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# Restoration of Severely Impaired Eyesight in an Adolescent with Down Syndrome and Bilateral Cataract in South Asia

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## Key Words

Down syndrome · Cataract · Low- and middle-income countries · International cooperation

## Abstract

**Purpose:** To report the recovery of visual function after phacoemulsification in a 16-year-old adolescent with Down syndrome (DS) from Saharsa, India. **Methods:** Interventional case report and short review of the pertinent literature. **Results:** A 16-year-old South Asian male with DS and bilateral cataract underwent successful surgery in both eyes after considerable difficulty for his parents including cross-border travel from India into Nepal. After the operation, the patient responded to visual stimuli (e.g. movement of hands) that had eluded him prior to surgery, and thus the additional obstacle to social participation imposed by the impaired eyesight could be removed. **Conclusions:** The establishment of basic ophthalmological surgery in developing countries is feasible and can yield extraordinary benefits for individual patients.

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## Introduction

Down syndrome (DS) is a frequent, genetically determined birth defect with substantial health and economic impacts around the world. Individuals with DS not only typically present with the pathognomonic symptom constellation of a reduced upper facial depth and length, epicanthal folds, short nose and low nasal bridge, downturned mouth and protruding tongue, dental abnormalities, small ears, wide neck and short hands with single palmar creases [1], but suffer from a – individually highly diverse – plethora of other ailments, most

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notably neurocognitive impairment [2, 3]. In contrast to children without genetic abnormalities, those with DS rely more heavily on social interaction when presented with cognitive challenges [2], and therefore they depend even more on intact communication functionality, i.e. eyesight, hearing, and speech. Children with DS face serious challenges trying to keep up with things on an intellectual level, and the frequent impairment of eyesight (e.g. due to astigmatism, strabismus, retinal abnormalities, hyperopia/myopia, amblyopia, nystagmus, and cataract) [4–7] raises the bar for social participation considerably.

A relatively infrequent but serious cause of vision impairment in children with DS is cataract, occurring in approximately 5–15% of afflicted patients [6, 7]. Whereas refraction abnormalities are relatively easy to remedy, cataract treatment requires ocular surgery that is frequently unavailable in developing and low- and middle-income countries. The author (C.-L.S.) has contributed to the development of eye clinics in Kenya, Myanmar, and Nepal. The adolescent was operated on by the coauthor (A.H.) in both eyes due to DS-related cataract in Lahan, Nepal.

### Case Report

A 16-year-old South Asian male with DS from Saharsa, India, grew up as the beloved son of a landless laborer and a housewife. Only 3 months before presentation, his parents realized that there might be an impairment of vision contributing to the general cognitive difficulties of the adolescent. Despite the relatively modest road distance of about 130 km, the Sagarmatha Choudhary Eye Hospital in Lahan, Nepal, is hard to reach for the villagers of the region; nevertheless, its reputation in the area is remarkable. The patient's parents knew about the facility and managed to overcome major difficulties (the journey took 16 h by train and bus and stressed the economic means of the parents to their very limits) to bring their son to Lahan for treatment. The adolescent was diagnosed with cataract in both eyes (fulfilling all clinical criteria [1] in the absence of the opportunity for genetic testing) and underwent surgery (phacoemulsification and implantation of a posterior chamber lens) in both eyes on 2 consecutive days (February 20 and 21, 2013, respectively). Surgery was successful from a macroscopic point of view (fig. 1, fig. 2), and the patient responded to visual stimuli (e.g. movement of hands) that had eluded him prior to surgery. The adolescent had not been able to contribute to domestic chores at all previous to surgery, but might be able to do so in the future due to the apparent improvement in eyesight.

### Discussion

Intact vision is a prerequisite for communication and cognitive development of children irrespective of other diseases and impairments, and the majority of conditions inducing vision impairment are curable by employing technically relatively simple procedures. The limited availability of such procedures is the main cause for the monumental dimension of the problem of global blindness that is focused on developing countries; out of an – probably too conservatively – estimated number of 37 million blind people in 2002, 80% lived in developing or least developed countries [8]. The WHO assumes about 75 million people will suffer from blindness by the year 2020 without well-directed measures [9, 10].

The present case report graphically illustrates the additional burden imposed on patients with DS by reduced eyesight. It has to be emphasized that the parents of the patient underwent a very considerable effort to get the cataract corrected almost as soon as they

learned about its existence in the first place; this effort comprised lengthy and expensive cross-border travel from India into Nepal. The motivation for this is in part economic. In least developed countries (such as Nepal and the India-Nepal border region), children and adolescents in general contribute substantially to their families' welfare (often not very effectively) without necessarily being in questionable or illegal forms of employment [11]. In the absence of government-provided remedies, parents have been known to undergo substantial efforts in order to improve their children's intellectual abilities [12].

This report also underscores the imperative necessity of eye care facilities in low- and middle-income countries and the huge catchment area of such facilities once they are instituted.

### Disclosure Statement

The authors have no conflicts of interest to declare. The authors have supported surgical eye care in developing countries since 1998 and were directly involved in the treatment of the patient described in this case report.

### References

- 1 Preus M: A diagnostic index for Down syndrome. *Clin Genet* 1977;12:47–55.
- 2 Fidler DJ: The emergence of a syndrome-specific personality profile in young children with Down syndrome. *Downs Syndr Res Pract* 2006;10:53–60.
- 3 Fidler DJ, Nadel L: Education and children with Down syndrome: neuroscience, development, and intervention. *Ment Retard Dev Disabil Res Rev* 2007;13:262–271.
- 4 Afifi HH, Abdel Azeem AA, El-Bassyouni HT, Gheith ME, Rizk A, Bateman JB: Distinct ocular expression in infants and children with Down syndrome in Cairo, Egypt: myopia and heart disease. *JAMA Ophthalmol* 2013;131:1057–1066.
- 5 Aslan L, Aslankurt M, Aksoy A, Altun H: Preventable visual impairment in children with nonprofound intellectual disability. *Eur J Ophthalmol* 2013, E-pub ahead of print.
- 6 Creavin AL, Brown RD: Ophthalmic abnormalities in children with Down syndrome. *J Pediatr Ophthalmol Strabismus* 2009;46:76–82.
- 7 da Cunha RP, Moreira JB: Ocular findings in Down's syndrome. *Am J Ophthalmol* 1996;122:236–244.
- 8 Pizzarello L, Abiose A, Ffytche T, Duerksen R, Thulasiraj R, Taylor H, Faal H, Rao G, Kocur I, Resnikoff S: VISION 2020: The Right to Sight: a global initiative to eliminate avoidable blindness. *Arch Ophthalmol* 2004;122:615–620.
- 9 World Health Organization: What is 'VISION 2020: The Right to Sight'? Geneva, WHO Press, 2007.
- 10 Yorston D: The global initiative vision 2020: the right to sight childhood blindness. *Community Eye Health* 1999;12:44–45.
- 11 UNICEF Division of Communication: Poverty and children. Lessons of the 90s for least developed countries. 2001. [http://www.unicef.org/publications/index\\_4424.html](http://www.unicef.org/publications/index_4424.html) (accessed August 19, 2013).
- 12 Einfeld SL, Stancliffe RJ, Gray KM, Sofronoff K, Rice L, Emerson E, Yasamy MT: Interventions provided by parents for children with intellectual disabilities in low and middle income countries. *J Appl Res Intellect Disabil* 2012;25:135–142.



**Fig. 1.** Pronounced opacity in the left eye of the adolescent patient with DS.



**Fig. 2.** Our patient after phacoemulsification and implantation of a posterior chamber lens.