

A Case Report and Review of Literature: The Possibility of Marcus Gunn Jaw-winking and Monocular Elevation Deficiency Being the Same Disease



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ABSTRACT

Introduction: Marcus Gunn Jaw-Winking (MGJW) synkinesis is characterized by drooping eyelid and retraction with jaw movement. MGJW is usually seen alone, although familial cases have been documented. One of the problems that may occur with MGJW is Monocular Elevation Deficiency (MED). The concomitant MGJW, congenital ptosis, and MED represent a congenital misdirection syndrome that involves oculomotor nerve.

Case Presentation: In this report, we present a 13-year-old girl with MGJW with severe ptosis and concomitant MED in her left eye. A brief review was performed about this case too.

Conclusions: MED and MGJW maybe two extremes of one disease, because there is of a high probability of the same inheritance in the two conditions (both are due to a gene defect). Also, in our case with more severe ptosis, MED appeared, while in her aunt with less ptosis and milder MGJW, MED was absent.

1. Introduction

Marcus Gunn Jaw-Winking (MGJW) refers to the synkinetic movement of the eyelid associated with jaw movement [1]. MGJW synkinesis is one of the spectrum of congenital cranial aberrant connection between branches of cranial nerves [2, 3]. In 1883, Robert Marcus Gunn introduced the disease as unilateral congenital ptosis that was re-

tractable during bilateral two-sided pterygoid muscle movements [4]. MGJW is commonly seen as sporadic cases, although familial cases have been documented [5]. It is almost always unilateral, but in severe abnormality ipsilateral superior rectus underaction is happened simultaneously [6]. MGJW is found in approximately 2-13% of newborns with congenital ptosis [7].

Monocular Elevation Deficiency (MED) is defined as a unilateral congenital defect in the eye elevation both in

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the abduction and in adduction. Clinical diagnostic tests of both the restrictive and the paretic causes have indicated that a tight inferior rectus muscle and superior rectus palsy may coincide. Additionally, the clinical picture of MED may be a symptom of a supranuclear problem. The concomitant MGJW, congenital ptosis, and MED represent a congenital misdirection syndrome that involves oculomotor nerve [8]. Treatment may include surgery in severe cases, but it usually depends upon the amount of ptosis and degree of jaw winking. There can be a high frequency of strabismus and amblyopia, so ophthalmologic evaluation is mandatory to prevent vision loss and to rule out related abnormalities. On the whole, it is a challenging task [9].

2. Case Presentation

A 13-year-old girl with drooping of the left upper eyelid since birth without amblyopia referred to the Eye Clinic of Bu-Ali Sina Hospital, Sari, Iran. The girl was born at 38 weeks of normal pregnancy from healthy parents. The neurological examination did not reveal any pathologies. Her childhood development was normal; she had no past medical history. Her eyes examination revealed the following results: both eyes visual acuity, 10/10; her cycloplegic right refraction, +1.5 and left eye, +1.25; her right eye ptosis, zero and left eye ptosis, six mm. Also, her levator function was in the range of 16mm in the right eye and five mm in the left eye.

The marginal reflex distance -1 of her right eye was +4mm and that of the left eye -2mm. When her jaw

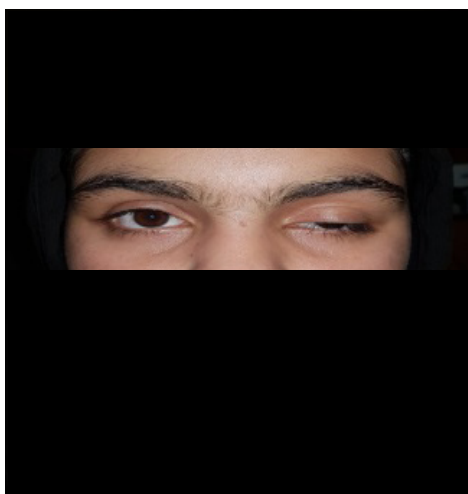
moved to the right (opposite) side, the eyelid opened up to eight mm, and when the jaw moved to the left, the eyelid did not change. In her left eye, she had 30 prism diopter hypotropic in primary position, 40 prism diopter in abduction, and 27 prism diopter in adduction. Bell's phenomenon existed (about four mm). Forced duction test in the right and left eye was negative. Extraocular muscle movements were abnormal in the left eye (two mm limitation in upward gaze) and normal in the right eye. The evaluation of the anterior and posterior segments of both eyes was normal. The systemic evaluation did not show any abnormal signs (Figure 1).

