

Visual Disorders in Children with Congenital Ptosis: A Cross-Sectional Analysis of Amblyopia, Strabismus and Refractive Error

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ABSTRACT

Purpose: The aim of this study was to investigate the rates, types and characteristics of visual disorders in children diagnosed with congenital ptosis.

Methods: An institution based cross sectional study was conducted for children with congenital ptosis aged 6 months to 16 years between October 2020 and September 2021 using an interviewer administered structured questionnaire. Data was collected including age at the time of presentation, sex, ethnicity, laterality and severity of ptosis and types of visual disorders. Statistical analysis was performed to determine any significant associations between the severity of congenital ptosis and the occurrence of visual disorders.

Results: We enrolled 63 children with congenital ptosis, mean age was 5.86 years (SD \pm 4.3). The overall rate of visual disorders consisted of amblyopia was 23.8%; strabismus was 19.7%; refractive errors were 41.3%. Male and unilateral ptosis was predominating among children with visual disorders. Family history was observed in 53.3% amblyopia, 42.3% strabismus and 58.3% refractive error. Furthermore, a significant association was found between the severity of congenital ptosis and the occurrence of amblyopia ($p < 0.05$).

Conclusion: Visual disorders including amblyopia, strabismus and refractive errors in children with congenital ptosis are common, early detection and appropriate intervention is importance. Family history was observed. Amblyopia was associated with severity of ptosis. Effective strategies for the management and treatment of visual disorders in children with congenital ptosis are recommended.

Keywords: Congenital ptosis; Amblyopia; Strabismus; Refractive errors; Visual disorders

INTRODUCTION

Congenital ptosis, a condition characterized by drooping of one or both eyelids from birth, can have significant implications for visual development in affected individuals, potentially leading to the development of visual disorders such as amblyopia, strabismus and refractive errors [1,2]. Amblyopia, commonly known as "lazy eye," is a neurodevelopmental visual disorder that results in reduced visual acuity and impaired binocular vision due to abnormal visual development during childhood [3]. Amblyopia often occurs due to the presence of a significant eyelid droop obstructing the visual axis during the critical period of visual development [4]. It can also be due to high astigmatism, anisometropia or strabismus [5]. Strabismus refers to the

misalignment of the eyes, which can result in impaired binocular vision [6]. Refractive error, including astigmatism, myopia and hyperopia, cause blurred vision due to abnormalities in the eye's ability to focus light accurately.

Several studies have reported an association between congenital ptosis and visual disorders in children. According to a study, it was found that children with congenital ptosis had a higher prevalence of amblyopia compared to the general population. Additionally, a higher incidence of strabismus was reported in children with congenital ptosis [7]. These findings highlight the importance of understanding the prevalence and characteristics of visual disorders in children with congenital ptosis to ensure appropriate diagnosis and intervention.

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Despite the available literature on the association between congenital ptosis and visual disorders, there is a need for further research to assess the specific prevalence rates and characteristics of amblyopia, strabismus and refractive errors in Ethiopian population. This cross-sectional analysis aimed to fill this research gap by examining the occurrence of these visual disorders in a population of children diagnosed with congenital ptosis. By conducting this analysis, healthcare professionals can gain a deeper understanding of the impact of congenital ptosis on visual development and identify potential risk factors or patterns associated with the occurrence of these visual disorders. This knowledge will facilitate early detection, appropriate intervention and targeted treatment strategies for affected children. Moreover, it will contribute to the development of guidelines for regular vision screenings and comprehensive eye care in children with congenital ptosis.

