Published in final edited form as:
Ophthalmology. 2011 June ; 118(6): 1180–1183. doi:10.1016/j.ophtha.2010.10.026.

Incidence and Demographics of Childhood Ptosis

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Abstract

Purpose—To report the incidence and demographics of childhood ptosis diagnosed over a 40-year period in a well-defined population.

Design—Retrospective, population-based cohort study.

Participants—Patients (< 19 years) diagnosed with childhood ptosis as residents of Olmsted County, Minnesota, from January 1, 1965, through December 31, 2004

Methods—The medical records of all potential patients identified by the Rochester Epidemiology Project were reviewed.

Main Outcome Measures—Calculated annual age- and sex-specific incidence rates and demographic information.

Results—A total of 107 children were diagnosed with ptosis during the 40-year period, yielding an incidence of 7.9/100,000 < 19 years (95% confidence interval [CI]: 6.4-9.5) of age. Ninety-six (89.7%) of the 107 were congenital in onset, 81 (75%) of which had simple congenital ptosis, yielding a birth prevalence of 1 in 842 births. A family history of childhood ptosis was present in twelve percent of queried patients with simple congenital ptosis. Three (4%) of the simple congenital ptosis cases were bilateral and 55 (68%) of the unilateral cases involved the left upper eyelid (95% CI: 57%-78%, p<0.001).

Conclusion—Childhood ptosis was diagnosed in 7.9 per 100,000 patients less than 19 years (95% CI: 6.4-9.5). Simple congenital ptosis was the most prevalent form, occurring in 1 in 842 births, and significantly more likely to involve the left side.

Ptosis, a drooping of the upper eyelid below its normal position, is a relatively common form of eyelid malposition in children and adults. While adult ptosis generally results in reversible obscuration of the visual axis, childhood ptosis may be associated with amblyopia.[1] There are a number of classification schemes based on the underlying etiologic mechanism or age of onset.[2-5] Prior reports have described the clinical characteristics and management of congenital ptosis from large tertiary referral-based practices.[1,6-9] There are, however, to the best of our knowledge, no population-based studies of childhood ptosis in the literature. The purpose of this study is to report the prevalence and clinical features of

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The author(s) have no proprietary or commercial interest in any materials discussed in this article.

childhood ptosis diagnosed during a 40-year period among patients < 19 years of age who were residents of Olmsted County, Minnesota.

Subjects and Methods

The medical records of all patients younger than 19 years of age who were residents of Olmsted County, Minnesota when diagnosed with ptosis between January 1, 1965, and December 31, 2004, were retrospectively reviewed. Institutional Review Board approval was obtained for this study. Potential cases of ptosis 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.[10] 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 were excluded from the study.

Unilateral ptosis was defined as either a measured palpebral fissure asymmetry of ≥ 1mm between the two upper eyelids or a marginal reflex distance (MRD) of < 2.5 mm. Bilateral ptosis was defined as a MRD of < 2.5 mm in both eyes. The medical records of all potential patients were diagnosed and reviewed by an ophthalmologist. Patients were considered to have a congenital form of ptosis if they presented to a physician within the first few months of life unless an acquired etiology was specifically noted in the chart. Late-presenting cases were deemed congenital if symptoms were observed within the first few months of life and verified by a photograph or parental history. A family history of congenital ptosis was collected if reported within the medical record. Ptosis as a result of ocular or other surgeries were specifically excluded, as were patients with mechanically occlusive eyelid lesions such as neurofibromas or hemangiomas.

To determine the incidence of childhood ptosis in Olmsted County, annual age- and sex-specific incidence rates were constructed using the age-and sex-specific population figures for this county from the United States Census. For those cases of childhood ptosis deemed to have been congenital in onset, the birth prevalence was also calculated from the number of births occurring from January 1, 1965, through December 31, 2004, using the annual birth incidence for this county. The 95% confidence intervals were calculated using assumptions based on the Poisson distribution.

Results

A total of 107 patients less than 19 years of age were diagnosed with childhood ptosis in Olmsted County, Minnesota, during the 40-year study period. This number corresponds to an age-and gender-adjusted incidence of 7.9 per 100,000 residents less than 19 years of age (95% confidence interval [CI]: 6.4-9.5). There were 59 (55%) males (M) with an age-adjusted incidence rate of 8.4 (95% CI: 6.3 – 10.6) per 100,000 compared to and 48 (45%) females (F) with an age-adjusted incidence rate of 7.4 (95% CI: 5.3 – 9.5) per 100,000 (P=0.42). The individual forms and demographics of these patients are shown in Table 1. An additional 11 children (7 of which were thought to have simple congenital ptosis) were diagnosed with ptosis but were excluded from the study for a lack of eyelid measurements. If these 11 were included, the incidence of childhood ptosis would be 8.8 (95%CI: 7.2-10.4) per 100,000 < 19 years of age.

