

MACROGLOSSIA AS A CAUSE OF ATYPICAL SWALLOWING: COMPARISON OF EVALUATION AND LOGOPEDIC TREATMENT BETWEEN BECKWITH-WIEDEMANN AND DOWN PATIENTS

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Abstract. Atypical swallowing is the persistence of childlike deglutition at the end of dental eruption. One of the main causes is macroglossia, that is the abnormal enlargement of the tongue. The treatment is logopedic and/or surgical. Children with macroglossia have an increased incidence of respiratory diseases and infections, as well as malocclusions, articulatory defects and aesthetic damage. In this study we focused on two genetic syndromes with macroglossia: Beckwith-Wiedemann Syndrome (BWS) and Down Syndrome (DS): 7 patients were evaluated for logopedic treatment: 3 are affected by BWS, 2 are affected by DS. In addition, 2 patients with isolated atypical swallowing were included in the study to emphasize problems connected with atypical swallowing. All the patients underwent a global examination and a personalized logopedic therapy scheme was planned. With the exception of one of them who was lost to follow up and who did not continue with the exercises the speech therapist had recommended, all the children showed good response and compliance with remarkable improvements, thus proving the importance of an early, constant and intensive logopedic treatment.

Key words: atypical swallowing, macroglossia, genetic syndromes, logopedic treatment

INTRODUCTION

Swallowing is the skill to transfer solid, liquid and gas substances from the external environment to the stomach. Swallowing pathology can be divided into four big chapters: (1) malformations, (2) defects in the passage from childlike deglutition to the adult one, (3) adult swallowing pathology, (4) dysfunctional diseases. In particular, the second form is known as atypical swallowing and it is characterized by the persistence of childlike swallowing at the end of dental eruption (at the age of seven years), when normally the adult form appears [1-2]. One of the main causes of this alteration is macroglossia.

Macroglossia is the abnormal enlargement of the tongue, that at rest leaks over the teeth. It can be divided into: (1) true macroglossia, when the increased volume is caused by histological alterations, and (2) relative macroglossia, when tongue volume is normal but there is an insufficient space in the oral cavity. Diagnosis is clinical, treatment is logopedic and surgical, according to tongue dimensions, risk of complications and patient compliance. Complications, if not identified and treated, can be lethal. In fact, children with macroglossia have an increased incidence of respiratory diseases, especially Obstructive Sleep Apnea Syndrome (OSAS) and an increased risk of infections of the upper and lower respi-

ratory tract, ear and Central Nervous System due to oral breathing. Moreover, there are dental malocclusions, errors in the articulation of words and aesthetic damages that can create many problems in the social relationships with other children [3-4].

In this study we focused on two causes of macroglossia: Beckwith-Wiedemann Syndrome and Down Syndrome.

Beckwith-Wiedemann Syndrome (BWS) is a rare genetic disorder with an incidence of 1/13700 births. The syndrome is due to an anomaly in the imprinting region on chromosome 11p15. BWS is characterized by prenatal and postnatal overgrowth, macroglossia, anterior abdominal wall defects, ear anomalies, facial *nevus flammeus*, organomegaly, genitourinary abnormalities, hypoglycemia, microcephaly, hemihyperplasia, heart dysfunction, mental retardation and increased risk of neoplasms, especially Wilms Tumour. Diagnosis is clinical and it is confirmed by cytogenetic and molecular analyses. Macroglossia is observed in 97% of BWS cases, it is a true macroglossia and it represents one of the main diagnostic criteria. In the first year of life there is a high risk of respiratory and alimentary disorders, but in the following period there is often an improvement and the prognosis *quoad vitam* is good, even if morbidity remains elevated [5-8].

Down Syndrome (DS) is a frequent condition with

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an incidence of about 1/800 births, caused by a chromosomal aberration. Diagnosis of DS is clinical and it is confirmed by karyotype analysis [9-11]. Macroglossia is relative, in the context of an overall skull dysplasia. In fact, between the 6th and 12th week of gestation an overall reduction in eyes, brain, hands and heart development takes place. A study performed at the Radiology Department of Cincinnati Children's Hospital showed reduced skull and facial parameters with tongue proportionally larger in relation to them in patients with Down Syndrome vs. healthy children [12]. In DS macroglossia leads to atypical deglutition, breathing difficulties and increased risk of infections and contributes significantly to three orthodontic alterations: (1) anatomic deep bite, (2) functional openbite and (3) class III malocclusion [13].

EVALUATION AND LOGOPEDIC TREATMENT OF MACROGLOSSIA

Evaluation

The correct evaluation of patients is global: the speech therapist has to consider expression and comprehension of the language, reading, writing and speaking abilities, voice technical parameters like frequency and timbre, graphic capabilities as well as the psychological aspects of the child's behaviour. Logopedic evaluation is structured in three moments: (1) collection of information by talking with parents (anamnesis) and evaluation of clinical data written by the paediatrician who sent the patient; (2) observation of the patient; (3) discussion with parents about the results of the examination and the therapeutic program.

