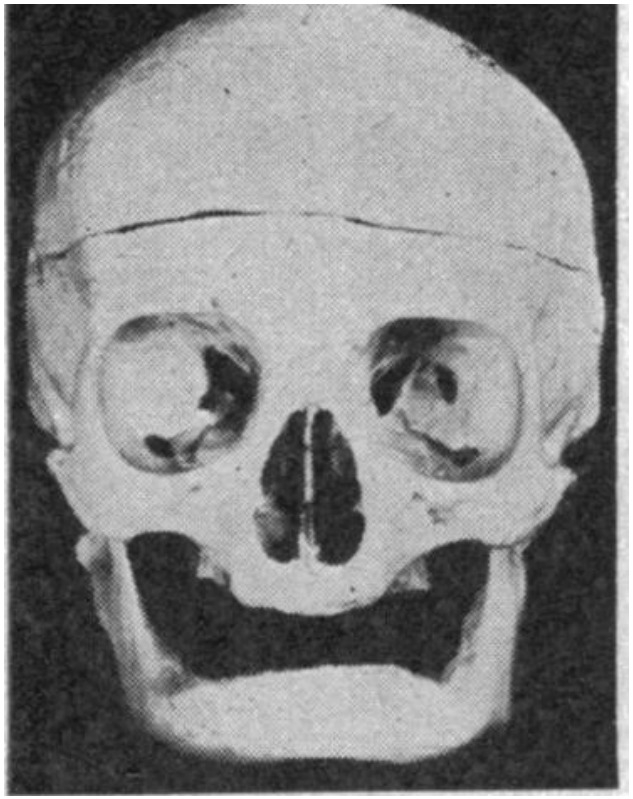
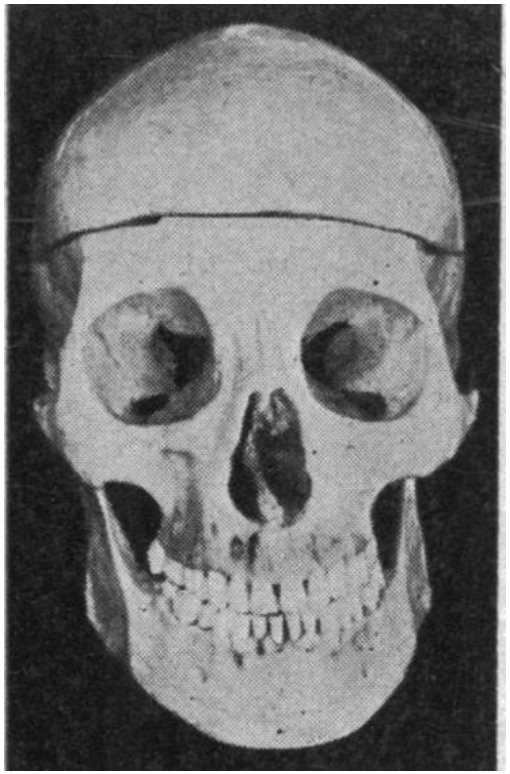
Down Syndrome (Trisomy 21) – Ophthalmic Overview

