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# Epidemiology, aetiology and management of visual impairment in children

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Received 30 March 2013

Revised 19 September 2013

Accepted 24 September 2013

Published Online First

22 October 2013

## ABSTRACT

An estimated 19 million of the world's children are visually impaired, while 1.4 million are blind. Using the UK as a model for high income countries, from a population-based incidence study, the annual cumulative incidence of severe visual impairment/blindness (SVL/BL) is estimated to be 6/10 000 by age 15 years, with the incidence being highest in the first year of life. The population of visually impaired children within high, middle and lower income countries differ considerably between and within countries. The numerous and mainly uncommon disorders which can cause impaired vision result in heterogeneous population which includes a substantial proportion (for SVI/BL, the majority) of children with additional systemic disorders or impairments whose needs differ substantially from those with isolated vision impairment. Paediatricians and other paediatric professionals have a key role in early detection and multidisciplinary management to minimise the impact of visual impairment (VI) in childhood.

## INTRODUCTION

Visual impairment (VI) has a significant impact on the affected child's psychological, educational and socioeconomic experiences, during childhood and beyond. As the disorders which cause VI in childhood are uncommon, the population of children with VI is complex and heterogeneous, but essentially comprises two main groups: those with isolated VI and those with VI in addition to, or associated with, another disorder or impairment. These two populations differ significantly with respect to their clinical management and their health, educational and social care needs. Here we set out current data on frequency and causes and the general principles of management of all-cause VI in childhood.

## Normal visual development in childhood

Vision comprises several interconnected functions such as colour vision, depth perception and higher level cognitive functions such as visuo-spatial processing, but the key function is acuity. Acuity is quantified using gratings or optotype symbols such as shapes and letters. It is now most commonly measured on a logarithmic scale (logMAR) in which 0.0 is 'normal' acuity, and 1.0 logMAR indicates a 10-fold decrease in acuity (table 1). Previously, the geometric Snellen scale was more widely used, where 6/6 is normal vision, and 6/60 means the subject sees at a distance of 6 m the symbol that would be seen at 60 m by a person with 'normal' vision (table 1).

Vision rapidly matures during the first few years of life as ocular anatomy and visual pathways

circuitry develop. Newborns have an average acuity of approximately 1.5 logMAR, which rapidly improves to an average acuity 0.35 logMAR by 24 months of age, and 0.0 ('normal' adult acuity) by 5 years of age.<sup>1 2</sup> Methods for assessing vision need to be appropriate to the age and developmental stage of the child. By the age of 5 years, the majority of (otherwise developmentally normal) children are able to comply with simple quantitative shape/letter-based acuity chart testing. In young, preverbal children and those with developmental, cognitive or communication disorders, gaze behaviour responses ('preferential-looking') to graded visual stimuli can be used to give some estimate of acuity level.

## Sensitive periods and amblyopia

Animal experimental research has shown that the development of mammalian sensory modalities involves a crucial sensitive period, a time window during early development when experience has a profound effect on the consequent structure and function of the brain.<sup>3 4</sup> Within the sensitive period is a critical period, during which visual experience is absolutely necessary for the creation of neural networks and subsequent normal function. Evidence from clinical (human) research supports the existence of this 'critical period' in early infancy.<sup>5</sup> The visual system is progressively less responsive (sensitive) until the age of about 8 years, although in some individuals sensitivity persists into late childhood and even occasionally into adult life.<sup>4</sup> Any disorder which prevents normal visual experience will result in failure of normal visual development, that is, amblyopia (a form of developmental cerebral VI). Amblyopia is treatable within the window of sensitivity, but beyond this period amblyopia is associated with permanent impairment. In managing any ophthalmic disorder, its direct visual impact and its indirect potential impact through amblyopia have to be considered.

