ORIGINAL ARTICLE ASIAN JOURNAL OF MEDICAL SCIENCES

Pattern of congenital eye anomalies in children: A study from rural tertiary care hospital

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Submission: 07-11-2021 Revision: 05-01-2022 Publication: 01-02-2022

ABSTRACT

Background: Congenital eye anomalies are an important cause of childhood blindness. Worldwide, there are a lot of variations in the spectrum of congenital eye anomalies. The key to preventing childhood blindness is early detection and intervention of the congenital eye anomalies. **Aims and Objectives:** This study aims to describe the type and frequency of congenital eye anomalies in children. **Materials and Methods:** This cross-sectional study was done on children attending pediatric OPD and the newborn babies born in the hospital in the period of 1 year from June 2019 to May 2020. They were screened for any ocular abnormality and then detailed ocular examination was done by the ophthalmologist. The data were collected for demographic characteristics, clinical diagnosis, age of presentation of anomaly, laterality, involvement of one or both eye, and type of anomaly. Variables were expressed as proportions or percentages. **Results:** A total of 32,858 children were screened for ocular abnormalities. Out of these, 1571 (0.047%) children had ocular abnormalities. Among them, 1011 (64.35%) presented in the 1st year of life, 381 (24.25%) in the 2nd year of life, and the remaining 179 (11.4%) were diagnosed after 2 years of life. Among various anomalies, congenital nasolacrimal duct obstruction was the most common anomaly (65%) followed by coloboma of iris and choroid (13.8%). Congenital ectropion and anopthalmos were found in five and three children, respectively. **Conclusion:** The most common anomaly was congenital nasolacrimal duct blockage followed by coloboma of iris, choroid, and cataract. Early detection and intervention of the congenital eye anomalies is necessary to prevent blindness.

Key words: Childhood blindness; Congenital; Early diagnosis; Eye anomalies

Access this article online

Website: http://nepjol.info/index.php/AJMS

DOI: 10.3126/ajms.v13i2.40720

E-ISSN: 2091-0576

P-ISSN: 2467-9100

INTRODUCTION

To begin with, there are 36 million people in the world are blind.[1] Congenital eye abnormalities are a leading cause of childhood blindness,[2] accounting for approximately 60% of all cases.[3] In the age category of 0–14 years, 1.4 million children throughout the world are blind.[4] Congenital abnormalities of the eyes are the leading cause of childhood blindness worldwide. In children, ocular abnormalities account for 15–20% of blindness and severe vision impairment.[5] Early onset of blindness in a child results in a large number of blind years, which has a substantial socioeconomic impact.[6-8]

Any anomaly in the eye and surrounding tissue can occur on its own or as part of a multisystem miscreation syndrome. The cause of most of these abnormalities is unclear, but genetic and environmental factors both play a role.[9-11] The specific genetic location of many eye abnormalities is known, but for many others, it is unknown. Ocular miscreations can be caused by both germline and somatic mutations.

Ocular congenital abnormalities include malformation, distortion, disruption, and dysplasia. Congenital cataract and glaucoma are the most prevalent diseases in poor

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countries, and they are frequently caused by preventable causes.[12,13] Anophthalmos, microphthalmos, and coloboma are the most common congenital abnormalities in affluent nations, and are primarily due to unavoidable reasons.[14] Congenital abnormalities are most commonly seen at birth, and in certain cases, even in pregnancy, however, the age of onset might vary. Maternal factors, in addition to hereditary ones, have a major influence. Maternal malnutrition, diseases such as rubella, CMV, toxoplasmosis, and varicella, as well as a history of drug use and alcohol consumption, all have a role.

In this area, the literacy rate is very low with poor health awareness and there are high chances of consanguinity. There is coexistence of maternal malnutrition and infections. All these contribute to high chances of congenital abnormality. Hence, this study was planned to find out the spectrum of congenital eye anomalies.

Aims and objectives

The study was conducted with the aim and objective to describe the type and frequency of congenital eye anomalies.

MATERIALS AND METHODS

This cross-sectional observational study was carried out at the Department of Pediatrics, SHKM, GMC, Nalhar, Nuh, Haryana, in coherence with the Department of Ophthalmology. Ethical approval was obtained from the Institutional Ethics Committee of our institute and informed consent was taken before performing all procedures. All the patients attending pediatric OPD and the newborn babies born in the hospital in the period of 1 year from June 2019 to May 2020 were included in the study. They were screened for any ocular abnormality. Exclusion criteria for the study were the cases of retinopathy of prematurity and retinoblastoma. The data were collected for demographic characteristics, clinical diagnosis, age of presentation of anomaly, laterality, involvement of one or both eye, type of anomaly, and any associated systemic complaints.

