Miogenic ptosis in oculopharyngeal muscular dystrophy

Ptose miogênica na distrofia muscular oculofarinênea

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ABSTRACT

The authors report a case of oculopharyngeal muscular dystrophy, an autosomal dominant genetic disease, which leads to miogenic ptosis. This patient presented bilateral palpebral ptosis and dysphagia and underwent ptosis surgical treatment, with a good functional and aesthetic result.

Keywords: Blepharoptosis/surgery; Muscular dystrophy, oculopharyngeal; Dysphagia; Oculomotor muscles/physiopathology; Case reports

RESUMO

Relato de caso de distrofia muscular oculofaríngea, doença genética de herança autossômica dominante e uma das causas de ptose miogênica adquirida. A paciente apresentou quadro de ptose palpebral bilateral e disfagia, achados clínicos típicos da doença, foi submetida a tratamento cirúrgico da ptose, com bom resultado estético e funcional.

Descritores: Blefaroptose/cirurgia; Distrofia muscular oculofaríngea; Disfagia; Músculos oculomotores/fisiopatologia; Relato de casos

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INTRODUCTION

Ptosis is an anatomical disorder characterized by the positioning of the upper eyelid (UE) below its normal position, usually located about 2 mm below the upper limb in the primary eye position(1).

Eyelid ptosis can be classified as congenital or acquired, and this differentiation is important because it determines the surgical technique to be used in the correction(2).

Acquired ptosis is classified according to its pathophysiology in aponeurotic, neurogenic, myogenic, traumatic and mechanical. Myogenic ptosis is relatively rare and manifests as a severe eyelid ptosis with reduced or absent function of the levator of the UE, decreased extrinsic ocular motility and strength of the muscles of the face, including the eyes orbicular muscle and the shoulder girdle in 20% of cases(1).

The causes of acquired myogenic ptosis include: (1) mitochondrial myopathy (progressive chronic external ophthalmoplegia, Kearns-Sayre syndrome, and myopathy associated to encephalopathy); (2) oculopharyngeal muscular dystrophy; (3) oculopharyngeal distal myopathy and (4) myotonic dystrophy(3).

Patients with myogenic ptosis have a vicious head position, with elevation of the mento associated to a frontal muscle spasm in an attempt to free the visual axis obstructed by the severe eyelid ptosis, which may progress to chronic muscle fatigue(1).

Oculopharyngeal muscular dystrophy is a progressive disease of dominant autosomal inheritance characterized by bilateral blepharoptosis, dysphagia, involvement of extraocular muscles and proximal muscle weakness(4).

The genetic alteration that causes the disease was described by Brais in 1995. The disease-related gene is found in the DNA of all body cells located in chromosome 14q. Its normal sequence is comprised of ten basic elements encoding molecules to form alanine, which in turn is part of the composition of PABPN1 protein (poly (A) binding protein, nuclear 1)(5).

The first symptoms usually appear between 45 and 55 years of age(5). Dysphagia is initially noted with solid food, but can evolve with liquid swallowing difficulty(4).

Oculopharyngeal Muscular Dystrophy (OPMD), a disease of dominant autosomal inheritance, is characterized by dysphagia, paresis of the extraocular muscles, proximal paresis of the extremities and progressive bilateral ptosis, forcing the patient to use the front muscles and retroflex the head to compensate the loss of field of vision(3).

The diagnosis is based on clinical presentation, and can be confirmed by genetic testing. Unfortunately, there is no cure for oculopharyngeal muscular dystrophy. Current therapies are aimed at improving the symptoms that result from ptosis and dysphagia(4).

DISCUSSION

Oculopharyngeal Muscular Dystrophy (OPMD), a disease of dominant autosomal inheritance, is characterized by dysphagia, paresis of the extraocular muscles, proximal paresis of the extremities and progressive bilateral ptosis, forcing the patient to use the front muscles and retroflex the head to compensate the loss of field of vision(3).

Most patients with OPMD are those with French-Canadian ancestry who reside in Quebec.(6) Other cases of this disease are among the Bukhara Jews immigrants in Israel.(7)

OPMD usually begins insidiously during the fifth or sixth decade of life. These patients develop progressively with myogenic bilateral ptosis. Dysphagia is a prominent characteristic of OPMD, and is initially perceived with the ingestion of solid food, which can later develop into liquid swallowing difficulties.(8)
Besides ptosis and dysphagia, patients may also have proximal weakness of the limbs in varying degrees of severity. Often there is also an element of extraocular muscle weakness (usually affecting the supraduction), but complete external ophthalmoplegia is rare.

The diagnosis of OPMD is based on the history and clinical presentation and can be confirmed by genetic testing. Before the feasibility of such tests, the diagnosis was confirmed by muscle biopsy, which shows vacuoles in the muscle fibers, small and angled fibers, as well as intranuclear inclusion corpuscles.(9)

Aspiration pneumonia and malnutrition are the main causes of death in patients with OPMD, but it does not decrease the life expectancy because they tend to occur in the later stages of the disease.(5)

Regarding the treatment, there is still no cure for OPMD. Current treatment methods are based on improving the signs and symptoms related to eyelid ptosis and dysphagia. The surgical techniques for correction of ptosis in these patients are difficult to be performed. Some authors prefer to advance the levator muscle of the upper eyelid or combine this technique with Muller’s muscle advance in cases of MEPS function preserved.(10)

Another technique also widely used is the primary suspension of the frontal muscle due to the progressive nature of the disease. Fortunately, many patients with OPMD maintain good function of the orbicularis muscle and a good Bell’s phenomenon, which greatly reduces the risk of corneal commitment postoperatively.(9)

CONCLUSION

There is no treatment to cure oculopharyngeal dystrophy. Genetic studies are important not only in the diagnosis but also in the identification of asymptomatic patients and genetic counseling. It is for the health professional to offer biopsychosocial multidisciplinary support in order to minimize the symptoms, such as surgical correction of ptosis, cricopharyngeal myotomy or gastrostomy to relieve dysphagia, as well as physiotherapy and psychological support, promoting better quality of life.

REFERENCES