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A study of etiological causes of severe visual impairment and blindness in children of Western Rajasthan

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Abstract

Childhood blindness are group of conditions that occurs in childhood or early adolescence (<16 years of age), and if left untreated, results in severe visual impairment or blindness that is likely to be not treatable later on. The prevalence ranges from 0.3 per 1000 children aged between 0 and 15 years in prosperous countries to 1.5 per 1000 children in extremely poor communities.

Materials and Methods: This study was done at Department of Ophthalmology at Mathura Das Mathur Hospital, Jodhpur. It is non randomized, observational study conducted from January 2015 to June 2016 (18 months) and all blind or severely visually impaired children with less than 16 years age attending Ophthalmology OPD or children living in blind schools near and within Jodhpur city were enrolled after receiving informed written consent by their parents/guardian.

Results: In this research study, we found that hereditary causes were accountable for blindness in 160(56.34%) patients but we were not able to specify any inheritance pattern in 153(53.87%) patients. Intrauterine factors such as rubella and toxoplasmosis were responsible in 6(2.11%) patients. Perinatal/Neonatal factors such as cerebral hypoxia/injury and ROP were responsible in 6(2.11%) patients. Postnatal factors such as trauma, measles, neoplasms and meningitis were responsible in 14(4.93%) patients. In 98(34.51%) cases factor/factors causing cataract and glaucoma could not be determined but probably genetic factors appear to be responsible.

Conclusions: We concluded that the most important causes of preventable blindness were genetic in 160(56.34%) patients followed by TORCH infections in 6 (2.11%), trauma in 4(1.41%), measles in 4(1.41%) and meningitis in 3 patients i.e. 1.06%. The most significant cause of disease which can be treated was cataract in 76(26.76%) patients followed by glaucoma in 12(4.23%) and ROP in 2(0.70%) patients. Timely recognition and treatment is necessary to prevent advancement of dense amblyopia.

Keywords: Childhood blindness, Hereditary causes of blindness, Severe visual impairment

Introduction

Childhood blindness are group of conditions that occurs in childhood or early adolescence, i.e. <16 years of age, and if left untreated, results in severe visual impairment or blindness that is probable to be not treatable later on.[1] The prevalence ranges from 0.3 per 1000 children aged between 0 and 15 years in prosperous countries to 1.5 per 1000 children in extremely poor communities.[2]

Blindness in children is around 10 times lower than adults, but it remains on a high priority for the reason that of the expected number of years to be lived in with this disease.

Childhood blindness is more common in deprived regions due to two reasons: (a) there are risk factors and diseases which can lead to blindness from causes that used to occur in industrialized countries earlier (example, Vitamin A deficiency, measles, malaria, ophthalmia neonatorum (b) there are smaller amount of well equipped eye departments with ophthalmic paramedics, nurses and ophthalmologists trained in managing treatable causes of blindness (example, glaucoma and cataract). The incidence is therefore higher, and fewer blind children have their sight restored. On the whole, in India, there are most likely 280,000–320,000 blind children. In developed countries, there are approx 60 blind children per million total population, on the contrary in India they are likely to be between 100 and 400.[3]

WHO classified causes of childhood blindness depending upon the aetiology in following groups:[4]

1. Intrauterine causes (at the time of pregnancy): rubella
2. Hereditary causes (at conception): genetic and chromosomal abnormalities
3. Childhood causes: trauma, measles and vitamin A deficiency
4. Prenatal causes: birth injury, retinopathy of prematurity and neonatal conjunctivitis/ophthalmia neonatorum)
5. Unknown in which cause cannot be determined

In poor countries complications of ophthalmia neonatorum, measles, harmful traditional eye medicines and Vitamin A deficiency were common causes of blindness.

Uncorrected refractive errors are major contributor to the visual disability and could be managed or treated easily. Retinopathy of Prematurity (ROP) and congenital cataract can be treated as well as prevented. Modalities to diagnose, prevent and manage these conditions are available.[5,6] Visual prognosis after cataract surgery has improved significantly, especially

in young children. Across the globe, ROP is the 5th foremost cause of childhood blindness. But luckily, infants who are at risk, screened and treated for ROP have better structural as well as functional results.

ICEH (International Centre for Eye Health), London, developed the standard reporting form for recording the causes of visual loss in children, for the WHO/PBL program.[7]

Since, in developing world insufficient data is available on prevalence and causes of ocular morbidities in children. So, we conducted this study to estimate the prevalence and causes of ocular morbidity amongst children so that we can plan interventional measures to reduce avoidable blindness.

