

**TITLE: Congenital Laryngeal Anomalies**

**SOURCE: Grand Rounds Presentation, UTMB, Dept. of Otolaryngology**

**DATE: November 2, 2005**

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## **Laryngeal Anatomy**

There are certain differences between the adult and the newborn larynx that are significant in understanding the presentation and progression of the clinical picture in the different congenital laryngeal anomalies. The newborn larynx is about one third the size of the adult counterpart. The diameter of the subglottic and glottis are narrower which leads to an increase propensity for airway obstruction and compromise. The subglottic region is about 4 to 5 millimeters in diameter. The epiglottis is also narrower in infants. The larynx lies at the level of the fourth cervical vertebrae at birth. By fifteen years of age it has descend to the level of the six to seventh vertebrae.

## **Laryngeal embryology**

The larynx develops from the fourth and fifth branchial arches. At the third week of gestation, the respiratory primordium is derived from the primitive foregut to later form the lung bud and later the bronchial bud which will eventually develop into the tracheobronchial tree. At the fourth and fifth week of gestation the tracheoesophageal folds fuse to form the tracheoesophageal septum leading to the separation of the tracheal airway lumen from the esophageal digestive tract. Failure in the adequate development of this structure results in a communication between these two lumens and will place the patient at risk for aspiration. Examples of developmental failure of the tracheoesophageal septum are laryngeal and laryngotracheoesophageal cleft.

The laryngotracheal groove is the primitive opening of the larynx during development. This structure will develop into the primitive laryngeal aditus which is formed by three eminences. The hypobranchial eminence is the most cephalad of these structures and will later develop into the epiglottis. The other two eminences will form the arytenoid cartilages. The laryngeal lumen obliterates and later recanalizes by the tenth week of gestation. Failure of complete recanalization will result in anomalies producing airway obstruction. Examples of incomplete recanalization are laryngeal webs and congenital subglottic stenosis.

## **Laryngeal function**

The larynx has an array of functions including breathing passage, airway protection, and aids in the clearance of secretions and in vocalization. In laryngeal pathologies, symptoms are a direct effect of the malfunction of the coordinated activities within the larynx. The laryngeal airway can be reduced by anomalies like laryngeal webs, laryngomalacia and subglottic stenosis among others producing symptoms of airway obstruction. Symptoms of airway obstruction can be benign presenting with mild stridor, but can also present with increased work of breathing associated to retractions, nasal flaring and tachypnea, apnea episodes, cyanosis and even sudden death. It is important to be able to recognize the different characteristics of stridor because this leads to recognition of the location and a more accurate differential diagnosis. Inspiratory stridor can be observed in pathologies of the supraglottis and glottis due to collapse during negative inspiratory pressure. Biphasic stridor may be seen in subglottic laryngeal and cervical tracheal anomalies. Expiratory stridor is seen in the distal trachea and the main stem bronchi.

There are three level of airway protection. The first level involves the epiglottis, aryepiglottic folds and arytenoids. The second level of airway protection involves the false vocal folds and the third level is the true vocal folds. Anomalies on any of these structures may lead to aspiration and swallowing dysfunction with symptoms of coughing, choking and gagging episodes, stasis of secretion and recurrent pneumonia. Feeding difficulties in these patients can be associated to aspiration or due to intolerance to the interruption of breathing while being fed.

Phonatory abnormalities are dependent on the level of abnormality. For example, a muffled cry suggests supraglottic obstruction while a high pitched or absent cry is associated with glottic abnormalities (webs & atresia). Other phonatory abnormalities can include hoarseness, weak cry and even aphonia.

## **Laryngomalacia**

Laryngomalacia is the most common congenital laryngeal anomaly accounting for 60% of them. It is also the most common cause of stridor in children. Laryngomalacia is two times more frequent in males than females. It is characterized by flaccidity of the supraglottic tissue leading to inward collapse of supraglottic structures during inspiration causing airway obstruction. Laryngomalacia is said to be caused by a derangement of supraglottic anatomy, histology or neurologic function.

The stridor in patients with laryngomalacia is characterized by intermittent low-pitched inspiratory stridor usually starting within the first two weeks of life. The stridor increases in severity during the first few months after birth followed by gradual improvement. It peaks at six months and most are symptom free by 18 to 24 months (75%).

In mild to moderate cases, stridor may be the only symptom of laryngomalacia. It may be exacerbated by exertion including crying, agitation, feeding or supine position. In more severe obstruction, intercostals and substernal retraction along with other signs of respiratory distress may be seen. In cases of chronic severe obstruction the patient may develop pectus excavatum. Other complications of laryngomalacia include feeding difficulties, GERD, failure to thrive, cyanosis, intermittent complete obstruction, cardiac failure & death.

