ORIGINAL RESEARCH

Ocular findings in Down's syndrome: evaluation of 66 Hong Kong Chinese children

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Abstract

Aim: To identify the most common ocular findings in a group of children with Down's syndrome in a retrospective case-series study at a regional hospital in Hong Kong.
Patients and methods: Sixty six children with Down's syndrome aged between 2 and 18 years underwent ocular examination from June 1998 to December 2001. The presence of ocular abnormalities after examination, including visual acuity assessment, slit-lamp biomicroscopy, ocular motility, cycloplegic retinoscopy, and indirect ophthalmoscopy, were measured.
Results: Matching tests were found to be the most useful method of visual acuity assessment. Ocular abnormalities in decreasing frequency were as follows: refractive errors (85.0%), strabismus (24.0%), nystagmus (24.0%), cataract (18.0%), severe myopia (11.0%), myopic fundal changes (6.0%), myelinated nerve fiber (4.5%), and optic disc coloboma (3.0%). No patient had Brushfield spots or keratoconus. The frequency of ocular abnormalities increased with age.
Conclusions: Early diagnosis of the ocular abnormalities in patients with Down's syndrome together with the treatment of refractive errors, strabismus, cataract, and amblyopia may minimize handicaps. Therefore, referral programs should be advocated for all children with Down's syndrome and annual reassessment by ophthalmologists is recommended.

Key words: *Cataract, Down's syndrome, Matching tests, Refractive errors, Strabismus*

Introduction

Down's syndrome, first described in 1866,[1] is the most common chromosomal derangement in live births. In Hong Kong, the incidence is 1.28 per 1000 live births,[2] which is slightly higher than the 1.00 per 1000 live births reported in the USA.[3] Individuals with Down's syndrome have a higher incidence of functional and structural abnormalities of the eyes. Some ocular abnormalities such as epicanthic folds and oblique slanting of the palpebral fissures are commonly found on pediatric examination and have no functional significance.

Other anomalies such as refractive errors, congenital cataracts, nystagmus, and strabismus may have important functional and therapeutic implications. Unfortunately, these conditions are frequently missed on routine pediatric examination and not all patients with Down's syndrome are referred to an ophthalmologist for assessment. It is expected that their quality of life could be enhanced by early ophthalmological assessment and treatment.

In this study, the ocular features of 66 consecutive children with Down's syndrome were investigated from June 1998 to December 2001. The characteristic ocular abnormalities were compared with those of 3 similar studies and whether an earlier referral program should be advocated for all children with Down's syndrome is discussed.

Patients and methods

In this retrospective case series, 66 patients were examined by the same ophthalmologist at Yuen Long Yung Fung Shee Ophthalmic Center from June 1998 to December 2001.

The children were referred from various sources, including hospitals, child assessment centers, other regional eye clinics, and private practitioners. Ocular examination included visual acuity assessment, ocular motility, slit-lamp biomicroscopy, cycloplegic retinoscopy, and indirect ophthalmoscopy.

Snellen E test and the Matching tests (including Ffook's symbols, isolated E or Sheridan-Gardiner test) were the preferred methods for checking visual acuity. For those patients unable to understand, preferential looking test with the Cardiff acuity cards was used. Catford drum was used as a final option. Orthoptists or optometrists were responsible for carrying out the visual acuity assessments.

Visual acuity test results were divided into 3 categories:
- good — 6/12 or better
- fair — worse than 6/12 but equal to or better than 6/36
- poor — worse than 6/36.

For any unequal visual acuity between 2 eyes in a patient, only the better eye was included.

The patients underwent slit-lamp biomicroscopy for examination of the cornea, iris abnormalities, and the lens. Placido's disc was used for those patients suspected having keratoconus. Cataracts were defined as any opacity of the lenses involving 1 or both eyes.

Cycloplegia was attained after instillation of 1 drop of cyclopentolate 1% and phenylephrine 2.5% twice with a separation time of 5 minutes, and objective refraction was performed 45 minutes later. Emmetropia was defined as a refractive error between -0.5 diopter (D) and +0.5 D spherical equivalent. Myopia was defined as less than -0.5 D spherical equivalent, hyperopia as more than +0.5 D spherical equivalent, and astigmatism was defined as refractive error greater than 1.0 D of the cylinder. Severe myopia was defined as greater than -7.0 D of spherical equivalent, and severe astigmatism as greater than 3.0 D of cylinder.