Shivanand Sheth



Down's syndrome Adult Skull



Normal Adult Skull

*The eyes in Mongolism – Ronald Lowe, BJO, March 1949

Ophthalmic abnormalities Not affecting vision

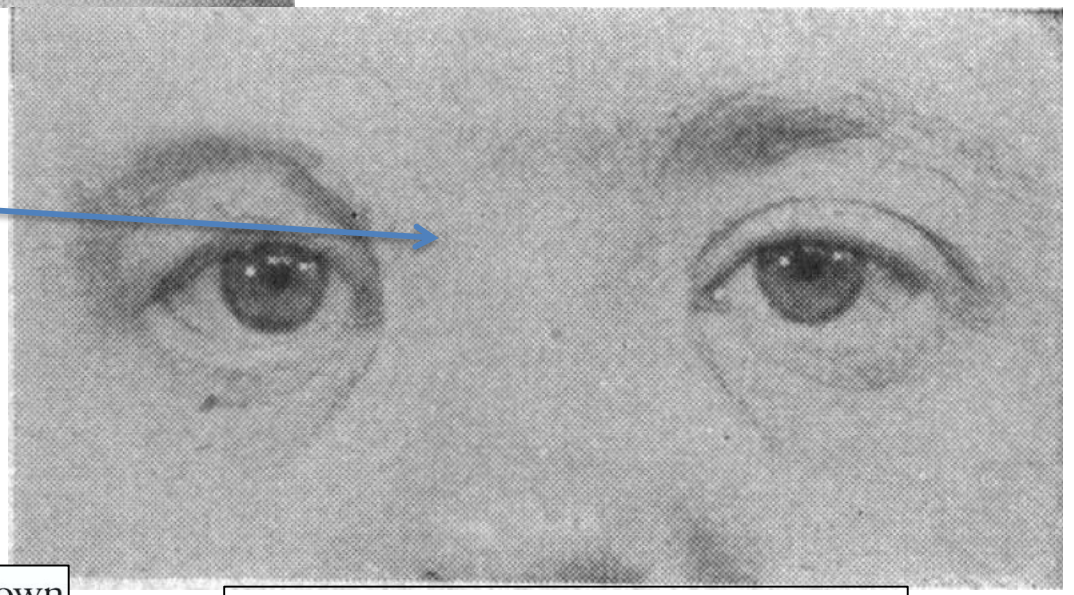
Up-Slanting Palpebral fissures

Lateral Canthus higher than Medial Canthus

Prevalence 60 - 80%

Wide - epicanthal folds

Prevalence: > 60%



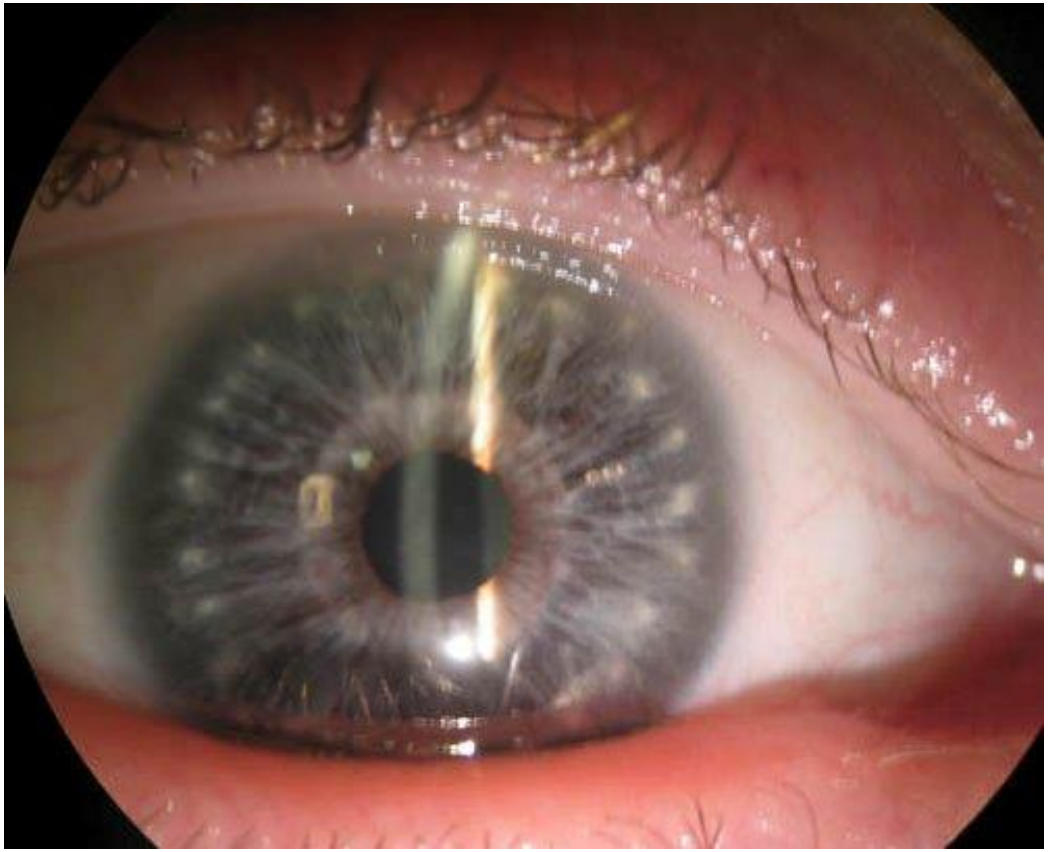
Ophthalmic Abnormalities in Children With Down Syndrome

Alexandra L. Creavin; Ray D. Brown, FRCOphth

***The eyes in Mongolism – Ronald Lowe, BJO, March 1949**

Ophthalmic abnormalities Not affecting vision

- Brushfield spots on iris
- Prevalence: 0 – 80%



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Ophthalmic abnormalities affecting Vision

- **Structural:**
  - Lens: Cataracts (15-37%)
  - Lacrimal duct obstructions (17- 36%)
  - Cornea: Keratoconus (12%)
  - Optic nerve abnormalities (3%)
  - Retinal abnormalities (< 10%)

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Ocular Findings in Down's Syndrome

ROSANA PIRES DA CUNHA, M.D., AND JOSÉ BELMIRO DE CASTRO MOREIRA, M.D.

Ophthalmic abnormalities affecting Vision

- **Strabismus:**

Author	Year	Population	No. patients	Age	Eso-deviations	Exo-deviations	Vertical deviations
Lowe	1949	selected	67	5–60 years	33%	0	
Skeller & Øster	1951	selected	81	<6–58 years	31%	4%	1%
Hiles et al.	1974	selected	123	"children"	28%	6%	
Rochels et al.	1977	selected	1047	1 month–23 years	70%	0	
Jaeger	1980	selected	75	15–64 years	37%	3%	1%
Caputo et al.	1989	selected	187	3.5 months–26 years	52%	2%	3%
Hestnes et al.	1991	selected	26	21–72 years	65%	4%	
Wesson & Maino	1995	selected	134	average age 56 months	34%	4%	
Shapiro & France	1985	unselected	53	7–36 years	42%	2%	
Riise	1986	unselected	123	2 weeks–66 years	40%	4%	
Berk et al.	1996	unselected	55	2 months–25 years	20%	2%	
Roizen et al.	1996	unselected	77	2 months–19 years	26%	1%	
da Cunha & Moreira	1996	unselected	152	2 months–18 years	34%	1%	3%
Woodhouse et al.	1997	unselected	92	3 months–12 years	19%	0	
Present study	2001	unselected	60	2–12 years	35%	3%	3%

