Foregut Duplication Cysts in the Head and Neck

Presentation, Diagnosis, and Management

Stephen M. Kieran, MB; Caroline D. Robson, MB, ChB; Vânia Nosé, MD, PhD; Reza Rahbar, DMD, MD

Objective: To review the presentation, diagnosis, and management of foregut duplication cysts of the head and neck in our institution.

Design: An institutional review board–approved retrospective review of all patients treated for foregut duplication cysts of the head and neck over an 18-year period.

Setting: Pediatric otolaryngology tertiary referral center.

Patients: Twenty-two patients with 23 pathologically confirmed foregut duplication cysts of the head and neck were identified. Fourteen patients (64%) were male. The median age at diagnosis was 1.5 years (age range, 5 days to 7 years).

Main Outcome Measures: Clinical data, including age, presenting symptoms, anatomical site(s), evaluation, treatment, and complication, were recorded and analyzed.

Results: Presentation varied depending on anatomical site of involvement, with 12 patients (55%) being asymptomatic. The cysts were found in the oral cavity (n=12), oropharynx (n=6), supraglottis (n=2), and neck (n=3). Imaging, which was performed in 13 patients and consisted of magnetic resonance imaging (n=8), computed tomography (n=5), and ultrasonography (n=1), demonstrated the cystic nature of the lesions. All patients underwent surgical excision, which focused on excising the cyst, while preserving surrounding normal tissues. No patient demonstrated recurrence at follow-up.

Conclusions: Foregut duplication cysts of the head and neck, although uncommon, should be included in the differential diagnosis of cystic head and neck lesions. Preoperative imaging is recommended to differentiate these lesions from other congenital head and neck masses. Surgical excision biopsy with complete removal of the mucosal lining is curative, with no instances of recurrence in our series.


In the developing embryo, the foregut gives rise to the pharynx, lower respiratory tract, and upper gastrointestinal tract (esophagus, stomach, duodenum, and hepatobiliary system). During the first trimester, heterotopic rests of foregut-derived epithelium may persist, resulting in foregut duplication cysts. Such cysts can occur anywhere along the alimentary tract; however, they are most frequently seen in the thorax or abdomen. Foregut duplication cysts in the head and neck have rarely been reported. Their clinical presentation can mimic that of other masses in the head and neck, many of which are optimally managed differently. We report our institution’s experience with these cysts (to our knowledge the largest such series to be reported in the English-language literature) and delineate their clinical presentation, radiologic appearance, pathologic characteristics, and management.

Foregut duplication cysts are benign developmental anomalies that contain foregut derivatives. Traditionally, there are 3 criteria that must be met to make a diagnosis of foregut duplication cyst: they must (1) be covered by a smooth muscle layer, (2) contain epithelium derived from the foregut, and (3) be attached to a portion of the foregut. Duplication cysts are lined by 1 or more types of epithelium: gastric mucosa, ciliated respiratory-type epithelium, stratified squamous epithelium, and simple cuboidal epithelium. All types of cysts may show squamous metaplasia, mucosal ulceration, inflammation, and necrosis, making distinction between the cysts sometimes impossible.

Based on their epithelial type and other features, foregut duplication cysts may appear to closely resemble airway, esophageal, or small intestine. Therefore, the term foregut duplication cyst includes bronchogenic cyst, esophageal duplication cyst, and enteric duplication cyst. Bronchogenic cysts represent 50% to 60% of all mediastinal cysts. They have a ciliated columnar, cuboidal, or pseudostratified epithelial cell layer, with cartilage and respiratory glands.

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Author Affiliations:
Departments of Otolaryngology and Communication Disorders (Drs Kieran and Rahbar), Radiology (Dr Robson), and Pathology (Dr Nosé), Children’s Hospital Boston, Boston, Massachusetts.
and a fibromuscular connective tissue, which identifies them as bronchial in origin. Esophageal duplication cysts have a mucosa with either ciliated columnar epithelium or stratified squamous epithelium and a layer or 2 of muscularis propria. Enteric duplication cysts may be lined by gastric and/or respiratory mucosa and are located in the posterior mediastinum, distinct from the esophagus.3

### METHODS

We retrospectively reviewed all patients treated for foregut duplication cysts of the head and neck over an 18-year period at Children’s Hospital Boston, Boston, Massachusetts. Patients were identified from a prospectively maintained head and neck database and the hospital pathology database. Local institutional review board approval for this study was obtained before the study began. All patients who underwent surgical excision of a foregut duplication cyst between 1989 and 2007 were included in the study, and their clinical and operative records were reviewed. In each case, pathologic specimens were reviewed by a senior pathologist (V.N.), and all available images were reviewed by a senior radiologist (C.D.R.).

