The CHARGE Association

The Role of Tracheotomy

Gilles Roger, MD; Marie-Paule Morisseau-Durand, MD; Thierry Van Den Abbeele, MD; Richard Nicollas, MD; Jean-Michel Triglia, MD; Philippe Narcy, MD; Veronique Abadie, MD; Yves Manac’h, MD; Erea-Noèl Garabedian, MD

Objectives: To evaluate the need for a tracheotomy and its timing during the evolution of an association of malformations, including coloboma, heart defects, choanal atresia, developmental and growth retardation, genitourinary malformation, and ear anomalies (CHARGE association).

Design: Retrospective study from January 1988 through December 1997.

Setting: Four academic tertiary care centers.

Patients and Methods: Forty-five patients with CHARGE association having at least 3 cardinal malformations (growth retardation excluded) and review of the malformations and respiratory manifestations encountered. All the patients underwent endoscopic exploration on several occasions. We reviewed the nature and the timing of therapeutic interventions performed on the airway.

Results: Two patients died (one patient of septicemia, the other of unknown causes). Abnormalities of blood gas levels and/or sleep were found in 30 patients (67%), were responsible for cardiorespiratory arrest in 9 (20%), and required admission to the intensive care unit in 21 (47%). Pharyngolaryngeal anomalies leading to dyspnea (discoordinate pharyngolaryngomalacia, glossophtosis, retrognathia, laryngeal paralysis, cleft, stenosis, and difficult intubation) were found in 26 patients (58%). Tracheobronchial anomalies (esophagotracheal fistula, esophageal atresia, and tracheomalacia) were present in 18 patients (40%). Resection of the aryepiglottic folds was attempted 3 times, but without success. Tracheotomy was necessary in 13 patients (29%) at a median age of 2.4 months (mean duration, 25 months). Among these infants, the posterior nasal choanae were patent in 10 patients at the time of tracheotomy. Gastroesophageal reflux was encountered in 36 patients (80%). Prolonged enteral feeding was necessary in 21 patients (47%), with gastrostomy in 16 (of whom 9 needed a tracheotomy). These feeding difficulties and airway problems were highly correlated.

Conclusions: We encountered multiple, complicated airway abnormalities. Resection of aryepiglottic folds was inadequate. Often, a tracheotomy could not be avoided in these patients, regardless of choanal patency. Tracheotomy needs to be performed early to avoid hypoxic events. In some selected patients, ventilation using bi-level positive airway pressure may be an alternative.


HALL1 FIRST described and Pagon et al2 later codified an association of malformations, including coloboma, heart defects, choanal atresia, growth or developmental retardation, genitourinary abnormalities, and ear anomalies (with possible deafness) (CHARGE association). Numerous other malformations that are not part of the acronym have been described frequently in these children, and many involve the airways. Indeed, the following have often been reported: retrognathia, glossophtosis, intubation difficulties, esophagotracheal fistula with or without esophageal atresia, stenosis or laryngeal paralysis, and clefts.3-10 A tracheotomy is performed in 10% to 30% of patients.11 Few authors have specifically and analytically studied the obstructive respiratory manifestations in these patients. These obstructive features have often been attributed to the choanal atresia. In our experience, the main abnormalities responsible for dyspnea are situated at the level of the pharyngolarynx and/or trachea. Therefore, we performed a retrospective study of patients with this association to evaluate precisely the severity and the consequences of these airway anomalies.

RESULTS

Forty-five patients received a diagnosis of CHARGE association. The distribution among the 4 hospitals was 16, 15, 11, and
PATIENTS AND METHODS

A retrospective review of all the patients with CHARGE association was performed in 4 pediatric teaching hospitals in France, from January 1988 through December 1997. Three of the hospitals were located in Paris, and 1 hospital was located in Marseille. We collected the data on the various malformations that could be part of the CHARGE association and on the airway endoscopies performed. The endoscopies were performed under general anesthesia, during spontaneous ventilation while awakening from the anesthesia and using a nasolaryngofiberscope, to study the pharyngolaryngeal dynamics, or using rigid optics and/or bronchoscopes and assisted ventilation, to study the subglottic and tracheal morphologic characteristics. The Cotton classification for laryngeal stenosis was used to quantify the degree of pharyngeal, laryngeal, and tracheal obstruction. The percentage of stenosis was defined by the maximal degree of obstruction evaluated during several respiratory cycles. We focused particularly on the malformations or anomalies involving any part of the airway tract and their repercussions as measured by arterial blood gas levels and/or results of sleep recordings. All the interventions performed to improve respiratory function and all the abnormalities involving the digestive system were also noted.

