The eye and Down's syndrome

Down's syndrome is a common genetic abnormality that can affect most parts of the eye. Common ocular features are described, as well as those which cause reduced vision, requiring referral to an ophthalmologist.

Down's syndrome is a genetic syndrome with a wide range of characteristic clinical findings. Ocular associations of the syndrome have been reported in every part of the eye except for the vitreous. None of the ocular features are pathognomonic of Down's syndrome and there are large variations in phenotypic expression, such that no individual exhibits every ocular abnormality and there are no identified groups of concordant findings (Catalano, 1990). This article describes the common ocular features associated with a diagnosis of Down's syndrome and identifies those features that could affect sight and therefore require referral to an ophthalmologist.

Features impacting on vision

Refractive errors

Refractive errors are very common in Down's syndrome and include hypermetropia, myopia and astigmatism (Shapiro and France, 1985; Catalano, 1990; Berk et al, 1996; Da Cunha and Moreira, 1996; Haugen et al, 2001; Yurdakul et al, 2006). Uncorrected refractive errors give rise to reduced vision and affect quality of life. Thirty per cent of the population with Down's syndrome have high levels of myopia and there is a strong correlation between congenital heart disease and this condition.

However, the majority of individuals with Down's syndrome have mild refractive errors that can be readily corrected by spectacles. An annual optometric review from the age of 4 years is recommended to screen for this.

Strabismus and amblyopia

Between 23 and 43% of children with Down's syndrome have squints, most of which are convergent, with the average age of onset being 4 years (Shapiro and France, 1985; Catalano, 1990; Berk et al, 1996; Da Cunha and Moreira, 1996; Haugen et al, 2001; Yurdakul et al, 2006). The majority of these squints can be corrected by wearing spectacles and in many cases binocular vision is restored (Haugen and Hovding, 2001).

Amblyopia is defined as defective vision in one or both eyes that persists after the correction of refractive errors and any pathological obstacle to vision. It has been described in up to 25% of individuals with Down's syndrome (Da Cunha and Moreira, 1996). Once again, optometrists can screen for this and, if detected, occlusion treatment to the good eye can be carried out under the supervision of an ophthalmologist and orthoptist.

Mr Sidath Liyanage *is Specialist Registrar in Ophthalmology, Moorfields Eye Hospital, London EC1V 2PD and* **Mr Jonathan Barnes** *is Consultant Paediatric Ophthalmologist, Luton and Dunstable Hospital, Luton*

Correspondence to: Mr S Liyanage

Cataracts

The true prevalence of cataracts in Down's syndrome is not known, with estimates ranging from 13 to 85% (Shapiro and France, 1985; Catalano, 1990; Berk et al, 1996; Da Cunha and Moreira, 1996). Higher rates of congenital cataract have been reported and all infants are screened for this at their postnatal and 6-week checks. Flake-like lens opacities are commonly encountered during the first decade. Also, nuclear sclerotic cataracts, which are typically found in the elderly, occur at a much younger age in the Down's syndrome population. This may represent an example of the premature ageing process associated with the syndrome.

As in the general population, cataract surgery is the most common intraocular procedure performed in Down's syndrome. Surgery for juvenile and adult-onset cataracts is considered when the patient complains of problems with his/her vision. In cases where the individual is unable to vocalize difficulties, carers may identify changes in behaviour that suggest that the patient's vision is impaired because of a cataract.

Nystagmus

Nystagmus represents an involuntary rhythmic oscillation of the eyes that can contribute to decreased vision. It has been noted in up to 30% of individuals with Down's syndrome (Shapiro and France, 1985; Catalano, 1990; Da Cunha and Moreira, 1996). This high prevalence is thought to represent abnormal integration of visio-spatial information (Averbuch-Heller et al, 1999). Children with nystagmus need to be referred to an ophthalmologist for further assessment. Prismatic lenses can sometimes help to dampen the nystagmus and improve vision.

Keratoconus

Keratoconus is a corneal abnormality characterized by central corneal thinning that subsequently deforms the shape of the cornea. In mild cases, this produces high degrees of corneal irregularity or astigmatism. In extreme cases, the central portion of the cornea becomes conical and scarred, and may perforate. Keratoconus is commonly associated with Down's syndrome, with a prevalence of 15% (Shapiro and France, 1985). The precise

aetiology is not known, but chronic eye rubbing is believed to be a contributing factor. This is likely to be influenced by blepharitis, which can cause eyelid irritation. In mild cases, it can be managed by fitting contact lenses in those individuals who can tolerate them. Corneal grafts may be considered for severely scarred central corneas or in cases where the cornea has thinned to the point of perforation.