Results

The study population were all Chinese children with Down's syndrome, with 42 boys (63.6%) and 24 girls (36.4%) examined between June 1998 and December 2001. They ranged in age from 2.0 to 18.0 years, with a mean age of 9.0 years (standard deviation [SD] ± 4.2 years).

Visual acuity

The results were analyzed according to the 3 age-groups. Group 1 consisted of patients younger than 5 years (n = 11), group 2 comprised patients aged from 5 to <12 years (n = 38), and group 3 consisted of patients aged from 12 to 18 years (n = 17).

Best corrected visual acuity was successfully evaluated in all patients by different methods, as follows: 13.6% by Catford drum, 13.6% by Cardiff acuity test, and 66.7% by Matching tests. Overall, only 4 patients (6.1%) could perform the Snellen E test, presumably because a higher level of functioning was required for this test.

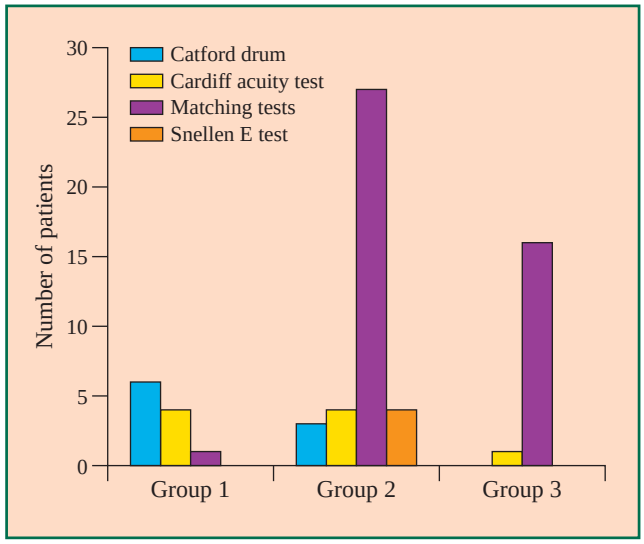

Figure 1. Methodology of visual acuity testing.

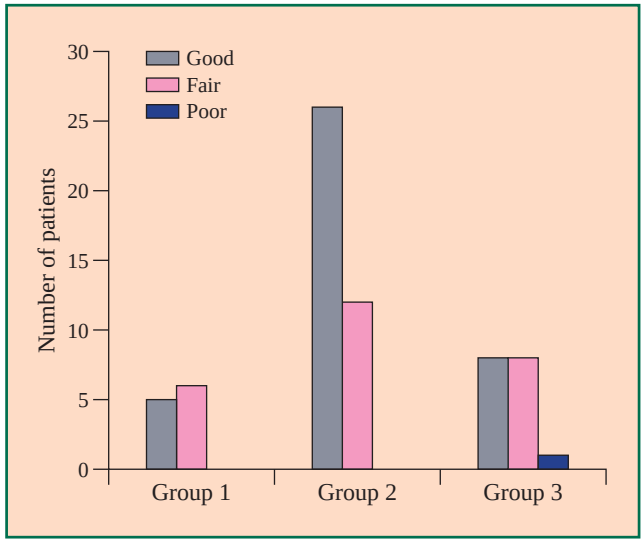

Figure 2. Distribution of visual acuity.

As shown in **Figure 1**, the Catford drum and Cardiff acuity tests were more useful for assessing children younger than 5 years (10 of 11 patients). Matching test and Snellen E test were more successful in assessment of children older than 5 years (47 of 55 patients).

As shown in **Figure 2**, the majority of patients (65 of 66) attained visual acuity better than 6/36. However, only 70% of children in group 2 and 50% in groups 1 and 3 attained visual acuity ≥6/12.

Refractive error

Cycloplegic refraction was successfully measured in 64 patients. Two patients younger than 3 years were not cooperative enough to perform refraction. Distribution of refractive errors of either eye was similar so to simplify the results, only the refractive data for the right eyes are shown in **Table 1**.

