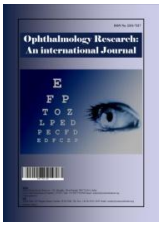

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Study of Congenital Ocular Anomalies Prevalent in Routine Eye OPD in a Tertiary Care Hospital

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Authors' contributions

This work was carried out in collaboration among all authors. Author AK designed the study, performed the statistical analysis, wrote the protocol and wrote the first draft of manuscript. Authors AS and DM managed the analyses of the study. Author DM managed the literature searches. All authors read and approved the final manuscript.

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ABSTRACT

Aims: Congenital ocular anomalies require treatment, visual rehabilitation and genetic counselling. Goes undetected in neonates and children due to lack of routine eye checkup. Diagnosed later during eye examination or when come for disability certificate.
Study Design: Prospective and observational study.
Place and Duration of Study: We conducted study at tertiary care hospital, over a period of six months (from January 2018 to June 2018).
Methodology: Patients with congenital ocular anomalies irrespective of their age were identified. Findings were confirmed by detailed ocular examination and investigations when required. Whatever best management like correcting refractive error, surgery, low vision aids or rehabilitation was advised. Those having acquired defects giving similar appearance as in congenital anomalies, cases of ocular trauma, infections, with prior surgical intervention were excluded.
Results: Over a period of six months 128 eyes were detected with congenital ocular anomalies. The most common anomalies were microphthalmos, microcornea and iris coloboma seen in 85 eyes. Other congenital anomalies were congenital cataract, nystagmus, anophthalmos, aniridia, congenital glaucoma, Ankyloblepharon, coloboma of disc, congenital esotropia, lid coloboma, congenital ptosis, axenfield anomaly, limbal dermoid.

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Conclusion: Patient with congenital ocular anomalies and their parents should be counseled regarding possible current treatment, visual prognosis and genetic counselling regardless of age and time of presentation.

Keywords: Congenital ocular anomalies; microphthalmo; microcornea; iris coloboma.

1. INTRODUCTION

Worldwide, an estimated of 36 million peoples live with blindness [1]. Prevalence of blindness by age distribution, around 1.4 million children aged between 0–14 years are currently living with blindness, whereas approximately 17.5 million are at a risk of developing low vision [2]. The estimated burden associated with the blindness among children is 70 million blind person years [3]. The number of blind years resulting from the blindness is alarmingly high in the children, and this has an immense social and economic impact [4-6]. The congenital ocular anomaly is one of important cause of childhood blindness. 16.7% of total childhood blindness are caused by major structural childhood blindness like anophthalmos, microphthalmos and coloboma globally [7].

The magnitude and causes of visual impairment and blindness vary according to region, owing to socio-developmental diversification [4]. Analyses of global data showed that around 90% of blind people reside in developing countries [8]. A majority of the causes of childhood blindness are avoidable even in the minimal resource settings of developing countries [8]. In India, the conditions are significantly worse in the rural areas, in which there are enormous issues pertaining to the delivery of healthcare. Owing to socioeconomic and cultural constraints, and the medical poverty trap settings, it could be assumed that India, especially its rural areas, has poor eye care facilities. Over the last few years, a number of initiatives have been commenced with the aim of achieving the VISION 2020 goal [9]. However, there is further scope for improvement. Moreover, few existing studies have investigated the burden and capacity of the healthcare system in India in terms of childhood blindness. Very little information is explored and documented about epidemiology of childhood blindness. However, for designing a prevention strategy comprehensive epidemiological information is crucial.