The diagnosis of CHARGE association was based on the presence of at least 3 cardinal malformations (excluding growth retardation) and required that the malformations could not be classified as part of another syndrome of multiple malformations. Patients with trisomy 21 syndrome and with craniosenosis were excluded from the study.

3 patients. There were 23 boys and 22 girls (male-female ratio, 1.04). The mean follow-up was 4.1 years (SD, 25.6 months).

The details regarding the malformations encountered are shown in Table 1. The abnormalities of the upper airway tract are detailed in Table 2, and those of the digestive system, in Table 3.

All the patients underwent evaluation at least once using nasolaryngotraceobronchial endoscopy in the operating room. The details of the therapeutic interventions are shown in Table 4. Only 6 patients (13%) did not have any clinical, endoscopic, or biological abnormality of the upper airway tract. Eight patients (18%) had no other involvement of the airway than choanal atresia. On the other hand, 26 children (58%) presented with at least 3 respiratory anomalies as defined by the criteria in Table 2. Treatment of these respiratory abnormalities included, depending on the case, a variable association of continuous or nocturnal oxygen therapy, prolonged antibiotic treatment, respiratory physiotherapy, nasopharyngeal aspiration, a temporary oropharyngeal airway, and postural physiotherapy. Treatment of gastroesophageal reflux (essentially with cisapride), sometimes associated with tube feeding, was also an important measure to improve breathing by decreasing the number of episodes of aspiration and reducing the laryngeal edema.

A tracheotomy was performed in 13 patients, at a mean age of 3.7 months (median, 2.4 months; SD, 3.1 months) and for a mean duration of 25 months (SD, 18 months). Five children still have a tracheotomy, at a mean age of 20 months. Among the 13 children with a tracheotomy, only 6 (46%) had bilateral choanal atresia (3 had unilateral atresia and 4 had normal choanae). Treatment for choanal atresia was performed in 8 patients before tracheotomy and was successful in 7. Thus, 10 patients (77%) had normal bilateral choanal patency at the time of tracheotomy. One patient (8%) had only 1 permeable choana at the time of tracheotomy. Additional anomalies found in these 13 patients are given in the following tabulation:

<table>
<thead>
<tr>
<th>Anomaly</th>
<th>No. (%) of Patients</th>
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</thead>
<tbody>
<tr>
<td>Bilateral choanal obstruction</td>
<td>2 (15)</td>
</tr>
<tr>
<td>Micrognathia</td>
<td>6 (46)</td>
</tr>
<tr>
<td>Discoordinate pharyngolaryngomalacia</td>
<td>12 (92)</td>
</tr>
<tr>
<td>Including isolated discoordinate pharyngolaryngomalacia</td>
<td>5 (38)</td>
</tr>
<tr>
<td>Left laryngeal palsy</td>
<td>3 (23)</td>
</tr>
<tr>
<td>Subglottic stenosis</td>
<td>3 (23)</td>
</tr>
<tr>
<td>Laryngeal cleft</td>
<td>1 (8)</td>
</tr>
<tr>
<td>Severe tracheomalacia</td>
<td>4 (31)</td>
</tr>
</tbody>
</table>

Nine (69%) of these 13 patients required a gastrostomy, and 2 patients (15%) required prolonged tube feeding because they could not suck or swallow. Only 2 children in this group had less than 3 months of enteral feeding. Among the group of children who did not have a tracheotomy (n = 32), gastrostomy or enteral feeding for more than 3 months was necessary in 7 patients (22%) and 3 patients (9%), respectively. The difference was statistically significant compared with patients with a tracheotomy (P = .01). Only 13 patients (29%) did not have any anomaly involving the digestive system.

One patient received mechanical ventilation using nasal bilevel positive airway pressure (PAP), instituted 6 months after decannulation, followed by failure of resection of the aryepiglottic folds and continued for 14 months. At 18 months of age, respiratory obstruction during sleep linked to a pharyngolaryngomalacia redeveloped. It was seen clearly at fiberscopy under inhaled general anesthesia, which allowed maintenance of spontaneous ventilation. While awake, this patient’s ventilation and blood gas levels were normal, but a significant hypoxia-hypercapnia was found during sleep. The bilevel PAP was well tolerated and allowed normalization of ventilation and blood gas levels during sleep.

