



RESEARCH ARTICLE

STUDY OF OCULAR ANOMALIES IN PEDIATRIC PATIENTS AT RURAL TERTIARY CARE HOSPITAL

Aditi Agresar¹, Shubhangi Nigwekar² and Sakshi Bagdiya³

1. Junior Resident, Department of Ophthalmology, DBVP Rural Medical College and Dr. Vitthalrao Vikhe Patil Pravara Rural Hospital, Loni.
2. Professor, Department of Ophthalmology, DBVP Rural Medical College and Dr. Vitthalrao Vikhe Patil Pravara Rural Hospital, Loni.
3. Junior Resident, Department of Ophthalmology, DBVP Rural Medical College and Dr. Vitthalrao Vikhe Patil Pravara Rural Hospital, Loni.

Manuscript Info

Manuscript History

Received: 27 November 2024

Final Accepted: 30 December 2024

Published: January 2025

Key words:-

Congenital Ocular Anomaly, Congenital Ocular Anomaly in Rural Population

Abstract

Introduction: The human eye develops from a complex interplay between three germ layers. i.e surface ectoderm, neuroectoderm, the mesoderm. The basic morphogenetic process of the eye is completed at the end of the 2nd month of gestation but the complete maturation takes place only after birth. Any derangement of the development process leads to a different congenital ocular anomaly. Many children with congenital ocular anomalies have low vision. Therefore, there is a growing need for early screening, detection, and treatment by pediatric or trained ophthalmologists who may provide the rehabilitation of these cases.

Aims and Objectives: To study the distribution of various ocular anomalies and to study its association with known risk factors (like consanguinity, antenatal insults like maternal malnutrition and infections, etc) in the pediatric age group at tertiary care hospital.

Materials and Methods: An Observational, Descriptive, Cross-Sectional, and Hospital Based Study in patients age group upto 18 years attending Ophthalmology OPD and referred patients at Dr. VitthalraoVikhePatilPravara Rural Hospital, Loni over 12 months period from March 2023 to February 2024. A total of 50 patients with congenital anomalies were screened and evaluated. Results: In this study, the range was from birth to 18 years of age. 40% of the cases(20 patients) were in the age group < 1 year. Out of 50 patients, there were 33 males (66%) and 17 females (34%). History of consanguinity present in 26% (13 patients) of cases. Antenatal insults present in 10% (5 patients) of cases. In this study, 18 patients in pre-school age were following the torch light as they were <1 yr of age. Total 9 (18%) patients had vision <3/60. Severe visual impairment (<6/60-3/60) seen in 4 (8%) patients. Moderate visual impairment (<6/18-6/60) seen in 11 (22%) patients. Mild or no visual impairment (6/18 or better) in 6 (12%) patients. The most common anomaly in our study was congenital dacryocystitis (30%), followed by congenital cataract (20%), followed by squint (18%), followed by ptosis (18%) and others were 14%.

Conclusion: Congenital ocular anomaly is one of the important cause of childhood blindness. Proper knowledge of the developmental pathogenesis of congenital ocular anomalies is highly important for correct diagnosis. The findings of our study can act as a reference guide for clinicians and health professionals for counselling, health planning and creating community awareness.

Copyright, IJAR, 2025,. All rights reserved.

Introduction:-

- Surface ectoderm, neuroectoderm, and mesoderm play a complex role in the development of the human eye.
- Although basic morphogenetic process of the eye is completed by the end of the second month of gestation, complete maturation takes place after birth⁽¹⁾.
- During this period, any disruption in the development process results in a different congenital ocular anomaly.
- Genetics, environmental factors,⁽²⁾ various teratogens, intrauterine infections, consanguinity of marriage, maternal malnutrition, drugs, smoking and alcohol consumption, as well as maternal metabolic disorders such as folic acid deficiency, diabetes, cretinism or chromosomal abnormalities in the developing embryo during intrauterine life, can all result in congenital ocular abnormalities. In addition to maternal infections and medications taken during pregnancy, radiation also play a role in causing congenital anomalies⁽³⁾.
- Low vision is common in children with congenital ocular abnormalities. Preventable causes of childhood blindness, such as cataracts, can be avoided with early treatment. Anatomically correctable anomalies, like iris and lens coloboma, can be vision rewarding in few cases with certain limitations, provided there are no associated complications. If congenital ocular anomalies are not treatable, early counselling is essential to guide in decisions regarding education, career choices, and support services.
- Therefore, there is a growing need for early screening, detection, and treatment by pediatric or trained ophthalmologists who may offer therehabilitation of these cases⁽⁴⁾.

