

# REPERCUSSIONS OF SPEECH THERAPY IN SECKEL SYNDROME: STUDY CASE

## *Repercussões fonoaudiológicas na síndrome de Seckel: estudo de caso*

Aline Ferreira de Brito <sup>(1)</sup>, Sílvia Elaine Zuim de Moraes Baldrighi <sup>(2)</sup>

### ABSTRACT

Seckel syndrome, it's rare condition of autosomal recessive hereditary nature of, characterized by severe growth retardation, intrauterine, proportionate short stature, microcephaly with small and weak chin, nose large and curved, in some cases mental retardation, multiple congenital anomalies in the face skull and skeleton, among other malformations. The aim of this study was to identify the phonological manifestations found in this syndrome, from a clinical case report. The assessment speech that addressed: physical description, aspects of stomatognathic functions, the oral and written language, speech and hearing, and cognitive aspects. To complement the speech diagnosis, orthodontic evaluations were performed, otorhinolaryngology and physical therapy. With respect to stomatognathic functions, the child had breathing so oral, chewing with the teeth hold food side, unilateral mastication, with parted lips and exaggerated movements of the perioral muscles, swallowing with tongue interposition and participation of the orofacial muscles. Speak with imprecise articulation and locked due to limited mouth opening. Regarding the voice, the patient had reduced maximum phonation time, high larynx position, nasal resonance, vocal attacks soft and weak intensity. It was not observed any change in oral and written language. The scarcity of studies reporting on phonological manifestations of Seckel syndrome associated with the rarity of occurrence, justifies the interest in performing this study, to collaborate more knowledge on the part of speech therapists and health professionals.

**KEY WORDS:** Speech, Language and Hearing Sciences; Evaluation; Diagnosis

### ■ INTRODUCTION

In recent years, speech pathology and genetics have been a complementary way. This integrated partnership has contributed to the improvement of procedures aimed at the diagnosis, prognosis and intervention of individuals with genetic syndromes and communication disorders as early as possible.

The investigation of the clinical history of syndromic patient should begin by gathering information about the speech-language disorder and a previous history. For the characterization of the complaint, it is important to obtain information

from period pre, peri and post-natal, to describe the manifestation evolved, if it has improved or if the frame remained stable. How the general and specific motor development of the child was, as well as all the familiar one. Information about the development and auditory function are also relevant. This assessment focuses on characterizing the performance of language-related areas, in oral and written forms and in the defining areas of skills and difficulties related to this process, involving the processing of auditory and visual information, speech and oral functions / swallowing<sup>1</sup>.

First described as "dwarfism bird head" by Rudolf Virchow in 1892<sup>2</sup>. Afterwards, Seckel, in 1960, based on two case studies in Chicago and 13 in cases of microcephaly dwarfs, characterized the syndrome as it is currently described in the literature<sup>3</sup>. In 1967, McKusick and colleagues documented this condition in three of 11 brothers and suggested

<sup>(1)</sup> Pontifícia Universidade Católica de São Paulo, São Paulo, SP, Brasil.

<sup>(2)</sup> Universidade Federal de Sergipe (UFS), Aracaju, SE, Brasil.

Conflict of interest: non-existent

that the inheritance was probably recessive autosomal <sup>4</sup>, being characterized by a genetic mutation on chromosomes 3, 18y and 14 and its frequency less than 1 in each 1,000,000 born<sup>2,5</sup>.

This syndrome equally affects both men and women, without presenting geographic<sup>2</sup> or ethnic dominance, characterized by intrauterine retardation, low birth weight, microcephaly, dwarfism, "bird head" and frequent higher airway infections<sup>6</sup>. Mental retardation may be present, but at a much lower degree than the expected one, considering the skull size<sup>7</sup>. Those patients are generally nice and friendly, hyperkinetic and easily distracted children <sup>2</sup>.

The craniofacial characteristics represent a turning point in the differentiation of other syndromes. Seckel syndrome (SCKL) presents genotypic<sup>8</sup> and phenotypic heterogeneity. Due to this fact, some patients have additional clinical findings, such as large bulging skull, cleft palate, atresia palate, abnormalities in dentition, among these, dental crowding may occur due to the narrowing of the jaw and maxilla<sup>6</sup>, ocular manifestations, pancytopenia, chromosomal instability, limb anomalies, dislocation of the femoral head, and scoliosis gastrointestinal<sup>7</sup> malformation.

The diagnosis can be made by prenatal ultrasound, clinical features, radiological bone age retardation, hip dysplasia and dislocation of the radial head and the weight ranges from 450 to 1600g and size between 33-43 cm.

In this scenario, the audiologist plays a vital role in working with individuals with craniofacial malformations and syndromic. The performance of this professional multidisciplinary team together contributes to diagnosis and previous intervention<sup>9</sup>.

Thus, the aim of this study was to identify and analyze the speech language expressions found in Seckel syndrome, from the report of a clinical case.

## ■ CASE PRESENTATION

This study was approved by the Ethics Committee of Universidade Federal de Sergipe, CEP / UFS with No. CAEE-0157.0.107.000-10.

This is a case report of a child nine years old at the date of assessment, male, evaluated and diagnosed with SCKL at nine months old by a geneticist at the National Health System of the State of Sergipe.

