FREQUENCY AND ASSOCIATIONS OF MARCUS GUNN PHENOMENON IN CONGENITAL PTOSIS

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ABSTRACT

Objective: To determine the frequency and various associations of Marcus Gunn Phenomenon in patients of congenital ptosis.
Study Design: Cross-sectional observational study.
Place and Duration of Study: Department of Ophthalmology, Khyber Teaching Hospital, Peshawar, from Jun to Sep 2020.
Methodology: A total of 100 patients with congenital ptosis were included. Patients with any cause of secondary or acquired ptosis were excluded. Congenital ptosis was classified as simple and complex. Assessment for ptosis severity, presence of jaw winking ptosis, jaw winking severity, refractive error, amblyopia, strabismus, systemic association and family history for congenital ptosis was performed.
Results: Simple congenital ptosis was the most common type overall (84%). A total of 60% patients were males and 40% were females. More than 90% of the patients had severe ptosis. Ninety eight percent cases had onset since birth. No patient had systemic association or positive family history. Nine percent frequency of Marcus Gunn Phenomenon was noted with congenital ptosis. All the patients with Marcus Gunn Phenomenon had unilateral presentation. Left eye was affected predominantly (88.89%). A total of 22.22% of the patients with Marcus Gunn Phenomenon had anisometropic amblyopia. No other ocular or systemic association of Marcus Gunn Phenomenon was observed.
Conclusion: Simple congenital ptosis is the most common type of congenital ptosis. The authors report 9% frequency of Marcus Gunn Phenomenon with congenital ptosis and left side effected predominantly. Anisometropic amblyopia was the major ocular association of Marcus Gunn Phenomenon observed.

Keywords: Association, Blinking, Blepharoptosis, Congenital, Jaw, Marcus gunn phenomenon, Reflex, Synkinesis.

INTRODUCTION

Blepharoptosis or ptosis is the drooping of eyelid margin below its anatomical location with the eyeball in its primary position. Normally in adults, the upper eyelid is highest nasally and covers the cornea almost 1.5-2 mm below superior limbus[1,2]. According to time of onset, ptosis can be either congenital or acquired and according to etiology, acquired ptosis can be traumatic, neurogenic, myogenic, mechanical, and aponeurotic[3].

Congenital ptosis is drooping of eyelid below its anatomical position either at birth or within first year of life[4]. Congenital ptosis occurs due to dysgenesis of levator palpebrae superioris (LPS) muscle where normal muscle fibers are replaced by adipose and fibrous tissue resulting in decreased ability of LPS to contract and relax[5]. It can also occur due to interrupted innervation to LPS muscle thus representing a form of congenital cranial dysinnervation disorder[6]. Incidence of congenital ptosis was reported as 7.9 per 1 lac patients below 19 years of age[7].

Marcus Gunn Phenomenon (MGP) or Jaw winking ptosisis a synkinesis where ptotic upper eyelid moves with the movement of jaw. This reflex consists of temporary upper eyelid elevation or retraction to the same or higher level as that of normal fellow eyelid, when there is stimulation of pterygoid muscles of same side. These patients have quite variable degrees of ptosis in resting primary position. It is usually unilateral but can also present as bilateral in rare cases[8]. Pathophysiology of MGP suggests an abnormal connection between motor division of trigeminal nerve that supply external or internal pterygoid muscle and the superior branch of oculomotor nerve which supply the ipsilateral LPS muscle of upper lid[9]. Clinically, jaw winking ptosis can be associated with amblyopia, refractive errors, double elevator palsy, and superior rectus palsy. Jaw winking can be either mild (<2mm), moderate (2-5 mm), or severe (>5mm)[9]. There are various approaches for its management but overall surgical outcome is less favorable in congenital ptosis patients who have MGP rather than for those who don't have jaw-winking[10].

In literature, data is lacking about the prevalence of MGP in patients with congenital ptosis. In addition whether it is more prevalent in patients who have congenital ptosis at birth or who develop later on within first year of life. Thus, the rationale of this study was

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made to determine the frequency of MGP in congenital ptosis patients and to determine its association with amblyopia, refractive error, and strabismus.

