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Childhood Blindnessdue to Optic Nerve And Retinal Disorders

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Abstract: *The pattern of childhood blindness changes 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. 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.[1] This study will be aimed to find out the pattern of blindness due to optic nerve and retinal disorders in age group 0-16 years and address the patients through investigation, prevention, counselling, treatment, and low vision aids.*

Keywords: *childhood blindness, retina, optic nerve, visual pathways, corneal scarring, Retinopathy of prematurity*

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I. Introduction

Visual impairment in childhood has implications in all areas of a child's development. It possess educational, occupational and social challenges, with affected children being at risk of behavioral, psychological and emotional difficulties, impaired self-esteem and poor social integration. A visually impaired child is a great burden and challenge, not only to the family and society but also to the nation. There are variations in the causes of blindness and visual impairment across and within countries and the pattern changes with time.

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.[2] Reliable, population-based data on the causes of blindness in children are difficult to obtain in developing countries as registers of the blind do not exist, and very large sample sizes would be required for formal cross-sectional surveys.

II. Materials And Methods

This will be a cross sectional study conducted over a period of 18 months on 240 eyes of 120 patients, aged up to 16 years presenting to 'Retina Clinic' of the Institute of Ophthalmology, Aligarh Muslim University, Aligarh from July 2017 to December 2018.All the patients will be examined by a single examiner and the information will be collected on a predesigned proforma. Patients will undergo visual acuity estimation, external ocular examination, fundoscopy and fundus photography. Refraction, NCT, low vision workup, OCT, FFA, MRI will done where indicated. The patients will be divided into two groups for analysis, Group A will consist of patients with optic nerve disorders and Group B will consist of patients with retinal disorders. They will be further categorized on the basis of visual impairment according to the table given below.

Category	Presenting distance visual acuity	
	Worse than	Better than or equal to
0 - mild or no impairment		6/18
1- moderate visual impairment	6/18	6/60
2- severe visual impairment	6/60	3/60
3 - blindness	3/60	1/60 or CF at 1 m
4 - blindness	1/60	Perception of Light
5 - blindness	No light perception	

Categorization of visual impairment[3]

Inclusion criteria:

- All diagnosed cases of optic nerve disorders with reasonably clear media
- All diagnosed cases of retinal disorders with reasonably clear media
- Age 0-16 years

Exclusion criteria:

- The patients with media not clear (where fundus examination or photograph is not possible)
- Patients over the age of 16 years
- Non co-operative patients
- Unstable patients

III. Review Of Literature

The WHO has defined blindness as visual acuity worse than 3/60 in the better eye with best correction, the UNICEF defines childhood as lasting till the age of 16 years. A blind child is one who is aged less than 16 years and has a visual acuity in the better eye of <3/60 after best possible correction. However, many studies do not use this definition, which makes it difficult to compare the findings of different studies.

The drawback of this definition is the methodology followed for measuring visual acuity, particularly in population based studies, is to use a "pin hole" in patients whose "presenting" vision is below a certain cut off point (currently 6/18). The use of "best corrected" vision overlooks a large proportion of persons with visual impairment, including blindness, due to uncorrected refractive error, a common occurrence in many parts of the world. Uncorrected refractive error is now considered to be a major cause of visual impairment and estimations are under way to calculate the loss in terms of DALYs (disability-adjusted life years) resulting from this cause.

Another major drawback is the definition does not include monocular causes of visual impairment overlooks a large proportion of persons with visual impairment, including blindness in one eye.

The current definition does not make a distinction between those who have "irreversible" blindness (No perception of light) and those that have light perception but are still less than 3/60 in the better eye. The management of these two categories is different and categorization based on this would be useful.

"A person with low vision is one who has impairment of visual functioning even after treatment and/or standard refractive correction, and has a visual acuity of less than 6/18 to light perception, or a visual field of less than 10 degree from the point of fixation, but who uses, or is potentially able to use, vision for planning and/or execution of a task."