### RESULTS

Of the 22 patients, 14 were male (64%) and 8 were female. The mean age at surgery was 1.5 years, with a range of 5 days to 7 years. Twelve patients (55%) were asymptomatic. Symptoms in the other 10 patients included airway obstruction (n=3), feeding difficulties (n=2), tongue swelling (n=1), speech difficulties (n=1), odynophagia (n=2), and otitis media (n=1). The clinical, radiologic, and pathologic findings are summarized in the Table.

Presenting signs on physical examination included anterior cervical masses (n=3), apparent vallecular cysts (n=2), tongue masses (n=6), palatal cysts (n=3), tonsillar mass (n=1), retropharyngeal mass (n=1), and masses in the floor of the mouth and anterior aspect of the tongue (n=7). Imaging was performed in 13 patients (59%). Four patients underwent more than 1 imaging study. Imaging included ultrasonography (US) (n=1), computed tomography (CT) (n=5), magnetic resonance imaging (MRI) (n=8), and thyroid uptake scanning (n=3). In 1 patient, the mass was noted on prenatal US.

Thyroid uptake scanning was performed if a lingual thyroid was considered in the preoperative workup. Imaging studies, which were available for review in 8 patients, consisted of CT (n=4) and MRI (n=4). All examinations revealed cystic, nonenhancing masses with density or signal intensity similar to cerebrospinal fluid in all (Figures 1, 2, and 3) but 1 patient who had proteinaceous or hemorrhagic contents within the cyst. Lesions typically involved the floor of the mouth (Figure 1), appearing to extend into the anterior third of the tongue (n=6) (Figure 2), and were midline (n=5) or midline and extending to the left (n=1). Less commonly, the cysts were located slightly to the left of midline (n=2), adjacent to the supraglottic airway (n=1) and trachea (n=1) (Figure 3). The cysts usually appeared unilocular (n=6). Less frequently, a serpiginous tubular component extended posterior to the cyst (n=2) (Figure 2).

