Refractive errors and visual anomalies in Down syndrome

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Abstract — A comparatively high incidence of ocular and orbital abnormalities has been reported in persons with Down syndrome. Eighty six children (50% male, 50% female) with Down syndrome in several institutions for individuals with learning difficulties (age range 5-18 years, mean 12.5) were examined for visual impairment in order to relate the ocular impairment to the level of learning difficulty. 6% had mild, 7% moderate, 45% severe and 42% profound learning difficulty. 9% of the children had no refractive errors. A significant (P<0.01) positive correlation was found between progressive amounts of strabismus and ocular pathology with increasing amount of learning difficulty. On the other hand no correlation was found between refractive errors and the level of learning difficulty. Due to the significant number of ocular disorders found it is recommended that all children with Down syndrome should have an eye examination during the first six months of life and annually thereafter.

Keywords — Refractive errors, visual anomalies, Israel

Introduction

Ocular and orbital abnormalities are common features in persons with Down syndrome and several researchers have reported their findings. Wong and Ho (1997) evaluated 140 Chinese children in Hong Kong with detailed opthalmologic examination, da Cunha and Moreira (1996) examined 152 children in Brasil, Berk et al. (1996) examined 55 patients in Turkey, Castane et al. (1995) saw 25 patients in Spain, Roizen et al. (1994) evaluated 77 children in Chicago, Courage et al. (1994) tested 51 children in Canada, Hestnes et al. (1991) saw 30 patients in an institution for individuals with learning difficulties in Norway and Caputo et al. (1989) evaluated 187 medical records in New Jersey.

In the present study we would like to present ocular findings in a group of children with Down syndrome and relate the ocular defects to the level of learning difficulty.

Methods

Children with Down syndrome in several institutions for individuals with learning difficulties were examined for visual impairment by one of the authors (KK). The children had mild, moderate, severe and profound learning difficulties

Distance refractive status was determined by cycloplegic examination or by using assistants to maximize fixation on a

distant target, when consent for the use of cycloplegia could not be obtained. Opthalmoscopy was also performed.

The refraction results were divided into low myopia/hyperopia (less than or equal to 2.00 diopters), moderate (2.25-6.00) and (>6.00) high myopia. In cases of astigmatism the spherical equivalent was used and in cases of anisometropia the result was expressed on the basis of the better of the two eyes.

The Chi-squared test and Pearson correlation coefficient were used in the statistical analysis of the data.

Results

A total of 86 children (aged 5-18 years with a mean age of 12.5 years) were examined in the institutions. Sex distribution was 50% male and 50% female. Mild learning difficulty was found in 6% of the children, 7% had moderate, 45% severe and 42% profound learning difficulty. 9% of the children did not have any refractive errors.

The group with low or no refractive errors included all of the children with mild learning difficulty, 50% of those with moderate learning difficulties, 34% of those with severe learning difficulties and 50% of those with profound learning difficulties. None of the children with mild learning difficulties had moderate refractive errors, but 50% of the subjects with moderate learning difficulties had moderate refractive errors as did 38% of the children with severe learning difficulties and 42% of the children with profound learning difficulties. Twenty eight percent of those with severe learning difficulties and 8% of those with profound learning difficulties had high refractive errors while none of

Group Con	rrelation coefficient	Significance
Strabismus versus learning disability	0.988	P < 0.01
Low refractive error versus learning disability	0.818	Not significant
Moderate refractive error versus learning disal	oility 0.448	Not significant
High refractive error versus learning disability	0.789	Mot significant

Table 1. The relationship between refractive errors and visual anomalies to the level of learning difficulty in persons with Down syndrome.

the children with mild or moderate learning difficulties had this degree of refractive error.

No strabismus was found in the children with mild learning difficulties, but strabismus was found in 25% of those with moderate learning difficulty, 41% of those with severe learning difficulties and 50% of the children with profound learning difficulty. No other types of ocular pathology was found in the group of children with mild learning difficulty, but 25% with moderate learning difficulties, 34% with severe learning difficulties and 50% of the children with profound learning difficulties had other types of disorder. The following conditions were found: cataract (in 37%), corneal anomalies (in 6%), retinal anomalies (in 9%), blepharitis (in 9%), keratoconus (in 13%) and nystagmus (in 9%).