In 2 other patients, a resection of the aryepiglottic folds was performed without any improvement in the dyspnea.

One patient died of septicemia at 7 months of age; another died of unknown causes at 3 months of age.

A recent review of the selection criteria for diagnosing the CHARGE association proposed that this acronym should only be used if at least 3 cardinal criteria were met, excluding growth retardation but including at the very least coloboma and/or choanal atresia. We have used this
definition to obtain a homogeneous group of patients for evaluation. In particular, children with craniostenosis or trisomy 21 can have other anomalies of the airways (eg, macrognathia in trisomy 21), even if they fulfill the diagnostic criteria for the CHARGE association, and these anomalies may interfere with the respiratory consequences directly linked to the CHARGE association.

Airway manifestations of the CHARGE association (excluding choanal atresia and cleft lip and palate) were found in 28 (56%) of 50 patients in the literature.4,5

**CHOANAL ATRESIA**

Apart from choanal atresia, no other abnormalities of the aerodigestive tract form part of the CHARGE association. Choanal atresia is found in only 60% of patients on average.12 Coniglio et al8 reported that these patients can have a narrow nasopharynx and a narrow choanal region. These anatomical and radiological observations could explain the high failure rate and the high rate of reintervention noted in patients with CHARGE association.4,8 The therapeutic difficulties resulting from the choanal atresia in these patients led Asher et al4 to suggest a temporary tracheotomy in cases of bilateral atresia. Morgan et al12 did not observe any particular problems in the treatment of such atresia, with a mean of 2.3 interventions, which is similar to our experience. We have observed that the average number of sessions necessary to achieve good choanal patency was slightly greater in patients with bilateral atresia, but the difference was not significant. However, 4 of the children with unilateral atresia still have not been treated for their atresia. One of these required a tracheotomy.

The role of choanal atresia in determining the need for a tracheotomy has not been specifically evaluated in the literature. It would be of value to know the degree of choanal patency at the time of tracheotomy in all patients with the CHARGE association. We found normal choanal patency in 10 (77%) of 13 patients at the time of tracheotomy. This finding reinforces the idea that choanal atresia only has a role as an aggravating factor in the respiratory instability. Therefore, choanal pat-
ency should be a minor indication for an eventual tracheotomy.

**ESOPHAGEAL ATRESIA**

Kutiyanawala et al\(^1\) reported 7 deaths before the age of 1 year in 10 patients with esophageal atresia (mainly due to cardiopathies, apart from 2 patients with recurrent pneumopathy). These 10 patients formed part of a larger series (n = 61) observed by the group at The Hospital for Sick Children, London, England. In the other publications concerning this series of patients,\(^3,5\) esophageal atresia was not associated with major complications. In our series, 4 patients had esophageal atresia. There were no deaths, and the respiratory outcome was favorable (none of the children needed a tracheotomy) after a mean follow-up of 70.2 months. These 4 children did not have cyanotic heart disease.

**MICROGNATHIA**

Like Stack and Wyse,\(^3\) we found nearly a 40% incidence of micrognathia. However, this did not specifically lead to any significant problems with intubation or laryngeal exposure (5 of 18 patients [28%] vs 11 of 27 patients [41%] without micrognathia). It is probable that these problems were diminished by the fact that all observations concerning laryngeal exposure were made during endoscopy of the upper airways by trained pediatric laryngologists in ideal conditions for examination.

**LARYNGOMALACIA OR DISCOORDINATE PHARYNGOLARYNGOMALACIA**

The most frequently observed endoscopic anomaly in this study (61.4%) was pharyngolaryngeal hypotonia. Discoordinate pharyngolaryngomalacia (DPLM) was first used to describe this inability to maintain the patency of the pharyngolaryngeal passage.\(^1\) Typically, this abnormality varies with time and with respiration, becoming more marked during sleep and during inspiration. We believe that the systematic involvement of the larynx and the oropharynx should lead to the abandonment of the term laryngomalacia for this condition. Indeed, the following associations of different types of abnormalities have been observed: anteroposterior flattening of the larynx without any real curling of the epiglottis on itself, and no significant displacement of the aryepiglottic folds toward the laryngeal lumen, whereas the epiglottis often comes into contact with the posterior pharyngeal wall without deviating toward the arytenoids. The arytenoids are usually edematous, probably due to systematic gastroesophageal reflux, which occurs in these conditions. There is an associated pharyngeal collapse involving the lateral walls, but also the posterior wall, with more or less complete obstruction of the airway.

