TOPICS IN PEDIATRICS

Congenital macroglossia: clinical features and therapeutic strategies in pediatric patients

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Abstract Congenital macroglossia is a condition that consists in an enlarged tongue protruding beyond the alveolar ridge in a resting position. It has been classified into two categories: true macroglossia, which occurs in congenital or acquired forms, and relative macroglossia. As this alteration may be due to different causes, its incidence is not known. It is more frequently associated with Beckwith-Wiedemann syndrome, mucopolysaccharidoses, and Pompe disease, and it has been less frequently associated with isolated muscle hypertrophy or hemangioma or lymphangioma. Macroglossia is characterized by an enlarged and thick tongue that may have ulcers and fissures, cause tongue alterations, difficulties for feeding and swallowing, sialorrhea and recurrent infections or even the obstruction of the upper airway. The clinical evaluation must include a complete clinical chart with careful physical exploration and a pedigree of at least three generations, besides identifying the presence or absence of an associated hereditary syndrome. Macroglossia management is complex. More than twenty different surgical options to reduce the tongue size have been proposed; however, there is not a general agreement in this respect so far. The objective of this work was to review clinical and surgical aspects related to macroglossia from non-surgical pediatricians and geneticists, addressed to the different medical specialists, including the maxillofacial surgeons who are involved in the management of these patients.

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1. Embryological development of the tongue

Tongue develops from a series of ventral thickenings (on the floor of the pharynx),\(^1\) which occur in the fourth week of gestation, and a pair of lateral thickenings and one medial thickening (tuberculum impar) that take place during the fifth week of gestation. These thickenings, which are on the inner side of the mandibular arches, originated from the first pharyngeal arch\(^2\) in such a way that their union is finally represented by the median sulcus of the tongue.\(^3\)

Behind the tuberculum impar, there is a splanchnic elevation called hypobranchial eminence, which is formed by the mesoderm from the second, third, and part of the fourth-branchial arches.\(^4\)

The growth of the tongue is due to an expansion of the lateral tongue thickenings and the tuberculum impar, which join to form the two anterior thirds of the body of the tongue. The root of the tongue is derived from the hypobranchial eminence and the ventromedial tissue.\(^2\) The fusion line of the anterior and posterior portions of the tongue is indicated by the groove in the form of \(V\), which is called terminal sulcus.\(^7\)

The mesenchyme of the branchial arches forms the connective tissue and both blood and lymph vessels.\(^1\) The intrinsic muscles of the tongue develop from myoblasts of the occipital somites.\(^1,5\) During the migration of these cells, \(PAX-3\) gene is expressed.\(^4,5\) Innervation of the two anterior thirds of the tongue is given by the fifth cranial nerve (derived from the first branchial arch), while the posterior third is innervated by the ninth (derived from the third branchial arch) and the tenth cranial nerves. The motor innervation is given by the ninth and twelfth cranial nerves.\(^5\)

2. Definition and classification of macroglossia

The tongue is a structure of the oral cavity that is indispensable for the production of speech, swallowing, and breathing.\(^6,7\) The greatest growth of the tongue occurs in the first eight years of life, reaching its final size at the age of 18 years; its mean length is 25.3 cm and 22.6 cm in males and females, respectively,\(^3,8,9\) although a correlation between size and age in children has not been established.\(^1\)

It is accepted that the term macroglossia refers to a tongue which protrudes beyond the alveolar ridge (Fig. 1).\(^10\) This alteration is important in the pediatric practice because of the complications that this presentation can cause. Therefore, it is necessary to identify if it is an isolated entity or a syndromic presentation (which would require genetic counseling). Both situations are transcendent to decide the course of action regarding surveillance and the need for surgical intervention.

Macrognosia can cause dental-muscular-bone deformities, alterations in chewing, speech and airway obstruction.
The correct understanding of the signs and symptoms orients to the diagnosis and possible treatment of patients. It also helps to identify those who may benefit from surgical treatment, either to improve function, stability of dental conditions or just the aesthetic appearance (without a precise surgical indication).  

