

Original Research Article

Study on ocular anomalies in pediatric patients with special reference to congenital anomalies

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Received: 27 January 2021

Accepted: 03 March 2021

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ABSTRACT

Background: Aim of the study was to study the proportion, clinical and etiological profile of congenital ocular anomalies in pediatric patients and limit its magnitude wherever possible so that preventable blindness can be dealt effectively.

Methods: Present study was carried out in AMCH, Dibrugarh during the period of July 2018-June 2019. Total 1850 pediatric cases were examined, VA testing was done by Snellen's chart or clinical judgment by HM, PL and PR also with Allen pictures in preschool children and in infants' pupillary response and OKN were done. Examination under general anesthesia was done when necessary for posterior segment evaluation and IOP estimation.

Results: Proportion congenital ocular anomalies were found to be 13.72%. Male:female ratio was found to be 1.8:1 where majority of cases were within 4 years of age. Most common congenital anomaly was congenital cataract followed by uveal coloboma, microphthalmos and anophthalmos etc. Majority of congenital cataract were in first birth rank and in 4.33% cases parents gave history of consanguinity.

Conclusions: The prevalence of congenital anomaly varies in different aspects like age, sex, dwelling, socioeconomic class etc., factors like maternal infection, medication during pregnancy and maternal nutrition may have influence on production of congenital anomalies. Treatable congenital anomaly like congenital cataract, congenital glaucoma, eyelid coloboma etc. should be managed by ophthalmologist. Various demographic and mainly maternal factors which are found in study give a clue for further study on those group of people to get a conclusion for the etiology of ocular congenital anomalies concerned.

Keywords: Ocular malformation, Birth defect, Congenital anomaly, Chromosomal malformation, Cataract, Coloboma, Microphthalmos, Iris coloboma, Optic nerve hypoplasia

INTRODUCTION

Congenital eye anomalies are major cause of childhood blindness and account for 60% of cases.¹ Understanding the prevalence and types of childhood blindness provides useful information for its prevention. Major abnormalities like microphthalmos, anophthalmos and colobomas accounts for 16.7% childhood blindness globally.¹ Congenital anomalies are mostly present long before the time of birth, some in the embryonic period (up to the 7th week of gestation) and others in the fetal period (8th week

to term). Defect arising in germinal period due to some teratogen are not concerning as they lead to gross abnormality, which seldom or never comes to term. Those arising the organogenesis period form the bulk of well-known abnormalities of eye.

The congenital anomalies cover all the major classes of abnormalities of development which occur due to malformation, deformation, disruption and dysplasia. Visual loss in childhood affects all aspect of child's development i.e., educational, occupational and social

challenges. There is also risk of behavioral, psychological, emotional difficulties, impaired self-esteem and poor social integration. It is believed that overall, multifactorial etiology account for 20-25% of all abnormalities, 6-8% is monogenic, that is caused by mutations in the single gene, 6-8% by chromosomal abnormalities, and 6-8% by environmental factors such as maternal illness, infections, drugs, radiation and alcohol.² Major cause is maternal infection during pregnancy, caused by some important infectious agents as follows.² Rubella, Varicella, Cytomegalovirus, Toxoplasmosis. Worldwide there is inter-regional variation in the spectrum of congenital eye anomalies. In the developing nations, congenital cataract and glaucoma are the most common anomalies and are often attributable to avoidable causes.³ In contrast, in developed countries, anophthalmos, microphthalmos and coloboma (AMC) are the leading anomalies seen at birth and are mainly due to unavoidable causes.

Purpose of the study was to study the proportion and clinical profile of congenital ocular anomalies in paediatric patients with congenital ocular anomalies.

METHODS

The present study is a hospital based cross-sectional study, conducted in the department of ophthalmology, Assam medical college and hospital Dibrugarh, Assam from July 2018 to June 2019.

Selection of cases

All the patient who fulfils the inclusion and exclusion criteria are taken in the study group.

Inclusion criterion

All the pediatric patients in the age group 0-12 years attending ophthalmology OPD of Assam medical college and hospital, Dibrugarh were included in the study.

Exclusion criteria

Patients with any acquired abnormality, patients with any history of trauma to eye, patients with nutritional deficiencies and all the congenital ocular anomalies associated with other acquired abnormalities and nutritional deficiencies were excluded from the study.

Study done by taking interview with parents or attendants of the concerned patients, comprehensive examination, retrospective analysis of parent’s environment, birth history, family tree examination wherever feasible, recording of demographic characteristics of affected individuals. Collected data’s were tabulated, compiled and provisional diagnosis was reached in all the cases.

