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CASE REPORT

Bilateral functional ptosis in adolescence: a case report

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Abstract

Introduction: Bilateral eyelid ptosis is an extremely rare form of functional neurological disorder (FND). FNDs are a group of disorders that have in common the absence of a structural pathology of the nervous system and the presence of common neurological symptoms, and can be somewhat challenging in regard to diagnosis, approach, and treatment. **Case report:** We present the case of an adolescent diagnosed with sudden asymmetric ptosis. To consider the different potential diagnoses (cranial polyneuropathy, mitochondrial disease, and neuromuscular junction disorder), the patient underwent several complementary diagnostic tests, which gave inconclusive results. Although there had been no recent stressful events, the observed inconsistency and incongruity, as well as the observation of certain details in the neurological exam, led to a diagnosis of FND. Six days after clinical discharge, the patient recovered suddenly and spontaneously from all neurological problems. **Discussion:** Relevant symptoms and an accurate assessment of psychological factors are fundamental to ensuring a correct diagnosis and positive prognosis.

Keywords: Functional neurological disorder. Functional eyelid ptosis. Child and adolescence psychiatry. Neuropediatrics. Case report.

Ptose funcional bilateral na adolescência: um reporte de caso

Resumo

Introdução: As perturbações neurológicas funcionais (PNF) são um grupo de doenças que têm em comum a ausência de patologia "estrutural" do sistema nervoso e a presença de manifestações neurológicas comuns, constituindo um desafio no que concerne ao diagnóstico, abordagem e tratamento. **Relato de caso:** Apresenta-se o caso clínico de um adolescente com ptose assimétrica de início súbito. Tendo em conta os diagnósticos diferenciais considerados (polineuropatia craniana, doença da placa motora e doença mitocondrial), realizou vários exames complementares que foram inconclusivos. Apesar de não se identificarem eventos stressores recentes, a inconsistência e incongruência observadas, e o reconhecimento de detalhes no exame neurológico permitiu o diagnóstico de PNF. Seis dias após alta, o doente recuperou súbita e espontaneamente de todos os défices neurológicos. A ptose palpebral bilateral é uma apresentação extremamente rara de PNF. **Discussão:** A fenomenologia clínica e a correta avaliação de fatores psicológicos são aspetos essenciais para estabelecer o diagnóstico e para um bom prognóstico.

Palavras-chave: Perturbação neurológica funcional. Ptose palpebral funcional. Pedopsiquiatria. Neuropediatria. Adolescência.

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Keypoints

What is known

- Functional neurological disorders (FNDs) are a highly prevalent group of disorders that present certain challenges in terms of diagnosis, approach, and treatment.
- Eyelid ptosis is a rare form of FND, with only a few cases of functional unilateral ptosis reported.

What is added

- Until this article was published, no cases of bilateral functional ptosis during childhood had been published; this is a unique case.
- Diagnosis of FND without an identified recent stressful event or associated psychopathology – factors that increase the difficulty in correct diagnosis and management.
- Clear and complete description of the clinical judgement behind the evaluation and diagnosis of this case report, enabling the reader to acquire knowledge about functional ptosis and its differential diagnosis.

Introduction

Functional neurological disorders (FNDs) are a group of disorders that have in common the absence of a "structural" pathology of the nervous system and the presence of often persistent and disabling neurological manifestations[1]. These are a group of highly prevalent disorders, representing 16% of the total number of referrals for Neurology consultations[2]. They can be somewhat challenging in terms of diagnosis, approach, and treatment[3], which can result in an excessive consumption of time and resources.

In the last decade, there has been a paradigm shift, recommending a positive approach to FND diagnosis, through the recognition of clinical patterns, rather than a diagnosis based on exclusion or the bizarre and/or unusual character of the semiology[4]. A better understanding of the pathophysiological and neurobiological mechanisms associated with FND[5], such as the identification of alterations in cerebral perfusion or metabolism in associated areas[4], has led to a lesser appreciation of psychological factors as a single and causal explanation. The terms 'conversion' and 'psychogenic' have been replaced by the term 'functional'[6,5], and the presence of psychopathology or the identification of a precipitating stressor is not currently required for diagnosis[7].

The most common manifestations of FND in the field of neuro-ophthalmology are blindness, diplopia, and ophthalmoparesis[8]. Eyelid ptosis is a rare form of FND, with only a few cases of functional unilateral ptosis reported and, until this article was published, no cases of bilateral functional ptosis during childhood had been reported. We present the case of a 14-year-old boy admitted to the Emergency Unit with asymmetric palpebral ptosis.

Clinical case

A 14-year-old male adolescent was admitted to the Emergency Unit with a sudden onset of asymmetric palpebral ptosis, frontal headache, and generalized asthenia. There was no fatigability, appendicular weakness, fever, or symptoms suggesting increased intracranial pressure. Four episodes of right eyelid ptosis in the previous two years were reported, in the context of a respiratory infection of the upper tract, from which he completely recovered following a cycle of antibiotic therapy. No personal history of head or face trauma was reported. The young man lived with his mother and father and was undergoing his 9th year of schooling, displaying medium-low school performance, with several changes in the educational establishment in the last year being described, and periods of school absenteeism.

