



Effect of Visual Acuity and Quality of Life in Different Grades of Congenital Ptosis

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ABSTRACT

Background: Congenital ptosis (blepharoptosis) is characterized by drooping of the upper eyelid due to abnormal development or dysfunction of the levator palpebrae superioris. It may affect the visual axis and visual function. **Objective:** To describe the clinical characteristics and visual acuity of patients with congenital ptosis and assess the association between palpebral fissure height and visual acuity. **Methodology:** A descriptive cross-sectional study was conducted among 40 patients with congenital ptosis at DHQ Vehari Eye Complex, Pakistan. We recruited participants aged 1-30 years using convenience sampling. We excluded patients with acquired ptosis, previous treatment or surgery, or ocular conditions affecting visual acuity or assessment. Clinical assessment included head posture, extraocular movements, palpebral fissure height, marginal reflex distance (MRD), levator palpebrae superioris function, and visual acuity. The association between palpebral fissure height and visual acuity was analyzed using Pearson's chi-square test. **Results:** Among 40 participants, 55.0% were male and 45.0% female; 62.5% were aged 1-10 years, 32.5% 11-20 years, and 5.0% 21-30 years. Normal head posture was present in 95.0%, while full extraocular movements were present in 96.0% of the right and 94.0% of the left eyes. Visual acuity of 6/9-6/12 was observed in 75.0% of the right and 57.5% of the left eyes. Poor levator function was found in 13.0% of the right and 24.0% of the left eyes. Palpebral fissure height and MRD showed variation between the eyes. No significant association was found between palpebral fissure height and visual acuity in the right ($p=0.309$) or left eye ($p=0.105$). **Conclusion:** Congenital ptosis showed variation in eyelid measurements and levator function, while visual acuity was relatively good in most participants. Palpebral fissure height was not significantly associated with visual acuity in either eye. Clinical assessment should therefore combine eyelid measurements and levator function with visual acuity and evaluation of refractive status, ocular alignment, and amblyopia.

Keywords: Congenital ptosis, Levator palpebrae superioris, Marginal reflex distance, Palpebral fissure height, Visual acuity, Visual function

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INTRODUCTION

Congenital ptosis, also termed congenital blepharoptosis, is characterized by an abnormally low position of the upper eyelid margin present at birth or developing during early infancy. It may occur unilaterally or bilaterally and may present as an isolated condition or in association with ocular, neurologic, or systemic disorders. Clinical presentation varies by laterality, severity, levator function, eyelid position, and associated ocular abnormalities.¹

In most cases of simple congenital ptosis, the underlying abnormality is developmental dysgenesis of the levator palpebrae superioris muscle, the principal elevator of the upper eyelid. Impaired levator development may result in reduced levator function, an abnormal eyelid crease, incomplete eyelid elevation, and characteristic lag of the affected eyelid during downgaze. Congenital ptosis may also occur in association with conditions such as Marcus Gunn jaw-winking syndrome, blepharophimosis syndrome, Horner syndrome, and congenital cranial nerve abnormalities.² These variations contribute to considerable heterogeneity in the clinical presentation of patients with congenital ptosis.

Population-based evidence demonstrates that congenital-onset ptosis constitutes the predominant form of childhood ptosis. In a 40-year population-based cohort, 89.7% of patients with ptosis had congenital-onset disease. Simple congenital ptosis accounted for 75% of all childhood cases and had an estimated birth prevalence of approximately 1 in 842 births.³ These findings demonstrate that congenital ptosis is an important clinical condition requiring appropriate assessment of eyelid and ocular characteristics.

The etiological heterogeneity of congenital ptosis is further supported by genetic evidence. Although many cases are sporadic, familial forms and syndromic presentations have been described, and several genetic mechanisms have been implicated in congenital ptosis and related developmental abnormalities.⁴ This genetic and phenotypic heterogeneity may contribute to differences in the clinical characteristics observed among affected patients and reinforces the importance of comprehensive clinical assessment. Beyond its

anatomical appearance, congenital ptosis may have important functional implications for vision. Drooping of the upper eyelid may partially or completely obstruct the visual axis and produce stimulus-deprivation effects, while associated refractive errors, anisometropia, and strabismus may independently compromise visual function. A systematic review and meta-analysis reported pooled prevalences of 22.7% for amblyopia, 22.2% for astigmatism, 19.6% for strabismus, and 17.3% for anisometropia among patients with congenital ptosis, demonstrating the substantial burden of associated visual abnormalities.⁵

Visual acuity is therefore an important component of the clinical assessment of patients with congenital ptosis. Reduced visual acuity may result from direct visual-axis obstruction or from associated refractive and binocular abnormalities. In patients with congenital ptosis, the risk of amblyopia has been associated with factors including ptosis severity and visual-axis involvement, emphasizing that visual function cannot be reliably inferred from eyelid position alone.⁶ Assessment of visual acuity alongside the clinical characteristics of ptosis is consequently important for identifying patients with potential visual impairment.