### Table. Summary of Clinical and Radiologic Findings in Patients With a Foregut Duplication Cyst of the Head and Neck

<table>
<thead>
<tr>
<th>Patient No./ Sex/Age</th>
<th>Clinical Features</th>
<th>Clinical Findings</th>
<th>Imaging Findings</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/M/7 y 2/F/7 y 3/F/1 y</td>
<td>Asymptomatic</td>
<td>Lower anterior neck mass</td>
<td>CT: monocystic mass inferior to thyroid</td>
</tr>
<tr>
<td>4/F/8 mo</td>
<td>Asymptomatic</td>
<td>Midline tongue mass</td>
<td>MRI: circular tongue base mass and sinus tract</td>
</tr>
<tr>
<td>5/M/2 y 6/M/2 y 7/F/6</td>
<td>Asymptomatic</td>
<td>Midline anterior mass of tongue</td>
<td>MRI: circular tongue base mass and sinus tract</td>
</tr>
<tr>
<td>8/M/5 y</td>
<td>Intermittent tongue swelling</td>
<td>Midline tongue mass posterior to circumvallate papillae</td>
<td>MRI, thyroid scan, and US: cystic mass extending to tongue base</td>
</tr>
<tr>
<td>9/M/9 mo</td>
<td>Asymptomatic</td>
<td>FOM and tongue mass</td>
<td>None</td>
</tr>
<tr>
<td>10/M/6 mo</td>
<td>Asymptomatic</td>
<td>Midline mass in FOM and tongue</td>
<td>Thyroid scan and CT: midline cyst anterior to hyoid</td>
</tr>
<tr>
<td>11/F/5 d</td>
<td>Feeding problems</td>
<td>Sublingual mass</td>
<td>MRI: midline sublingual mass</td>
</tr>
<tr>
<td>12/F/4 mo</td>
<td>Asymptomatic</td>
<td>Midline mass in anterior undersurface of tongue</td>
<td>MRI and thyroid scan: posterior tongue mass</td>
</tr>
<tr>
<td>13/F/1 mo</td>
<td>Asymptomatic</td>
<td>Midline mass in posterior tongue</td>
<td>Prenatal US: tongue mass</td>
</tr>
<tr>
<td>14/M/7 y</td>
<td>Asymptomatic</td>
<td>Mass posterior to left tonsil</td>
<td>None</td>
</tr>
<tr>
<td>15/M/9 d</td>
<td>Possible otitis media</td>
<td>Mass anterior to left tonsil</td>
<td>None</td>
</tr>
<tr>
<td>16/M/5 mo</td>
<td>Possible otitis media</td>
<td>Two lesions: cyst in right uvula and mass in right side of palate</td>
<td>None</td>
</tr>
<tr>
<td>17/M/7 mo</td>
<td>Mild snoring</td>
<td>Blue vallecular cyst</td>
<td>CT and carotid arteriography: left parapharyngeal space mass</td>
</tr>
<tr>
<td>18/F/2 mo</td>
<td>Respiratory distress</td>
<td>Left bluish retropharyngeal mass</td>
<td>None</td>
</tr>
<tr>
<td>19/M/3 y</td>
<td>Loud snoring</td>
<td>Blue vallecular cyst</td>
<td>None</td>
</tr>
<tr>
<td>20/M/7 y</td>
<td>Odynophagia</td>
<td>Right soft palate mass</td>
<td>None</td>
</tr>
<tr>
<td>21/M/4 y</td>
<td>Asymptomatic</td>
<td>FOM mass</td>
<td>CT: cystic FOM mass</td>
</tr>
<tr>
<td>22/F/3 y</td>
<td>Asymptomatic</td>
<td>Anterior neck mass</td>
<td>CT: level of hyoid, oval lesion with rim enhancement, fluid filled, 1.2 × 0.8 cm</td>
</tr>
</tbody>
</table>

Abbreviations: CT, computed tomography; FOM, floor of mouth; MRI, magnetic resonance imaging; US, ultrasonography.

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All patients underwent surgical excision of their cysts. Simple excision with preservation of surrounding structures was performed in all cases. Twenty lesions were excised transorally. The remaining lesions were located in the anterior aspect of the neck and were excised via local neck incisions.

Pathologic examination was performed in all cases. Grossly, the median maximum dimension of the cysts was 1.4 cm, with a range of 0.5 to 8.5 cm. The lesions were mostly spherical cystlike structures with a pink, tan, and smooth glistening surface. The cysts were found to contain gastric mucosa in 5 cases and respiratory epithelium in 16 cases. They were filled with clear, mucinous fluid. A thick, layered muscular wall lined by diverse epithelium characterized the specimens. In some areas, the epithelial cells were of the ciliated type, while in others, there was gastric-foveolar–type mucosa or focally squamous epithelium. Occasional seromucous glands were seen. Areas of cartilaginous tissue were also present. Ganglion cells were found between the muscular layers, and a few mixed inflammatory cells were also seen.

Only 1 patient had a postoperative complication: a tongue wound dehiscence after excision of a foregut duplication cyst on the anterior aspect of the tongue. All patients were followed up in the pediatric otolaryngology clinic. Duration of follow-up ranged from 11 months to 6 years, with a mean follow-up of 2.4 years.

**COMMENT**

The primitive foregut gives rise to the oropharynx, the lower respiratory system, the esophagus, the stomach, parts 1 and 2 of the duodenum, the liver, the pancreas, and the biliary apparatus. The mucosa observed in the cyst may not correspond to the normal gastrointestinal mucosa at the level of the cyst, and multiple mucosal types may occur within a single cyst. Therefore, even though pure esophageal, gastric, and colonic enteric cysts are seen, they often defy anatomical categorization and are termed foregut duplication cysts. A number of theories exist as to the etiology of foregut duplication cysts: (1) duplication abnormalities may occur because of disturbed recanalization with abnormal foregut rests forming cysts; (2) cystic duplications arise from a supernumerary lung bud found in the foregut during the fifth and seventh week of embryogenesis; and (3) part of the developing stomach may become trapped between the lateral lingual swellings as they close over the tuberculum impar. The latter theory is supported by the finding that columnar and goblet cells in foregut duplication cysts tend to be well differentiated, with local inducive factors possibly acting on primitive endothelial cell rests.

