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Refractive Errors and Strabismus in Children With Down Syndrome: A Controlled Study

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Abstract

Purpose—To evaluate the prevalence of refractive errors, strabismus, nystagmus, and congenital cataract in children with Down syndrome and control subjects of similar age.

Methods—Seventy-seven children with Down syndrome and 151 control subjects were evaluated for the prevalence of ocular findings.

Results—Ocular findings were discovered in 97.4% of children with Down syndrome and 42.4% of control subjects ($P < .0001$). The point prevalence of nystagmus, strabismus, hypermetropia, astigmatism, and congenital cataract was significantly higher in children with Down syndrome ($P < .0001$ for the first four categories, and $P < .01$ for congenital cataract).

Conclusion—Evaluation, treatment, and regular review of ocular and refractive findings in children with Down syndrome is urgently needed.

INTRODUCTION

Ocular manifestations of Down syndrome have been well described in numerous studies and include eyelid anomalies such as prominent epicanthal folds, upward slanting of the palpebral fissures, epiblepharon, nasolacrimal duct obstruction, blepharitis, keratoconus, retinal abnormalities, Brushfield spots, iris abnormalities, glaucoma, and amblyopia due to strabismus, refractive errors, and media opacities.[1–12] However, in all of these studies an appropriate control group was lacking and the authors compared their findings with previous normative studies. This is not an optimal approach because comparing groups across discrepant studies fails to take confounding factors such as sampling bias, age, and socioeconomic status into account. Only one such study was conducted in Turkey, and subsequently there is little data on ocular characteristics in Turkish children with Down syndrome. In the current study, our aim was to evaluate the ocular findings and refractive errors in children with Down syndrome and control subjects of similar age and socioeconomic background.

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PATIENTS AND METHODS

The index cases included 77 consecutively referred children with Down syndrome applying to the child psychiatry outpatient clinic of a children's hospital for eligibility certification for publicly funded special educational services. The diagnosis of Down syndrome was made through clinical and genetic findings. The control subjects consisted of 151 children of average intelligence who were also consecutively referred from the same child psychiatry outpatient clinic and were assessed for various emotional and behavioral problems at intake. The study was approved by the institutional ethics board and all participating families gave their informed consent.

All of the case and control subjects underwent standardized formal intelligence quotient (IQ) tests, which included the Stanford-Binet Intelligence Scale and the Weschler Intelligence Scale for Children-Revised, appropriate to the child's developmental functioning at the child psychiatry clinic. All of the children were also evaluated by the pediatric neurology department and underwent formal eye examinations as part of the concurrent medical evaluations. All of the control subjects recruited into the study were similarly offered formal psychological testing and were noted to have IQ scores greater than 80 and did not have any known neurological or medical disorders primarily related to ocular functioning.

The control subjects with or without known syndromic diagnoses who had IQ scores lower than 80 were excluded because they may be independently associated with ocular findings. Therefore, we chose to control for IQ at the outset.

All children underwent cycloplegic autorefraction (with cyclopentolate), retinoscopy, or both, slit-lamp biomicroscopy, and detailed dilated fundus examination. Ocular movements were checked and ocular alignment was assessed by the Hirschberg corneal reflex test, Krimsky prism test, or prism cover test. Because the distance measurement of deviation was not reliable in some patients with Down syndrome who had strabismus, the near measurements were taken into account. Visual acuity testing by Snellen chart and intraocular pressure measurement were performed only in cooperating children. Myopia is defined as spherical equivalent refraction of at least −0.50 diopter (D), hyperopia as spherical equivalent refraction of at least 2.0 D, astigmatism as cylinder of at least 1.0 D, and anisometropia as spherical equivalent difference of at least 2.0 D between the two eyes of the same child. The prevalence of refractive errors, strabismus, nystagmus, and congenital cataract were compared between the Down syndrome and control groups.

The chi-square test was used to compare categorical variables and analysis of variance was used to compare age ($P < .05$ was regarded as statistically significant). The SPSS 13.0 statistical program (SPSS Inc., Chicago, IL) was used for all analyses.