Information regarding the medical history of the individual was collected from medical records of the University Hospital of Universidade Federal de Sergipe (UFS). Data relating to pre, peri and post-natal and past medical problems were considered important for the delineation of the syndrome, among the main findings include: the low-set hair,

short neck, and large prominent eyes, dwarfism in the pre, peri and post-natal, presence of microcephaly and episodes of choking. This information was collected by reading the medical record.

In the initial interview with the mother, the child is the second child of healthy, non-consanguineous parents. Maternal age at birth was 39 years old. The child was born vaginally at term, weighing 2.130g, measuring 42 cm, head circumference 34 cm, it was not necessary to stay in the incubator.

After being diagnosed with SCKL the child was referred to the University Hospital of the institution, due to delayed growth, as presented short stature and low weight.

At nine months old, the child had frequent seizures, when drug treatment started with gardenal-70 drops / day in order to control the frame. This treatment was extended to eight years old, a period in which happened the last convulsion.

The child had recurrent infections frames of upper airway, with loss of consciousness and gastroenteritis, being hospitalized for a long period of time. On that occasion, he/she was admitted to hospital more than once a month. These facts were extended to eight years old, but less frequently.

Clinical histories were reported no developmental delays. The child began talking at 12 months old and he/she was always well understood and does not require use of gestures to effect communication. As for the insertion in the school environment, at first he/she had difficulty in adaptation, yet managed to overcome and currently he/she develops well the school activities, so that there are no complaints from the teacher, or parent in relation to learning.

The speech complaint was due to change in breathing pattern viewed and forwarded by pediatric dentistry.

## ■ RESULTS

With regard to the aspects of independent judges, the synthesized protocol (intra-oral examination, oral function and mobility) MBGR (2009) <sup>10</sup> (Annex A) During the extra and intraoral examination, it was observed : long type with the most inferior facial, hard palate with increased depth and narrow width, palatal veil symmetrical with good mobility and functionality, usual uvula; habitual posture parted lips, length of the upper covering only half of the upper incisors (labial incompetence), both with normal color, but with external mucosa showing cracks and dryness; little sharp mentolabial angle; adequate labial frenulum; habitual position of the interdental tongue with decreased tone and tremor, extension of short frenulum, but not limiting. Ment with increased tone. Mixed dentition with

good dental conservation, with presence of anterior, upper and lower crowding, Class I canine and molar. Concerning mobility difficulties presented to the tongue, lips and cheeks. With respect to the temporomandibular joint (TMJ), unable to perform jaw movements of laterality, but presented isolated movement of opening and closing the mouth, with restricted movement.

With respect to oral functions, breathing manifested by the average type / superior oral mode, with possibility of nasal use between 1 and 2 minutes. The chewing pattern occurred bilaterally simultaneously with lateral incision, the labial closure occurred with the systematic parted lips, the muscular contractions of the perioral muscles were exaggerated, showed increased velocity, reported no pain when chewing but also showed no noises in the temporomandibular joint (ATM). During swallowing, we observed partial lip lock with positioning of the tongue between the teeth during swallowing of liquid and pasty consistency and sharp contraction of the mentalis and orbicularis muscle, inadequate food and containment of this head movement. Choking was not observed in any of the evaluated consistency (solid, liquid and pasty), change in facial color, dyspnea or cough and / or vocal change after swallowing.

The phonological inventory could be assessed through the infantile language test in the areas of phonology, vocabulary, fluency and Pragmática<sup>11</sup>, (Annex B) consisting of two tests to verify the phonological system: imitation and naming and spontaneous speech. The analysis was performed by two students of speech and an experienced professional. And just observed the production of consonant with the phoneme / r / (tepe) and simplification of final consonant phoneme / R / changings<sup>11</sup>.

The articulation of words took inaccurately and with reduced mouth opening, making most often impaired intelligibility due to restriction of mouth opening, as well as reduced loudness. During the speech he/she had excessive accumulation of saliva at the corners of the lips<sup>10</sup>. And no disfluency was observed.

The use of oral language as communicative resource proved to be suitable. The patient started and continued dialogue and had oral emissions with some words with a bit of intelligibility. Regarding oral language it was not observed syntactic changings, semantic, phonological and pragmatic behavior.

It was observed that the patient had comprehension and vocabulary adequate for her age, with properties that a text must present to be meaningful<sup>12</sup>. As the cognitive aspects, the child presented notions of the body, colors, shapes and

sizes, he/she presented no difficulty appointments. The presence of functionality and symbolic playing at the stage of complex elaboration was observed. He/she presented spatial orientation notion, did cognitive relationship with amount and performed categorisations. He/she didn't present difficulties in abstract logical reasoning and memory difficulties.

During the writing test, it was used dictation and balanced assessments of reading fluency and comprehension. For the application of balanced dictation it was given a white sheet of paper and the words were said. If errors occurred the child was asked not erase them and write them beside the correct word<sup>13</sup>.

In the assessment of silent reading, time and the articulatory support using were observed. The assessment of oral reading was performed with the same text used in silent reading when they were observed: fluency, the use of time, volume, punctuation, omission or addition of words and / or phonemes, repetition of word or phrases and replacing words or phonemes<sup>14</sup>.

In written language it was not observed alteration in print, dictation, spontaneous writing of words, single words and phrases, as well as silent reading and reading comprehension. Regarding to reading and writing learning, the researchers came to the school of the child, according to the teacher's reports, it accompanies the class, not presenting learning difficulties yet, despite he/she is easily distracted.

