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# Beckwith Wiedemann Syndrome: Presentation of a case report

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## Abstract

Beckwith Wiedemann Syndrome (BWS) is a rare congenital disease of low prevalence. However, it presents a high prevalence within the genetic pathologies of overgrowth. This syndrome presents typical manifestations such as macroglossia, macrosomy at birth, omphalocele and defects of the anterior abdominal wall. Its origin is known to be genetic, but its mechanism of generation is not clear. This syndrome has been the object of wide studies since investigators have established a relationship between the methods of assisted fertilization (assisted reproduction treatment, ART) and its appearance.

Currently, research is oriented towards the improvement of the prenatal diagnostic techniques, which would allow a preparation of the multidisciplinary medical team to treat the pathologies with which these patients are born.

Next we present the case of a 1 year-old child who consults this service with a diagnosis of macroglossia associated with BWS.

***Key words:*** *Macroglossia, Beckwith Wiedemann syndrome, glossectomy.*

## Introduction

Beckwith Wiedemann Syndrome (BWS) was described independently by two investigators. In 1963, Beckwith presented 3 postmortem cases with macroglossia, omphalocele, cytomegaly of the fetal adrenal cortex, renal medullar dysplasia and visceromegaly. On the other hand, Wiedemann in 1964 (1) reported 3 cases of siblings with similar clinical characteristics, adding diaphragm defects and hypoglycemia.

It is a disease of low prevalence; however, it represents one of the most common overgrowth syndromes (2-4). In Spain, 0.13 for every 10000 live births (5). In the United Kingdom, it was reported a prevalence of 1% of live births (6).

This genetic syndrome has its apparent origin in an alteration of the expression of genes from de chromosome 11 region p15.5, which can be sporadic (85%), inherited (15%) or because of chromosomic abnormalities (1%), (2, 3, 6-10). This alteration has been found primarily on the IGF2 genes, which is a fetal growth factor, and in the H19 gene, which is thought to be a tumor suppressor gene (4).

The diagnosis of this syndrome is done mainly on basis of the existence of 5 major criteria such as macroglossia, defects of the anterior abdominal wall, hypoglycemia at birth or previous to parturition (3, 7, 10). In patients with some systemic alteration of growth, such as a hyperinsulinemia refractory to treatment and without a clear diagnosis, a genetic study becomes important (7). For parents with familial background genetic counseling gains importance,

whereas it would establish opportune norms of treatment and limit the consequences that a hypoglycemia could provoque at birth, fundamentally the development of neurologic damage (2).

Clinically, it presents in diverse forms, its most common features being macroglossia (97-100%) which can be asymmetric, defects of the abdominal wall (77-80%), hypoglycemia (63%) and macrosomy (68%) (2, 3, 5, 6, 8-11). There are other minor clinical expressions (10) that sometimes are unnoticed such as the predisposition to neoplasms or embrionary tumors, placentomegaly, grooves in the earlobe, cleft palate, renal alterations, visceromegaly, refractory hyperinsulinemia, polydactyly, mental retardation (3, 6, 10, 12). However, the neurologic engagement is rare (2).

The prevention of this type of syndrome doesn't have a defined protocol, but there have been orientations established for parents in risk situation. The risk situations correspond to a family history of BWS, utilization of some type of ART (7-9). The ART have been under investigation, since there has been an increase observed in the number of cases of BWS or alterations in gene expressions associated to BWS in children born by ART, unlike controls born by natural means (7, 8). No relation has been found between socioeconomic status and BWS (8).

Regarding the diagnosis of these patients, it must be as precocious as possible (13). With current imagenology methods it is possible to determine previous to birth if a baby presents some feature of BWS. This exam could be reinforced by direct diagnostic techniques such as amniocentesis or chorionic villus sampling, with the technical difficulties and consequences that these could carry (14). During pregnancy, by ultrasonic studies it is possible to determine renal function or the presence of embrionary tumors (2), own expressions of this pathology.

The local treatment of this syndrome is focused to limiting the functional alterations such as those caused by macroglossia (10, 11).