Ninety-six (89.7%) of the 107 cases were congenital in onset, of which 81 (84.3%) were diagnosed with simple congenital ptosis, yielding a birth prevalence of 1 in 842 live births. The median age at diagnosis for simple congenital ptosis was 1.3 years (range, 32 days to

16.7 years). Three (4%) of the simple congenital ptosis cases were bilateral and 55 (68%) of the unilateral cases involved the left upper eyelid (95% CI: 57%-78%, p<0.001). Prematurity was noted in 2 (3.7%) of 54 simple congenital ptosis cases for which birth records were available. Seven (11.7%) of the 60 simple congenital ptosis cases for which records were available, had a family history of ptosis.

Discussion

Childhood ptosis was diagnosed in 107 children or 7.9 per 100,000 patients less than 19 years of age as residents of Olmsted County, MN during the 40-year study period. Simple congenital ptosis was the most prevalent subtype of childhood ptosis diagnosed, comprising 76% of all cases and occurring in 1 in 842 live births. A presumed congenital onset occurred in 90% of patients and the left eyelid was involved in two-thirds of patients with unilateral simple congenital ptosis.

This is the first population-based report on the incidence of childhood ptosis. Berry-Brincat and coauthors reported the relative proportion of various forms of ptosis in a large 10-year review of 155 children in the United Kingdom (UK) who presented for corrective surgery.[6] The most common form of ptosis noted was myogenic developmental abnormality (114 patients; 74%), which in the current study is termed simple congenital ptosis. Although not designed to determine population-based rates of prevalence or incidence, the predominance of myogenic development ptosis closely correlates with the prevalence of simple congenital ptosis (76%) in the present study. Other types of ptosis noted in the UK study were third nerve palsy (7.7%), Marcus-Gunn jaw winking ptosis (5%), mechanical ptosis (5%) from an eyelid or orbital mass, blepharophimosis (4.5%), orbital fibrosis syndrome (2%), Horner's syndrome (0.6%), myasthenia gravis (0.6%) and post-traumatic ptosis (0.6%).[6]

Left eye predominance in unilateral simple congenital ptosis has not been noted in prior reports. The statistically significant p-value of < 0.001 was calculated by assuming a right to left ratio of 50:50. Another childhood ocular disorder with left eye predominance is Duane's retraction syndrome (DRS), which is reported to occur in 59% of patients, although the reason for this predominance is unknown.[11-13] The pathophysiology of DRS appears to be a result of congenital "miswiring" of the medial and lateral rectus muscles associated with an absent or hypoplastic abducens nerve.[13] Aberrant innervation of the lateral rectus by motor fibers that derive from a branch of the inferior division of the oculomotor nerve leads to synkinesis with fibrosis. The curious predominance of left side involvement in simple congenital ptosis may suggest, in some cases, like DRS, an underlying failure of proper motor innervation. There have also been reports of left eye predominance in patients with Marcus Gunn jaw winking synkinesis,[14,15] although these finding have not been replicated by others.[16,17] A recent non-population based consecutive case series found a slight predominance of left eye involvement that did not reach statistical significance.[18]

A higher frequency of males has been reported in two large cohorts of patients undergoing corrective childhood ptosis surgery.[6,8] In the current study, there was a trend but no statistically significant difference in sex distribution of patients with childhood ptosis (M 54(56%): F 42(44%), p=0.54) or within the subcategory of simple congenital ptosis (M 46(57%): F 35 (43%), p=0.65). Moreover, a family history of childhood ptosis was noted in seven (11.7%) patients with simple congenital ptosis which, to the best of our knowledge, has not been previously commented on in the literature.

Simple congenital ptosis has traditionally been categorized as a myogenic disorder based on histopathologic characteristics.[2-5, 19-24] More recently, cases of oculomotor synkinesis in congenital unilateral ptosis and an improved understanding of neural innervation as a

fundamental prerequisite for embryonic differentiation of striated muscle fibers, have led others to argue for an underlying neurogenic origin to many cases of simple congenital ptosis.[25-28] Dysgenesis of the oculomotor nerve may result from either abnormal development of third nerve nuclei motor neurons or a failure of migration involving the superior division of the oculomotor nerve destined for the levator palpebrae superioris.[27-28]

The relatively rare incidence of other forms of childhood ptosis in this cohort is consistent with prior reports.[6,8] Four cases of aponeurotic dehiscence were due to hard contact lens use. The two cases each of acquired third cranial nerve (CNIII) palsy and acquired Horner's syndrome were due to trauma. An additional two cases of trauma resulting in architectural damage to the eyelid was classified as traumatic structural ptosis.

