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Glossoptosis

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ABSTRACT

Glossoptosis causes varying degrees of airway obstruction and feeding difficulties. It can occur as a consequence of micrognathia in Robin Sequence, but can also occur in children with hypotonia. Despite several attempts to classify severity in Robin Sequence patients, taking into account symptoms, presence of concomitant syndromes or malformations, and even endoscopic findings, there is still no general consensus. Furthermore, several management recommendations have been reported without an agreement about indications, efficacy, or risks of each treatment option. The present article provides an overview of clinical presentation, diagnosis, management, and prognosis of patients with glossoptosis.

Introduction

Glossoptosis is defined by the backward and downward fall of the base of the tongue causing obstruction to the airway while the posterior pharyngeal wall remains in a stationary position. It is important to differentiate this condition from another cause of airway obstruction, the pharyngomalacia. In the latter, the anterior wall of the pharynx moves posteriorly, and its posterior wall moves anteriorly. This condition differs from glossoptosis, in which only the tongue moves posteriorly.[1] The diagnosis is suggested by micrognathia in the so-called Robin Sequence (RS), but hypotonia of the pharyngeal and glossal musculature can also result in glossoptosis in children with neurologic impairment. Glossoptosis was a cause of airway obstruction in 25% of children referred for fluoroscopic sleep studies and micrognathia was present in only 3 of the 17 patients in this series.[1] Clinical manifestations of glossoptosis are heterogeneous, and therefore treatment has to be individualized, aiming at adequate breathing and optimized growth and development. That the care of these patients should be multidisciplinary is unequivocal, but there is no consensus on the diagnostic workup and treatment.

Clinical manifestations

Glossoptosis is the cause of varying degrees of respiratory and deglutition dysfunction, ranging from mildly affected children who require no support to severely affected ones who present with disabling airway obstruction and are unable to feed. The presence of other malformations, aside from the possibility of cleft palate, contributes for the variations in clinical manifestations in these patients. Also, abnormal maxillary morphology causing midface hypoplasia has also been described with RS and may contribute to airway obstruction.[2]

Airway obstruction symptoms can occur spontaneously or with feeding, during wakefulness or sleep.

Obstructive sleep apnea (OSA), associated with failure to thrive, behavioral deficits, and sudden infant death, has a high prevalence in patients with glossoptosis, not always accompanied by snoring. Accordingly, snoring did not appear to correlate with OSA severity.[3] The obstruction may be present at birth, may become progressively worse in the first few weeks of life, or even develop much later in life. Premature infants tend not to develop obstruction until nearly term gestational age.[4] It is important to emphasize that upper airway obstruction may not be clinically apparent at birth, and OSA in these patients may manifest as failure to thrive instead of obvious obstruction. The children with glossoptosis expend a great deal of energy to breathe against an obstructed airway. OSA in combination with known feeding difficulties can further exacerbate growth failure.[3,4] Airway disorders can also be triggered by incidental surgical procedures.[5]

Printzlau and Andersen[6] described a possible correlation between the severity of respiratory problems with the severity of retro/micrognathia, the presence of other malformations and the intrauterine impairment.

Swallowing disorders are frequent in patients with glossoptosis[7] and are characterized by low oral intake, feeding times greater than 30 min, fatigue, and coughing, gagging, and vomiting with intake. These feeding difficulties are thought to be secondary to airway obstruction, inasmuch as impaired breathing can lead to

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incoordination of sucking and swallowing, and glossoptosis prevents forward positioning of the tongue required for suction. Besides airway obstruction, the swallowing disorder can be further compounded by additional neurologic disorders, through diffuse hypotonia and an uncoordinated swallowing mechanism. In addition, if cleft palate is present, it becomes increasingly difficult to generate the negative intraoral pressure necessary to suck. In isolated RS children with feeding difficulties, early airway intervention can dramatically reduce the need for feeding intervention. Chronic feeding difficulties and eventual gastrostomy tube placement are higher in glossoptosis when associated with concomitant syndromes, other malformations, and neurologic abnormalities despite the timing of airway intervention.[8]

Gastroesophageal reflux is also prevalent in RS patients and can complicate airway obstruction, since it causes airway inflammation and edema, increasing secretion production. The evaluation of gastroesophageal reflux is particularly important if there is a persistence of some degree of airway obstruction despite optimal medical management.

There are several attempts to classify patients with RS, based on symptomatology, presence of concomitant syndrome, malformation, or regarding endoscopic findings. Although there is no consensus on what should be the best approach for these patients, those classifications are useful, especially when comparing treatment results. Based on the severity of symptoms and signs, Cole et al.[9] devised a classification for RS: *grade 1*: no respiratory distress when nursed supine, inconsistent glossoptosis, feeding assessment satisfactory; *grade 2*: intermittent evidence of mild respiratory obstruction when nursed supine, none nursed on side, consistent glossoptosis, feeding precipitates some respiratory distress; *grade 3*: moderate-to-severe respiratory distress when nursed supine, evidence of airway obstruction remains when nursed on side, consistent glossoptosis, unable to feed orally.