During voice evaluation, it was observed that the child had maximum phonation time for the vowel / e / voiceless: 3.9 seconds-(s), fricative / s / : 6,2s, vowel / e / : 3 8s, fricative / z / : 3,9s; relationship / and / and voiceless / e / : 0.1 milliseconds; larynx was elevated position; nasal resonance, absence of hoarseness, roughness, breathiness, asthenia and strain in the voice (RASATI / 0); smooth vocals and weak intensity 10,21 attacks.

Audiological testing showed hearing thresholds within normal limits, presence of acoustic reflexes and tympanometry type or The Air, indicative of stiffness of the tympanic-ossicular system curve.

It may be noted that throughout the diagnostic process, the child had difficulty sustaining attention during the execution of some proposed activities. The same was dispersed easily with any external noise, with existing materials or objects in the evaluation room.

## ■ DISCUSSION

The SCKL is a spectrum of craniofacial and vertebral abnormalities. The craniofacial characteristics, according to the authors studied

above<sup>6,7</sup> represent a turning point in the differentiation of other syndromes<sup>2,8</sup>. The study of this condition is still scarce due to its complexity and scope of clinical aspects. In fact, there are few studies to correlate their findings with the literature, mainly with the area of speech.

Collected data from the patient's medical records revealed that the diagnosis of SCKL was given because of the present alterations as: intrauterine retardation, low birth weight, microcephaly, dwarfism, frequent convulsions with unconsciousness, recurrent respiratory infections, gastroenteritis, ocular manifestations, membership changings and bone age delay<sup>2,6-9</sup>.

According Kilinc et al.<sup>7</sup>, individuals with this syndrome may be mentally retarded, but to a much smaller degree than the expected one, taking into consideration the size of the skull. In the case studied, it was not, so far, described changings related to cognitive impairment and mental or intellectual retardation.

Regarding craniofacial characteristics, the child is dolichofacial with atretic palate, abnormalities in dentition due to the mandible and maxilla narrowing, anterior crowding, upper and lower, Class I canine and molar<sup>6,7,15</sup>.

During the diagnostic process, it was observed that the child studied was pleasant, friendly and easily distracted. To Gómez<sup>2</sup>, these characteristics are present in these children.

Despite the literature does not present findings to correlate with speech therapy SCKL manifestations, then, child characteristics are described, based on the normal range quoted by the authors studied.

In her studies, the author<sup>16</sup> said stomatognathic functions are altered when they occur as follows: oral breathing mode, costodiaphragmatic type; with chewing, chewing pattern simultaneous bilateral lateral incision, with parted lips and exaggerated movements of the perioral muscles and increased speed; swallowing with tongue interposition and orofacial muscles participation<sup>17,18</sup>.

According to Fraive et al.<sup>8</sup>, microcephaly is present in SCKL. Allied to this condition, according to Tanigute<sup>19</sup>, when the breathing mode is not given through the nose, it can harm the proper development of orofacial structures. Therefore, the following changings were found in children: habitual posture parted lips, habitual posture of the anterior, hypotonic tongue and tremor. Due to the presence of these changings, mobility and tone of orofacial structures found to be inadequate<sup>13</sup>. It is believed that, according to the studied authors<sup>18,20-22</sup> chewing, once installed, should be alternating bilateral, with significant rotation movements jaw, which is the standard considered correct by allowing the

switching working and the muscles relaxing. Thus, the food is distributed homogeneously both in the left and right tooth, also enabling uniform distribution of masticatory forces, voltage stability and the activity of bilateral sync masticatory muscles. So, to the authors, the bilateral pattern, would be the ideal model of mastication, due to functional harmony of the various components of the estomatognathic system<sup>18,21,22</sup>. This information was not observed in the present study.

Facial typology is the variation of the craniofacial skeleton, which is composed of bone and muscle structures. To diagnose the facial type is important because each type has its own characteristics according to dental occlusion, facial harmony, the orofacial muscles, in addition to the shape and configuration of craniofacial structures. It is known that these aspects directly influence the functions of chewing, swallowing, voice, breathing and speech. In this study, the individual had a long face, it can be observed: altered posture of the lips, oral or oronasal breathing, speaking and swallowing with anterior tongue, chewing and altered tongue posture on the floor during the pause<sup>20</sup>, this information can be correlated with the findings described here, since the face type with SCKL was not found. Swallowing presented in a modified form, but the child is in the mixed dentition<sup>21</sup>, therefore, the result requires consideration in relation to this changing, it can not be regarded as a pathological functional changings and not necessarily the syndrome studied. It is important to emphasize that the individual variations that occur during this phase, as they must always be related to the severity of muscle involvement, the presence or absence of dental malocclusion segment, the exchange of teeth and altered anatomical conditions.

Regarding the speech articulation succeeded inaccurate and reduced form, due to the limitation of maximum mouth opening, which in this case was 33millimeters (mm) caused by the small size of the structures orofaciais<sup>8</sup>, the contents of which Bianchini<sup>22</sup> refers normality for maximum active mouth opening less than 35mm in the child, it is a warning of possible muscle and/or joint<sup>22,23</sup> problems. The child presented simplification of consonant and final consonant, phonological processes, according to Wertzner<sup>24</sup>, it should not be present in children with nine years old.

