

BASIC MEDICAL SURVEILLANCE ESSENTIALS FOR PEOPLE WITH DOWN SYNDROME

OPHTHALMIC PROBLEMS – BACKGROUND NOTES

These background notes should be read in conjunction with the DSMIG surveillance guideline on ophthalmic problems. To avoid confusion references which appear here and in the guideline are numbered as in the guideline. Numbers for new references relevant to these background notes start at 30.

Point 1. Gives fully referenced background information about the nature and prevalence of ocular disorder in this population.

Point 2. With reference to the need for examination for cataract both in the newborn period and at age 6 weeks we follow recommendations for all children made by the Children's Subgroup of the National Screening committee (NSC) of the UK in 2000 ⁽¹⁰⁾ and Hall ⁽³⁰⁾ The NSC guideline was reviewed in 2009 (www.nsc.nhs.uk) and no changes were recommended. We note that the effectiveness of this 2 stage approach has not been fully evaluated ⁽¹⁰⁾. However it is salutary to read that previously, at a national level, a study by the British Congenital Cataract Interest Group found that despite specific guidance at that time less than half of newly diagnosed cataracts had been detected at the newborn and 6 week examinations. ⁽³¹⁾

In response to this finding the same group addressed the training needs for those carrying out these early ophthalmic examinations ⁽³²⁾. Hence we have stressed that for this high risk group of babies with Down syndrome it is essential that these examinations are undertaken by a 'trained person'.

Point 3. This good practice point is not evidence based but by consensus we have stressed the need for ongoing paediatric monitoring of gaze, visual behaviour, and attention in the first 18 months. This is because of the risk of cataract having been missed in the first weeks of life, because of ongoing low risk of developing infantile glaucoma ^(7,35) and because of the high risk of other ocular abnormalities.(see guideline. point 1).

We did not consider that formal ophthalmic screening was necessary until age 18 months because the possibility of significant ophthalmic abnormality in the intervening months should be clinically obvious to an experienced paediatrician who can then refer for expert ophthalmologic review. It is however essential that this need for paediatric monitoring is included in local review protocols covering the first 18 months of life.

Point 4. It is from the age of 18 months that our recommendations differ from those for the general population as set out in the National Screening Guideline ^(10, 30). For these children Hall et al ⁽³⁰⁾ do not recommend vision screening before school age but they state that 'all children with dysmorphic syndromes or neurodevelopmental problems should undergo a specialist eye examination as some may have serious defects of vision'. We have recommended two such examinations, one at 18 months – 2 years and another at age 4 years for the following reasons.

Investigations by Woodhouse et al ^(4,5), Haugen et al ⁽²⁾ and Courage et al ⁽¹⁸⁾ have all revealed very distinct, complex and aberrant visual development in children with Down syndrome

from the early months of life until school age. Furthermore Roizen et al³⁷ in a comparison study demonstrated that whilst a developmental paediatrician found abnormalities of vision in 40% of a group of children with Down syndrome a paediatric ophthalmologist identified a further 21% with problems, thus supporting a need for specialist examination.

In typically developing children, distribution of refraction is wide at birth and narrows over the first few years of life so that by school age very few children have a significant refractive error; this is the process of emmetropisation. However development in this respect is different for children with Down syndrome. For these children, although the distribution of refraction in early infancy is similar to that in typically developing children, longitudinal studies of refraction have shown that the distribution widens over time, rather than narrows, and that failure of emmetropisation is common. Data from Woodhouse and colleagues⁽³⁾ shows that only 25% of those who had significant refractive errors in infancy showed emmetropisation and 20% who had no significant refractive errors went on to develop refractive errors. In addition however research findings also show that the majority of children with Down syndrome have below normal acuity even in the absence of ocular anomalies and with refractive errors fully corrected^(18, 19).

Both Woodhouse and colleagues^(4, 5) and Haugen et al⁽²⁾, using dynamic retinoscopy, have demonstrated poor accommodation in many of these young children. They tend to under-accommodate and this results in blurred near vision which undoubtedly contributes to the below normal acuity. Courage⁽¹⁸⁾ and others also speculate that neurodevelopmental problems may play a part.

All of these findings^(2, 3, 4, 5, 13, 18) indicate an incontrovertible need for frequent ocular appraisal in the early years. Because problems with near vision are exacerbated by poor accommodation, we have stressed the need to check distance and near functioning at every review, whenever this is developmentally possible, (see guideline point 7) and at all ages to consider a prescription for near correction or bifocals. Practitioners should have a high index of clinical suspicion and note peering or other unusual visual behaviour especially in young children who are unable to co-operate fully.

