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Research Article

A CLINICAL STUDY OF CONGENITAL AND DEVELOPMENTAL OCULAR ANOMALIES IN PAEDIATRICS AGE GROUP

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A R T I C L E I N F O

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A B S T R A C T

Aim: To evaluate the magnitude and pattern of congenital ocular anomalies among paediatric eye patients attending OPD and to correlate them with demographic and environmental factors. **Materials and Methods**: -In this study, number of all paediatric eye patients and also the number of ocular anomalies were recorded. From this, incidence of eye anomalies were calculated. After taking detail history, all patients were subjected to systemic and comprehensive eye examination. Bio-chemical and radio-imaging investigations were performed wherever required.

Result: Out of 3102 eye patients, 140 patients had ocular anomalies, incidence being 4.5%. Maximum number of patients were in the age group between 0-5 years (43.57%) with a male preponderance. History of consanguinity were observed in only 3.5%. 11 cases(7.86%) gave history of pre-term delivery. Maximum patients presented in between 0-5 years (45%) while 32 patients (22.86%) had unilateral and 108 patients (77.14%) had bilateral involvement. 65.71% of patients had anterior segment involvement , 1.43% patients showed posterior segment involvement while globe involvement was observed in 32.86%.Congenital cataract, globe anomalies, congenital dacryocystitis and ptosis were most frequently detected anomalies.

Conclusion: Congenital ocular anomalies are involved in considerable size of paediatric eye patients which often result in ocular morbidity and even blindness. So, there is a great need for early detection and management of these patients.

INTRODUCTION

A congenital ocular anomaly is an abnormal presentation of the eye present at birth which may remain undiagnosed until months or years later. It may appear in isolation, in combination or part of a syndrome consisted of multiple anomalies and disorder in other parts of the body. Congenital defects including ocular anomalies cover all the major classes of abnormalities of development which consist of four categories.[1] namely-Malformation, Deformation, Disruption and Dysplasia. In the human embryo, the eyes are formed by a delicate and complex process and problems in this process can lead to congenital ocular malformations. These conditions occur in approximately five per 10,000 live births. [2] In a survey conducted for blindness in India (in 1968), a total of 4047 cases of blindness were noted. Out of these 48 were due to a congenital defect forming 1% of the total [3]. There are many records of various forms of blindness and those due to congenital defect form at least a small percentage for causes of blindness.[4] It is worth mention here that childhood blindness is one of the diseases that have been given priority in the on-going WHO''s vision programme "The Right to Sight" because of higher number of blindness years a blind child has[5]. Besides, the genetic factors, maternal illness and maternal infection caused by Rubella, Varicella, Cytomegalovirus and Toxoplasmosis, drugs, alcohol, radiation etc. play a major role in causing congenital anomalies[6]. 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 and are often attributable to avoidable causes[7][8]. In contrast, in the developed countries anophthalmos, microphthalmos and coloboma(AMC) are the leading anomalies seen at birth and are mainly due to unavoidable causes[9]. The off springs of couples engaged in consanguineous marriages are more prone to develop congenital anomalies in different parts of the body including ocular malformations. Such marriages are widely prevalent in different parts of the globe and our India is also no exception to this. Preconception and premarital counselling on consanguinity should be a part of the training of health care providers particularly in highly consanguineous populations [10].

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MATERIALS AND METHOD

This prospective study was undertaken to evaluate the magnitude of congenital and developmental ocular anomalies among the paediatric eye patients attending outpatient

department (OPD) of Regional institute of Ophthalmology, Gauhati Medical College, Guwahati for a period of 1 year and also to explore the patterns of ocular anomalies. It was also aimed to correlate the congenital eye anomalies with environmental and demographic factors. It included all paediatric eye patients attending ophthalmology OPD and their numbers were recorded. Among them, the number of congenital ocular anomalies are also recorded. From this, we calculated the incidence of ocular anomalies among the paediatric eye patients. All paediatric ocular anomaly cases (0-18 years) were included in the study. Those above 18 years of age, patients with history of ocular trauma and with inflammation of eye were excluded from the study. After obtaining informed consent, patients were enrolled for study.

A detail of past history, birth history, family history and maternal history in respect of each patient was taken and recorded. After general examination, all patients were subjected to comprehensive ocular examination which included visual acuity assessment, intraocular pressure measurement, slit lamp biomicroscopy and ophthalmoscopy. Examination under general anaesthesia was performed as and when necessary. Biochemical tests and radio-imaging investigations were done wherever required.

