Research Paper

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A Study of Congenital Ocular Anomalies in Paediatric Age Group

Medical Science

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ABSTRACT

Purpose: To study the pattern of congenital ocular anomalies in paediatric age group. Materials & Methods: In this study total 9350 patients were screened. The age and sex of the patient, gestational age, occurrence of consanguineous, distribution of various subtype of congenital anomalies, subtype of congenital cataract, age at presentation and diagnosis were noted. Results: The age variation in the study was between 0-12 years. The maximum number of patients were in the age group of 0-2 years. Male: female ratio was 1:1.4. More number cases were reported in anterior segment with full term delivery.32 cases having no positive history of consanguineous marriage. Total 12 cases were found about chronic dacryocystitis, 8 cases of coloboma of iris and choroid and each 5 cases of congenital cataract and Microhthalmos were found. None of the cases had any history of antenatal, obstetric complication, radiation and drug intake. Conclusion: A prevalence of 0.053% of congenital ocular anomalies. Most common anomaly was congenital dacryocystitis (24%), congenital cataract and microphthalmos being the second most common anomalies (14%) each

Introduction-

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 the7th week of gestation) and other in the fetal period (8th week to term). The anomaly covers all the major classes of abnormalities of development which there are four major categories as follow [1]

- Malformation
- Deformation
- Disruption
- Dysplasia

Congenital anomalies contribute a significant proportion of infant morbidity and mortality, as well as fetal 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 etiology account for 20-25% of all abnormalities; 6-8% are monogenic, that is cause 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.

Major cause is maternal infection during pregency, caused by some important infectious agents as follow [2]

1. Rubella
2. Varicella
3. Cytomegalovirus
4. Toxoplasmosis

In a survey conducted for blindness in India 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 at least a small percentage of causes. [4]

Congenital deformities are due to two etiological causes-

1. Primary due to germinal causes
2. Secondary due to environmental causes

Here, the title, Prevalence of congenital ocular anomalies in the pediatric age group is chosen with deliberation in order to limit, it's scope for an immense range of abnormalities conditions, indeed much of the medicine could be included under the umbrella of anomalies of development.

MATERIAL AND METHODS

Study was conducted at Department of Ophthalmology, in a tertiary care teaching hospital located in rural area of western Maharashtra. The study was carried out over a period of two years. These included all the new born babies in the pediatric ward, all patients attending ophthalmology OPD and camps. Cases were of the age group 0-12 years.

Screening consisted of name, age sex, residence, religion and opd number of the patients. Detailed antenatal history was taken which included consanguinity, any unwanted event in the early pregency, drug intake, radiation during pregency, any disparity detected between period of gestation.

Nature of delivery, full term or premature and natural, assisted or operative was also taken into consideration. APGAR score in relevant cases was also noted. General examination and systemic examination for other congenital anomalies.

A complete detail examination was carried out with the help of torch light (and slit lamp wherever possible). Rough assessment of vision was done in all new born with torch light, pre-school children (3-5 years) was done by illiterate E-cutout test. Measurement of vision in school children (above 5 years) was done with Snellen's chart.

A case which required investigations like ocular tension, indentation tonometry and measurement of corneal diameter and gonioscopy to rule out bupthalmos were undertaken in general anesthesia.

A case of ptosis was examined for the presence of degree of ptosis, squint, Marcus- Gunn phenomenon, presence and absence of Bell's phenomenon, MRD 1 & 2 to measure the amount of ptosis, leavator function test performed.

In a case of squint cover test, cover-uncover test, alternate cover test and Hirschberg test were performed.

All patients of congenital anomaly were investigated in detail for base line investigations like X-ray of the chest, complete haemogram, urine routine and microscopic, USG abdomen and pelvis to assess the complete nature of anomaly. Fundus examination in all cases with help of direct ophthalmoscope.