With respect to voice, in the literature, it was not found findings of vocal correlations with the syndrome, yet the child had reduced maximum phonation time (MPT). Behlau<sup>25</sup> to the accompanying figures TMF average value of the numbers of years. Thus, it would be expected to sustain the child



for approximately nine seconds since currently, he/she is nine years old.

The child also had elevated position in the larynx, which features a higher voice, nasal resonance, which, according to Servilha<sup>26</sup>, can result from frequent infections of the upper airways, smooth vocals and low intensity attacks, which are related to microcephaly, the size of the structures and possible timidity of the patient during the assessment, since in nasolaryngoscopy, no significant structural changings were observed.

In accordance with Vitto e Feres<sup>27</sup>, language is a complex process that requires the participation and interaction of phonology, semantics, morphology, syntax and pragmatics. She also reported on changings in oral communication found in individuals with certain genetic syndromes. In this case, with respect to the oral skills and written language at the time of evaluation, no changings were observed.

Hearing thresholds were within normal limits, with the presence of contralateral acoustic reflexes ipsie, yet tympanogram was the type Ar, which refers to an increase in the rigidity of the eardrum-ossicular system<sup>28</sup>. Since there is no data in the

literature regarding the audiological findings in these children, it is not possible to say whether this type of curve is present in other cases. What justifies further research on other cases where it is detected the presence of SCKL.

## ■ FINAL CONSIDERATIONS

In this study, speech therapy manifestations were observed in the orofacial motor skills, speech and voice areas, however these cannot be generalized to all cases with SCKL mainly by the range of changes that this syndrome can present, and because the study was conducted only with an individual. The results of this study suggest the need for speech therapy as part of the interdisciplinary work of these children.

A few studies in the literature, associated to the rarity of occurrence, justified the interest in this work aiming to expansion of scientific knowledge by both audiologists and health professionals, and it is useful as an incentive for more research in this field, still little explored.

## RESUMO

Síndrome de Seckel, trata-se de condição rara e de herança autossômica recessiva, hereditário, caracterizada por um severo retardo de crescimento intra-uterino, baixa estatura proporcional, microcefalia com queixo retraído e pequeno, nariz grande e curvo, em alguns casos retardo mental, várias anomalias congênitas em face, crânio e esqueleto, entre outras malformações. O objetivo deste estudo foi identificar as manifestações fonoaudiológicas encontradas nesta Síndrome, a partir do relato de um caso clínico, de um paciente do gênero masculino, de 09 anos de idade, encaminhado pela odontopediatria, com queixa de alteração no padrão respiratório. Foi realizada avaliação fonoaudiológica que abordou: descrição física, aspectos das funções estomatognáticas, da linguagem oral e escrita, da voz e audição e dos aspectos cognitivos. Para complementação do diagnóstico fonoaudiológico, foram realizadas avaliações ortodôntica, otorrinolaringológica e fisioterapêutica. Com relação às funções estomatognáticas, a criança apresentou respiração de modo oral, mastigação com preensão dos alimentos nos dentes laterais, mastigação unilateral, com lábios entreabertos e movimentos exagerados da musculatura perioral; deglutição com interposição de língua e participação da musculatura orofacial. Fala com articulação imprecisa e travada, devido à limitação de abertura de boca. No tocante à voz, o paciente apresentou tempo máximo de fonação reduzido, laringe em posição elevada, ressonância nasal, ataques vocais suaves e intensidade fraca. Não fora observada nenhuma alteração na linguagem oral e escrita. A escassez de estudos relatando manifestações fonoaudiológicas na Síndrome de Seckel, associada à raridade de ocorrência, justifica o interesse em realizar este estudo, para colaboração de maior conhecimento por parte dos fonoaudiólogos e profissionais de saúde.