RESULTS

Proportion congenital ocular anomalies were found to be 13.72% out of 1850 paediatric cases as shown in Figure 1. Figure 2 and 3 shows male:female ratio as 1.8:1 where majority of cases were within 4 years of age. Congenital cataract is the most common anomaly followed by uveal colobomatous disorder. 27.55 % cases have congenital cataract, 22.04% have coloboma of iris and choroid, 16.53% have anophthalmos, 10.62% cases have microphthalmos and microcornea. 7.08 % have congenital NLD obstruction, 4.33% have congenital glaucoma 3.93% have hypoplastic optic disc, 3.54% cases are found to present with congenital esotropia, 1.96% cases have ptosis and congenital corneal opacity and 0.39% cases have PHPV as shown in Figure 4. Among all cataract cases most common was total cataract followed by lamellar cataract, posterior subcapsular cataract and nuclear cataract.

Majority of congenital cataract were in first birth rank and in 4.33% cases parents gave history of consanguinity. majority of the mother of affected children has not consumed any unknown medication. 20.86% cases have given the history of consumption of some unknown medication in their early gestational period. 85.03% cases of affected children were born in full term and 12.20% cases have congenital anomaly in their families.

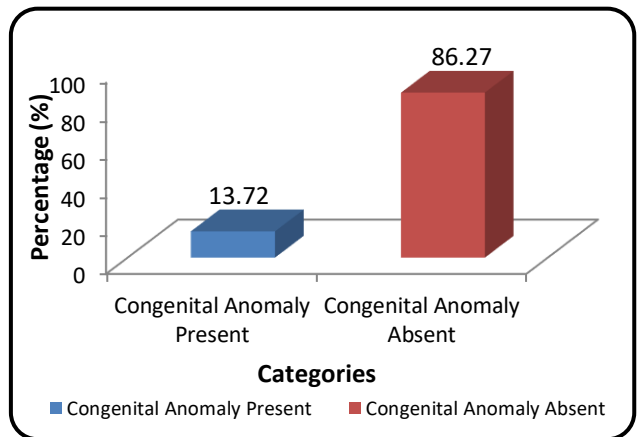


Figure 1: Total number of cases with congenital anomaly.

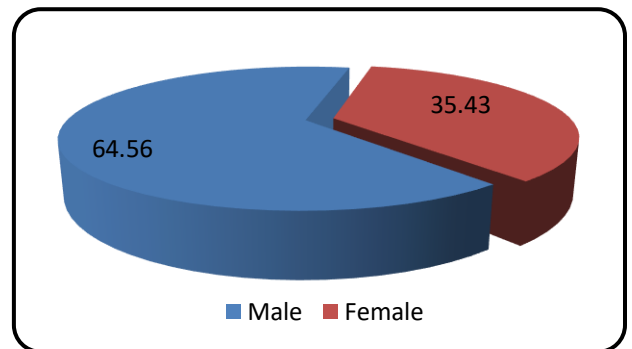


Figure 2: Sex distribution.

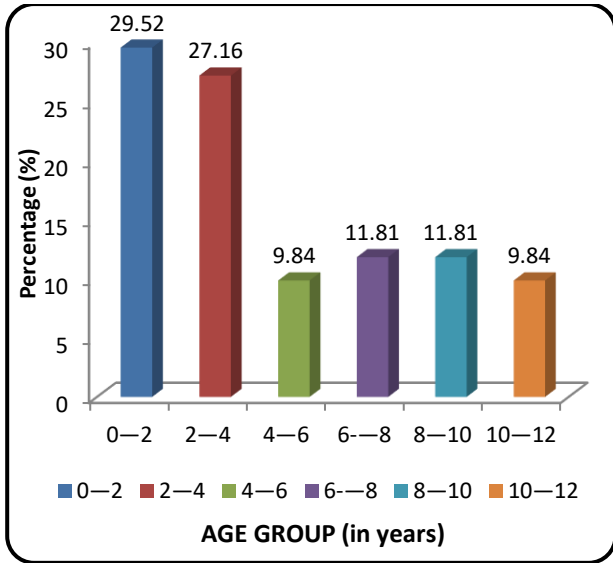


Figure 3: Age distribution.

