Diagnosis and Therapy for Airway Obstruction in Children With Down Syndrome

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Objectives: To document the causes of upper airway obstruction in a population of children with Down syndrome and to highlight the role of associated comorbidities.

Design and Setting: Review of 23 cases involving children with Down syndrome who were referred for upper airway obstruction over a 2½-year period to the Pediatric Otolaryngology Service of the University of New Mexico, Albuquerque.

Methods: Data on the following variables were obtained: reason for referral, demographics, diagnosis, surgical procedures, complications, and comorbidities.

Results: The children ranged in age from 1 day to 10.2 years (mean age, 1.8 years; median age, 6 months). Thirteen children were male and 10 were female. None of the children had subglottic stenosis. Laryngomalacia was the primary diagnosis in 10 children (43%), 8 of whom were younger than 1 month. Obstructive sleep apnea was the primary diagnosis in 11 children (48%), 8 of whom were older than 2 years. All children with obstructive sleep apnea and 4 children with laryngomalacia had a secondary ear, nose, and throat disorder. Gastroesophageal reflux was a comorbidity in 14 children (61%).

Conclusions: The causes, severity, and presentation of upper airway obstruction in children with Down syndrome are related to the age of the child and to associated comorbidities. The treatment of comorbidities and secondary ear, nose, and throat disorders is an integral component of the surgical management of upper airway obstruction in such cases.


THE INTERPLAY between oto-laryngologic disease and systemic comorbidities in children with Down syndrome has been discussed in a recent report. However, there are few reports that have specifically addressed upper airway obstruction and associated comorbidities in these children. Children with Down syndrome have midface hypoplasia, macroglossia, a narrow nasopharynx, and a shortened palate. These anatomical abnormalities along with generalized hypotonia, an immature immune system, and a tendency to obesity predispose children with Down syndrome to upper airway obstruction. Furthermore, gastroesophageal reflux disease (GERD) and chronic lung disease are also common in these children and may worsen airway problems.

Our goal is to describe the causes of upper airway obstruction in a population of children with Down syndrome, the surgical therapy used to treat this obstruction, and the role of associated comorbidities.

METHODS

A retrospective review of cases involving children with Down syndrome who were referred for upper airway obstruction to the Pediatric Otolaryngology Service at the University of New Mexico Health Sciences Center, Albuquerque, was undertaken. Approval (Human Research Review Committee Approval No. 01-301) was obtained from the institutional review board of the University of New Mexico School of Medicine. All pediatric surgical procedures and inpatient consultations between July 1, 1999, and December 31, 2001, were entered into a spreadsheet (Excel; Microsoft Corp, Redmond, Wash). Children with Down syndrome were identified from this database, and those with a primary diagnosis of upper airway obstruction were included in the study. Primary otolaryngologic diagnosis was defined as the primary disorder underlying the reason for referral. Secondary otolaryngologic diagnosis was defined as a disorder that was revealed by history and physical examination in addition to the primary disorder. A single surgeon (R.B.M.) was responsible for all otolaryngologic procedures performed on children in the study population. The following parameters...
were obtained from the medical records of children included in the study: gestational age; birth weight; reason for referral; number and types of previous surgical procedures; complications; presence and nature of cardiac disease; and presence of other comorbidities, such as GERD and chronic lung disease.

The diagnosis of GERD was based on the results of a radiologic contrast study and clinical findings. Reflux was identified by the presence of contrast medium above the lower esophageal sphincter. In children younger than 6 months, 3 or more episodes of reflux were considered a positive finding. In children older than 6 months, a positive finding was 1 or more episodes of reflux. The severity was defined according to the following classification: mild, the presence of contrast medium confined to the lower third of the esophagus; moderate, up to the middle third of the esophagus; and severe, up to the level of the cricopharyngeus muscle, with dilatation of the esophagus. The clinical findings relevant to GERD included the number of respiratory tract infections, posturing, weight loss, and failure to thrive.

A total of 23 children were included in the study. Ethnicity was classified as Hispanic or Latino, Caucasian, African American, Native American, Asian, or other. The gestational age of the infant at birth was recorded as follows: term, 40 ±2 weeks (mean ±SE); nonterm, younger than 38 weeks; or unknown. Both diagnostic and therapeutic surgical interventions were recorded. Comorbidities were recorded to include the nature of comorbidity, surgical intervention, and outcome. Therefore, a total of 55 fields were maintained for each child. A unique identifier was used for each record so that medical record numbers and names of patients could be deleted from the database to protect the privacy of the patients.

