Distonia Cervical: A Doença Infrequente como Paradigma dos Cuidados Diferenciados em Cuidados de Saúde Primários

Cervical Dystonia: The Infrequent Disease as a Paradigm of Differentiated Care in Primary Health Care

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RESUMO

A distonia compreende um conjunto de movimentos complexos anormais, sendo a maioria de causa idiopática. A terapêutica farmacológica não mostra resultados favoráveis, pelo que as injeções de toxina botulínica são o tratamento de escolha. Neste artigo, relatamos um caso de distonia cervical com resposta favorável a levodopa.

Trata-se de um caso de distonia cervical idiopática, sem resposta favorável a injeções de toxina botulínica e a vários fármacos, que foi encaminhado para um centro de estimulação cerebral profunda e, simultaneamente, para a consulta de dor crónica da unidade de saúde familiar. Nesta consulta foi tentado tratamento com levodopa com resposta favorável, evitando o procedimento invasivo.

Os resultados positivos alcançados, vão de encontro a outros autores que assumem que a levodopa pode ser tentada em casos atípicos de distonia. Este caso mostra a importância dos cuidados de saúde primários cuja intervenção evitou um procedimento invasivo.

PALAVRAS-CHAVE: Distonia/tratamento farmacológico; Levodopa/uso terapêutico; Torcicolo/tratamento farmacológico

ABSTRACT

Dystonia is a comprised of complex abnormal movements, with most cases being idiopathic. Pharmacological therapy has unfavorable results, and therefore, local botulinum toxin injections are the treatment of choice. In this paper, we report a case of cervical dystonia with a favorable response to levodopa.

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It is a case of idiopathic cervical dystonia, without a favorable response to local injections of botulinum toxin and a lot of drugs, which was sent to a deep brain stimulation center and, simultaneously, to a Chronic Pain Medical Consultation at the family health unit. At this consultation, treatment with levodopa was tried with a favorable response, avoiding the invasive procedure.

The positive results achieved using levodopa agree with other authors who assume that levodopa could be attempted in atypical dystonia. This case highlights the importance of primary healthcare, which can prevent the need for an invasive procedure.

KEYWORDS: Dystonia/drug therapy; Levodopa/therapeutic use; Torticollis/drug therapy

INTRODUCTION

Dystonia is considered a sign or symptom, which is comprised of complex abnormal and dynamic movements of different etiologies. A specific cause is identified in approximately 28% of patients, which only occasionally results in specific treatment.[1] Idiopathic cervical dystonia (ICD), the most common adult segmental dystonia is characterized by recurrent or sustained abnormal head postures.[2,3] Whereas the cause of dystonia in most affected individuals remains elusive, a genetic etiology is identified in a larger proportion of patients in whom the disorder was previously classified as "idiopathic".[4] A lot of drugs have been tried for treatment with unfavorable response and therefore, local botulinum toxin injections are currently the treatment of choice.[5]

In this paper, we report a case of cervical dystonia that showed a favorable response to levodopa.

CASE REPORT

The patient, a woman born in 1958, began to suddenly develop blepharospasm at the age of 48 (2007), with a partial favorable response to botulinum toxin. At the time, no pharmacologic therapy was used, nor was she submitted to any medical exam. In 2015, she started, once again, to show involuntary and continuous movements of the neck, backwards and to the left. She was admitted for investigation, and, having excluded any primary cause, it was concluded that it was a case of idiopathic cervical dystonia. She started a series of medications with higher and higher doses of clonazepam, cinnarizine and trihexyphenidyl, never with a favorable response. Periodically, she received botulinum toxin injections, not yet with favorable response. Bearing in mind the negative impact that these involuntary movements were having on the patient´s day-to-day life and the fact that no treatment seemed to be having any result, she was then referred to the Center for Deep Brain Stimulation. The family doctor remained a constant presence throughout this process and simultaneously indicated the patient for Chronic Pain Medical Consultation in his own family health unit. On the first medical appointment, the patient was being treated with clonazepam 2 mg (twice a day), diazepam 10 mg (once a day), pantoprazole 20 mg (once a day), tapentadol 50 mg (once a day) and was using a neck brace to relieve her discomfort. When questioned, she referred to the involuntary movements were what most bothered her. As to the pain, she attributed 8 on a scale of 10, according to the Visual Analogue Scale. When submitted to a physical exam, it was clear that she maintained both the neck movements, backwards and to the left, and the bilateral blepharospasm. She started baclofen 10 mg (twice a day), morphine 10 mg (sos) and the initial dose of tapentadol to 100 mg (twice a day). The remaining therapeutics continued the same as before, as well as the periodic botulinum toxin injections. Monthly consultations became part of her medical plan to adjust medication in case it was necessary. In April 2016, the pain was under control, but the hyperkinetic movements continued and deprived her from working. Due to the fact that this dystonia was refractory to therapeutics used until that moment, a different therapeutic treatment was initiated with carbidopa/levodopa (25/100), twice a day. Simultaneously, the patient also began a physical and rehabilitation program. In June 2016, the patient was still under controlled pain but there was an improvement of the hyperkinetic symptoms. The dose was increased to 5 tablets a day (2+1+2). In 2017, it became evident that the treatment with carbidopa/levodopa 25/100 mg was having satisfactory results on the patient, thus avoiding deep brain stimulation. At this stage, the patient started carbidopa/levodopa 50/200 twice a day. One year later (2018) the dose was reduced to one tablet of carbidopa/levodopa 50/200 in the morning and one tablet of carbidopa/levodopa 25/100 in the evening. In 2020, the dose was again reduced to carbidopa/levodopa 25/100 every 12/12 hours, a dose that the patient is currently maintaining. At the moment, the patient states no pain, and the hyperkinetic movements seem to have significantly improved.

DISCUSSION

The main limitations inherent in case reports are the absence of a control group and the fact that they do not allow for statistical analysis.

When analyzing this case, it was clear that there were symptomatic improvements after having introduced a dopamine agonist. However, the author consider that rehabilitation program and botulinum toxin injections were also fundamental in the success of this case. It was then possible to avoid an invasive procedure (deep brain stimulation), giving back the life quality she had lost.

Dopa-responsive dystonia typically occurs in childhood, starts in the lower limbs and with marked diurnal variation. Other Parkinson's symptoms can occur and there are often other family members affected.[3,6] It appears that our patient was an atypical case of dopa-responsive dystonia, since the initial symptoms occurred during adulthood. Besides, the patient never showed kinetic alterations of the members, nor is there any knowledge of diurnal variations of the symptoms. Up to the present date, there seems to be no other family member affected. The case is unusual for these reasons.

Several genes and genetic tests are under study in this area, although none that can the diagnose of dopa-responsive dystonia.[3] The *GCH1* gene (located on chromosome 14q 22.1-q 22.2) is a potential candidate gene for the disorder.[7] Our patient is still under genetic study.

Thus, we conclude that further research is necessary to correctly diagnose this disorder. Bearing in mind the positive results achieved using levodopa, we agree with other authors who assume that a therapeutic trial of levodopa could be attempted in atypical cases.

This case shows the importance of primary healthcare, whose intervention avoid an invasive procedure.

RESPONSABILIDADES ÉTICAS

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