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Clinical Manifestations of Choanal Atresia in Infancy

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● **The clinical features of 41 infants with various types of nasal obstruction were reviewed to study the correlation between the degree of nasal obstruction and clinical manifestations. Twenty-one subjects had bilateral choanal atresia/stenosis; 12 had unilateral choanal atresia/stenosis, and eight who were referred with suspected choanal atresia had a simple inflammatory nasal obstruction. Patients with bilateral choanal obstruction and patients with unilateral choanal obstruction or rhinitis showed no differences in clinical picture or in referral age. Many infants with either unilateral or bilateral choanal obstruction had one or more symptoms not fully relieved after surgical repair, although the choanae were widely patent. At long-term follow-up the surviving patients showed spontaneous recovery and good nasal respiration. Overall, five patients died. Since the common syndrome appeared to be related to a dysfunction of the autonomic nervous system, we conclude that any type of nasal obstruction may exacerbate or precipitate an underlying maturational autonomic disturbance.**

INDEX WORDS: Choanal atresia; sudden infant death syndrome; sleep-apnea syndrome; gastroesophageal reflux; vagal bradycardia.

IT IS WELL ESTABLISHED that infants with bilateral choanal atresia who are unable to breathe through the mouth may succumb to asphyxia soon after birth; those who are able to breathe through the mouth will experience an intermittent dyspnea relieved by crying ("cyclic dyspnea") or a "dyspnea while suckling."[1,2] However, Medovy and Beckman did point out that of nearly 300 collected cases of congenital obstruction of choanae, 46 patients with untreated bilateral choanal atresia were first recognized between the age of 20 and 45 years.[3]

Unilateral choanal atresia causes symptoms that are not well defined and, as a rule, is not diagnosed until later life.[1,2] Cozzi and Pierro have reported that infants with unilateral or bilateral choanal obstruction have clinical manifestations similar to the glossoptosic syndrome described by Pierre Robin in infants with micrognathia.[4] In addition, some infants referred with suspected choanal atresia had a simple inflammatory nasal obstruction associated with a similar clinical picture.[4]

Awareness by pediatricians on the staff of our hospital of the possibility that one or more features of glossoptosis-apnea syndrome may be associated with an upper airway obstruction has resulted in an increasing referral to us over the past 5 years. This study is a review of all our infants with a diagnosis of nasal obstruction.

MATERIALS AND METHODS

We have studied the case notes of 41 patients seen at our hospital who had a final diagnosis of nasal obstruction. Of 20 patients admitted over 12 years prior to 1982, ten had bilateral choanal atresia/stenosis (BCA), six unilateral choanal atresia/stenosis (UCA), and four a simple rhinitis. In addition, nine subjects with bilateral choanal atresia, two with bilateral choanal stenosis, two with unilateral choanal atresia, four with unilateral choanal stenosis, and four with rhinitis were admitted between January 1982 and December 1986.

We have previously reported our diagnostic criteria and definitions of clinical and laboratory findings.[4] In the present study we have classified as reflex apnea or bradycardia all apneic or bradycardic episodes observed "during feeding" or "after feeding" or "after pharyngeal suctioning." All patients with an anatomic obstruction underwent surgery at the time of diagnosis. We removed the choanal obstruction through a transpalatal approach in the first two infants and through a transnasal approach in the others. All patients underwent stenting for a period of time as long as was necessary to avoid narrowing of the choanae due to scarring. All surviving patients were followed on a regular basis.

The differences in the distribution of clinical features and the complications among the 21 patients with BCA and the 20 with UCA or rhinitis were tested by means of χ^2 test applying Yate's correction.

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RESULTS

Sex Ratio and Gestational Age

The entire series includes 21 patients with BCA (12 female), 12 with UCA (five female), and eight with rhinitis (four female). The gestational age was known in 38 patients. Eleven babies (29%) were born after a gestation of 37 weeks or less (five with BCA, four with UCA, and two with rhinitis). Three preterm (two with UCA and one with rhinitis) and seven full-term babies (five with BCA, one with UCA, and one with rhinitis) had a birth weight that was below the tenth percentile.

Age of Referral

Twelve infants with BCA, six with UCA, and seven with rhinitis were admitted during the first month of life. Eighty-three percent of all patients were admitted during the first 3 months of life. Five patients with BCA and two with UCA were admitted between 6 months and 3 years of age.

