# Clinical Profile Of Congenital Eye Abnormalities In Children Aged 0-5 Years And Their Correlation With Demographic Factors

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**KEYWORDS**

Congenital eye anomalies, pediatric ophthalmology, visual impairment, congenital cataract, demographic factors

**ABSTRACT:**

***Background:*** Congenital eye abnormalities encompass a wide range of structural and functional defects present at birth, contributing significantly to childhood visual impairment and blindness. Understanding their clinical profile and demographic associations is essential for early detection and prevention. ***Aim of the study:*** To evaluate the clinical spectrum of congenital eye abnormalities in children aged 0-5 years and analyze their correlation with demographic factors. ***Methods:*** This hospital-based observational study was conducted over one year in Bangladesh. Eighty-two pediatric patients presenting with congenital ocular anomalies were included. Data on clinical features, antenatal history, and demographic variables were collected using a structured questionnaire. Comprehensive ophthalmologic examinations were performed, and findings were analyzed using SPSS v26.0. ***Result:*** Children aged 4-5 years constituted the largest group (56.10%), with a male predominance (64.63%). The most common presenting symptom was decreased ocular vision (18.29%), followed by watering of eyes (13.41%). Congenital cataract was the most prevalent anomaly (46.74%), followed by coloboma of the iris and choroid (33.62%), and anophthalmos (19.68%). Bilateral involvement was more frequent than unilateral presentation. ***Conclusion:*** Congenital cataract, coloboma, and whole globe anomalies are the predominant ocular abnormalities in early childhood. Timely screening and intervention strategies are essential to minimize long-term visual disability, especially in resource-limited settings.

## INTRODUCTION

Congenital eye abnormalities (CEAs) represent a spectrum of structural and functional ocular defects present at birth, arising from disruptions during embryonic development of the visual system [1]. These anomalies can involve the anterior or posterior segment of the eye, extraocular structures, or the entire globe, and often result in significant visual impairment or irreversible blindness during early childhood [2]. Globally, congenital eye anomalies are estimated to affect approximately 12-15 per 10,000 live births, contributing considerably to pediatric visual disability and accounting for nearly 7% of childhood blindness [3]. In Bangladesh, the prevalence of childhood visual impairment due to congenital causes has been reported at 11.5%, reflecting a significant national public health concern [4]. CEAs encompass a wide range of ocular disorders, including relatively minor conditions such as epicanthus, microcornea, or congenital nasolacrimal duct obstruction, as well as severe malformations like anophthalmia, coloboma, congenital glaucoma, and congenital cataract [5]. These abnormalities may present as isolated ocular defects or occur in association with multisystem syndromes. The etiology of CEAs is multifactorial, often involving genetic mutations, chromosomal anomalies, intrauterine infections (particularly TORCH complex), teratogenic exposures, maternal metabolic conditions, or nutritional deficiencies during pregnancy [6]. In addition to biological factors, several demographic and environmental variables have been associated with an increased risk of CEAs. These include advanced maternal or paternal age, consanguinity, low socioeconomic status, inadequate prenatal care, poor maternal nutrition, and exposure to environmental toxins [7]. Understanding the interplay between these factors is crucial for early identification and prevention strategies. Early diagnosis and prompt intervention are essential to prevent permanent visual impairment, support visual rehabilitation, and ensure proper neurovisual development [8]. However, in many low-

and middle-income countries (LMICs), including Bangladesh, children with CEAs often present late due to a lack of neonatal screening programs, limited parental awareness, socio-cultural barriers, and insufficient pediatric ophthalmology services [9]. This delay in diagnosis significantly compromises treatment outcomes, particularly for vision-threatening conditions such as congenital cataracts or glaucoma [10]. Despite the global burden, there remains a dearth of region-specific epidemiological data on CEAs, especially regarding their clinical spectrum and demographic associations in early childhood [11]. This gap in knowledge is even more pronounced in South Asia, where studies focusing exclusively on congenital ophthalmic disorders in children aged 0–5 years are limited. Such data are essential to develop targeted screening initiatives, allocate resources, and design culturally appropriate public health interventions aimed at preventing childhood blindness [12]. Congenital eye abnormalities are defined as structural or functional ocular defects present at birth due to disturbances in the embryonic development of the eye [13]. This study aims to evaluate the clinical profile of congenital eye abnormalities among children aged 0–5 years and investigate their correlation with key demographic factors, thereby contributing to early diagnosis, prevention, and comprehensive pediatric eye care planning.

