Case Report

Relato de Caso

Spinocerebellar ataxia type 7

Ataxia espinocerebelar tipo 7

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Keywords

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ABSTRACT

The aim of this study was to verify the possible alterations observed in a case of spinocerebellar ataxia type 7. The patient was referred to the Laboratory of Neurotology of Universidade Tuiuti do Paraná (Brazil), and was submitted to the following procedures: anamnesis, otoscopy, auditory and vestibular assessments. The case described is a 34-year-old woman with a genetic diagnosis of spinocerebellar ataxia type 7, who referred imbalance when walking, speech difficulties, headache, dizziness, and dysphagia. The audiological evaluation showed normal hearing thresholds and tympanometric curve type "A", with bilateral presence of stapedius reflex. In the vestibular evaluation, it was observed the presence of spontaneous and gaze nystagmus with central characteristics, altered optokinetic nystagmus and pendular tracking, and hyperreflexia during the caloric test. Labyrinth alterations were found, indicating central vestibular system affection and evidencing the importance of this evaluation. The existence of a possible relationship between the findings and the neurotological symptoms presented by the patient leads us to a new issue, that is, the importance of the applicability of rehabilitation exercises that act in central structures of neuroplasticity. These exercises accelerate and stimulate natural compensation mechanisms, which may provide the patient with ataxia a better performance of its functions.

Descritores

Degenerações espinocerebelares
Ataxias espinocerebelares/diagnóstico
Ataxias espinocerebelares/complicações
Doenças vestibulares/etiologia
Eletronistagmografia/utilização
Nistagmo fisiológico

RESUMO

O objetivo deste estudo foi verificar possíveis alterações vestibulococleares em um caso de ataxia espinocerebelar tipo 7. O paciente foi encaminhado para o Laboratório de Otoneurologia da Universidade Tuiuti do Paraná e foi submetido aos seguintes procedimentos: anamnese, inspeção otológica, avaliações audiológica e vestibular. Trata-se de indivíduo do gênero feminino, de 34 anos de idade, com diagnóstico genético de ataxia espinocerebelar tipo 7, que referiu desequilíbrio à marcha, dificuldade para falar, cefaléia, tontura e disfagia. Em avaliação audiológica, apresentou limiares auditivos dentro dos padrões de normalidade e curva timpanométrica do tipo "A" com presença dos reflexos estapedianos bilateralmente. No exame vestibular, observou-se presença de nistagmos espontâneo e semi-espontâneo com características centrais, nistagmo optocinético e rastreio pendular alterados e hiperreflexia à prova calórica. Constatamos alterações labirínticas que indicam afecção do sistema vestibular central e evidenciam a importância dessa avaliação. A existência da possível relação entre os achados com os sintomas otoneurológicos apresentados pela paciente nos remete a uma nova questão, ou seja, à importância da aplicabilidade dos exercícios de reabilitação que atuam em estruturas centrais de neuroplasticidade. Eles aceleram e estimulam mecanismos naturais de compensação, que poderão proporcionar ao portador de ataxia um melhor desempenho de suas funções.

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INTRODUCTION

Spinocerebellar ataxias (SCA) are a heterogeneous group of neurodegenerative diseases. They are characterized by the presence of progressive cerebellar ataxia, whose initial clinical manifestations are eye disorders and the deterioration of balance and coordination[1].

Ataxias can be classified in: sensitive, frontal, vestibular and/or labyrinth and cerebellar[2]. In the SCA type 7, the cerebellar ataxia is associated with progressive visual loss that results from the retina degeneration (progressive macular degeneration). Pyramidal signs, ophthalmoplegia, Parkinsonism, slow saccades and weakness[3-5] can also be associated with it. Studies[4,5] have identified the occurrence of olivopontocerebellar degeneration associated with the reduction of retinal ganglion cells and pigmentary macular dystrophy. The clinical condition may develop during childhood until the 60 years of age, and the younger the individual affected, the faster the disease progresses[5].

SCAs have a variable geographic prevalence. The SCA type 7, whose identified chromosome is 3p located at 3p14, gene SCA 7, mutation CAG, ataxin-7 protein, has a high incidence in Sweden, Finland, United States and China[1,4-6].

Some ataxias are caused by genetic abnormalities, such as the case of SCA type 7. The SCA type 7 is an autosomal dominant neurodegenerative disorder caused by mutations characterized by the presence of a trinucleotide repeat, that is expanded and unstable in the encoded region of the gene[1,7]. Such protein mutation results in neuronal loss, which mainly affects the cells of the cerebellum, regions of the brainstem, inferior olivary complex and retina[7].