Under this definition persons who would benefit from low vision care also exist among those who are currently categorized as blind. This has led to miscalculations in the estimation of persons requiring LOW VISION care.

In 1993,**Rahi et al**[4]studied a total of 1411 severely visually impaired or blind (SVI/BL) children in 22 schools from nine states of India in different geographical zones and observed that Retinal dystrophies and albinism contributed for 19.3% of all the cases. In 2003, **Dandona and Dandona**[5]**,** studied 6935 children ≤15 years of age and observed that 50% of the blindness was due to causes that are currently not treatable or preventable, of which a major proportion was of congenital eye anomalies (16.7%) and retinal degeneration (16.7%).A study by **Gogate et al.**[6]in 2007, of1778 blind and severely visually impaired children (best corrected visual acuity <20/200 in the better eye, aged up to 16 years) in a total of 35 residential schools in Maharashtra, India and found that congenital anomalies (41%)and retinal disorders (11.2%) together accounted for more than 50% of the cases of blindness, which was higher than in a similar study conducted 10 years ago.

Changing pattern of childhood blindness between 1993 and 2005[6]

	1993 n (%)	**2005 n (%)**
Cornea	32 (20.4)	395 (22.2)
Lens	20 (12.7)	107 (6.0)
Retina	22 (13.8)	199 (11.2)
Anophthalmos/ Microphthalmos	27 (17.2)	735 (41.3)
Other causes	51 (35.9)	342 (19.3)
Total	152 (100)	1778 (100)

Bhattacharjee et al. (2008)[7], in their study of 258 students in schools for the blind in the northeastern states of India found that retina (5.8%) optic nerve (5.4%) are amongst the most affectedanatomical sites of visual loss.**Ozturk et al. (2016)**[8]**,** in a study of 11,871 patients who had SVI or blindness found the most common anatomic sites of SVI to be retina (24.6%). The prevalence of CVI was found to be relatively increased due to the significant reduction in the frequency of preventable causes of SVI.**Prakash et al. (2017)**[9], in their study of 302 students, observed that optic nerve atrophy and retinal dystrophy are the emerging causes of blindness and among the anatomical causes of blindness, the optic nerve was found to be the affected site in

24.8% cases, whereas retinal disorders were seen in 18.2% together which accounted for 43.0% of the cases, underlining the need for genetic counseling and low vision rehabilitation centers, along with a targeted approach for avoidable causes of blindness.

IV. Diagnosis And Evaluation

Diagnostic evaluation of visual impairment or blindness in children should always begin with thorough medical history, systemic examination and pedigree analysis asgenetic eye disease (GED) is one of the leading causes of blindness and includes disorders affecting all structures of the eye, such as albinism, corneal dystrophy, retinitis pigmentosa, Stargardt disease, and hereditary optic neuropathy.[10]

A systemic evaluation is necessary as many congenital anomalies of the retina and optic nerve may bea part of a larger systemic malformation syndrome, especially neurologic, endocrinologic and renal abnormalities e.g., Bardet-Biedl syndrome, Usher syndrome, Bassen-Kornzweig syndrome, Refsum disease, Leber Congenital Amaurosis, Alport syndrome,Aicardi syndrome, Stickler syndrome, etc. And many genetic syndromes like Marfan syndrome, Cri-du-chat syndrome, Tuberous sclerosis, von Hippel-Lindau syndrome have various ocular manifestations. [11,12]

Fundus photography andoptical coherence tomography (OCT) is crucial for accurate diagnosis and detailedanalysis of structural anomalies and their resulting pathologies, and their potential visual outcomes.[13]

Next-generation genetic sequencing: Inherited retinal dystrophies are a significant cause of vision loss and are characterized by the loss of photoreceptors and the retinal pigment epithelium (RPE). Mutations in approximately 250 genes 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%.[14,15]

V. Management

Maternal and Child Health (MCH) Services

Incidence of diseases like Retinopathy of prematurity, toxoplasma retinochoroiditis, congenital rubella syndrome (CRS) can be minimized by good antenatal care services like prevention of preterm child births, nutritional supplementation, immunization, identification and treatment of sexually transmitted infections, providing good post-natal care to preterm and low birth weight (LBW) babies. Adequate post-natal care is crucial for the early diagnosis and management of ROP.

