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HEMIFACIAL SPASM IN A PATIENT WITH NEUROFIBROMATOSIS AND ARNOLD-CHIARI MALFORMATION

A unique case association

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ABSTRACT - *Background:* The association of hemifacial spasm (HFS), Chiari type I malformation (CIM) and neurofibromatosis type 1 (NF1) has not been described yet. *Case report:* We report the case of a 31-year-old woman with NF1 who developed a right-sided HFS. On magnetic resonance imaging (MRI) a CIM was seen without syringomyelia. The patient has been successfully treated with botulinum toxin type A injections for 5 years without major side effects. *Conclusion*: Clinical features of HFS, CMI and NF1 are highlighted together with their possible relationship. Also, therapeutic strategies are also discussed.

KEY WORDS: hemifacial spasm, neurofibromatosis, Arnold-Chiari malformation.

Espasmo hemifacial em paciente com neurofibromatose e malformação de Arnold-Chiari: uma associação rara

RESUMO - *Introdução:* A associação entre espasmo hemifacial (EHF), malformação de Chiari tipo I (MCI) e neurofibromatose tipo I (NFI) ainda não foi descrita. *Relato do caso:* Relatamos o caso de mulher com 31 anos com NFI que desenvolveu EHF à direita. Na ressonância magnética (RM) foi observada MCI sem seringomielia associada. A paciente foi tratada com sucesso com toxina botulínica tipo A por 5 anos sem efeitos colaterais. *Conclusão*: Ressaltamos as características clínicas do EHF, MCI e NFI assim como uma possível relação entre elas. Além disto, discutimos também estratégias terapêuticas.

PALAVRAS-CHAVE: espasmo hemifacial, neurofibromatose, malformação de Arnold-Chiari.

Hemifacial spasm (HFS) is a common movement disorder characterized by involuntary tonic or clonic contractions of the facial muscles innervated by the ipsilateral seventh nerve. In the majority of cases HFS is associated with underlying vascular abnormalities around the root exit zone of the facial nerve. Secondary causes of HFS are very unusual[1]. Chiari type I malformation (CIM) originates from a developmental pathological condition and is characterized by caudal migration of the cerebellar tonsils. There are few case reports on the literature of HFS associated to CIM[2-4]. Neurofibromatosis type 1 (NF1), on the other hand, is a genetic autosomal dominant disease (loci 17q11.2) in which individuals develop both benign and malignant tumors at an increased frequency[5]. Single case reports describes the association of CIM and NF1[6]. The association of CMI, NF1 and HFS, however, has not been described yet.

We report an unique case of HFS probably related to CIM in a patient with NF1. We also highlight that this association may not be merely fortuitous and that even in this scenario long-term use of botulinum toxin type A (BTX-A) may successfully relief symptoms of facial spasm.

CASE

A Brazilian 31-year-old white woman was referred with a 4-year history of paroxystic and irregular facial twitches that affected firstly her right upper face and afterwards the ipsilateral lower face, without sleep improvement. She had no previous history of peripheral facial palsy and was not taking any medication. At the age of fifteen she noticed few spots on her back skin. There were no other mem-

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bers of her family with movement disorders but her elder sister had the same skin lesions. Clinical examination showed numerous neurofibromas especially on the face, chest and back surfaces. There were also six café-au-lait macules. Based on the skin lesions and family history of a first-degree relative with the same disease, clinical diagnosis of NF1 was made. Neurological examination showed a right-sided HFS. There were no other abnormalities in the remaining cranial nerves, such as in cerebellar functions and other long motor or sensory tracts. On magnetic resonance imaging (MRI) a CIM was seen without syringomyelia (Figure). The cerebellar tonsils were herniated caudally until the top level of C2. Treatment was started with BTX-A after informed consent and permission for data publication. Five years later a great improvement is still noticed without major side effects. The patient signed informed consent.