It should be noted that MGJW syndrome in our patient was severe and was accompanied by MED. In our case, ptosis was present and associated with MED that needed surgical intervention. However, her aunt, a 33-year-old female with MGJW syndrome (with a milder degree than her niece' disease), did not have MED. The patient and her family were consulted for surgical intervention as the child was at high risk of developing poor psychosocial outcomes as observed in some other cases. Informed consent was obtained from the patient for presentation of this case report. We also performed a brief review in PubMed and Google scholar databases about the presented case which would be discussed here.

3. Discussion and Literature Review

MGJW is a nerve dysfunction syndrome that involves simultaneous retraction of the upper eyelid with the pterygoid muscles movement. It is accompanied by

a



b

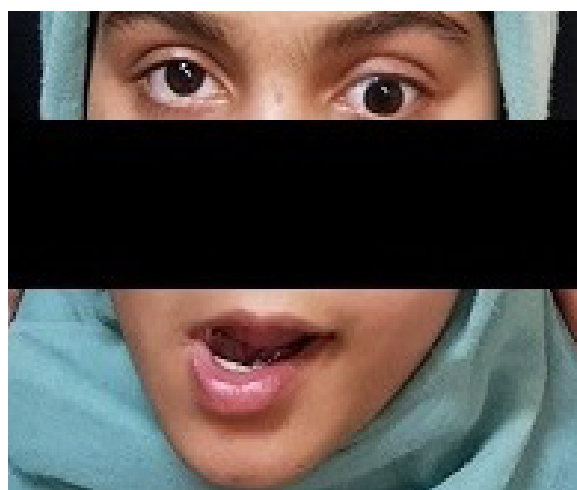


Figure 1. The photograph of the patient

A. Left-sided Marcus Gunn phenomenon; B. Retraction of the left upper eyelid during the pterygoid muscles right-side movement and mouth opening

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varying degrees of ptosis of the upper eyelid [10]. The incidence of MGJW in males and females and both eyes are equal. MGJW is often unilateral. It occurs alone, but in a few cases, it was comorbid with systemic or ocular abnormalities such as upper rectus muscle paralysis and Duane's syndrome, pseudo inferior oblique overaction, and iris heterochromia [6]. The differential diagnosis of the disease includes chronic progressive external ophthalmoplegia, congenital fibrosis syndrome, and myasthenia gravis.

There are a few cases in the literature on MGJW synkinesis and MED. Saemeh Nuzhat Zafar et al. performed a study on 22 patients with MED, nine (40.9%) of them had MGJW. MGJW with variable severity in 56% of patients with MED and ptosis was seen that is higher than reported in other studies. Two patients (9.99%) cases of MED patient were without ptosis. Considering the high association between the MGJW and MED, common pathogenesis with variable presentations can be considered for these two conditions [8]. In this study, unlike ours, the probability of genetic inheritance of MED with the variable range of ptosis and MGJW synkinesis was not investigated. Pearce et al. reported a six-year-old girl who had MGJW without ptosis in the left eye since her birth. A positive family history of paternal family with MGJW was found without ptosis in her cousin [10]. In our study, the patient had a positive familial history, although both of our patients with same genetic studied in this report had MGJW and ptosis, the severity of MGJW was different, which indicates a different penetrance in the disease.

Ilaria Biagini et al. reported that more than half (59%) of the patients with MGJW and ptosis had amblyopia. They also reported that one-half of the patients had strabismus with isolated superior rectus palsy in one quarter of the patients and double elevator palsy in another quarter of the patients. Other ocular associations have been reported in rare cases such as synergistic divergence and Duane retraction syndrome [11].

However, our patient's visual acuity in both eyes was 10/10, even though the visual axis was closed that was probably prevented by the compensatory mandibular maneuver to counteract ptosis. Although ptosis was severe, the absence of amblyopia suggests that ptosis is less effective in the incidence of amblyopia and strabismus, and compensatory maneuver to control ptosis is a determining factor in the incidence of amblyopia. To confirm this theory, studies should be conducted on more cases.

