

Article ► Visual Status of Children with Down Syndrome

Robert H. Duckman, OD, MA, State University of New York, College of Optometry, New York, New York

ABSTRACT

Background: Children with Down syndrome (DS) are known to have a high prevalence of visual anomalies including strabismus, high refractive errors, Brushfield spots, nystagmus, keratoconus, and external pathologies such as blepharitis and conjunctivitis. These anomalies can impair children with DS from maximum functional capabilities.

Methods: This study looks at 42 children from Stepping Stone Day School between the ages of 2.33 years and 5.17 years. All 42 children received a comprehensive visual evaluation. Testing included visual acuity (either forced preferential looking (FPL) or Lea Picture Cards), ocular motor status (motilities and cover test/Hirschberg test), dry and damp retinoscopy, and ocular health examination with dilation.

Results: DS children showed a significantly higher prevalence than normal children of strabismus (43%), ocular-motor deficiencies (100%), decrease in visual acuity (31; 74% of the children had uncorrected visual acuity between 20/60 and <20/400), and high refractive errors (one third had hyperopia $>+1.50$, 23.81% had myopia > -1.00 , and 28.57% had astigmatism > 1.00 diopter; one third had no significant refractive error).

Conclusion: It is clear that children with DS should have a comprehensive visual evaluation as early as possible and annually thereafter.

Keywords: Down syndrome, refractive error, strabismus

Introduction

Although recognized before that time, John Langdon Down (thus Down syndrome, DS) was the first to describe the syndrome in 1866. It had previously been known for its characteristic physical signs and had been called mongoloidism. Down syndrome is the most common genetic trisomy and affects chromosome #21. This chromosomal abnormality was not identified until 1959. Many other trisomy anomalies exist, but viability of the fetus is far less likely.¹ Therefore, the trisomy we hear about most frequently is trisomy 21. Down syndrome is the most commonly identifiable cause of intellectual disability, occurring in 100% of affected children. The degree of retardation is highly variable, ranging from mild to severe.

There is a form of DS known as mosaicism which results in less severe retardation. Mosaicism is characterized by a mixture of normal cells and cells which have trisomy 21. The greater the number of normal cells, the greater the probability of higher cognitive function, although function remains adversely affected. The only way to know if there is mosaicism is through genetic testing. The incidence of Down syndrome is about 1 in 600 live births, but it increases dramatically with the age of the mother at the time of delivery:^{2,3}

- a. At 20 years = 1/2000
- b. At 30 years = 1/1000
- c. At 35 years = 3/1000
- d. At 40 years = 1/100
- e. At 45 years = 36/1000
- f. >49 years = $\frac{1}{4}$

In 1972, Lyle et al.⁴ reviewed the literature and observed that “the appearance of those with Down’s Syndrome is so characteristic that affected children are often noted to resemble each other more closely than they resemble their actual relatives.”

The physical signs of DS are very consistent and are found in Table 1.² The visual signs are listed in Table 2.⁵ Lyle, Woodruff, and Zuccaro⁴ looked at 44 patients with Down syndrome. They found that 36% had a strabismus, 56% were emmetropic (-0.25 to +2.50), 26% were mildly myopic (-0.37 to -5.87), 6% were highly myopic (-6.00 or greater), and 10% were hyperopic (+2.62 or greater).

Wesson and Maino⁵ reported that there are more hyperopes than myopes in this population, but those who are myopic tend to be more severely affected. They also noted that 49% of the children and adults had astigmatism which averaged 1.09 D across the age groups (range = -0.25 to -3.75). There was a predominance of with-the-rule cylinder in the younger children.

Woodhouse et al.⁶ looked at 92 infants and children between the ages of 4 months and 12 years. They found that the prevalence of refractive errors (including astigmatism) is higher among young children with DS than among controls. The higher prevalence of refractive defects cannot be explained by the presence of strabismus or other pathologies.

Cregg et al.⁷ examined the development of refractive error and strabismus in 55 children with DS. They found that “despite the high prevalence of large refractive errors in children with Down syndrome, longitudinal data showed that these are not always present in early infancy.” Twenty-one of their

Table 1: Physical Signs of DS

short stature
protruding tongue
intellectual disability
simian creases
hypotonia
shortened skull with occipital flattening
small underdeveloped ears
excessive skin about the neck
abnormal fingerprints
cardiac, gastrointestinal, and skeletal abnormalities

Table 2: Visual Signs of DS

short and oblique palpebral fissures with large epicanthal folds which do not resolve over time
large refractive errors
strabismus
nystagmus
cataracts
keratoconus
blepharitis
speckling of the iris (Brushfield's spots)

children were emmetropic throughout the study period. Of the 24 children with significant refractive error at the outset, only 6 (25%) showed emmetropization. The others retained or increased their refractive errors. There were 10 children who were emmetropic at the outset but who then developed significant refractive errors.