- **20 – 70% of Down's Syndrome have Strabimus with Esodeviations being much more common than exodeviations**
- **Nystagmus:** 10 -20 % of Down's Syndrome have Nystagmus

Strabismus and binocular function in children with Down syndrome. A population-based, longitudinal study

Olav H. Haugen and Gunnar Høvding
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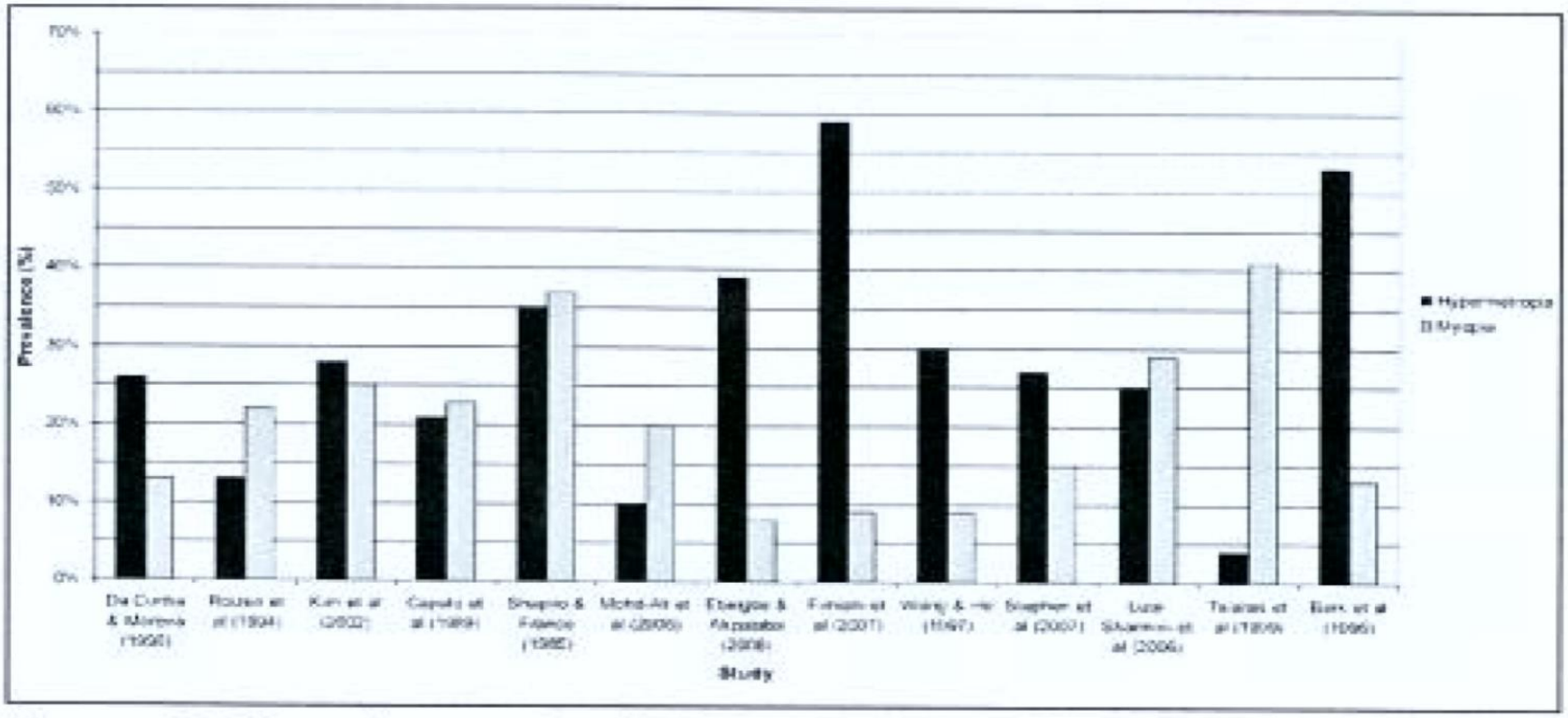
Ophthalmic Abnormalities in Children With Down Syndrome

Alexandra L. Creavin; Ray D. Brown, FRCOphth

Ophthalmic abnormalities affecting Vision

- **Optical: Refractive errors**
  - Refractive errors are common in Down's (more than general population)
  - Overall hyperopia more common than myopia in most studies
  - Astigmatism more common too



Ophthalmic abnormalities affecting Vision

- **Optical: Accommodation –** Excellent articles and research on accommodation in Down's Syndrome

Association Between Accommodative Accuracy, Hypermetropia, and Strabismus in Children with Down's Syndrome

RUTH E. STEWART, PhD, BSc, MOptom, J. MARGARET WOODHOUSE, PhD, BSc, FSMC, MARY CREGG, PhD, DipOptom, and VALERIE H. PAKEMAN, BSc, MOptom