RESULTS AND OBSERVATIONS

Prevalence of Congenital ocular Anomalies Among Paediatric eye Patient Attending eye OPD

Table 1 Prevalence of ocular anomalies

Groups	No.	Percentage (%)
Total No.of paediatric eye patients	3102	100%
No. of children with ocular anomalies	140	4.5%

Distribution of Ocular Anomalies Among Different age Group

Table 2 Age distribution

Age Groups in years	Numbers	Percentage (%)
0-5	61	43.57%
6-10	39	27.86%
11-15	27	19.29%
>15	13	9.29%
Total	140	100

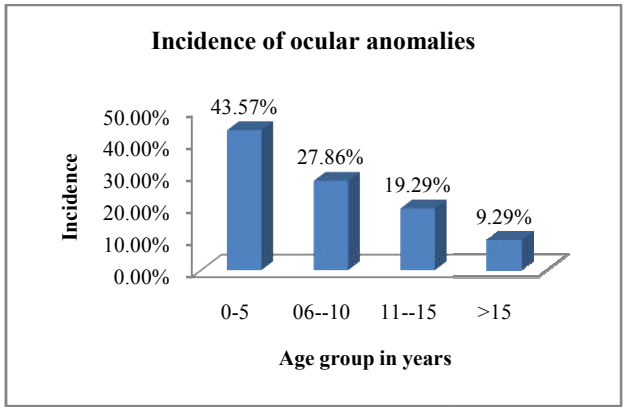

Figure 1

Gender Distribution of Congenital ocular Anomalies

Table 3 Gender distribution

Gender	Numbers	Percentage (%)
Male	81	57.86%
Female	59	42.14%
Total	140	100%

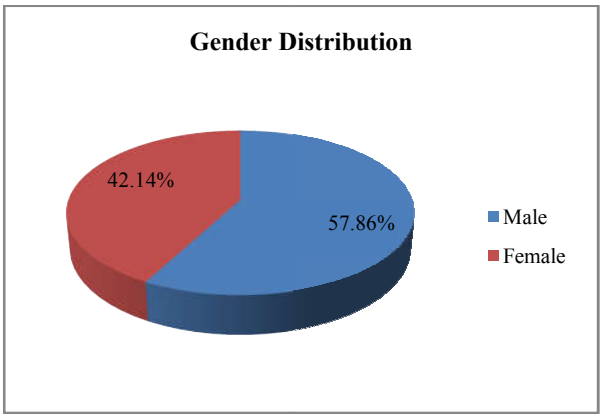

Figure 2

Occurrence of Consanguinity in the present study

Table 4 Consanguinity

Consanguinity	Number	Percentage (%)
Present	5	3.57%
Absent	135	96.43%
Total	140	100%

History of consanguinity was found in only 3.57%

Gestational age at birth among the patients with congenital eye anomalies

Table 5 Gestational age at birth

Gestational age	Number	Percentage (%)
Preterm	11	7.86%
Full term	129	92.14%
Total	140	100%

Age of the Patient at Presentation

Table 6 Age at presentation

Age at presentation (in years)	Number	Percentage (%)
0-5	63	45%
6-10	39	27.86%
11-15	28	20%
16-18	10	7.14%
Total	140	100%

Laterality of Involved eyes in Ocular Anomaly Patients

Table 7 Laterality

Laterality of involved eyes	Number	Percentage (%)
Unilateral involvement	32	22.86%
Bilateral involvement	108	77.14%
Total	140	100%

Distribution of cases in Relation to Involvement of Anterior and Posterior Segment

Table 8 Involved segment

Involved segment	Number	Percentage (%)
Anterior segment	92	65.71%
Posterior Segment	2	1.43%
Both	46	32.86%
Total	140	100%

Distribution of Various types of Congenital Anomalies

Table 9 Distribution of anomalies

Sl No	Ocular anomalies	No. of patients	Percentage
1	Congenital cataract	43	30.71%
2	Globe anomalies (uveal coloboma, microcornea etc.)	30	21.42%
3	Congenital Dacryocystitis	25	17.86%
4	Congenital ptosis	15	10.72%
5	Squint	10	7.14%
6	Corneal opacity	6	4.29%
7	Dermoid	6	4.29%
8	Congenital Glaucoma	5	3.57%
9	Anterior staphyloma	3	2.14%
10	Lipoma	2	1.43%
11	Optic disc anomaly	2	1.43%
12	Keratoconus	1	0.71%
13	Dystrichiasis	1	0.71%
14	Megalocornea	1	0.71%
	Total	140	100%

DISCUSSION

In our hospital based prospective study, the prevalence of congenital and development ocular anomalies was noticed to be 4.5% .TupuParageN *et al* (2016) observed in their study that the incidence of ocular malformation was 0.53%[11] whereas Stoll *et al* on the epidemiology of congenital eye malformation in Strasbourg, France from 1978 to 1988, reported the prevalence to be 0.75% [12]which are lower than our study . Besides , in the study of Singh *et al* [13] and Bermejo *et al* [14], the incidence rates were 0.105% and 0.037% respectively . In our study, the age distribution of patients with congenital eye anomalies range from birth to 18 years of age. Maximum number of patients belonged to the age group between 0-5 years (43.57%). This finding was similar to studies of TupuParage *et al*[11] with 54% in the age group 0-2 years and also that Bermejo *et al* (14). In our study, there were 81 male patients (57.86%) and 59 female patients (42.14%) showing male preponderance. Punita Garg *et al* (2016) also found more male patients (112 patients: 56%) than females patients (88 patients:44%).(15) Similar results were revealed by C.M. Chuka –Okisa *et al* (2005) with 29 males and 25 females[16].