A congenital anomaly is an abnormality that is present at birth, even if not diagnosed until months or years later. Most congenital anomalies are present long before the time of birth, some in the embryonic period (up to the 7th week of gestation) and other in the foetal period (8th week to term). The anomaly covers all the major classes of abnormalities of development which there are four major categories as follow [10]. Malformation, Deformation, Disruption, Dysplasia. Congenital anomalies contribute a significant proportion of infant morbidity and mortality, as well as foetal mortality. As a consequence, it is essential to have basic epidemiological information on these anomalies. The precise of congenital malformations is not known for as many as 50 – 60% of the total. It is believed that overall, multifactorial aetiology account for 20-25% of all abnormalities; 6-8% are monogenic; 6-8% by chromosomal Abnormalities; and 6-8% by environmental factors such as maternal illness, infections, drugs, radiation and alcohol [11]. Major cause is maternal infection during pregnancy, caused by some important infectious agents as follow [11]. Rubella, Varicella, Cytomegalovirus, Toxoplasmosis.

2. AIMS AND OBJECTIVE

2.1 Aim

To study the profile of congenital ocular anomalies in routine eye OPD.

2.2 Objective

To create awareness about their anomalies among patients and relative.

To provide proper treatment of congenital ocular anomalies.

3. MATERIALS AND METHODS

A Prospective study was carried out over a period of six months (from January 2018 to June 2018) in tertiary care center. Patients with congenital ocular anomalies irrespective of their age were identified. Findings were confirmed by detailed ocular examination and investigations when required. Whatever best management like

correcting refractive error, surgery, low vision aids or rehabilitation was advised. Those having acquired defects giving similar appearance as in congenital anomalies, cases of ocular trauma, infections, with prior surgical intervention were excluded.

4. RESULTS

Over a period of six months 84 cases (128 eyes) were detected with congenital ocular anomalies out of which the most common congenital anomalies were microphthalmos, microcornea and iris coloboma seen in 56 cases (85 eyes). Other congenital anomalies were congenital cataract, nystagmus, anophthalmos, aniridia, congenital glaucoma, Ankyloblepharon, coloboma of disc, congenital esotropia, lid coloboma, congenital ptosis, axenfield anomaly, limbal dermoid (Table 1). The family histories in our few cases showed that one of the sibling had a similar congenital eye anomaly.

Most cases showed involvement of both eyes, no preferred involvement of either right or left eye seen (Table 2) (Fig. 1).

On studying the pattern of age distribution most of patients with congenital ocular anomalies presented between 18 to 25 years of age with male predominantly affected (male: female; 2.2:1) (Table 3) (Fig. 2).

In 15.48% cases consanguinity was present while 9.52% cases had maternal infection during antenatal period. Simultaneous systemic involvement was observed in 13.09% cases (Table 4) (Fig. 3).

Table 1. Congenital ocular anomalies observed

S. N.	Congenital ocular anomalies	No. of cases	No. of eyes affected
1	Microphthalmos, microcornea, iris coloboma	56	85
2	Congenital cataract	8	9
3	Nystagmus	4	8
4	Anophthalmos	4	7
5	Aniridia	3	6
6	Congenital glaucoma	1	2
7	Ankyloblepharon	1	2
8	coloboma of disc	2	3
9	congenital esotropia	1	1
10	Lid coloboma	1	1
11	Congenital ptosis	1	1
12	Axenfield anomaly	1	2
13	Limbal dermoid	1	1
	Total	84	128

Table 2. Eyes involvement

S. N.	Congenital ocular anomalies	Cases with BE	Cases with RE	Cases with LE
1	Microphthalmos, microcornea, iris coloboma	29	16	11
2	Congenital cataract	1	2	5
3	Nystagmus	4	0	0
4	Anophthalmos	3	0	1
5	Aniridia	3	0	0
6	Congenital glaucoma	1	0	0
7	Ankyloblepharon	1	0	0
8	coloboma of disc	1	1	0
9	congenital esotropia	0	0	1
10	Lid coloboma	0	0	1
11	Congenital ptosis	0	1	0
12	Axenfield anomaly	1	0	0
13	Limbal dermoid	0	0	1
	Total	44	20	20

Fig. 1. Graphical representation showing eye preference
(BE - Both eyes, RE - Right eye, LE - Left eye)