The first step consists in the voice registration that is fundamental to have an idea of its quality. The second step is the physical examination, both at rest and dynamic, which has to include also a phonetic examination. Clinical examination at rest permits the evaluation of the oropharyngeal morphology: shape and dimensions of the tongue, presence of macroglossia, ogival palate, short lingual frenulum, hypersalivation, tonsillar hypertrophy, nose stenosis. Dynamic examination is useful to observe oral, facial (masseter, temporal and mentalis) and velopalatine muscles function, lips and tongue mobility, temporo-mandibular joint and presence of dyspraxia and apraxia. The diagnosis of atypical swallowing due to macroglossia is made according to the results of anamnesis and clinical examination. Afterwards the speech therapist requests information regarding feeding (breast-feeding or bottle-feeding), sucking (dummy and/or finger, until what age), dental development, feeding and sleep habits, presence of infections, allergies, diseases of tonsils or adenoids and about any surgical treatment such as tonsillectomy and/or adenoidectomy. It is fundamental to remember that logopedic evaluation has to be global

and not limited to macroglossia, because this is a single aspect of a wider clinical status, especially if the patient is affected by a genetic syndrome [14-15].

Treatment

After the evaluation and before starting the treatment, the patient undergoes other medical examinations: otorhinolaryngological, dental, audiometric and phoniatric, he then is subjected to a radiologic exam of the oral cavity and to polysomnography, if necessary. Patients should undergo the first examination during their first year of life, in order to start the treatment as soon as possible. Until the seventies, treatment methods did not include a global vision, but focused on the specific pathologic aspect. On the contrary, in the last ten years, experts have begun to consider the oral function in an unitary way, evaluating breathing, feeding, sucking, swallowing, chewing, speech articulation, taste, and facial expression altogether. The treatment has to be personalized and adjusted according to the global characteristics of the patient, and not only based on the disease. The rehabilitative intervention in newborns and children with macroglossia focuses at first on lips and cheeks tonicity, on the increase of the strength of perioral and oral muscles and on control and coordination of tongue movements. Once these objectives are achieved, the speech therapist can start working on his main objective: the reduction of tongue protrusion, which, in turn, leads to a reduction of dental, feeding and breathing alterations and of the risk of infections. The speech therapist has to work, from the first year of a patients' life, on pre-speech abilities, that are the basis of the articulated language [16].

Sucking can be difficult in new-borns with macroglossia, especially in DS patients, because of the poor muscle tissue: an improvement can be achieved by teaching the mother specific positions that help the baby to concentrate his energy only on sucking. The use of feeding bottles is not recommended because the tongue takes a low position, and after the second year dummies should be avoided, too. The intervention has to allow the patient the acquisition of all the fundamental motor skills before he is 3, because after this period the modification of anatomy and habits becomes very difficult. In fact, if the patient undergoes the first examination when he is older, the therapist has less possibilities of intervention and, in many cases, an orthodontic apparatus or a surgical intervention may become a necessity.

There are a large variety of exercises, divided into 3 phases. At the beginning the activity of metacognition is crucial in order to help the patient to know the structure and the function of his own face, lips, tongue, mouth and nose; the second phase consists in the passive functional training, with the speech therapist performing exercises on the patient's face; the third phase consists in the active functional training, with the patient doing exercises by himself [16].

The utility of odontostomatologic rehabilitative treatments in children with DS and/or mental retardation has been demonstrated. One of the most useful instruments in this field are Castillo-Morales plates; they are mobile devices that can stimulate the movements of the tongue, lips and chewing muscles and facilitate the closure of the mouth correcting the incorrect tongue and lips position through stimulation elements. The aim is to improve nasal breathing and to acquire physiologic swallowing and sucking [17-18].

PATIENTS, METHODS AND RESULTS

This study is based on a series of 7 patients presented for evaluation to the Department of Pediatrics of the University of Siena: 3 of them with diagnosis of BWS, 2 with diagnosis of DS and 2 with isolated atypical swallowing. The study has a wide age range (between 4 and 14 years of age); 5 patients presented true or relative macroglossia, 4 patients presented atypical swallowing (the last three patients weren't evaluable because they were less than seven years old); 7 patients had hypotonic perioral muscles, 6 patients presented dyslalic speech (the seventh patient wasn't evaluable because he was too young), 3 patients presented mental retardation of different degrees. The patients were submitted to logopedic evaluation and an individualized therapeutic schedule was planned. BWS and DS patients underwent follow up examinations every 3 months; patients with isolated atypical swallowing underwent logopedic examinations and treatments only, since the diagnosis was made by a dentist when they were 7. 1 patient didn't continue the therapy and she was lost to both medical and logopedic follow up; 1 patient started the therapy with little delay; the other 5 patients correctly followed the planned schedule.