Although this type of dysfunction has not been reported in previous studies on the CHARGE association, the cases of laryngomalacia described do not correspond to typical situations. The airway patency was very difficult to maintain in all 4 patients described by Stack and Wyse,\(^3\) requiring long-term ventilation with continuous PAP or intubation and tracheotomy in 2 children. Asher et al\(^4\) mentioned hypopharyngeal incoordination as one of the factors predisposing to dyspnea. Oley et al\(^4\) described 5 patients with velopharyngeal incoordination. Morgan et al\(^3\) described 5 patients with laryngomalacia among 50 patients, 2 of whom required a tracheotomy. We are unaware of any published successful cases of endoscopic resection of the aryepiglottic folds in a child with the CHARGE association (we have observed 3 failures in our practice), which is in sharp contrast to the higher rate of success in cases of isolated laryngomalacia.\(^15,16\) In addition, these children systematically have particularly severe problems with alimentation;

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**Table 3. Malformations and Anomalies Involving the Digestive Tract Encountered in 45 Patients With CHARGE Association**

<table>
<thead>
<tr>
<th>Malformation or Anomaly</th>
<th>No. (%) of Patients</th>
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<tbody>
<tr>
<td>Gastroesophageal reflux (pH probe monitoring)</td>
<td>36/42 (86)</td>
</tr>
<tr>
<td>Aspiration</td>
<td>31/42 (74)</td>
</tr>
<tr>
<td>Anomalies of sucking and swallowing</td>
<td></td>
</tr>
<tr>
<td>None</td>
<td>13/44 (30)</td>
</tr>
<tr>
<td>Slow swallowing</td>
<td>9/44 (20)</td>
</tr>
<tr>
<td>Sucking but no swallowing</td>
<td>5/44 (11)</td>
</tr>
<tr>
<td>No sucking or swallowing</td>
<td>17/44 (39)</td>
</tr>
<tr>
<td>Electromyography of sucking and/or swallowing</td>
<td>9/45 (20)</td>
</tr>
<tr>
<td>Feeding</td>
<td></td>
</tr>
<tr>
<td>Normal</td>
<td>13/44 (30)</td>
</tr>
<tr>
<td>Transient enteral feeding (&lt;3 mo)</td>
<td>19/44 (23)</td>
</tr>
<tr>
<td>Prolonged enteral feeding (&gt;3 mo)</td>
<td>5/44 (11)</td>
</tr>
<tr>
<td>Gastrostomy</td>
<td>16/44 (36)</td>
</tr>
<tr>
<td>Esophageal atresia (all type III)</td>
<td>4/45 (9)</td>
</tr>
</tbody>
</table>

*CHARGE association is described in the first paragraph of the text. Missing patients indicate status unknown.
†Results were abnormal in all 9 patients in whom electromyography was performed.

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**Table 4. Therapeutic Management of Airway Problems**

<table>
<thead>
<tr>
<th>Airway Problem (No. of Patients)</th>
<th>No. of Choanal Surgeries</th>
<th>Transnasal Repairs</th>
<th>Carbon Dioxide Laser Repairs</th>
<th>Transpalatine Surgeries</th>
<th>All Choanal Procedures</th>
<th>No. of Tracheotomies</th>
</tr>
</thead>
<tbody>
<tr>
<td>Unilateral choanal atresia (12)</td>
<td>8</td>
<td>3 (1.6)</td>
<td>4 (2.2)</td>
<td>5 (1.0)</td>
<td>18 (2.2)</td>
<td>3</td>
</tr>
<tr>
<td>Bilateral choanal atresia (15)</td>
<td>15</td>
<td>9 (1.2)</td>
<td>7 (2.6)</td>
<td>9 (1.0)</td>
<td>38 (2.5)</td>
<td>6</td>
</tr>
<tr>
<td>Normal choanae (18)</td>
<td>0</td>
<td>. . . . . . . . . . .</td>
<td>. . . . . . . . . . .</td>
<td>. . . . . . . . . . .</td>
<td>. . . . . . . . . . .</td>
<td>4</td>
</tr>
</tbody>
</table>

*Some patients underwent successive transnasal and/or carbon dioxide laser repairs and/or transpalatine surgeries, explaining the number of cases in each column. Ellipses indicate not applicable.*
among 27 children with DPLM, only 2 (7%) did not need tube feeding. Prolonged tube feeding was necessary in 5 of 27 patients (18%) or was followed by gastrostomy in 16 patients (59%), which is also exceptionally rare in isolated laryngomalacia.

