Clinical patterns of congenital ocular anomalies in the pediatric age group (0 to 5 years) and its association with various demographic parameters

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Purpose: Congenital ocular anomalies are rare but important cause of childhood blindness. This study aimed to observe the clinical patterns of congenital ocular anomalies in the pediatric age group (0 to 5 years) and its association with various demographic parameters. Methods: Hospital-based cross-sectional study done on all pediatric patients in the 0-to-5-year age group presenting with congenital ocular anomalies to the Ophthalmology department of a tertiary care hospital in Eastern India between October 2018 and October 2020. Thorough clinical history was obtained, and comprehensive ocular examination was done in each case. Results: A total of 5868 patients in the 0 to 5 years age group attended the eye OPD during the study period. Congenital ocular anomalies were seen in 140 patients. The prevalence of ocular anomalies was 2.46%. Average age of patients was 3.32 ± 1.42 years. There were 74 (52.9%) males and 66 (47.1%) females. Unilateral and bilateral involvement was seen in 100 (71.45%) and 40 (28.6%) cases, respectively. Antenatal period was uneventful in 92.14% cases. Decreased vision was the most common presentation (40%). Congenital nasolacrimal duct obstruction was the most common anomaly seen in 29 (20.71%) cases followed by congenital cataract in 21 (15%) cases. Conclusion: Few of the congenital ocular anomalies can be prevented by increasing community awareness. Findings of the study can act as a reference guide for clinicians and health professionals for counseling and health planning.

Key words: Birth defects, childhood blindness, congenital, maternal malnutrition, ocular anomalies

Congenital eye diseases, though rare, are an important cause of childhood blindness. It can occur in isolation or in combination as a part of a syndrome. Congenital ocular anomalies are the result of defective development of ocular tissues during the intrauterine life, which can result from adverse genetic effects, environmental factors, teratogens, or chromosomal anomalies in the developing embryo. The array of ocular findings associated with these developmental disorders, metabolic and systemic diseases, and chromosomal anomalies is endless. Around 16.7% of total childhood blindness is caused by congenital ocular anomalies like anophthalmos, microphthalmos, and coloboma.[3]

People in the developing world are living amid risk factors for birth defects, e.g., universal marriage, high fertility, large number of unplanned pregnancies, poor coverage of antenatal care, poor maternal nutritional status, high consanguineous marriages rate, and high carrier rate for hemoglobinopathies. Despite the high risk of congenital malformations, there are no well-accepted preventive measures in most developing countries. This indicates that strong preventive measures for congenital anomalies are needed in this region. Increasing awareness about maternal care during pregnancy, educational programs on congenital malformations, and the consequences of consanguineous marriages need to be highlighted to decrease the incidence of congenital anomalies and their comorbidities.

Treatment of congenital ocular anomalies is usually very disappointing.[2] However, we can prevent some of the congenital ocular anomalies by spreading simple awareness among the community. In the present study, we have aimed to study the clinical patterns of congenital ocular anomalies in the pediatric age group (0 to 5 years) and its association with various demographic parameters.

Methods

The present study is a hospital-based, cross-sectional study done on patients with congenital ocular abnormalities attending the Ophthalmology outpatient department of a tertiary care referral hospital in Eastern India over a 2-year period. The study adhered to the basic tenets of Declaration of Helsinki. Institutional ethical committee clearance was obtained before the start of the study. Informed consent was obtained from each patient (parent accompanying the child) before enrolling in the study.

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All patients below 5 years of age with new or previously diagnosed congenital ocular abnormalities, presenting to the Ophthalmology outpatient department from October 2018 to October 2020 were included in the study. Children, who expired after the ocular diagnosis was made, were also included in the study. Exclusion criteria included age of the child >5 years, inadequate data, death of the child before ocular diagnosis, improper diagnosis, normal biological or developmental variations, and when the cause could not be ascertained to be of congenital origin.

Data was collected by interviewing the mother or adult accompanying the child. The data was recorded in pretested semi-structured questionnaires. A thorough history including age, sex, symptoms, and laterality of lesion was obtained. History of any significant antenatal events like prevailing chronic disease, infectious diseases (Toxoplasma, Rubella, Cytomegalovirus, and Herpes) during the antepartum period, intake of toxic substances or medications, radiation exposure, etc. was obtained. Information on maternal nutrition, food supplements taken during antepartum period, and duration of pregnancy (full-term or premature) was collected. Any significant family history in the form of any problems in other siblings was obtained. History of consanguineous marriage was also obtained.