Aim:

To study the distribution of ocular anomalies in pediatric age group at Dr. VitthalraoVikhePatilPravara Rural Hospital, Loni.

Objectives:

- 1)To study the distribution of various ocular anomalies in pediatric age group at Dr. VitthalraoVikhePatilPravara Rural Hospital, Loni.
- 2)To study its association with known risk factors(like consanguinity, antenatal insults like maternal malnutrition and infections, etc.).

Criteria For Inclusion Of Study Patients:

- 1)All patients in the age group upto 18 years attending Ophthalmology OPD and referred patients at Dr. VitthalraoVikhePatilPravara Rural Hospital, Loni.
- 2)Patients or parents willing to give written informed consent to participate in the study.

Criteria For Exclusion Of Study Patients:

- 1)Patients with Retinopathy of prematurity.

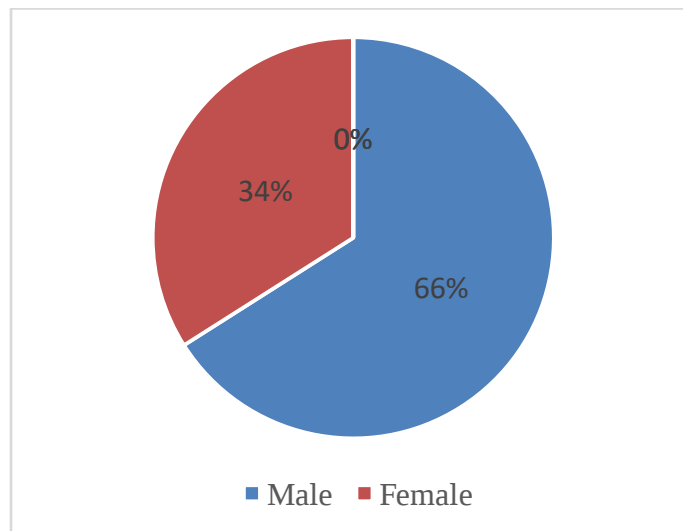
Materials And Methodology:

An observational, descriptive, cross-Sectional, and hospital based study conducted after Institutional Ethical Committee (IEC) approval in patients age group up to 18 years attending Ophthalmology OPD and referred patients at our OPD. A total of 50 patients with congenital anomalies were examined and documented over 12 months period from March 2023 to February 2024. We studied the patients age group up to 18 yrs with ocular anomalies. A complete and detailed examination carried out for any congenital ocular anomaly.

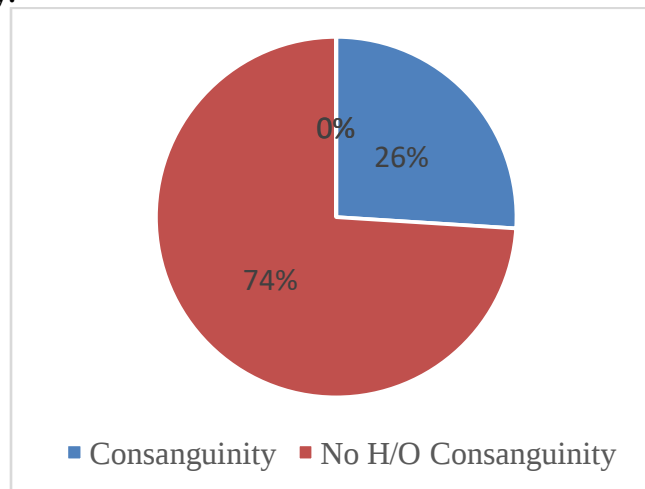
Results:**Age Group:****Table 1:-** Study showing age-wise distribution of 50 study patients with congenital ocular anomalies.

Age group	Number of cases	Percentage
< 1 year	20	40%
1-4 years	10	20%
4-5 years	9	18%
> 5 years	11	22%

