

SPEECH-LANGUAGE CLINICAL PATTERN IN A NEUROGENIC DYSPHAGIA CASE THROUGH OCULO-PHARYNGEAL MUSCULAR DYSTROPHY

Conduta fonoaudiológica em um caso de disfagia neurogênica por distrofia muscular oculofaríngea

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ABSTRACT

The Muscular Dystrophy Oculo-pharyngeal myopathology is a hereditary autosomal dominant, late-onset, with clinical signs of ptosis and oral pharyngeal dysphagia, this may be associated to the proximal muscle weakness. This study examines the speech therapy conduct in a case of oculo-pharyngeal dystrophy. The subject was treated in the speech-language therapy outpatient since March 2013, with biweekly sessions, being changed to weekly sessions from July of the same year. The therapy consists of improving mobility of oral facial structures; organization of the pneumonic articulatory coordination; frequent reevaluations of the swallowing disorders, with the register of the episodes of signals and symptoms of swallowing disorders in the habitual feeding, as well as, by the verification of the usual diet consistencies with security maneuver (chin-chest); weight check; using the habitual daily swallowing disorders diary, and orientation for the accomplishment of the daily oral facials exercises at home. She had stabilization of the clinical picture and the corporal weight during the year of attendance, with posterior retaken of disease progression. The suggested clinical aiming is of interdisciplinary boarding, having as contribution from speech and language therapy, the aiding of a safe feeding, the facilitation of the verbal communication and improvement of the quality of life.

KEYWORDS: Deglutition Disorders; Muscular Dystrophy, Oculopharyngeal; Nervous System Diseases; Voice; Speech; Language

■ INTRODUCTION

Myopathies are structural and/or functional muscle diseases, which are resulted from various etiologies, what is evident as muscle weakness, being required to set the differentiation of motor neuron diseases, peripheral neuropathies and/or neuromuscular junction disorders¹.

The patients evaluations are to, firstly, identify the myopathy, which are likely to be caused by channel defects, structural alteration or metabolic disorder. It was required to determine if the myopathy in

question is hereditary or acquired, as well as if there is an available treatment, even paliative¹.

Oculopharyngeal muscular dystrophy (OPMD) was first described by Taylor, in 1915, as being characterized as an hereditary autosomically transmitted dominant myopathy of late manifestations (between the fifth and sixth decade of life), with clinical signs of ptosis of the eyelids, oropharyngeal Dysphagia, the latter being possibly associated with proximal muscle weakness^{2,3}. Cases, such as the report of a Jewish family from 1962, disseminated its designation as Oculopharyngeal muscular dystrophy – OPMD⁴.

OPMD is characterized by Dysphagia in different levels, accordingly to the severity or progression of the disorder, being necessary to modify the diet's firmness. The deglutition disorders are, therefore, progressive, starting as difficulty ingesting solid

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food, leading softer food firmness and, subsequently, suppressing the ingestion of food orally. Consequently, the carriers of such disorders are likely to present episodes of bronchoaspiration, oral regurgitation and/or nasal reflux, asphyxiation crises, pneumonia, as well as bronchial asthma². The clinical profile might be accompanied by hoarseness (dysphonia)², dysarthria⁵ and weight loss². Voice, articulatory and deglutition alterations characterize the profile of dysarthrophonia, which negatively impacts the subjects quality of life⁶.

The genetic alteration that is responsible for the disorder is described as being caused the short repetitive expansions of GCG trinucleotides encoded for the polyalanine (poly-A) tract-binding protein 2 (PABP2)⁷. The evaluation counts on manometry data and on the radiologic contrast study, however the safe diagnosis is obtained through the genetic study PABPN1, observing the gene of the 14th chromosome³.

The estimative of the frequency of the gene is one in every thousand inhabitants in the French-Canadian population⁷, but it varies according to the region of a determined country and of the world, being estimated in one in every one hundred thousand European inhabitants, and one in every six hundred Israeli Jews^{8,9}.

This study is a report of the clinical case, whose objective is to describe the clinical and speech-language evolutionary data of a patient who is a carrier of OPMD.

■ CASE PRESENTATION

This study was approved by the Ethics Committee of Human Beings Research of the Federal University of Health Sciences of Porto Alegre (CEP – UFCSPA), under the opinion issued #042/12 and the Free and Elicited Consent Term (TCLE) was signed by the participating subject.

The subject who carries OPMD in this report is a 65 year-old female, with severe Eyelid Ptosis, admitted in the Speech Therapy Ambulatory, along with the Neurology department, of a hospital in Porto Alegre, Brazil, whose record states an initial complaint with weight loss.