The relationship between ocular disorder and the level of learning difficulty is shown in Table 1. A significant correlation was found between greater levels of cognitive impairment and increasing amounts of strabismus and additional ocular pathology. On the other hand no correlation was found between refractive errors and the level of learning difficulty.

Discussion

The faces of persons with Down syndrome display narrow interpupillary distance, upward and outward slanting palpebral fissures and epicanthus. These clinical features were of diagnostic importance before it became possible to base the diagnosis of Down syndrome on chromosomal analysis.

Ocular and orbital abnormalities in persons with Down syndrome are common and have been reported with varying frequency in different surveys (Catalano, 1992): blepharitis (2-47%), keratoconus (5-8 %), glaucoma (less than 1%), iris anomalies (38-90%), cataract (25-85%), retinal anomalies (0-38%), optic nerve anomalies (very few cases), strabismus (23-44%), amblyopia (10-26%), nystagmus (5-30%) and refractive errors (18-58%). The earlier mentioned researchers and clinical centres for persons with Down syndrome reported on the overall occurrence of ocular problems, but we have not been able to find reports relating the ocular findings with cognitive level.

In our study we found a significant correlation between increased ocular pathology and strabismus with an increased level of learning difficulty. The results for refractive disorders did not follow such a pattern. While low refractive errors were present in the entire group of persons with mild learning difficulties they were present to the same extent in

persons with moderate and profound learning difficulties. While moderate refractive errors were absent in the group with mild learning difficulties, they were fairly evenly distributed in the other groups. The one factor that seemed to emerge was the relatively high prevalence of high refractive errors in the children with severe learning difficulties.

Conclusion

A very significant number of persons with Down syndrome have undiagnosed or underdiagnosed ocular problems that can affect their quality of life if uncorrected. It is therefore recommended that all children with Down syndrome should have an eye examination during the first six months of life and annually thereafter.

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References

Berk, A.T., Saatci, A.O., Ercal, M.D., Tunc, M. & Ergin, M. (1996). Ocular findings in 55 patients with Down's syndrome. *Opthalmic Genetics*, 17(1), 15-19.

Caputo, A.R., Wagner, R.S., Reynolds, D.R., Guo, S.Q. & Goel, A.K. (1989). Down syndrome. Clinical review of ocular features. *Clinical Pediatrics of Philadelphia*, 28(8), 355-358.

Castane, M., Peris, E. & Sanchez, E. (1995). Ocular dysfunction associated with mental handicap. *Ophthalmic Physiological Optometry*, 15(5), 489-492.

Catalano, R.A. (1992). Opthalmologic concerns. In S.M. Pueschel and J.K. Pueschel (eds), *Biomedical concerns in persons with Down syndrome*. Baltimore: Paul Brookes. pp. 59-68.

Courage, M.L., Adams, R.J., Reyno, S. & Kwa, P.G. (1994). Visual acuity in infants and children with Down syndrome. *Developmental Medicine and Child Neurology*, 36(7), 586-593.

da Cunha, R.P. & Moreira, J.B. (1996). Ocular findings in Down's syndrome. *American Journal of Ophthalmology*, 122(2), 236-244.

Hestnes, A., Sand, T. & Fostad, K. (1991). Ocular findings in Down's syndrome. *Journal of Mental Deficiency Research*, 35(3), 194-203.

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- Roizen, N.J., Mets, M.B. & Blondis, T.A. (1994). Opthalmic disorders in children with Down syndrome. *Developmental Medicine and Child Neurology*, 36(7), 594-600.
- Wong, V. & Ho, D. (1997). Ocular abnormalities in Down syndrome: An analysis of 140 Chinese children. *Pediatric Neurology*, 16(4), 311-314.