Akash et al. reported an 18-year-old man who had asymmetric ptosis and MGJW in both eyes since birth, and MED in his right eye. In the examination, the magnitude of the right eye ptosis was four mm and left eye one mm. Also, the levator function test of his right eye was one mm and of his left eye 14mm. Their study suggests the relationship between the severity of MGJW and ptosis with the incidence of MED [6].

This study demonstrates the association of MED in the eye with severe ptosis. Also, by comparing the levator function test in two eyes, it was found that the eye with severe ptosis had less innervated levator and superior rectus complex. rectus complex that resulted in MED and severe MGJW.

Based on these studies, we conclude that the levator and superior rectus complex have a common innervation from the upper branch of the third cranial nerve. If there is a defect in the innervation of this complex, the superior rectus muscle dominates over the other muscle. Therefore, if the innervation is defective, the levator and superior rectus muscles weakness will coincide with the incidence of MED, which is associated with a more severe eyelid droop.

Melissa Brigham de Figueiredo et al. reported a newborn girl with left eyelid ptosis and synchronous movement of the abduction of the eyelid during suckling or crying. Her cranial ultrasound imaging revealed a left side lenticulostriate vasculopathy and bilateral grade one periventricular/ intraventricular hemorrhage. The infant was diagnosed with Marcus Gunn syndrome [12].

Karolina Dżaman et al. reported an olfactory nerve disturbance in association with MGJW synkinesis. They postulated that this pathological process in the central nervous system might be associated with the developmental disorder of other nerves [13].

Different theories have been presented about the etiology of this disorder. The first theory states that the innervation of the levator palpebrae superioris muscle from both oculomotor nucleus and the external pterygoid portion of the trigeminal nucleus is being disturbed. In the normal state, this muscle is only innervated from the oculomotor nucleus. The second theory of nerves describes a reflex arc from the trigeminal nervous system to the Gasserian ganglion. It extends between the neuronal connections to the nucleus of the oculomotor and ultimately reaches the upper eyelid muscle. This theory is based on the embryonic documentation that

the nuclei of two cranial nerves have been in a closely spaced area at one time [7].

In the lack of innervation, the distal end of the third nerve prefers superior rectus over levator palpebral muscle. If the innervation is more, the upper rectus muscle is fully innervated, and if innervation is less, both superior rectus and levator palpebral muscles get involved, and MGJW and MED present simultaneously. Due to the lack of innervation to levator and superior rectus muscle complex, aberrant innervation induced MGJW. Presumably, this disease belongs to the family of congenital cranial dysinnervation disorders. Considering the same inheritance that most likely exist between the patient and her aunt, MED and MGJW appear to be two extremes of one disease because MED appeared in the child with severe ptosis, while MGJW with milder ptosis appeared in her aunt.

The management generally depends upon the amount of ptosis and the degree of jaw winking. It is a challenging task. Therefore, a comprehensive medical and ophthalmologic evaluation and a detailed history taking are mandatory before undertaking the successful treatment of patients with this syndrome. It has been shown that great success can be achieved when practicing a suitable treatment after comprehensive medical evaluation and doing standard tests on patients with MGJW [14].

In the end, we should mention that there was no study about the severity of the ptosis and its relation to the severity of MGJW and the incidence and severity of MED. These issues should be investigated in subsequent studies with more cases.

4. Conclusion

MGJW and MED can be two extremes of one disease spectrum with variable severity. In the family of our case, the inheritance of each extreme of this disease has been presented. However, further studies are needed with more cases and the particular emphasis on the relationship between MED and severity of MGJW as well as the degree of ptosis and the incidence of MED. In treating these patients, ophthalmologists should make an early diagnosis to deliver a proper, timely, and individually-based treatment according to the severity and coincident diseases.

Ethical Considerations

Compliance with ethical guidelines

All ethical principles were considered in this article, and the participant was informed about the purpose of the research.

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Authors contribution's

Study concept and design, critical revision of the manuscript for important intellectual content, and drafting of the manuscript: Reza Jafari; Review of the literature and writing the manuscript: Sogand Shahabinia; Review of the literature: Somayeh Rostami Maskopaii.

Conflicts of interest

The authors declared no conflict of interests.

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