Cregg et al.⁸ examined accommodation and refractive error cross-sectionally and longitudinally in children with DS. The children they studied ranged in age from 4-85 months. They used the Nott dynamic retinoscopy technique to measure accommodative responses to stimuli. They found that accommodative responses were poor and under-accommodation is substantial even when there is no, or a fully corrected, refractive error.

Children with DS have reduced visual acuity in the absence of any clinically evident ocular condition.^{9,10} Little et al.¹¹ measured Vernier acuities and found them to be reduced. They concluded that this indicated that cortical visual function is compromised and the magnitude of the cortical deficits is significant. They state that it “should be considered along with previously reported poor optical quality.”

Roizen et al.¹² reported that the percentage of children with ophthalmic disorders increased with age, from 38% in the two- to twelve-month-old group to 80% in the five- to twelve-year group. They concluded that children with DS should receive visual evaluation before six months of age and yearly thereafter.

Haugen and Hovding¹³ examined 60 children with DS and looked at the frequency, type, age of onset, and binocular potential of strabismic patients. Subjects were examined and followed longitudinally. The mean follow-up time was 55 +/- 23 months. Hirschberg reflex test and cover test were used to assess alignment. Titmus stereo fly and Lang stereo tests were used to evaluate binocular function. Twenty-five (42%) of their 60 DS children demonstrated strabismus (21 esotropia, 2 exotropia, and 2 vertical deviations). Of the 21 subjects with esotropia, only one was infantile with the other 20 being acquired. The mean age of onset was 54 +/- 35 months. Of the acquired esotropias, 15 were associated with hyperopia. Seventeen of the strabismic patients had an “accommodation weakness.” Eleven of the strabismics gave a definite positive response to one or both stereo tests.

Stephen et al.¹⁴ recognized that these children have a higher prevalence of ocular disorders than age-matched normally developing children. He retrospectively looked at 81 children with DS. All children had a neonatal eye examination and a comprehensive eye examination (cycloplegic refraction, ophthalmoscopy, and orthoptic assessment) by at least the age of 3 years and preschool follow-up as indicated. They found that by school age, 43% of the study population had significant refractive errors, with 27% having hyperopia and astigmatism. Prevalence of strabismus was 47%, naso-lacrimal duct obstruction 36%, cataracts 7.8%, and nystagmus 16%.

Methods

This study evaluated the visual function of children with DS. The data presented are from 42 consecutive DS children who were assessed at the Stepping Stone Day School (SSDS) in Kew Gardens, New York during the 2007-2012 program years. SSDS is an early intervention/preschool facility which provides rehabilitative and educational services to children from birth to approximately five years of age. Children from birth to two and a half years receive home-based services, while those between two and a half and five years receive center-based services. The children described in this paper were all from the center-based program.

The 42 children ranged in age from 2.33 years to 5.17 years with an average age of 3.74 years. All children had been diagnosed with DS at the time of birth or in utero. Intellectual function varied from child to child, and standardized test scores were not available to the author. There were 23 (54.76%) boys and 19 (45.24%) girls. All 42 children were given a comprehensive eye examination including visual acuity; ocular-motor evaluation (ocular motility was tested with various finger puppets on a transilluminator) to assess range of movement, comitancy vs. non-comitancy, and quality of movement; retinoscopy (refractive error was determined using both dry and damp retinoscopy using 1% tropicamide 30 minutes after instillation); and ocular health assessment including a dilated fundus evaluation. Thirty-four (80.95%) of the children had no previous eye examination. Six of the eight children who had had a previous visual examination had a constant strabismus. Thirty-five of the children were tested with Teller Acuity Cards (TAC) in a forced choice preferential looking (FPL) paradigm. Seven of the children were able to respond to Lea Picture Symbol testing. Acuity, using FPL, was tested binocularly

unless there was a constant, unilateral strabismus, in which case the monocular acuity of the strabismic eye was tested first. Lea Picture Symbols acuity testing was done monocularly for each eye. It should be noted that for twelve children (28.57%), acuity values were based on the limits of the child's attention to the task rather than on threshold value (minimal acuity).

Results

Ocular Motor Status

Eighteen (42.86%) of the children had strabismus: 61% of these were esotropia and 39% were exotropia (including intermittent XT). Of the 11 children with esotropia, 100% had constant strabismus, while only two of the seven exotropes (28.28%) were constant. Twenty-four of the children (57.14%) were aligned at distance and near.

Almost all children (90.48%) had full range of movement and were comitant. Four (9.52%) of the children showed non-comitancy. All children (100%) had poor quality tracking skills as judged subjectively by the author. Observed behaviors included poor fixation, head/neck movement, frequent loss of fixation, and inattention. Saccadic eye movements were attempted, but most of the children could not understand what they had to do.