The speech-language assessment from 2013 shows: facial-oral muscle tension loss by denervation, limitation of the opening of the mouth, atrophy of the palatine pillars, limited palate mobility, alteration of the vocal quality (nasal, hoarse and moist), multiple deglutition, nasal reflux, articulatory imprecision and alteration of the pulmonary phonation and articulatory coordination.

The speech-language diagnosis is of flaccid dysarthrophony, obtained through the Dysarthria

protocol¹⁰, with diminished sensibility in the oral pharyngeal region the reduced mobility of the soft palate, what caused nasal resonance to her speech and neurogenic oral pharyngeal dysphasia with flaccid predominant characteristics of a moderate degree for liquid and solid food through the AFDN protocol¹¹.

The patient was subjected to sessions at every two weeks in the Speech Therapy Ambulatory, from March 2013, being that, from July 2013 she was to have weekly sessions.

The speech therapy for this particular case consisted in: Facial–oral Myofunctional Therapy through exercises of the mobile structures; tactile and thermal stimulation; Pulmonary Phonation and Articulatory Coordination Organizing; swallowing practice with protection maneuver and frequent reassessments of the Dysphagia condition, with the registering of the modifications of softness in diet to pasty foods, ingested with the head protection maneuver – chin tuck – and, by daily assessing the deglutition disorder¹², having weekly checks of the patient's weight; and orienting and training the patient to perform the Facial-oral Myofunctional Therapy exercises at home.

■ RESULTS

In the speech therapy process of the patient, observed signs are of clinical stabilization, recovery and worsening, the latter due to the evolution of the disease.

In September 2013, the patient observed a greater easiness to speak and better understanding from the interlocutor and improved intraoral sensibility, with fewer episodes of coughing and choking by using the safety maneuver, considerably improving the capacity to have liquids. The result of the video fluoroscopy was: inadequate oral phase swallowing in the bolus preparation, premature loss and changes in esophageal clearance for solid food requiring to alternate with liquids (taken in 2013). There was body weight stabilization. By having weekly sessions the patient had observed clinical improvement in the aspects of sensibility, facial-oral mobility and motor function.

During the first semester of 2014 (between January and August 2014) the clinical evolution was of low expiratory capacity for the gender, age and body weight, assessed through spirometry; reported choking, coughing, aspiration, respiratory alterations and tiredness or sleepiness after daily nourishment, assessed through the Dysphagia diary; the improvement of the pulmonary phonation and articulatory coordination, but with glottis cooptation, what was verified by the s/z ratio. There

was no alteration in the body weight between August 2013 and August 2014.

The patient's records stated that vocal characteristics were maintained, with hoarse and breathy voice; there was an improvement in the aspects of sensibility, facial-oral mobility and motor function, which reflected in the pulmonary phonation and articulatory coordination and speech intelligibility; and worsening on the signs and symptoms of Dysphagia.

■ DISCUSSION

With the advancements in genetic medicine, nowadays, it is available a better access to the diagnosis instrumentation, what are now important tools for medical diagnosis and, thus, a broader access to data obtained through scientific evidence.

The OPMD research was benefitted from such advancements, since it permits the diagnosis of the molecular genetic mutation of the disease, which has a late development, whose symptoms are present in the family history from two or more generations, affecting both genders⁹.

It is a degenerative disease of the skeleton muscles, of hereditary nature, which is differentiated from other muscular dystrophy pictures by the distribution of the muscular deficit, the age when the first symptoms appear, the speed of its evolution, the gravity of the symptoms and the family background, whose clinical relevant sign is the precocious incapacity of swallowing^{9,13}.

Its variable incident is considered to be high, depending on the region of the world, being identified in more than 30 countries⁹. It is slow progressing and the symptoms are manifested by bilateral ptosis of the eyelids and oral pharyngeal dysphagia, due to weakness of the eyelid lifting muscle and the pharyngeal muscles¹⁴. The data collected from observing the patient make evident Dysarthrophonia, with alterations in the articulation, voice and swallowing.

The ptosis of the eyelids is classified according to the capacity of the lifting muscle, what could be a *mild ptosis*, when the edge of the upper lid is between 2 mm and 4 mm under the corneal limbus, *moderate ptosis*, when it is 4 mm to 6 mm under the corneal limbus, and *severe ptosis* when it is 6 mm or more under the corneal limbus¹⁵. According to this classification¹⁵, the patient of this study presented the edge of the upper eyelid under 6 mm from the corneal limbus, what set the gravest degree of ptosis of the eyelids.