**DESCRITORES:** Fonoaudiologia; Avaliação; Diagnóstico

## ■ REFERENCES

1. Giacheti CM. Fonoaudiologia e Genética: Estudos Contemporâneos. In: Piccolotto L, Befi-Lopes DM, Limongi SCO. Tratado de fonoaudiologia. ed 1ª. São Paulo: Roca LTDA; 2005. p1041-53.
2. Gómez FC. Síndrome de Seckel (Dwarfismo Primordial), Reporte de um caso. Disponível em <<http://soniped.org/articulos/FCKeditor/UserFiles/File/articulo%20DR.CAJINA.pdf>>. Acesso em: 20. 05. 2011.
3. Reddy S, Christopher S. Seckel syndrome and spontaneously dislocated lenses. *Journal of Cataract & Refractive Surgery*. Manhattan. 2007;33:910-2.
4. HarshaVardhan BG, Muthu MS, Saraswathi K, Koteeswaran D. Bird-Headed Dwarf of Seckel. *J Indian Soc Pedod Prevent Dent*. Índia. 2007;25(5):8-9.
5. Maroteaux PP. Le maladiesoseuse de l' infant. *Flamarin medicine Sciencies Paris*. Apud: Borba J. Aspectos ortopeditos en la Síndrome de Seckel. *Rev.Esp.de Cir.Ort*. 1974;12:183-9.
6. Premby M, Thompson E. Seckel syndrome: an overdiagnosed syndrome. *Journal of Medical Genetics*. 1985;22:192-200.
7. Kilinc MO, Ninis VN, Ugur SA, Tüysüz B, Seven M, Balci S, Goodshio J, Tolun A. Is the novel SCKL3 at 14q23 the predominant Seckel locus?. *European Journal of Human Genetics*. 2003;11:851-7.
8. Faivre L, Cormier-Daire V. Seckel Syndrome. *Orphanet encyclopedia*. França. abril. 2005.
9. Brito MC, Misquiatti ARN. Intervenção fonoaudiológica na síndrome de kabuki: relato de caso. *Rev. CEFAC*. 2010;12(4):693-9.
10. Genaro KF, Berretin-félix G, REHDER MIBC, MARCHESAN IQ. Avaliação Miofuncional Orofacial: Protocolo MBGR. *RevCefac*. 2009;11(2):237-55.
11. Andrade CRSPD, Befi-Lopes DM, Fernandes FDM, Wertzner HF. ABFW- Teste de linguagem infantil nas áreas de fonologia, vocabulário, fluência e pragmática. ed. 2ª. Baurueri-SP: Pró-fono; 2004. p.97.
12. Navas ALGP, Santos MTM. Linguagem Escrita: aquisição e desenvolvimento. In: Piccolotto L, Befi-Lopes DM, Limongi SCO. Tratado de fonoaudiologia. ed 1ª. São Paulo: Roca LTDA; 2005. p 825-43
13. Salles JF, Parente MAMP. Funções neuropsicológicas em crianças com dificuldades de leitura e escrita. *Psicol Teor Pesqui*. 2006;22(2):153-62.
14. Saraiva RA, Moojen SMP, Munarski R. Avaliação da compreensão leitora textos expositivos. São Paulo: Casa do Psicólogo; 2005.
15. De Coster PJ, Veerbeek RMH, Holthaus V, Martens LC, Vral A. Seckel syndrome associated with oligodontia, microdontia, enamel hypoplasia, delayed eruption, and dentin dysmineralization: a new variant?. *Oral Pathol Med*. Ghent. 2006;35:639-64.
16. Felício CM. Desenvolvimento Normal das Funções Estomatognáticas. In: Ferreira LP, Béfi Lopes DM, Limongi S. Tratado de Fonoaudiologia. ed 1ª. São Paulo: Roca; 2004. p.195-210.
17. Furkim AM. Fisiologia da deglutição. In: Ferreira LP, Béfi Lopes DM, Limongi S. Tratado de Fonoaudiologia. ed 1ª. São Paulo: Roca, 2004. p.212-9.
18. Marchesan IQ. Avaliação e Terapia dos Processos de Respiração. In: Marchesan IQ. Fundamentos em Fonoaudiologia: Aspectos Clínicos da Motricidade Oral. ed 2ª. Rio de Janeiro: Guanabara Koogan, 2005. p.28-43.
19. Tanigute, CC. Desenvolvimento das Funções Estomatognáticas. In: Marchesan IQ. Fundamentos em Fonoaudiologia: Aspectos Clínicos da Motricidade Oral. ed 2ª. Rio de Janeiro: Guanabara Koogan, 2005. p.1-9.
20. Pereira AC, Jorge TM, Ribeiro Júnior PD, Berretin-Felix G. Características das funções orais de indivíduos com má oclusão classe III e diferentes faciais. *Rev Dental Press Ortodon Ortopedi Facial*. 2005;10(6):111-9.
21. Bertolini MMB. Estudo da deglutição adaptada e sua associação com variáveis de interesse epidemiológico, em crianças na fase inicial de denteição mista. [Tese de Doutorado]. Campinas(SP): Universidade Estadual de Campinas. Pós-Graduação da Faculdade de Ciências Médicas. 2004
22. Bianchini EMG. Mastigação e ATM: avaliação e terapia. In: Marchesan I Q. Fundamentos em fonoaudiologia: aspectos clínicos da motricidade oral. Rio de Janeiro: Guanabara Koogan, 2005. p. 37-49.
23. Cattoni DM, Fernandes FDM. Distância interincisiva máxima em crianças na dentadura mista. *Rev. Dent. Press Ortodon. Ortop. Facial*. 2005;10(1)
24. Wertzner HF. Fonologia: Desenvolvimento e Alterações. In: Ferreira LP, Béfi Lopes DM, Limongi S. Tratado de fonoaudiologia. ed 1ª. São Paulo: Roca LTDA, 2005. p.772-86.
25. Behlau M, Azevedo R, Pontes P. Conceito de Voz normal e classificação das disfonias. In: Behlau M. Voz o livro do especialista I. ed 1ª. Rio de Janeiro: Revinter, 2008. p. 53-76.
26. Servilha EAM. Voz na infância. In: Ferreira LP, Béfi Lopes DM, Limongi S. Tratado de fonoaudiologia. ed 1ª. São Paulo: Roca LTDA, 2005. p.118-26.
27. De Vitto MMP, Feres MCLC. Distúrbios da Comunicação Oral em Crianças. *Medicina*. 2005;38:229-34.

28. Carvalho RMM. Timpanometria. In: Bevilacqua MC, Martinez MAN, Balen SA, Pupo AC, Reis ACMB, Frota S. Tratado de Audiologia. ed 1ª. São Paulo: Santos, 2011.p.121-33.

