Clinical treatment of oral manifestations of Beckwith-Wiedeman syndrome in a child

Aldo Giancotti* / Gaia Romanini** / Raffaella Docimo*** / Claudio Arcuri****

Orthodontic treatment in a young patient with Beckwith-Wiedeman Syndrome (BWS) is reported and a multidisciplinary approach to the management of this syndrome is reviewed. The patient presented with a tendency to Class III malocclusion, an open-bite and a slight macroglossia, which was treated at an early age by glossotomy. It was decided to monitor growth without treatment and to wait for the best time to begin therapy. It was based only on functional rehabilitation, without any fixed appliances, in which optimum intercuspation of the teeth and the skeletal Class I relationship was achieved and maintained after the retention period of three years. In conclusion the treatment of BWS patients requires a multidisciplinary approach that includes orthodontics, orthopaedics and surgical intervention. It is also necessary to underline the significance of diagnosis at an early age and timely treatment to reduce the development of dento-skeletal alterations.
J Clin Pediatr Dent 27(4): 377-380, 2003

INTRODUCTION

The Beckwith-Wiedeman Syndrome (BWS) is a rare genetic pathology linked to an alteration on the short arm of chromosome 11, specifically on 11p15.[5] band, though the process responsible for clinical manifestation is still uncertain.[1,2] This disease is characterized by various symptoms, among which is a distinct symptomatic triad: Omphalocele-Macroglossia-Gigantism. In addition, other frequent symptoms are: maxillary underdevelopment, cleft palate, closure defects of the abdominal wall, facial nevus flammeus, advanced bone age, visceromegaly, nephromegaly, combined immunodeficiency and hypertension, mental retardation resulting from neonatal hypoglycemic attacks, and a high propensity of malignant childhood neoplasms.[3-8] To date, there are more than 500 documented cases with additional clinical symptoms still added to those mentioned above. The BWS has an incidence that fluctuates in approximately 1:13.700 subjects, and is non-specific for sex.[9]

One of the most common features of BWS is macroglossia;[10,11] the increased volume of the tongue tends to seek space in an insufficiently wide oral cavity, leading to a distinctive appearance of a Class III skeletal malocclusion with a protrusion and increased length of the mandible.[12] From an orthodontic point of view, BWS patients have to undergo a double therapeutic treatment. First, there is the surgical reduction of the tongue to eliminate the principal cause of odontogenic problems; and then there is orthodontic treatment to rebalance neuromuscular function in order to modify the dynamic space of the tongue and the occlusal balance of the upper and lower arches.

In practice, the maxillofacial alterations that BWS patients may exhibit require a multidisciplinary approach. This multidisciplinary approach may require: surgery, orthopaedic treatment, functional rehabilitation and speech therapy. The goal is to recover not only a correct position in the oral cavity for global improvement of physiological activity, but also providing proper neuromusculoskeletal inter-dynamics.

CLINICAL ASPECTS

One of the most salient signs of BWS is macroglossia, which is present in 97% of the affected individuals, though it is not an obligatory manifestation of the disease.[11,13] It usually takes the form of a large and

* Dott. Aldo Giancotti, Assistant Professor, Department of Orthodontics, Univeristy of Rome "Tor Vergata", Ospedale Fatebenefratelli, Isola Tiberina.
** Dott. Gaia Romanini, Postgraduate resident, Department of Orthodontics, Univeristy of Rome "Tor Vergata", Ospedale Fatebenefratelli, Isola Tiberina.
*** Prof. Raffaella Docimo, Associate Professor, Department of Pediatric Dentistry, Univeristy of Rome "Tor Vergata", Ospedale Fatebenefratelli, Isola Tiberina.
**** Prof. Claudio Arcuri, Associate Professor, Department of Oral Pathology, Univeristy of Rome "Tor Vergata", Ospedale Fatebenefratelli, Isola Tiberina.

Send all correspondence to Dr. Aldo Giancotti, Viale Gorizia 24/C, 00198 Rome, Italy.