School of Optometry & Vision Sciences, Cardiff University, Cardiff, United Kingdom

Optometry and Visual science, Feb 2007

Reduced Accommodation in Children With Down Syndrome

J. Margaret Woodhouse, Jennifer S. Meades, Susan J. Leat, and Kathryn J. Saunders

Investigative ophthalmology and Visual science, 1993

Accommodation and Refractive Error in Children with Down Syndrome: Cross-Sectional and Longitudinal Studies

Mary Cregg,[1,2] J. Margaret Woodhouse,[1] Valerie H. Pakeman,[1] Kathryn J. Saunders,[1,3] Helen L. Gunter,[1,4] Margaret Parker,[1] William I. Fraser,[5] and Prema Sastry[6]

Investigative ophthalmology and Visual science, 2001

Accommodation and Refractive Error in Children with Down Syndrome: Cross-Sectional and Longitudinal Studies

Mary Cregg,[1,2] J. Margaret Woodhouse,[1] Valerie H. Pakeman,[1] Kathryn J. Saunders,[1,3] Helen L. Gunter,[1,4] Margaret Parker,[1] William I. Fraser,[5] and Prema Sastry[6]

Investigative Ophthalmology & Visual Science, January 2001, Vol. 42, No. 1

- Examines the relationship between defective accommodation and refractive errors in children (with and without spectacle correction) with Down's Syndrome

Studies have shown:

- Many children with Down's syndrome have a *lag of accommodation* at all distances tested

- Accommodation is influenced by refractive errors, which have a higher prevalence in Down's

- Hypermetropia + Down's + Accommodative Lag = Higher degree of Hypermetropia

1. Woodhouse JM, Cregg M, Gunter HL, et al. The effect of age, size of target and cognitive factors on accommodative responses of children with Down syndrome. *Invest Ophthalmol Vis Sci*. 2000; 41:2479–2485.
2. Woodhouse JM, Pakeman VH, Saunders KJ, et al. Visual acuity and accommodation in infants and young children with Down syndrome. *J Int Dis Res*. 1996;40:49–55.
3. Woodhouse JM, Meades JS, Leat SJ, Saunders KJ. Reduced accommodation in children with Down syndrome. *Invest Ophthalmol Vis Sci*. 1993;34:2382–2387.
4. Woodhouse JM, Pakeman VH, Cregg M, et al. Refractive errors in young children with Down syndrome. *Optom Vis Sci*. 1997;74: 844–851.

Methods:
Subjects Chosen

- Cross Sectional data: 75 children with Down's
- Longitudinal data: 69 children with Down's
- Age range : 4 - 85 months

(Mean age: 42.7 ± 23.4 months)

- Control data of 121 normal kids aged 1 to 45 months
- The longitudinal data from the study was compared to the accommodative response by a 36 year old emmetropic adult.

Methods: Procedures

- Refractive error assessed by Mohindra Retinoscopy technique and Cycloplegic retinoscopy

- Accomodation assessed by Nott Dynamic Retinoscopy technique at 10 cm (10 D), 16.6 cm (6 D) and 25 cm (4 D)

Methods: Procedures

- Nott's dynamic retinoscopy – The accommodative target was presented at 3 distances (10, 16.6 and 25 cm) and the retinoscope was moved towards or away from the eye till neutrality was observed – Dioptric equivalent of the distance was the "Accommodative response"

- Mohindra Retinoscopy: Dry Retinoscopy done in dark room – considered equal to cycloplegic retinoscopy as patient doesn't accommodate in dark.