## Defining visual impairment

Most systems for classifying VI are based on acuity in the better eye and to a lesser degree the visual field. A child with impaired vision (of any severity) in only *one* eye due to unilateral or asymmetric disease is thus not formally considered 'visually impaired'. The WHO categorisation system for VI (table 1) has been very widely but not universally adopted. The UK criteria for the certification of children as being sight impaired (previously termed 'partially sighted') or severely sight impaired (previously termed 'blind') are also set out in table 1. Although children with vision better than 0.5 logMAR but worse than 0.4 in the better seeing



► <http://dx.doi.org/10.1136/archdischild-2012-301970>

**To cite:** Solebo AL, Rahi J. *Arch Dis Child* 2014;**99**:375–379.

eye are not formally classified as sight impaired, this level of vision is below the threshold for driving and is increasingly termed as socially significant VI.<sup>6</sup> In this article, we have adopted wherever possible the WHO terminology referring to VI and severe visual impairment/blindness (SVI/BL).

**BURDEN OF VISUAL IMPAIRMENT**

An estimated 19 million of the world’s children are visually impaired, while 1.4 million are blind, according to WHO criteria.<sup>6</sup> There are limited population-based data on the epidemiology of childhood VI and BL owing to the methodological challenges to obtaining accurate information on uncommon and heterogeneous disorders. In particular, robust data on the frequency of mild/moderate VI are lacking for many countries where data are available for those with severe sight or visual impairment/blindness (SVI/BL).

In some higher income countries, it is possible to estimate the prevalence of VI or SVI/BL using live registers of VI (eg, in the UK all children certified as sight impaired/severely sight impaired are then registered as such by the Social Services system), although these may be incomplete or biased if registration is voluntary (non-statutory). Data from middle and lower income countries have tended to be derived from studies of schools for blind children, but more recently there have been large-scale population-based studies.<sup>7</sup> These studies estimate the prevalence of childhood BL in middle and lower income countries at between 0.2 and 7.8 per 10 000.<sup>7</sup> The variation in prevalence of SVI/BL closely correlates with the under 5 years childhood mortality rate in the region,<sup>8</sup> with BL itself and the causes of BL being linked to the increased risk of mortality.

Using the UK as a model for industrialised countries, from a population-based incidence study, the annual cumulative incidence (in 2000) of SVI/BL is estimated to be 6/10 000 by age 15 years, with the incidence being highest (4/10 000) in the first

year of life.<sup>9</sup> The evidence on temporal trends is unclear. A decline in incidence of VI and SVI/BL between 1984 and 1998 was reported from an analysis of the Oxford Region Register of Early Childhood Impairments.<sup>10</sup> By contrast, a doubling of the number of children ‘registered’ as blind in England and Wales between 1982 and 2011, with the incidence increasing from 0.17/10 000 to 0.41/10 000 during that period, has recently been reported.<sup>11</sup> While changes in certification practices may partly account for the latter, on balance it is likely that the overall trend in the UK is indeed of increasing frequency through a combination of an increase in the population at risk, and an increasing incidence of disorders which cause VI and improved survival of children with VI.

**CAUSES OF CHILDHOOD VISUAL IMPAIRMENT**

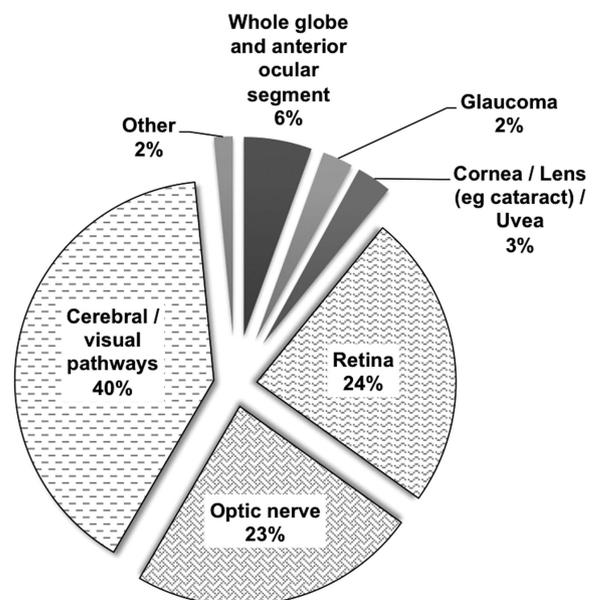
The pattern of underlying disorders (‘causes’) of VI and BL vary considerably between and within (rural/urban settings) countries, reflecting the regional balance of the determinants of specific diseases, and the available resources to execute preventive strategies. Globally, the most frequent causes of childhood VI/SVI/BL are retinal disorders, glaucoma, corneal scarring (primarily due to Vitamin A deficiency), cataract and cerebral causes.<sup>7</sup>