Results-

Table 1:

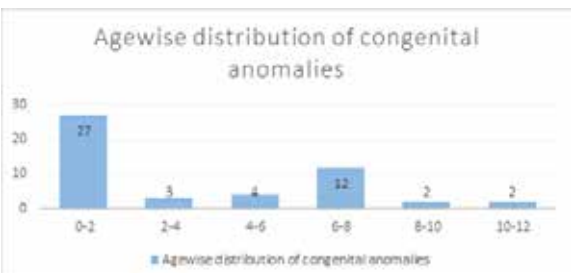
Magnitude of congenital ocular anomalies-

	Number of cases	Percentage
Children screened	9350	100%
Congenital ocular anomalies	50	0.53%

During study period, 9350 children below the age of 12 years were examined. The number of children that were detected to have congenital ocular anomalies were 50, giving prevalence of 0.53%.

Graph no1-

Age of distribution of the patients with congenital ocular anomalies-



In this study, the age range was from birth to 12 years of age. From below graph maximum 27 cases were found in the age group 0-2 years, giving percentage of 54%.

Graph no- 2

Gender distribution of patients:

This graph shows that the male: female ratio in our study was 1:1.4.

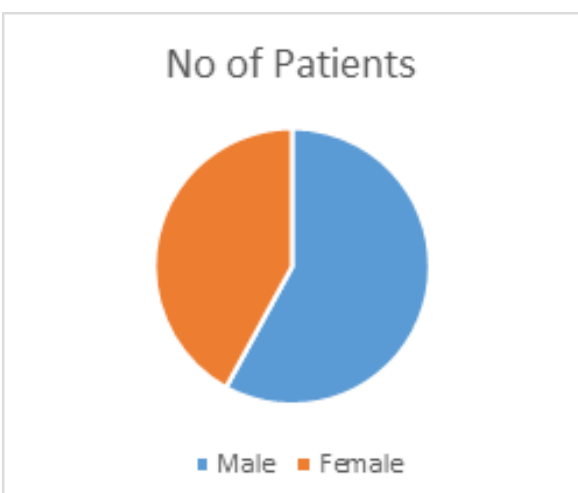


Table no -2:

Gestational age at birth

Sr no.	Gestational age	No of cases	Total number of cases
1	Preterm	02	370
2	Full term	48	8980

Most of the children with congenital anomalies were full term deliveries ,48 cases. Two cases with preterm birth one with congenital cataract and one with megalocornea were detected.

Table no-3

Distribution of cases in anterior and posterior segment:

Cases	Number of cases	Percentage
Anterior segment	41	82%
Posterior segment	4	8%
Both	5	10%

It was observed that anterior segment had more cases 41 (82%) and only 4 cases of posterior segment anomalies were detected. 5(10%) cases were found to have both anterior and posterior segment.

Table no 4-

Occurrence of consanguinity in the study:

Consanguinity	Number of cases with degree			Percentage
	1st	2nd	3rd	
Present	3	14	1	36%
Absent	32			64%

It was observed that in 18 cases (36%) parents gave a history of consanguinity. Of these 16% had 1st degree of consanguinity, 78% had 2nd degree consanguinity and only 6% subjects 3rd degree consanguineous relations in marriage were present.

Table no 5-

Distribution of various subtypes of congenital anomalies:

Sr.	Anomaly observed	Both eye	Right eye	Left eye	Total	percentage
1	Anophthalmos	3	-	-	3	0.03%
2	Microphthalmos and microcornea	5	-	-	5	0.05%
3	Orbital cyst with rudimentary eye	1	-	-	1	0.01%
4	Congenital dacryocystitis	8	4	-	12	0.11%
5	Congenital ptosis	1	1	-	2	0.02%
6	Congenital ectropion	1	-	-	1	0.01%
7	Congenital esotropia	-	1	-	1	0.01%
8	Congenital corneal opacity	2	-	-	2	0.02%
9	Megalocornea	-	1	-	1	0.01%
10	Aniridia	2	-	-	2	0.02%
11	Coloboma of the iris and choroid	3	5	-	8	0.09%
12	Heterochromia iridum	-	-	1	1	0.01%
13	Congenital cataract	5	-	-	5	0.05%
14	Congenital glaucoma	3	-	-	3	0.03%
15	PHPV	1	1	-	2	0.02%
16	Coloboma of the disc	-	1	1	2	0.02%
17	Leber optic atrophy	1	-	-	1	0.01%
18	Crouzon's disease	2	-	-	2	0.02%