E-mail: giancott@uniroma2.it

uniform increase in the length and width of the tongue, and differs in degree of enlargement from case to case.[8,14] The causes of macroglossia are associated with an hypertrophy of the muscle fibers, because of an increase size in the fiber cells, shown by the histological examinations on biopsies of lingual tissues from BWS patients.[15,16] Frequently, it is evident at birth, while less often it appears as a subordinate symptom that occurs during early childhood, with a frequent, but not total, spontaneous regression with age.[17,18]

Orofacial alterations associated with macroglossia can affect musculoskeletal growth. Indeed, the lack of space in the oral cavity obliges the patient to keep the mouth and the mandible in an advanced position. Initially, the result is a tendency to Class III profile with an obtuse gonial angle. It is not caused by a true prognathic mandible, but by the rotation and forward displacement of the mandible in opening the mouth, with the consequence of retroinclination of the inferior incisors and anterior open-bite.[13-15] This situation represents the adaptation of the facial skeleton to an enlarging tongue.[4]

However, in time, the protracted action of the tongue has a direct influence on the surrounding bony structures, inducing the mandible to grow in a disorderly manner to accommodate the tongue more comfortably in the oral cavity, and ultimately leading to an actual skeletal Class III.

The results of macroglossia are loud breathing, possible obstruction of the upper airway passage, under nourishment from difficulties in introducing and chewing food, speech impediments and tongue infections from constant exposure to the outside ambient.[14] Additionally, there are the consequential psychological problems caused by the quasi-retarded features that accompany BWS patients.

The indications for tongue reduction are few. Surgical intervention is suggested only when the tongue protrudes beyond the labial seal, undermines skeletal growth and position of the teeth and hinders the pronunciation of words causing a speech impediment.[15,19-21] We suggest that glossotomy should generally be performed at an early age, usually within the first two to three years, to prevent the aberrant function derived from macroglossia that can modify the quality and quantity of prognathic mandible growth. In such cases, surgical techniques may be chosen from four different approaches (wedge incision, side incision, amputation of the tip, or a combination of these), each of which considers the individual differences.[20]

CASE REPORT

The patient was diagnosed as having Beckwith-Wiedeman Syndrome at birth, and she came under our care and observation at the age of four. Her parents appeared healthy, and without any known past and present family history of BWS on either side. The omphalocele was diagnosed during the pregnancy through an ultrasound exam and was confirmed at birth with the concomitance clinical manifestation of macroglossia.

The objective examination of the patient showed the presence of a large omphalocele, macroglossia (Figure 1), symmetric facies, asymmetric anomalies of the pavilions of the ears, cylindrical chest, physiological MV, red skin and red visible mucosa. In reference to the cardiovascular system, the tones were clean and sound. To sum up the conditions of the patient, she was in good health and her nutritional status was acceptable.

A previous surgical operation was performed two days after her birth to remove the umbilical cord, which was the seat of the omphalocele. After being discharged, the patient was checked regularly; however, she appeared to be in pain and the parents reported that the child had breathing difficulties especially at night. Also, the child suffered from snoring and apnoea, particularly when lying on her back or when she has a cold, which was attributed to the growth in volume of the tongue. So, the patient was hospitalised for a second operation at the age of 13 months, and the anterior third of the tongue was removed. There were no post-operative complications, and at one month after surgery, the remaining scar tissue of the tongue was minimal.

The patient came under our observation at 4 years of age, with an evident protruded profile, open bite, macroglossia and a dental and skeletal Class I with a tendency to Class III malocclusion. It was decided to monitor the growth of the patient without any therapy until shedding of the deciduous teeth. The patient was seen regularly up to the age of seven, where she presented with the following orthodontic situation: an essentially dolichocephalic face with concave profile, an abnormal development of the soft tissues with a protruding lower lip, a marked growth of the third medium of the face, an apparent maxillary retraction with mandibular protrusion, an increased nose-labial angle, tendency to Class III malocclusion and open bite (Figures 2, 3, 4).

Since the above clinical picture remained unaltered during early adolescence (she was now 13 years of age) the treatment plan was initiated and based only upon functional orthopaedic therapy, without the help of any fix appliance (Figures 5, 6).

This was done to improve the neuromuscular function, to exploit the advantages of growth and to keep the eruption of the teeth under control. But, most importantly, the least invasive therapy was chosen to avoid further psychological trauma to the patient, since she was already under strain from having had contact with so many doctors and therapies.

The treatment protocol included two stages: (1) a rebalancing of the muscular function with a reduction of the dynamic space of the tongue, and (2) accomplishing a proper occlusion. At the end of these two therapeutic procedures, the patient had a Class I skeletal and dental relationship, an optimal occlusion and a total elimination of the open bite deformity (Figures 7, 8).