**Causes of visual impairment in higher income countries**

Due to the challenges discussed earlier, the epidemiology of the individual causative disorders underlying childhood VI is uncertain. The most common cause of childhood SVI/BL in industrialised countries such as the UK and USA is neurological or cerebral disorder affecting the visual system, due to ischaemic, developmental or unknown insults.<sup>12 13</sup> While the USA data are based on studies of children registered in schools for the blind, the UK estimates are drawn from the British Childhood Visual Impairment Study (BCVIS), the only national population-based incidence study of childhood SVI/BL. Of the 493 children newly diagnosed with SVI/BL in 2000, almost 50% of children had cortical VI (figure 1).<sup>12</sup> Optic nerve pathology accounted for 28% of childhood SVI/BL, and retinal disorders (including

**Table 1** Categorising vision: logMAR and Snellen measurement scales, the WHO and UK categories of visual impairment (VI)

Acuity measurement scales		Vision categories (using best achievable acuity with both eyes open)	
LogMAR (linear scale)	Snellen (geometric scale)	WHO definition of visual impairment	UK criteria for visual impairment certification Full field of vision   Very reduced field of vision
-0.1	6/4.8	No sight impairment	No sight impairment
0.0	6/6		
0.1	6/7.5		
0.2	6/9		
0.3	6/12		
0.4	6/15	No sight impairment*	No sight impairment
0.5	6/18		
0.6	6/24	Moderate visual impairment	Sight impairment (previously termed partially sighted)
0.7	6/30		
0.8	6/36		
0.9	6/48		
1.0	6/60	Severe visual impairment	Severe sight impairment
1.1	5/60		
1.2	4/60		
1.3	3/60	Blindness	Severe sight impairment
1.4	2/60		



**Figure 1** The different causes of childhood severe visual impairment/blindness (SVI/BL) in the UK British Childhood Visual Impairment Study (BCVIS) by anatomical site affected (JR).

retinopathy of prematurity) 29%. These three causes (insult to the cortex, optic nerve and retina) are also the most common causes of childhood SVI/BL in other higher income countries.<sup>13</sup> The majority of children in BCVIS (77%) had an additional associated non-ophthalmic disorder, as has commonly been described in similar populations.<sup>13</sup> An increased risk of SVI/BL in children from ethnic minority groups, socio-economically deprived families, and those of low birth weight (<2500 g) was clearly identified, as was a 10% mortality risk in the first year after diagnosis; these findings being echoed by subsequent studies in industrialised countries.<sup>14 15</sup> Preterm birth, inevitably associated with low birth weight, has increased significantly over the last two decades in the UK<sup>16</sup> and the neurological sequelae of low birth weight are well recognised, with these children being more likely to have white matter damage affecting the visual system or developmental anomalies of the optic nerve.<sup>17</sup> There is also an increasing recognition of the impact of late preterm birth (between 34 and 36 weeks' gestation) on adverse neurodevelopmental outcomes.<sup>18 19</sup> The continued increase in the number of preterm births can be expected to have an impact on the frequency of VI globally, as can the increased survival of children with neurological or neurodevelopmental disorders.

### MANAGEMENT OF CHILDHOOD VISUAL IMPAIRMENT

It is beyond the scope of this review to discuss the ophthalmic management of individual disorders that cause VI. It is accepted that children affected by these disorders require multidisciplinary specialist teams with appropriate training and facilities and ancillary structures, in keeping with the Royal College of Ophthalmologist Quality Standards and Quality Indicators for Ophthalmic Care and Services for Children and Young People (<http://www.rcophth.ac.uk/page.asp?section=444&sectionTitle=Quality+Standards+for+Paediatric+Ophthalmology>) and its standing report 'Ophthalmic Services for Children' (<http://www.rcophth.ac.uk/page.asp?section=293&sectionTitle=Ophthalmic+Services+Guidance>). Here we discuss the management of 'all cause' VI in terms of primary, secondary and tertiary prevention.

