

Case Report

Relato de Caso

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Keywords

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Descritores

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Manifestações neurológicas
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Dysfunctions of the stomatognathic system and vocal aspects in Fahr disease: case report

Disfunções do sistema estomatognático e aspectos vocais na doença de Fahr: relato de caso

ABSTRACT

The aim of this study is to report the case of a patient with Fahr's Disease in order to describe the main stomatognathic and vocal changes that can be found in individuals with this disease. In order to establish the diagnosis, an assessment of the conditions of orofacial motor system and speech production, as well the efficiency of swallowing, was realized. Based on these assessments, there were difficulties in coordinating and sustaining muscle during speech and presence of oropharyngeal dysphagia. Speech disorders found in Fahr's disease manifest themselves in complex and cover various aspects of phonological knowledge and the diseases that affect the basal ganglia have similar frames of speech-language disorders of the stomatognathic system, being able to present a picture of dysarthria.

RESUMO

O objetivo deste estudo consiste em relatar o caso de uma paciente com Doença de Fahr buscando descrever as principais alterações estomatognáticas e vocais que podem ser encontradas em indivíduos com essa doença. A fim de estabelecer o diagnóstico fonoaudiológico, foi realizada avaliação das condições motoras orofaciais e produção da fala, além de eficiência da deglutição. Com base nessas avaliações, observaram-se dificuldades na coordenação e na sustentação muscular durante a fala e presença de disfagia orofaríngea. Os achados fonoaudiológicos na Doença de Fahr manifestam-se de forma complexa, incluindo disfagias e disartria e as doenças que acometem os núcleos da base apresentam quadros semelhantes de alterações fonoaudiológicas do sistema estomatognático, podendo apresentar quadro de disartria.

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INTRODUCTION

Fahr's disease, also known as idiopathic basal ganglia calcification, is characterized by accumulation of calcium deposits in the regions of the basal ganglia, which may also affect regions of the caudate nucleus, the putamen, thalamus, the white matter, and the cerebellum^(1,2).

The appearance of calcium deposits, usually bilateral and benign, is associated with genetic, metabolic, or infectious changes. Among the pathologies that can also cause calcium accumulation, one can highlight hypothyroidism, pseudo-hypothyroidism, mitochondrial myopathy, Wilson's disease, lupus erythematosus, Down syndrome, and neurobrucellosis. Despite this, some individuals develop these calcium deposits and have no well-defined etiology, cases in which the cerebral calcification is characterized as Fahr's disease⁽¹⁻³⁾.

The diagnosis of Fahr's disease is considered when there is exclusion of other conditions that may also cause bilateral calcification of the basal ganglia and other brain regions. Brain imaging shows the presence of whitish concentrated radiopaque regions according to the affected area and with well-defined areas, although the highest concentration is observed in a particular region of calcification, such as the basal ganglia⁽¹⁻³⁾.

Regarding the clinical findings on the disease, the literature data show that there are changes in social behavior and motor functions, as well as motor and phonic tics and inappropriate behavior according to the situation experienced⁽⁴⁾, features that imply modifications in speech and orofacial motor functions.

The modification in speech is closely related to the ability to communicate orally and motor coordination. Patients with this disease show difficulty in articulating sounds due to the inability to coordinate motor functions⁽⁵⁾. It is known that injury to brain areas, especially those involving the coordination of movements, maintenance of motor rhythm, and muscle tone, such as the basal ganglia region, highly affected by Fahr's disease, can result in changes in muscle function, changing stomatognathic functions. In this disease of oral expressiveness, the individual has difficulty or inability to express themselves through speech because of the difficulty in maintaining speech⁽⁴⁻⁶⁾.

Thus, this study aims to report the case of a patient with Fahr's disease, seeking to describe the main stomatognathic characteristics and vocal aspects found.

CASE REPORT

This case study was conducted at the Neurology Ambulatory of Hospital Santa Clara – Complexo Hospitalar Santa Casa de Misericórdia de Porto Alegre by the Speech-Language Pathology and Audiology Team of Universidade Federal de Ciências da Saúde de Porto Alegre (UFCSPA).

For this study, approved by the research ethics committee of UFCSPA under protocol no. 1850/12, a patient with a medical diagnosis of hyperparathyroidism and Fahr's disease, under the care of Speech-Language Pathology and Audiology students of UFCSPA, was recruited. The patient, henceforth identified as I.C.G., female, 66 years old, was informed about the objectives and procedures of this study and signed the free and informed consent form.

In search of the characterization of her clinical condition, the patient was evaluated according the following protocols: Protocol for Speech-Language Pathology and Audiology Evaluation of Oropharyngeal Dysphagia of Neurogenic Etiology – AFDN⁽⁷⁾, Protocol of Dysarthria⁽⁸⁾, and Protocol Alpha (Montreal Protocol – Toulouse Alpha 1)⁽⁹⁾. The evaluations were performed by two observers independently at the same time. At the end of the assessment, results were compared and discussed to obtain the final data and presentation in this study.

Regarding the results of Protocol AFDN, good communicative interaction was observed, with the patient being able to understand and respond to inquiries made. Furthermore, it was found that the sensibility of speech organs was preserved, but the patient showed slowness of movements, especially in relation to the mouth opening and the tongue lateralization, also presenting sagging of the orofacial muscles. Regarding food intake, anterior mastication was observed for solid foods; difficulty in controlling saliva, with drooling through the labial commissures; multiple swallowing for liquid and soft foods; in addition to the anterior dribbling of liquids. By cervical auscultation, the presence of cervical noises, compatible with stasis of food, was observed, being manifested by the patient as a pharyngeal globus sensation. On the basis of the findings observed in the evaluation, the presence of oropharyngeal dysphagia, characterized by mild changes in oral control and pharyngolaryngeal dysmotility, was observed.