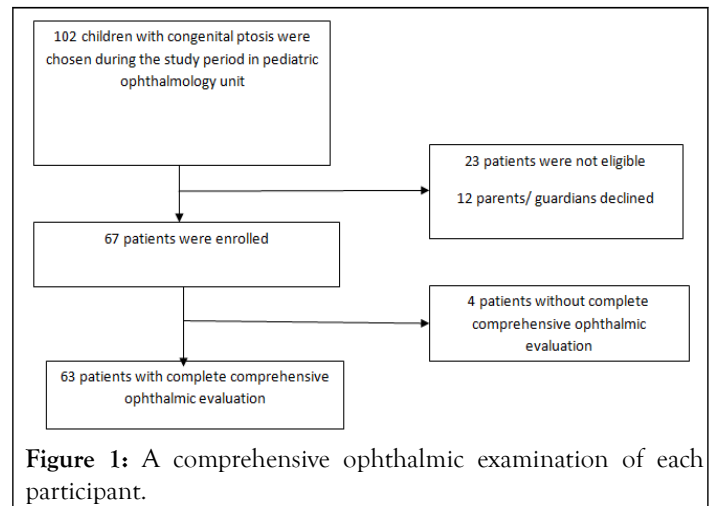
MATERIALS AND METHODS

Patients and methods

A cross-sectional study included consecutive patients diagnosed with congenital ptosis for the first time, as well as those children with settled diagnoses returning for follow-up visits at pediatric ophthalmology unit of a comprehensive specialized hospital, aged between 6 months and 16 years.

Children with acquired causes of ptosis (e.g. trauma, eyelid tumors), those who had neurogenic conditions like oculomotor nerve palsy and marcus gunn winking syndrome, and those who had associated syndromes like blepharophimosis syndrome, other structural ocular diseases such as media opacity and retinal disease, those who had previous ptosis surgery, were excluded from the study. Children whose parents or guardians did not provide informed consent were also excluded from the study.

The study protocol was reviewed and approved by the institutional review board, ensuring that the study adhered to ethical guidelines and protected the rights and welfare of the participants under the 1975 Helsinki declaration, as revised in 1983. The study was conducted over a period of one year, from October 2020 to September 2021. Children with congenital ptosis were chosen and invitation to participate in the study was extended to the parent/guardian and written informed consent was obtained for all participant children before their inclusion in the study. By assigning a code to each patient, confidentiality and anonymity were preserved. Investigators were present to clarify any questions or concerns raised by the participants' parents/guardians (Figure 1).



Data collection was carried out using an interviewer administered structured questionnaire the questionnaire used in this study was adapted from a previous study [8]. The questionnaire was piloted to test its effectiveness and identify any areas for improvement in data collection. Based on the results of the pilot study, necessary modifications were made to enhance the questionnaire's reliability and validity. The modified questionnaire was then utilized to collect data and gather relevant information during the routine clinical practice regarding to demographic information of the child including age at the time of presentation, sex, and ethnicity, laterality and severity of ptosis and comprehensive ocular examination and refraction.

A comprehensive ophthalmic examination of each participants including assessment of visual acuity using Snellen chart or use appropriate assessment methods for each age group and assessing the severity of ptosis, eyelid position, presence of associated factors such as strabismus or nystagmus and any associated ocular abnormalities. Ptosis measurement was done and confirmed by the pediatric ophthalmologist, utilizing measurement of Marginal Reflex Distance 1 (MRD1) which is measured from upper eyelid to the central corneal light reflex using a ruler [9].

Refraction data were obtained to determine the refractive error status of each participant using cycloplegic retinoscopy or autorefractor or subjective refraction. Additionally, slit lamp bio microscopy was performed to examine the anterior segment of the eye, while fundus examination (direct ophthalmoscopy or +90 D sph lens) was carried out to assess the posterior segment.

The diagnosis and type of amblyopia was made based on the criteria established by the American Association for Pediatric Ophthalmology and Strabismus (AAPOS) [10].

Operational definition

Children are persons from 6 months to 16 years.

Mixed amblyopia: The presence of both significant interocular difference in refractive error and strabismus, along with reduced visual acuity in the affected eye.

Severity of congenital ptosis was assessed based on MRD1 measurement.

Mild ptosis: MRD1 measurement greater than 2 mm.

Moderate ptosis: MRD1 measurement between 1 mm-2 mm.

Severe ptosis: MRD1 measurement less than 1 mm or absent (MRD1=0).

Types of refractive error

Astigmatism was defined if it was less/more than ± 0.5 D Cyl.