There are several characteristic anatomic abnormalities that can be visualized in laryngomalacia. The epiglottis can be long and tubular, a pathological exacerbation of the normal omega shape. The aryepiglottic folds are also shortened and there is a posterior inspiratory displacement of the epiglottis against the posterior pharyngeal wall or inferior collapse to the vocal folds. There may be inward collapse of aryepiglottic folds, primarily the cuneiform cartilages and an anteromedial collapse of the arytenoid cartilages.

There is no concrete pathophysiologic explanation for the supraglottic collapse in laryngomalacia. One of the proposed explanations for this collapse is laryngeal cartilage immaturity leading to weakness and collapse on inspiration. However, arguing against this theory is that the incidence of laryngomalacia is not increased in premature infants. Also there are no microanatomic abnormalities found on histologic exam except for pervasive subepithelial edema. There has also been an association between laryngomalacia with central apnea, hypotonia, mental retardation and early speech delay, which has led to theories of disturbance of neuromuscular control. It is suggested that muscles involved in dilation of the larynx like the stylopharyngeus, palatopharyngeus, hyoglossus & digastric muscles are affected in this laryngeal pathology.

There is an association of more than 50% Gastroesophageal reflux disease in patients with laryngomalacia. In laryngomalacia, the increased negative intrathoracic pressure with collapsed supraglottic structures leads to retrograde gastric contents. Posterior supraglottic edema and erythema is also seen and may lead to further airway compromise due to reduction of the laryngeal lumen.

The diagnosis of laryngomalacia is best made by awake fiberoptic laryngoscopy in which the anatomic abnormalities and the supraglottic collapse can be visualized. Dysphagia can be observed and diagnosed by fiberoptic endoscopy & videofluoroscopy. A chest radiograph and soft tissue neck radiographs should also be done in order to visualize the subglottis and tracheal airway. Direct laryngoscopy and bronchoscopy should be performed by evaluating for synchronous lesions because patients with laryngomalacia have a 27% association with synchronous lesions.

Since most symptoms of laryngomalacia resolve spontaneously within the first 18 months after birth, observation is indicated in the majority of cases. Medical management for GERD should be started after diagnosis in order to decrease airway edema and further airway compromise.

In laryngomalacia, surgical management is reserved for severe symptoms and for failure of medical management. In 1922, Iglauer performed the first surgical approach for the management of laryngomalacia by amputation of epiglottic redundant tissue with a wire snare. Today the most common technique is supraglottoplasty, which consists of an array of procedures directed to treat the individual abnormalities. Part of the procedure involves trimming mucosa from lateral edges of the epiglottis, aryepiglottic folds and arytenoids and corniculate cartilages with the aid of a CO2 laser, microlaryngeal scissors and microdebrider.

There are several complications associated with supraglottoplasty. If an aggressive approach is taken in which several anatomic abnormalities are addressed in the same surgery, the

patient may develop supraglottic stenosis, exacerbation of dysphagia with aspiration and in rare cases, massive collapse of the supraglottic framework leading to tracheotomy placement. The best approach for preventing these complications is a conservative excision.

## **Laryngoceles & Saccular Cysts**

In order to understand this laryngeal pathology it is crucial to understand the anatomy of the saccule. The laryngeal ventricle is a fusiform fossa bound by the vocal folds and the false vocal folds. The saccule is a cecal pouch of mucous membrane found in the anterior roof of the ventricle. It is bound medially by the false vocal cords, anterolaterally by the thyroid cartilage and superior anteriorly by the epiglottis.

Laryngoceles are abnormal dilations or herniations of the saccule which communicates with the lumen of the larynx and is filled by air or mucous. There are three types: internal, external and combined. The internal extends posterosuperiorly into the aryepiglottic fold. The external extends cephalad to protrude through the thyrohyoid membrane. The combined have characteristics of both external and internal laryngocele. Laryngoceles can be congenital or acquired. In the newborn, laryngoceles are typically congenital while in the adult, laryngoceles occur in those with occupations with increased pressure on the laryngeal lumen.

Saccular cysts, also called congenital cysts of the larynx or laryngeal mucocele, are differentiated from laryngoceles in that the lumen is isolated from the interior of the larynx and it doesn't contain air. There are two types of saccular cysts, lateral and anterior. The lateral saccular cyst extends posterosuperiorly into the false vocal cords and aryepiglottic folds. The anterior saccular cyst extends medially and posteriorly protruding into the laryngeal lumen between the true vocal cords and false vocal cords.