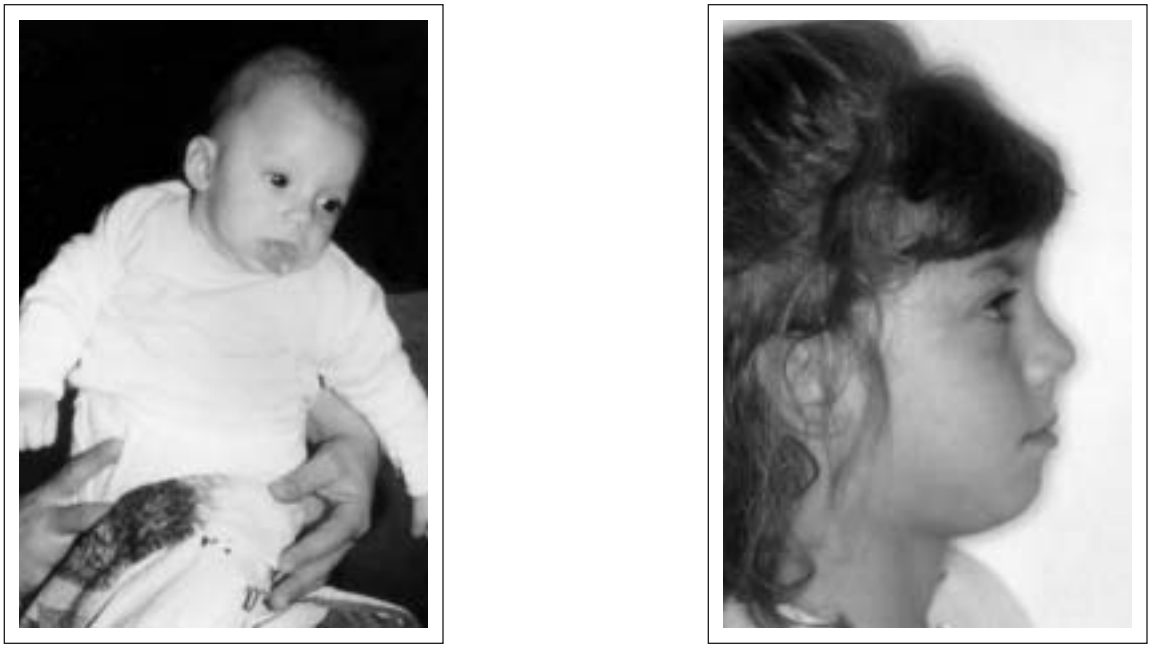
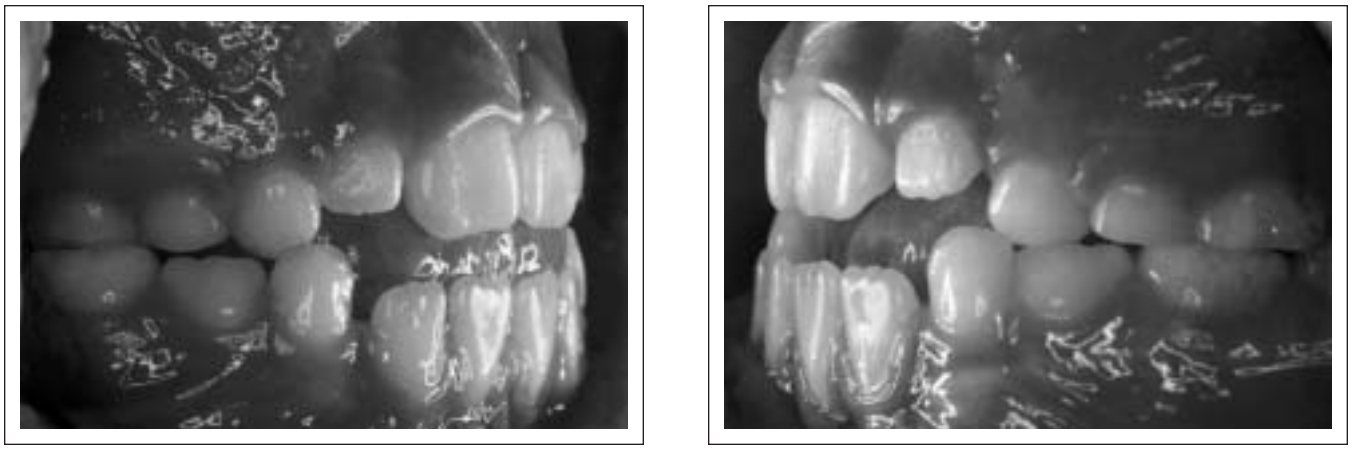
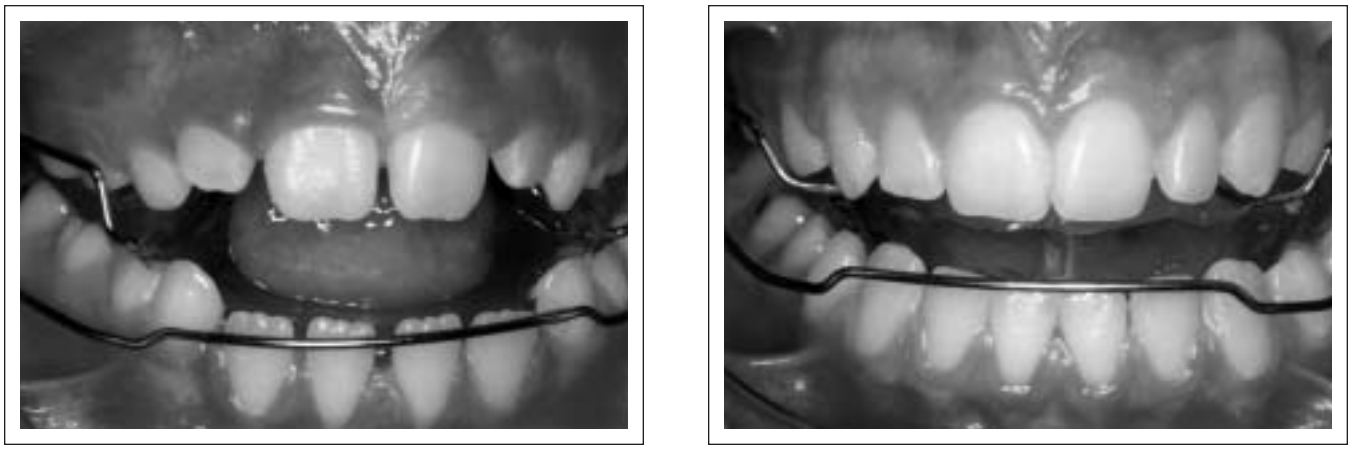