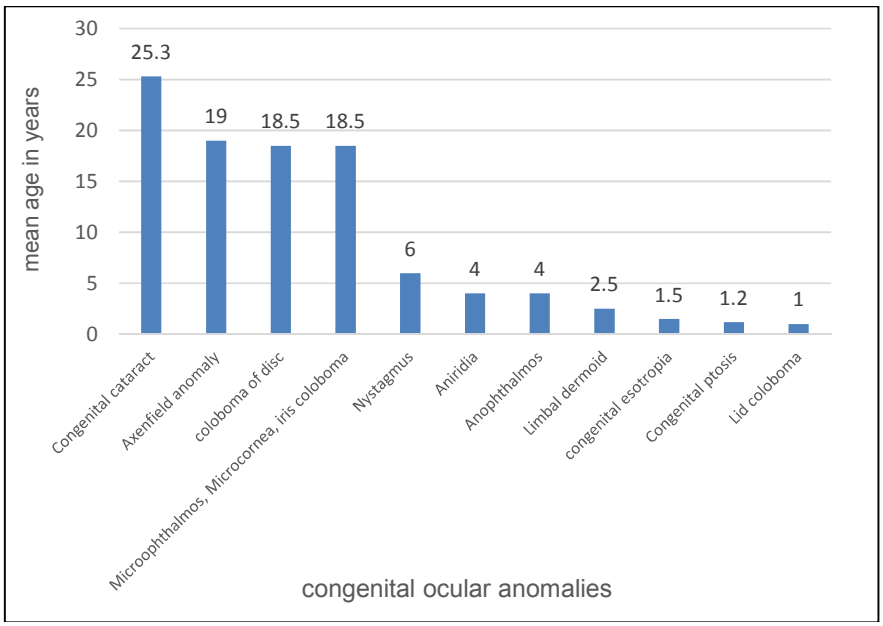

Fig. 2. Graphical representation of mean age distribution of various congenital ocular anomalies

5. DISCUSSION

Congenital ocular anomalies require treatment, visual rehabilitation and genetic counselling. Usually they go undetected in neonates and children due to lack of routine eye checkup in children and neonate. Ocular anomalies are diagnosed later during routine eye examination or when patients come to get a disability certificate. As the treatment of congenital ocular anomaly is very discouraging, the blindness due to this cause has not given importance in most of the studies. However, we can prevent some of the congenital ocular anomalies by simple awareness among the community.

Severe visual impairment and blindness in infants must be detected as early as possible to initiate immediate treatment to prevent deep amblyopia. The causes of severe visual

impairment and blindness may be prenatal, perinatal, and postnatal. Congenital anomalies such as anophthalmos, microphthalmos, coloboma, congenital cataract, infantile glaucoma, and neuro-ophthalmic lesions are causes of impairment present at birth. Leukocoria or white pupillary reflex can be caused by congenital cataract, persistent hyperplastic primary vitreous and retinoblastoma. Current antenatal ultrasound protocols for imaging of the fetal eye are inconsistent and inadequate to screen for the spectrum of ocular malformations, and there are no clear guidelines on detection of these rare abnormalities.

Table 3. Age and sex distribution

S. N.	Congenital ocular anomalies observed	Mean Age distribution	Male cases	Female cases
1	Microphthalmos, microcornea, iris coloboma	18.5 yrs	37	19
2	Congenital cataract	25.3 yrs	6	2
3	Nystagmus	6 yrs	3	1
4	Anophthalmos	4 days	3	1
5	Aniridia	4 yrs	2	1
6	Congenital glaucoma	2 mnth	1	0
7	Ankyloblepharon	1 mnth	0	1
8	coloboma of disc	18.5 yrs	1	1
9	congenital esotropia	1.5 yr	1	0
10	Lid coloboma	1 yr	1	0
11	Congenital ptosis	1.2 yr	1	0
12	Axenfield anomaly	19 yrs	1	0
13	Limbal dermoid	2.5 yr	1	0
	Total		58	26