Foregut duplication cysts most commonly occur in the chest and abdomen. The estimated incidence of alimentary tract duplications is 1:4500, approximately one-third of which are foregut duplication cysts. While foregut duplication cysts may occur anywhere from the mouth to the anus, occurrence in the head and neck is uncommon, with approximately 65 reported cases by 1997. The oral cavity is the most common location in the head and neck, with approximately 50 reported cases. However, to our knowledge, no reports have mentioned foregut duplication cysts of the pretracheal neck.

In our institution, we have treated 22 patients with 23 foregut duplication cysts of the head and neck over the past 18 years. These cysts may contain only a mucosal lining or mucosa, submucosa, and muscularis propria. The mucosal lining may be squamous, respiratory, intestinal, or mixed, with the secretory epithelium causing gradual enlargement of the cyst. The most common presenting clinical features are feeding difficulties, odynophagia, stridor, tongue edema, and speech difficulties. The neonate, however, may present with frank respiratory distress due to airway obstruction from the mass. Only 1 of the 3 patients who presented perinatally did so with respiratory distress, while a second patient presented with feeding problems. Overall, 12 of our 22 patients (54%) were asymptomatic at presentation.

The most common site of foregut duplication cysts in the head and neck is the oral cavity; our study supports this finding. Of the 23 cysts described in our study, 12 (52%) presented in the oral cavity, with 5 masses in the anterior aspect of the tongue and 7 masses in the floor of the mouth. Five of these oral cavity lesions were asymptomatic; the others presented with feeding or speech-related problems. Foregut duplication cysts in the oropharynx are also uncommon.

Figure 1. A 7-week-old girl with a tongue mass. A, An axial fast spin-echo inversion recovery magnetic resonance image reveals a unilocular, sharply circumscribed midline lesion that is isointense with cerebrospinal fluid located within the anterior aspect of the floor of the mouth. B, A gadolinium-enhanced, fat-suppressed sagittal T1-weighted image reveals a cystic mass that does not enhance.
with possibly only 2 previously reported cases.\textsuperscript{16,17} We identified 6 oropharyngeal foregut duplication cysts, with 3 soft palate lesions (2 lesions were synchronously identified in 1 patient), 1 tonsil lesion, 1 retropharyngeal mass, and 1 tongue base mass. As might be expected from their location, oropharyngeal lesions are unlikely to present on routine neonatal examination; in our series, all 6 oropharyngeal cysts presented with symptoms.

We have identified 3 patients who presented with anterior or pretracheal neck masses that on histologic examination were most consistent with a foregut duplication cyst. This phenomenon is highly unusual. According to our experience, these cysts would have been expected to have arisen from the floor of the mouth or the anterior third of the tongue, pharynx, or esophagus. One possible explanation is that a transition occurs, from a bronchogenic cyst to a less differentiated foregut duplication cyst containing enteric mucosa. Two patients presented with cysts in the region of the vallecula and epiglottis: one with odynophagia and drooling, and the other with airway distress from birth.

In evaluating these lesions, imaging (CT or MRI) is invaluable. Previous authors have found that endoscopic examination and barium swallow studies have only limited utility in the diagnosis and preoperative planning of the esophageal duplication cyst.\textsuperscript{18} Endoscopically, a submucosal mass with normal overlying mucosa is typically found, making it difficult to distinguish from other lesions, such as a leiomyoma. Similarly, extraluminal compression is seen on barium swallow. While technically difficult, endosonography has been used to assist in diagnosis.\textsuperscript{19} For the rare lesion that communicates with the spinal canal, MRI is usually performed. Although a foregut duplication cyst is indistinguishable from a thyroglossal duct cyst and a dermoid cyst in terms of appearance on CT and routine MRI pulse sequences, the presence of a cystic mass in the anterior floor of the mouth or anterior third of the tongue distinguishes a foregut duplication cyst from a thyroglossal duct cyst or a vallecular cyst. Thyroglossal duct cysts are usually located between the foramen cecum and hyoid bone or within the infrahyoid neck. Vallecular cysts are located in the vallecula. Mucous retention cysts may appear indistinguishable from foregut duplication cysts in terms of both imaging characteristics and location. Studies have demonstrated the association of vertebral anomalies with foregut duplication cysts, termed neurenteric cysts, as the cyst interferes with anterior fusion of the vertebral mesoderm. The patients in our series who underwent neck CT or MRI did not have any evidence of vertebral or spinal anomalies.