Congenital Saccular Cysts are caused by a developmental failure to maintain patency of the saccular orifice. Acquired cases are due to inflammation, trauma or tumors occluding the saccular orifice. Laryngopyocele are infections of the laryngocele, saccular cyst or large saccule.

Laryngoceles have a more benign presentation than other laryngeal anomalies with symptoms of intermittent hoarseness and dyspnea, weak cry or aphonia. In contrast, saccular cysts usually present with respiratory distress, inspiratory stridor, inaudible or muffled cry and occasional dysphagia. The diagnosis is made by visualization of the mass and its position with a flexible laryngoscopy followed by direct laryngoscopy to evaluate the extension and the relation to the saccular orifice. A high KV soft tissue radiograph of the neck may aid in the diagnosis of laryngocele if it is distended with air. Combined laryngocele will show a mass in the neck that protrudes with a modified Valsalva maneuver on physical exam. The presence of a saccular cyst is confirmed by needle aspiration.

The treatment of saccular cysts starts with aspiration or unroofing with cup forceps or CO2 laser. However, there is a high incidence of recurrence with this technique. For recurrence or more definitive treatment, an endoscopic excision is performed with dissection of the cyst to its base at the saccule orifice and removal of remnants with a CO2 laser. If it recurs, excision through a lateral cervical approach is performed incising the thyrohyoid membrane and carrying the dissection to the base at which point the saccule is ligated. During this procedure, it is crucial

to protect the superior laryngeal nerve and it is rarely necessary to remove thyroid cartilage. Intubation, however, may be needed until edema subsides

## **Vocal Cord Paralysis**

Vocal cord paralysis is the third most common congenital laryngeal anomaly producing stridor. Unilateral and Bilateral cord paralysis share the same incidence. There is a 50% association of congenital cord paralysis to other anomalies. Of acquired paralysis, 70% are associated to congenital neurologic abnormalities (meningocele, Arnold Chiari Malformation and Hydrocephalus) or neurosurgical procedure to treat them. Unilateral are often associated to cardiovascular anomalies like PDA and left side paralysis is more common.

Bilateral paralysis produces high-pitched inspiratory stridor and inspiratory cry. There is a paradoxical function in which the vocal cords close during inspiration due to negative lumen pressure and open during expiration due to positive intralumen pressure. In unilateral vocal cord paralysis, less symptoms are present. These include weak cry, occasional breathy and feeding problems secondary to laryngeal penetration and aspiration.

The diagnosis is best made by awake flexible laryngoscopy in which the larynx is visualized and recorded for evaluation of vocal cord motion. In a small child, recording the FFL will enable slow motion replay for better visualization of the cord motility. Imaging of head and chest are done to evaluate for neurologic or cardiovascular abnormalities. Direct laryngoscopy with palpation of the glottis and even laryngeal EMG can be performed to assess for vocal cord fixation versus neurogenic paralysis. Laryngeal lesions can be diagnosed by evaluation under anesthesia with passive motion of arytenoids and by visualization of webbing or scarring.

In the majority of cases, the treatment of idiopathic vocal cord paralysis involves watchful waiting. This is because there is spontaneous resolution in 70% of patients and most resolve within the first six months. Feeding difficulties associated with paralysis are managed by thickening of liquids. Rare surgical management is required in which case the first line should be tracheotomy awaiting for spontaneous resolution. If increased intracranial pressure is present, early shunting or posterior fossa decompression is typically performed for better outcome. In bilateral vocal cord paralysis, a tracheotomy may be necessary. If tracheotomy is not performed, continuous evaluation of failure to thrive is needed as airway requirements increase with growth. The surgical goal is to improve glottic airway by lateralizing one or both paralyzed vocal cords. Reinnervation technique has unclear utility and is rarely used. Surgical lateralization procedures are injurious to the developing larynx. Also excisional procedures in which tissue is removed from the posterior glottis by open or endoscopic technique with laser has been performed with success (laser arytenoidectomy and posterior cordotomy). More consistent results are achieved by external approaches including arytenoidectomy, arytenoidopexy or laryngeal expansion with costal cartilage to the posterior cricoid plate. Voice long term results are typically acceptable and rare aspiration problems are seen. Late complications include failure to achieve an adequate airway.