RESULTS

Twenty-three children with Down syndrome and upper airway obstruction were included in the study. The children ranged in age from 1 day to 10.2 years (mean age, 1.8 years; median age, 6 months). Thirteen children were male and 10 were female. The caregivers for 15 children identified themselves as Hispanic or Latino, 5 as Native Americans, 2 as non-Hispanic whites, and 1 as Asian. Fourteen children were full-term neonates and 9 were preterm.

Eleven children (48%) had obstructive sleep apnea, 10 (43%) had laryngomalacia, 1 had tracheomalacia, and 1 had inhaled a foreign body (Table 1). The age range for children with obstructive sleep apnea was 3 months to 10 years. The majority of these children (73%) were older than 2 years. The age range for children with laryngomalacia was 1 day to 2 years. Eight of these children were 1 month or younger.

All children with a primary diagnosis of obstructive sleep apnea had a secondary ear, nose, and throat disorder, most commonly recurrent otitis media (73%). One child with obstructive sleep apnea also had laryngomalacia. A secondary ear, nose, and throat disorder was present in 4 children with laryngomalacia. One child with laryngomalacia also had an unilateral vocal cord palsy. Secondary ear, nose, and throat disorders were present in all children 2 years or older and in 7 (54%) of the children younger than 2 years.

All children in the study population underwent at least 1 diagnostic or therapeutic procedure for upper airway obstruction. The most common diagnostic procedure was flexible laryngoscopy (Table 2). Ten children who underwent flexible laryngoscopy had laryngomalacia, 3 had obstructive sleep apnea, and 1 had tracheomalacia. All 11 children who had a diagnosis of obstructive sleep apnea underwent full-night polysomnography (Table 2).

Eight children underwent bronchoscopy (Table 2). Four of these children had laryngomalacia and episodes of cyanosis. Two children had obstructive sleep apnea and stridor. One child with stridor had no evidence of laryngomalacia but had tracheomalacia diagnosed on bronchoscopy. One child underwent bronchoscopy to remove a foreign body. Nine children had obstructive sleep apnea and no evidence of stridor. These children did not undergo bronchoscopy. The remaining 6 children of the total study population of 23 children had mild symptoms that did not justify bronchoscopy. A tracheotomy was performed in 6 children, 2 of whom had laryngomalacia that had been treated unsuccessfully with laser aryepiglottoplasty. Two other children had GERD and severe aspiration in addition to laryngomalacia. One child had tracheomalacia, and 1 child had obstructive sleep apnea that did not improve after adenotonsillectomy.

Nineteen (83%) of the children in the study population had systemic comorbidities. Gastroesophageal reflux, which was the most common systemic comorbidity, was present in 14 children (61%). Chronic lung disease was present in 13 children (56%), 6 of whom were preterm. Congenital cardiac disease was present in 11 children (48%). Pulmonary hypertension was diagnosed in 7 children (30%). Seventeen children (74%) had 2 or more systemic comorbidities. Four children (17%) had 3 comorbidities, which included GERD, chronic lung disease, and congenital heart disease.

COMMENT

In children with Down syndrome who are younger than 1 month, the combination of laryngomalacia and GERD may lead to upper airway obstruction that presents as stridor. In the present study, this was the most common reason for a neonate with Down syndrome to require a tracheotomy. In children 2 years and older, obstructive sleep apnea is the major cause of upper airway obstruction that presents as sleep-disordered breathing. Gastroesophageal reflux disease was the most common comorbidity in all age groups and in many instances required additional surgical therapy.
Laryngomalacia was described by Holinger as a condition that may lead to stridor in neonates. The disorder is characterized by folding of the epiglottis into an Omega shape, flaccidity of the aryepiglottic folds, and medial movement of the arytenoids on inspiration. Three factors appear to determine whether laryngomalacia leads to stridor in neonates: (1) the extent of the morphological changes, such as the shape of the epiglottis and the degree of infolding of the aryepiglottic folds; (2) the relative flaccidity and hypotonia of the muscles of the supraglottis; and (3) the presence and severity of GERD.