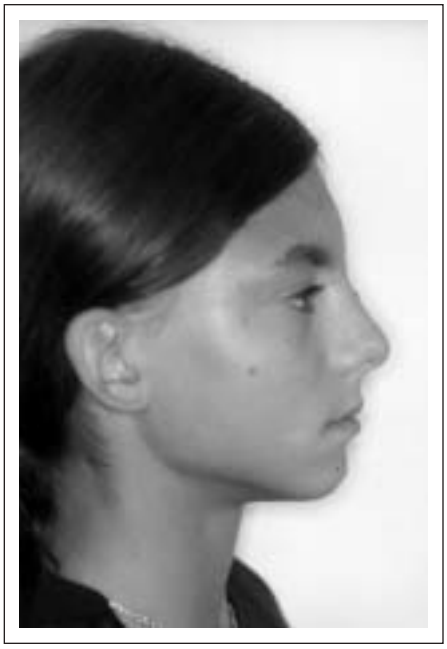
Figure 1. The patient at 4 months of age. It is evident the protruding tongue out of the oral cavity.

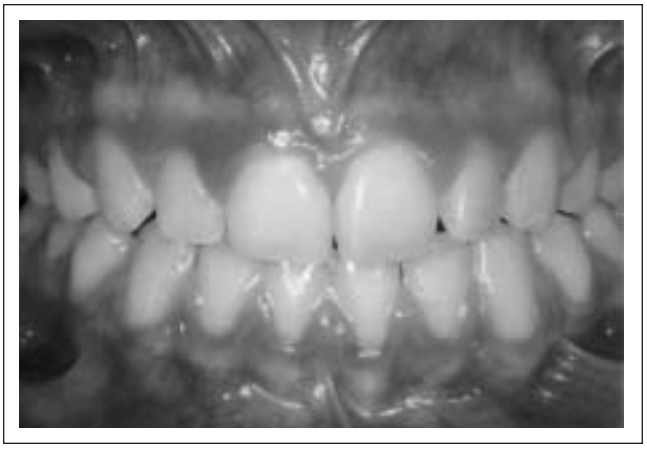


Figures 3-4. Intraoral photographs with evident open-bite.



Figures 5-6. Orthopaedic appliances to reduce the tongue's dynamic space and to improve the neuromuscular function.





Figures 7-8. Frontal and intraoral view at the end of therapy.

Furthermore, a lateral cephalometric radiograph was taken three years after the end of the therapy, showing that the above results were maintained.