METHODOLOGY

After getting permission from ethics review committee (908/ADR/KMC), Khyber Medical College, Peshawar, a cross-sectional observational study was conducted at department of ophthalmology, Khyber Teaching Hospital, Peshawar, Pakistan. Sample size of 96 was obtained with 95% confidence level, anticipated population proportion of 50%, and with absolute precision of 10% using WHO prevalence formula[7]. To make round figure 100 patients were included in this study. Consecutive sampling was used as a sampling technique. All the consenting patients, of both genders diagnosed as idiopathic congenital ptosis were included in the study. Patients with any cause of secondary or acquired ptosis like traumatic, third nerve palsy, Myasthenia gravis, Horner syndrome, chronic progressive external ophthalmoplegia, Duane syndrome and blephrophimosis syndrome were excluded from this study. Informed consent was taken from every patient/ guardian.

Patients were explained about the purpose and the benefits of the study and a specially designed proforma was used as data collection tool. Congenital ptosis was classified as simple and complex on the basis of ocular and systemic association. Assessment for refractive error or associated amblyopia was performed for all the patients included. The prevalence MGP was determined along with its association with amblyopia, refractive error, and strabismus.

All the included patients underwent the routine ophthalmic examination. Complete ptosis and squint evaluation was carried out. Slit-lamp bi-microscopy and fundoscopy was performed as well to exclude other causes of reduced visual acuity. Data was transferred from the pro-forma to SPSS-20. Mean of age of all the presenting patients was computed. Frequencies were calculated to describe all the nominal variables. Chi-square test was applied to check association of Marcus Gunn Phenomenon with gender, laterality, amblyopia, strabismus and refractive error.

RESULTS

Mean age of included patients was 11.93 ± 7.737 years. All the patients had congenital ptosis. Gender distribution, age of onset (as noted by parents i.e. since birth or during first year of life) and laterality are shown in table-I.

Table-IV: Associations of marcus gunn jaw wink.

Jaw Winking Ptosis	Gender		Laterality			Amblyopia	Strabismus	Refractive Error
	Male	Female	R	L	B			
n=9	5 (55.55%)	4 (44.44%)	1 (11.11%)	8 (88.88%)	-	2 (22.22%)	-	2 (22.22%)
p-value	0.520		0.000			0.514	0.457	0.572

Eighty four patients had simple congenital ptosis i.e. with no ocular or systemic manifestation. Sixteen patients had complex congenital ptosis. On severity grading, 78 patients had ptosis >4mm, 14 patients had ptosis between 3-4mm, 7 patients had ptosis between 1-2mm. Manifestations of complex congenital ptosis and patients who had associated refractive error, amblyopia and positive family history for congenital ptosis are given in table-II.

Table-I: Demographic detail of patients with congenital ptosis.

	Mean
Age at presentation (years)	11.93 ± 7.737
Gender Distribution	**Frequency (%)**
Male	60 (60%)
Female	40 (40%)
Age of Onset	
Since Birth	98 (98%)
Since Childhood (1st Year)	2 (2%)
Laterality	
Right Side	41 (41%)
Left Side	36 (36%)
Bilateral	23 (23%)

Jaw winking ptosis was graded as mild (<2mm), moderate (2-5 mm), or severe (>5 mm) (table-III). Cross tabulation of Marcus Gunn phenomenon with gender, laterality, amblyopia, strabismus and refractive error is shown in table-IV.

DISCUSSION

Incidence of congenital ptosis is reported as 7.9 per 100,000 patients in a population based report of 40 years[7]. In a study done at Al-Shifa Eye Hospital, Rawalpindi, Pakistan frequency of congenital ptosis was reported (8.8%) among all the cases of congenital ophthalmic malformations. It was concluded that it is the 3rd most common congenital ophthalmic malformation[11]. Niazi observed (55.55%) occurrence in male and (44.44%) in females[11]. Similar trends were observed in

Table-II: Classification, severity and associations.

Associations of congenital ptosis	n (%)
Congenital Ptosis	
Simple	84 (84%)
Complex	16 (16%)
Ptosis Severity (n=100)	
Mild (1-2 mm)	7 (7%)
Moderate (3-4mm)	14 (14%)
Severe (>4mm)	78 (78%)
Refractive Error, Amblyopia or Family History Association (n=100)	
Refractive Error	26 (26%)
Amblyopia	28 (28%)
Positive Family History for congenital ptosis	2 (2%)
Complex Congenital Ptosis (n=16)	
Marcus Gunn phenomenon	9 (56.25%)
Associated Squint	7 (43.75%)
Systemic Association	-

Table-III: Jaw winking ptosis severity (n=9).