We established a scale to quantify the degree of obstruction by using the Cotton classification. Of course, this method is subjective, and grading the degree of obstruction is not easy, as it depends on the ventilatory and anesthetic conditions. Nevertheless, the use of this type of scale of severity can have important practical consequences. Among the 13 patients who were tracheotomized, there were 12 (92%) with greater than 89% obstruction of the upper larynx and 9 (69%) with greater than 89% obstruction of the upper pharynx. This degree of obstruction was only seen in 3 patients (9%) (for the pharynx and the larynx) among the 32 patients with evaluable conditions who did not have a tracheotomy, the difference being statistically significant for the larynx and the pharynx ($P<.001$).

**OTHER LARYNGEAL ANOMALIES**

The presence of DPLM was thus the most important anatomico-functional criterion for performing a tracheotomy. However, the ventilatory instability of these children may be compromised by other events. In particular, the onset of left laryngeal palsy after cardiac surgery would appear to be critical. In our series, 3 patients of 4 who had iatrogenic palsy on the left were tracheotomized. Stack and Wyse\(^1\) reported 7 tracheotomies among 50 patients. In 2 of these 7 patients, the tracheotomy was performed during the postoperative period following cardiac surgery to aid long-term ventilation and weaning. These 2 patients may have had a left laryngeal palsy.

The observation of subglottic stenosis would also appear to be relatively frequent (8%, according to Morgan et al\(^3\)). Acquired stenoses are attributable to repeated and prolonged interventions involving the laryngotracheal axis. The presence of congenital stenosis (in 2 [4%] of our patients and 2 [4%] of 50 patients seen by Morgan et al\(^3\)) carries a better prognosis, but may aggravate a precarious ventilatory state. Given that there are no other laryngotracheal anomalies, management can often be undertaken without a tracheotomy.\(^17\) In fact, our 3 patients with subglottic stenosis did require a tracheotomy. All 3 patients had a DPLM, and 1 of them had a laryngeal palsy following cardiac surgery as well.

Two (4%) of our patients had a type 1 posterior laryngeal cleft. One of them underwent tracheotomy (coexistence of a laryngeal palsy and DPLM), whereas the other patient was treatable endoscopically, as was the case for the 2 patients in the series by Morgan et al.\(^3\) In both series, clefts were observed at the same frequency as congenital subglottic stenosis. This point is particularly noteworthy, given that the normal incidence of both malformations is very different.\(^18\)

**TRACHEOBRONCHIAL ANOMALIES**

Tracheomalacia is frequently associated with esophageal fistulae in the context of esophageal atresia.\(^19\) This was also the case in 4 of our patients, none of whom required a tracheotomy. Thirteen other patients had tracheomalacia without esophageal atresia or any evidence of extrinsic compression of the trachea, whereas hypotonia of the posterior wall was responsible for an obstruction of greater than 89% in 5 patients. Four of them underwent a tracheotomy, and all 4 had DPLM. The fifth patient (who did not have DPLM) was treated with respiratory physical therapy and continuous alternating antibiotics for 2 years, in the absence of any serious modification of the blood gas levels.

Bronchopulmonary dysplasia was observed in 1 patient who was premature, rather than being directly related to the CHARGE association. Similarly, an abnormality of bronchial segmentation probably occurred by chance.

**RESPIRATORY CONSEQUENCES OF THE DIFFERENT ANOMALIES**

The diverse anomalies of the upper airways found in patients with the CHARGE association need to be assessed according to the degree of dyspnea, clinically and with sleep recordings and/or measurement of arterial blood gas levels. In fact, among the 19 patients without clinical dyspnea, results of polysomnographic recording identified 4 patients (21%) with hypopnea, 2 of whom had an arterial oxygen saturation level of less than 90%. In these patients, we were able to establish the indications for tracheotomy using a combination of polysomnography results and continuous monitoring of percutaneous oxygen and carbon dioxide tensions (or at least, the capillary oxygen saturation level). Repeated episodes of hypoxia can lead to perturbed cerebral development and chronic hypercapnia, indicating the presence of respiratory insufficiency. The large variations with time of these phenomena and the possibility of sudden unexpected acute obstructive symptoms favor prolonged monitoring of these children.