RESULTS: Relation Between Accommodation & Refractive error

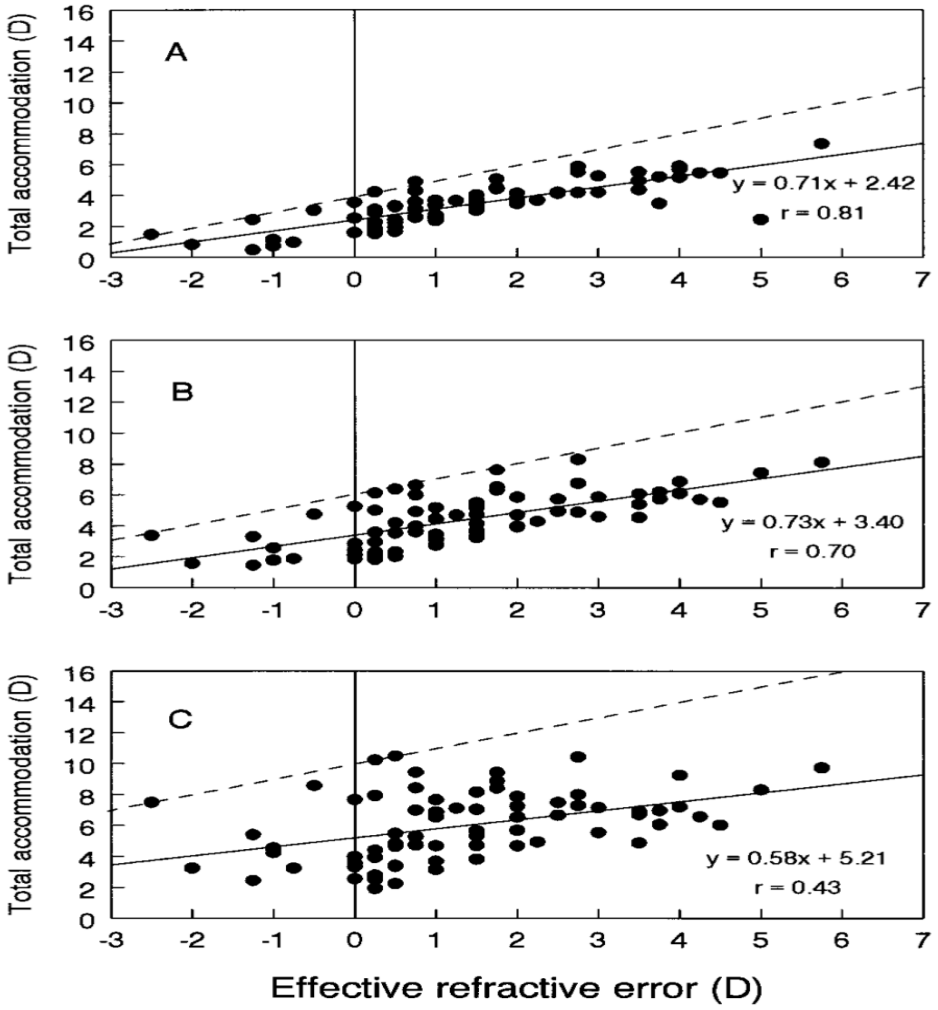


FIGURE 1. Total accommodation and effective refractive error in 75 children with Down syndrome, at three target distances: (A) 25 cm (4.00 D), (B) 16.6 cm (6.00 D), (C) 10 cm (10.00 D). The least-squares regression line through the data are shown in each case (*solid line*) with its equation and correlation coefficient. In each figure the *dashed line* represents accurate accommodation to the target.

Effective refractive error Range:
-2.59 D to +5.75 D
(Mean +1.35 ± 1.64)

Astigmatism range:
0.00 to 3.00 D

RESULTS: Relation Between Accommodation & Refractive error

- The total accommodation produced was dependent on the target distance
- ***The discrepancy between demand and response increased as the target distance decreased***

RESULTS: Effect of Spectacle Correction

TABLE 1. AEI for Children with Down Syndrome

	n	Effective Refractive Error	AEI
Emmetropes	41	+1.20 ± 0.89	3.07 ± 1.42 (0.00–5.29)
Myopes (no spectacles)	6	−1.50 ± 0.61	2.63 ± 1.52 (0.00–4.34)
Myopes (with spectacles)	4	+0.56 ± 0.66	4.77 ± 0.66 (3.82–5.27)
Hypermetropes (no spectacles)	14	+3.96 ± 0.75	4.78 ± 0.78 (3.50–6.24)
Hypermetropes (with spectacles)	10	+0.50 ± 0.61	4.93 ± 0.90 (3.55–6.68)

Children were divided into groups according to full refractive error and spectacle wear. Median AEI for normally developing children is 0.00 and 95% have AEI between 0.00 and 2.20D.[1] Data are in mean diopters ± SD. AEI range is in parentheses.

Patients categorized into 4 groups:

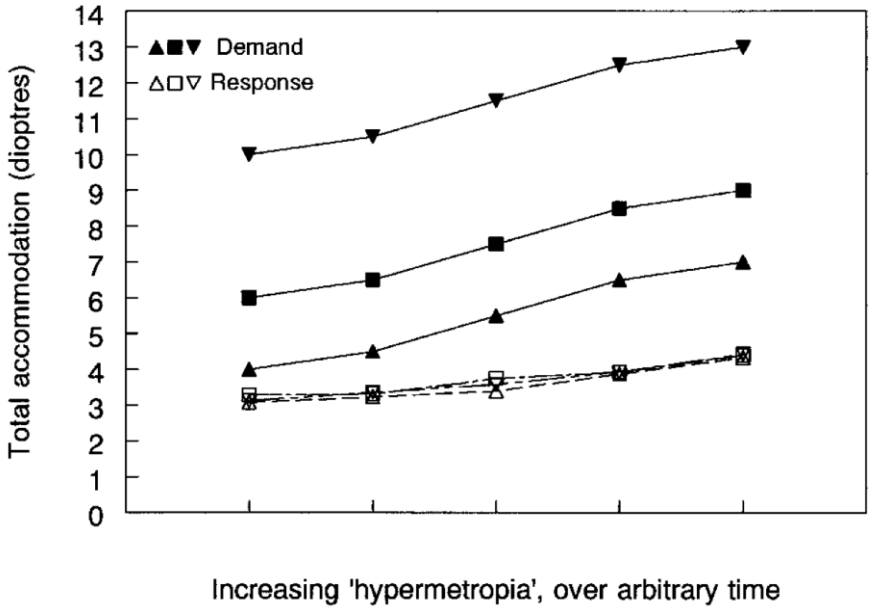
- Myopia > -0.75 D
- Emmetropia
- -0.75 to +2.75 D
- Hypermteropia > +3.00D

15. Saunders KJ. Early refractive development in humans. *Surv Ophthalmol*. 1995;40:207–216.
16. Ingram RM, Walker C, Wilson JM, Arnold PE, Dally S. Prediction of amblyopia and squint by means of refraction at age 1 year. *Br J Ophthalmol*. 1986;70:12–15.