## **Congenital Laryngeal Web**

This is an uncommon laryngeal pathology. Most cases of congenital laryngeal web are

glottic and extend into the subglottic larynx. Symptoms depend on location and degree of involvement and are associated with some degree of vocal dysfunction. In the case of thin anterior web with visible vocal folds, there is little airway obstruction and mild hoarseness. Thicker webs and extension into the subglottic leads to increased airway obstruction and weaker voice. More than 75% glottic involvement and significant subglottic extension leads to aphonia and severe airway obstruction requiring tracheostomy soon after birth. Supraglottic webs are rare, most represent a fusion of the ventricular bands anteriorly causing mild symptoms of airway obstruction. In posterior supraglottic or interarytenoid webs, the larynx is difficult to visualize and there may be limited vocal cord motility. Complete laryngeal atresia is incompatible with life and requires emergent tracheostomy.

Thin anterior glottic web may require incision or dilation. More significant glottic lesions require incision along one vocal fold and dilation, with revision surgeries for better results. Subglottic involvement is usually accompanied by anterior cricoid plate abnormality requires an external approach with division of the web and the cricoid plate and reorientation of the anterior cricoid plate. A cartilage graft can be used to expand or a laryngeal keel can be placed for 7-14 days. If the anterior cricoid plate is not amenable to repositioning, it can be resected submucosally followed by anterior costal cartilage augmentation or completion of a partial cricotracheal resection in severe cases.

## **Congenital Subglottic Stenosis**

Congenital subglottic stenosis is the second most common cause of stridor in neonates, infants and children. It is characterized by a narrowing of the subglottic lumen in the absence of trauma. Full term newborn larynx diameter is 5mm. Less than 4mm in newborn or 3mm in the premature infant is consistent with subglottic stenosis. The etiology of congenital subglottic stenosis is incomplete recanalization of the laryngeal lumen during embryogenesis.

There are two types of subglottic stenosis, membranous and cartilaginous. The membranous type is circumferential, soft and dilatable. The mucosal lining is thickened by an increased fibrous connective tissue layer of the submucosa or by hyperplasia and dilation of the mucous glands in the subglottis. The cartilaginous type shows thickening and deformation of the cricoid cartilage. There are two types of cricoid deformities in the cartilaginous type, a flattened cricoid and an elliptical cricoid. The flattened cricoid has a smaller anteroposterior diameter while the elliptical has a smaller transverse diameter. The elliptical is also associated to laryngeal cleft.

Symptoms of upper airway obstruction predominate in Congenital subglottic stenosis. The stridor is inspiratory with progression to biphasic. Agitation worsens the symptoms due to increased airway flow. In mild to moderate cases, patients are asymptomatic. During bouts of URI, patients develop airway edema which decreases the laryngeal lumen and the patient develops symptoms of croup. Congenital subglottic stenosis patients may have a history of recurrent or prolonged croup. In severe obstruction, the patient may exhibit respiratory distress needing intubation. This changes the category from congenital to acquired.

Diagnosis of congenital subglottic stenosis is made by rigid endoscopy with visualization of the entire larynx. The distinction between soft membranous versus cartilaginous as well as

documenting the relation to the vocal cords should also be made at this time. A full exam of the tracheobronchial tree is performed in order to assess for synchronous lesions and measurement of the stenosis with sequential size ET tube should also be done. Connection to the anesthetic circuit pressure can be done to find the ET tube that permits a leak of 10-25 mm of H<sub>2</sub>O. The ET tube circumference is an accurate measure of airway size.

Classification of subglottic stenosis is made based on the degree of obstruction. Grade I is characterized by less than 50% obstruction, grade II 51-70% obstruction, grade III 71-99% obstruction, and grade IV has no detectable lumen. Also consideration of the extension out of the subglottis, general medical condition, swallowing ability, age & weight are made.

Treatment of subglottic stenosis is based on classification and degree of obstruction. In grade I, supportive care awaiting for growth is usually the best management option while cases with more than 50% obstruction may require some intervention. Patients with grade II-III usually present with history of multiple failed extubation. In these cases, tracheostomy may be needed until the cricoid cartilage grows for decannulation. Anterior laryngotracheal decompression or cricoid split is utilized predominantly in neonates or young children with anterior subglottic or glottic narrowing in which failed multiple attempts at extubation leads to extubation in 66-78% and tracheotomy decannulation in 75-78%. In this procedure, a horizontal skin incision over the cricoid is followed by a vertical midline incision that splits the entire cricoid, first two tracheal rings and lower 1/3 of thyroid cartilage. Two Prolene sutures are then placed on each side of incised cricoid and the patient is kept intubated for 7-14 days for stenting of the airway. In cases that involve the posterior glottis or subglottic stenosis or severe grade III & IV, an anterior, posterior and possible lateral laryngotracheal decompression with long term above the stoma stenting and reconstruction with costal cartilage to the posterior and possible anterior cricoid regions with 2-4 wks stenting is performed.