Materials and Methods

The research was conducted for providing the data on the major causes of severe visual impairment in children of Western Rajasthan using the WHO standard reporting form.

This study was conducted for 18 months, i.e. from Jan. 2015 to June 2016, at Ophthalmology Department, Mathura Das Mathur hospital under Dr S. N. Medical College, Jodhpur. Ophthalmology Department at Mathura Das Mathur hospital is a tertiary care centre with facilities for diagnosis and management of common eye disorders.

Children <16 years of age coming to eye OPD of Mathura Das Mathur hospital and associated group of hospitals with visual problems and found to be blind or severe visual impairment were enrolled for the study. Children living in blind schools within and near Jodhpur were also enrolled for this study to get sufficient size of the sample.

The permission for examination of the children was obtained from the school principal. Children were enrolled in the study after getting informed written consent from parent/guardian.

Blindness is defined "as presenting visual acuity in the better eye of less than 0.05 (3/60; 10/200) and severe visual impairment as presenting visual acuity in better eye of less than 0.1 (6/60; 20/200) to 0.05 (3/60; 10/200) 'functional' low vision as presenting visual acuity in the better eye of less than 0.3 (6/18; 20/60) to light perception".

Students of the blind school who are aged <16 years have been included. Whenever possible, the appropriate information was collected from parents and teachers. Medical and family history and brief demographic details of each child were recorded.

Exhaustive eye examination of children was conducted. In one case, through Snellen tumbling E visual acuity test chart, visual acuity was assessed and recorded using WHO categories of visual impairment before and after refraction. In another case, children were assessed for the ability to percept and follow light. To classify a child under low visual category, functional vision tests were used. They were, the capability to navigate approximately two chairs set two meters away from each other, unaided with a visual acuity of <20/60 to light perception and to recognize faces at a distance of three meters.

Anterior segment of eye was examined using torch light or slit lamp and the posterior segment was examined using indirect and direct ophthalmoscope after dilatation of pupil. Examination findings were recorded in standard form, WHO/PBL Eye Examination Record for Children with low vision and blindness.

The etiological factor is identified and classified depending on the onset time of the condition leading to blindness (intrauterine, hereditary, childhood, prenatal and unknown).

Statistical analysis: Appropriate statistical tools and technique were applied as per data collection.

Inclusion criteria:

1. Children aged 0 to 15 years.
2. Children with finest corrected visual acuity of <6/60 (equal to 6 m finger count).
3. Children whose parents/ guardians agree or give consent for the study.

Exclusion criteria:

1. Children with low vision and finest corrected visual acuity >6/60 in better eye.
2. Children with uniocular blindness.
3. Parents/ guardians did not agree or refused consent for study.

Results

Table 1: Age (at the onset of visual loss) and sex wise distribution of blind children

Time of visual loss	Males (%)	Female (%)	Total (%)
Since birth (Hereditary/Intrauterine)	164(57.75%)	62(21.83%)	226(79.58%)
First year of life (Perinatal/New born/Infancy)	05(01.76%)	02(00.70%)	07(02.46%)
1 – 15 year (Childhood)	28(09.86%)	12(04.23%)	40(14.08%)
Unknown/Undetermined	08(02.82%)	03(01.06%)	11(03.87%)
Total	205(72.18%)	79(27.82%)	284(100%)

Table 2: Blind school Vs Outdoor patients

	Males (%)	Females (%)	Total (%)
From Blind School	47(16.55%)	09(03.17%)	56(19.72%)
Hospital OPD Patients	158(55.63%)	70(24.65%)	228(80.28%)
Total	205(72.18%)	79(27.82%)	284(100%)

Table 3: Family history of similar condition in any other family member

	Males (%)	Females (%)	Total (%)
Positive family history	31(10.92%)	16(05.63%)	47(16.55%)
Negative family history	174(61.27%)	63(22.18%)	237(83.45%)
Total	205(72.18%)	79(27.82%)	284(100%)

Table 4: Categories of visual impairment and blindness

Category	Males (%)	Females (%)	Total (%)
Severe visual impairment	21(07.39%)	05(01.76%)	26(09.15%)
Blind	184(64.79%)	74(26.06%)	258(90.85%)
Total	205(72.18%)	79(27.82%)	284(100%)