The identification of a patient with SCA is done through the multiplicity of clinical forms and frequent associations that may occur due to the evolution of the disease. Nowadays, 30 types of SCA have been diagnosed, from which type 2 (characterized by cerebellar atrophy and the presence of peripheral neuropathy) and type 3 (characterized by cerebellar, pons, globus pallidus, frontal and temporal lobe atrophy), are the most common forms according to neuroimaging studies. Type 3 is the most common one in Brazil, and the severity of clinical manifestations and the age at which the symptoms start to appear will depend on which parent the expanded allele was inherited from[1,8].

The essential element for the vestibular analysis is the nystagmus, i.e. set of eye movements with fast and slow components, opposite directions which happen in turns. Through the tests that comprise the vestibular examination it is possible to assess the relationship of balance to the function of the posterior labyrinth, vestibular branches of cranial nerve VII, vestibular nuclei of the floor of the fourth ventricle, vestibular pathways and, above all, the vestibulo-oculomotor, vestibulocerebellar, vestibulospinal and vestibular proprioceptive-cervical interrelations. In central neurotological syndromes it is noticeable that the signs of affection of the central vestibular system are prevalent in several labyrinth tests, which does not occur in peripheral syndromes[9].

The SCA type 7 causes oculomotor changes, especially in both saccadic and slow pursuit movements, which thus show important alterations in structures located in the region of the cerebral cortex[7]. The SCAs are part of a list of diseases that show important alterations which concern the field of Speech Therapy, more specifically neurotology. This study is justified due to its contribution to the formulation of knowledge that subsidizes therapeutic and evaluative procedures aimed at the overcoming and/or reduction of the adverse effects of this disease.

Given the above considerations, the aim of this study is to examine the vestibulocochlear alterations observed in a case of SCA type 7.

CLINICAL CASE PRESENTATION

In the Neurotology Sector of the Universidade Tuiuti do Paraná (UTP), in the city of Curitiba, a 34-year-old woman with a genetic diagnosis of spinocerebellar ataxia type 7, who referred imbalance when walking, speech difficulties, headache, dizziness, and dysphagia was evaluated. The ataxia diagnosis was carried out through genetic testing with the use of the PCR (Polymerase Chain Reaction).

The research was approved by the Research Ethics Committee of the UTP under protocol number 058/2008, and carried out after the patient's agreement through the signature of the informed consent form.

The following procedures were carried out: anamnesis, otorhinolaryngologic, audiological and vestibular evaluations, and acoustic immittance measures.

Anamnesis

A questionnaire was administered with emphasis on neurotological signs and symptoms.

Otorhinolaryngological evaluation

The otorhinolaryngological evaluation was carried out with an aim to rule out any alteration that could interfere with the examination.

Audiological evaluation

The conventional pure-tone audiometry was carried out with the use of a two channel Itera audiometer from Madsen-GN Otometrics®, with TDH-39 headphones, and thresholds in dBNA. The piece of equipment was calibrated according to ISO Standard 8253. Then, both the speech recognition threshold and the percentage index of speech recognition were obtained in a sound-proof booth in order to prevent the interference of strange noises with the evaluation. Criteria were established for the determination of the degree and type of hearing loss[10].

Acoustic immittance measures

This procedure was carried out in order to evaluate the integrity of the tympanic-ossicular system through the tympanometric curve and the acoustic reflex research, by using Jerger's criteria for the interpretation of the results[11]. The

piece of equipment used was the Madsen Otoflex 100™ impedanciometer.

Vestibular evaluation

The patient underwent several tests that comprise the vestibular examination. At first, dizziness and spontaneous and semi-spontaneous positional nystagmus were searched for.

In order to carry out the vectoelectronystagmography (VENG), a thermo-sensitive VN316 Berger® device was used, with three recording channels. An active electrode was placed in the lateral angle of each eye and on the frontal middle line and bonded with electrolytic paste, forming an isosceles triangle. This allows for the identification of horizontal, vertical and oblique ocular movements and mainly the measurement of the angular speed of the slow component (ASSC) of nystagmus.

A Ferrante® decreasing pendular rotary chair, an EV VEC Neurograff® visual stimulator, and an NGR 05 Neurograff® air otocalorimeter device were used. The following eye and labyrinth tests were carried out during VENG[12]: eye motion calibration, search for spontaneous and semi-spontaneous nystagmus, search for pendular tracking, search for optokinetic nystagmus, pre and post-rotational, pre and post-caloric. The caloric stimulation in each ear at 42ºC and 18ºC lasted 80 seconds for each temperature. The responses were recorded with patients closing their eyes and then with their eyes opened, in order to observe the inhibitory effect of eye fixation (IEEF).

