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## Amblyopia in Childhood Eyelid Ptosis

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### Abstract

**Purpose**—To report the prevalence and causes of amblyopia among children with ptosis diagnosed in a well-defined population over a 40-year period.

**Design**—Retrospective, population-based cohort study.

**Methods**—We retrospectively reviewed the charts of one hundred and seven patients (< 19 years) for the prevalence and causes of amblyopia who were diagnosed with childhood ptosis as residents of Olmsted County, Minnesota, from January 1, 1965, through December 31, 2004.

**Results**—Amblyopia was diagnosed in 16 (14.9%) of the 107 patients with childhood ptosis. Fourteen (14.6%) of 96 patients diagnosed with a congenital form of ptosis demonstrated amblyopia. Twelve (14.8%) of the 81 patients diagnosed with simple congenital ptosis had amblyopia, 7 (8.6%) of which were due solely to eyelid occlusion of the visual axis. The causes of amblyopia in the remaining 5 patients were due to significant refractive error in 3 patients and strabismus in 2 patients.

**Conclusions**—Amblyopia occurred in 1 in 7 children diagnosed with ptosis in this population-based cohort. Approximately half of those with amblyopia, or less than 10% of all patients, were due solely to eyelid occlusion of the visual axis.

Amblyopia is the most common cause of monocular vision loss in children.<sup>1,2</sup> Among patients with childhood ptosis, however, the incidence of amblyopia has been reported to be higher than that in the general population.<sup>3–11</sup> Although this disparity is thought to be the result of an increased prevalence of strabismus, significant refractive error, and eyelid occlusion of the visual axis, none of the prior reports are population-based. The purpose of this study is to report the prevalence and causes of amblyopia among a cohort of 107

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### Disclosure

The authors have no financial disclosures.

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children diagnosed with ptosis over a 40-year period while residing in Olmsted County, Minnesota.

## Methods

The medical records of 107 patients younger than 19 years of age with childhood ptosis, who were residents of Olmsted County, Minnesota and diagnosed between January 1, 1965, and December 31, 2004, were retrospectively reviewed. The inclusion criteria for study, incidence and demographic data of these 107 patients have been previously reported.<sup>12</sup> Institutional Review Board approval was obtained for this study. The cases were identified using the resources of the Rochester Epidemiology Project, a medical record linkage system designed to capture data on any patient–physician encounter in Olmsted County, Minnesota.<sup>13</sup> The population of this county is relatively isolated from other urban areas and virtually all medical care is provided to its residents by Mayo Clinic, Olmsted Medical Group, and their affiliated hospitals. Patients not residing in Olmsted County at the time of their diagnosis with childhood ptosis were excluded from the study.

Amblyopia was defined as two lines or more difference between the two eyes while wearing the proper prescription, or lack of central, steady and maintained fixation in the ptotic eye with presence of normal central, steady and maintained fixation in the nonptotic eye. Each patient diagnosed with amblyopia was managed by an ophthalmologist (pediatric, oculoplastic or comprehensive ophthalmologist). The initial and subsequent refractions were determined in the majority of patients following the topical administration of 1% cyclopentolate in younger patients and by a manifest refraction for older patients. All refractions were converted into their spherical equivalent. Significant refractive error was defined as the presence of anisometropia of at least one diopter difference between the spherical equivalents of each eye, hyperopia  $\geq 3$  diopters or astigmatism  $\geq 1$  diopter. Amblyopia as a result of strabismus was diagnosed in those with an intermittent or constant horizontal deviation of at least 10 prism diopters, or a vertical deviation of at least 2 prism diopters, and in whom no other cause for decreased vision was apparent. If the exam met the above criteria for both significant refractive error and strabismus, the cause of amblyopia was listed as “combination.” Cases of amblyopia secondary to eyelid occlusion of the visual axis alone, without frontalis muscle recruitment or chin-up posture, were determined to have neither significant refractive error nor strabismus.

## Results

Sixteen (14.9%) of the 107 study patients were diagnosed with amblyopia in Olmsted County, Minnesota, during the 40-year period at a median age of 4.0 years (range, 1 month to 10.2 years) (Table). Fifteen cases of amblyopia were diagnosed in patients with unilateral ptosis while one case was diagnosed in a childhood myasthenia gravis patient with bilateral ptosis. Of the 96 patients with a congenital form of ptosis, 14 (14.6%) patients were diagnosed with amblyopia, while 2 (14.3%) of the 11 patients with an acquired form of ptosis had amblyopia. All cases of amblyopia occurred in patients with unilateral ptosis with the exception of one patient with childhood myasthenia gravis. Further information concerning the forms of ptosis and causes of amblyopia are shown in the Table. There were no cases of amblyopia due to a combination of significant refractive error and strabismus. All of the patients had unilateral ptosis with the exception of the patient with childhood myasthenia gravis.