Medical comorbidities

As cited, the association of glossoptosis, micrognathia, and resultant airway obstruction is known as RS, a congenital craniofacial anomaly. A cleft palate can be an associated malformation.[10,11] The incidence of RS varies from country to country, from 1/5000[12] to 1/14,000 live births.[6] This broad range of incidence can be a result of regional variations, but is potentially a consequence of the lack of clearly defined diagnostic criteria, one being able to find up to 15 different definitions in the literature.[13,14]

RS patients can be divided according to their presentation in isolated RS, RS plus (associated with additional congenital malformations without a known specific diagnosis), and syndromic RS.[15] More than 40 syndromes with RS have been described[16] and the most common associated syndromes are Stickler syndrome, Velocardiofacial syndrome, Treacher Collins, and facial and hemifacial microsomia.[17] The proportion of syndromic diagnosis in RS patients varies between 14.6% and 46%.[17,18]

A distinction is made between micrognathia and retrognathia. Micrognathia refers to size, whereas retrognathia to position. In Treacher Collins syndrome, for example, the mandible is short. In deletion 22q11.2 syndrome, the mandible is essentially normal in size, but retrognathic in position because the cranial base angle is larger than normal.[16] This differentiation is not always easy to discern in childhood.

Because of this frequent association with syndromes, a geneticist should be involved in the multidisciplinary assessment of the RS patients to aid in the identification of a specific syndromic diagnosis and provide recommendations for genetic testing. Furthermore, the high frequency of associated anomalies, with or without a diagnosed syndrome, warrants an active investigation with an echocardiogram, neonatal hearing screening, ophthalmological evaluation, and ancillary-specific investigations based on clinical suspicion.[18]

Evaluation

Anatomical airway evaluation

There is uniform agreement on the need for a careful evaluation of the upper airway with flexible fiberoptic laryngoscopy (FFL) in all infants with RS, since a multitude of airway abnormalities may be present.[19] Aside from the mechanical obstruction of glossoptosis, other factors can contribute to ventilation compromise in these patients. Understanding the site of airway obstruction seems to be critical for determining optimal therapy. There is a growing debate on whether awake[20–22] or slight sedation[19,23–25] endoscopic evaluation would be a better option for these patients. In our opinion, since the worst moment of obstruction in RS is during sleep, the sleep endoscopy would be the best method to evaluate obstruction in RS patients. However, sleep endoscopy was not yet described in such a population.

Attempts to classify glossoptosis severity were already reported, although none specifically seems to be consensually accepted worldwide. There is certainly a need for dedicated research on their informative value in a prospective context and their association with clinical and polysomnographic parameters.

Sher et al.[20] described a classification of airway findings (not of glossoptosis). They described 4 mechanisms of obstruction in 33 patients with craniofacial anomalies, including RS, from birth to 24 years of age, examined while awake. The mechanisms described are *type 1*: the tongue contacting the posterior pharyngeal wall below the soft palate (true glossoptosis); *type 2*: posterior contraction of the tongue toward the posterior pharyngeal wall, but the palate becomes sandwiched between the tongue and velum; *type 3*: medial contraction of the lateral pharyngeal walls; and *type 4*: sphincteric, the tongue does not contact the posterior pharyngeal wall.

Yellon[24] examined 14 children under light sedation and graduated epiglottis and base of tongue prolapse from 0 to 3 (Figure 1).

de Sousa et al.[21] evaluated 56 children with RS, examined while awake and graduated glossoptosis in mild, moderate, and severe (Figure 2). They described a poor correlation between the severity of glossoptosis and the severity of clinical manifestations.

An important tool in endoscopic evaluation is jaw-thrust maneuver performed under anesthesia, in which the mandible is brought forward manually with direct endoscopic visualization and can predict dynamic airway change that occurs with mandibular advancement.[26]

Objective evaluation of airway obstruction

Clinical signs alone seem to be insufficient to ascertain the degree of upper airway obstruction, while the use of polysomnography (PSG) greatly improves the diagnostic accuracy of upper airway obstruction severity assessment.[4]

PSG is the gold standard for the diagnosis of airway obstruction, particularly if the clinical picture is not clear. It is well known that young infants have central nervous system pauses in breathing, and PSG is useful to differentiate them from obstructive pauses. However, it must be performed concomitantly to endoscopic evaluation, since it does not evaluate the degree of obstruction. An obstructive apnea index of 1 is chosen as the cutoff for the reference range and an apnea index of 10 is considered to be severe, diverging from adults.[27,28]