RESULTS: Effect of Spectacle Correction

- Emmetropes: Better response than Hypermetropes (both corrected and uncorrected) ($P=0.049$ and $P<0.001$)
- Emmetropes: Better response than corrected myopes. ($P= 0.049$)
- Uncorrected Myopes: Better response than Hypermetropes (both corrected and uncorrected) ($P= 0.002$ and $P= 0.003$)
- NO significant difference between Corrected and Uncorrected Hypermetropes ($P = 0.991$)

Results: Longitudinal Changes in refractive error: Presbyopic Adult



- Compared to Presbyopic adult:
  - Tested at 3 distances
  - Tested by simulating increasing hypermetropia with Minus lenses
- The curves of the accommodative response of the adult for the 3 distances superimposed – indicating that the normal presbyopic adult can accommodate to a maximum amplitude required at all target distances, even when hypermetropic refractive error was increased

Results: Longitudinal Changes in refractive error: Down's Syndrome

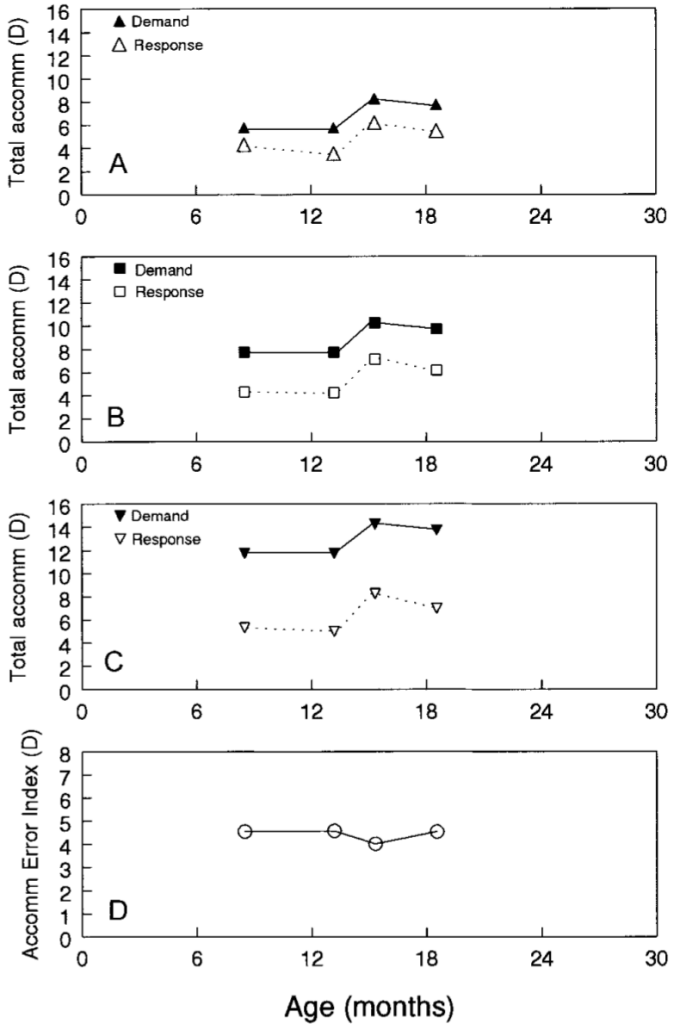


FIGURE 3. Total accommodative demand (*filled symbols*) and response (*open symbols*) in one child with Down syndrome whose refractive error changed over time, with stimulus at (**A**) 4.00 D (*triangles*); (**B**) at 6.00 D (*squares*); (**C**) at 10.00 D (*inverted triangles*). (**D**) AEI.

+1.75 D at 8.5 months

+3.75 at 18.5 months

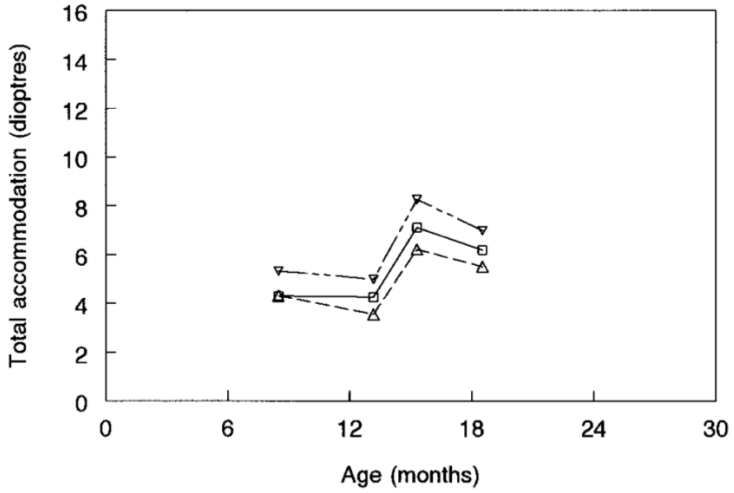


FIGURE 4. Total accommodative response, from Figure 3A through 3C, plotted on a single figure. Stimulus at 4.00 D (△); 6.00 D (□); and 10.00 D (▽).