The neurological examination carried out during admission showed bilateral eyelid ptosis, almost complete closure of the right eye and the left eye had coverage of about one third ptosis of the right eyebrow and elevation of the left eyebrow, more pronounced eyelid creases on the right side, absence of folds on the right in the contraction of the frontalis muscle, and hypoesthesia of the right hemiface including the angle of the mandible. The remaining neurological examination displayed no further changes.

The young man was admitted to the Adolescents Unit and underwent several complementary diagnostic tests, including orbital and cranioencephalic CT and MRI, electromyographic study, and laboratory evaluations (anti-acetylcholine receptor antibodies, ACE measurement, and study of mitochondrial DNA deletions) that were considered unaltered. He underwent a therapeutic test with pyridostigmine (15 mg, 4 id, in one day) that come out negative. During hospitalization, in addition to slight fluctuations, a single episode of total ptosis reversal was observed after the eyelid was forcedly closed and subsequently opened, lasting a few seconds. The Child and Adolescence Psychiatry and Psychology departments followed up his development, and no recent stressful events were identified.

Six days after discharge, in the context of a Neuropediatric Appointment, a sudden and spontaneous recovery of all

neurological deficits was observed. The patient is now accompanied by a district hospital, with no recurrence, as evaluated by telephone interview.

Discussion

Bilateral eyelid ptosis is an extremely rare manifestation of FND[8], especially during childhood[9]. The differential diagnosis of bilateral eyelid ptosis is extensive[10], including cranial polyneuropathy (inflammatory, infectious, or autoimmune), motor end plate disorder, and mitochondrial disorder.

In the clinical case presented, despite not having identified recent stressful events, the inconsistencies and incongruities observed, the recognition of details in the neurological examination, including spasm of the orbicularis muscle, the existence of folds in the upper and lower eyelids, the position of the eyebrows (ptosis and elevation of the left eyebrow), and hypoesthesia of the right hemiface with no anatomical relation to innervation of the trigeminal nerve, as well as the involvement of different specialties, made it possible to establish a diagnosis.

There are some diagnostic clues that make it possible to differentiate functional ptosis from other causes of ptosis, including: (1) the one-sidedness of symptoms, as the bilaterality of the symptoms in the clinical case presented prolonged the etiological investigation and made the final diagnosis difficult; (2) the exposure to stress factors or adverse life events that, though not necessary for a diagnosis, constitute an important clue[11]; (3) the presence of details in the neurological examination, including (a) the position of the eyebrows: on the ptosis side, there may be ptosis of the eyebrow[10], while on the contralateral side, the eyebrow is higher, which is due to the spasm of the frontal muscle to compensate for the decreased activity of the levator palpebrae muscle on the same side; (b) the existence of folds in the upper and lower eyelids due to the spasm of the orbicularis oculi muscle[10]; (c) inverse lower eyelid ptosis[9]; (d) no attempt by the patient to open the eye using the fingers or tilt the chin[8].

FNDs are a very heterogeneous group of pathologies, requiring careful assessment and holistic discussion, involving collaboration between several specialties[3]. An error or delay in establishing a diagnosis can bring harm to the patient, namely by subjecting him/her to unnecessary diagnostic and therapeutic methods, affecting the prognosis. Reliable knowledge about the expected manifestations of a given clinical syndrome, associated with the recognition of clinical patterns, will pave the way for a positive approach to diagnosing FNDs.

Authors' contribution

Sofia Vaz Pinto: Conception and design of the study, report, review or other type of work or paper; Acquisition of data either from patients, research studies, or literature; Analysis or interpretation of data either from patients, research studies, or literature; Drafting the article; Critical review of the article for important intellectual content; Final approval of the version to be published; Agreement to be accountable for the accuracy or integrity of the work. Ana Isabel Gonçalves and Inês de Oliveira: Conception and design of the study, report, review or other type of work or paper; Acquisition of data either from patients, research studies, or literature; Final approval of the version to be published; agreement to be accountable for the accuracy or integrity of the work. Andreia Pereira: Critical review of the article for important intellectual content; Final approval of the version to be published; Agreement to be accountable for the accuracy or integrity of the work.

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Conflicts of interest

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Ethical disclosures

Protection of human and animal subjects. The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Confidentiality of data. The authors declare that they have followed the protocols of their work center on the publication of patient data.

Right to privacy and informed consent. The authors have obtained the written informed consent of the patients or subjects mentioned in the article. The corresponding author is in possession of this document.

Use of artificial intelligence for generating text. The authors declare that they have not used any type of generative artificial intelligence for the writing of this manuscript, nor for the creation of images, graphics, tables, or their corresponding captions.

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