Severity of Jaw winking ptosis	n (%)
Mild (<2mm)	3 (33.33%)
Moderate (2-5 mm)	-
Severe (>5 mm)	6 (66.66%)

our study with 60% and 40% male and female distribution. Left sided congenital ptosis is reported more frequently in literature[7,9,12]. In our study, right sided congenital ptosis was more common. In our study, only the patients with congenital ptosis were included. Acquired ptosis was not included. This can be the reason for this difference in laterality. Left sided ptosis is reported to be associated with Duane retraction syndrome most of the time[7], which was excluded in our study. Twnty three percent patients in our study had bilateral congenital ptosis. Bilateral congenital ptosis is reported about 3-7% in different reports[4,7,12]. In our study bilateral cases are reported in far major number. This can be because of single center study setting or due to referral of patients with severe congenital malformation to tertiary care teaching hospital.

A total 98% patients in our study had presumed congenital onset of ptosis. Majority of the included patients (84%) had simple congenital ptosis. In a 24-year report of blepharoptosis in Korea, simple congenital ptosis was reported to be the most common type of congenital ptosis overall with a frequency of 73.7%[12]. Gregory *et al*, also reported simple congenital ptosis as the most common type with 76% of the patients with childhood ptosis labelled as simple congenital ptosis[7]. Similar trends were observed in our study. More than 90% patients in our study had moderate to severe ptosis. Severe ptosis is more common in case of congenital ptosis while in acquired ptosis mild-moderate ptosis severity is encountered more frequently[12].

In a meta-analysis by Yijie *et al*, the prevalence of amblyopia in congenital ptosis ranged from 18.5–27.8%[13]. In our study 28% patients had associated amblyopia with congenital ptosis. Twenty six percent patients in our study also had associated refractive error. Associated strabismus was found in 8% of the patients. Higher prevalence (19.6%) of strabismus is reported in recent meta-analysis[13]. No patient had any systemic association or family history for ptosis in our study.

The incidence of Marcus Gunn phenomenon (MGP) ranges between 2-13% in patients with congenital ptosis[9]. A 10 year review on childhood ptosis in UK has shown 5% prevalence of jaw winking ptosis[14]. In a 3 year review of congenital ptosis at Mayo Hospital, Lahore 11% patients had jaw winking ptosis[15]. In our study 9% prevalence of Marcus Gunn phenomenon was noted in patients of congenital ptosis.

No gender dominance was noted. Five patients (n=9) were males, while 4 (n=9) were females. Pearce *et al* in their second largest case series on MGP noted a trend toward female gender with MGP and congenital ptosis but this was reported to be statistically insignificant[16]. A total of 88.89% patients in our study had left sided jaw winking ptosis. Left sided predominance in jaw winking is reported in literature[9,16,17]. Qirat *et al* in a case series on jaw winking ptosis treatment, performed in Karachi, Pakistan, reported 72.7% occurrence on left side[18]. All the patients in our study had unilateral MGP. Bilateral MGP is also reported in literature but is rare overall[16,19].

Severity of jaw winking ptosis was graded on the basis of excursion of upper eyelid on mouth opening[9]. Six patients (n=9) had severe jaw winking while 3 patients (n=9) has mild jaw winking. Ocular associations with MGP observed in our study were amblyopia and refractive errors. In our study, 2 patients (n=9) had associated amblyopia with MGP. Both patients had anisometropic amblyopia. In literature, the most common ocular association of MGP reported is strabismus (50-60%) followed by amblyopia (30-60%)[20]. Other ocular associations of MGP include double elevator palsy (25%)[15,20] and superior rectus palsy (25%)[20]. These ocular associations were not observed in our study because of small sample size and study duration. Recent case reports also show association of MGP with morning glory optic disc anomaly[21]. It is also reported that MGP can even present without blepharoptosis[22].

Limitations of our study include single center, small duration of study and small sample size.

CONCLUSION

Simple congenital ptosis is the most common type of congenital ptosis. The authors report 9% prevalence of MGP with congenital ptosis and left side effected predominantly. Anisometropic amblyopia was the major ocular association of MGP observed.

CONFLICT OF INTEREST

This study has no conflict of interest to be declared by any author.

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