Clinical cohorts have also demonstrated a relationship between congenital ptosis and visual function. Amblyopia in patients with congenital ptosis has been associated with refractive errors, anisometropia, and strabismus, while some cases may be attributable directly to stimulus deprivation caused by severe ptosis.⁷ These findings indicate that visual acuity may vary according to the clinical characteristics of ptosis and associated ocular abnormalities. Accordingly, evaluation of visual acuity provides an important functional dimension when characterizing patients with congenital ptosis.

Refractive abnormalities represent another important consideration in the visual assessment of congenital ptosis. Patients with congenital ptosis have been reported to have higher frequencies of astigmatism, anisometropia, and other refractive errors, which can contribute to the development of amblyopia.⁸ In particular, patients with unilateral congenital ptosis may develop refractive asymmetry between the affected and fellow eyes, including hyperopic anisometropia in the ptotic eye, which may contribute to amblyogenic visual

abnormalities.⁹ Thus, differences in visual acuity among patients with congenital ptosis may reflect not only the extent of eyelid-related visual-axis obstruction but also associated refractive and binocular abnormalities.

The ptotic eyelid may also exert mechanical effects on the cornea. Corneal topographic studies have demonstrated alterations in corneal curvature and astigmatism in eyes affected by congenital ptosis, suggesting that chronic eyelid pressure may contribute to changes in corneal shape and optical quality.¹⁰ Such changes provide an additional potential mechanism through which the clinical characteristics of ptosis may be associated with visual function.

Comprehensive assessment of congenital ptosis should therefore consider relevant clinical characteristics, including laterality, severity, levator function, eyelid position, and associated ocular findings, together with visual acuity. Evaluation of these features is particularly important because the anatomical severity of ptosis does not necessarily correspond directly to the degree of visual impairment. Differences in visual acuity may arise from a combination of eyelid-related visual-axis obstruction and associated refractive or binocular abnormalities.¹¹

Despite substantial evidence concerning the clinical manifestations and visual consequences of congenital ptosis, the distribution of clinical characteristics and visual acuity may vary across patient populations and clinical settings. A focused assessment of these features can better describe the clinical profile of affected patients and their associated visual function. Therefore, this cross-sectional study aimed to assess the clinical characteristics and visual acuity of patients with congenital ptosis.

METHODOLOGY

A descriptive cross-sectional study was conducted at DHQ Vehari Eye Complex, Vehari, Pakistan, to assess the clinical characteristics and visual acuity of patients with congenital ptosis. The study was conducted over four months following approval of the research synopsis. The study was conducted after obtaining the required ethical and institutional approval. We explained the study's purpose and procedures to all participants and, where appropriate, to their parents or legal guardians before enrollment. Participation was

voluntary, and we kept all collected information confidential throughout the study. We recruited 40 patients with congenital ptosis using convenience sampling. Both male and female participants aged 1-30 years were eligible for inclusion. We included participants with clinically diagnosed congenital ptosis who were available for clinical and visual-acuity assessment during the study period. We excluded patients with acquired ptosis, previous surgery or other treatment for ptosis, or coexisting ocular conditions that could affect visual acuity or clinical assessment.

We collected data using a structured questionnaire and standardized data collection proforma. We obtained relevant demographic and clinical information from participants or, where appropriate, from their parents or guardians. Recorded variables included demographic characteristics and clinical features of congenital ptosis, including head posture, laterality of ptosis, extraocular muscle movements, palpebral fissure height, marginal reflex distance (MRD), and levator palpebrae superioris function. Clinical findings were documented separately for the right and left eyes.

The diagnosis of congenital ptosis was based on the clinical assessment performed at the study site. Clinical characteristics were systematically documented on the standardized study proforma. Visual acuity was assessed separately for the right and left eyes using the visual-acuity assessment method routinely used at the study site. Visual acuity was categorized as 6/9-6/12, 6/18-6/24, 6/36-6/60, and >6/60 and recorded on the standardized data collection proforma together with the relevant clinical findings.

Completed data collection forms were reviewed for completeness and consistency before data entry. Data were entered and analyzed using SPSS version 24. Categorical variables were summarized using frequencies and percentages. Descriptive analysis characterized the study population's demographic profile, clinical characteristics, and visual-acuity findings. The association between visual acuity and palpebral fissure height was assessed separately for the right and left eyes using the Pearson chi-square test. A p-value of <0.05 was considered statistically significant.