There are several limitations to the findings in this study. While the majority of patients were diagnosed by a pediatric ophthalmologist and/or an oculoplastic specialist, some cases may have been evaluated by a non-ophthalmologist, and potentially excluded from the study, thereby underestimating the true incidence of the disease in this population. Second, although a relatively isolated county, some residents of Olmsted with ptosis may have sought care outside the region potentially further underestimating the incidence. Moreover, the ability to generalize these findings is limited by the demographics of Olmsted County, a relatively homogeneous semi-urban white population. Finally, mechanically occlusive eyelid lesions resulting in ptosis were specifically excluded from the current study. A preliminary review found that ptosis was often not coded in patients with such lesions or masses. Due to the wide variety of mechanical forms of ptosis from hemangiomas, neurofibromas, dermoid tumors, blepharochalasis, metastatic tumors, trachoma, and other causes, it was not practical to comment on their incidence in this study.

The findings of this study provide population-based prevalence and incidence rates for childhood ptosis diagnosed over a 40-year period. Simple congenital ptosis accounted for 76% of childhood ptosis, while all other forms were relatively rare. Unilateral simple congenital ptosis appears to have a predominance of left eye involvement.

Acknowledgments

This study was made possible in part by the Rochester Epidemiology Project (Grant # R01-AR30582 from the National Institute of Arthritis, Musculoskeletal and Skin Diseases) and by an unrestricted grant from Research to Prevent Blindness (RPB), Inc, New York, NY

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Table 1

Etiology and demographics of 107 patients <19 years diagnosed with childhood ptosis in Olmstead County, Minnesota from 1965 to 2004

Etiology	Number (%)	Mean Age at Diagnosis in years	Age and Sex adjusted Incidence: x/100,000 <19 years (95% CI)	Gender		Laterality		
				Male	Female	R	L	B
Congenital Ptosis								
Simple Congenital Ptosis	81 (75.7)	3.7 (0.1 - 16.7)	5.9 (4.6-7.2)	46(57%)	35(43%)	23	55	3
Blepharophimosis	3 (2.8)	1.4 (1.1 - 1.7)	0.21 (0.00-0.44)	2	1	0	1	2
Congenital CNIII Palsy	3 (2.8)	3 (0.1 - 5.1)	0.22 (0.00-0.46)	1	2	1	2	0
Marcus Gunn Jaw Wink	3 (3.8)	4.9 (0.4 - 13.8)	0.22 (0.00-0.46)	1	2	2	1	0
Congenital Horner's Syndrome	2 (1.9)	0.1 (0.1 - 0.2)	0.14 (0.00-0.33)	2	0	1	1	0
Central Core Myopathy	1 (0.93)	1.6 (1.6 - 1.6)	0.07 (0.00-0.21)	1	0	0	0	1
CFEOM	1 (0.93)	0.7 (0.7 - 0.7)	0.07 (0.00-0.21)	1	0	0	0	1
Myotonic Dystrophy	1 (0.93)	17.4 (17.4 - 17.4)	0.08 (0.00-0.24)	0	1	0	1	0
Noonan's Syndrome	1 (0.93)	5.6 (5.6 - 5.6)	0.07 (0.00-0.22)	0	1	0	0	1
Acquired								
Aponeurotic Dehiscence	4 (3.7)	17.8 (15.2 - 18.9)	0.35 (0.01-0.70)	1	3	1	3	0
Acquired CN III Palsy	2 (1.9)	16 (14 - 18)	0.17 (0.00-0.41)	1	1	1	1	0
Acquired Horner's Syndrome	2 (1.9)	5.5 (3.7 - 7.3)	0.15 (0.00-0.36)	1	1	2	0	0
Traumatic Structural Ptosis	2 (1.9)	15.2 (15 - 15.5)	0.17 (0.00-0.40)	2	0	0	2	0
Childhood Myasthenia Gravis	1 (1.9)	4.4 (4.4 - 4.4)	0.08 (0.00-0.23)	0	1	0	0	1
Total	**107**	**4.7 (.09 – 18.9)**	**7.9 (6.4-9.5)**	**59 (55%)**	**48 (45%)**	**31**	**67**	**9**

CI=confidence interval, R=right, L=left, B=bilateral, CNIII=third cranial nerve, CFEOM=congenital fibrosis of the extraocular muscles