Table no 6:

Age at presentation and diagnosis:

Sr.	Anomaly observed	No of cases	Laterality		Average age of diagnosis
1	Anophthalmos (fig no-1)	3	3 bilateral	-	Day 1
2	Microphthalmos and micro cornea	5	5 bilateral	-	4.4 years
3	Orbital cyst with rudimentary eye (fig no-2)	1	Bilateral	-	7 Days
4	Congenital dacryocystitis (Fig no-3)	12	8 Bilateral	4 Unilateral	2 Years
5	Congenital ptosis	2	1 Bilateral	1 Unilateral	8.5 years
6	Congenital ectropion	1	Bilateral	-	Day1
7	Congenital esotropia	1	-	Unilateral	1 Year

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8	Congenital corneal opacity	2	2 Bilateral	-	3.5 Month
9	Megalocornea	1	-	Unilateral	1 Month
10	Aniridia	2	2 Bilateral	-	8.5 Years
11	Coloboma of the iris and choroid	8	3 Bilateral	5 Unilateral	5.5Years
12	Heterochromia iridium (fig no-4)	1	-	Unilateral	7 Years
13	Congenital cataract (fig no-5)	5	5 Bilateral	-	5.1 Years
14	Congenital glaucoma	3	3 Bilateral	-	2.67 Months
15	PHPV	2	1 Bilateral	1 Unilateral	2.5 Months
16	Coloboma of the disc	2	-	2 Unilateral	9 Months
17	Leber optic atrophy	1	Bilateral	-	6 Years
18	Crouzon's disease (fig no-6)	2	Bilateral	-	1.5 Years

Fig no-1 Anophthalmos

Fig no-2 Orbital Cyst

Fig no-3 Congenital Dacryocystitis

Fig no-4 Heterochromia Iridium

Fig no-5 Congenital Cataract

Fig no-6 Crouzon's Disease

Discussion-

The complexity of the process by which a fully fertilized egg develops into a fully formed individual and the extreme rapidity with which revolutionary changes occur especially in the early stages of growth astonished us that so many of us are born normal.

In the magnitude of congenital anomalies, we noted a prevalence of 0.53%. In a study conducted by Stoll, et al [5] on the epidemiology of congenital eye malformation in Strasbourg, France 1978 to 1988, the reported prevalence was 0.75% which was similar to our study.

In a study conducted by Singh, et al [6] the incidence of congenital anomalies was 0.105%. the differed between this study and ours is statistically insignificant. Bermejo, et al [7] found, a prevalence of congenital malformations to be only 0.037%. However, this difference is of no statistical significance.

In a survey conducted for blindness in India (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 [7], which also correlates with our study.

In our age distribution of the patients with congenital ocular anomalies, the age range was from birth to 12 years of age. We found maximum 27 number of cases were found in the age group 0-2 years, giving a percentage of 54%. This finding was similar to a study, by Bermejo et al [7]. This may be because of literacy and early detection of congenital anomalies.

In our gender distribution shows male to female ratio 1:1.4. This finding was similar to a study by chukka-Okosa, et al [8]. this study also reported a male preponderance of congenital ocular anomalies with male to female ratio 1:1.2. In a study by Stoll, et al [5] the sex ratio was 1:1.22 which corroborates with our study.

In our gestational age birth 4 % cases of congenital ocular anomalies gave a positive history of premature birth, however this percentage is statistically insignificant when compared to the total number of children examined in both full term and premature birth categories.

In a study by Rahi, et al [9], it was reported that in 60% of severely visually, impaired /blind children, vision loss was attributable to factors operating in the prenatal period, in 47% the prenatal factors were known and definite, and in 13 prenatal factors were the most probable causes.

In our distribution of cases in anterior and posterior segment was observed that anterior had more cases 41 and only 4 cases of posterior segment anomaly. 5 cases were found to have both anterior and posterior segment.