Nine patients had 1 or several episodes of cardiorespiratory arrest. Eight of them (89%) also had a cardiology (cyanotic in 3 patients). Cardiopathy was less frequent (23 [74%]) in the 31 patients who did not have cardiorespiratory arrest, but the difference was not statistically significant. Five (56%) of the 9 patients later underwent tracheotomy, without subsequent relapse. In comparison, the tracheotomy rate was much lower if there was no history of cardiorespiratory arrest (7 [22%] of 31 patients; $P = .04$). Asher et al\(^1\) has previously highlighted the relationship between episodes of severe dyspnea and delayed development.

We believe that the occurrence of an episode of cardiorespiratory arrest without any specific treatable cause (eg, septic shock) is an important argument in favor of performing a tracheotomy. We believe that repeated episodes of desaturation below 85% and/or a hypercapnia of greater than 50 mm Hg and/or apnea-hypopnea with desaturation or bradycardia represent absolute indications for tracheotomy. Moreover, the origin of these manifestations is often multifactorial, and the specific treatment of only 1 of these factors is usually inadequate, as was the case in the 3 patients in whom we attempted resection of the aryepiglottic folds.
Thus, 13 children (ie, 29% in our series) had a tracheotomy. Although the tracheotomy can cause nonnegligible morbidity and mortality, especially when performed early and when of long duration, we nevertheless prefer this option rather than waiting for a possible respiratory catastrophe. With this approach, one can also hope for an improved cerebral development. However, the presence of multiple sensory handicaps in these patients (visual, auditory, and vestibular) makes it very difficult to assess developmental progress.22

Intermittent positive pressure ventilation using bi-level or continuous PAP may represent an effective alternative in some cases.5,13 However, good nasal patency is a prerequisite20 for this option, and it can only be used intermittently. We have successfully used this treatment for a recurrence of symptoms in 1 patient following decannulation. A combined adenoidectomy and tonsillectomy can help to improve the patency of the upper airways, although such surgery is not of primary importance during the first months of life, when most obstructive events occur.

FEEDING DIFFICULTIES AND GASTROESOPHAGEAL REFLUX

The neuromuscular incoordination that occurs with DPLM15 and the frequency and the severity of the gastroesophageal reflux may explain the particularly severe feeding difficulties encountered. Major perturbations of sucking and swallowing are frequently found and contribute to the high risk for aspiration, which itself can aggravate the ventilatory status of these patients (repeated pneumopathies) and lead to an irreversible change in their pulmonary function.

There is no absolute parallel between the severity of respiratory and digestive symptoms. Nevertheless, a strong correlation was found in this series. Among the 21 patients who needed prolonged tube feeding and/or a gastrostomy, only 3 patients (14%) did not have dyspnea or abnormal blood gas levels. Conversely, the absence of dyspnea was much more common among the children who needed only temporary tube feeding or who had normal alimentation (17 [71%] of 24; P < .001). Furthermore, there was a close link between the need for a gastrostomy and for a tracheotomy (9 tracheotomies among 16 gastrostomies).

CONCLUSIONS

Patients with the CHARGE association present with multiple abnormalities affecting the upper aerodigestive tract. The variability of these lesions and their range of severity explain the large variations observed between patients. A systematic assessment, including in particular endoscopic investigation, and monitoring of respiratory function are essential to accurately define the therapeutic options. Although choanal atresia is easy to detect, it has only a minor role in determining the severity of the respiratory manifestations. Discordinate pharyngolaryngomalacia appears to be a more frequent and more important cause of respiratory obstruction and feeding difficulties. Resection of the aryepiglottic folds is ineffective. Regardless of the degree of choanal patency, tracheotomy is necessary in a significant number of patients. Early tracheotomy may help the development of these children, which is already precarious due to their multiple handicaps.

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Reprints: Gilles Roger, MD, Ear Nose and Throat Department, Hôpital d’Enfants Armand Trousseau, AP-HP, 26 Ave du Dr A Netter, 75571 Paris Cedex 12, France (e-mail: orl.trousseau@trs.ap-hop-paris.fr).

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