## METHODOLOGY & MATERIALS

This was a hospital-based observational study conducted over a one-year period, from January 2020 to December 2020, at the Vitreo-Retina, Bangladesh Eye Hospital, Dhaka, Bangladesh. The aim of the study was to investigate the clinical profile of congenital eye abnormalities in children aged 0 to 5 years and examine their correlation with demographic factors. Patients were enrolled in the study based on predefined inclusion and exclusion criteria. All eligible participants who met the criteria were included in the study sample.

### Inclusion Criteria:

- Pediatric patients aged 0 to 5 years who presented to the ophthalmology outpatient department with congenital ocular anomalies were included.

### Exclusion Criteria:

Children were excluded if they had:

- Acquired ocular abnormalities,
- A history of ocular trauma,
- Ocular changes due to nutritional deficiencies.

### Ethical Considerations

The study received prior approval from the Institutional Ethics Committee, and written informed consent was obtained from the parents or guardians of all participants.

### Data Collection

Data were collected using a semi-structured, pretested questionnaire administered to the parent or adult accompanying the child. Relevant demographic details (age, sex), presenting symptoms, and clinical signs, and types of congenital anomaly were recorded. Detailed antenatal history including any maternal illness, medication use, radiation exposure, nutritional intake during pregnancy, and family history of ocular disorders were also documented.

Vision assessment in infants was done using torchlight, pupillary responses, and the illiterate E-chart. Anterior segment examination was conducted using a slit-lamp biomicroscope. Lacrimal syringing was performed for suspected congenital nasolacrimal duct obstruction (CNLDO), and intraocular pressure was measured using Schiotz tonometry to identify congenital glaucoma. Additional investigations included keratometry and A-scan ultrasonography for cases of congenital cataract to assess corneal diameter and intraocular lens power. Fundus examination was performed under pharmacological dilation using direct and indirect ophthalmoscopes. Strabismus evaluation (if present) included cover tests and the Hirschberg test. Examination under anesthesia was carried out when necessary.

### Statistical Analysis

Data were entered into Microsoft Excel and analyzed using SPSS version 26.0. Descriptive statistics including frequencies and percentages were used to summarize demographic variables, clinical symptoms and signs, and the distribution of various congenital ocular anomalies.

## RESULT

The majority of the children (56.10%) were in the 4-5 years age group, followed by 36.59% in the 1-4 years group, and 7.32% were under 1 year of age. In terms of gender distribution, males were more commonly affected, accounting for 64.63% of the cases (Table 1). Table 2 shows that the most commonly reported symptom was

dimness of vision (DOV), observed alone in 18.29% of cases, and in combination with other signs in several others. Watering of the eyes was also frequently reported, either alone (13.41%) or alongside other symptoms such as discharge, swelling, or foreign body sensation. Other notable findings included white reflex with DOV (9.76%), absence or small size of the eye (each 7.32%), large eyes with watering (6.10%), and small cornea (6.10%). Less common presentations included keyhole iris, fused eyelids, pupillary abnormalities, and ocular growths, each accounting for 1.22%–4.88% of cases (Table 2). The most common anomaly observed was congenital cataract, affecting both eyes in 57 cases (46.74%). Coloboma of the iris and choroid was the second most prevalent, seen bilaterally in 41 children (33.62%) and unilaterally in 5 right eyes (4.10%). Anophthalmos was present bilaterally in 24 cases (19.68%), with additional unilateral involvement in both left (3.28%) and right eyes (4.92%). Microphthalmos with microcornea affected both eyes in 15 children (12.3%), while congenital nasolacrimal duct obstruction (CNLDO) was also exclusively bilateral (12.3%). Other less frequent anomalies included congenital glaucoma (7.38%), optic disc hypoplasia (6.56%), congenital esotropia (4.92% bilateral), congenital corneal opacity (3.28%), congenital ptosis (2.46% combined), and PHPV (0.82%). Bilateral involvement was notably more common than unilateral presentation across most anomalies (Table 3).

