The rare phenomenon of Marcus-Gunn jaw winking without ptosis: Report of 14 cases and review of the literature

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Purpose: To report a rare case series of 14 patients of the Marcus-Gunn jaw-winking phenomenon (MGJWP) without ptosis. Methods: This was a retrospective noninterventional case series. The medical records of all patients diagnosed with MGJWP over the past 10 years were retrieved. Patients with documented evidence of absence of ptosis were segregated and analyzed for visual acuity, the severity of Marcus-Gunn, the presence of squint and amblyopia, and the presence of other aberrant regenerations. Results: A total of 207 patients were diagnosed with MGJWP, out of which 14 (6.76%) patients had isolated MGJWP without blepharoptosis. The mean age of presentation was 9.5 years and males and females were equally affected. The left eye was involved more commonly (57.2%) than the right eye. Twelve patients were congenital and two were presumed to be of traumatic origin. The most common refractive error in this cohort was astigmatism (10, 71.42%), followed by hyperopia (3, 35.71%). One patient had anisometropic amblyopia. Marcus-Gunn was found to be mild (≤2 mm of lid excursion) in all cases. None of the patients had strabismus or any other aberrant innervations. None of the patients underwent surgery and did not develop ptosis or worsening or improvement of Marcus-Gunn after a mean follow-up period of 2.3 years. Conclusion: Isolated MGJWP in the absence of ptosis is a very rare entity and this is the largest series to date to report such an occurrence. All patients had a mild form of MGJWP with no intervention required in any of the cases.

Key words: Aberrant innervation, Marcus-Gunn jaw-winking phenomenon, Primitive reflex, without ptosis

Despite many theories regarding the neurological basis of this entity, the underlying etiology is yet not clearly understood.

Since 1888, there has been abounding literature on this entity; however, MGJWP has always been mentioned in association with blepharoptosis. A thorough review of the literature revealed only five cases of MGJWP in absence of ptosis.[1,4]

In our study, we discuss the unique clinical presentation of a case series of 14 patients who were diagnosed with Marcus-Gunn jaw winking in the complete absence of ptosis. The present study is by far the largest case series to date on such a rare occurrence.

Methods

This was a retrospective noninterventional case series. Electronic medical record search was done for all cases of Marcus-Gunn jaw-winking phenomena over a period of 10 years from January 2005 to January 2015. MGJWP without ptosis was defined as the presence of MGJWP with a normal orbit, oculoplasty, reconstructive and aesthetic services, Aditya Birla Sankara Nethralaya, (A Unit of Medical Research Foundation, Chennai) Kolkata, West Bengal, ‘M.N. Eye Hospital, ‘Department of Pediatric Ophthalmology and Strabismus, Sankara Nethralaya, Medical Research Foundation, ‘Orbit, Oculoplasty, Reconstructive and Aesthetic Services, Sankara Nethralaya, Medical Research Foundation, Chennai, Tamil Nadu, India.

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marginal reflex distance-1 (MRD-1) without any evidence of ptosis in the involved eye. All patients were subjected to a minimum of two evaluations, by a senior oculoplastic consultant, with measurement of ptosis performed both with and without jaw movement.

Demographic data analyzed included age at presentation, sex, laterality, and family history. Clinical data analyzed included visual acuity, the severity of Marcus-Gunn, presence of squint and amblyopia, presence of other aberrant regenerations, LPS action, and extraocular motility. Consent for identifiable photographs was taken from the patient or guardian. The institutional review board approved of the study and it adhered to the tenets of the Declaration of Helsinki. Data analysis was done using the Chi-square test and the student’s T-test (SPSS version 14).

Results

A total of 207 patients were diagnosed with MGJWP, out of which 14 (6.76%) patients had isolated MGJWP without blepharoptosis. There were 7 males and 7 females. The mean age at presentation was 9.5 years (range 18 days to 40 years). The median age was 5.75 years. All patients presented with unilateral MGJWP. The right eye was involved in 6 patients (42.8%) and left eye in 8 (57.2%) patients. None of the patients had see-saw MGJWP [Table 1].

