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PEDIATRIC THEME

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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PALABRAS CLAVE
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Macroglosia congénita: características clínicas y estrategias de tratamiento en la edad pediátrica

Resumen La macroglosia congénita es una condición que se caracteriza por una lengua que en posición de reposo protruye más allá del borde alveolar; se ha clasificado en dos categorías: verdadera, que puede ser congénita o adquirida, y relativa. Debido a la asociación de esta alteración con múltiples causas, su incidencia es variable. Es más frecuente que la macroglosia se asocie al síndrome de Beckwith-Wiedemann, a las mucopolisacaridosis y a la enfermedad de Pompe, y con menor frecuencia a linfangioma, hemangioma o hipertrofia muscular aislada. La macroglosia se caracteriza por una lengua alargada, engrosada y/o ancha, protruida crónicamente en reposo, con presencia o no de fisuras y úlceras, alteraciones del lenguaje, dificultad para la alimentación y deglución, sialorrea e infecciones recurrentes de la vía respiratoria superior u obstrucción de la misma. Su valoración en niños debe iniciarse con una historia clínica y exploración física completas y con la elaboración de un árbol genealógico de al menos tres generaciones, además de investigar la presencia o no de una entidad sindrómica. Se han propuesto más de veinte técnicas quirúrgicas para resolver la macroglosia congénita; sin embargo, a la fecha no existe consenso para la aplicación de una técnica en particular para reducir su tamaño. En esta revisión se pretende destacar los aspectos clínicos y quirúrgicos de la macroglosia, desde la perspectiva de pediatras no cirujanos y genetistas, hacia los cirujanos maxilofaciales que atienden a estos pacientes.

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 larger 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.[3]

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 the twelfth cranial nerves.[3]

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 transcendental to decide the course of action regarding surveillance and the need for surgical intervention.

Macroglossia can cause dental-muscular-bone deformities, alterations in chewing, speech and airway obstruction.

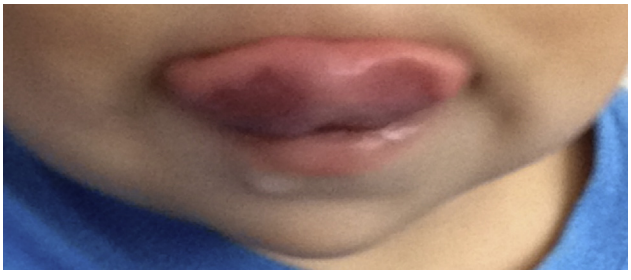

Figure 1 Macroglossia present in a patient with Beckwith-Wiedemann syndrome.