Ptosis of the eyelids is observed, in average, at the age of 48, and Dysphagia at the age of 50 in cases of OMPD¹⁴. The early symptoms of

Dysphagia are⁹ increased eating time and refusing dry food. Swallowing problems often evolve gradually suppressing the ingestion of liquids as well. The evolution of the symptoms might lead to bronchoaspiration episodes, oral regurgitation and nasal reflux, asphyxiation crises, pneumonia and undernourishment. Hoarseness is also observed, as well as moist voice due to the laryngeal invasion of liquids in 67% of the patients and weakness of the facial muscles in 43% of them^{3,9}. In the most severe cases, which represent 5% to 10% of the carriers, ptosis of the eyelids and oral pharyngeal dysphagia are observed before the age of 45⁹.

In the clinical assessment of Dysphagia¹¹ that was initially done, the patient presented: articulatory imprecision; pulmonary phonation and articulatory incoordination; incoordination of the oral motor sensory system, which is observed by the difficult mobility of the phonation and articulatory organs and facial-oral sensibility; difficulty in posteriorizing the bolus for liquids, by noticing signs of Laryngeal invasion of liquids, and solid food with premature loss; altered cervical auscultation; and the presence of coughing before, during and/or after swallowing.

The degree of the swallowing difficulty is set by noticing the presence of signs and symptoms of disorders while performing it. By using the protocol¹¹, it is possible to classify the Dysphagia in: *mild*, when there is articulatory adequacy, pulmonary phonation and articulatory coordination, incoordination of the motor oral sensory system, difficulty in posteriorizing the bolus and presence of food or saliva stasis in small quantity; *moderate*, when there is difficulty to repeat certain words or articulatory imprecision, pulmonary phonation and articulatory incoordination, incoordination of the motor oral sensory system, difficulty in posteriorizing the bolus, stasis of food or saliva in small quantities, altered cervical auscultation and the presence of coughing during and/or after swallowing; and *severe*, with the occurrence of mental confusion, non-cooperation of the patient with the performing of the test, affected comprehension, difficulty in repeating words or articulatory imprecision, pulmonary phonation and articulatory incoordination, incoordination of the oral motor sensory system, difficulty in posteriorizing the bolus, stasis of food or saliva in large quantities, altered cervical auscultation, presence of coughing before, during and/or after swallowing, or by the absence of coughing reflex. The degree of the subject in question is moderate. From clinical data, the degree of functionality could be set for food intake by using the Functional Oral Intake Scale (FOIS)¹⁶, that comprises a scale of food intake in 7 levels (picture 1). The functionality level was assessed as IV.

Levels	Oral Intake Functionality
Level I	Nothing by mouth;
Level II	Tube dependent with minimal attempts of food or liquid, for palatal stimulus;
Level III	Tube dependent with consistent intake of food or liquid, proving eating pleasure;
Level IV	Total oral diet of a single consistency;
Level V	Total oral diet with multiple consistencies but requiring special preparation or compensations;
Level VI	Total oral diet with multiple consistencies without special preparation, but with specific food limitations;
Level VII	Total oral diet with no restriction.

Figure 1 – Functional Oral Intake Scale – FOIS¹⁴

The level of severity of the vocal alterations could be established through a perceptual analysis. Considering that analysis and using the parameters of the GRBASI scale¹⁷, in which G refers to the grade of Dysphonia, R – Roughness, B – Breathiness, A – Asthenia, S – Strain and I – Instability, that scores each feature in a grade from 0 to 3, in which 0 refers to the absence of alterations, 1 to a mild alteration, 2 to a moderate alteration and 3 for an intense alteration, it is noticed that the subject in question fits the grade 2, the moderate degree.

The weight loss is referenced throughout the diseases' progressive process^{3,18}. Frequently it is reported a weight loss larger than 10% of the body weight, associated with the signs of dysphagia and pneumonia by aspiration¹⁴. The clinical records of the patient of this study and her initial complaint are corroborated by literature on the issue.

Another point that calls attention is the diminished sensibility in the oral pharyngeal region and reduced mobility of the soft palate, causing nasal resonance to her dysarthric speech. Dysarthria was already mentioned in a study⁵ as one of the diagnosis characteristics of the disease. In literature, it is not found, data of the classification of Dysarthria or Dysarthrophonia in those of OPMD, although there is the citing of the presence of Dysphonia²,

Dysarthria⁵, Dysphagia^{2,3,5,9,13,14}, pharyngeal muscle compromising^{2,5,14} and voice nasality².