Medical examination comprised physical, laboratory and instrumental exams. An accurate familiar anamnesis was made, especially focusing on the research of any genetic diseases. We evaluated parameters related to pre- and postnatal period. Logopedic evaluation consisted in: registration of voice parameters (intensity, height, timbre, vocal attack), phonetic and articulation exam (isolated phonemes and phonemes at the beginning, in the middle and at the end of the words), gestural communication, oral communication (vocalise, holophrasis, spontaneous contracted sentence, spontaneous structured sentence, correct use of articles and verbs, correctness of speech, lexical richness), oral comprehension (execution of gestures on request, simple exercises, identification of objects, figures and actions), evaluation of graphic, writing, reading, rhythmic and perceptive levels (association and abstraction capabilities; chromatic, time and space, direction and orientation perception, identification of measures and forms, knowledge of body scheme and denomination of vari-

ous body's components), global and fine motricity, behavioural scheme. Great attention was paid to the execution of buccal-lingual movements, to the observation of perioral muscles, tongue, lips and jaw and to the evaluation of breathing, swallowing and chewing abilities. Once the evaluation was completed, an individualized therapeutic schedule was planned.

Patient 1, female (twin birth). She presented at birth the typical BWS features. Clinical diagnosis was confirmed by genetic studies. The patient presented mild mental retardation, medium hypoacusis and true macroglossia. At the age of 3 she underwent surgery to remove a Wilms Tumour. A speech therapist evaluated the patient when she was 4 and 6 and specific exercises were planned, but the parents decided to interrupt the follow up. In February 2009 the patient was contacted by the Department of Pediatrics and she was evaluated in March 2009: the parents, who had not accepted their daughter's genetic disease, reported that she had never done any logopedic exercise and in 2008 the dentist had to implant Castillo-Morale plates, without receiving any benefit. During the examination emerged the permanence of all the defects presented in previous examinations and atypical swallowing was detected. The patient has a progressive disease and atypical swallowing worsened through the years.

Patient 2, female, presented at birth the typical BWS features. Clinical diagnosis was confirmed by genetic studies. In October 2009 she was examined by a speech therapist who identified the presence of macroglossia and a mild defect of language, swallowing and chewing. She showed a spontaneous improvement of macroglossia during the growth, but some articulatory, swallowing and aesthetical disabilities remained, with an high risk of developing atypical swallowing in the future; for this reason she started, when she was 4, logopedic therapy to stimulate lips and tongue hypotonic muscles and some praxis exercises for tongue and lips were recommended to better contain the tongue into the mouth. After only few sessions of therapy we observed an improvement of her conditions, with a reduction of tongue protrusion at rest.

Patient 3, male (twin birth), presented at birth some of the typical BWS features. At the age of 10 months he underwent pediatric, otorhinolaryngological, psychological and logopedic examinations. The presence of macroglossia was detected. No alterations of feeding and breathing were present. Some praxis exercises were recommended to strengthen lips and to improve tongue motility. He achieved remarkable improvements in concentration, behavioural, attention and phonatory abilities, tongue protrusion was reduced, muscular tone became stronger and his compliance improved.

Patient 4, male, presented at birth the typical DS characteristics. Clinical diagnosis was confirmed by genetic analyses. At the age of 3 years he underwent the first psychological and logopedic examination and a logopedic treatment was started. At the beginning he did

not collaborate, but during the following examinations he established a good interaction with the therapist and his interest for the exercises improved. Exercises to correct macroglossia and to improve language, attention and concentration capabilities were made. As for macroglossia, great attention was paid to the stimulation of buccal-facial praxis. He achieved remarkable improvements in concentration, behavioural, attention and phonatory abilities, tongue protrusion was reduced, muscular tone became stronger.

Patient 5, female, presented at birth many of the characteristics of DS. Clinical diagnosis was confirmed by genetic analyses. She presented a global and heavy hypotonia with a notable difficulty with sucking and a delayed psychomotor development. The presence of macroglossia was detected. At the age of 7 months she had epileptic crises and diagnosis of West Syndrome was made. At the age of 16 months hypothyroidism was diagnosed. The speech was dyslalic and difficult to understand, there was a lack of coordination of oral-facial praxis and the writing was difficult. When she was 7 atypical swallowing was detected. Since the age of 14 months she did logopedic exercises 3 times a week, with very good response in all fields, especially with regard to macroglossia, atypical swallowing and language comprehension.

In this study, in order to emphasize problems connected with atypical swallowing, 2 patients with isolated atypical deglutition were followed during logopedic evaluation and treatment.