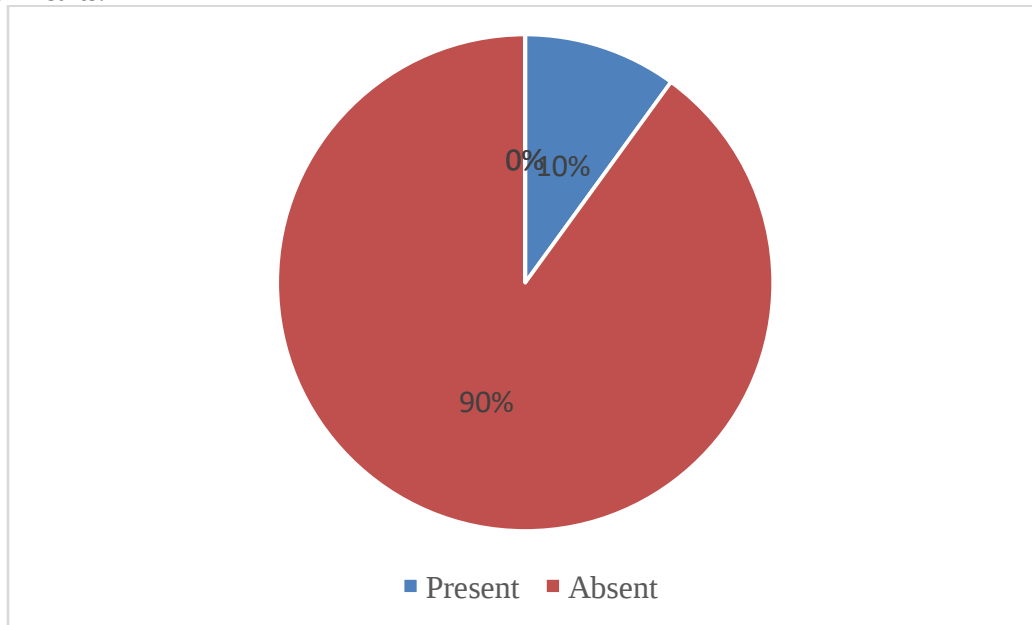
We studied the patients from birth to 18 years of age. 40% of study patients (20 patients) were in the age group < 1 year.

Gender:**Graph 1-** Gender-wise distribution.

Out of 50 study patients, there were 33 males (66%) and 17 females (34%).

History Of Consanguinity:**Graph 2-** History of consanguinity.

In our study, history of consanguinity present in 26% (13 patients) of cases.

Antenatal Insults:**Graph 3-** History of antenatal insults.

In our study, history of antenatal insults present in 10% (5 patients) of cases.

Visual Acuity Testing:**Table 2:-** Study showing visual acuity at presentation in 50 study patients with congenital ocular anomalies.

	Follows torch light	No PL	<3/60 to PL	<6/60-3/60	<6/18-6/60	6/18 or better	Not co-operative
Pre-school(<4 yrs)	18	2	1	1	4	2	2
School-going (4-18 yrs)	-	-	6	3	7	4	-

In our study, 18 patients in pre-school age were following the torch light as they were <1 yr of age.

Total 9 (18%) patients had vision <3/60.

In 4 patients (8%), severe visual impairment (<6/60-3/60) was seen.

In 11 (22%) patients, moderate visual impairment (<6/18-6/60) was seen

Mild or no visual impairment (6/18 or better) in 6 (12%) patients.

Congenital Ocular Anomalies:**Figure 1:-** (LE) Congenital dacryocystitis **Figure 2:-** (RE) Congenital cataract