RESULTS

The cases included 77 children with Down syndrome (42 boys and 35 girls; mean age ± standard deviation = 8.5 ± 3.7 years; age range = 1 to 17 years) and the control subjects included 151 children (109 boys and 42 girls; mean age ± standard deviation = 8.9 ± 2.4 years; age range = 3 to 15 years). Although the age distribution was similar (F = 0.96; P = .

33), there was a significant preponderance of males among control subjects, reflecting the referral pattern in the child psychiatry clinic ($\chi^2 = 6.9$; $P = .01$).

All of the children in the control group had IQ scores greater than 80, whereas all of the children with Down syndrome had intellectual disability. Of the children with Down syndrome, 32% (24 of 77) had mild intellectual disability (IQ = 50 to 69), 48% (36 of 77) had moderate intellectual disability (IQ = 35 to 49), and 20% (15 of 77) had severe intellectual disability (IQ < 35). The prevalence of ocular findings was not significantly different among the intellectual disability severity groups ($\chi^2 = 0.18$ to 0.83, $P > .58$). Ocular findings were discovered in 97.4% (75 of 77) of the children with Down syndrome and 42.4% (64 of 151) of children in the control group. The total prevalence of having any ocular finding or refractive error was significantly higher among the children with Down syndrome ($\chi^2 = 66.8$; $P < .0001$).

The prevalence of nystagmus, strabismus, hypermetropia, astigmatism, and congenital cataract was significantly higher in the Down syndrome group ($P < .0001$ for nystagmus, strabismus, hypermetropia, and astigmatism, and $P < .01$ for congenital cataract), whereas the prevalence of myopia and anisometropia was similar in both groups ($P = .17$ and $P = .13$, respectively). Data regarding the prevalence of ocular findings and refractive errors in both groups are summarized in the table. Among the 25 children with Down syndrome who had strabismus, 24 had alternating esotropia and 1 had alternating exotropia. Among the 151 control subjects, 2 had strabismus, which was refractive accommodative esotropia.

The prevalence of hypermetropia was 58.3% (14 of 24) in the children with Down syndrome with esotropia, whereas it was 64.7% (33 of 51) in children with Down syndrome without strabismus. Hypermetropia was not more common in the children with Down syndrome with esotropia than in the children with Down syndrome without strabismus ($\chi^2 = 0.67$; $P = .71$). Among the 15 children with Down syndrome who had nystagmus, 13 had horizontal jerk (12 were congenital and 1 was latent) and 2 had rotatory nystagmus.

DISCUSSION

Our results have significant public health implications, especially with respect to the ophthalmic care of children with Down syndrome. First, the current study clearly shows the need to evaluate every child with Down syndrome because the point prevalence of the ocular or refractive finding in our subjects was 97.4%. Additionally, we found that nystagmus, strabismus, hypermetropia, astigmatism, and congenital cataract were significantly more common in children with Down syndrome than in control subjects, whereas the prevalence of myopia and anisometropia was similar between groups. Several of the ocular abnormalities among children with Down syndrome are amblyogenic and amenable to treatment. Therefore, early and regular assessment is likely to contribute to preventing visual loss in children with Down syndrome.

Davis reported that people with Down syndrome have characteristic ocular findings and associated problems and should have periodic ophthalmic examinations. Early recognition and treatment can minimize added problems of visual deprivation and permit children with

Down syndrome to function at an optimal level.[9] Roizen et al. evaluated the total prevalence of ocular findings in children with Down syndrome and reported that the percentage of children with ophthalmic disorders increased with age from 38% in the 2- to 12-month-old group to 80% in the 5- to 12-year-old group; the authors concluded that children with Down syndrome should be evaluated by a pediatric ophthalmologist in the first 6 months of life and undergo annual or biannual examinations thereafter.[10]

It has been shown that, relative to people with no visual impairment, individuals with bilateral mild, unilateral, or bilateral moderate or severe visual impairments report greater difficulties in performing activities of daily living in addition to having poorer mental health outcomes. Children with intellectual disability have enormous social, behavioral, and emotional difficulties. Visual impairment would be an important additional burden on these individuals and present a significant impediment to optimal functioning and quality of life.

