International Journal of Anatomy and Research,
Int J Anat Res 2019, Vol 7(1.2):6138-43. ISSN 2321-4287
DOI: https://dx.doi.org/10.16965/ijar.2018.422

Original Research Article

A CLINICO-EMBRYOLOGICAL STUDY OF CONGENITAL OCULAR AND ITS ADNEXAL ANOMALIES IN A TERTIARY CARE HOSPITAL

Sarita Behera [1], Ravindra Kumar Chowdhury *[2], Mamata Sar [3].

[1,3] Department of Anatomy, V.S.S.Institute of Medical Science & Research, Burla, Sambalpur, Odisha, India.

*[2] Ophthalmology, V.S.S.Institute of Medical Science & Research, Burla, Sambalpur, Odisha, India.

ABSTRACT

Aim: This study aims at studying different congenital ocular and its adnexal anomalies over a period of two years in a tertiary care hospital and their association with embryological development.

Materials and Methods: 75 eyes of 60 patients having congenital ocular anomalies as diagnosed by ophthalmologists from June 2015 to June 2017 were included in the study. The demographic profile, perinatal history, associated systemic diseases were studied. The embryological development of all the anomalies has been discussed in details.

Results: Out of 60 patients, 35 (58.3%) were male and 25 (41.7%) were female. Bilateral involvement was seen 'in 14(23.3%). Nasolacrimal duct anomalies were found to be the most common (33.3%) followed by congenital cataract(29.3%),coloboma of uveal tract(20.0%), microphthalmous (4.0%), anophthalmous (4.0%), persistent pupillary membrane (2.7%), congenital glaucoma(1.3%), congenital ptosis(1.3%), Heterochromia iridis (1.3%), coloboma of lids (2.7%) of total eyes. History of consanguinity was present in 13.13% and a history of maternal infection during the antenatal period was found in 8.3%. Systemic involvement was seen in 6.06%.

Conclusion: Proper knowledge of the developmental pathogenesis of congenital ocular anomalies is highly important for correct diagnosis and early intervention. Preventive measures can be applied if history is taken properly during evaluation of the patients.

KEY WORDS: Congenital ocular anomaly, Congenital cataract, Nasolacrimal duct anomaly.

Address for Correspondence: Dr Ravindra Kumar Chowdhury, Assistant professor,Department of Ophthalmology, V.S.S.Institute of Medical Science & Research, Burla, Sambalpur, Odisha, India.
E-Mail: ravindrachowdhury@gmail.com

Access this Article online	Journal Information
Quick Response code	International Journal of Anatomy and Research
	ICV for 2016 90.30
	ISSN (E) 2321-4287 | ISSN (P) 2321-8967
	https://www.ijmhr.org/ijar.htm
	DOI-Prefix: https://dx.doi.org/10.16965/ijar
DOI: 10.16965/ijar.2018.422	

Article Information	
Received: 11 Nov 2018	Accepted: 03 Jan 2019
Peer Review: 12 Nov 2018	Published (O): 05 Feb 2019
Revised: None	Published (P): 05 Feb 2019

INTRODUCTION

The human eye develops from a complex inter-play between three germ layers i.e surface ectoderm, neuroectoderm and mesoderm The neural tube which differentiates into optic vesicles at cranial part results in the development of the neuronal retina, retinal pigment epithelium, epithelial layers of iris & ciliary body and iris muscles [1].

The first evidence of primitive eye formation is seen at the 3rd week of gestation. The surface ectoderm invaginates into the optic vesicle to form the optic cup. The thickened surface ectoderm at optic vesicle results in the formation of lens placode at 27th day of gestation. The lens placode is the source of lens development. In the meantime, mesoderm differentiates to form the different blood vessel. 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 development process leads to the different congenital ocular anomaly. The major insult which results in congenital ocular anomaly are intrauterine infections, drugs, consanguinity of marriage and maternal metabolic disturbance like the folic acid deficiency, diabetes, cretinism, and alcoholism. The congenital ocular anomaly is one of the important cause of childhood blindness.16.7% of total childhood blindness are caused by major structural childhood blindness like anophthalmos, microphthalmos and coloboma globally [2]. The prevalence of congenital eye malformations by Kainbo et al in Zaire and Bermajo et al in Spain are 2.2% and 3.68/10,000 newborns respectively [3,4]. El-Gilary et al in their study at Egypt found that congenital causes accounted for the half of the cases of the 113 blind people that were studied [5]. As the treatment of congenital ocular anomaly is very discouraging, the blindness due to this cause has not given importance in most of the studies. However, we can prevent some of the congenital ocular anomalies by simple awareness among the community. In this study, we have tried to discuss the clinico-embryological association of all the congenital anomalies of eye and its adnexa diagnosed in our institute over a study period of two years.