#### Primary prevention: preventing the insult to the visual system

In the BCVIS, the majority of children with SVI were blind due to disorders or pathologies attributable to one or more prenatal insults to the developing visual system. For many of these children, the pathophysiology of the insult was associated with the developmental consequences of preterm birth. Preterm birth (and low birth weight) is, globally, the most significant cause of newborn mortality and is rising in prevalence across many countries.<sup>20</sup> Prematurity is associated with multiple interrelated risk factors including maternal age, health and socio-economic status.<sup>21 22</sup> Thus, it is a considerable challenge to direct preventive strategies towards this population. An active area of research that may impact on cerebral VI in addition to other adverse neurological outcomes is therapeutic hypothermia, with a recent Cochrane systematic review supporting its efficacy in hypoxic ischaemic encephalopathy in late preterm and term infants.<sup>23</sup>

A comparison of the relative importance of causes of childhood VI in higher, middle and lower income countries provides some indirect evidence of the beneficial impact of general public health preventive strategies such as immunisation against measles and rubella, and childhood nutritional programmes targeted against vitamin A deficiency in reducing the burden of BL due to corneal opacity, cataract and other ocular anomalies.<sup>7 8 13</sup>

As hereditary causes account for a third of childhood SVI/BL in the BCVIS cohort, genetic counselling is potentially a tool in the prevention of childhood VI due to known Mendelian eye conditions.

Retinopathy of prematurity (ROP) is a globally important cause of childhood SVI/BL. Screening for ROP aims to detect infants with early stage disease to enable timely treatment to prevent the development of advanced disease leading to retinal detachment and BL. In higher income countries, routine screening is undertaken of infants born earlier than 32 weeks' gestational age or with birth weight of less than 1500 g, according to national guidance ([http://www.rcophth.ac.uk/core/core\\_picker/download.asp?id=180](http://www.rcophth.ac.uk/core/core_picker/download.asp?id=180)). In low and middle income countries, screening criteria and national programmes differ as infants of greater gestational age and/or birth weight are also considered to be at risk of developing ROP.<sup>24</sup>

#### Secondary prevention: early detection of visual impairment

Prompt identification of childhood VI is essential as it allows early intervention of ophthalmic and developmental interventions which are necessary to maximise visual outcomes. Early ophthalmic intervention will be directed at the disorder itself and the associated amblyopia. Early detection will also be important to the success of emerging novel therapies, such as genetic therapies for inherited retinal dystrophies.<sup>25</sup>

Paediatricians and other paediatric health professionals have a key role in the early detection of children with impaired vision and/or visually impairing ophthalmic disorders. While parents and caregivers are often the first to suspect some degree of impaired vision in their child, in the UK almost half of all children with SVI/BL first present to hospital-based paediatricians.<sup>9</sup> This is particularly the case for the large population of children who have additional associated systemic disorders.

In approximately a fifth of children with SVI, reduced vision is discovered in the context of the routine NHS Newborn and Infant Physical Examination Programme (NIPE).<sup>9</sup> Childhood screening programmes to detect disorders which cause VI exist in varying forms in most industrialised countries. In the UK, the National Screening Committee (NSC) agrees standards for and appraises the programmes for childhood vision screening, which currently comprises the examination of the eyes of all children in the first days following birth, and a second examination between the ages of 6 and 8 weeks as part of NIPE. Within the Healthy Child Programme (previously the Child Health Promotion Programme), screening by testing of acuity is undertaken at school entry age, of 4–5 years, to detect children with impaired vision—predominantly aimed at those with unilateral amblyopia (eg, secondary to refractive error and/or strabismus) as children with significantly reduced vision in both eyes are generally symptomatic and thus present earlier.