To our knowledge, this is the first controlled study involving adult or pediatric populations in which the ocular findings and refractive errors in children with Down syndrome have been systematically compared with those in control subjects of similar age and socioeconomic background referred consecutively within the same setting. Previous studies have had serious limitations in terms of lack of appropriate control groups.[1–12] Although the current study involves clinical populations, it was conducted in a public children's hospital clinic serving a catchment population of children seeking special educational services rather than being referred for ophthalmological concerns. Therefore, eye examinations in both the case and control subjects were conducted as part of routine assessments.

Yurdakul et al. described the distribution of refractive errors and clinical characteristics of strabismus in patients with Down syndrome.[12] The authors compared refractive errors, anisometropia, and amblyopia for the patients with Down syndrome with and without strabismus. They reported a higher frequency of hypermetropia in patients with Down syndrome with strabismus. We noted a similar prevalence of hypermetropia in patients with Down syndrome with esotropia than in those without strabismus. However, according to our results, the higher prevalence of esotropia in children with Down syndrome cannot be attributed to the higher prevalence of hypermetropia observed in people with Down syndrome.

Cregg et al. investigated the development of refractive errors and strabismus in a cohort of 55 children with Down syndrome participating in a longitudinal study of visual development.[14] They reported that despite the high prevalence of large refractive errors in children with Down syndrome, these findings were not always present in early infancy. Twenty-one (38%) of the children were emmetropic throughout the study. Of the 24 children with a significant refractive error at the outset, only 6 (25%) showed emmetropization. The other children retained or increased their refractive errors. The remaining 10 children were emmetropic at the outset, but then had significant refractive errors develop. The authors found a high prevalence of strabismus in children with Down syndrome (29% of the total group) that could not be attributed to the presence of hypermetropia or anisometropia. They concluded that the retention or development of infantile refractive errors in many children with Down syndrome indicates a failure of

emmetropization and that all children were at risk of strabismus regardless of the refractive error.[14]

There were some limitations of the current study. First, the case–control study was conducted within a clinical population of children. Although the samples may not be epidemiologically based, they represent children referred for special educational eligibility assessments within the catchment base of a municipal children's hospital in a developing middle-income country. Although our results clearly may not be valid for all individuals with Down syndrome, they represent a significant picture of their clinical needs compared to control subjects of similar age and socioeconomic circumstances. It also needs to be noted that the control group was referred from the same clinical setting and may represent a higher base prevalence of ophthalmological disorders.

An overwhelming number of children with Down syndrome (97.4%) in our study had ocular findings or refractive errors, compared to 42.4% of children in the control group. The prevalence figures for nystagmus, strabismus, hypermetropia, astigmatism, and congenital cataract were all significantly higher in children with Down syndrome than in those in the control group. Therefore, the evaluation, treatment, and coordinated aftercare of ocular and refractive findings in children with Down syndrome is essential and likely to significantly enhance the quality of life of individuals with Down syndrome.

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TABLE

Prevalence of Ocular Findings and Refractive Errors in Both Groups

Ocular Findings	Down Syndrome Group (n = 77)	Control Group (n = 151)	χ^2 (df = 2)	*P*
Nystagmus	15 (19.4%)	0	32.4	< .0001
Strabismus	25 (32.5%)	2 (1.3%)	46.3	< .0001
Hypermetropia	48 (62.3%)	24 (15.9%)	56.1	< .0001
Unilateral	5 (6.4%)	6 (4%)		
Bilateral	43 (55.8%)	18 (11.9%)		
Astigmatism	46 (59.7%)	38 (25.2%)	32.7	< .0001
Unilateral	9 (12%)	17 (11.3%)		
Bilateral	37 (47.7%)	21 (13.9%)		
Myopia	6 (7.8%)	26 (17.2%)	3.5	.17
Unilateral	3 (3.9%)	12 (7.9%)		
Bilateral	3 (3.9%)	14 (9.3%)		
Anisometropia	7 (9%)	6 (4%)	2.65	.13
Congenital cataract	4 (5.1%)	0	8.2	< .01