Main Clinical Features

We found no significant differences in distribution of the main clinical manifestations and no correlation between the degree of nasal obstruction and the severity of the symptomatology (Table 1). All but one with UCA had an inspiratory dyspnea characterized by a discrepancy between severity of respiratory distress and lung lesions clinically or radiologically detectable. The patients had long periods of poor air entry associated with noisy respiration. Twenty-nine infants had short episodes of complete upper airway obstruction (glossoptosis-apnea) with vigorous inspiratory efforts not associated with air entry. The infants reestablished a patent airway by crying (cyclic dyspnea). Occasional opisthotonos was sometimes observed.

Table 1. Distribution of Main Clinical Features of 21 Infants With Bilateral Choanal Obstruction and 20 Infants With Unilateral Choanal Obstruction or Rhinitis Associated With Glossoptosis-Apnea Syndrome

	Bilateral Nasal Obstruction		Unilateral or Inflammatory Nasal Obstruction	
	No.	%	No.	%
Respiratory problems				
Dyspnea	21	100	19	95
Glossoptosis-apnea	16	76	13	65
Noisy respiration	13	62	18	90
Expiratory wheezing	10	48	8	40
Opisthotonus	3	14	7	35
Feeding problems				
Oropharyngeal dysphagia	16	76	12	60
Vomiting	9	43	13	65
Reflex bradycardia	3	14	2	10
Reflex apnea	2	10	8	40
Abdominal distension	0	0	5	25
Hyperhydrosis	4	19	4	20
Sialorrhea	3	14	5	25
Hyperthermia	2	10	4	20

Eighteen patients also had expiratory dyspnea with wheezing or audible grunting. Two subjects, referred for minor surgical procedures, had a history of "recurrent bronchiolitis" or "asthmatic attacks" during the night. Passage of nasal catheters revealed choanal stenosis. Dilatations were followed by marked improvement. Factors precipitating or exacerbating respiratory problems included supine position, crying, respiratory infections, sleeping, and anesthesia.

Sucking and/or swallowing difficulties occurred in 28 patients and vomiting episodes in 22. Four infants with BCA, five with UCA, and four with rhinitis had episodes of reflex bradycardia and/or apnea. A full-term baby with choanal stenosis had recurrent episodes of tachycardia associated with transient glycosuria. This patient also had severe dysphagia, vomiting, and recurrent reflex apneas during gavage or after vomiting. A feeding gastrostomy and fundoplication avoided vomiting and apneas.

Excessive salivation and sweating (mainly during sleeping), and defective temperature regulation with periodic fever were slightly more frequent in infants with a minor degree of nasal obstruction.

Main Complications

When the dyspnea was more severe, cyanosis was seen in the perioral region and the nails of the fingers and toes in 16 with BCA and 14 with UCA or rhinitis. In addition, ten had severe cyanotic attacks during crying, sometimes followed by loss of consciousness and brief seizure.

PO_2 and PCO_2 were measured in 27 patients; eight with BCA and six with UCA or rhinitis had hypoxemia that in many instances was not associated with hypercapnia and was not relieved by oxygen administration. Chronic hypoxemia and/or persistent upper airway obstruction and/or feeding problems were associated with failure to thrive in 11 with BCA and eight with UCA or rhinitis. Of the 18 patients with wheezing, six with BCA and two with UCA had a deformity of the sternum and/or Harrison's groove that improved or disappeared after regression of asthmatic-like problems.

One patient with BCA and one with UCA were microcephalic while two with UCA and one with rhinitis had a mild mental and motor retardation probably linked to severe prematurity. In two subjects with BCA and two with rhinitis, the brain damage was judged to be secondary to asphyxia.

Two newborn babies with BCA had severe neonatal asphyxia. One died soon after admission and before surgery. The other had recurrent episodes of bradycardia and/or cyanotic attacks after anesthesia; a sudden cardiorespiratory arrest was followed by an irreversible bradycardia and cardiac arrest. One infant with unilateral choanal atresia was found dead in his cot; autopsy revealed no plausible cause of death. Another infant with microcephalus and unilateral choanal stenosis died at home at the age of 3 months during a severe cyanotic attack.

Finally, a premature baby with BCA had persistent respiratory and feeding problems after surgery although the choanae were widely patent. She also had recurrent episodes of bradycardia and two sudden cardiorespiratory arrests during anesthesia. She developed chronic hypoxemia and cor pulmonale. Nearly all symptoms disappeared by the age of 2 years. This patient then developed bouts of noninfectious fever and died elsewhere during an episode of uncontrolled hyperthermia. Cor pulmonale was diagnosed in two other infants with rhinitis.