RESULTS

The study included 40 patients with congenital

ptosis. Participants ranged in age from 1 to 30 years. Figure 1 presents the demographic distribution. Males constituted a slightly larger proportion of the sample than females, with 22 (55.0%) males and 18 (45.0%) females. Most participants were aged 0-10 years ($n = 25$, 62.5%), followed by those aged 11-20 years ($n = 13$, 32.5%), whereas only 2 (5.0%) participants were aged 21-30 years.

Table 1 summarizes the principal clinical characteristics of the affected eyes. Normal head posture was reported in 38 (95.0%) participants, whereas 2 (5.0%) had a tilted head posture. Full extraocular muscle movements were reported in 96% of right eyes and 94% of left eyes; corresponding absolute frequencies were not available in the source data and therefore are reported as percentages only. Palpebral fissure height differed between the two eyes. In the right eye, 21 (52.5%) participants had a fissure height of 1-2 mm, 9 (22.5%) had 3-4 mm, and 10 (25.0%) had >4 mm. In the left eye, 9 (22.5%) had 1-2 mm, 16 (40.0%) had 3-4 mm, and 15 (37.5%) had >4

mm. For marginal reflex distance (MRD), an MRD of 4 mm was reported in 23 (57.5%) right eyes and 13 (32.5%) left eyes, while an MRD of <4 mm was reported in 17 (42.5%) right eyes and 27 (67.5%) left eyes. Levator palpebrae superioris function was categorized as normal, good, fair, or poor. In the right eye, the reported proportions were 38%, 34%, 15%, and 13%, respectively, while in the left eye they were 18%, 38%, 20%, and 24%, respectively.

Visual acuity was assessed separately for both eyes, and results are presented in Table 2. In the right eye, most participants (30; 75.0%) had visual acuity of 6/9-6/12, whereas 7 (17.5%) had visual acuity of 6/18-6/24, while 3 (7.5%) had 6/36-6/60. No participant reported right-eye visual acuity worse than 6/60. In the left eye, 23 (57.5%) participants had visual acuity of 6/9-6/12, 11 (27.5%) had 6/18-6/24, and 5 (12.5%) had 6/36-6/60. One participant (2.5%) had visual acuity worse than 6/60. Thus, reduced visual acuity was more frequently observed in the left eye.

Figure 1: Demographic characteristics of participants with congenital ptosis

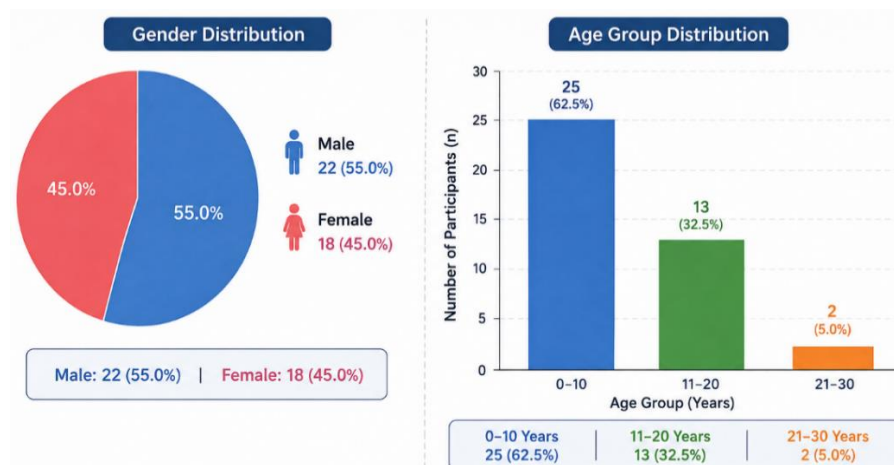


Table 1: Clinical characteristics of participants with congenital ptosis by eye

Clinical characteristic	Category	Right eye n (%)	Left eye n (%)
Extraocular muscle movements	Full	96.0%	94.0%
	Restricted	4.0%	6.0%
Palpebral fissure height	1-2 mm	21 (52.5)	9 (22.5)
	3-4 mm	9 (22.5)	16 (40.0)
	>4 mm	10 (25.0)	15 (37.5)
Marginal reflex distance	4 mm	23 (57.5)	13 (32.5)
	<4 mm	17 (42.5)	27 (67.5)
Levator palpebrae superioris function	Normal	38.0%	18.0%
	Good	34.0%	38.0%
	Fair	15.0%	20.0%

	Poor	13.0%	24.0%
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Table 2: Distribution of visual acuity in the right and left eyes