Physical characteristics including the general appearance, body shape and size, craniofacial examination, neck examination as regards length, webbing, and neck swelling and examination of extremities regarding symmetry, shortening of limbs, and abnormalities of the fingers and toes were done. Rough assessment of vision was done in all newborns with torchlight. Visual acuity assessment in preschool children (3–5 years) was done using illiterate E-chart cut outs. Slit-lamp examination of the anterior segment was done to confirm any morphological or structural anomalies. Lacrimal syringing was done to confirm any congenital nasolacrimal duct obstruction (CNLDO). Intraocular pressure was recorded using Schiotz tonometer to diagnose primary congenital glaucoma.

Keratometry and A-scan ultrasonography were done to determine corneal diameter and ascertain the power of intraocular lens to be implanted in cases of congenital cataracts. Fundoscopy was done using both direct and indirect ophthalmoscopes after dilating both pupils with a combination of tropicamide (0.5%) and phenylephrine (10%) eye drops. In a case of squint, cover, cover-uncover, alternate cover, and Hirschberg tests were performed. Clinical examination and tests were individualized and performed as per requirement. The examination was done under anesthesia as per the requirement.

Data obtained was entered into a Microsoft excel sheet. Descriptive statistical analysis was performed to calculate the means with corresponding standard deviations. Data has been presented as actual numbers, percentage, mean, and standard deviation. Test of proportion was used to find the Standard Normal Deviate (Z) to compare the difference in proportions and Chi-square ($\chi^2$) test was performed to find the associations. $P$ value <0.05 was taken to be statistically significant. Statistical analysis was performed with help of EpilInfoTM version 7.2.2.2.

### Results

A total of 5686 patients in the age group of 0–5 years attended the Ophthalmology outpatient department during the study period. Out of these, 140 patients were found to have congenital disorders of the eye. The study included 180 eyes of these patients. As per the present study, the prevalence of congenital anomalies was found to be 2.46%. There were 74 (52.9%) males and 66 (47.1%) females. Right and left eye involvement was noted in 52 (37.1%) and 48 (34.3%) cases, respectively. Bilateral involvement was seen in 40 (28.6%) cases. The mean age of the patients was 3.32 ± 1.42 years.

#### Table 1: Salient demographic features of the patients

| Variable            | Integer |
|---------------------|---------|
| Total number of patients | 140     |
| Sex                  |         |
| Male                | 74 (52.9%) |
| Female              | 66 (47.1%) |
| Average age (years) | 3.32±1.42 |
| Age groups          |         |
| <1 year             | 10 (7.1%) |
| 1-4 years           | 51 (36.4%) |
| 4-5 years           | 79 (56.4%) |
| Laterality          |         |
| Right eye           | 52 (37.1%) |
| Left eye            | 48 (34.3%) |
| Bilateral           | 40 (28.6%) |
| Consanguinity       | 8 (5.7%) |

#### Table 2: Prevalence of congenital ocular anomalies

| Age groups   | Unilateral (%) | Bilateral (%) |
|--------------|----------------|---------------|
| <1 year      | 37.1%          | 7.1%          |
| 1-4 years    | 36.4%          | 10.0%         |
| 4-5 years    | 56.4%          | 14.0%         |

Corrected Chi-square test showed a significant association between age and congenital anomaly of patients ($P < 0.001$). CNLDO was more prevalent in patients <3 years and congenital cataract was more prevalent in the 4–5-year age group. The proportion of males was slightly higher as compared to females, but it was not found to be statistically significant ($Z = 0.84$, $P = 0.39$). CNLDO was prevalent more significantly in males and congenital cataract in females ($p < 0.001$). No statistically significant association was noted between age and gender of the patients ($P = 0.31$). Thus, in this study, ocular congenital anomalies were equally prevalent over age of the male and female patients. The proportion of children with CNLDO (20.7%) followed by congenital cataract (15.0%) was significantly higher than that of other congenital anomalies ($Z = 2.37$; $P = 0.017$).