In the vestibular examination, we were able to notice: absence of positional nystagmus and dizziness; regular eye motion calibration, presence of spontaneous nystagmus with eyes opened, of the right-horizontal type with ASSC at 8º/s and absence of it with eyes closed; presence of semi-spontaneous nystagmus of the multiple type, right-horizontal, non fatiguing, without paroxysm, without latency and with ASSC at 8º/s; pendular tracking type IV; abolished optokinetic nystagmus; pre-rotational nystagmus – stimulation of the symmetric lateral semicircular canals, with nystagmus frequency (counterclockwise CCW=6 and clockwise CW=8) with directional preponderance of the nystagmus of 14% to the right. Stimulation of the symmetric posterior semicircular canals, with nystagmus frequency (CCW=9 and CW=10) with directional preponderance of the nystagmus of 5% to the right.

Stimulation of the symmetric anterior semicircular canals, with nystagmus frequency (CCW=6 and CW=10) with directional preponderance of the nystagmus of 25% to the right and post-caloric nystagmus with temperature at 42ºC right ear (RE) with ASSC at 20º/s, 42ºC left ear (LE) with ASSC at 18º/s, 18ºC RE with ASSC at 40º/s and 18ºC with ASSC at 34º/s. The patient didn't refer dizziness and showed the presence of IEEF in the four stimulations.

As a synthesis of the findings: spontaneous nystagmus with eyes opened, semi-spontaneous multiple type, abolished optokinetic, pendular tracking type IV and caloric hyperreflexia.

Conclusion of the examinations

In the audiological evaluation the patient showed the presence of hearing thresholds within normal standards and tympanometric curve type "A" with bilateral presence of stapedius reflex, and the vestibular examination showed "suggestive of bilateral irritative central vestibular dysfunction".

DISCUSSION

Progressive disorders are characterized by the degeneration of the spinocerebellar tracts. Amongst the neurological manifestations present, the most frequent ones are visual loss and nystagmus[6].

The cerebellum receives afferents from visual, auditory, vestibular, brainstem somatosensory structures, and information from somatosensory receptors of the limbs and motor, premotor, prefrontal areas of the cerebral cortex. It has three anatomical regions: medial zone (worm of the cerebellum and fastigial nucleus), which responds for the control of posture, balance, locomotion; intermediate zone (intermediate hemisphere and interpositus nucleus), responsible for the control of movements and ipsilateral discrete reflexes of the limbs; and lateral zone (lateral hemisphere and dentate nucleus) responsible for motor planning and for the complex movements of the limbs guided visually[13].

Several eye movements are performed by specific regions of the cerebellum and such movements may be classified as saccadic, slow pursuit or convergence. Saccadic movements are voluntary rapid eye movements in which the eyes move from one visual target to the other, or reflexes in response to a visual target that intensely appears in the peripheral visual field. The slow pursuit movement, aims to reduce visual image towards velocities that are too slow to allow clear view. Medial and floccular regions of the cerebellum are responsible for these movements. The convergence movement allows breaking the horizontal parallelism of the eyes for visual targets seen at close range, and both the medial and intermediate regions of the cerebellum[14] are responsible for it.

The unilateral extraocular movements are performed by six extrinsic muscles of the eye bulb. These muscles are innervated by three oculomotor nerves (cranial nerves III, IV and VI), which are located in the brainstem and are originated from the nuclei, which receive information from the cerebellar cortex, as well as from the vestibular nuclei[14,15].

Amongst the functions performed by the cerebellum is the control of eye movements. Any abnormality of these functions may cause oculomotor alterations, such as nystagmus. In this case, the vestibular findings (presence of spontaneous nystagmus with eyes opened, semi-spontaneous of the multiple type; pendular tracking type IV (anarchic); abolished optokinetic nystagmus and caloric hyperreflexia) indicate central alterations originated from the degeneration of the cerebellum and its afferent and efferent pathways.

The findings from this study contribute to the thought on the importance of evaluation and speech-language pathology monitoring of these patients, because they may show important otoneurologic and stomatognathic system alterations. There are no reports of vestibular referring to SCA type 7 in neither national nor international literature. Besides, there is

no treatment that delays or prevents the progression of the disease.

FINAL COMMENTS

In the case studied, we found labyrinth alterations that indicate affection of the central vestibular system and point to the importance of this evaluation. The existence of a possible relationship between the findings and the neurotological symptoms presented by the patient leads us to a new issue, that is, to the importance of the applicability of rehabilitation exercises that act in central structures of neuroplasticity. These exercises accelerate and stimulate natural compensation mechanisms, which may provide the patient with ataxia a better performance of its functions.

In view of the above, we evidence the importance of the speech therapist's presence at the diagnosis and monitoring/ treatment of patients with SCA, given that the neurotogical symptoms appear in the initial stages of this disease.

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