Visual acuity	Right eye n (%)	Left eye n (%)
6/9-6/12	30 (75.0)	23 (57.5)
6/18-6/24	7 (17.5)	11 (27.5)
6/36-6/60	3 (7.5)	5 (12.5)
>6/60	0 (0.0)	1 (2.5)
Total	40 (100.0)	40 (100.0)

The distribution of visual acuity according to palpebral fissure height in the right and left eyes is presented in Table 3. In the right eye, most participants had visual acuity of 6/9-6/12 (n = 30), of whom 18 had a palpebral fissure height of 1-2 mm, while 6 each had heights of 3-4 mm and >4 mm. Among participants with visual acuity of 6/18-6/24 (n = 7), fissure heights of 1-2 mm, 3-4 mm, and >4 mm were observed in 3, 2, and 2 participants, respectively. The three participants with visual acuity of 6/36-6/60 had fissure heights of 3-4 mm (n = 1) or >4 mm (n = 2). Overall, the reconstructed Pearson chi-square analysis indicated no statistically significant association between right-eye visual acuity and palpebral fissure height ($\chi^2 = 4.794$, df = 4, p = 0.309).

Table 3: Association of visual acuity with palpebral fissure height in the right and left eyes

Right-eye	1-2 mm	3-4 mm	>4 mm	Total
6/9-6/12	18	6	6	30
6/18-6/24	3	2	2	7
6/36-6/60	0	1	2	3
Total	21	9	10	40
Pearson $\chi^2 = 4.794$, df = 4, p = 0.309				
Left-eye	1-2 mm	3-4 mm	>4 mm	Total
6/9-6/12	9	8	6	23
6/18-6/24	0	5	6	11
6/36-6/60	0	3	2	5
>6/60	0	0	1	1
Total	9	16	15	40
Pearson $\chi^2 = 10.492$, df = 6, p = 0.105				

For the left eye, 23 participants had visual acuity of 6/9-6/12, including 9 with a fissure height of 1-2 mm, 8 with 3-4 mm, and 6 with >4 mm. Among the 11 participants with visual acuity of 6/18-6/24, fissure heights of 3-4 mm and >4 mm were recorded in 5 and 6 participants, respectively. The five participants with visual acuity of 6/36-6/60 had fissure heights of 3-4 mm (n = 3) or >4 mm (n = 2), while the single participant with visual acuity

worse than 6/60 had a fissure height of >4 mm. The reconstructed Pearson chi-square analysis likewise showed no statistically significant association between left-eye visual acuity and palpebral fissure height ($\chi^2 = 10.492$, df = 6, p = 0.105). Thus, although visual acuity varied descriptively across palpebral fissure-height categories in both eyes, neither association was statistically significant.

DISCUSSION

The present study characterized the clinical features and visual acuity of 40 patients with congenital ptosis and demonstrated substantial variation in eyelid measurements and levator palpebrae superioris function. Most participants were children, with 62.5% aged 0-10 years, while males constituted a slight majority (55.0%). The predominance of younger patients is clinically important because congenital ptosis is primarily encountered during childhood and may affect visual development during the period when amblyopia can develop. However, including participants up to 30 years of age also shows that congenital ptosis may remain clinically relevant beyond early childhood.

The clinical measurements in the present study showed considerable variation between the two eyes. The left eye had a higher proportion of MRD <4 mm (67.5%) and a greater proportion of fair or poor levator function than the right eye. These findings are consistent with the recognized relationship between levator muscle dysfunction and the external manifestations of congenital ptosis. Bagheri et al., in a study of 77 patients with unilateral congenital ptosis, demonstrated that levator muscle function was directly correlated with both lid fissure height and MRD, confirming that impaired levator function contributes to reduced eyelid aperture and greater ptosis severity.¹² Their findings are relevant to the present study because the same clinical

parameters - levator function, palpebral fissure height, and MRD were assessed in this cohort.

The predominance of normal head posture in the present study is also notable, with 38 (95.0%) participants maintaining a normal head position. Although abnormal head posture can occur as a compensatory response to ptosis, the present findings indicate that such compensation was uncommon in this cohort. Because head posture was recorded descriptively and no visual-field assessment was performed, the present study cannot establish a relationship between head posture and visual function. Therefore, the finding should be interpreted as a clinical characteristic rather than as evidence of preserved visual function.