Myopia was defined if it is less than -0.5 D Sph.

Hyperopia was defined if it is more than +1.00 D Sph.

Data analysis procedure

Statistical analysis was conducted using appropriate software, and rates of amblyopia, strabismus and refractive error in patients with congenital ptosis were calculated. Descriptive statistics such as mean, standard deviation and frequency distributions were used to summarize the characteristics of the study population. To find any patterns or trends in the rate of various types of amblyopia, strabismus and refractive error, a statistical analysis utilizing Pearson's *chi-square* test was

conducted. Additionally, a *chi-square* test was used to examine the relationships between the existence of amblyopia and the degree of ptosis, the kind of strabismus and the type of refractive error.

RESULTS

A total of 63 children with congenital ptosis were enrolled in the study. The mean age of the participants was 5.86 years (SD=4.3), the participants ranged in age from 6 months to 15 years. Table 1 shows, the majority of children with congenital ptosis in the study was between the ages of >5 and 12 years. There was a higher prevalence of males. Ptosis was more commonly observed in the left eye compared to the right eye with a smaller percentage having bilateral involvement. A significant proportion of children had a positive family history of ptosis. The ethnic distribution was varied, with Oromo and Amhara being the most common ethnic groups represented. In terms of severity, the majority of cases were classified as moderate and fewer were severe.

Table 1: Demographic characteristics of children with congenital ptosis.

Characteristics	Frequency	%
Age group (yr)		
0-1	15	23.8
>1-3	8	12.7
>3-5	9	14.3
>5-12	26	41.3
>12-16	5	7.9
Sex		
Male	44	69.8
Female	19	30.2
Laterality		
Right	16	25.4
Left	40	63.5
Bilateral	7	11.1
Family history		
Yes	21	33.3
No	42	67.7
Ethnicity		

Afar	5	7.9
Amhara	15	23.8
Gambela	2	3.2
Gurage	10	15.9
Oromo	22	34.9
Somale	3	4.8
Tigre	6	9.5
Severity		
Mild	14	22.2
Moderate	38	60.3
Sever	11	17.5

Out of 70 eyes with congenital ptosis, 18.6% (n=13) had Best Corrected Visual Acuities (BCVA) less than or equal to 6/24 and 5.7% (n=4) had Un-Central, Steady and Un-Maintained (UCSUM) (Table 2).

Table 2: Visual acuity in ptotic eyes (n=70).

BCVA*	Frequency	%
6/6-6/18	31	40
6/24-6/60	8	15.71
5/60-3/60	2	2.86
<3/60	3	4.29
CSM**	22	31.43
UCSUM***	4	5.7
Total eyes	70	100

Note: *Best corrected visual acuity; **Central steady maintained; ***Un-central steady un-maintained

Information was available for 15 (23.8%) amblyopic children of these, 8 (53.3%) had anisometric amblyopia. (Figure 2), 5 (33.3%) children with congenital ptosis had mixed amblyopia, which is attributed to a combination of strabismus and refractive error. Additionally, one participant had strabismic amblyopia and another had deprivational amblyopia.

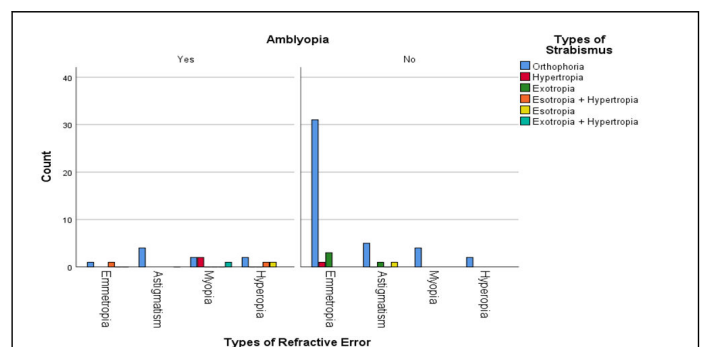


Figure 2: Types of amblyopia, strabismus and refractive error in children with congenital ptosis.