First and foremost, clinical examination was done by the pediatrician and all the children with suspected ocular abnormalities were referred to the ophthalmology department. The detailed ocular examination was done by the ophthalmologist which included broad light examination, slit-lamp examination, fundus examination, and IOP recording with applanation tonometer whenever possible. Vision examination was done in detail along with wet retinoscopy, auto-refractokeratometer, and refraction.

Statistical analysis

Quantitative variables, demographic variables, were expressed as a number and frequency, while qualitative variables were expressed as proportions or percentages.

RESULTS

The total pediatric OPD from June 2019 to May 2020 was 31,258 and total live births were 1600. Hence, a total of 32,858 children were screened for ocular abnormalities. Out of these, 1571 (0.047%) children had ocular abnormalities. The frequency of various congenital anomalies with sex wise distribution is depicted in Table1.

Hence, 0.047% of the children attending the hospital and born in the hospital were having ocular abnormalities. Out of these, 902 were male children which comprised 57.42% of the affected children. The remaining 669 were female children which comprised 42.58% of the affected children.

Out of 1571 children, 1011 (64.35%) presented in the 1st year of life. Three hundred and eighty-one (24.25%) of these were diagnosed in the 2nd year of life, the remaining 179 (11.4%) were accidently diagnosed after 2 years of life. These 11.4% had visited hospital for other reasons and were incidentally diagnosed with ocular abnormalities. The most common eye anomaly was congenital NLD blockage (65%) followed by coloboma of iris and choroid (13.8%) depicted in Figures 1,2. Congenital ectropion and anopthalmos were found in five and three children, respectively. Congenital esotropia was detected in 23 (1.46%) cases (Figure 3) and congenital ptosis was observed in 8 (0.5%) children (Figure 4).

Many of these children were managed very well in our center and only few were referred to higher center for further management.

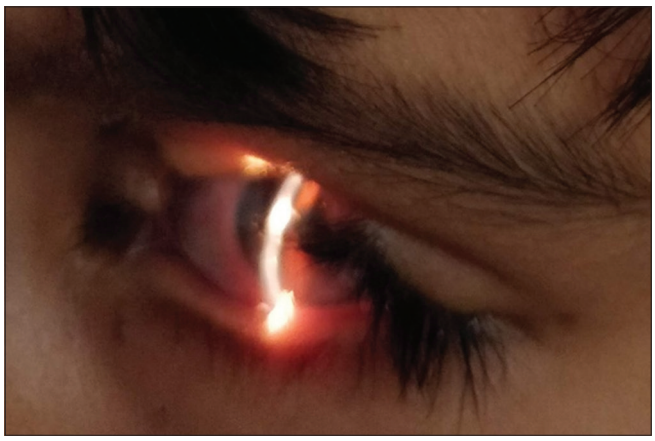

Figure 1: Lid coloboma

Table 1: Distribution of congenital eye anomalies

S. No.	Eye anomaly	Total children, no. (%)	Male children	Female children
1.	Congenital NLD blockage	1021 (65%)	578	443
2.	Coloboma of iris and choroid	218 (13.8%)	118	100
3.	Congenital cataract	167 (10.6%)	102	65
4.	Congenital corneal opacity	35 (2.2%)	22	13
5.	Abnormalities of the Iris	33 (2.1%)	21	12
6.	Heterochromia iridium	31 (1.97%)	19	12
7.	Congenital esotropia	23 (1.46%)	14	9
8.	Microphthalmos with microcornea	18 (1.14%)	11	7
9.	Congenital ptosis	8 (0.5%)	6	2
10.	Minor lid abnormalities (symblepharon, ankyloblepharon, and epicanthal folds)	9 (0.57%)	6	3
11.	Congenital ectropion	5 (0.3%)	3	2
12.	Anophthalmos	3 (0.2%)	2	1