Results: Longitudinal Changes in refractive error: Down's Syndrome

- With increasing Accommodative demand the actual accomodative response changed in accordance to the increasing effective refractive error, while maintaining a constant AEI over the age range

- The curves of the Total accommodative response did not superimpose unlike the presbyopic control

Results: Effect of Spectacles in Hypermetropes: Subject A

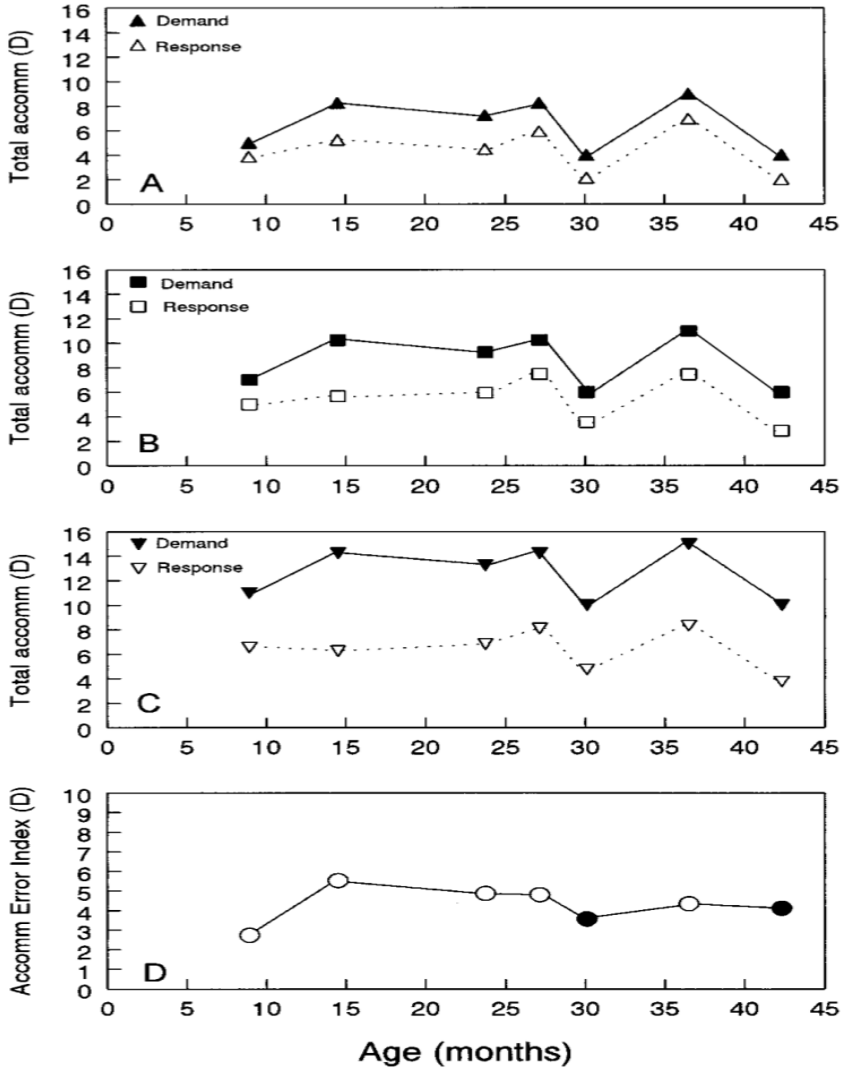


FIGURE 5. (A, B, and C) Total accommodative demand (*filled symbols*) and response (*open symbols*) in one child with Down syndrome wearing spectacles to correct hypermetropia during visits at 30 months and 42 months. (A) Stimulus at 4.00 D (*triangles*); (B) at 6.00 D (*squares*); (C) at 10.00 D (*inverted triangles*). (D) AEI without (*open symbols*) and with (*filled symbols*) spectacles.

- Accomodative response were noted for atleast 3 visits without spectacles and then with Spectacle during 2 occasions: 30 months and 42 months in this example child
- Result: Child maintained same level of underaccommodation with and without spectacles

Results: Effect of Spectacles

TABLE 2. AEI and Effective Refractive Error for Children with and without Spectacles

Subject	Age Interval (mo)	AEI (D)		Effective Refractive Error (D)	
		Without	With	Without	With
A	27.14–29.96	4.81	3.58	+4.25	0.00
B	36.67–52.07	4.09	3.96	+4.50	+0.75
C	60.35–65.87	5.38	4.98	+5.00	+0.50
D	22.30–48.53	3.76	4.41	+4.50	+1.00
E	45.80–78.06	6.28	4.97	+6.00	0.00
F	23.95–36.44	5.21	5.07	+4.75	+1.25
G	12.20–39.69	4.47	5.15	+4.25	0.00
I	43.37–55.36	5.74	4.09	+5.50	+0.50
H	46.00–54.54	4.05	4.89	+5.00	+1.00
J	16.30–41.59	5.60	3.37	+4.00	0.00
K	48.50–66.73	3.62	4.41	−2.50	0.00
L	34.00–53.98	3.01	5.68	0.00	+2.00
M	35.00–42.25	0.73	3.56	−6.50	−0.50

The AEI is given for the visit immediately before spectacle wear commenced (without) and for the latest visit at which spectacles prescribed during the study were worn (with). The age interval column gives the age of each child at the two relevant visits. Data for subject A are shown in Figure 5, for subjects B through J in Figure 6, and for subjects K, L, and M in Figure 7.