Refractive Status

Two of the children were wearing plus lens correction for accommodative esotropia. Of the 42 children: 14 children were hyperopic ($>+1.50$, 33.33%, Range $+1.75$ to $+5.50$); 10 children were myopic (>-1.00 , 23.81%, Range -1.25 to -9.00); 12 children were astigmatic (>-1.00 , 28.57%, Range -1.25 to -4.50); 14 children (33.33%) had no significant refractive error.

There was one child with a moderate anisometric refraction (3D hyperopic difference), and three were accommodative esotropes (two were already wearing correction for the accommodative strabismus). Prescription lenses were given to 28 (66.67%) of the children for full time use. Three of those children (10.71%) were given bifocal adds to achieve ocular alignment at near. On follow up it was noted that 23 (82.14%) of these children adapted nicely to their glasses. Three children (10.71%) would not leave the glasses on, one child (3.57%) had thrown them away, and one child (3.57%) never had the prescription filled.

Visual Acuity

Eleven (26%) children had normal visual acuity (20/40 or better) and 31 (74%) had uncorrected visual acuity ranging from 20/60 to $<20/400$. Corrected visual acuities improved significantly for 21 (91.3%) of the 23 children who tolerated the application of lenses. The average visual acuity improvement was 3.22 lines. Corrected acuity ranged from 20/25 – 20/200 tested binocularly with either TAC or Lea Picture Symbols.

Ocular Health Status

Ocular health was normal in 73.81% of this population. There was one case of optic atrophy (2.38%), one case of cataract (bilateral, 2.38%) and three children with nystagmus (7.14%). There were four children with blepharitis (9.52%) and two with conjunctivitis (4.76%). Therefore, there was ocular pathology in 26.19% of the children. All active pathologies were referred to local eye care professionals because the author's visits were two months apart and follow-up care could not be rendered.

Discussion

Children with DS have been shown to have a higher prevalence of ocular and visual problems than their age-matched controls.⁴⁻¹⁴ In the young DS child, there is a high prevalence of strabismus, ocular motor dysfunction, refractive error, reduced visual acuity (uncorrected), and ocular pathology. Cregg et al⁸ studied refraction and accommodation in DS children. They used the Nott dynamic retinoscopy technique to measure accommodative responses to stimuli. They found that accommodative responses were poor and under-accommodation is substantial even when there is no, or a fully corrected, refractive error. Reduced accommodative function in DS children has been treated successfully with bifocal lenses. Nandakumar and Leat¹⁵ did a longitudinal study on 14 DS children, all of whom had reduced accommodative function both before and after single vision lenses were prescribed. They demonstrated that the application of distance refractive correction significantly improved near visual acuity. They concluded that all subjects might benefit from bifocal lenses. Al-Bagdady et al.¹⁶ concluded that a large number (62.16%) of their DS patients who wore bifocal lenses improved in visual acuity both at distance and near after using these lenses. Nandakumar¹⁷ found that the use of single vision lenses did not necessarily improve distance acuity, but that near visual acuity improved significantly. Al-Bagdady et al.¹⁸ showed that with the application of bifocals, accommodative function improved for 65% of their DS patients. They further noted that accommodative responses did not improve with age before the children began wearing bifocal lenses. In addition, accurate accommodation was sustained after the children returned to single vision lenses. Woodhouse¹⁹ found that about three quarters of her DS children consistently under-accommodated and that this response continues even when the hypermetropia is corrected. She states that "bifocals are successful in improving the accommodative response and rendering near work in focus."

Conclusion

With the high prevalence of visual problems in children with DS, early optometric evaluation is important for identification and early intervention of these anomalies. Thereafter, vision should be assessed annually at a minimum.