## **Laryngeal & Laryngoesophageal Clefts**

Congenital Laryngeal & Laryngoesophageal Clefts are rare with an incidence of <0.1%. They are characterized by posterior midline deficiency in the separation of the larynx and trachea from hypopharynx and esophagus and incomplete development of the tracheoesophageal septum. Congenital laryngeal & Laryngoesophageal Clefts have a strong association with other anomalies (56%), most commonly tracheoesophageal fistula in 20-27%. They are associated with Pallister-Hall syndrome, an autosomal dominant, hypothalamic hamartoblastoma, laryngeal cleft, hypopituitarism, imperforated anus and polydactyly. They are also associated with G syndrome/Opitz-Friaz syndrome dysphasia, hypospadias, hypertelorism, cleft lip and palate.

Minor clefting involves only a failure of interarytenoids muscle development. In major cleft, the defect can extend to the carina and even mainstem bronchi. It is important to differentiate the length of the cleft as laryngeal (interarytenoids only, partial cricoid or complete cricoid) and laryngotracheoesophageal clefts that extend into cervical or intrathoracic trachea.

The symptoms of laryngeal and laryngotracheoesophageal clefts are inspiratory stridor, cyanotic attacks associated with feeding, aspiration, and recurrent pulmonary infections. The length of the defect is proportional to the severity of symptoms with 100% aspiration of laryngotracheoesophageal clefts. The best single diagnostic study is endoscopic examination by

direct laryngoscopy and bronchoscopy in which the arytenoids need to be parted for adequate visualization due to redundant esophageal mucosa prolapsing into the glottic and subglottic lumen.

Most clefts are limited to the supraglottic larynx and do not require surgical intervention. Evaluation and treatment for GERD (gastroesophageal reflux disorder) as well as swallowing therapy should be performed. If surgical management is required, endoscopic repair is successful in 80% of cases. Surgical repair is required with extension below the vocal cords. An anterior approach through a laryngeal fissure is most commonly used with excellent exposure of the entire defect without risk to laryngeal innervations. Complete laryngotracheoesophageal cleft may require a posterolateral approach to allow a two layer closure. A tracheostomy is present or performed at the time of the reconstructive surgery.

The mortality associated with laryngeal clefts is usually due to other anomalies or excessive delay in making the diagnosis rate of 11% and 46%. Mortality of intrathoracic laryngotracheoesophageal is as high as 93%.

## **Subglottic Hemangiomas**

Subglottic Hemangioma is a benign vascular malformation with histological findings of endothelial hyperplasia. It is more frequent in females with a 2:1 ratio. There is a 50% association with cutaneous hemangioma and is usually asymptomatic at birth. 85% of patients present with stridor by 6 months of age. This lesion has a rapid growth phase in the 1<sup>st</sup> year follow by slow resolution with most having complete resolution by 3-5 years of age. There is a 30-70% mortality rate if untreated. The priority is to maintain the airway while minimizing potential long term sequella.

The diagnosis of subglottic hemangiomas is made by direct laryngoscopy to visualize and palpate the lesion which is compressible, asymmetric, and usually posterolateral with bluish or reddish discoloration. CT and MRI can be performed to assess extension and characteristics.

The principal medical treatment of subglottic hemangioma is systemic steroids, which has been shown to induce partial regression in most patients (82-97%). Steroid treatment, however is associated with a risk of growth retardation and increased susceptibility to infection. This risk is reduced by alternate-day dosing regimen in the smallest doses possible. Also intralesional corticosteroid treatment has been employed with successful avoidance of tracheotomy. Another successful medical treatment is Interferon alpha-2a with 50% or greater regression of lesion in 73% of patients. This treatment requires prolonged therapy and blocks various steps of angiogenesis. Side effects include neuromuscular impairment, skin slough, fever and liver enzyme elevation.

Surgical management of subglottic hemangiomas includes tracheotomy which bypasses the obstructing lesion while waiting for the expected involution. There are risks of tracheostomy as well as delay in speech and language associated. Laser CO2 and KTP have been used with success, but are associated with a significant risk of inducing subglottic stenosis in up to 20% of cases. Surgical excision has been successful with decannulation shortly after surgery and avoiding tracheostomy in 85% of patients, however, there is risk of laryngeal distortion or

damage with this procedure.

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