Summary of the study

- Not only Hypermetropic children, but Down's children with all types and amounts of refractive error demonstrate underaccommodation, even emmotropes
- Increased under-accommodation seen with increased accommodative demand
- Saturation of accommodation was not demonstrated in Down's (maximum accommodation was not ascertained)
- Down's Syndrome don't behave like adult presbyopes

Implications of study

- Children with Down's syndrome are constantly in state of blur for near distances – can affect normal development and contribute to overall developmental delay

Possible theories to explain underaccommodation

- Anomalous set point of accommodation (predetermined amount of underaccommodation for given accomodative demand)
- Large blur tolerance (retinal or cortical?)
- Abnormal convergence driven-accommodation?
- Increased depth of focus (but no discrepancy in pupil size noted compared to normal children)

Limitations of the Study

- No control data of normal accommodative response in normal kids with and without glasses included in this study – (Done in a previous study which they have referred to)
- Compared longitudinal data with single control adult presbyopic of 36 years – maybe too young – could have compared with range of presbyopic patients
- Tested only one eye – other eye might have driven total accommodative response
- Tested astigmatism along only more myopic meridian – can single meridian testing be reflective of overall accommodative response?

Merits of Study

- Great study! – Makes us aware of the need to consider testing and correcting near vision/ prescribe bifocals in all Down's Children

- Throws up potential for studying near visual acuity in Children with Down's with and without corrections

- Normal kids have decreasing hyperopia, down's children have increasing hyperopia - Maybe due to under accomodation? Can give an insight into role of accommodation in the emmetropization process.

Ophthalmic abnormalities affecting Vision

- **Optical: Accommodation –** Summary of findings:
  - Many children with Down's syndrome have a lag of accommodation at all distances tested.
  - Accommodation is Influenced by refractive errors, which have a higher prevalence in Down's
  - Not only Hypermetropic children, but children with all types and amounts of refractive error demonstrate underaccommodation, even emmotropes
  - Increased under-accommodation seen with increased accommodative demand
  - Down's Syndrome don't behave like adult presbyopes

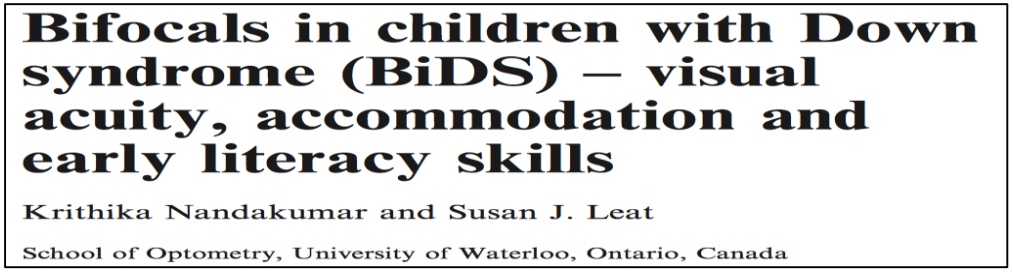
Ophthalmic abnormalities affecting Vision

- **Optical: Accommodation – Implications and Recommendation**

- Children with Down's syndrome are constantly in state of blur for near distances – can affect normal development and contribute to overall developmental delay

- Prescribing Bifocals to all children with hypermetropia or even emmetropia is a rational solution

- Leave myopic Down's undercorrected – full correction can induce accommodation lag at all distances



Bifocals in children with Down syndrome (BiDS) – visual acuity, accommodation and early literacy skills

Krithika Nandakumar and Susan J. Leat

School of Optometry, University of Waterloo, Ontario, Canada

Summary

- Down's Syndrome have multiple ocular issues which can cause poor visual function
- Apart from un-treatable structural abnormalities, many other issues can be treated effectively.
- Need good optical correction and refraction keeping in mind they do not have normal accommodation. Bifocals can be precribed with a lower threshold even in kids.
- Strabismus can be treated with surgery – No difference in surgical results compared to normal subjects *

Outcomes of Strabismus Surgery for Esotropia in Children With Down Syndrome Compared With Matched Controls

W. Walker Motley, III, MS, MD; Andrew T. Melson, BA; Michael E. Gray, MD; Shelia R. Salisbury, PhD

Clinical Implications

- Very often, Children with Down's syndrome are not 6/6, even if they are emmetropic on Retinoscopy
- We do not focus on issues of near vision and give only distance prescription.
- Need to have lesser threshold for prescribing Bifocals in Down's Syndrome to account for increased underaccommodation for near, especially if they have poor near vision with glasses.
- Keep option of prescribing readers to even an emmetropic Down's Child.
- Do not fully correct myopic Down's, as some myopia gives them clear vision at near.
- Fully/over correcting myopia will induce under-accommodation causing blur at all distances.

- Thank You