The characteristics of the different visual disorders amblyopia, strabismus and refractive error are shown in Table 3. Refractive error is the most prevalent visual disorder among those studied, followed by amblyopia. On average, individuals with strabismus tend to be slightly older than those with amblyopia or refractive error. Males make up the majority in each visual disorder group

and predominantly unilateral. A significant percentage of individuals with visual disorders have a family history of visual disorders. This suggests a potential genetic or familial component in the development of these visual disorders. More refractive error had moderate and severe types of ptosis, but this difference didn't reach statistical significance.

Table 3: Descriptive characteristics of the different visual disorders, mean \pm SD or N (%).

Characteristics	Amblyopia	Strabismus	Refractive error
N (%)	15 (23.8%)	12 (19.0%)	26 (41.2%)
Age (yrs)	6.27 \pm 2.79	7.27 \pm 4.51	6.08 \pm 3.25
Male	12 (80%)	9 (75%)	19 (73.1%)
Unilateral	13 (86.7%)	9 (75%)	22 (84.6%)
Family history	8 (53.3%)*	7 (58.3%)	11 (42.3%)
Severity of ptosis			
Mild	0 (0%)	2 (16.7%)	2 (7.7%)*
Moderate	9 (60%)**	6 (50.0%)	18 (69.2%)*
Severe	6 (40%)**	4 (33.3%)	6 (23.1%)*

Note: *p=0.06; **p<0.05; ***p=0.08, *chi-square* test

DISCUSSION

This study aimed to investigate the rates and characteristics of visual disorders, including amblyopia, strabismus and refractive error, in pediatric patients with congenital ptosis at a comprehensive specialized hospital. The findings of this study contribute to the understanding of visual health in this specific population and have important implications for clinical practice and healthcare policies in Ethiopia.

The prevalence of amblyopia in our study population was found to be 23.8%, which is consistent with previous studies conducted in congenital ptosis with overall pooled prevalence of 22.7%. Amblyopia is a common visual disorder associated with congenital ptosis and early detection and intervention are crucial to prevent long-term visual impairment. Our findings emphasize the importance of routine screening for amblyopia in pediatric patients with congenital ptosis, as prompt management can lead to improved visual outcomes.

Strabismus was found to be present in 19.7% of the study population, which is similar to the prevalence reported in the study done by Junior G, et al. This highlights the increased risk of strabismus in children with congenital ptosis. Strabismus can have significant implications for binocular vision and can lead to amblyopia if not addressed early.

Refractive error was detected in 41.3% of the study population, with astigmatism (42.3%) being the most common refractive error. These findings align with previous studies that have

reported a higher prevalence of refractive error in children with congenital ptosis compared to the general population. Correcting refractive errors through appropriate prescription of glasses or contact lenses is crucial to optimize visual acuity and promote normal visual development.

The results of this study underscore the importance of a multidisciplinary approach in the management of pediatric patients with congenital ptosis. Collaboration between pediatric ophthalmologists, orthoptists and oculoplastic surgeons is essential to ensure comprehensive evaluation, accurate diagnosis and appropriate management of visual disorders in these patients. Early identification and intervention can significantly improve visual outcomes and enhance the quality of life for affected children.

While this study provides valuable insights into the rates and characteristics of visual disorders in pediatric patients with congenital ptosis at a comprehensive specialized hospital, there are some limitations to consider. The study was conducted at a single hospital, which may limit the generalizability of the findings to the broader Ethiopian population. Future studies with larger sample sizes and multi-center designs would be beneficial to obtain a more representative picture of visual disorders in this population.

CONCLUSION

In conclusion, this study highlights the high rate of amblyopia, strabismus and refractive error in pediatric patients with congenital ptosis at a comprehensive specialized hospital. The findings emphasize the importance of early screening, prompt intervention and regular follow-up visits to optimize visual outcomes in these patients. Further research is warranted to explore the long-term visual prognosis and effectiveness of different treatment modalities. These findings can guide clinical practice and inform healthcare policies to improve the visual health of pediatric patients with congenital ptosis in Ethiopia.

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