All patients had observed the onset of MGJWP by a parent since birth, except in a 6.5-year-old boy, whose parents became aware of it after the incidental blunt injury to the left eye with a cricket ball. Another 18-day-old child had a suspected history of birth trauma, with an incised wound on scalp post caesarian surgery. The family history was positive in a 1-year-old boy. All except two patients had been delivered at full term by normal vaginal delivery. Two patients were delivered by cesarean.

Visual acuity was measured by Snellen’s distance acuity chart in 10 patients, out of which 9 had visual acuity of 20/20. A 22-year-old male had a visual acuity of 20/60 in the involved eye due to anisometropic amblyopia. Four patients were preverbal, and their vision was assessed by the fixation pattern, which was central, steady, and maintained. The most common refractive error in this cohort was astigmatism (10, 71.42%), followed by hyperopia (5, 35.71%).

One 11-year-old child had an associated congenital nasolacrimal duct obstruction of the same side as the MGJWP.

The MGJWP was mild in all cases (less than or equal to 2 mm of lid excursion) [Figs. 1 and 2]. None of the patients had associated squint or extraocular motility restriction. Levator excursion could be measured in nine patients and was excellent. All patients were explained the management option in the form of LPS excision with tarsofrontalis sling, along with the possible adverse effects and complications. None of the patients opted for surgery.

The mean follow-up was 2.3 years (range 1–8 years). During this period, none of the patients went on to develop ptosis and the MGJWP remained stable.

Discussion

MGJWP is usually associated with a variable amount of ptosis that manifests at the same time as the synkinesis itself. In our case series, all 14 patients had MGJWP in the complete absence of ptosis [Figs. 1 and 2]. This is the second case series, and the largest to date to report such an occurrence. [9] One case of MGJWP without ptosis was reported by Pratt et al. in their review of 71 cases and this was the only case reported over the past 150 years, till the case series of 4 patients reported by Pearce et al. recently. [14] Pearce et al. reported that 6% of the patients with MGJWP had no ptosis; we noticed a similar rate (6.76%) in our cohort.

All the patients in our series showed unilateral presentation. MGJWP usually presents unilaterally, but rarely can be seen bilaterally.[9] All 14 patients demonstrated typical upper eyelid elevation on chewing or sucking. Although lid elevation is commonly associated with chewing or sucking movements, it has been reported with other movements such as lateral mandibular movement, yawning, smiling, sternocleidomastoid contraction, tongue protrusion, Valsalva maneuver, or even during inspiration.[10-12] We did not analyze different maneuvers triggering lid elevation, and since there were no remarks regarding any rare maneuver triggering it we presume that most of them were associated with chewing, sucking and jaw movement. There were no dental anomalies associated with our subset of patients; however, there have been a few case reports that describe malocclusion, proclined upper anterior teeth, severe lower crowding and mandibular micrognathia associated with MGJWP.[13,14]

All patients in our study had MGJWP of congenital origin except two, one of whom observed it after trauma with cricket ball injury while another 18-day-old neonate with a suspected history of birth trauma during delivery. In the above two cases, it is unclear if trauma was the definitive causative factor. None of the cases reported by Pearce et al. had a history of trauma.

There was no gender preponderance in our series of patients, which concurs with previous studies.[15] The left eye was more commonly affected (57.2%) in our study. Doucet et al. also reported a slight left eye preponderance in their series of

### Table 1: Baseline characteristics of the study population

| Parameters                  | Number and percentages |
|-----------------------------|------------------------|
| Total number of MGJWP patients | 207                   |
| Incidence of MGJWP without ptosis | 14/207 (6.76%)        |
| Age                         | Mean: 9.5 years (range: 18 days-40 years) |
| Male: Female                | 1: 1                   |
| Laterality                  | 100% unilateral         |
| Right eye: Left eye         | 6: 8 (42.8%: 57.2%)     |
| Severity of MGJWP           | Mild in all cases       |
| Mean visual acuity in LogMar| 0.05(Measurable 10 eyes) |
| Follow-up period            | Mean: 2.3 years (range: 0-8 years) |
| Refractive error            | Astigmatism, 71.4% Hyperopia, 35.7% |
| Associated features         | Associated trauma in 2 Familial tendency in 1 Anisometropic amblyopia in 1 Associated nasolacrimal duct obstruction in 1 |