The classification performed by Vogel et al. in 1986, which is currently used, considers two categories: true and relative macroglossia. True macroglossia, which can be congenital or acquired, is caused by a primary tongue condition or alteration. Histological deformations correlate with clinical findings of an elongated tongue. Relative macroglossia consists of a small oral cavity or a neurological dysfunction, as it occurs in children with Down syndrome, who tend to maintain an open mouth with a protruding tongue due to the characteristic hypotonia of this syndrome. Another classification, described by Myer et al. in the same year, places macroglossia as generalized or localized, subdividing it according to its etiology in congenital, inflammatory, traumatic, metabolic and neoplastic.

3. Etiology of macroglossia

Because of the association of macroglossia with multiple genetic syndromes, the true incidence of this disorder is unknown. Regarding its etiology—which is perhaps the most appropriate strategy—Balaji described macroglossia according to its causes into four categories.

Tissue overgrowth, as in Beckwith-Wiedemann syndrome (BWS), congenital hypothyroidism, chromosomal abnormalities, hemihyperplasia, and mucopolysaccharidosis, among others. Tissue infiltrate, as it is the case of lymphatic or venous malformations, hemangiomas, neoplasms, mucopolysaccharidoses, and neurofibromatosis. Relative macroglossia, as in Down syndrome, micrognathia, muscular hypotonia, and angioedema. Inflammatory or infectious causes.

The most common causes of macroglossia include hemangioma, glandular hyperplasia, and lymphangioma. The latter, which etiology is unknown, originates in the lymphatic vessels; 75% of the cases are located in the head and neck, and its occurrence in the oral cavity is rare. However, when present, it is located on the two-thirds of the dorsal surface and the ridge side of the tongue. Hemangiomas are located in the head and neck approximately in 60% of cases and less commonly in gums, lips, tongue or palate. The least frequent causes of macroglossia comprehend some tumors, such as dermoid cyst or rhabdomyoma.

Mucopolysaccharidosis is associated with the BWS, mucopolysaccharidoses or Pompe disease; it can be observed in multiple syndromes of genetic origin. For example, Moreno-Salgado et al. identified 19 patients with BWS from January 2007 to December 2012 at the Hospital Infantil de México Federico Gómez, from which 89% presented macroglossia. In the case of acquired macroglossia, the causes can be systemic, such as amyloidosis, myxedema, lymphoma or carcinoma, or because of reactive local changes as in angioneurotic edema.

4. Clinical manifestations and complications

It is important to identify the signs and symptoms of macroglossia to determine if glossectomy is necessary: the presence of an elongated thickened and widened tongue, open anterior or posterior bite, prognathism, malocclusion with or without a crossbite, chronic protrusion of the resting tongue, fissured tongue and ulcers. Also, the presence of glossitis due to the prevalence of oral breathing should be noted, as well as alterations of the speech, particularly articulation problems, asymmetry of the maxillary or mandibular arches, difficulties in feeding and swallowing, sialorrhea, recurrent infections of the upper airway, airway obstruction and sleep apnea.

Due to the relationship with physical appearance, macroglossia has aesthetic implications since it may be perceived as an intellectual disability, which generates anxiety on the family and the patient. X-ray techniques allow to detect if the tongue occupies all of the oral cavity, if it protrudes through an anterior open bite, if a mandibular dentoalveolar protrusion or bimaxillary dentoalveolar protrusion exist, or the presence of over angulation of anterior maxillary and mandibular teeth and mandibular disproportionate overgrowth.