Clinical surveillance of groups at increased risk of VI is also an important strategy. In the UK, it is advocated that high risk groups such as children with neurological or neurodevelopmental disorders, or with systemic disorders with known ophthalmic associations, or with a family history of ocular disorders, or those with sensori-neural hearing loss should undergo targeted ophthalmic assessments.<sup>26</sup>

#### Tertiary prevention: managing the child with established visual loss

All children with established visual loss require specialist training and support for development, education and independent mobility in order to minimise the adverse impact of impaired sight. Thus, tertiary prevention of further burden from VI

involves continued ophthalmic input to limit the risk of further visual loss and provision of general care, support and training to minimise the potential impact of impaired vision. A key principle is that children with VI should be assessed and managed by a multidisciplinary team, which can be virtual rather than co-located, to ensure comprehensive and integrated intervention. The benefits to parents/families of a 'key worker' type service in which a dedicated professional provides information, support and liaison from the time around diagnosis are also well established.<sup>27</sup>

### Early developmental support

The importance of early, vision-specific developmental support for all children with VI is well established. In the UK, approaches include the use of the Department of Education Early Support Developmental Journal for babies and children with VI, which enables parents and carers to track their child's visual development in order to create an individualised framework identifying current and potential future needs.<sup>28</sup>

### Certification and registration of childhood visual impairment

Certification of a child's VI (by the ophthalmologist) enables registration of the child as sight impaired by Social Services or an equivalent governmental body, allowing the family improved access to educational and welfare support. Certification remains non-statutory in the UK, and although there is evidence that most eligible children are offered certification in a timely manner, variations in practice exist.<sup>29</sup> In some specialised tertiary ophthalmic units, certification of VI is facilitated by a 'key worker' for example, an Eye Clinic Liaison Officer, who is also the point of contact for families, providing information and assistance.

### Educational support

Certification is not a prerequisite for referral to childhood VI services or vision-related educational needs assessment in the UK. Early referral/notification of children with VI/SVI/BL and early involvement of a Peripatetic Visual Impairment Service/Peripatetic Teachers of Children with VI is recognised to be of value. A formal low vision aids assessment can be provided by ophthalmic services or by educationalists and covers aids such as hand-held or self-supporting magnifiers. More advanced adaptive technologies include video magnifiers, closed circuit television relays of printed material or electronic relay or material on laptop-based/tablet-based large font displays and voice activated/voice recognition and text to speech software. Simple strategies such as sitting a child nearer the whiteboard in the classroom can also be valuable. In the UK, currently half of school-aged visually impaired children receive education in mainstream schools with or without specialist visual resources, while, due to the significant population of children with multisystem disease, a third of children with SVI/BL attend schools for children with physical disabilities or learning difficulties.<sup>30</sup>

### SUMMARY

The population of visually impaired children in high, middle and lower income countries differ considerably between and within countries. The numerous and mainly uncommon disorders which can cause impaired vision results in heterogeneous population which includes a substantial proportion (for SVI/BL, the majority) of children with additional systemic disorders or impairments whose needs differ substantially from those with isolated vision impairment. Paediatricians and other paediatric professionals have a key role in early detection and

multidisciplinary management to minimise the impact of VI in childhood.

**Contributors** Both authors contributed to conception, design, data analysis and interpretation, and article drafting, revision and final approval.

**Funding** The Centre for Paediatric Epidemiology and Biostatistics at ICH benefits from funding support from the Medical Research Council in its capacity as the MRC Centre of Epidemiology for Child Health. This work was undertaken at UCL Institute of Child Health/Great Ormond Street Hospital for children which received a proportion of funding from the Department of Health's NIHR Biomedical Research Centres funding scheme. JR is supported in part by the National Institute for Health Research (NIHR) Biomedical Research Centre based at Moorfields Eye Hospital NHS Foundation Trust and UCL Institute of Ophthalmology. JR and ALS are members of the Ulverscroft Vision Research Group. The views expressed are those of the author(s) and not necessarily those of the NHS, the NIHR or the Department of Health. The funding organisations had no role in the design or conduct of this research.

**Competing interests** None.

**Provenance and peer review** Commissioned; externally peer reviewed.

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