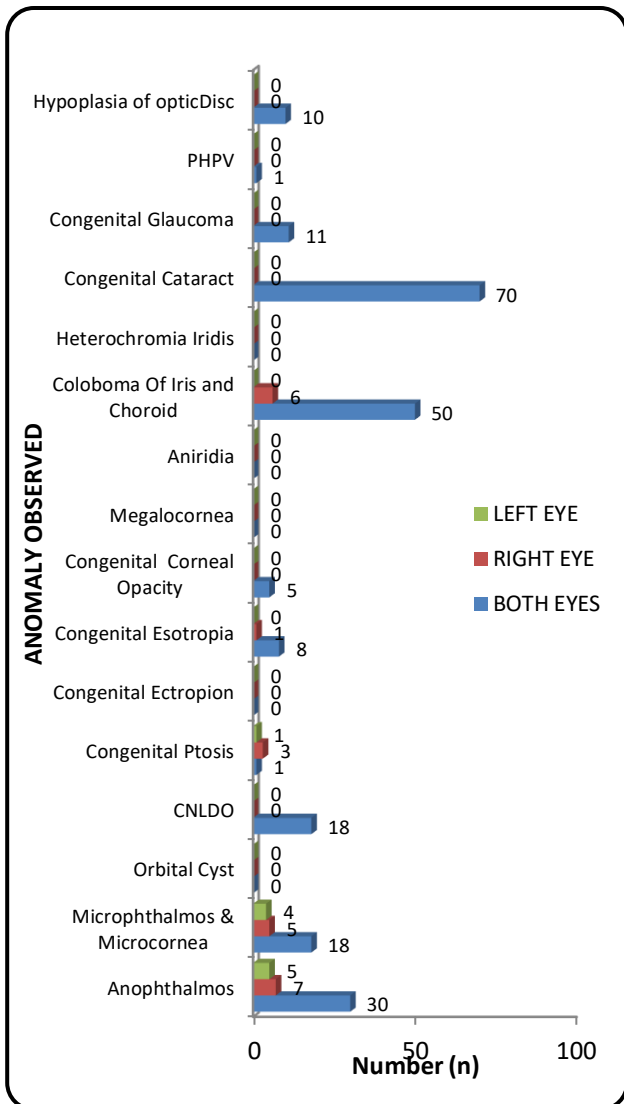


Figure 4: Distribution of various types of congenital anomaly.



Figure 5: (A) Congenital ptosis, (B) Anophthalmos, (C) Microphthalmos, (D) Congenital cataract.

DISCUSSION

In our study congenital ocular anomaly is found to be 13.72% i.e., 254 cases out of total 1850 paediatric patient which is corroborative to other study but there are other studies where prevalence of congenital ocular anomaly was found low.⁴⁻⁶ Reason behind estimation of higher number of anomalies in our study may be because AMCH, Dibrugarh is tertiary care hospital which covers large number of populations. Sex distribution was found to be higher in males and prevalence of congenital ocular anomaly was higher within 4 years of age which is comparable with other study.^{3,6,7}

Most common disorder in our study is congenital cataract followed by uveal coloboma than whole globe anomaly like microphthalmos and anophthalmos, which is similar to the other studies.^{3,6,7} Our study showed majority of congenital cataract were in the first birth rank and rest of anomaly like microphthalmos, anophthalmos, uveal coloboma and congenital glaucoma are more common in second birth rank, which is similar to another study done by Dutta and Bhattacharjee where incidence of congenital cataract is highest in second birth rank Incidence of colobomatous defect of the globe is highest in second birth rank (27%); next frequencies in third (20.43%) and then in fourth birth order (17.51%). 4.33% of the parents gave history of consanguineous marriage whereas other studies have found higher prevalence of consanguinity.⁸⁻¹⁰ Low prevalence of consanguinity in our study may be due to different marriage pattern in these parts of Assam and consanguineous marriage is not being practiced. Present study also shows 20.86% cases with history of consumption of any unknown medication in their early

gestational period there can be a possible link between the use of specific herbal medicines during the first trimester of pregnancy and increased risks of specific groups of congenital malformations.¹¹

Our study has showed majority of the cases were born in full term of gestational age and 7.87% children are born before term. In similar study done by Tupe et al have found 4% cases of congenital anomaly are born below term.¹⁰ We have also found that 12.20 % cases have ocular anomaly in their families like congenital cataract, uveal coloboma, congenital glaucoma run in families. According to our study 21.4% cases have positive family history of cataract and out of these 11.4% cases has given history of congenital cataract among their siblings which can be compared with study done by Wirth et al.¹² Uveal coloboma and glaucoma are also present among family members in proportion of 20% and 9% respectively. Recurrence rate of uveal coloboma for subsequent child of healthy unrelated parents has been estimated as 9%; for the children of an affected person, this figure is as high as 46%.¹³ this suggests importance of genetic analysis and prenatal diagnosis of congenital anomaly and avoidance of marriage among couples with similar disorder.

CONCLUSION

Congenital cataract-related blindness in children represented a large burden of avoidable diseases in our sample population. These children often present late to ophthalmic surgeons, resulting in poor outcomes due to the extended period of visual deprivation. Awareness regarding maternal and child health care, immunization against measles, rubella should be improved. Early diagnosis and prompt referral from peripheral hospital will lead to prevention of most of the childhood blindness. Encouragement of genetic counselling, low vision classes with print education in blind school including braille classes, would require investment and political interest. This study is like a screening procedure in detecting the different varieties of congenital anomaly of eye and to initiate the future advance researches in genetic and molecular biology for early detection of the cases and treating the factors causing these anomalies.

Funding: No funding sources

Conflict of interest: None declared

Ethical approval: The study was approved by the Institutional Ethics Committee

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Cite this article as: Paul L, Kuli JJ. Study on ocular anomalies in pediatric patients with special reference to congenital anomalies. Int J Res Med Sci 2021;9:1102-5.