In our study occurrence of a history of consanguinity as high as in 36% cases, but this incidence is statically insignificant. Our finding matched with that of Narchi, et a [10]. He undertook a study of congenital anomalies diagnosed in AL-Hasa area in Saudi Arabia between Jan 1987 and Dec 1992. In a study conducted by Stoll, et al [5] on the epidemiology of congenital eye malformations in Strasbourg, France 1978to 1988, a significant association reported.

The incidence of anophthalmos in our study was 6% of congenital anomalies. In a study conducted by Bermejo, et al [7] found a prevalence of anophthalmos to 5%. In a study, Stoll, et al [5] was 4.6%.and in Hormby, et al [11], was 2.35%.

In our study 5 cases microphthalmos were detected, which make prevalence of 0.5 per thousand populations. According to Alberta [12] it was 0.09per thousand and Kallen, et al [13] to be about 1.5 per 10000 populations.

One case of orbital cyst with rudimentary eye noted. The prevalence was 1/10000 population. The cases have bilateral involvement. In the Jain, et al [14] bilateral orbital cyst is more commonly associated with major systemic abnormalities.

In our study, 12 cases congenital dacryocystitis were recorded. The prevalence was 0.12%. In Alberta [12] study it was 0.08%.

In our study two cases of congenital ptosis were recorded. Yilmaz, et al [10] reported a case of congenital ptosis with associated multiple ocular and congenital malformation were the associated ocular malformations.

One case of congenital ectropion was detected. Therefore, prevalence is very low i.e. 0.01 per hundred populations. Ruben, et al [13] has reported 0.3 per hundred populations.

One case of congenital esotropia found in our study. In hunter et al [15] reported the incidence of associated congenital ocular and systemic was much more with congenital exotropia than congenital esotropia.

Only one case of primary congenital corneal opacity was detected in our study.In a study conducted by Rezende et al [13] reported that only 6.9%of corneal opacities.

One cases of megalocornea was found in our study. No associated ocular or systemic malformations were detected.

We reported thee cases of bupthalmos, incidence 0.3 per thousand populations. All cases were bilateral. Levy, et al [16] reported incidence of congenital glaucoma as 0.1 per thousand populations. The Alberta [12] reported congenital glaucoma to be 0.03 per thousand populations.

We found 5 cases of congenital cataract means 0.3 per thousand populations in our study. In Alberta [12] has reported 0.13 per thousand populations. Koraszewska et al [17] reported a prevalence of 0.07%.

We found 8 cases of coloboma of iris, choroid and both. The uveal coloboma 0.08 per thousand populations. According to Alberta [12] it is 0.10 per thousand populations. Clarke found 2.4 per thousand populations.

Conclusions –

1. In our study we noted a prevalence of 0.053% of congenital ocular anomalies in the total population.
2. The age wise distribution of congenital anomalies showed that the peak age at presentation is in the first two years of life (56%)
3. We found a male preponderance in occurrence of congenital ocular anomalies, with a sex ratio of 1:1.4
4. The incidence of infants with congenital ocular anomalies that had premature birth was 4%in our study.
5. We found a positive history of consanguinity in 36%of our study.
6. Amongst the ocular anomalies 82% involved the anterior segment and only 8% posterior segment.
7. We found that 40% of the congenital anomalies caused severe visual impairment or blindness. All of these cases were bilateral.
8. Most common anomaly in our study was congenital dacryocystitis (24%), congenital cataract and microphthalmos being the second most common anomalies (14%) each.
9. The incidence of congenital systemic anomalies associated with ocular anomalies in our study was 10%.
10. We noted that only 17% of colobomatous defect of the uvea were complete.
11. None of the cases had any history of antenatal, obstetric complication, radiation and drug intake.
12. Most of cases occurred sporadically, suggesting more often environmental factor.

Limitations of the study-

There are a varying number of congenital anomalies which appear much later in life. This study restricts to a study of mainly cases with gross anatomical abnormalities. Also, cases of retinopathy of prematurity and retinoblastoma were not included in this study. As serological examinations like TORCH are not routinely done in this hospital, blood samples were sent outside in relevant cases only.

Conflict of Interest: Nil

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