Dysarthria is the speech impediment resulting from paresis, paralysis, incoordination or spasticity of the muscles. The lesion of the inferior motor neuron determined the flaccid type of the case, which is characterized by the flaccidness, weakness, atrophy and fasciculation observed^{6,18,19}.

During the speech therapeutic process the patient was managed in order to avoid weight loss, her weight was checked during the sessions, and an estimate 10% of her body weight was lost, and subsequently maintained, until August 2014.

The weekly records of the signs and symptoms of Dysphagia, through the diary of swallowing disorders¹², was critical in this patient's therapeutic process as it was possible to view and correspond the clinical signs and symptoms of Dysphagia in her daily meals and, thus, to identify them in her daily diet, what improved the effectiveness of the treatment and conduced to an adequacy of the speech therapy techniques.

The diary of swallowing disorders was idealized from the reports of patients from the Speech Therapy Ambulatory, and was adjusted until its final version (Picture 2).

No clinical reports of OPMD in the area of Speech Therapy were found, even when characterized by the presence of Oral Pharyngeal Neurogenic Dysphagia and/or Dysarthrophonia.

■ CONCLUSION

Oculopharyngeal muscular dystrophy is considered to be a high incidence disease, with variable occurrence in the world population, varying by region, and it is also a progressive and hereditary disease.

The account of this case shows a speech therapy clinical condition of Oral Pharyngeal

Neurogenic Dysphagia of a moderate degree, with voice alteration characterized by hoarseness and breathiness of moderate gravity, and flaccid type dysarthria, what characterizes the case as flaccid dysarthrophonia. The speech therapy intervention promoted the stabilization of the clinical condition and the body weight during the year of treatment, with later progression of the disease.

The clinical guidance suggested for the cases of OPMD is of an interdisciplinary approach, in which the contribution of speech therapy is to provide a safer nourishment condition, easing the oral communication and improving the quality of life.

RESUMO

A Distrofia Muscular Oculofaríngea é uma miopatia hereditária de transmissão autossômica dominante, de início tardio, com sinais clínicos de ptose palpebral e disfagia orofaríngea, essa podendo estar associada à fraqueza muscular proximal. O presente estudo analisa a conduta fonoaudiológica em um caso de distrofia oculofaríngea. O sujeito atendido no ambulatório de Fonoaudiologia, desde março de 2013, com sessões quinzenais, frequência essa modificada para sessões semanais a partir de julho do mesmo ano. A terapia constituiu-se com os objetivos de: aprimorar a mobilidade das estruturas orofaciais; organizar a coordenação pneumofonoarticulatória; reavaliar frequentemente a disfagia, por meio do registro dos episódios de sinais e sintomas de disfagia na alimentação habitual, assim como, da verificação das consistências da dieta usual com manobra de segurança (queixo no peito); verificar o peso; utilizar o diário de distúrbios da deglutição, e orientar a realização dos exercícios orofaciais em casa. Houve estabilização do quadro clínico e do peso corporal durante o ano de atendimento, com posterior retomada de progressão da doença. O direcionamento clínico sugerido é de uma abordagem multidisciplinar, cuja contribuição da fonoaudiologia é do favorecimento para uma alimentação segura, a facilitação da comunicação oral e melhoria da qualidade de vida.

DESCRIPTORIOS: Transtornos da Deglutição; Distrofia Muscular Oculofaríngea; Doenças do Sistema Nervoso; Voz; Fala; Linguagem

■ REFERENCES

1. Fiorini VCC. Mioptias Hereditárias. 2009. [atualizada em 2009 Aug 29; acesso em 2014 Jun 14]. Disponível em: http://www.medicinanet.com.br/conteudos/revisoes/2239/miopatas_hereditarias.htm.
2. Medici M, Pizzarossa C, Skuk D, Yorio D, Emmanuelli G, Mesa R. Oculopharyngeal muscular dystrophy in Uruguay. *Neuromuscular Disorders*. 1997; 7(Suppl.1):50-2.
3. Liaño AD, Fernandez R, Yáñez C, Artieda C, González G, Artajona A et al. Distrofia muscular oculofaríngea. Tratamiento quirúrgico. *Rev. Chilena de Cirugía*. 2009;61(4):360-5.
4. Victor M, Hayes R, Adams RD. Oculopharyngeal muscular dystrophy: familial disease of late life characterized by dysphagia and bprogressive ptosis of the eyelids. *N Engl J Med*. 1962;267:1267-72.
5. Morales SG, Méndez LA, Rivero AR, Sánchez VML. Distrofia muscular progresiva. *Rev Hosp Jua Mex* 2002;69(1): 30-7.
6. Barata FL, Miguel LS, Silva SAC, Carrara-de Angelis E. Caracterização da fonoarticulação e sua relação com a disfagia em pacientes com disartrofonia em um hospital oncológico. *Distúrb Comum*. 2009;21(1):79-91.
7. Brais B, Bouchard J-P, Gosselin F, Xie Y-G, Fardeau M, Tome Fms et al. Using the full power of linkag e analysis in I I French Canadian families to