**Table 1: Demographic characteristics of the study population (n=82)**

| Variables | Frequency (n) | Percentage (%) |
|---|---|---|
| Age Group (Years) | | |
| <1 | 6 | 7.32 |
| 1–4 | 30 | 36.59 |
| 4–5 | 46 | 56.10 |
| Gender | | |
| Male | 53 | 64.63 |
| Female | 29 | 35.37 |

**Table 2: Symptoms and sign of the patients (n=82)**

| Symptoms and Sign | Frequency (n) | Percentage (%) |
|---|---|---|
| DOV | 15 | 18.29 |
| Watering | 11 | 13.41 |
| White reflex, DOV | 8 | 9.76 |
| Absence of eye | 6 | 7.32 |
| Small eye | 6 | 7.32 |
| Large eyes, watering | 5 | 6.10 |
| Small cornea | 5 | 6.10 |
| Keyhole Iris, DOV | 4 | 4.88 |
| Watering, discharge, swelling | 4 | 4.88 |
| Inward deviation of eye, DOV | 2 | 2.44 |
| Watering, Periocular crusting | 2 | 2.44 |
| DOV, different iris color | 1 | 1.22 |
| Fused eyelids | 1 | 1.22 |
| Keyhole Iris | 1 | 1.22 |
| Painless cystic swelling | 1 | 1.22 |
| Painless swelling | 1 | 1.22 |
| Pupillary notch, DOV | 1 | 1.22 |
| Thread-like strand in pupil | 1 | 1.22 |
| Absence of eye, DOV (OS) | 1 | 1.22 |
| DOV, pupillary notch | 1 | 1.22 |
| Small eye (OU) | 1 | 1.22 |
| Watering, FB sensation | 1 | 1.22 |
| Watering, FB sensation, Growth in limbal area | 1 | 1.22 |
| Watering, FB sensation, Growth in limbus | 1 | 1.22 |
| Watering, FB sensation, Growth in superotemporal limbus | 1 | 1.22 |

**Table 3: Distribution of various types of congenital anomaly (n=82)**

| Anomaly Type | Left Eye | | Right Eye | | Both Eyes | |
|---|---|---|---|---|---|---|
| | n | % | n | % | n | % |
| Anophthalmos | 4 | 3.28 | 6 | 4.92 | 24 | 19.68 |
| Microphthalmos & Microcornea | 3 | 2.46 | 4 | 3.28 | 15 | 12.3 |
| CNLDO (Congenital NLD Obstruction) | 0 | 0.00 | 0 | 0.00 | 15 | 12.3 |
| Congenital Ptosis | 1 | 0.82 | 2 | 1.64 | 1 | 0.82 |
| Congenital Esotropia | 0 | 0.00 | 1 | 0.82 | 6 | 4.92 |
| Congenital Corneal Opacity | 0 | 0.00 | 0 | 0.00 | 4 | 3.28 |
| Coloboma of Iris and Choroid | 0 | 0.00 | 5 | 4.10 | 41 | 33.62 |
| Congenital Cataract | 0 | 0.00 | 0 | 0.00 | 57 | 46.74 |
| Congenital Glaucoma | 0 | 0.00 | 0 | 0.00 | 9 | 7.38 |
| PHPV (Persistent Hyperplastic Primary Vitreous) | 0 | 0.00 | 0 | 0.00 | 1 | 0.82 |
| Hypoplasia of Optic Disc | 0 | 0.00 | 0 | 0.00 | 8 | 6.56 |