Evaluation After Surgery

Twenty infants with BCA and 12 with UCA underwent surgical removal of the anatomic obstruction. One infant with BCA and one with UCA died during the postoperative period. Thirty patients were followed on a regular basis for 3 months to 17 years after surgical repair. Seven patients with BCA and four with UCA became symptomless after surgery. One patient with microcephaly and UCA died at home during the last of a series of recurring cyanotic attacks. The other 18 infants showed a marked improvement in their symptoms after surgery. However, in infants with either BCA or UCA, one or more symptoms were not relieved although the choanae were most often widely patent. The main triggering factor was sleeping in the supine position, respiratory tract infections, crying, and exercise. The patients who manifested the most severe symptoms after surgery included nine infants born prematurely and the surviving infant with microcephalus. A gradual and spontaneous improvement of symptomatology was observed after the first year of life. Some of the preterm infants and the patient with microcephalus had persistent symptoms for a long period of time. All surviving patients showed nearly complete recovery and good nasal air flow bilaterally at follow-up.

DISCUSSION

Clinical[5] and radiologic[6,7] observations indicate that in infants with bilateral choanal obstruction the main pathophysiologic manifestation is a recurrent functional airway obstruction, mainly due to a backwards displacement of the tongue. Measurement of intrathoracic pressure during strenuous inspiratory efforts led to the conviction that strong negative pressures and the consequent pharyngeal vacuum play an important role in the pathogenesis of glossoptosis-apnea.[5,6]

In the present clinical study we found that about 50% of all patients with symptomatic nasal obstruction had only a minor degree or an absence of anatomic obstruction associated with a clinical picture not different from that of infants with bilateral choanal atresia. The lack of correlation between degree of nasal obstruction and severity of symptomatology gives further support to the concept that neuromuscular dysfunction of genioglossus may play a role in the pathogenic mechanism of glossoptotic pharyngeal obstruction.[4]

The persistent symptomatology after removal of anatomic obstruction indicates that neuromuscular dysfunction is present also in some infants with BCA. Factors that increase the negative intrathoracic pressure (respiratory tract infections, crying, exercise, etc) and decrease the upper airway dilating muscle activity (sleeping, sedatives, anesthesia, etc) may cause an upper airway functional obstruction even in the absence of an anatomic narrowing.

Many clinical manifestations that we found are remarkably similar to those described in infants with vagal bradycardia.[8] A parasympathetic over-activity affecting one or more of the clinical sites of vagal activity may be responsible for pharyngeal dysphagia, asthmatic-like problems, gastroesophageal reflux (GER), pylorospasm, reflex bradycardia or apnea after swallowing or GER, and inability to maintain body temperature.[8] In our patients we found additional evidence of autonomic disturbances; hyperhydrosis, sialorrhea, abdominal distension, reflex tachycardia, and transient glycosuria clearly indicate autonomic dysfunction.

Animal studies have provided evidence that the vagus plays a role in the maintenance of upper airway patency.[9-12] The efferent hypoglossal nerve discharge is characterized by a rapid rise to peak activity early in inspiration, followed by gradually decreasing activity due to the inhibition of motor neuron activity by phasic vagal volume feedback. Since in infancy the afferent limb of the vagal airway feedback system plays an important role in the regulation of respiratory center output, we speculate that a vagal dysfunction may also contribute to an upper airway instability in the young human infant.

Recent investigations have altered the previous concept that the newborn infant is an obligatory nasal breather since it has been demonstrated that he or she

may use the oral airway by detaching the palate from the tongue both spontaneously and in response to nasal obstruction.[13,14] Premature infants acquire the ability to use the oral route of breathing between 31 and 34 weeks of postconceptional age: this ability increases with advancing postnatal maturation.[15] The abnormal response of some infants to a simple inflammatory nasal obstruction may be related to a genioglossus neuromuscular dysfunction.[4]

The glossoptosis-apnea syndrome provides a model of sudden infant death that may be related to sudden infant death syndrome.[4-6,16,17] Preterm infants and sudden infant death syndrome victims have a lower number of myelinated vagus fibers, which gradually increase with postconceptional age.[18] It is conceivable that infants with glossoptosis-apnea syndrome, preterm infants, and sudden infant death syndrome victims have a naturational neural dysfunction affecting particularly the autonomic nervous system.

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