Table 5: Aetiological classification of vision loss in children with blindness

Cause	Males (%)	Females (%)	Total (%)
Hereditary disease	115(40.49%)	45(15.85%)	160(56.34%)
a. Chromosomal	00	01(00.35%)	01(00.35%)
b. Mitochondrial	00	00	00
c. Autosomal dominant	01(00.35%)	00	01(00.35%)
d. Autosomal recessive	03(01.06%)	02(00.70%)	05(01.76%)
e. X linked	00	00	00
f. Cannot specify	111(39.08%)	42(14.79%)	153(53.87%)
Intrauterine factor	04(01.41%)	02(00.70%)	06(02.11%)
a. Rubella	03(01.06%)	01(00.35%)	04(01.41%)
b. Toxoplasmosis	01(00.35%)	01(00.35%)	02(00.70%)
c. Drug/Alcohol	00	00	00
d. Others	00	00	00
Perinatal/Neonatal factor	05(01.76%)	01(00.35%)	06(02.11%)
a. Cerebral hypoxia/Injury	03(01.06%)	01(00.35%)	04(01.41%)
b. ROP	02(00.70%)	00	02(00.70%)
c. Ophthalmianeonatorum	00	00	00
d. Others	00	00	00
Postnatal/Infancy/Childhood (1-15Y) factor	11(03.87%)	03(01.06%)	14(04.93%)
a. Vitamin A deficiency	00	00	00
b. Measles	04(01.41%)	00	04(01.41%)
c. Neoplasms (Cerebral tumours)	02(00.70%)	01(00.35%)	03(01.06%)
d. Trauma	03(01.06%)	01(00.35%)	04(01.41%)
e. Traditional harmful practices	00	00	00
f. Others (Meningitis)	02(00.70%)	01(00.35%)	03(01.06%)
Cannot determine/Unknown aetiology	70(24.65%)	28(09.86%)	98(34.51%)
a. Cataract	56(19.72%)	20(07.04%)	76(26.76%)
b. Glaucoma/Buphthalmos	07(02.46%)	05(01.76%)	12(04.23%)
c. Retinoblastoma	00	00	00
d. Abnormality since birth	04(01.41%)	02(00.70%)	06(02.11%)
e. Others	03(01.06%)	01(00.35%)	04(01.41%)
Total	205(72.18%)	79(27.82%)	284(100%)

Discussion

Children suffering with any kind of blindness need to be identified timely so that they can be examined, treated and referred. This is essential if they want to have the best possible chance of proper childhood development, education, and participation inbroader social life. In order to set priorities in control programs of blindness, baseline epidemiological data of the occurrence and main causes of childhood blindness are necessary. Indeed, it is essential to identify major treatable and preventable causes in each country and to monitor the changing patterns over the period of time.

Nearly 75 percent of early learning comes from vision. Early start of visual loss can have consequences on a child's social, motor, psychological and emotional development.[8] If this deficit is not treated on time, permanent visual deficit and amblyopia can occur. Hence, early diagnosis and timely treatment is important.

In our study, out of total 284 enrolled patients 205(72.18%) patients were males and 79(27.82%) patients were females. This difference is probably due to the greater value accorded to male children in this part of the countryand visit hospitals and blind schools to seek medical treatment or rehabilitate them.

In our study, 226 (79.58%) patients were blind since birth due to hereditary or intrauterine factors, 7(2.46%) patients developed blindness in first year of life and 40(14.08%) patients developed blindness in between 1-15 years of age. We were not able to determine exact time of onset of blindness in 11(3.87%) patients.

Out of total enrolled patients 228(80.28%) patients were enrolled from the patients visiting outdoor of MDM hospital. Rest 56(19.72%) patients were enrolled from blind schools present in vicinity of Jodhpur, Rajasthan.

Only 10 percent of the blind children attend blind schools in developing countries.[9] Inaccessibility to the schools and ignorance and unawareness of parents are main factors amongst those residing in remote and deprived areas. A culture of scepticism and mistrust exists in some village and tribal communities regarding such centres, which further hinders access.[10]

There are some biases in any study of children from blind school. This is due to causes of blindness in children with numerous disabilities, deaths of some children, lower socioeconomic areas, and those from rural communities are likely to be under-represented in schools for the blind compared with population-based studies.[11] Most of the blind schools do not give admission to children below 5 years of age, so we need to enrol patients visiting OPD of our hospital along with blind children studying in school.

Positive family history of similar eye condition was present in 47(16.55%) patients. Most of the patients had positive family history in their siblings. Family history was negative in rest 237(83.45%) patients.