Facing a macroglossia, which represents the most evident physical feature in these patients, a differential diagnosis must be made with conditions of lymphangioma, idiopathic muscular hypertrophy, hemangioma, radbomyomas, amyloidosis, cretinism and acromegaly (5, 10, 13, 15). When it is not treated it provoques dentoesqueletal alterations, protrusion of teeth, respiratory alterations, open bite, increased goniac angle, increase of mandibular dimensions and swallowing (10).

The defects of the anterior abdominal wall must be treated immediately or within few months from birth (13).

Another expression present is macrosomy. In regards to it the literature indicates that the diagnosis must be strict because there is a percentage of patients with size alterations without apparent cause, which we could find in genetic alterations (2, 6) especially if it associates to other systemic alterations that could orient towards BWS (2, 6).

## Case Report

Male sex 1 year-old patient consults the Pediatric Maxillofacial Surgery Service of San Juan de Dios Hospital for a Macroglossia. This patient comes with a diagnosis of Beckwith Wiedemann Syndrome from his treating physician. Within his morbid background: controlled pregnancy, diagnosis of macroglossia and omphalocele in the prenatal controls, birth by cesarean at 33 weeks, operated at birth for omphalocele, congenital hypothyroidism in treatment with levothyroxine sodium (18 mgs. daily). Without familial background. Segmentary general exam without alterations. Good general state. Psychomotor state with mild hypotony, height/weight relationship with 9 kgs. overweight. Extraoral exam: open mouth with slight lingual protrusion (figure 1), grooves in earlobes (figure 2). Intraoral exam: slight lingual depapilation and protrusion of tongue. Requested exams: Hemogram, Glycemia, Hepatic and Thyroid Tests, within normal ranks.

Later he underwent a Partial Glossectomy intervention by rhomboid technique (figures 3-5) under general anesthesia which was performed without inconveniences. At discharge the patient is well. At 7 day control he is normal.

He then continues controls with his treating physician.

