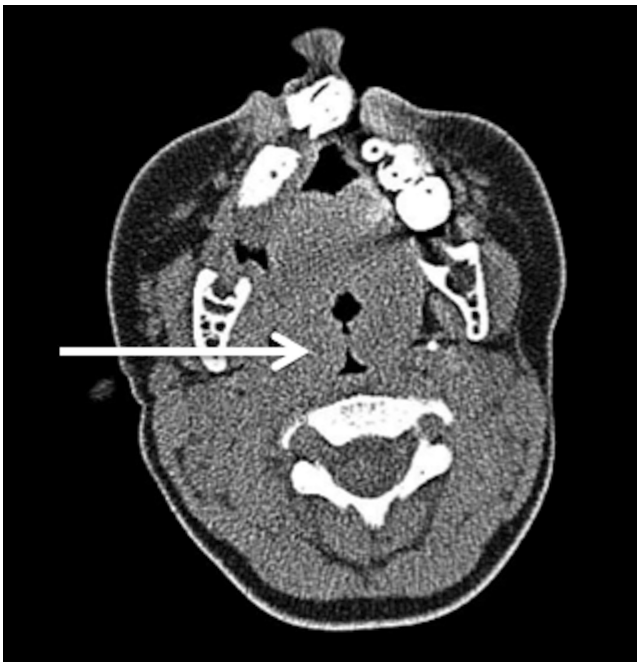
Left photograph: front view shows facial asymmetry; center photograph: the right lateral view shows microtia; right photograph: left lateral view shows maxillary hypoplasia.

Figure 2—Three-dimensional computed tomography images of the head.



Left image: front view shows clefting of the palate, inverted tooth obstructing the nasal passages, and deviated septum; center image: left lateral view shows maxillary clefting and deformity; right image: nasal obstruction with an inverted tooth is shown.

Figure 3—Computed tomography transverse image shows tonsillar hypertrophy with airway obstruction (arrow).



In another retrospective study of nine patients with Goldenhar syndrome, severe OSA was found in 4 patients, isolated hypoxemia in 1, and hypercapnia in 5. Three of the 5 patients with hypercapnia had neurologic abnormalities.[4]

The pathophysiology of sleep-disordered breathing in Goldenhar syndrome may be secondary to maxillomandibular hypoplasia, glossoptosis, and abnormal control of breathing in patients with neurological compromise.[4] Other anatomic abnormalities, as shown in our patient, can contribute to narrowing of the nasal and oropharyngeal airway.

Management of patients with Goldenhar syndrome should include a multidisciplinary team including otolaryngology staff, sleep physician, orthopedic surgeon, plastic surgeon, nutritionist, and pediatrician. Options for treatment include adenotonsillectomy, mandibular distraction, maxillary advancement, CPAP, and tracheostomy.[3] Due to the rarity and heterogeneity of this syndrome, there is little evidence with which to standardize the management of OSA in children with Goldenhar syndrome and studies comparing treatment modalities are lacking.

The complete nasal obstruction secondary to the aberrant growth of the maxillary incisor certainly contributes to this child's OSA as well as his inability to tolerate nasal CPAP. An inverted tooth obstructing the nasal airway is an uncommon source of nasal obstruction, although it should be on the differential for an obstructing mass of the anterior nasal cavity in a child with an ipsilateral or bilateral cleft lip and alveolus. Another possible, yet uncommon, source of nasal obstruction in the same patient could be the instability and migration of the premaxillary segment into the nose, which may occur in the setting of surgical manipulation of that segment during repair of a bilateral cleft lip.

The obstruction in our patient was multifactorial. Adenotonsillectomy was performed, as it is the first-line treatment for OSA in children. The success rate for adenotonsillectomy varies in children. The tooth removal was done simultaneously, because it is a simple procedure with minor risks. The skeletal abnormalities including micrognathia and septal deviation place our patient at higher risk for worsening obstruction in the future. Close follow-up and other surgical options (mandibular distraction) should be considered.

Children with Goldenhar syndrome and sleep-related complaints should be evaluated for early diagnosis and treatment of sleep-disordered breathing.

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