CHILDHOOD BLINDNESS: CHALLENGES AHEAD

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Childhood blindness is an important cause contributing to the burden of blindness globally. Worldwide, 19 million children are visually impaired, of whom 1.4 million are irreversibly blind and need visual rehabilitation interventions for a full psychological and personal development[1]. Visual impairment in childhood has implications in all areas of a child's development. It poses educational, occupational and social challenges, with affected children being at risk of behavioral, psychological and emotional difficulties, impaired self-esteem and poor social integration. The pattern of childhood blindness varies depending on the accessibility of affordable health care services as well as socio-cultural factors. A major cause of childhood blindness in one country can be insignificant in another, and even over a decade the causes of childhood blindness can change quite dramatically in the same country[2,3]. The variations in the major causes of blindness in pediatric age group from different parts of the world is determined by socioeconomic factors and the availability of primary eye care services. In developed countries, lesions of the retina, optic nerve and higher visual pathways predominate as the cause of blindness, whereas corneal scarring is the major cause in low-income countries. Retinopathy of prematurity is an important cause in middle-income countries.

Early work by Rahi et. Al. (1993) showed that retinal dystrophies and albinism are emerging causes of childhood blindness"[4]. Similarly, Dandona and Dandona (2003), observed that 50% of the blindness were due to causes that are currently not treatable or preventable, of which a major proportion was of congenital eye anomalies and retinal degeneration[5]. Likewise, Gogate et. al. (2007), found that congenital anomalies and retinal disorders together accounted for more than 50% of the cases of blindness, which was higher than in a similar study conducted 10 years ago[6]. Another study by Bhattacharjee et al. (2008) observed that retina and optic nerve are amongst the most affected anatomical sites of visual loss[7]. Again Ozturk et. al. (2016) in agreement to previous studies, observed that in severe visual impairment (SVI) or blindness, the most common anatomic site is retina[2]. Latest research by Prakash et. al. (2017), observed that optic nerve atrophy and retinal dystrophy are the emerging causes of blindness underlining the need for genetic counseling and low vision rehabilitation centers, along with a targeted approach for avoidable causes of blindness[8].

The prevalence of so-called 'non-treatable' causes of childhood blindness amongst all studies is found to be relatively increased due to the significant reduction in the frequency of preventable causes of visual impairment and blindness, underlying the need for trained professionals, newer diagnostic techniques and multi-disciplinary approach. The exact reason for the changing trend is difficult to ascertain, but increased health services might have a role to play. There might be regional differences in the trends depending on whether the study is conducted in a rural population in remote areas or an urban setup with good access to health care facilities.

While optical coherence tomography (OCT) is crucial for accurate diagnosis and detailed analysis of structural anomalies [9], Next-generation genetic sequencing are emerging as a vital tool for accurate diagnosis and patient-tailored therapy as mutations in approximately 250 genes have been linked to cause inherited retinal degenerations with a high degree of genetic heterogeneity. New techniques in next-generation sequencing are allowing the comprehensive analysis of all retinal disease genes thus changing the approach to the molecular diagnosis of inherited retinal dystrophies. These new sequencing tools are highly accurate with sensitivities of 97.9% and specificities of 100%.[10]

More recently, data suggest that the prevalence of functional low vision (corrected visual acuity in the better

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eye ranging from <6/18 to, and including, light perception from untreatable causes) is approximately twice the prevalence of blindness: there are almost 3 million children worldwide who have the potential to benefit from low vision care. It is, therefore, essential that low vision services be part of eye care services for children at all levels of service delivery.[11]

Future Research

The U.S. Food and Drug Administration on December 19, 2017 approved Luxturna (voretigene neparvovec-rzyl), a new gene therapy, to treat children and adult patients with an inherited form of vision loss that may result in blindness. Luxturna is the first directly administered gene therapy approved in the U.S. that targets a disease caused by mutations in a specific gene. Luxturna is approved for the treatment of patients with confirmed balletic RPE65 mutation-associated retinal dystrophy that leads to vision loss and may cause complete blindness in certain patients.–[1214]

Retinal gene therapy clinical trials are underway for multiple genes including RPE65, ABCA4, CHM, RS1, MYO7A, CNGA3, CNGB3, ND4, and MERTK for which a molecular diagnosis may be beneficial for patients. Recent developments in genetic testing and gene therapy has now given new hopes for the diseases which were previously considered 'incurable'.

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