Unilateral eye involvement was noted in 71.4% cases, which was significantly higher than bilateral (28.6%) ($Z = 5.93$; $P < 0.0001$) affliction. A significant association was noted...
Table 2: Distribution of presenting symptoms and signs of the patients

| Presenting symptoms and sign | n  | %   |
|------------------------------|----|-----|
| DOV                          | 25 | 17.9% |
| Watering                     | 19 | 13.6% |
| White reflex, DOV            | 14 | 10.0% |
| Absence of eye               | 11 | 7.9%  |
| Small eye                    | 11 | 7.9%  |
| Large eyes, watering         | 10 | 7.1%  |
| Small cornea                 | 8  | 5.7%  |
| Keyhole Iris, DOV            | 6  | 4.3%  |
| Watering, discharge, swelling| 6  | 4.3%  |
| Inward deviation of eye, DOV | 4  | 2.9%  |
| Watering, Periocular crusting| 4  | 2.9%  |
| DOV, different iris color    | 3  | 2.1%  |
| Fused eyelids                | 2  | 1.4%  |
| Keyhole Iris                 | 2  | 1.4%  |
| Painless cystic swelling     | 2  | 1.4%  |
| Painless swelling            | 2  | 1.4%  |
| Pupillary notch, DOV         | 2  | 1.4%  |
| Thread-like strand in pupil  | 2  | 1.4%  |
| Absence of eye, DOV (OS)     | 1  | 0.7%  |
| DOV, pupillary notch         | 1  | 0.7%  |
| Small eye (OU)               | 1  | 0.7%  |
| Watering, FB sensation       | 1  | 0.7%  |
| Watering, FB sensation, Growth in limbal area | 1 | 0.7% |
| Watering, FB sensation, Growth in limbus | 1 | 0.7% |
| Watering, FB sensation, Growth in superotemporal limbus | 1 | 0.7% |

DOV = decrease of vision, FB: foreign body

Table 3: Distribution of congenital anomaly of the patients

| Congenital anomaly          | n  | %  |
|------------------------------|----|----|
| CNLDO                        | 29 | 20.7% |
| Congenital cataract          | 21 | 15.0% |
| Anophthalmos                 | 12 | 8.6%  |
| Coloboma IRIS                | 10 | 7.1%  |
| Microphthalmos               | 10 | 7.1%  |
| Congenital glaucoma          | 8  | 5.7%  |
| Microcornea                  | 8  | 5.7%  |
| PPM                          | 8  | 5.7%  |
| Congenital esotropia         | 6  | 4.3%  |
| MNFL                         | 5  | 3.6%  |
| Dermoid cyst                 | 4  | 2.9%  |
| Limbal dermoid               | 4  | 2.9%  |
| Aniridia                     | 3  | 2.1%  |
| Heterochromia IRIDIS         | 3  | 2.1%  |
| Optic disc coloboma          | 3  | 2.1%  |
| Cryptophthalmos              | 2  | 1.4%  |
| Microphthalmia               | 2  | 1.4%  |
| Coloboma choroid             | 1  | 0.7%  |
| Optic nerve hypoplasia       | 1  | 0.7%  |

CNLDO = congenital nasolacrimal duct obstruction

between laterality and congenital anomaly of the eye (P < 0.001). Microphthalmia was prevalent significantly in higher proportion in both eyes and CNLDO was prevalent in significantly higher proportion in single eye (P < 0.001). Consanguinity was noted in only 5.7% cases. No statistically significant association was obtained between laterality of eye and consanguinity of patients (P = 0.41). However, the proportion of patients with history of consanguinity was higher in bilateral (7.5%) than unilateral (5.0%) cases.

Discussion

The prevalence rate of congenital ocular anomalies in the present study was 2.46%. A similar prevalence rate of 2.4% has been reported by Tomairek et al.[3] Prevalence rates of 1.7% and 0.75% have been reported by Lawan,[4] and Stoll et al.[5] Most of the patients in the present study (56.4%) were in the 4–5 years age group. Tupe and Chaudhari[6] in their study on the prevalence of congenital ocular anomalies have reported maximum patients in the 0–2 year’s age group. There was a significant association between age and congenital anomaly of the patients (P < 0.001). CNLDO was prevalent significantly in higher proportion among the patients with age ≤3 years and congenital cataract was prevalent in a significantly higher proportion among the patients in the 4–5-year age group (P < 0.001). Our findings on the prevalence of congenital cataract differ from the findings of Rahi and Dezateaux[7] who stated that the adjusted annual age-specific incidence of new diagnosis of congenital and infantile cataract was highest in the first year of life, being 2.49 per 10,000 children.