As regards, prevalence of consanguinity among parents of patients with ocular malformation, 5 cases (3.57%) gave positive history of consanguineous marriage in the present study. 36% of the parents disclosed a history of consanguinity in a study conducted by Tupeparag N *et al*[11] which is higher than our finding. Our finding matched with that of Narachi *et al* [17] and Stoll *et al* [12]. On evaluating the gestational age of the ocular anomaly cases at birth in our study, 11 cases (7.86%) gave history of preterm delivery while 129 cases (92.14%) had full term delivery history. Similar results were encountered by Tupuparag *et al* showing 2 cases (4%) with preterm and 48 cases (96%) with full term delivery (11). The average age of presentation of cases ranged from 0 to 18 years in the present study whereas in the study of Tupeparag *et al* average age of at presentation and diagnosis ranged from Day 1 to 8.5 years [11].22.86 %(32 cases) of ocular anomalies were unilateral and 77.14% (108 cases) were bilateral in this study . TupuParagN *et al* also found more of bilateral ocular anomalies than unilateral lenses [11]. Our study revealed that most of the ocular anomalies were confined to anterior segment (65.71%) and few cases (1.43%) were found in the posterior segment and whole globe (32.86%). This corroborates with that of TupuParag N *et al* with 82% lesions in anterior segment, 8% in posterior segment and 10% involving the both segment [11]. A.Ilechie *et al* (2013) also revealed that major locations of the eye anomalies were lens with 20.5%(anterior segment), retina with 13%(posterior segment) and whole globe with 17% (both segments)[18].

Congenital cataract was observed to be the commonest ocular anomaly in our study with 43cases (30.71%). This co-relates with the finding of PunitaGarg*et al* with 19% of cataract [15] and that for A Ilechie *et al* with 16.8%of cataract [18]. The incidence of congenital Dacryocystitis was revealed to be 17.86%. Its incidence varies in different author's study: Alberta with 0.08%[19], Tupeparag N *et al* with 0.12% [11] and A Ilechie *et al* with 2.3%[18]. Prevalence of congenital glaucoma in our study was 3.57% whereas TupeParag*et al* showed it be 6%(3cases)[11] and A.Ilechie found it be 14.1%[18] which are higher than our results . There were 30 cases (21.42%) globe anomalies and 1 cases (0.71%) of megalocornea in present study. In a study conducted by Rahi JS *et al* [20] and Alla Foster [21], the prevalence of globe anomalies was 25% which correlates with our results. megalocornea prevalence was 2% and 2.3% in the findings of TupeParage *et al* [11] and AA Ilechie *et al* [18] respectively which are higher than our observation.As regards congenital ptosis, there were 10.72% of cases in the instant study while PunitaGarg *et al* [15] found it to be 6% and this matches with our result. 7.14% of congenital squints were encountered in our study and PunitaGarg *et al* [15] also found similar finding with 3.5% of congenital strabismus. Besides, congenital corneal opacities accounted for 4.29% and all the cases had vascularization. Our result tallies with findings of PunitaGarg *et al* with 5% incidence [15] whereas finding of TupeParage N *et al* [11] was as low as 0.02% only. Besides, some other anomalies as stated in result were noticed in very small percentage.

CONCLUSION

An incidence of 4.5% of congenital ocular anomalies is revealed among the paediatric eye patients. The peak age at the presentation is in the age between 0 - 5 years. There is a male preponderance in gender distribution and majority of cases had bilateral involvement. Most patients are full-term children. There is a positive history of consanguineous marriage among

few parents of the study population. The anterior segment of eyes are more involved than posterior segment. Congenital cataract, congenital dacryocystitis, globe anomalies and ptosis are mostly prevalent eye anomalies among others. These congenital eye disorders, in isolation or in combination, or as part of a syndrome, though rare are important causes of childhood blindness which adversely affects development, morbidity, education and working avenues of a child. So, childhood blindness has rightly been placed in priority under Vision 2020.There is a great need for instituting screening programmes at maternity care centres for prompt detection of congenital ocular anomalies before discharge of new-borns and early treatment of those which can result in blindness. Besides such babies should have a thorough systemic examination to explore anomalies in other systems of the body forming parts of a syndrome.There is also a need for premarital counselling to eliminate or reduce congenital ocular malformations. Paediatric ophthalmology with optimal infrastructures should be introduced in all referral hospitals.

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