- fine map the oculopharyngeal muscular dystrophy gene. *Neuromusc Disord.* 1997;7(Suppl. 1):70-4.
8. Relatórios Orphanet – coleção de doenças raras. No 1, 2014 May. [atualizada em 2014 Jan 20; acesso em 2014 Jun 14]. Disponível em: http://www.orphanet/orphacom/cahiers/docs/PT/Prevalencia_das_doencas_raras_por_ordem_alfabetica.pdf.
9. Trollet C, Gidaro T, Klein P, Périé S, Butler-Browne G, St Guily JL. Oculopharyngeal Muscular Dystrophy. In: Pagon RA, Adam MP, Ardinger HH, Bird TD, Dolan CR, Fong CT, Smith RJH, Stephens K, eds. *Gene Reviews*. Seattle (WA): University of Washington, Seattle; 1993-2014. [atualizada em 2014 Feb 20; acesso em 2014 Jun 14]. Disponível em: <http://www.ncbi.nlm.nih.gov/books/NBK1126/>.
10. Ortiz KZ. Avaliação da Disartria. In: Ortiz KZ. *Distúrbios neurológicos adquiridos: Fala e deglutição*. São Paulo. Editora Manole, 2010, Cap. 5. P. 73-96.
11. Cardoso MCAF. Avaliação Fonoaudiológica das Disfagias Orofaríngeas de Etiologia Neurogênica – AFDN. In: Cardoso MCAF. *Disfagia Orofaríngea de causa neurogênica: verificação do índice de saturação de O2 na avaliação clínica fonoaudiológica*. [dissertação]. Santa Maria (RS): Universidade Federal de Santa Maria; 2004.
12. Lemos IO, Kurtz L, Bello JZ, Cardoso MCAF. The construction of a daily instrument to evaluate swallowing disorders in patients with dysphagia. *International Archives in Otorhinolaryngology.* 2013;17(Suppl 1):428.
13. Distrofia muscular, NINDS. Julio 2007. Publicación de NIH 07-77s. [atualizada em 2009 Dec 18; acesso em 2014 Jun 14]. Disponível em: http://espanol.ninds.nih.gov/trastornos/distrofia_muscular.htm.
14. Onófrio FQ, Oliveira JH, Reichert R, Arruda CA, Lopes AB, Freitas CPF et al. *Distrofia Muscular Orofaríngea: relato de caso*, Porto Alegre. 29ª Semana Científica do Hospital de Clínicas de Porto Alegre, 2009. [atualizada em 2009 Oct 29; acesso em 2014 Jun 14]. Disponível em: <http://hdl.handle.net/10183/21360>.
15. Saito FL, Gemperl R, Hiraki PY, Ferreira MC. Cirurgia da ptose palpebral: análise de dois tipos de procedimentos cirúrgicos. *Rev. Bras. Cir. Plást.* 2010;25(1):11-7.
16. Crary MA, Mann GDC, Groher ME. Initial psychometric assessment of a functional oral intake scale for dysphagia in stroke patients. *Arch Phys Med Rehabil.* 2005;86:1516-20.
17. Bodt MS, Wuyts FL, Heyning PHV, Croux C. Test-Retest of the GRBAS Scale: Influence of experience and professional background on Perceptual rating of Voice Quality. *J Voice.* 1997;11(1):74-80.
18. Lundy-Ekman L. Cérebro e aplicações clínicas. In: Lundy-Ekman L. *Neurociência: Fundamentos para reabilitação*. Elsevier Editora, 2008. P.378-97.
19. Ortiz KZ. Disartria. In: Ortiz KZ. *Distúrbios neurológicos adquiridos: Fala e deglutição*. São Paulo. Editora Manole, 2006. P. 54-72.
20. Plourde A. *La dystrophie musculaire oculopharyngée (DMOP)*. Québec: 2009. [atualizada em 2009 Apl 20; acesso em 2014 Ago 31]. Disponível em: <https://www.orpha.net/data/patho/Pub/fr/DMOP-FRfrPub664.pdf>.

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