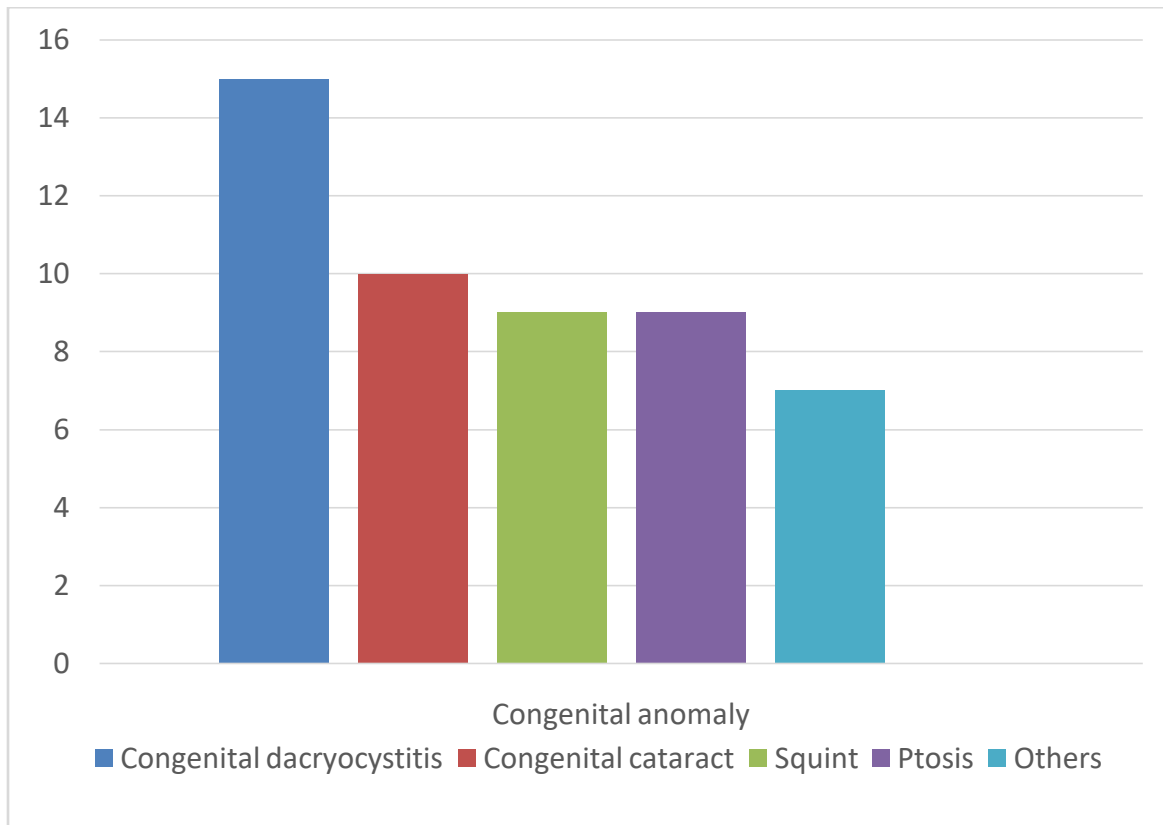


Figure 3:- (RE)Congenital mild ptosis**Figure 4:-** (BE)Alternate esotropia.

Table 3:- Study showing distribution of congenital ocular anomalies in 50 study patients.

Congenital anomaly	Number of cases	Percentage
Congenital dacryocystitis	15	30%
Congenital cataract	10	20%
Squint	9	18%
Ptosis	9	18%
Others	7	14%

In our study, the most common anomaly was congenital dacryocystitis (30%), followed by congenital cataract (20%), followed by squint (18%), followed by ptosis (18%) and others were 14% which included each one case of microphthalmia with total coloboma, microphthalmia with microcornea, buphthalmos, megalocornea, retinoblastoma, cryptophthalmos and crouzon's syndrome.



Graph 4 – Distribution of congenital ocular anomaly

Discussion:

From March 2023 to February 2024, an observational, descriptive, cross-sectional, and hospital based study was conducted at tertiary care hospital. The study involved all patients in the age group upto 18 years attending Ophthalmology OPD and referred patients at our OPD. A total of 50 patients with congenital anomalies were examined and documented over 12 months period. A complete and detailed examination carried out for any congenital ocular anomaly.

For simplicity, in table no. 2, we have tabulated the visual grading in 2 parts only.

1. Pre-school
2. School going

However, in pre-school, which contains early postnatal to 4 years age group. Visual acuity testing methodology obviously varies in these patients according to the age, IQ, developmental milestones, and the co-operativeness of child. Thus, the visual acuity results are not uniformly evaluated with snellen's chart. However, only 8 patients could answer correctly with snellen's chart.

Age:

In this study, the range was from birth to 18 years of age. 40% of the cases (20 patients) were in the age group < 1 year.

Tupe and Chaudhari⁽⁵⁾ in their study on the prevalence of congenital ocular anomalies have reported maximum patients in the 0–2 year's age group giving percentage of 54%. This may be because of literacy and early detection of congenital anomalies.

Gender:

In our study, out of 50 patients, there were 33 males (66%) and 17 females (34%). i.e. male to female ratio of 1.9:1.

In a study conducted by Dash P, et al⁽⁶⁾, there were 74 (52.9%) males and 66 (47.1%) females with male to female ratio of 1.1:1.

This finding was also similar to study by Chuka-Okosa, et al⁽⁷⁾. This study also reported male preponderance of congenital anomalies with male to female ratio of 1.2:1. This may be due to inheritance pattern of the genetic diseases.

History of consanguineous marriage:

We revealed the history of consanguineous marriage in 26% (13 patients) of cases.

A study conducted by Nirmalan *et al.*⁽⁸⁾ in Andhra Pradesh found that 26.7% of all screened subjects had a consanguineous parent. A hospital-based study in south India found that 28.8% of the patients who had a history of consanguinity had ocular genetic disorders⁽⁹⁾. This finding was similar to our study.

Antenatal insults:

According to the literature, genetic effects, environmental factors,⁽²⁾ various teratogens, maternal metabolic disturbances like folic acid deficiency, intrauterine infections, maternal malnutrition, drug abuse, smoking and alcohol consumption are known risk factors.