Genetic Testing and Counselling

Genetic testing should be offered to patients with clinical findings suggestive of a Mendelian disorder whose causative gene(s) have been identified and the patient should receive counseling from a physician with expertise in inherited disease or a certified genetic counselor.[16]

Low Vision Services

Recent data suggest that the prevalence of functional low vision (corrected visual acuity in the better 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. Also,optic nerve atrophy and retinal dystrophy are the emerging causes of blindness. It is, therefore, essential that low vision services be part of eye care services for children at all levels of service delivery.[9,17–20]

Gene Therapy

The U.S. Food and Drug Administration on December 19, 2017 approved Luxturna (voretigeneneparvovec-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 biallelic RPE65 mutation-associated retinal dystrophy that leads to vision loss and may cause complete blindness in certain patients. [14,21,22]

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.[14]

VI. Results and Discussion

Magnitude

The prevalence of childhood blindness varies in line with the socioeconomic development and under-5 mortality rates (U5MR). In low-income countries with high under-5 mortality rates, the prevalence may be as high as 1.5 per 1000 children, while in high-income countries with low under-5 mortality rates, the prevalence is around 0.3 per 1000 children. The number of blind children in the world

is approximately 1.4 million. Approximately three-quarters of the world's blind children live in the poorest regions of Africa and Asia.[1]

The prevalence of blindness in the pediatric age group in India is estimated to be 0.8/1,000 with around 280,000 blind children.[23]"About 30 per cent of the blind in India are said to lose their eyesight before they reach the age of 20 years."[24]Due to the low 'prevalence' of blindness in children, accurate population-based data on the causes of childhood blindness are difficult to obtain in a developing country like India, as very large sample sizes would be required for cross-sectional surveys, which exponentially increases the cost, so much of the data used for estimating prevalence and etiology of childhood blindness comes from blind school surveys. Unfortunately, these may not be reflective of the true picture of magnitude of childhood blindness as they make up only a small proportion of the total blind in the community and children with multiple disabilities are unlikely to be enrolled in these schools. Other sources of information are examination of children identified as blind in community-based rehabilitation programs, special institution and tertiary care centers, but these too do not provide the real picture and possiblesources of bias must be borne in mind due to differences in the population under study.The standard reporting form for recording the causes of visual loss in children, developed by the International Centre for Eye Health, London for the WHO/PBL program[25] has been used in various states of India.

Accurate population-based data on the prevalence and causes of blindness in children is pivotal for planning and evaluating preventive and curative services. Information on the major causes of blindness in children is vital to plan effective programs for prevention and treatment of the same.[26]

VII. Conclusions

Recent studies indicate that incurable conditions such as optic atrophy, retinal dystrophies, and congenital anomalies including whole globe anomalies are emerging as the major cause of blindness and visual impairment in children.[9] This is in contrast to previous studies, as well as the data collected from underdeveloped countries, in which corneal causes, retinopathy of prematurity, and lens-related causes were important. 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. In the past couple of decades, diagnostic imaging such as OCT, FFA, MRI has emerged as a crucial tool for identifying and studying various retinal and optic nerve disorders. Recent developments in genetic testing and gene therapy has now given new hopes for the diseases which were previously considered 'incurable'.

Abbreviations

CVI: Cerebral Visual Impairment
FFA: Fundus Fluorescein Angiography
MRI: Magnetic Resonance Imaging
NCT: Non-contact Tonometry
NPCB: National Programme for Control of Blindness
OCT: Optical Coherence Tomography
SVI/BL: Severe Visual Impairment/Blindness
UNICEF: United Nations Children's Fund
WHO: World Health Organization
WHO/PBL: World Health Organization/Prevention of Blindness

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