

**Paediatric Ocular Diseases in a Tertiary Care Eye Centre**Ashwani Meherda<sup>1</sup>, Deepak Choudhury<sup>2</sup>, D. Anuradha<sup>3</sup><sup>1</sup>Chief Medical Officer, Safdarjung Hospital, New Delhi, India<sup>2</sup>Assistant Professor, Department of Ophthalmology, SCB Medical College, Cuttack, Odisha, India<sup>3</sup>Assistant Professor, Department of Ophthalmology, MKCG Medical College, Berhampur, Odisha, India

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Conflict of interest: Nil

**Abstract:****Background:** Congenital ocular anomalies are one of the important causes of childhood ocular morbidity and blindness. Visual impairment is one of the most common disabilities affecting children. A chromosomal/ genetic abnormality, an intrauterine infection, or maternal toxin is often involved.**Aim:** To study various paediatric ocular diseases in a tertiary care eye centre of Southern Odisha.**Material & Methods:** This was a prospective observational type of study of 2 years duration (from October 2017 to September 2019). 155 eyes of 118 patients of pediatric age group of both sexes with congenital ocular anomalies were included in the study. Detailed patient history and maternal obstetrics history was taken. Detailed systemic examination was done to rule out presence of any other malformations. Detailed ocular examination was done and recorded. All laboratory investigations were done.**Results:** Out of 118 patients, 69 (58.47%) cases were males & 49 (41.53%) cases were females. Male to female ratio was 1.4:1. The age of presentation of congenital ocular anomalies was found to be highest (40.67%) in the age group of < 1 years. History of consanguineous marriage was found in 11 (9.32%) cases. The most common mode of presenting symptom was watering (41.52%), followed by defective vision (28.82%). Nasolacrimal duct obstruction was the most common anomaly, found in 52 (44.06%) cases, followed by coloboma, found in 30 (25.43%) cases.**Conclusion:** Majority of cases were reported in less than 1 year of age group. Congenital nasolacrimal duct obstruction was found to be the most common congenital ocular anomaly followed by coloboma and congenital cataract.**Keywords:** Congenital ocular anomalies, Nasolacrimal duct obstruction, Coloboma, Genetic abnormality, Intrauterine infection.

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**Introduction**

Congenital ocular anomalies are one of the important causes of childhood ocular morbidity and blindness[1]. Visual impairment is one of the most common disabilities affecting children. There is an estimated 1.4 million children worldwide who are blind, two-thirds of whom live in developing countries[2]. The genetic and hereditary eye diseases account for 11–39% of childhood blindness with more common in developed than in the developing world[3]. Anomalies may involve a single ocular tissue, a region of the eye, or the entire eye. They may be unilateral or bilateral and may occur in association with a constellation of cranial, facial, or systemic abnormalities. A chromosomal/genetic abnormality, an intrauterine infection, or maternal toxin is often involved. Overlapping clinical manifestations from different genetic and non-genetic causes occur as a result of spatial and temporal targeting of specific developmental processes and embryonic events[4]. Early childhood

screening is advocated to enable early detection and prompt intervention[5]. This study is done to review the clinical profile of congenital ocular anomalies with various epidemiological parameters.

**Materials & Methods**

This was a prospective observational type of study of 2 years duration (from October 2017 to September 2019).

**Inclusion criteria**

Patients of pediatric age group of both sexes with congenital ocular anomalies.

**Exclusion criteria**

Patients having ocular diseases due to other acquired causes like trauma.

155 eyes of 118 cases with congenital ocular anomalies were included in the study. Detailed

patient history and maternal obstetrics history was taken. Any significant antenatal events were noted. Detailed systemic examination was done to rule out presence of any other malformations.

Visual acuity was measured by Snellen's chart. Ocular movements were examined. Slit lamp examination was done to evaluate the anterior segment. Lacrimal passage irrigation was done to test the patency.