In the present study, the proportion of males affected was significantly higher than females (Z = 3.95, P < 0.001). The male to female ratio was 1.1:1. Behera et al.[3] have reported a male and female ratio of 1.4:1 in their study. But in a retrospective non-comparative case-series study of 54 cases in 8 years in Enugu by Chuka-Okosa et al.[8] in 2005, the male:female ratio was found to be 1:1.2. CNLDO was prevalent significantly in higher proportion among the male patients and congenital cataract prevalent in a significantly higher proportion among the female patients (P < 0.001). However, Rahi and Dezateaux[7] have reported equal incidence of cataract in either sexes. CNLDO was the most common ocular anomaly detected in our study (20.07%). This is in agreement with findings of other studies,[3,8,9] Tomairek et al.[3] reported congenital cataract to be the most common congenital ocular anomaly in their study. In his study on congenital and adnexal anomalies, Lawan[4] has reported buphthalmos to be the most common anomaly in 38% cases followed by cataract and CNLDO in 35% and 14% cases, respectively. Decreased vision was the most common presenting symptom in our study followed by watering and leukocoria. Watering from eyes was the most common complaint in cases with CNLDO. Watering from eyes was the most common presenting symptom in studies done by Behera et al.[3] and Tupe and Chaudhari,[6] Tomairek et al.[3] have reported leukocoria to be the most common presenting symptom in their study on the prevalence of congenital ocular anomalies among children with genetic disorders.

Unilateral involvement was noted in 71.4% cases, which was significantly higher than bilateral cases (28.6%). Lawan[4] has reported unilateral and bilateral involvement in 57.9% and 42.1% of their cases, respectively. Microphthalmia and iris
coloboma were prevalent significantly in higher proportion bilaterally and CNLDO was prevalent in significantly higher proportion unilaterally ($P < 0.001$). In our study, congenital cataract was prevalent bilaterally in 57.7% cases and unilaterally in 42.3% cases. Similar results have been reported in the study by Rahi and Dezateaux,[1] who have stated that the incidence of bilateral congenital cataract was higher than unilateral cases. Consanguinity can be an important cause of congenital ocular defects. The prevalence of consanguinity was 5.7% in our study. This is much less than the prevalence rate of 39.2% reported in the study by Mohammed et al.[8] The low prevalence in our study can be attributed to increasing modernization due to which the frequency of such marriages is on the wane.

Maternal malnutrition, infection, and exposure to toxic drugs during the pre-natal period can be associated with significant birth defects.[10–12] There was a history of fever and flu-like symptoms in 4 of our cases. One case had a history of malnutrition. There was a history of maternal alcohol intake and use of anti-tubercular therapy in one each case. One patient had vitamin-A deficiency. The congenital abnormalities noted in the above cases were congenital cataracts, congenital glaucoma, iris colobomas, and microphthalmia. Microphthalmia has been associated with maternal alcohol use during pregnancy.[13]

There is a tendency of ocular anomalies to be simultaneously associated with other systemic findings.[14,15] Systemic associations were noted in 2.14% of cases. Congenital heart disease was seen to be associated with a case of optic disc coloboma (0.7%). Limb deformity and webbed neck were seen in a case of anophthalmia (0.7%) and limb anomalies, flat nasal bridge, slanting palpebral fissure, and webbed neck were seen in a case of congenital cataract (0.7%).

Limitations of the present study include the small sample size and absence of genetic studies. Genetic study could not be performed because of its nonavailability in our setting.

**Conclusion**

Congenital ocular anomalies can be associated with poor prognosis. We can prevent some of the congenital ocular anomalies by simple awareness among the community. Proper knowledge of the developmental pathogenesis of congenital ocular anomalies is highly important for correct diagnosis and early intervention. Preventive measures can be adopted if history is taken properly during evaluation of the patients because maternal infection and systemic involvement have a great impact in this context. Community genetic services (counseling) should be integrated into the primary healthcare systems. Preconceptional counseling of prospective parents can maximize the chances of the couple having a healthy baby. Premarital counseling for consanguinity and screening for carriers of genetic diseases can help prevent many congenital diseases. We believe that our study estimates will serve as a reference guide for clinicians and other health professionals in counseling and public health planning.

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**Conflicts of interest**

There are no conflicts of interest.

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