We have been criticized for citing unpublished data^(11, 12) as the basis for our statement that one third of children will have ocular/visual defects by age 18 months to 2 years. This information is enmeshed in Woodhouse's papers⁽¹⁾ giving data on prevalence of refractive errors, strabismus and nystagmus, but has not been published as such. Unfortunately Crofts' data on children attending a specialist Down syndrome clinic in Oxford though collated and presented at meetings has never been put forward for publication.

It has however been suggested to us that even if at least a third of children have significant visual or ocular disorder by age 18 months to 2 years there is no evidence that benefit necessarily results from correcting refractive and/or accommodative errors at this early age. Some argue that screening is better delayed until nearer the time that the child will go to school.

A difficulty with this approach is that between age 2 and 3 years children increasingly need to use their near vision for activities both at home and in nursery or playschool environments. Uncorrected hypermetropia will confer additional disadvantage for a child already struggling with learning difficulties. Defective accommodation will also contribute to blurred near vision. Early identification and consideration of full correction of hypermetropia with spectacles or bifocal lenses should reduce the burden of disadvantage. Even those without a

refractive error in infancy should be re-examined again age 4 as this could develop. This approach has been implemented in some centres following publication of DSMIG Guideline, and is effective and practical⁽³³⁾

Point 5. The evidence for ongoing failed emmetropisation (see above) and robust evidence that 50% of those with Down syndrome have refractive errors by age 4 years⁽¹⁾ underlies our recommendation that further full ocular review is essential at age 4 even for those without refractive error at age 2.

Points 6, 7 and 8. The guideline hereafter is mainly straightforward and includes many 'good practice' points. Many hospital clinics are now prescribing bifocals as the evidence for their benefit mounts, including improvement in visual perceptual skills and literacy^(17,36). There is no doubt that they are effective at improving focus at near^(17,38) and many children choose to wear them all the time. Furthermore extrapolating from the experiences of older presbyopes who experience great subjective relief when increasing hypermetropia and poor accommodation are corrected by spectacles, it is reasonable to expect that young children with Down syndrome will benefit likewise. It is interesting to note that evidence is emerging which suggests that some children who wear bifocal lenses 'learn' to accommodate and are subsequently able to give them up⁽³⁸⁾. Hence our consensus view is that the provision of bifocal glasses is good practice.

Point 9. We have pointed out that nasolacrimal duct obstruction occurs commonly in young children with Down syndrome^(20,22). Stephens et al⁽³⁴⁾ found that although this may take longer to resolve than in typically developing children spontaneous resolution commonly occurs by age 2 years and the majority resolve by 3 years. However specialist referral may be necessary in persistent or troublesome cases.

References for guideline

1. Woodhouse JM, Pakeman VH, Cregg M, Saunders KJ, Parker M, Fraser WI, Sastry P, Lobo S. (1997). Refractive errors in young children with Down syndrome. *Optometry & Vision Science* **74** 844-851
2. Haugen OH, Hovding G. (2001). Strabismus and binocular function in children with Down syndrome: A population-based, longitudinal study. *Acta Ophthalmologica Scandinavica*. **79** 133-139
3. Cregg M, Woodhouse JM, Stewart RE, Pakeman VH, Bromham NR, Gunter HL, Trojanowska LD, Parker M, Fraser WI. (2003). Development of refractive error and strabismus in children with Down syndrome. *Investigative Ophthalmology & Visual Science* **44** 1023-1030
4. Woodhouse JM, Cregg M, Gunter HL, Sanders DP, Saunders KJ, Pakeman VH, Parker M, Fraser WI, Sastry P. (2000). The effect of age, size of target and cognitive factors on accommodative responses of children with Down syndrome. *Investigative Ophthalmology & Visual Science* **41** 2479-2485
5. Cregg M, Woodhouse JM, Pakeman VH, J SK, Gunter HL, Parker M, Fraser WI, Sastry P. (2001). Accommodation and refractive error in children with Down syndrome: cross sectional and longitudinal studies. *Investigative Ophthalmology & Visual Science* **42** 55-63
6. Kallen B, Mastroiacovo P, Robert E. (1996). Major congenital malformations in Down syndrome. *American Journal of Medical Genetics*. **65** 160-166