Twelve (14.8%) of the 81 patients with simple congenital ptosis were diagnosed with amblyopia, of which 7 (8.6%) cases were due to occlusion of the visual axis from the ptotic eyelid. Of the remaining five cases of amblyopia, 3 patients had a significant refractive error

including anisometropia in 2 patients and astigmatism in 1 patient, while the remaining 2 patients had an associated exotropic deviation.

## Discussion

Amblyopia occurred in 1 in 7 patients with any form of childhood ptosis in this population-based cohort diagnosed over a 40-year period. Fourteen (14.6%) of 96 cases were diagnosed in patients with a congenital form of ptosis and amblyopia as a result of visual axis occlusion alone occurred in less than ten percent of the study patients. These rates are at the low end of the range of previous non population-based estimates (14–48%) of amblyopia for all forms of congenital ptosis.<sup>3–11</sup>

Simple congenital ptosis is the most common form of childhood ptosis.<sup>12</sup> Within the group of 96 patients with any congenital form of ptosis, 81 patients were diagnosed with simple congenital ptosis, of which, twelve (14.8%) were diagnosed with amblyopia. This rate similarly falls at the low end of the range of previous non-population-based estimates, although it is comparable to some recent reports including Srinagesh and colleagues (25.3% of 87 patients) and Lin and colleagues (21.5% of 130 patients).<sup>8,11</sup>

Amblyopia has an estimated prevalence of 3.0% to 3.2% in the general population.<sup>9,14,15</sup> The rate among patients with congenital ptosis has been reported to be higher than that of the general population.<sup>3–11</sup> Previous clinical studies have examined the causes of amblyopia in the general population and have shown that approximately one third are due to anisometropia, one third to strabismus, and the remaining third to a combination of both disorders or a form of occlusive stimulus deprivation.<sup>1,16,17</sup> Occlusive stimulus deprivation amblyopia appears to be the least frequent subtype based on the relative rarity of the primary causative factors such as infantile cataract (2 to 4.5 of every 10,000 births) and childhood ptosis (7.9 per 100,000 less than 19 years of age).<sup>1,12,18,19</sup>

The precise cause of the increased prevalence of amblyopia among patients with congenital ptosis is controversial. Although some authors have argued that the occlusive effect of the ptotic eyelid(s) does not interfere with visual development, subsequent reports have demonstrated that between 1.6% and 12.3% of patients with a diagnosis of congenital ptosis will have amblyopia due solely to occlusive stimulus deprivation.<sup>3,5–7,9,19–22</sup> In the current report, within the group of 96 congenital ptosis patients, 7 (7.3%) cases of amblyopia were due to occlusion of the pupillary axis by the ptotic eyelid. These 7 cases of occlusion amblyopia accounted for half of all the cases (n=14) of amblyopia diagnosed in patients with congenital ptosis. All 7 patients were also diagnosed with simple congenital ptosis, the most prevalent subtype of congenital ptosis. Two of the 7 patients were recorded as having occlusion as the cause of ptosis despite not having their refractive error measured. Each child was seen only once in clinic. It is possible that each child had significant refractive error, although both patients were reported in the charts as having “severe” ptosis in the absence of a compensatory head tilt. One patient had a margin reflex distance of 0.5mm in the ptotic eyelid while the other patient had complete occlusion of the pupil due to the ptotic eyelid. Of the remaining 5 cases of amblyopia in patients with simple congenital ptosis, 3 were due to significant refractive error and 2 were due to strabismus.

Occlusion of the visual axis was the leading cause of amblyopia in patients with congenital ptosis in this report. This finding is in contrast to large referral-based retrospective studies of congenital ptosis in which the leading causes of amblyopia were strabismus or significant refractive error.<sup>5,7,9,11</sup> In a study by Harrad et al. of 216 cases of simple congenital ptosis referred for oculoplastics evaluation, 37 (17%) patients developed amblyopia, of which 20 (9.3%) cases were due to strabismus and 5 (2.3%) cases were due to stimulus deprivation of

the visual axis from the ptotic eyelid.<sup>5</sup> Likewise, in a study by Dray and Leibovitch of 130 patients who had surgical correction of their ptosis, 30 (23%) patients were diagnosed with amblyopia, of which 16 (12.3%) cases were due to strabismus and 9 (6.9%) cases were due to occlusion.<sup>7</sup> More recently, in a study of 92 patients with congenital ptosis, 22 (23.9%) of patients were diagnosed with amblyopia, with almost every case occurring in the context of coexisting anisometropia or strabismus.<sup>11</sup> The same authors note that congenital ptosis patients are at risk of developing anisometropic and strabismic amblyopia even if not originally detected, and routine monitoring that includes regular cycloplegic refractions is recommended.<sup>11</sup>