Other ocular features of Down's syndrome

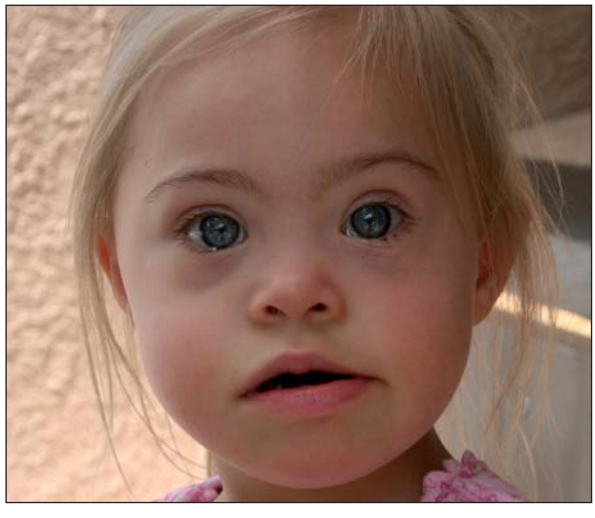
External appearance

Particular physical characteristics of Down's syndrome include a flattish, oval face with slanting eyes and epicanthic folds. The palpebral fissure (the area exposed between the upper and lower eyelids) is upward slanting such that the outer canthus of the eyelid is around 2 mm higher than the inner canthus (*Figure 1*). The palpebral aperture is narrowed horizontally (less than 30 mm) but of normal height (around 11 mm) (Shapiro and France, 1985; Catalano, 1990; Da Cunha and Moreira, 1996). Epicanthic folds are folds of skin that arise from the nasal margin of the upper eyelid and insert below the inner canthus (*Figure 2*). They have been noted in 60–100% of individuals with Down's syndrome and usually regress with age (Shapiro and France, 1985; Catalano, 1990; Da Cunha and Moreira, 1996).

Iris abnormalities

Brushfield spots are distinct markings on the surface of the iris (*Figure 3*). Histologically, they represent a hypercellular area of the iris tissue with a surrounding area of relative hypoplasia. Ninety per cent of people with Down's syndrome have Brushfield spots compared with 24% of the total population (Catalano, 1990). In Down's syndrome, these spots are more numerous and closer to the pupil margin. Brushfield spots do not correlate with mental disability, and are rarely encountered in the Asian population with Down's syndrome.

Figure 1. Young girl with Down's syndrome demonstrating upward slanting palpebral apertures.



Blepharitis

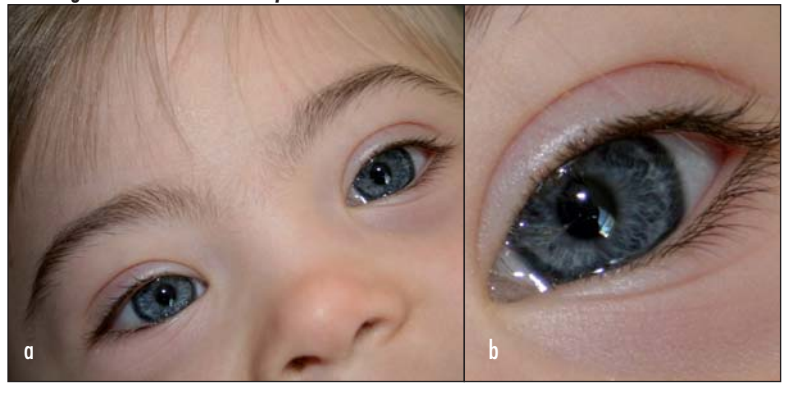
Although common in the general population, up to 46% of patients with Down's syndrome have blepharitis (Shapiro and France, 1985). Decreased resistance to infection and poor hygiene may contribute to the increased risk of this inflammatory eyelid margin condition. Significant discomfort, including redness, soreness and grittiness, may result from this inflammation. Treatment is initially conservative, using regular lid margin cleaning with baby shampoo or a solution of bicarbonate of soda. Indolent cases may need oral antibiotic treatment with doxycycline.