## DISCUSSION

Congenital eye abnormalities are structural or functional defects of the eye present at birth, often resulting in visual impairment or blindness if not detected early. These anomalies can vary widely in presentation and are influenced by genetic, environmental, and demographic factors. Understanding their clinical profile in early childhood is crucial for timely diagnosis and intervention. This study investigated the clinical spectrum and demographic associations of congenital eye abnormalities in children aged 0-5 years. The majority of cases were observed in the 4-5-year age group (56.10%). Tupe and Chaudhari (2015) reported that the majority of congenital ocular anomaly cases were observed in children aged 0-2 years, with a statistically significant correlation between age and the presence of congenital anomalies ($P<0.001$) [14]. In contrast, our findings regarding the prevalence of congenital eye abnormalities differ from those of Rahi and Dezateaux (2001), who noted that the highest age-specific incidence of newly diagnosed congenital and infantile cataracts occurred within the first year of life, at a rate of 2.49 per 10,000 children [15]. In the present study, a male predominance was observed, with 64.63% of the cases being male. Behera et al observed a male-to-female ratio of 1.4:1 in their study [16]. In this study, decreased ocular vision (DOV) was the most common presenting symptom (18.29%), often associated with anomalies like congenital cataract or optic nerve hypoplasia. Watering of the eyes (13.41%) was the next frequent complaint, commonly linked with CNLDO and anterior segment anomalies. Watering from the eyes was the most frequently reported presenting symptom in the studies conducted by Tupe and Chaudhari (2015) and by Behera et al [14,16]. White reflex (9.76%) was another significant sign, raising concern for serious conditions like congenital cataract. In the study on the prevalence of congenital ocular anomalies in children with genetic disorders, Tomairek et al identified leukocoria as the most commonly observed presenting symptom [17]. Absence of the eye and small eye each occurred in 7.3% of patients. Rare signs and combinations such as keyhole iris with DOV, pupillary notch with DOV, FP thread-like strands, limbal growths, fused eyelids were present in low frequencies (1-5%). While these are individually uncommon, they emphasize the heterogeneity of congenital ocular presentations and the necessity for detailed anterior segment evaluation. The presence of complex mixed features (e.g., DOV plus different iris colour, or watering plus limbal growths) suggests syndromic or anterior segment dysgenesis presentations. The most frequently observed disorder in our study was congenital cataract, followed by uveal coloboma and then whole globe anomalies such as microphthalmos and anophthalmos. This pattern is consistent with findings from other studies [17-19]. Coloboma, the second most frequent anomaly in our cohort, was present in 33.62% of cases, primarily affecting both eyes. Study reported that coloboma is one of the most common ocular malformations in children with an estimated incidence of 10–20 cases per 100,000 newborns [20]. Anophthalmos was present bilaterally in 19.68% of cases. Microphthalmos with microcornea was detected in 12.3% of patients, consistent with findings from Bermejo and Martínez-Frías (1998), who also documented a similar incidence of microphthalmic conditions in congenital ocular anomaly databases [21]. Congenital nasolacrimal duct obstruction (CNLDO) accounted for 12.3% of bilateral cases in our study. This condition, although generally underreported in anomaly registries, is one of the most common causes of epiphora in infants and was similarly highlighted in the work of MacEwen and Young (1991), who reported an incidence ranging from 5–20% in various populations [22]. Less frequent but noteworthy anomalies in our study included congenital glaucoma (7.38%), optic disc hypoplasia (6.56%), and congenital esotropia (4.92%). Interestingly, rare anomalies such as persistent hyperplastic primary vitreous (PHPV) were also documented (0.82%), reinforcing the need for early and comprehensive ocular screening in neonates and infants, particularly those with risk factors such as consanguinity or low birth weight.

***Limitations of the study:***

- The data were collected over a one-year period, which is insufficient to assess seasonal variations in birth defects or track delayed diagnoses.
- No genetic testing or pediatric syndromic screening was performed, limiting the ability to identify underlying genetic syndromes or chromosomal abnormalities associated with ocular defects.

## CONCLUSION

This study highlights that congenital cataract, coloboma, and anophthalmos are the most frequently encountered congenital eye anomalies in children aged 0–5 years. A significant number of cases presented with bilateral involvement and late diagnoses, underscoring the need for early and comprehensive pediatric ophthalmologic screening programs in Bangladesh. Increased parental awareness, improved access to neonatal eye care, and integration of genetic counseling are recommended to address preventable childhood blindness. Further large-scale, multicentric studies are needed to explore regional variations, risk factors, and long-term outcomes of CEAs.

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