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*Letter to editor*

# THE ROLE OF DENTIST IN ORAL TREATMENT OF BECKWITH-WIEDEMANN SYNDROME

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Beckwith-Wiedemann syndrome disease (BWD), originally termed exomphalos, macroglossia, and gigantism syndrome, was independently described by Beckwith, an American pathologist, and Wiedemann, a German pediatrician, in 1960 (1). BWD is an overgrowth syndrome that results in the overgrowth of any part of the body as one of its three main presenting features during infancy, along with abdominal wall defects and macroglossia. Patients with BWD may also present with other features of varying severities such as ear abnormalities, hemihyperplasia, enlarged abdominal organs, neonatal hypoglycemia, and increased predisposition to embryonal tumors during early childhood (1).

Variability in the mode of inheritance and clinical presentation of BWD has led to an underestimation of the exact prevalence and severity of the disease. Nonetheless, BWD is considered the most common congenital overgrowth disorder, despite its relatively low prevalence of 51 in 10,340 live births as reported in different ethnicities. Approximately 5% of BWD cases are sporadic, while 40% of BWD are inherited.

The complex underlying genetic mechanisms of BWD involving molecular aberrations of the genes within chromosome 11p15.5 include translocation, duplication, or inversion of this chromosome. The most extensively studied genes of the chromosome 11p15.5 region implicated in BWD are potassium voltage-gated channel subfamily Q member 1 (KCNQ1OT1), insulin-like growth factor 2 (IGF2), and cyclin-dependent kinase inhibitor 1C (CDKN1C) and imprinted maternally expressed transcript (H19) genes. Epigenetic changes were detected on chromosome 11p15.5, and it is a major characteristic of imprinted genes associated with BWD (2).

To address the phenotypic variety of BWD, is recommended the use of a clinical scoring system for the diagnosis of BWD. The scoring system of BWD relies on the cardinal and suggestive features. Cardinal features are those that when

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present are strongly suggestive of BWD; thus, two points are assigned for each feature. Cardinal features of BWD include macroglossia, omphalocele, lateralized overgrowth, hyperinsulinism, bilateral Wilms tumors, and specific pathological findings such as placental mesenchymal dysplasia or adrenal cytomegaly.

By contrast, suggestive features of BWD are those characterized as being independent of the general pediatric population. In fact, in BWD children birth weight is greater than two standard deviations above the mean: these children manifest ear creases or pits, polyhydramnios or placentomegaly, facial nevus simplex, transient hypoglycemia, nephromegaly or hepatomegaly, embryonal tumors, and umbilical hernia or diastasis recti. Each of these features of BWD was assigned one point. Based on this scoring scheme, a patient with a score of ≥4 satisfied the clinical diagnosis of classical BWD. A multidisciplinary team is often recommended for the management and care for a patient with BWD depending on both their phenotypic presentation and molecular subtype.

The complex manifestations of BWD require different management protocols to provide coordinated health care for patients. One of the target features to be addressed is macroglossia, since approximately 90% of patients diagnosed with BWD exhibit this feature (3).

Similarly, most cases of macroglossia during childhood are due to BWD. Approximately 40% of children diagnosed with BWD undergo a surgical tongue reduction because it may otherwise lead to functional difficulties with feeding, breathing, drooling, and speech as well as affect facial appearance. The goal of the surgical procedure in BWD is to reduce tongue bulk while preserving its normal shape and improving function.

Macroglossia of BWD is diagnosed based on morphology and on whether growth, feeding, functional, or psychological problems arise from tongue protrusion. Tongue reduction is the optimal choice of treatment of BWD patient with macroglossia to reduce drooling and feeding, breathing, and speech difficulties and prevent problems associated with facial appearance and occlusion. The appropriate timing for surgery of macroglossia in BWD children is before age 2 years to obtain favorable functional and esthetic results.

Aside from the functional difficulties caused by macroglossia, BWD highly predisposes affected children to cancerous and non-cancerous tumors. Hepatoblastoma, a form of liver cancer, and Wilms tumor, a kidney cancer, can develop in BWD children. The risk of severity of these tumors depends on clinical and molecular findings. Therefore, in BWD children, follow-up is recommended to strictly screen for tumors or other manifestations by abdominal ultrasonography as first-line investigation for Wilms tumor and hepatoblastoma and to measure serum alpha fetoprotein levels every 3 months until age 8 (4).

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