DISCUSSION

Our case report describes the association between HFS, CIM and NF1. To our knowledge this is the first case on the literature with this association. Secondary HFS is uncommon and among the main causes tumors of the cerebello-pontine angle (CPA) such as meningiomas, epidermoid cysts or lipomas are well recognized. CIM is also seldom associated with HFS[2-4]. Current evidence support that narrowing of the posterior fossa could be one of the mechanisms responsible for HFS[8]. Moreover, patients with Asian origin have shallow posterior fossa and it seems that the incidence of HFS in this population is higher[1]. In Brazil, subjects of the northeastern region are at greater risk for developing CIM since they have a singular phenotype characterized by shorter neck and shorter stature[9]. Our patient was born in this region of Brazil but on clinical examination her neck and body stature were on the average size. Therefore, we believe that narrowing of cranio-cervical space due to hindbrain herniation might have contributed to the development of HFS in our case and that CIM was not associated to the classical northeastern Brazilian phenotype.

CIM has many neurological presentations. The main clinical symptoms of CIM are related to the compression of neural or dural structures by the inferiorly herniated cerebellar tonsils or may be related to the associated syringomyelia. Headache is the most frequent symptom followed by weakness, sensory abnormalities, lower cranial nerve dysfunction and "downbeating" nystagmus[10].

Colpan and Sekerci reported a case of an 18-years-old boy who presented with a 10-month history of HFS due to CIM without syringomyelia. Posterior fossa decompression, C1 laminectomy and duraplasty were performed with success and subsequent disappearance of HFS[4]. Surgical risks of facial nerve decompression (impaired hearing and facial weakness), however, should not be overlooked. In our case, neurosurgical procedure was not recommended due to excellent relief after BTX-A injections. Thus, if we consider that the good resolution of facial spasms were obtained and also that posterior fossa decompression may cause serious side effects such as hearing loss and facial palsy, each case may have to be separately analyzed to better decide treatment options.

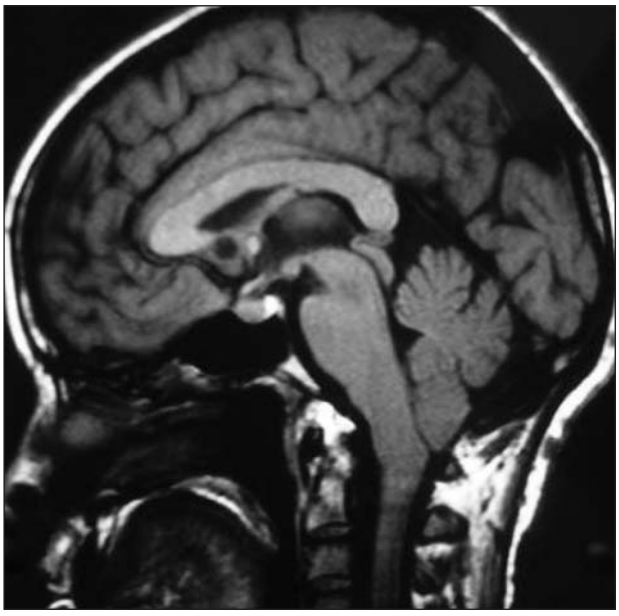

Figure. Sagittal T_1-weigthed brain MRI scan, showing Chiari type 1 malformation. Ectopia of the cerebellar amygdalae descending to top level of C2 is seen.

Apart from HFS and CIM our patient was diagnosed with NF1 which, similarly to CIM, has challenging clinical presentations. The recognized neurological complications of NF1 are nerve sheath tumors, optic gliomas, cognitive deficits, "unidentified bright objects" (hyperintense T_2 brain MRI lesions), seizures and macrocephaly[5].

Bony lesions are very frequent among patients with NF1 and are also considered as part of the clinical diagnostic criteria for NF1[5]. According to recent data CIM might not be a casual association to NF1. This is supported by a prevalence study on CIM and NF1[7]. It was shown that in 130 young patients with surgically addressed CIM 5.4% of them had NF1. On the other hand, 198 patients diagnosed with NF1 had in 8.6% of the cases a CIM radiologically confirmed, suggesting that CIM and NF1 are not spurious findings but rather true associations.

Thus, one may consider that CIM is part of the of NF1 spectra and ultimately may be responsible for

HFS. It is our opinion that patients with NF1 and HFS should be routinely scanned, preferably with MRI, with the purpose to find cranio-cervical malformations or tumors arising from the CPA.

In conclusion, although the majority of cases of HFS are related to vascular abnormalities around the root exit zone of the ipsilateral facial nerve, secondary cases due to CIM should always be remembered, especially in patients with NF1. Moreover, even in this scenario, management with BTX-A may provide convincing relief.

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