Figure 2: Bilateral limbal dermoid with iridochoroidal coloboma

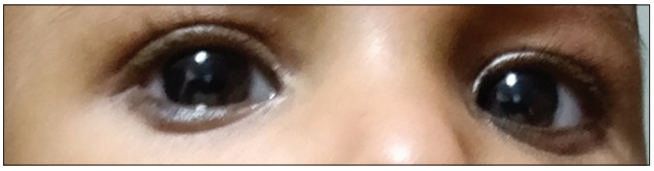

Figure 3: Esotropia

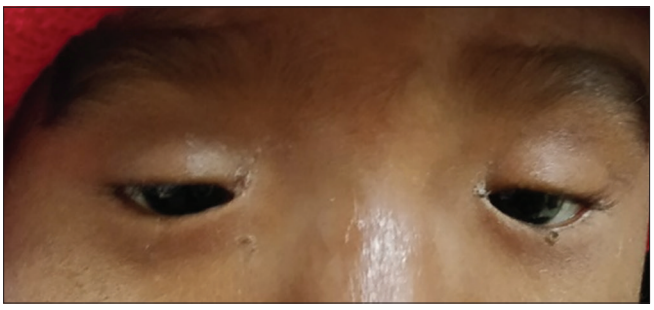

Figure 4: Congenital ptosis (lid) abnormalities

DISCUSSION

This research was carried out in a tertiary hospital in one of India's most impoverished regions. Muslims make up the majority of the population. All patients are directed to this medical college and hospital since it is the only tertiary care facility within a 100 km radius. The data represent the real data of the region because it is the only tertiary hospital in the area. There is a low literacy rate, as well as a lack of health knowledge and a significant risk of consanguinity. There is coexistence of maternal malnutrition and infections. All these contribute to high chances of congenital abnormality. Ocular morbidity and death can be greatly decreased if these instances are detected and treated early.

The prevalence of congenital eye anomalies was found to be 0.047% in our study area. In a study done by Parag and Sagar, the prevalence was reported to be 0.75%.[15] In our study, males made up 57.4% of the patients, while females made up 42.6%. This matches the findings of Behera et al., who found that out of 60 patients, 35 (58.3%) were male and 25 (41.7%) were female.[16] Other studies have also revealed a male preponderance.[12,15] We discovered that 64% of patients were under the age of 1 year, with 24% in the age bracket of 1–2 years. This is comparable to another study done in Indian children, where 54% of the cases were children under the age of 2 years.[15] It is possible that this is due to the early identification of congenital abnormalities.

Among the ocular anomalies, the most common anomaly in our study was congenital nasolacrimal duct blockage (65%), followed by coloboma of iris, choroid, and cataract. Other two studies in Indian patients found similar results, with the most common ocular abnormality being dacryocystitis (24%) and (33.3%), respectively.[15,16] Another study showed that microphthalmos, microcornea, and iris coloboma (67%) were the most common congenital ocular anomalies,[17] while in this study, microphthalmos with microcornea was observed in 18 (1.14%) patients. In our study, anophthalmos accounted for 0.2% of all congenital anomalies. In a study, Bermejo and Martines-Frias reported a 5% prevalence of anophthalmos.[14]

Eye anomalies are usually difficult to diagnose for the non-specialist. In order to prevent ocular morbidities, pediatricians need to be aware of the importance of these anomalies and report them to ophthalmologist at birth. Greater coordination of these two specialties can prevent many cases of blindness.

Limitations of the study

This study restricts to the pattern of congenital anomalies; we could not assess the etiological correlation with anomalies.

CONCLUSION

It is necessary to discover the spectrum of congenital ocular abnormalities as well as the etiological diagnosis of patients to allow efficient prevention and health promotion. The key to preventing childhood blindness is early detection and intervention of the congenital eye anomalies. Non-specialists sometimes have difficulty diagnosing eye abnormalities. For preventing ocular morbidity, it is essential to understand the relevance of these abnormalities. It is also important for the Paediatrician to understand the significance of referring such children to the ophthalmologist at birth. Patient with congenital ocular anomalies and their parents should be informed about their visual prognosis, possible current therapies, and genetic counseling.

ACKNOWLEDGMENT

The authors acknowledge Department of Pediatrics ad Ophthalmology, SHKM,Nuh for giving there valuable support in undertaking this study.

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Authors Contribution:
RJ- Concept and design of the study, prepared first draft of manuscript; **VP**- Preparation and drafting of manuscript; and **NS**- Concept, collection of data, preparation of manuscript, and revision of the final manuscript

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Source of Support: Nil, **Conflict of Interest:** None declared.