The same oral motor aspects were observed in the Protocol of Dysarthria. Regarding the vocal aspects evaluated in this protocol, the patient showed mixed oral and nasal breathing and a predominantly nasal resonance. The maximum phonation times found were low, with an average of 7 seconds, and the S/Z ratio was 0.8 seconds. Markers of vocal quality observed were: hoarseness, tremor, monotony, and nasality, caused by the decreased mobility of the soft palate. Also, little range of motion was observed in articulation. In evaluating combined basis involving breathing, phonation, articulation, and resonance, the patient showed slowness of speech, the occurrence of tremors, and pneumophonoarticulatory incoordination, which are similar to hypokinetic dysarthria.

Regarding the evaluation performed using Protocol Alpha, no variations suggesting alterations in understanding and expression of language were found.

Table 1 shows the main stomatognathic changes and vocal aspects observed in the case presented.

Table 1. Stomatognathic changes and vocal aspects observed in the case

Aspect assessed	Changes presented
Orofacial musculature	Preserved facial sensibility
	Slowness of movements (smaller mouth opening and tongue lateralization)
	Sagging of facial muscles
Mastication and swallowing	Difficulty in controlling saliva (drooling through labial commissures)
	Anterior mastication
	Multiple swallows for liquid and soft foods
	Anterior dribbling for liquids
	Presence of cervical noises compatible with stasis of food
Breathing and vocal production	Pharyngeal globus sensation
	Mild oropharyngeal dysphagia
	Mixed oral and nasal breathing
	Pneumophonoarticulatory incoordination
	Predominantly nasal resonance
	Maximum time of phonation reduced
	Hoarse, trembling and monotonous voice quality
Nasality due to low mobility of the soft palate	
	Slowness of speech and the presence of tremors

DISCUSSION

Currently, it is considered that the brain is a dynamic and plastic structure, and that the integration of systems occurs in a generalized way, i.e., the set of neurons in one region is able to carry out other functions⁽¹⁰⁾. In spite of this, the distribution of brain function occurs in an organized manner, with each region having a predominance to carry out brain commands and, therefore, to assume a function for a given region. Considering this aspect, a brain injury leads to loss of functional capacity of the individual, according to its location and extent⁽¹¹⁾.

The region of the basal ganglia is an extremely specialized location that, in terms of motor function, has as its main function the maintenance of muscle tone and movement coordination, together with the cerebellar system⁽¹²⁾. In relation to the functions of the stomatognathic system, diseases that affect this region also affect the orofacial motor functions and the capacity of expressiveness of speech, in addition to vocal aspects, and lead to difficulties in conducting the food bolus because of the change in muscle tone. Among the most studied diseases that affect this system are Parkinson's disease, extensively studied in the Speech-Language Pathology and Audiology field, and, more recently, diseases that result in the degeneration and calcification of the region, such as Fahr's disease^(1,12,13).

It is observed that the impairment of brain structures involved in the movement coordination process causes disability in performing combined motor functions, because the integration of motor information does not occur in an orderly manner⁽¹³⁾. Specifically regarding stomatognathic functions, this incoordination is evidently expressed, because all acts depend on a

complex combination of many muscles that will perform these functions, especially in swallowing and speech⁽¹⁴⁾.

According to the data in this case report, speech-language-related changes to the stomatognathic system presented by individuals affected by the disease are very similar to the characteristics of individuals with Parkinson's disease, such as the slowness of movements, changes in speech modulation with the presence of tremors and altered voice quality, and it is possible to observe a condition similar to the diagnosis of hypokinetic dysarthria⁽¹⁴⁾.

Other reports in the literature on individuals with Fahr's disease corroborate these findings, demonstrating that lesions in the basal ganglia due to calcifications cause changes in speech, oral motor functions, speech and swallowing functions, manifesting itself in a quite characteristic manner, which makes it possible to describe a fairly typical communicative and functional profile, but with varying degrees of impairment in accordance with the extent of injury^(1-3,5,15).

It is noteworthy that despite the distinction of the pathophysiology of the diseases discussed, the functionality of the system is compromised, generating important communicative impact. Thus, it is possible to observe that, according to reports in the literature and the clinical characteristics presented by the patient described, regardless of the type of impairment, speech-language features tend to perform similarly as reported in findings in individuals with Parkinson's disease, and this an important marker for suspecting diseases that do not complete a typical clinical condition of this disease, but that indicate changes in the basal ganglia, such as Fahr's disease⁽¹²⁻¹⁴⁾.

On the basis of these descriptions, one can highlight the importance of the speech-language professional in the accurate evaluation of stomatognathic functions. It appears that the association between changes in motor functions and coordination of movements, observed by changes of stomatognathic features and vocal aspects, may characterize some of the changes in the basal ganglia.

FINAL COMMENTS

The reports of speech-language disorders in Fahr's disease in the medical literature are still scarce and often describe difficulties in coordinating movements and changes in muscle activity. The case report in this study may facilitate the understanding of characteristics of Fahr's disease of interest to the Speech-Language Pathology and Audiology community, guiding the approach of medical action. This allows for interdisciplinary intervention, including approach from the Speech-Language Pathology and Audiology aspect, which should be preconized to minimize the difficulties described, aiming, among other things, to an improved quality of life.

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**All participants took part in the collection and interpretation of the collected data, as well as in the writing of the manuscript.*

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