7. *Traboulsi EI, Levine E, Mets MB, Parelhoff ES, O'Neill JF, Gaasterland DE* (1988) Infantile glaucoma in Down's syndrome (trisomy 21) *American Journal of Ophthalmology* **105** 389 – 394
8. *Pires da Cunha R, Belmiro de Castro Moreira J.* (1996). Ocular findings in Down's Syndrome. *American Journal of Ophthalmology* **122** 236-244
9. *van Schroyen Lantman-de Valk HMJ, Haveman MJ, Crebolder HFJM,* (1996) Comorbidity in people with Down's syndrome: a criteria based analysis. *Journal of Intellectual Disability Research*, **40** 385 – 399
10. *Rahi JS, Williams C, Bedford H, Elliman D.* (2001). Screening and surveillance for ophthalmic disorders and visual deficits in children in the United Kingdom. *British Journal of Ophthalmology* **85** 257-259
11. *Woodhouse JM*, unpublished data.
12. *Crofts B*, unpublished data
13. *Haugen OH, Hovding G, Lundstrom I.* (2001). Refractive development in children with Down's syndrome: a population based, longitudinal study. *British Journal of Ophthalmology* **85** 714-719
14. Guidelines for Children's Eye Care.(2002) Children's Eye Health Working Party, a joint working party comprising members of the Royal College of Ophthalmologists, College of Optometrists and British Orthoptics Society.
15. Guidance on Frequency of Eye Examinations (2001). Issued by the College of Optometrist
16. *Woodhouse JM.* Bifocal information leaflet. dsmig.org.uk/library/articles/bifocals.html
17. *Stewart RE, Woodhouse JM, Trojanowska LD.* (2005). In focus: the use of bifocals for children with Down's syndrome. *Ophthalmic and Physiological Optic.* **25** (6) 514-522
18. *Courage ML, Adams RJ, Reyno S, Kwa PG.* (1994). Visual acuity in infants and children with Down syndrome. *Developmental Medicine & Child Neurology* **36** 586-593
19. *John FM, Bromham NR, Woodhouse JM, Candy TR.* (2004). Spatial vision deficits in infants and children with Down syndrome. *Investigative Ophthalmology & Visual Science* **45** 1566-1572
20. *Fierson WM.*(1990).Ophthalmological aspects. In: *Clinical perspectives in Down syndrome* (Ed. Van Dyke D., Lang D., Heide F., Van Duyne S., Soucet M.) Springer-Verlag, New York, pp 26-54
21. *Oxford Eye Hospital.* Advice re lid hygiene. dsmig.org.uk/library/articles/blepharitis.html
22. *Berk AT, Saatci AO, Ercal MD, Tunc M, Ergin M.* (1996). Ocular findings in 55 patients with Down's syndrome. *Ophthalmic genetics.* **17** 15 – 19

Additional references for background notes:

30. *Hall and Elliman.* Health for all Children. Revised, illustrated 4th edition. 2006. Published by Oxford University Press, ISBN 0198570848, 9780198570844
31. *Rahi JS and Dezateux C.* (1999) National cross sectional study of detection of congenital and infantile cataract in the United Kingdom: role of screening and surveillance. *BMJ* **318** 362-5
32. *Rahi JS, Lynn R* (1998) A survey of paediatricians' practice and training in routine infant eye examinations. *Arch.Dis.Child* **78** 364-6
33. *Stephens et al* (2007) Surveillance of vision and ocular disorders in children with Down syndrome. *Dev Med and Child Neurology* **49**: 513-515
34. *Stephens et al* (2005) - presentation at DSMIG meeting November 2005)
35. *Creavin and Brown* (2009) Ophthalmic Abnormalities in Children With Down Syndrome. *Journal of Pediatric Ophthalmology & Strabismus* **46** No. 2: 76-82
36. *Nandakumar K, Leat SJ.*(2010) Bifocals in children with Down syndrome (BiDS) – visual acuity, accommodation and early literacy skills. *Acta Ophthalmologica* **88** 196-204

37. *Roizen NJ, Mets MB, Blondis TA (1994) Ophthalmic disorders in Children with Down Syndrome. Dev Med Child Neurol 36 594-600*
38. *Al-Bagdady M, Stewart RE, Watts P, Murphy PJ, Woodhouse JM, (2009) Bifocals and Down's syndrome: correction or treatment? Ophthalmic Physiol Opt.;29 416-421*

JD/PC/MW 13 February 2012