DISCUSSION

The therapy for BWS requires a multidisciplinary approach, in which both surgery and orthopaedics supplement one with the other to recover the correct position of the tongue in the oral cavity and to attain proper neuromuscular function. Although the treatment often requires long therapeutic cycles that can psychologically traumatize the patient, it does produce good results in obtaining a stable occlusion and a satisfactory condition of musculoskeletal dynamics.

The case report underlines the significance of early diagnosis with surgical intervention for the macroglossia to improve an abnormal dento-skeletal pattern. Glossectomy, done in the first months after birth, eliminates or at least reduces, the main cause of the dento-skeletal deformities.

REFERENCES

1. Bischoff FZ, Feldman GL, McCaskill C, Subramanian S, Hughes MR, Shaffer L. Single cell analysis demonstrating isodisomy and Beckwith-Wiedeman syndrome. Hum Mo Genet 4: 395-99, 1995.
2. Richard III CW, Boehnke M, Berg DJ *et al*. A radiation hybrid map of the distal short arm of human chromosome 11 containing the Beckwith-Wiedeman and associate embryonal tumor disease loci. Am J Hum Genet 52: 915-21, 1993.
3. Elliott M, Bayly R, Cole T, Temple IK, Maher ER. Clinical features and natural history of Beckwith-Wiedeman syndrome: presentation of 74 new cases. Clin Genet 46: 168-174, 1994.
4. Engström W, Lindham S, Schofield P. Wiedeman-Beckwith Syndrome. Eur J Pediatr 147: 450-7, 1988.
5. McManammy DS, Barnett JS. Macroglossia as a presentation of the Beckwith-Wiedeman syndrome. Plast Reconstr Surg 75: 170-6, 1985.
6. Weng EY, Mortier GR, Graham Jr JM. Beckwith-Wiedeman syndrome: an update and review for the primary paediatrician. Clinical Paediatrics 6: 317-26, 1995.
7. Wiedeman HR. Tumor and hemihypertrophy associate with Wiedeman-Beckwith's syndrome. Eur J Pediatr 141: 129, 1983.
8. Rimmel FL, Shapiro AM, Shoemaker DL, Kenna MA. Head and neck manifestations of Beckwith-Wiedeman syndrome. Otolaryngol Head Neck Surg 113: 262-5, 1995.
9. Thornburn MJ, Miller CG, SmithRead CH. Exomphalos-Macroglossia-Gigantism syndrome in Jamaican patients. Am J Dis Child 119: 316-21, 1970.
10. Beckwith JB. Macroglossia, omphalocele, adrenal cytomegaly, gigantism and hyperplastic visceromegaly. Birth Defects 5: 188-196, 1969.
11. Friede H, Figueroa AA. The Beckwith-Wiedeman syndrome: a longitudinal study of the macroglossia and dentofacial complex. J Craniofac Genet Dev Biol Suppl 1: 179-87, 1985.
12. Kamogashira K, Ito T, Nakagawa M, Kawagoe H, Ichikawa K, Matsumoto M. Orthodontic findings of a case of Beckwith-Wiedeman syndrome. J Jpn Orthod Soc 43: 564-72, 1984.
13. Malagola C, Barbato E, Fordellone P. Sindrome di Beckwith-Wiedeman: un raro caso. Dental Cadmos 1: 74-9, 1990.
14. Ruffino S, Frigerio M, GalvagnoG, Bosco G. Il problema della macroglossia nel lattante affetto da BWS: descrizione di un caso. Minerva Stomatologica 34, 1985.
15. Preda GE, Pasetti P, Caggiula S. Sindrome di Beckwith- Wiedeman: presentazione di un caso clinico. Dental Cadmos 16: 104-7, 1989.
16. Slater AD. Primary macroglossia. Clin Pediatr 7: 357- 63, 1968.
17. Farkas JG, (ed) Anthropometry of the head and face. 2nd ed. New York, Raven, p.244-333, 1994.
18. Moore ES *et al*. Heterogeneity in Wiedeman Beckwith Syndrome: anthropometrics evidence. Am J Med Gen 90: 283-90, 2000.
19. Sokoloski PM, Ogle RG, Waite DE. Surgical correction of macroglossia in Beckwith-Wiedeman syndrome. J Oral Surgery 36: 212-15, 1978.
20. Rizer FM, Schechter GL, Richardson MA. Macroglossia: etiologic considerations and management techniques. Inter J Paediatric Otorhinolaryngology 8: 225-36, 1985.
21. Kveim M, Fisher JC, Jones KC, Gruer B. Early tongue reduction for Beckwith-Wiedeman macroglossia. Ann Plast Surg 14: 142-4, 1985.