History suggestive of consanguinity in parents was present in only 13(4.58%) patients. In our study 271(95.42%) of patients were born to parents who were not had history suggestive of consanguineous marriage. Consanguineous marriages are not common in this part of country as compared to southern India. In a study conducted by Gogate et al from south India 32% cases of blindness had history suggestive of consanguineous marriage in parents.[12]

Out of total enrolled patients, 258(90.85%) patients were blind. According to WHO criteria, they had vision less than 3/60-PL or believed blind if aged less than 3 years (if vision could not be tested) in better eye. Severe visual impairment was present in 26 (9.15%) patients and it is considered, if child had vision less than 6/60-3/60 in better eye.

WHO classifies the causes of blindness in children according to the most affected anatomical site and the basic etiology.[13]

Hereditary causes were responsible for blindness in 160 (56.34%) patients but we were not able to specify any inheritance pattern in 153(53.87%) patients. Autosomal recessive mode of inheritance was present in 5(1.76%) patients. Autosomal dominant pattern of inheritance and chromosomal defects each was identified in 1(0.35%) patient.

Intrauterine factors were responsible in 6(2.11%) patients. Maternal rubella was positive in 4(1.41%) patients and toxoplasmosis was positive in 2(0.70%) patients. Prenatal/Neonatal factors were responsible in 6(2.11%) patients. Cerebral hypoxia/injury was present in 4(1.41%) patients. Blindness was due to ROP in 2(0.70%) patients. Postnatal factors were responsible in 14(4.93%) patients. Trauma and measles each was responsible in 4(1.41%) patients. Cerebral neoplasms and meningitis each was responsible in 3(1.06%) patients. Aetiology was unknown in 98(34.51%) cases.

In studies conducted in Asia, genetic diseases were responsible for 42% of severe visual impairment and blindness in Indonesia,[14] 35.0% in Sri Lanka,[9] 16.8% in Thailand and Philippines,[15] 23.0% in India,[16] 30.7% in China,[17] and 29.5% in Malaysia.[18] In our study, it was responsible for 56.34% of cases.

With the availability of newer diagnostic modalities of genetic disorders, prenatal diagnostic techniques and genetic counseling it is now possible to identify the genetic defects leading to congenital globe abnormalities, congenital and developmental cataract, inherited retinal disorders and congenital glaucoma.

It is not difficult to diagnose and detect severe visual impairment and blindness in infants. With proper treatment and care, formation of dense amblyopia can be prevented. Even if the ophthalmologist may not be able to help surgically or medically, rehabilitation and optical aids can help children reach their full capacity.

Conclusions

We found that hereditary causes were responsible for blindness in 160(56.34%) patients but we were not able to specify any inheritance pattern in 153(53.87%) patients. Intrauterine factors such as rubella and toxoplasmosis were responsible in 6(2.11%) patients. Perinatal/Neonatal factors such as cerebral hypoxia/injury and ROP were responsible in 6(2.11%) patients.

Postnatal factors such as trauma, measles, neoplasms and meningitis were responsible in 14(4.93%) patients. In 98(34.51%) cases factor/factors causing cataract and glaucoma could not be determined but probably genetic factors appear to be responsible.

We found that the most important causes of preventable blindness were genetic in 160(56.34%) patients followed by TORCH infections in 6(2.11%), trauma in 4(1.41%) measles in 4(1.41%) and meningitis in 3(1.06%) patients. Most significant treatable cause of blindness was cataract in 76(26.76%) patients followed by glaucoma in 12(4.23%) and ROP in 2(0.70%) patients.

Genetic counselling to educate individuals about hereditary diseases and their mode of transmission should be intensified and detailed genetic studies should be done to aid more precise diagnosis of these inherited disorders.

The significance of congenital cataract is escalating as a cause of blindness amongst children in developing countries. Blindness from corneal scarring appears to have dropped because of interventions like vitamin A supplementation, immunization, health education/ nutrition, and breast feeding practices.

Early detection and treatment is crucial to check development of dense amblyopia. Primary health care programs should incorporate a wide range of cost-effective services such as eye examination at birth, eye screening for pre-school and school children, early management of congenital cataracts, vaccination for infectious diseases in children, and initiatives to train health workers.

Early referral to a tertiary care center should be done so that treatable causes of blindness can be treated timely and amblyopia can be prevented. Optical or low vision services should also be available to maximize rehabilitation of the patients so that they can live independently and interact freely with their sighted peers.

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