With the increased use of maternal US, cystic lesions are being diagnosed before birth. One patient in our series had a tongue foregut duplication cyst diagnosed by antenatal US. Because the patient was asymptomatic at birth, with no evidence of respiratory distress, surgical intervention was deferred until the patient was 1 month old. Two cases of enteric duplication cyst of the oral cavity being detected in utero have been previously reported, both were initially decompressed before formal surgical excision.\textsuperscript{20}

Even though 9 of our 22 patients did not undergo preoperative imaging, we recommend imaging (either MRI or CT) in all patients to localize the lesion, to assess the extent of the mass, and to look for other pathologic entities. Those patients without imaging in our series were included almost exclusively at the beginning of our 18-year review, when such imaging modalities were not as readily available.

The differential diagnosis of pediatric cystic head and neck masses is extensive and includes mucocoele, ranula, dermoid, lymphatic malformation, venous malformation, teratoma, thyroglossal duct cyst, epidermoid cyst,
and lymphoepithelial cyst. The management of some of these lesions is substantially different from that of foregut duplication cysts. For example, for suspected thyroglossal duct cysts, the Sistrunk procedure (removal of the cyst in continuity with the central portion of the hyoid bone) is performed to avoid recurrence in contrast to the foregut duplication cyst, for which simple excision suffices. Furthermore, in the case of venous and lymphatic malformations, sclerotherapy or nonoperative management may be appropriate. Therefore, awareness of foregut duplication cysts as a differential diagnosis is essential, and preoperative imaging of suspected cases is recommended.

The treatment options proposed for such cysts include observation, resection, and aspiration. However, if the cyst is left untreated, there is the potential for complications to develop. Malignant transformation has been reported to occur in long-standing foregut duplication cysts of the head and neck, with 1 previous case report of an adenocarcinoma and 1 of metaplasia. In the chest, where foregut duplication cysts are more frequently encountered, early excision has traditionally been recommended to avoid serious complication. Because gastric epithelium may be a component, peptic ulceration can occur, resulting in bleeding or tissue perforation. If the cyst is left untreated, sinus formation with chronic mucous secretion may develop, as occurred in 1 patient in our series with an anterior neck cyst.

In certain circumstances, temporary aspiration of the cyst may confirm a benign diagnosis and temporarily alleviate symptoms such as respiratory distress and feeding difficulties. Asymptomatic simple cysts have the potential to grow and if they become symptomatic can result in higher rates of perioperative complications. Therefore, in view of the need for definitive histologic diagnosis and the risk of malignant potential without treatment, as well as to relieve associated symptoms, surgical resection is the treatment of choice.

In conclusion, foregut duplication cysts of the head and neck may present a diagnostic dilemma and should be included in the differential diagnosis of a congenital lesion of the head and neck, particularly in cases that involve the anterior floor of the mouth and tongue or that are located in proximity to the airway. Surgical intervention in the form of simple excision is recommended as it is both diagnostic and therapeutic. Preoperative imaging is used to provide a limited differential diagnosis and assists with surgical planning. Surgical excision has been shown to be both diagnostic and curative.

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Correspondence: Stephen M. Kieran, MB, Department of Otolaryngology and Communication Disorders, Children’s Hospital Boston, 300 Longwood Ave, Boston, MA 02115 (skieran@rcsi.ie).

Author Contributions: Drs Kieran and Rahbar had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. Acquisition of data: Kieran. Analysis and interpretation of data: Kieran, Robson, Nose, and Rahbar. Drafting of the manuscript: Kieran. Critical revision of the manuscript for important intellectual content: Kieran, Robson, Nose, and Rahbar. Statistical analysis: Kieran. Administrative, technical, and material support: Nose and Rahbar.

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REFERENCES