Table 4. Aetiological causes of congenital ocular anomalies

S.N.	Distribution of patients according to history	Number of cases	Percentage (%)
1	Patients with a history of consanguinity	13	15.48%
2	Patients with a history of maternal infection	8	9.52%
3	Patients with systemic involvement	11	13.09%

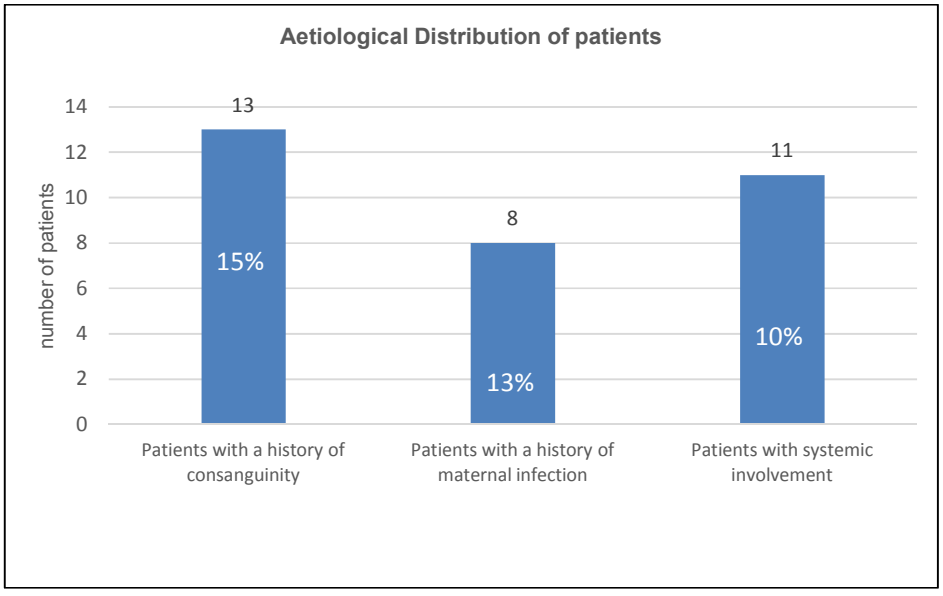

Fig. 3. Graphical representation of aetiological causes of congenital ocular anomalies

When we compared the our study to other study we founded some differences like Parag NT, Sagar VC [12] highlighted that most common congenital ocular anomaly was congenital dacryocystitis (24%) and male to female ratio was 1:1.4, maximum number of age group affected between 0-2 years (54%). Behera S, Chowdhury RK, Sar M [13] also highlighted that nasolacrimal duct anomalies were the most common (33.3%) congenital anomaly. But in our study we founded that most common congenital ocular anomalies was microphthalmos, microcornea, iris coloboma (67%), Male to female ratio was 2.2:1, and most common age group affected between 18 to 25 years.

6. CONCLUSION

Preventive measures can be adopted if the history is taken properly during the evaluation of the patients because maternal infection and systemic involvement have a great impact in this context. Early diagnosis is the key to early intervention for preventing childhood blindness. Patient with congenital ocular anomalies and their parents should be aware about visual prognosis, possible current treatment, and regarding to genetic counselling. Public awareness should be more regarding to MMR vaccination. In our opinion, termination of the pregnancy may be justifiable when severe eye malformations such as anophthalmia and cryptophthalmos are positively identified. Proper knowledge of the developmental pathogenesis of congenital ocular anomalies is highly important for correct diagnosis and early intervention.

CONSENT

All authors declare that written informed consent was obtained from the patient (or other approved parties) for publication of this case report and accompanying images.

ETHICAL APPROVAL

As per international standard written ethical approval has been collected and preserved by the author(s).

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COMPETING INTERESTS

Authors have declared that no competing interests exist.

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