Table 1. Refractive status of 64 patients with Down's syndrome.

Refractive status	Number of eyes (%)	Spherical equivalent Mean + SD (range)
Emmetropia	10 (15)	0.125 ± 0.350 (-0.375 - +0.500)
Hyperopia	19 (30)	+2.800 ± 1.600 (+0.625 - 6.000)
Myopia	35 (55)	-5.000 ± 4.200 (-0.625 - -15.500)

Table 2. Ocular abnormalities distribution.

Ocular abnormalities	Number of patients (%)
Nystagmus	16 (24.0)
Exotropia	1 (1.5)
Esotropia	15 (23.0)
Cataract	12 (18.0)
Myopic fundi	4 (6.0)
Myelinated nerve fiber	3 (4.5)
Optic disc coloboma	2 (3.0)
Brushfield spot	0
Keratoconus	0

Myopia (55%) was the most common refractive error in the study population. Twenty percent of this group had severe myopia with spherical equivalent ≥7.0 D and all were older than 8 years. Fifty five patients (86%) had significant astigmatism with mean -3.25 ± 1.7 D (range, -1.25 to -9.5 D) and 40% had severe astigmatism >3.0 D of cylinder.

Ocular abnormalities

The frequency of ophthalmic disorders increased with age with approximately 80% of the abnormalities occurring in patients older than 8 years. The 2 most common abnormalities among the overall population were strabismus and nystagmus (24.0% of patients each) [**Table 2**]. Esotropia was the most common type of strabismus diagnosed (15 of 16 patients). Exotropia was found in only 1 patient (1.5%), but its occurrence has been reported to be as high as 6.0%.[4] No vertical deviation was found. Cataract occurred in 12 patients (18.0%) and was found only in patients older than 8 years. The most common retinal abnormality was myopic fundal change (6.0%). The least common retinal abnormalities were myelinated nerve fiber (4.5%) and optic disc coloboma (3.0%). No Brushfield spot, keratoconus, or glaucoma was found.

Discussion

Ocular abnormalities are common in association with Down's syndrome but difficulties are encountered in obtaining accurate visual acuity test results and examining ocular structures. The different criteria used in the definition of ocular abnormalities and selection of the target population made comparison between studies difficult. We studied children with Down's syndrome referred from various sources to an ophthalmic clinic for visual screening or ocular abnormality assessment. Therefore, the overall incidence of ocular abnormalities may be greater than that previously thought for the general population with Down's syndrome.

The effectiveness of visual acuity testing methods varied for different age groups, and our findings revealed that the Matching test was the most useful method for measuring visual function of children with Down's syndrome, especially those older than 5 years. However, the success rate of the Snellen E test (6%) was the lowest, which was comparable with other studies. Wagner et al recorded Snellen visual acuity in 15 of 188 patients with Down's syndrome aged between 2 and 24 years (8%),[5] and Warshowsky recorded Snellen visual acuity in only 1 of 55 patients (2%).[6]

Several studies have reported that many people with Down's syndrome have below normal best corrected visual acuity.[7,8] Our data showed a similar finding with 41% of patients attaining less than 6/12 vision in the better eye. There are several possible explanations for this poor performance. First, visual acuity could be limited by certain neural deficits and ocular abnormalities commonly found in individuals with Down's syndrome.[9] Second, Woodhouse et al indicated that most children with Down's syndrome have reduced accommodative ability.[10] This disability might reduce visual acuity for near targets (such as the Cardiff acuity test which is used at 50 cm and 1 m). Since accommodation was not evaluated in our study, it is possible that reduced accommodation contributed, at least in part, to the poor visual acuity estimates that we found. As Woodhouse et al suggested, difficulty in seeing near visual targets clearly may well contribute to the poor intellectual achievement of these children, as well as to their frequent refusal to wear optical correction prescribed for refractive error.[10]

Fifty five percent of children had a myopic refraction (mean, –5.0 D), 30% were hyperopic (mean, +2.8 D), and 15% were emmetropic (within ± 0.5 D). In a similar age group of non-disabled children, one would expect approximately 83% emmetropia, 13% myopia, and 4% hyperopia.[11] Our refractive data support previous studies that emmetropisation does not occur in patients with Down's syndrome.[12,13]

We found that 62% of the study patients needed spectacles for better vision. Early prescription may have a positive effect on their cognitive and behavioral development. In Wong and Ho's study, the overall rate of failure in refractive error test requiring corrective lens prescription was 58%.[14] **Table 3** shows the percentage of ophthalmic disorders found in our patients compared with 3 similar studies.[14-16] The majority of the results were similar except for the distribution of different types of refractive errors that might be explained by different target population selection and definition of refractive errors.