It is likely that children with laryngomalacia who develop stridor represent a heterogeneous group in regard to the cause of the disorder.

Laryngomalacia was present in 10 children (43%) in the present study. Previous studies have shown that laryngomalacia is more common in children with neurologic disorders, particularly hypotonia. Children with Down syndrome have generalized hypotonia, leading to flaccidity of the supraglottis; anatomical changes in the epiglottis, arytenoids, and aryepiglottic folds; and a high prevalence of GERD. It is not surprising that these children have a high prevalence of laryngomalacia that leads to upper airway obstruction and presents as stridor.

Aryepiglottoplasty for laryngomalacia is a safe and effective procedure for children without significant comorbidities. Generalized hypotonia, the high incidence of GERD, and the likelihood of multiple intubations for further surgical procedures make aryepiglottoplasty a questionable procedure in children with Down syndrome. Outcome studies after aryepiglottoplasty in these children have not been reported, to our knowledge.

In the present study, 6 children underwent a tracheotomy. All 6 of them had multiple airway problems, and previous surgical therapy, including aryepiglottoplasty and adenotonsillectomy, had failed in 3 of them. Therefore, tracheotomy was performed only in those children in the study population either in whom previous surgical therapy had failed or who had severe aspiration or tracheomalacia. These children also had severe GERD in addition to the hypotonia and craniofacial abnormalities that characterize Down syndrome. As a consequence, a tracheotomy was the only effective mode of surgical therapy. In the present study, treatment of laryngomalacia involved therapy for GERD and tracheotomy for severe cases. It remains unclear what role aryepiglottoplasty for laryngomalacia may be in such cases.

Obstructive sleep apnea was the most common cause of upper airway obstruction in children with Down syndrome who were older than 2 years. The incidence of obstructive sleep apnea among children in the general population is approximately 3%. Because of macroglossia, mandibular hypoplasia, adenotonsillar hypertrophy, and generalized hypotonia, children with Down syndrome are more likely to have constriction of the pharyngeal airway during inspiration. Marcus et al found sleep-disordered breathing documented by full-night polysomnography in all 53 children with Down syndrome who were enrolled in their study. Levanon et al compared 23 children who had Down syndrome with 13 controls. They found that all children with Down syndrome whose parents reported a sleep disturbance had obstructive sleep apnea that was confirmed by full-night polysomnography. Even children with Down syndrome but without obstructive sleep apnea had significant sleep fragmentation that was manifested by frequent awakenings and arousals. This sleep fragmentation was not present in the control group.

Pediatric obstructive sleep apnea may have several different clinical presentations, including daytime somnolence, behavioral problems, poor school performance, and in younger children, developmental delay and enuresis. Because many of these factors are also associated with Down syndrome, a delay in diagnosis of obstructive sleep apnea in children with Down syndrome is usually the rule. Similarly, assessing improvement in behavior after surgery can be extremely complex.

In the present study, none of the children was diagnosed with subglottic stenosis. A relatively small trachea as well as the need for multiple intubations for surgery put children with Down syndrome at risk for the development of subglottic stenosis. Shott showed that children with Down syndrome should be intubated initially with an endotracheal tube at least 2 sizes smaller than would be used in a child of the same age without Down syndrome. The present findings agree with previous reports that the incidence of subglottic stenosis is low in children with Down syndrome and may reflect increasing awareness of the unique attributes of the airway in these children.

In conclusion, the causes of upper airway obstruction in children with Down syndrome are age related. In children younger than 2 years, the most common cause is laryngomalacia; in those older than 2 years, it is obstructive sleep apnea. The incidence of subglottic stenosis is low. Airway problems in children with Down syndrome rarely occur in isolation. The most common comorbidity in these children is GERD, but both anatomical abnormalities and systemic disease may exacerbate upper airway obstruction. It is not known whether neonates with Down syndrome...
drome progress from laryngomalacia to obstructive sleep apnea as they age. Equally, it is unknown to what extent airway problems worsen developmental delay or whether correction of these problems leads to behavioral improvements. Future research on the pathogenesis and treatment of airway problems in children with Down syndrome may resolve these issues.

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REFERENCES