All cases of amblyopia occurred in patients with unilateral ptosis with the exception of one patient with childhood myasthenia gravis. The child had bilateral ptosis with symmetric palpebral fissure heights (4mm) and 30 prism diopters of exotropia. Due to the significant degree of exotropia, strabismus was recorded as the cause of amblyopia, although it has been speculated elsewhere that ptosis may be the initial cause of amblyopia that leads to strabismus.<sup>7</sup>

Three of the 96 patients with congenital ptosis in the current study were diagnosed with Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome (BPES).<sup>12</sup> Although amblyopia was not noted in this small group, the rate of amblyopia in patients with BPES has been previously reported as high as 56.4% and early surgery is recommended.<sup>23</sup> Two of the three patients underwent surgery at a mean age of 54.4 (range, 44.4 – 64.4) months, while one patient diagnosed with BPES at birth with mild unilateral ptosis had no signs of amblyopia by age 14.

There are several limitations to the findings in this study. Although a relatively isolated county, some residents of Olmsted with ptosis may have sought care outside the region, potentially underestimating the incidence of amblyopia in this population. Secondly, the presence of a compensatory head tilt has been correlated with a high incidence of amblyopia in patients with congenital ptosis in the absence of significant anisometropia and strabismus.<sup>7,24</sup> In the current study, there were recording inconsistencies as to the presence or absence of this pertinent finding during the 40-year period. Due to the young age of patients in the study (median age at diagnosis for simple congenital ptosis: 1.3 years; range 32 days to 16.7 years)<sup>12</sup>, we were unable to assess stereoacuity to indirectly identify amblyopia, thus potentially underestimating the overall prevalence of amblyopia. Finally, the ability to generalize the findings of the current report is limited by the demographics of Olmsted County; a relatively homogeneous semi-urban white population.

The findings of this study provide population-based prevalence rates for amblyopia in childhood ptosis diagnosed over a 40-year period. Amblyopia affected approximately 1 in 7 patients diagnosed with childhood ptosis, of which nearly half were due solely to eyelid occlusion of the visual axis.

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Table

Amblyopia in 107 patients <19 years diagnosed with childhood ptosis in Olmsted County, Minnesota from 1965 to 2004

Ptosis Etiology	Number (%)	Causes of Amblyopia (%)				Total
		Occlusion (Ptosis)	Strabismus	Refractive Error		
<b>CONGENITAL</b>						
Simple Congenital Ptosis	81 (75.7)	7 (8.6) <sup>†</sup>	2 (2.5)	3 (3.7)	12 (14.8)	
Blepharophimosis	3 (2.8)	0	0	0	0	
Congenital CNIII Palsy	3 (2.8)	0	2 (66.6)	0	2 (66.6)	
Marcus Gunn Jaw Wink	3 (3.8)	0	0	0	0	
Congenital Horner Syndrome	2 (1.9)	0	0	0	0	
Central Core Myopathy	1 (0.93)	0	0	0	0	
CFEOM*	1 (0.93)	0	0	0	0	
Myotonic Dystrophy	1 (0.93)	0	0	0	0	
Noonan's Syndrome	1 (0.93)	0	0	0	0	
<b>ACQUIRED</b>						
Aponeurotic Dehiscence	4 (3.7)	0	0	1 (25)	1 (25)	
Acquired CN III Palsy	2 (1.9)	0	0	0	0	
Acquired Homer Syndrome	2 (1.9)	0	0	0	0	
Traumatic Structural Ptosis	2 (1.9)	0	0	0	0	
Childhood Myasthenia Gravis	1 (1.9)	0	1 (100)	0	1 (100)	
<b>Total</b>	<b>107</b>	<b>7 (6.5)</b>	<b>5 (4.6)</b>	<b>4 (3.7)</b>	<b>16 (14.9)</b>	

<sup>†</sup> Two children were seen only once in clinic and did not have a refraction.

\* Congenital fibrosis of the extraocular muscles