The initial approach to identifying the presence of macroglossia should be based on subjective clinical criteria (such as morphology and protrusion of the tongue) and alterations in the articulation, deglutition, and breathing. Subsequently, the evaluation should begin with a medical history and a complete physical examination, including a family tree of at least three generations. According to the suspected etiology, laboratory analysis can be requested: thyroid function, karyotype and expanded metabolic screening and lab studies (lateral skull radiography or magnetic resonance imaging). These studies will support the diagnosis of macroglossia by showing a tongue which occupies the complete oral cavity and protrudes beyond the alveolar ridge in the resting position. Functional tests may also be requested to identify alterations in speech, chewing or airway permeability. Invasive studies such as biopsies can be used in localized lesions of the tongue for the diagnosis of neoplasms and diseases of systemic origin. When macroglossia is suspected to be associated with syndromic entities, it is necessary to perform a complex diagnostic process due to the great diversity of syndromes associated with this disorder. A diagnostic algorithm has been proposed, where macroglossia is classified as isolated, probably BWS or associated to other syndromic entity. At least, an abdominal ultrasound should be requested for every patient to discard other BWS clinical manifestations since it is the most often associated with macroglossia syndrome.

Diagnosis and treatment should be focused on a multidisciplinary approach to reduce the risk of permanent maxillofacial speech alterations. Medical treatment includes conservative measures to reduce inflammation and bleeding. For example, in trauma by bites or lymphangiomas, sclerosing agents, cryotherapy, electrocautery, steroids, and embolization, among other treatments can be used. These actions allow the correction of complications such as airway obstruction, alterations in the
articulation of speech, mandibular deformation, dental and aesthetic deformities, but always trying to preserve the taste, sensitivity and the movement of the tongue.7,11,17,24 The type and size of the malformation, the anatomical structures involved and the infiltration of the surrounding tissue should be considered for the selection of the treatment.17

In some cases of macroglossia, the treatment of choice must be surgery. An absolute indication for this treatment is the airway obstruction, and a relative indication is altered deglutition due to macroglossia or dysphagia.15,24 At the present day, no precise clinical criteria indicative of glossectomy have been reported in the literature; however, there are cases of acquired macroglossia where the symptoms are so important that surgical procedures coupled with the treatment of the base pathology would be required.23 The surgical treatment should be selected in three situations: the presence of functional deficits (impaired swallowing, articulation of speech, sialorrhea or airway obstruction); dental alterations due to macroglossia; psychological impact due to the physical appearance of the patient, which gives a false impression of mental disability.7,23.

Surgical techniques reported in the literature can be divided into two groups: midline and peripheral glossectomy.7 However, a consensus where a particular technique is recommended does not exist. Therefore, every patient should be evaluated by an expert to select the most appropriate surgical approach.20 One of these techniques is the peripheral incision with marginal resection of tissue, which complications may be hypomobility and changes in the shape of the tongue which acquire a globular appearance.13 The V-shape incisions in the midline are effective to decrease the length; however, the width remains unchanged. Elliptical incisions in the midline without reaching the apex of the tongue decrease the width but not length.15 Resection of the central portions of the tongue has been recommended to preserve speech, sense, and taste.27 During the surgical treatment, the combination of a V-shaped and an elliptical incision is known as keyhole-shaped incision. This incision decreases both width and length of the tongue. In recent reviews, it is promoted to avoid the excision of the tip because this is the most mobile and sensitive portion of the tongue.25

Balaji14 recommended to preserve the tip and the side edge of the tongue as they areas of vital importance in the existing glossectomy surgical procedures. The final shape of the tip of the tongue is important aesthetically both for the patient and their parents. Moreover, it is the site for the identification of flavors. The preservation of the side edges prevents fibrosis of the muscle of the tongue.

It has been proposed that the optimal age for surgical treatment should be between four and seven years of age. However, an earlier correction may be necessary if the life of the patient is at risk due to the complications of macroglossia.12

The main post-surgical complications are the alteration of the movement and the decrease of taste, mainly in the perception of salty and bitter flavors.28 Finally, it is important to consider that every patient who has undergone a glossectomy should be subject to speech therapy for repairing or improving the acquired deficits.27

The bibliography in this review regarding the glossectomy in children with macroglossia of various causes29-50 shows that there is no consensus on the best surgical technique for correcting this alteration, even in cases of the same etiology.

Conflict of interest

The authors declare no conflicts of interest of any nature.

References