Retinal abnormalities

An increased number of vessels crossing the disc margin has been described in Down's syndrome, resulting in a spoke-like distribution (Catalano, 1990; Berk et al, 1996; Da Cunha and Moreira, 1996). This has no

Figure 2. Left epicanthic fold, arising at the nasal margin of the upper eyelid and inserting below the inner canthus. These folds can lead to the false impression of squint (note symmetrical appearance of the corneal light reflexes).

Figure 3. a. Brushfield spots appearing as hypopigmented (whitish) areas of the blue iris. b. Enlarged view of Brushfield spots.



known functional effect. Other retinal abnormalities, such as a pale fundal appearance, have been reported, occurring in up to 30% of the Down's syndrome population, but these may result from co-existent high myopia (Da Cunha and Moreira, 1996).

Conclusions

Down's syndrome is a relatively common condition and it is therefore important to consider whether any feature is truly associated with the condition, or if the increased prevalence simply reflects the chance occurrence of unrelated findings. Common ocular features highlighted in this article reflect true increased rates in large series of patients with Down's syndrome. While no ocular finding is pathognomonic of Down's syndrome, the presence of the above features are strongly associated with a diagnosis of Down's syndrome.

KEY POINTS

- Down's syndrome can affect most parts of the eye, resulting in reduced vision. It is important to consider this as most problems can be treated, leading to an improved quality of life.
- Ocular appearance can greatly contribute towards the diagnosis of Down's syndrome. External features include the upward slanting of the palpebral fissure and epicanthic folds.
- Children with Down's syndrome should be in contact with ophthalmic services in order to chart their visual development and screen for conditions that impact on vision.
- Regular optometric checks are essential in monitoring and treating refractive errors. Optometrists can also detect cataracts, which can occur at an earlier age in patients with Down's syndrome.
- Almost 50% of patients with Down's syndrome have blepharitis. This inflammatory eyelid condition can cause great discomfort and can be effectively treated with conservative management.

As this condition is the most common chromosomal abnormality in humans and patients have a relatively good life expectancy, it is important for physicians to consider significant but correctable ocular abnormalities that may impact on visual potential and quality of life.

All infants with Down's syndrome are screened for lens opacities at birth and at their 6–8-week check. Following this, children with Down's syndrome should be in contact with ophthalmic services in order to chart their visual development and screen for conditions which impact on vision. The UK Down's Syndrome Medical Interest Group have produced an insert for the personal child health record book (red book) which suggests a schedule of these checks for children with Down's syndrome (UK Down's Syndrome Medical Interest Group, 2000). **BJHM**

Figures 1, 2 *and* 3 *are reproduced from http://mdbeau.blogspot.com/2007/10/characteristics.html by kind permission of Michelle Helferich.*

Conflict of interest: none.

Averbuch-Heller L, Dell'Osso LF, Jacobs JB, Remler BF (1999) Latent and congenital nystagmus in Down syndrome. *J Neuroophthalmol* **19:** 166–72

Berk AT, Saatci AO, Ercal MD et al (1996) Ocular findings in 55 patients with Down's syndrome. *Ophthalmic Genet* **17:** 15–19

Catalano RA (1990) Down Syndrome. *Surv Ophthalmol* **34:** 385–98

Da Cunha RP, Moreira JBC (1996) Ocular findings in Down's syndrome. *Am J Ophthalmol* **122:** 236–44

Haugen OH, Hovding G (2001) Strabismus and binocular function in children with Down syndrome. A population-based, longitudinal study. *Acta Ophthalmol Scand* **79:** 133–9

Haugen OH, Hovding G, Lundstrom I (2001) Refractive development in children with Down's syndrome: a population based, longitudinal study. *Br J Ophthalmol* **85:** 714–19

Shapiro MB, France T (1985) The ocular features of Down's syndrome. *Am J Ophthalmol* **99:** 659–63

UK Down's Syndrome Medical Interest Group (2000) PCHR Insert. www.dsmig.org.uk/publications/pchr.html (accessed 28 October 2008)

Yurdakul NS, Ugurlu S, Meden A (2006) Strabismus in Down syndrome. *J Pediatr Ophthalmol Strabismus* **43:** 27–30