Table 3. Comparison of studies of ocular abnormalities in Down's syndrome.

Parameter	Present study	Kim et al[16]	Rosana et al[15]	Wong and Ho[14]
Year	2002	2002	1996	1997
Number of patients	66	123	152	140
Patient source	Eye clinic	Eye clinic	Eye clinic	Developmental assessment center
Age range	2 years – 18 years	6 months – 14 years	2 months – 18 years	3 months – 13 years
Refractive errors (%)	85	54	98	58
Hyperopia (%)	30	28	26	10
Myopia (%)	55	26	12	9
Astigmatism (%)	86	31	60	60
Severe myopia (%)	11	7	7	NA
Strabismus (%)	24	25	38	20
Nystagmus (%)	24	22	18	11
Cataract (%)	18	3	1	4
Brushfield's spots (%)	0	0	52	0
Keratoconus (%)	0	0	0	0

Abbreviation: NA = data not available.

In this study, the prevalence of strabismus (24%) was much higher than the prevalence of 3% reported for the general population,[17] of whom 94% were esotropic. We reported a similar incidence of strabismus and support a high incidence of esotropia in Down's syndrome when compared with other studies.[18-20] Congenital cataracts occur in 0.44% of the general population.[17] There is an increased prevalence of congenital cataracts in children and acquired cataracts in adults with Down's syndrome.[21] The observed prevalence of cataracts in our patients (18%) was higher than that of Wong and Ho's study of younger Chinese children.[14] In the present study, all cataracts occurred in patients older than 8 years. In Pueschel's study, 24% to 62% of patients with Down's syndrome had acquired cataracts after the first decade of life.[4] A maturational factor might account for the higher prevalence of lens opacities in our patients.

Brushfield's spots represent normal lighter colored stromal tissues of variable sizes situated at the junction of the middle and outer thirds of the iris. The adjacent iris is hypoplastic.[22] The prevalence of Brushfield's spots in Down's syndrome has been reported to vary between 13%[23] and 90%.[24] Similar to Wong and Ho, who assessed ocular abnormalities in 140 Chinese children with Down's syndrome,[14] our study patients exhibited no Brushfield's spots. Wong and Ho claimed that in Chinese people, the irides were much more pigmented and were dark brown or black in color.[14] This heavy pigmentation might explain the difficulty in detecting Brushfield's spots in our group of patients. The prevalence of keratoconus in Down's syndrome has been reported as being between 5.50% and 15.00% by various authors compared with that of the general population of approximately 50 per 100,000 (0.05%).[25] Keratoconus is essentially a post-pubertal disease and mostly presents between the ages of 15 and 25 years. It was absent in our patients, who might still be too young for keratoconus to develop, or the Placido's disc was not sufficiently sensitive to detect subclinical keratoconus. As the children mature, serial monitoring by slit-lamp examination and corneal topography might be helpful in detecting any genuine differences between our group and those in the other studies.

Conclusion

The high prevalence of refractive errors, strabismus, and cataracts demonstrated in our study should alert pediatricians and ophthalmologists that significant visual loss should be an avoidable event for patients with Down's syndrome because most of the eye problems are correctable.[18] The fact that the majority of patients were unable to obtain accurate visual assessment and corrective lenses from an optical shop further emphasizes this importance. Roizen et al found that 35% of children who had normal results on general pediatric physical examination had eye problems when examined by a pediatric ophthalmologist.[24]

Therefore, earlier eye referral programs should be advocated for all children with Down's syndrome. Since the occurrence of ophthalmic disorders appears to increase with age, annual reassessment by ophthalmologists is recommended.

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