Patient 6, female; the diagnosis was made by a dentist at the age of seven years. She underwent a logopedic evaluation, in which specific tests for atypical swallowing were performed with a positive result. The speech therapist planned Garliner myofunctional therapy [19-20] in association with praxis, respiratory and phonetic exercises. The following examinations were made weekly for 2 months, and then every other week, with good compliance and positive results after a few months.

Patient 7, female. The diagnosis was made by a dentist at the age of seven years. She was evaluated by a speech therapist and specific tests for atypical swallowing resulted positive. In addition, she presented phonetic and phonologic defects. Garliner myofunctional therapy was started with good compliance and positive results.

DISCUSSION

The main goal of this study was to prove the importance of early logopedic treatment in patients with genetic syndromes associated with macroglossia: Down Syndrome and Beckwith-Wiedemann Syndrome. This approach allows improvement in the outcome and facilitation of rehabilitation, with particular regard to atypical swallowing. A logopedic approach has been used in these syndromes for a long time, but often only

an evaluation is made and whether treatment schedule isn't planned or the therapy is started too late, or it is followed irregularly and/or incompletely. In fact logopedic treatment has often been left out or considered only a support for surgery.

In our study, despite the small sample, we were able to make interesting observations; first of all the approach has to be individualized according to the characteristics of the single patient. In some cases logopedic therapy can represent an alternative to, and not only a support for, surgery. Surgical treatment is still the main option when it is not possible to contain the tongue into the oral cavity or the tongue protrusion makes breathing more difficult.

Since macroglossia is one of the causes of atypical swallowing, in this study following the evaluation and treatment of patients 6 and 7 with isolated atypical deglutition, sent to logopedic observation when they were seven, was very useful. We evaluated the difficulties of these patients to set up "*de novo*" the mechanism of deglutition, even if there were no other problems and they complied well. In these patients logopedic treatment, started early, corrected the problem completely, while in patients with a worse clinical situation and significant comorbidities it didn't happened. In fact, patient 1, who didn't received a constant, intensive and continuative treatment, showed, at the age of ten, compromised general conditions, with a large hypotonic tongue protruding from the oral cavity and the lips, never stimulated, were hypotonic. Muscle hypotonia (with particular regard to perioral muscles), ogival palate, oral breathing, and evident macroglossia worked together to set up abnormal swallowing. In these conditions therapy was more complicated because we had to treat simultaneously tongue obstruction, muscle hypotonia and the features of adenoid facies. In 2008 Castillo Morales' device was applied but produced very little benefit. As for patient 5 she had a logopedic evaluation at the age of 14 months, she presented global hypotonia and a delayed psychomotor development; she was strictly followed in her treatment and she showed great improvements in using the tongue and in controlling the structures of the oral cavity, even if the speech is still difficult to understand. Analysing these two patients – the most serious in the study – the differences between the two genetic syndromes – BWS and DS – clearly emerge. In fact, patient 1 presented true macroglossia, mild mental retardation, no behavioural alterations, while patient 5 presented relative macroglossia, severe mental retardation and remarkable behavioural alterations. The children, who are around the same age, had very different progression and outcome: patient 5 had a number of problems at the beginning, but early treatment enabled considerable improvements, while patient 1 didn't show any improvements because of the rejection of logopedic therapy: this caused the progression of her deficit through the years. With age, incorrect habits, like oral breathing, got stronger becoming increasingly dif-

difficult to modify even with a correct therapy, because rehabilitation is more complicated than intervention before a wrong habit is consolidated.

CONCLUSIONS

Considering “genetic” as synonymous of “irreversible” is a wrong presupposition and it is an additional penalty for people with Down Syndrome or with Beckwith-Wiedemann Syndrome because they are clearly and unquestionably genetic. For this reason rehabilitative objectives are often very low (too low) and the patients are undertreated or treated too late.

Another historical mistake is that early physiotherapy (which is right) is not joined by logopedic therapy. This fact is justified with the statement: “it is too soon for logopedic treatment”; the truth is that in rehabilitation the concept of “too soon” doesn’t exist, but unfortunately the concept of “too late” does. Early and intensive intervention is the best weapon when a rehabilitative treatment is necessary.

Recently, the attitude of clinicians is changing: in fact they tend to leave out functional rehabilitative treatments such as mobile devices, while they recommend performing physiotherapy oral exercises precociously and perseveringly, in order to set up correct positions and feeding habits. Patients should be evaluated as soon as possible (the best thing would be to evaluate the child during his first year of life) by a team comprising surgeons, paediatricians, geneticists, childish neuropsychiatrists and speech therapists. Each expert should establish short, middle and long term objectives and use his creativity to propose various types of exercises keeping the child’s interest at high levels. Therefore a sensible balance between technicality and creativity is very important, and the final intention has to be care, not only cure.

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