Visual acuity was relatively well preserved in most participants. In the right eye, 75.0% had visual acuity of 6/9–6/12, compared with 57.5% in the left eye. Nevertheless, reduced visual acuity was more frequent in the left eye, including 12.5% with 6/36–6/60 and one participant with acuity worse than 6/60. This variation is clinically important because visual impairment in congenital ptosis may be influenced by factors beyond the degree of eyelid drooping alone. Wu et al. reported that congenital ptosis was associated with refractive error, strabismus, and amblyopia, with visual impairment being more pronounced in patients with unilateral ptosis and those with optical-axis obstruction.¹³

The principal finding of the present study was no statistically significant association between palpebral fissure height and visual acuity. In the right eye, the association was not significant ($\chi^2=4.794$, $df=4$, $p=0.309$), and the corresponding association in the left eye was also not significant ($\chi^2=10.492$, $df=6$, $p=0.105$). Although the descriptive distributions showed some variation in visual acuity across fissure-height categories, these differences were insufficient to demonstrate a statistically significant relationship. This finding is particularly important because palpebral fissure height represents an anatomical measure of eyelid aperture. In contrast, visual acuity reflects the combined effects of ocular optics, retinal image quality, binocular development, and amblyogenic factors.

The absence of a significant relationship in the present study is supported by population-based evidence demonstrating that visual impairment in childhood ptosis has multiple potential causes.

Griepentrog et al. studied 107 children with ptosis and found that amblyopia occurred in 14.9% of the cohort; among children with simple congenital ptosis, only 8.6% had amblyopia attributable solely to eyelid occlusion, whereas other cases were associated with refractive error or strabismus.¹⁴ These findings provide a strong clinical explanation for the present result: a larger or smaller palpebral fissure does not necessarily translate into a corresponding difference in visual acuity because visual function may be influenced by mechanisms other than direct visual-axis obstruction.

Similarly, Dray and Leibovitch reported strabismus in 20.8% and amblyopia in 23% of 130 patients with congenital ptosis, while amblyopia was attributable solely to ptosis in only a small subgroup.¹⁵ This supports the interpretation that congenital ptosis should be evaluated as a condition with both anatomical and functional dimensions. In the present study, the generally favorable visual-acuity distribution does not exclude clinically meaningful visual abnormalities, particularly because the study protocol did not include assessment of refractive error, anisometropia, strabismus, or formal amblyopia.

An additional study by Mocan et al. specifically examined whether clinical parameters were associated with visual-acuity levels in patients with unilateral congenital ptosis, making it particularly relevant to the present investigation.¹⁶ The study evaluated the possible associations of levator function and degree of ptosis with visual acuity, directly addressing the clinical question underlying the current analysis. This literature, together with the present non-significant chi-square findings, supports the view that structural ptosis measurements should not be used in isolation to predict visual acuity.

The study has several limitations. The sample was small and recruited through convenience sampling from a single eye-care center, limiting generalizability. In addition, the study did not quantify refractive error, anisometropia, strabismus, or amblyopia, which are important potential determinants of visual acuity in congenital ptosis. The categorical visual-acuity assessment may also have reduced statistical sensitivity compared with continuous logMAR measurements. Despite these limitations, the study provides clinically relevant descriptive data and demonstrates that eyelid morphology and visual

function should be assessed as related but distinct

CONCLUSION

The present study found considerable variation in palpebral fissure height, MRD, and levator function among patients with congenital ptosis. Most participants had relatively good visual acuity, although reduced vision was more frequent in the left eye. Importantly, palpebral fissure height was not significantly associated with visual acuity in either eye, indicating that eyelid aperture alone may not reflect visual function. Therefore, clinical assessment of congenital ptosis should not depend only on the visible degree of eyelid drooping. In children, assess visual acuity alongside MRD, palpebral fissure height, and levator function. Refractive error, anisometropia, ocular alignment, and amblyopia should also be specifically assessed because visual impairment may occur despite the absence of a significant relationship between eyelid measurements and visual acuity.

DECLARATIONS

Consent to participate: Written consent was obtained from all participants. All methods were performed in accordance with the relevant guidelines and regulations.

Availability of data and materials: Data will be made available upon request. The corresponding author will submit all dataset files.

Competing interests: None.

Funding: None.

CONSORT guidelines: All methods were performed in accordance with the relevant guidelines and regulations.

AUTHOR CONTRIBUTIONS

AI: Conceptualization, Supervision, Methodology, Writing - original draft, Writing - review & editing.

MA: Investigation, Formal Analysis, Writing - review & editing.

KA: Data curation, Formal analysis, Investigation.

EN: Formal analysis, Methodology, Validation.

HN: Validation, Writing - review & editing.

MR: Investigation, Validation, Formal analysis.

BH: Investigation, Writing - review & editing

All authors reviewed and approved the final version of the manuscript and agreed to be accountable for all aspects of the work.

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