References

1. Gaynon MW, Schimek RA. Down's syndrome: A ten-year group study. *Annals Ophthalmol* 1977;9:1493-7.
2. Batshaw ML, Perret YM. *Children with Disabilities: A Medical Primer*, 3rd Ed, Baltimore: Paul H. Brookes Publishing Co., 1992:271-8.
3. Catalano RA. Down syndrome. *Surv Ophthalmol* 1990;34(5):385-98.
4. Lyle WM, Woodruff ME, Zuccaro VS. A review of the literature on Down's syndrome and an optometrical survey of 4 patients with the syndrome. *Am J Optom Arch Am Acad Optom* 1972;49:715-27.
5. Wesson M, Maino DM. Oculo-Visual Findings in Down Syndrome, Cerebral Palsy, and Mental Retardation with Non-Specific Etiology. In: Maino D (ed). *Diagnosis and Management of Special Populations*, St. Louis: Mosby-Yearbook, Inc., 1995:17-54.
6. Woodhouse JM, Pakeman VH, Clegg M, Saunders KJ, et al. Refractive errors in young children with Down syndrome. *Optom Vis Sci* 1997;74(10):845-51.
7. Clegg M, Woodhouse MW, Stewart RE, Pakeman V, et al. Development of refractive error and strabismus in children with Down syndrome. *Inv Ophthalmol Vis Sci* 2003;44:1023-30.
8. Clegg M, Woodhouse JM, Pakeman VH, Saunders KJ. "Accommodation and refractive error in children with Down syndrome: Cross-sectional and longitudinal studies. *Invest Ophthalmol Vis Sci* 2001;42:55-63.
9. Courage ML, Adams RJ, Reyno S, Kwa PG. Visual acuity in infants and children with Down syndrome. *Dev Med Child Neurol* 1994;36:586-93.
10. Woodhouse JM, Pakeman VH, Saunders KJ, Parker M, et al. Visual acuity and accommodation in infants and young children with Down syndrome. *J Intellect Disability Res* 1996;40(pt 1):49-55.
11. Little J-A, Woodhouse JM, Lauritzen JS, Saunders KJ. Vernier acuity in Down syndrome. *Invest Ophthalmol Vis Sci* 2009;50:567-72.
12. Roizen NJ, Mets MB, Thomas AB. Ophthalmic disorders in children with Down syndrome. *Devel Med Child Neurol* 1994;36:594-600.
13. Haugen OH, Hovding G. Strabismus and binocular function in children with Down syndrome. A population-based, longitudinal study. *Acta Ophthalmologica Scand* 2001;79:133-9.
14. Stephen E, Dickson J, Kindley AD, Scott CC, Charleton PM. Surveillance of vision and ocular disorders in children with Down syndrome. *Devel Med Child Neur* 2007;49:513-5.
15. Nandakumar K, Leat SJ. Bifocals in Down syndrome study (BiDS): Design and baseline visual function. *Optom Vis Sci* 2009;86:196-207.
16. Al-Bagdady M, Stewart RE, Woodhouse JM. The success rate of wearing bifocals in children with Down's syndrome. *Ophthal Physiol Opt* 2008;28:101-5.
17. Nandakumar K. Bifocals in children with Down syndrome (BIDS). University of Waterloo. <http://hdl.handle.net/10012/5316>. Last Accessed June 1, 2014.
18. Al-Bagdady M, Stewart RE, Watts P, Murphy PJ, Woodhouse JM. Bifocals and Down's syndrome: Correction or treatment? *Ophthal and Physiol Opt* 2009;29:416-21.
19. Woodhouse JM. Accommodation in Down syndrome – More questions than answers. *J Vis* 2010;10(15): doi:10.1167/10.15.34.

Correspondence regarding this article should be emailed to Robert H. Duckman, OD, MA at rduckman@sunyopt.edu. All statements are the author's personal opinions and may not reflect the opinions of the representative organizations, ACBO or OEPF, Optometry & Visual Performance, or any institution or organization with which the author may be affiliated. Permission to use reprints of this article must be obtained from the editor. Copyright 2014 Optometric Extension Program Foundation. Online access is available at www.acbo.org.au, www.oepf.org, and www.ovpjjournal.org.

Duckman RH. Visual status of children with Down syndrome. *Optom Vis Perf* 2014;2(5):240-3.

The online version of this article contains digital enhancements.

Wolff Wands are available once again!

The Wolff Wands were developed by Dr. Bruce Wolff to provide suitable fixation targets for tracking and locating activities in visual testing and training. They were quickly adopted, and became the standard, for various retinoscopy and convergence testing procedures. They have also been adapted to other procedures requiring a fixation target. The lightweight aluminum rods allow the examiner/therapist to comfortably move and position the targets throughout the volume of the patient's near visual space. The use of shiny gold and silver colored surfaces minimizes the differences in accommodative demand created by some color pairs. The minified, reflected image of the patient from the highly polished surface provides an additional level of detail for fixation and accommodation.



The OEP Wolff Wand Fixation Targets duplicate the quality and durability of the original Wolff Wands. Each wand consists of a 1/2 inch (13mm) target mounted on a 14 inch (35 cm) aluminum wand. A set consists of a pair of wands - one with a highly polished, brass target and one with a silvery, nickel-plated target. The set of Wolff Wands comes packaged in a protective, black cloth bag for storage and transport. Wolff Wands may be purchased alone or with the Eye Tracking and Locating Skills manual.

- Wolff Wands (pair) **XBW100** \$24.00
- Wolff Wands (pair) with Eye Tracking and Locating Skills Manual **XBW200** \$28.00