In our study, we found antenatal insults present in 10% (5 patients) of cases. There was history of maternal infection in 3.33% (2 patients), history of fever and flu-like symptoms in 3.33% (2 patients) whereas history of folic acid deficiency in 1.66% (1 patient). This suggests that the congenital anomalies that occur due to genetic and environmental causes affects the fetus in the early trimesters of gestation i.e. in the critical period of organogenesis.

Visual acuity:

According to WHO, Blindness is defined as visual acuity less than 3/60 in the better eye with best correction or a visual field of less than 10° from the point of fixation in each eye.

According to NPCB⁽¹⁰⁾,

Low vision = < 6/18 – 6/60

Economic blindness = $< 6/60 - 3/60$

Social blindness = $< 3/60 - 1/60$

Manifest blindness = $< 1/60 - PL$

Absolute blindness = No PL.

In this study, 9 patients (18%) were blind according to WHO criteria suggesting congenital ocular anomaly is one of the important cause of childhood blindness.

Distribution of congenital ocular anomalies:

Congenital dacryocystitis (30%) was the most common anomaly, followed by congenital cataract (20%), followed by squint (18%), followed by ptosis (18%) and others were 14%. This shows that congenital dacryocystitis was most common congenital anomaly in this age group.

Tupe and Chaudhari ⁽⁵⁾ reported congenital dacryocystitis (24%) as most common anomaly followed by congenital cataract as second common anomaly (14%) in their study. This finding was similar to our study.

Dash P, et al ⁽⁶⁾ found congenital nasolacrimal duct obstruction as the most common anomaly in 29 (20.71%) cases followed by congenital cataract in 21 (15%) cases in their study.

Limitations:

The limitations of this study – 1. sample size is small.

2. study design is cross sectional.

3. challenging visual assessment in various pediatric age groups.

Conclusion:

- Congenital ocular anomalies are one of the major causes of childhood blindness.
- Proper knowledge of the developmental pathogenesis of congenital ocular anomalies is highly important for correct diagnosis.
- In this small sample sized study done at tertiary care hospital which is a referral centre, we found wide spectrum of congenital ocular anomalies, both preventable and non-preventable.
- The findings of our study can act as a reference tool for clinicians and health providers for counselling, health planning and raising community awareness.

References:

1. Tamm ER, Ohlmann A. Development of the human eye. *Ophthalmology*. 2012 Sep;109(9):911-28.
2. Helen Dolk, Pat Doyle, Ester Garne. A review of environmental risk factors for congenital anomalies. 1st edition. (Uploaded 29 April 2004):7-30.
3. Rahi JS, Sripathi S, Gilbert CE, et al. The importance of pre-natal factor in childhood blindness in India. *Dev Med Child Neurol* 1997;39(7):449-455.
4. Titiyal JS, Pal N, Murthy GVS, et al. Causes and temporal trends of blindness and severe visual impairment in children in schools for the blind in North India. *Br J Ophthalmol* 2003;87(8):941-945.
5. Tupe PN, Chaudhari SV. A study on prevalence of congenital ocular anomalies in paediatric age group. *Int J Med Res Heal Sci*. 2015;4:884-8.
6. Dash P, Rout JP, Panigrahi PK. Clinical patterns of congenital ocular anomalies in the pediatric age group (0 to 5 years) and its association with various demographic parameters. *Indian J Ophthalmol*. 2022 Mar;70(3):944-947. doi: 10.4103/ijo.IJO_1862_21. PMID: 35225547; PMCID: PMC9114588.
7. Chuka-Okosa CM, Magulike NO, Onyekonwu GC. Congenital eye anomalies in Enugu, South-Eastern Nigeria. *West Afr J Med*. 2005 Apr-Jun;24(2):112-4. doi: 10.4314/wajm.v24i2.28178. PMID: 16092309.
8. Nirmalan PK, Krishnaiah S, Nutheti R, Shamanna BR, Rao GN, Thomas R. Consanguinity and eye diseases with a potential genetic etiology. Data from a prevalence study in Andhra Pradesh, India. *Ophthalmic Epidemiol*. 2006;13:7-13. doi: 10.1080/09286580500473795.
9. Kumaramanickavel G, Joseph B, Vidhya A, Arokiasamy T, Shridhara Shetty N. Consanguinity and ocular genetic diseases in South India: analysis of a five-year study. *Community Genet*. 2002;5:182-5. doi:10.1159/000066334.

10. Ananda Kumar B, Ravinder S, Vishnu Preeti, VirijaVirija, Harish Harish. "OCULAR MANIFESTATIONS IN HIV AND AIDS INDIVIDUALS AT SAROJINI DEVI EYE HOSPITAL, HYDERABAD", Journal of Evolution of Medical and Dental Sciences, 2015.