Received on: January 22, 2014  
Accepted on: September 09, 2014

Mailing address:  
Aline Ferreira de Brito  
Rua Icarai, 161, Farolândia  
Aracaju – SE – Brasil  
CEP: 49032-380  
E-mail: alinebrito.fono@gmail.com

## ■ ANEXO A - MBGR

## EXAME MIOFUNCIONAL OROFACIAL (fonte: –GENARO et al., 2009, protocolo MBGR sintetizado)

## 3. INTRA-ORAL EXAMINATION

## Lips [ ]

Mucosa:	(0) normal	(1) wound
Frenulum higher:	fixation: (0) normal	(1) low
	thickness: (0) normal	(1) change (describe): _____

## Cheeks [ ]

Mucosa:	(0) normal	(1) dental brands R	(1) line Alba R	(1) resected R	(2) wound R
		(1) dental brands L	(1) line Alba L	(1) resected L	(2) wound L

## Tongue [ ]

Longitudinal groove:	(0) appropriate	(1) profound		
Usual position:	<input type="checkbox"/> unobservable	(1) on the floor		
		(1) dorsum of the tongue high (1) interdental: _____		
Symmetry:	(0) yes	(1) no (dicriver): _____		
Breadth:	(0) appropriate	(1) decreased	(2) increased	
Height:	(0) appropriate	(1) decreased	(2) increased	
Tremor:	(0) wanting	(1) in the usual position	(1) protrusion	(1) movements (four cardinal points)
Mucosa:	(0) normal	(1) Geographic	(1) fissured	
	(1) dental brands (local): _____	(1) Marked by device (local): _____		
	(1) wound (local): _____			
Frenulum: extension:	(0) normal	(1) short		
fixation on tongue:	(0) middle part	(1) preceding the middle part	(2) at the apex	
setting the floor:	(0) between caruncles	(1) between the alveolar crest and the caruncles	(2) alveolar crest	
lateralized to the tongue:	(0) normal	(1) lowers the apex R	(1) lowers the apex L	
stand out the tongue:	(0) normal	(1) lowers the apex	(1) depresses the central portion of the tongue	
to raise the tongue: (inside the mouth without touching the upper arch)	(0) normal	(1) square / rectangular apex	(2) heart Shaped	
other characteristics:	(0) wanting	(1) fixing longer visible in the alveolar ridge	(1) submerged (1) fibrous	

## Palate [ ]

Hard: depth:	(0) normal	(1) reduced (baixo)	(2) increased (high)
width:	(0) normal	(1) increased (large)	(2) reduced (narrowed)
Soft: symmetry:	(0) present	(1) wanting	
length:	(0) appropriate	(1) regular	(2) long (2) short
Uvula:	(0) normal	(1) change (Describe): _____	

## Tonsils [ ]

Presence:	<input type="checkbox"/> present	<input type="checkbox"/> removed	<input type="checkbox"/> not be observed
Size:	(0) normal	(1) hypertrophy R	(1) hypertrophy L
Colour:	(0) normal	(1) hyperemia R	(1) hyperemia L
Position in tonsils	<input type="checkbox"/> not visible	<input type="checkbox"/> mean: [ ] R [ ] L	

## 3.6. Teeth and Occlusion [ ]

Toothing:	<input type="checkbox"/> primary	<input type="checkbox"/> mixed	<input type="checkbox"/> permanent
No of teeth:	higher R _____	higher L _____	less R _____ less L _____
Dental flaw:	(0) wanting	(1) present (elements): _____	
Dental conservation:	(0) good (1) regular	(2) bad	
Gummy conservation:	(0) good (1) regular	(2) bad	
Dental flaw:	(0) appropriate	(1) diverted R	(1) diverted L
Occlusion:	(0) normal	(1) change	
Classification of Angle:	side R (0) Class I	(1) Class II div.1	(1) Class II div. 2 <sup>a</sup> (1) Class III
	side L (0) Class I	(1) Class II div.1	(1) Class II div. 2 <sup>a</sup> (1) Class III



Horizontal amendment: (0) wanting	(1) top bite (TH = 0mm)	(1) overjet (TH > 3mm)	(1) anterior crossbite (TH < 0mm)
Vertical change: (0) wanting	(1) top bite (TV = 0mm)	(1) overjet (TV > 3mm)	(1) anterior crossbite (TV < 0mm)
Transversal alteration: (0) wanting	(1) right posterior cross bite	(1) left posterior cross bite	(1) open bite posterior
Prosthesis use:	<input type="checkbox"/> não	<input type="checkbox"/> removável	<input type="checkbox"/> fixed
Use apparatus:	<input type="checkbox"/> não	<input type="checkbox"/> mobile	<input type="checkbox"/> fixed
	<input type="checkbox"/> partial	<input type="checkbox"/> total	

Describe the device and / or prosthesis: \_\_\_\_\_

Other changes: \_\_\_\_\_

**MOBILITY**

**Lips [ ]**

\*Run with the occluded teeth

- In alternating movements, run 3 times and pace.

	normal	approximate	attempts to perform	not perform
Protrude closed *	(0)	(1)	(2)	(3)
Retract closed *	(0)	(1)	(2)	(3)
Switch bulging / retract closed *	(0)	(1)	(2)	(3)
Protrude opened*	(0)	(1)	(2)	(3)
Retract opened*	(0)	(1)	(2)	(3)
Switch bulging / retract opened *	(0)	(1)	(2)	(3)
Protrude closed to R *	(0)	(1)	(2)	(3)
Protrude closed to L *	(0)	(1)	(2)	(3)
Switch bulging closed to R and L *	(0)	(1)	(2)	(3)
Click protruded	(0)	(1)	(2)	(3)
Clickretract	(0)	(1)	(2)	(3)
Switch snap protruded / retracted	(0)	(1)	(2)	(3)