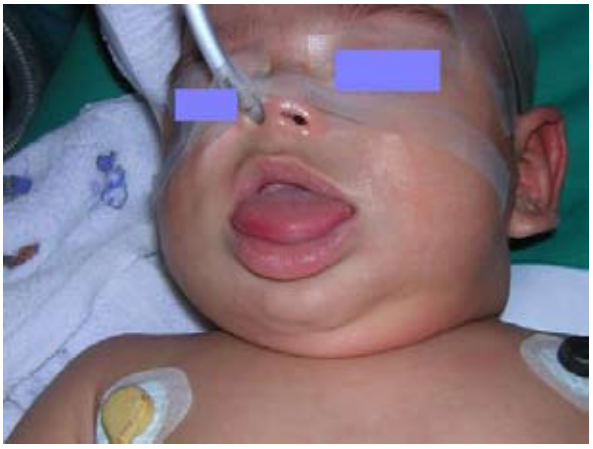

**Fig. 1.** Patient in general operating room, previous to glossoplasty.

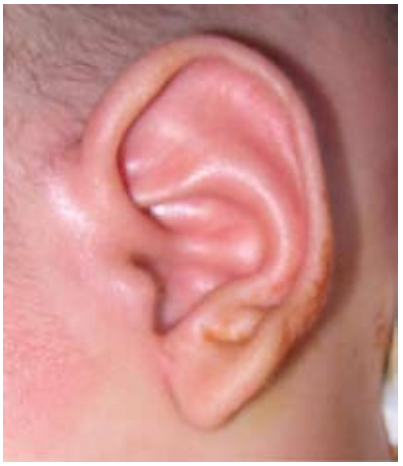

**Fig. 2.** Typical earlobes of a patient with BWS.

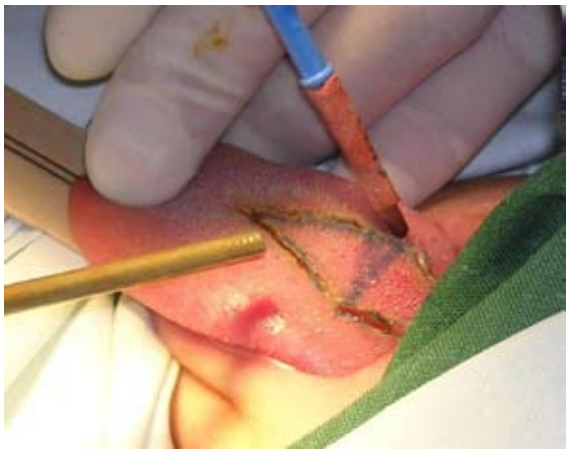

**Fig. 3.** After marking the tongue, the plasty is performed with electric scalpel.

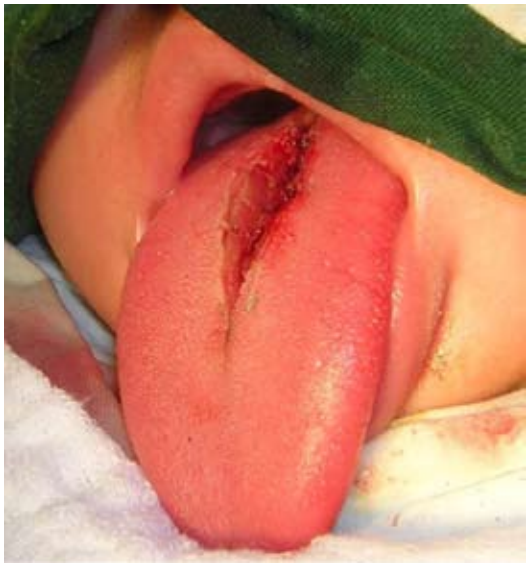

**Fig. 4.** Immediately after the incision of the planned section, the surgical site is appreciated previous to suturing.

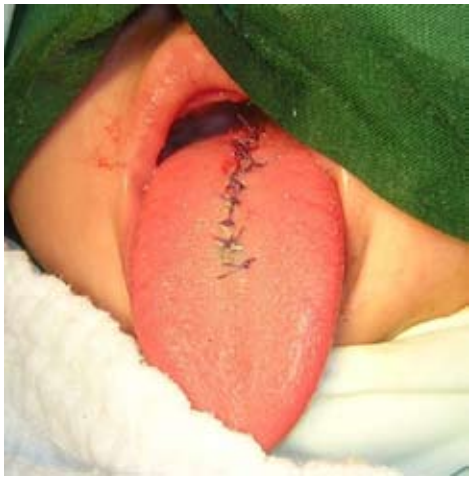

**Fig. 5.** After the plasty, proceeds the closure with 4-0 Vicryl. The tongue is clinically better inside the oral cavity.

## Discussion

Beckwith Wiedemann Syndrome (BWS) represents a genetic syndrome of low prevalence and diverse clinical expression. However, it is one of the most common overgrowth syndromes and presents macroglossia in almost all its forms (2, 5, 6). Facing a macroglossia of unknown cause or any other of the major diagnostic criteria, the clinician can suspect a BWS. The diagnosis of macroglossia is based on the morphology and protrusion; situation that carries functional, growth, psychological or feeding problems (10). In these cases macroglossia constitutes a true muscular hypertrophy, which is why glossectomy rarely relapses. Surgical glossectomy in this type of patients is recommended as soon as possible (10). This fact has been studied and some authors determined an alteration in taste perception and phonation (10) while other studies show the opposite (11).

The prenatal diagnosis, which with current technology is increasingly certain (2), represents an important tool because it allows two situations. On one hand, to prepare the parents, motivate them to have a periodic follow-up because of the increasing possibilities of developing tumors, and genetic counseling in case of the desire to have more children. The second situation is that it allows the planification of the surgical interventions necessary for the correction of defects present in the child, omphalocele/umbilical hernia, besides establishing a norm to prevent the consequences of metabolic alterations, for instance a neonatal hypoglycemia in the nervous system.

At the Maxillofacial level it is very relevant to detect macroglossia precociously whereas its opportune treatment results in the avoidance of any type of facial growth alterations (10, 11), such as dentoesqueletal alterations, protrusion of teeth, respiratory alterations, open bite, increased goniac angle, increase in mandibular dimensions with prognatic appearance (10) and swallowing . On the other hand, from the orthodontic point of view, the macroglossia intervention becomes necessary before starting the orthodontic treatment (10, 11).

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