Intraocular pressure (IOP) measurement was done by non-contact tonometer. Both direct and indirect ophthalmoscopy was done to rule out any posterior segment anomalies. B-scan ultrasonography was done when the media was hazy. Optical coherence tomography was done whenever required.

Laboratory investigations like differential count, total leukocyte count, hemoglobin, erythrocyte sedimentation rate, Montoux test, urine routine & microscopy was done. X-ray chest was done

whenever required. All abnormal findings were noted.

### Results

Out of 118 patients, 69 (58.47%) cases were males & 49 (41.53%) cases were females. Male to female ratio was 1.4:1. The age of presentation of congenital ocular anomalies was found to be highest (40.67%) in the age group of < 1 years and lowest (38.98%) in the age group of 1 to 5 years.

Age group of 6 to 14 years had 38.98% cases. Bilateral involvement was seen in 66.11% of cases. History of consanguineous marriage was found in 11 (9.32%) cases. Visual acuity (VA) could not be recorded in 55.08% cases. VA in 32 (27.12%) cases was found to be less than 6/60 and in 6 (5.08%) cases VA was more than 6/12. Associated systemic findings were found in 10 (8.47%) cases. 4 cases of coloboma were associated with mental retardation and another 4 were associated with deafness. Table-1 shows various modes of presentation.

**Table 1: Modes of presentation of symptoms**

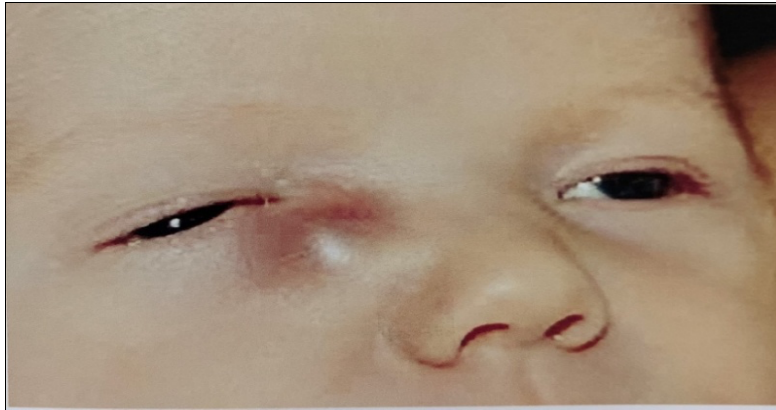
Presenting symptom	Number of cases	Percentage
Defective vision	34	28.82
Watering	49	41.52
Swelling	15	12.72
Large eye	4	3.38
Small eye	7	5.94
White papillary reflex	6	5.08
Absence of eye	2	1.69
Different iris color	1	0.85
Total	118	100

The most common mode of presenting symptom was watering (41.52%), followed by defective vision (28.82%) and swelling (12.72%). Table-2 shows various types of congenital anomalies.

**Table 2: Types of congenital anomalies**

Congenital ocular anomaly	Number of cases	Percentage
Nasolacrimal duct obstruction	52	44.06
Coloboma	30	25.43
Congenital cataract	16	13.57
Congenital glaucoma	5	4.25
Hemangioma	2	1.69
Congenital ptosis	2	1.69
Duane's retraction syndrome	2	1.69
Anophthalmos	2	1.69
Limbal dermoid	1	0.85
Crouzon's syndrome	2	1.69
Congenital entropion	2	1.69
Heterochromia iridium	1	0.85
Dermolipoma	1	0.85
Total	118	100

Nasolacrimal duct obstruction was the most common anomaly, found in 52 (44.06%) cases, followed by coloboma, found in 30 (25.43%) cases and congenital cataract in 16 (13.57%) cases. Typical coloboma was found in 24 (80%) cases. Figure 1 shows a case with lacrimal area swelling due to nasolacrimal duct obstruction and figure 2 shows a case with congenital cataract of left eye.