**Tongue [ ]**

In alternating movements, run 3 times and pace.

	normal	approximate	attempts to perform	not perform
Protrude	(0)	(1)	(2)	(3)
Switch bulging / retract	(0)	(1)	(2)	(3)
Raise the incisive papilla	(0)	(1)	(2)	(3)
Switch raise and lower the papilla	(0)	(1)	(2)	(3)
Raise the upper lip	(0)	(1)	(2)	(3)
Switch raise / lower lips touching	(0)	(1)	(2)	(3)
Play the labial R	(0)	(1)	(2)	(3)
Play the labial L	(0)	(1)	(2)	(3)
Switch to touch the corners R and L	(0)	(1)	(2)	(3)
Play the apex sequentially at the corners R / L and S / I lips	(0)	(1)	(2)	(3)
Internally touch the cheek R	(0)	(1)	(2)	(3)
Internally touch the cheek and	(0)	(1)	(2)	(3)
Switch touch the cheeks R and L	(0)	(1)	(2)	(3)
Click the apex	(0)	(1)	(2)	(3)
Click the body	(0)	(1)	(2)	(3)
Sucking the tongue on the palate	(0)	(1)	(2)	(3)
Vibrate	(0)	(1)	(2)	(3)

**Cheeks [ ]**

	normal	approximate	attempts to perform	not perform
Inflate	(0)	(1)	(2)	(3)
Inflate the right side	(0)	(1)	(2)	(3)
Inflating the left side	(0)	(1)	(2)	(3)
Switch Inflate right and left	(0)	(1)	(2)	(3)

**Soft Palate [ ]**

	normal		reduced movement		movement away		note
Talking [a] repeatedly	(0) R	(0) L	(1) R	(1) L	(2) R	(2) L	_____
Elicit the reflex nauseating	(0) R	(0) L	(1) R	(1) L	(2) R	(2) L	_____

**Mandible [ ]**

	normal	reduced	increased	does not perform	with deviation	
Mouth opening	(0)	(1) <40mm	(1) >55mm	(2)	(1) R	(1) L
Closing the mouth	(0)	-	-	-	(1) R	(1) L
Laterality right	(0)	(1) <6mm	(1) >12mm	(2)	-	-
Laterality left	(0)	(1) <6mm	(1) >12mm	(2)	-	-

Deviation of presence (in some movement)	(0) no	(1) yes
Pain (in some movement)	(0) no	(1) yes

**TONE [ ]**

	Normal	Decreased	Increased
Upper lip	(0)	(1)	(1)
Lower lip	(0)	(1)	(1)
Mento	(0)	(1)	(1)
Labial groove ment	(0)	(1)	(1)
Language	(0)	(1)	(1)
Floor of mouth	(0)	(1)	(1)
Right cheek	(0)	(1)	(1)
Left cheek	(0)	(1)	(1)

Perform palpation and visual observation, except the floor should only be observed.

**ORAL FUNCTION**

**Breath [ ]**

Type:	(0) medium / lower	(1) medium / higher	(1) Other (describe): _____
Mode:	(0) nasal	(1) oronasal	(2) oral
Nasal flow (use the mirror) to get:	(0) simetric	(1) Reduced right	(1) Reduced left
after cleaning:	(0) simetric	(1) Reduced right	(1) Reduced left
Possibility of nasal use:	(0) 2 minutes or more	(1) 1 to 2 minutes	(2) least 1 to 2 minutes

Notes: \_\_\_\_\_

**Chew:**  adequate  change: source [ ] functional [ ] anatomical [ ] articulate [ ] other (maſticatory assessment always use the same food)

**Habitual chewing [ ]**

Incision:	(0) previous	(1) Side	(1) other _____
Crushing:	(0) posterior teeth	(1) anterior teeth	(1) with the tongue
	(0) efficient	(1) inefficient	
Number of cycles:			
(via film):	right: first portion: _____	second portion: _____	third portion: _____
	left: first portion: _____	second portion: _____	third portion: _____
	total: first portion: _____	second portion: _____	third portion: _____
Chewing pattern:	(0) bilateral alternate	(0) unilateral	(1) bilateral (2) unilateral
		preferential: _____	simultaneous chronic: _____
Lip lock:	(0) systematic	(1) unsystematic	(2) wanting
Speed:	(0) appropriate	(1) increased	(1) decreased
Noise:	(0) wanting	(1) present	
Atypical muscle contractions:	(0) wanting	(1) present (describe): _____	

Others: \_\_\_\_\_

**Ask the patient**

Preferred chewing side:	(0) right and left	(1) right	(1) left	(0) do not know
Pain when chewing:	(0) wanting	(1) right	(1) left	
Noise at ATM:	(0) wanting	(1) right	(1) left	

