BASIC MEDICAL SURVEILLANCE ESSENTIALS FOR PEOPLE WITH DOWN SYNDROME

OPHTHALMIC CONDITIONS (Revised 2025)

A Down Syndrome Medical Interest Group (DSMIG(UK)) best practice recommendation

1. There is a high prevalence of ocular disorder among people with Down syndrome. Refractive errors and/or squint may be present from an early age and persist into adolescence and adulthood. The majority of children with Down syndrome have reduced accommodation at near (2,4,5). Compared to the general population there is a tenfold increase in congenital cataract (6) and infantile glaucoma may also occur (7). Nystagmus is present in at least 10% and severely impacts visual acuity (8, 9). Cataracts may develop in teenage years or later and studies suggest that these are approximately 4 times more common than in the adult general population, and are often bilateral (45%) (10, 11). Keratoconus onset is often in adolescence or early adulthood, and prevalence estimates vary from 5% in nationwide registry studies to nearly 70% having keratoconus-compatible corneal topography, although most studies suggest rates closer to 20-30% (12, 13, 14). People with Down syndrome have also been shown to have abnormal macular development and disc morphology, with a higher incidence of foveal hypoplasia, thicker ganglion cell and inner nuclear layers, and tilted and smaller discs (15, 16, 17). Most of these disorders are a significant cause of preventable secondary disability at all ages. Therefore, there should be extra vigilance at all ages.

2. As with all children, newborns with Down syndrome should be examined for congenital cataract and other eye anomalies within 72 hours by a trained person and this should be repeated at 6-8 weeks (18).

3. Visual behaviour should be monitored by the child's paediatrician particularly before the first formal ophthalmologic review. Those who start to squint or show other abnormalities of gaze, visual behaviour or attention should be referred for ophthalmological review.*

4. Between 18 months and 2 years, all children with Down syndrome should have formal ocular/visual assessment by an orthoptist and ophthalmologist/optometrist in accordance with local arrangements. This should include orthoptic assessment, refraction, and fundus examination, with vigilance for cataract, nystagmus, strabismus and refractive error in particular. At least one third will have ocular/visual defects by this age (1, 19, 20). Those with deviation from normal should be kept under appropriate specialist review. Refractive errors, most commonly hypermetropia, which often reduce spontaneously in other children, are likely to persist beyond infancy (3, 21). Correction for hypermetropia may be helpful at a younger age than that for typically developing children especially since the majority will have defective accommodation (2, 4, 5).

5. Those with no abnormality at first review should nevertheless have further full ocular/visual assessment including refraction around age 4 years (18, 22). As most subsequent abnormalities following first assessment are detectable with automated vision screening techniques or direct visual inspection of the eyes and ocular adnexa, formal optometrist visual screening or community orthoptic assessment could possibly replace a subsequent formal ophthalmological examination, if the preferable formal ophthalmological examination is not available (23). Contrast sensitivity is also commonly significantly reduced in this group and should be assessed at this review to guide any learning resource adaptation requirements in the early stages of schooling (24). At this age at least 50% are likely to have refractive errors (1).

6. After age 4, due to the increased prevalence of disorders, eye checks should be at least 2 yearly, preferably annually, until age 10 by professionals with appropriate skills and expertise in managing this client group (22, 25). These may be optometrists (hospital or high-street based), orthoptists or ophthalmologists. Refractive error should be monitored to ensure optimal correction and clinicians should be alert to the potential for myopia and astigmatism to develop at any age 9, 21).

7. Annual checks are advised from age 10 onwards, with particular emphasis on the early detection of keratoconus. Assessment should include detailed corneal evaluation using corneal topography, if available (12). Prompt specialist referral is essential to prevent disease progression, through reducing provoking factors such as eye rubbing, often exacerbated by ocular surface symptoms, and for consideration of corneal cross-linking (26, 27).

8. Children and adults with Down syndrome should be expected to respond to standard vision testing procedures at appropriate developmental age but a distraction-free environment and extra time may be necessary to optimise performance. Distance and near functioning vision should be checked at every review whenever developmentally possible and a prescription for near correction or bifocals considered at all ages* (21, 28, 29). Assessment of accommodative lag should ideally involve dynamic retinoscopy as there is a lack of correlation with near visual acuity (30). There is increasing evidence that bifocals are superior to unifocals, with improved near visual acuity in patients with accommodative lag, reduced manifest angle of strabismus and improved spectacle compliance demonstrated in studies (31, 32, 33). Visual acuity is likely to remain poorer than expected throughout life even when appropriate spectacles are worn, with a deficit of approximately 0.2 logMAR compared to children without Down syndrome (9, 34, 35).

9. Many high street opticians/optometrists give an excellent service particularly for older children, but younger children and those who find it difficult to tolerate this setting should be seen in a specialist clinic. The SeeAbility charity maintains a register of optometrists accredited to provide the Easy Eye Care service, a specialist eye care service for people of all ages with learning disabilities and autism (36). Children enrolled in special schools across England now benefit from on-site eye care services following a recent proposal from several UK eye care organisations (37).

10. Blepharitis has been reported to occur in up to 30% of children with Down syndrome (8, 38) and ideally can be managed in the usual way, although compliance issues may complicate management (39). Nasolacrimal duct obstruction (8.5%) and congenital lacrimal fistula (4%) also commonly occur (40, 41) and may need specialist referral. *

11. A significant proportion of children with Down syndrome will have visual perceptual problems. Cerebral Visual Impairment, or CVI, refers to difficulty in interpreting vision, even when visual acuity is reasonable or good. Common symptoms include difficulties in judging depth, difficulties finding an object in an array, poor judgement of movement, and problems with face and object recognition. Once any refractive error is corrected, any child who has unexplained visual difficulties should be referred for assessment of CVI (42).

12. In view of the high prevalence of ocular disorders (see item 1) and the communication difficulties encountered in people with Down syndrome, any child or adult with pain, and/or changing vision, and/or red eye should be referred in the normal way. *

* good practice point

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