**Figure 1: Case with lacrimal area swelling due to nasolacrimal duct obstruction.**



**Figure-2: Case with congenital cataract of left eye**

Out of 30 eyes of 16 patients with congenital cataract, cataractous lens extraction with PCIOL implantation was done in 28 eyes (93.33%). Out of 6 eyes with congenital glaucoma, trabeculectomy was done in 3 eyes (50%). All cases of CNLDO were first advised for massage of lacrimal sac area with maintenance of lid hygiene. 50 eyes out of 62 eyes responded to it. In 10 eyes of 6 cases, probing was done. In rest 2 cases dacryocystorhinostomy surgery was done. Excision was done in 1 dermoid case.

### Discussion

Out of 118 patients, 69 (58.47%) cases were males & 49 (41.53%) cases were females. Male to female ratio was 1.4:1. Present study is in agreement with the study of Lawan A[6]. He observed a male-female ratio of 2.3:1 in 69 patients, but Chuka-Okosa et al, in a retrospective non comparative case series study of 54 patients in Enugu found male to female ratio to be 1:1.2[7].

According to a study by Tupe PN et al, in which 9350 patients were screened, male: female ratio was 1:1.4[8]. The age of presentation of congenital ocular anomalies in our study was found to be highest (40.67%) in the age group of < 1 years and lowest (38.98%) in the age group of 1 to 5 years. In the study by Tupe PN et al, they found maximum

cases (54%) were in age group of 0-2 years[8]. According to Rahi JS et al, the adjusted 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. Adjusted cumulative incidence at 5 years was 3.18 per 10,000, increasing to 3.46 per 10,000 by 15 years[9].

In our study, bilateral involvement was seen in 66.11% of cases. According to Lawan A, in 57.9% cases were bilateral[6]. History of consanguineous marriage was found in 11 (9.32%) cases in our study. According to the study of Hornby S et al, history of consanguinity was found in almost 50% cases[10]. According to Nath A et al, out of 500 women studied, consanguinity was found in 36% of marriages[11]. Consanguinity was reported 76.7% by Tomairek et al in a study of congenital ocular anomalies in children with genetic disorders[12]. Associated systemic findings were found in 10 (8.47%) cases in our study. According to Bermejo et al, the tendency of eye malformation to be associated with other congenital abnormalities was also evident[13]. According to Bashour M et al, ocular abnormalities were present in 20% of patients, and systemic abnormalities were present in almost 25% of patients with serious nasolacrimal duct anomalies[14].

In our study, the most common mode of presenting symptom was watering (41.52%), followed by defective vision (28.82%) and swelling (12.72%). Martinez et al found the most frequent presenting feature was leucocoria, seen in 44% of patients[15]. In our study, nasolacrimal duct obstruction was the most common anomaly, found in 52 (44.06%) cases, and followed by coloboma, found in 30 (25.43%) cases, and congenital cataract in 16 (13.57%) cases. According to Lawan et al, the common congenital anomalies were buphthalmos (38%), cataracts (35%) and NLDO (14%). While according to Bermejo et al, anophthalmos/ microphthalmos to be most common (21.34/100000) followed by congenital cataract (6.31) followed by coloboma (4.89)[13]. But according to Stoll et al, anophthalmos/ microphthalmos was found to be only 3.9%[16]. On wising we did a hospital based study and found cataract to be the most common anomaly (30.7%) followed by squint (19.8%)[17].

In our study, out of 30 eyes of 16 patients with congenital cataract, cataractous lens extraction with PCIOL implantation was done in 28 eyes (93.33%), and out of 6 eyes with congenital glaucoma, trabeculectomy was done in 3 eyes (50%). In a study conducted by Lawan A, 268 children out of 4163 had undergone surgery. Surgery for congenital eye and adenexal anomalies accounted for 25.7% of eye surgeries in the study population[6]. According to Martinez et al, 58% of the congenital cataracts were treated by surgery[15].

### Conclusion

Majority of cases were reported in less than 1 year of age group. Congenital nasolacrimal duct obstruction was found to be the most common congenital ocular anomaly followed by coloboma and congenital cataract. Meticulous and skillful surgical intervention can bring about cure of few segments of these anomalies.

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