MGJWP: Marcus-Gunn jaw-winking phenomenon
MGJWP patients. This has been attributed to fibrin emboli preferentially traveling to left carotid vessels in-utero because of the asymmetry of thoracic vasculature. However, Demerci et al. found no statistically significant difference between either eye involvement.

All the patients of congenital MGJWP in our case series were born at full term. No significant history of prenatal insult was documented. Two patients in our case series had been delivered by cesarean delivery, while the other 12 were born of a normal vaginal delivery. One of the children born of cesarean delivery had a presumed birth trauma that might have been associated with the development of MGJWP in that eye.

MGJWP is usually considered a sporadic condition, although familial cases have been documented. Of our 12 congenital MGJWP patients, there was one patient (8.3%) with familial isolated MGJWP, in whom a hereditary pattern could not be identified in view of only one other affected family member (paternal grandfather). In a clinical and electrophysiological study of two patients of familial MGJWP, the hereditary pattern was found to be an incomplete autosomal dominant trait with variable expressivity.

Visual acuity was comparable in both the eyes in all except one of our patients, who had amblyopia in the affected eye due to anisometropia. Good vision can be explained by the fact that none of the patients had ptosis or strabismus which would otherwise have caused amblyopia. Similar findings were noted by Pearce et al., however reported visual acuity better than 20/40 only in 83% of his patients and 23% of the patients had stimulus deprivation amblyopia, as they were associated with varying grades of ptosis.

No other ocular aberrant innervation syndromes were associated with our patients of isolated MGJWP. However, MGJWP with ptosis has been reported to be associated with DRS in Mobius sequence, pseudo- inferior oblique over action (Y pattern exotropia due to lateral rectus activation on upgaze), trigemino-abducens synkinesis and gustatory sweating in poorly controlled diabetes. In our case series, we didn’t submit the patients to a pediatric neurologist’s opinion. However, it would be a good practice to rule out other systemic manifestations.

Despite numerous case reports and a number of case series on MGJWP, the exact mechanism of this fascinating phenomenon is yet to be clearly understood. There are two schools of thought. The first one believes in the “release hypothesis,” according to which aberrant connections between appositionally adjacent trigeminal mesencephalic nucleus and the oculomotor nucleus are a part of primitive reflex which got extinguished during phylogenetic development. This primitive reflex gets released and becomes active because of intrauterine trauma or some unknown causes, resulting in synkinesis on gaze deviation.

The second hypothesis is that MGJWP is due to some structural abnormality in the brainstem which causes neural misdirection of trigeminal motor axons to LPS. Other theories are pre-existing aberrant connections and ephaptic transmission between the mandibular division of trigeminal nerve innervating the pterygoids and the superior division of oculomotor nerve innervating LPS.

In our patients, the absence of ptosis further supports the central hypothesis, wherein a pre-existing primitive reflex could have been released or disinhibited during the intra-uterine period due to some unknown stimulus.

The mean follow-up period in our patients was for about 2 years, with the longest being 8 years. None of the patients demonstrated a change in the MGJWP or development of new-onset ptosis. None of the patients underwent any intervention and were asked to be under regular follow-up.

Retrospective design, no documentation of the type of jaw movement eliciting Marcus-Gunn, absence of EMG based documentation and short follow up period for few of the patients are some of the limitations of the present study.
Conclusion
To conclude, isolated MGJWP in the absence of ptosis is a very rare entity and this is the largest series to date to report such an occurrence. In this case series, we found that our subset of patients had mild MGJWP and hence required no intervention. Further studies on this subset of patients with neurophysiological and neuroradiological evidence, would shed more light on the pathogenesis of this otherwise elusive phenomenon.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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