Observations: \_\_\_\_\_

**Swallow:**  adequate  change: source [ ] functional [ ] anatomical [ ] articulate [ ] other

Lip lock:	(0) appropriate	(1) partial	(2) wanting
Tongue posture: <input type="checkbox"/> not seen	(0) behind the teeth	(1) against the teeth	(2) between teeth
Lower lip posture:	(0) contact top	(1) behind the upper incisors	
Food containment:	(0) appropriate	(1) partial	(2) inadequate
Contraction of the orbicularis:	(0) appropriate	(1) little	(2) sharp
Contraction of the chin:	(0) wanting	(1) little	(2) sharp
Contraction of the neck muscles:	(0) wanting	(1) little	(2) sharp
Nod of his head:	(0) wanting	(1) present	
Noise:	(0) wanting	(1) present	
Coordination:	(0) appropriate	(1) choking	(1) cough
Residue after swallowing:	(0) wanting	(1) present	

Comments: \_\_\_\_\_

**Ask the patient**

Tongue position:	(0) no	(1) yes (describe): _____
Tongue position:	(0) behind the upper teeth	(1) behind the lower teeth (2) between teeth (0) do not know

Comments: \_\_\_\_\_

## ■ ANEXO B - ABFW

## PHONOLOGY. PROTOCOL. IMITATION

Name:  
Exam date:  
Age:

Record	
word	Rescript
1 peteca	
2 bandeja	
3 tigela	
4 doce	
5 cortina	
6 gato	
7 foguete	
8 vinho	
9 selo	
10 zero	
11 chuva	
12 jacaré	
13 machado	
14 nata	
15 lama	
16 lápis	
17 prego	
18 café	
19 alface	
20 raposa	
21 borracha	
22 abelha	
23 carro	
24 branco	

Traditional analysis		
phoneme	initial	finale
P		
B		
T		
D		
K		
G		
F		
V		
S		
z		
ʃ		
ʒ		
M		
N		
ɲ		
l		
ʎ		
ɾ		
r		
Arqui /S/		
Arqui /r/		
pR		
bR		
tR		





PHONOLOGY. PROTOCOL. NOMINATION

Name:  
Exam date:  
Age:

Record	
word	Rescript
1 peteca	
2 bandeja	
3 tigela	
4 doce	
5 cortina	
6 gato	
7 foguete	
8 vinho	
9 selo	
10 zero	
11 chuva	
12 jacaré	
13 machado	
14 nata	
15 lama	
16 lápis	
17 prego	
18 café	
19 alface	
20 raposa	
21 borracha	
22 abelha	
23 carro	
24 branco	
25 travessa	
26 droga	
27 cravo	
28. grosso	
29 fraco	

traditionalanalysis		
phoneme	initial	finale
P		
B		
T		
D		
K		
G		
F		
V		
S		
z		
ʃ		
ʒ		
M		
N		
ɲ		
l		
λ		
τ		
r		
{S}		
{R}		
pR		
bR		
tR		
dR		
kR		
gR		
fR		
pl		

<b>Record</b>	
<b>word</b>	<b>Rescript</b>
30 plástico	
31 bloco	
32 clube	
33 globo	
34 flauta	
35 pastel	
36 porco	
37 nariz	
38 amor	
39 roupa	

<b>traditionalanalysis</b>		
<b>phoneme</b>	<b>initial</b>	<b>finale</b>
bl		
kl		
gl		
fl		

Hit: Omission: Replacement: distortion
---

PHONOLOGY.ANALYSIS OF THE PHONOLOGICAL PROCESSES.NOMINATION

Name: \_\_\_\_\_  
 Age \_\_\_\_\_

	palhaço	bolsa	tesoura	cadeira	galinha	vassoura	cebola	xícara	mesa	navio	livro	sapo	tambor	sapato	balde	face	fogão	total	
<b>Transcription</b>																			
Syllable reduction																			
harmony consonant																			
Stopping of fricative																			
Posteriorization to ensure																			
Backing for palatal																			
Fronting of velars																			
Palatal fronting																			
Liquid simplification																			
Simplification of consonant cluster																			
Simplification of the final consonant																			
Sound of plosives																			
Sound of fricative																			
Devoicing of plosive																			
Devoicing of fricative																			
Other																			
<b>Total</b>																			

PHONOLOGY: ANALYSIS OF THE PHONOLOGICAL PROCESSES: NOMINATION

Name:																	
Age																	
Transcription	peixe	relógio	cama	anel	milho	cachorro	blusa	garfo	trator	prato	pasta	zebra	girafa	braço	planta	cruz	total
Syllable reduction																	
harmony consonant																	
Stopping of fricative																	
Posteriorization to ensure																	
Backing for palatal																	
Fronting of velars																	
Palatal fronting																	
Liquid simplification																	
Simplification of consonant cluster																	
Simplification of the final consonant																	
Sound of plosives																	
Sound of fricative																	
Devoicing of plosive																	
Devoicing of fricative																	
Other																	
<b>Total</b>																	

PHONOLOGY.ANALYSIS OF THE PHONOLOGICAL PROCESSES.IMITATION

Name: \_\_\_\_\_  
 Age \_\_\_\_\_

Transcription	peteca	bandeja	tigela	doce	cofina	gato	foguete	vinho	selo	zero	jacaré	machado	nata	lama	lápiz	prego	café	alface	total	
Syllable reduction																				
harmony consonant																				
Stopping of fricative																				
Posteriorization to ensure																				
Backing for palatal																				
Fronting of velars																				
Palatal fronting																				
Liquid simplification																				
Simplification of consonant cluster																				
Simplification of the final consonant																				
Sound of plosives																				
Sound of fricative																				
Devoicing of plosive																				
Devoicing of fricative																				
Other																				
<b>Total</b>																				