MATERIALS AND METHODS

This study is a prospective & non-comparative study done in a tertiary care multispecialty hospital of India. It was conducted during the period from June 2015 to June 2017. All the blind schools in the nearby area were also visited to include the cases. The patients were diagnosed clinically by ophthalmologists. If necessary the examination was done under general anesthesia. Supporting investigations were done to confirm some of the diagnosis.75 eyes of 60 patients having congenital ocular anomalies were included in our study. Children more than 14 years were excluded from our study.The demographic profile, prenatal history, and associated systemic diseases were studied in all the patients. A thorough discussion has been done on the developmental basis of all the anomalies.

OBSERVATIONS

Out of 60 patients, 35(58.3%) were male and 25(41.7%) were female. Bilateral involvement was seen in 14(23.3%).Nasolacrimal duct anomalies were found to be the most common (33.3%) followed by congenital cataract(29.3%), coloboma of uveal tract(20.0%)[Fig-1], microphthalmos(4.0%) [Fig-2], anophthalmos(4.0%) [Fig-2],persistent pupillary membrane (2.7%), congenital glaucoma(1.3%)[Fig3], congenital ptosis(1.3%)[Fig-4],Heterochromia iridis(1.3%),coloboma of lids(2.7%)[Fig-5] of total eyes. History of consanguinity was present in 13.13% and a history of maternal infection during the antenatal period was found in 8.3%. Systemic involvement was seen in 6.06%.

Fig. 1: Congenital coloboma of Iris.

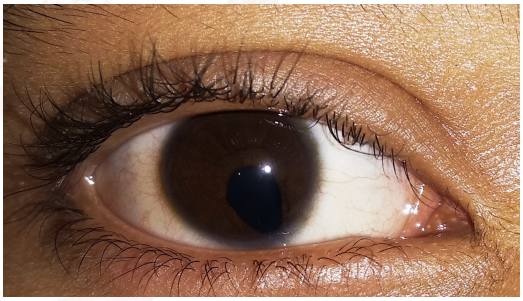


Fig. 2: Micropthalmos of right eye and Anopthalmos of left eye.

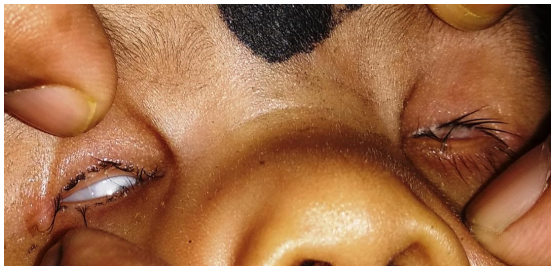


Fig. 3: Congenital glaucoma.

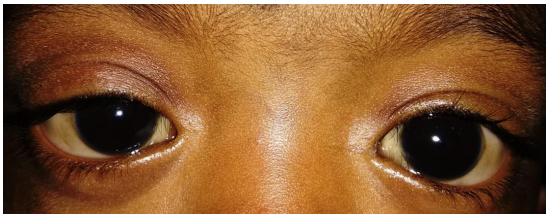


Fig. 4: Congenital Ptosis.

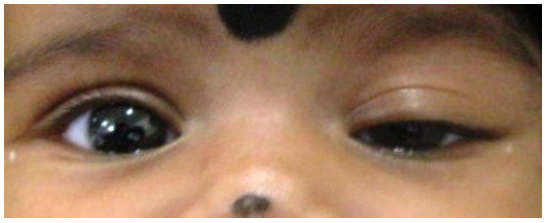
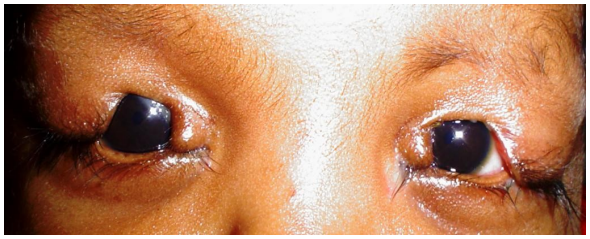


Fig. 5: Coloboma of Lids.



The observations were tabulated in the following tables

Table 1: Type of Ocular anomaly.

Type of anomaly	No of eyes	No of patients	Percentage
Nasolacrimal duct anomalies	25	20	33.30%
Congenital cataract	22	19	29.30%
Coloboma of uveal tract	15	13	20%
Microphthalmos	3	2	4%
Anophthalmos	3	2	4%
Persistent pupillary membrane	2	1	2.70%
Congenital glaucoma	1	1	1.30%
Congenital Ptosis	1	1	1.30%
Heterochromia iridis	1	1	1.30%
Congenital coloboma of lid	2	1	2.70%
Total	75	60	100%

Table 2: Sex Distribution.

Sex	No. Of patients	%
Male	35	58.3
Female	25	41.7
Total	60	100

Table 3: Presence of Consanguinity.

Total no. Of patients	Patients with a history of consanguinity	%
60	8	13.13

Table 4: Maternal infection.

Total no. of patients	Patients with a history of maternal infection
60	5

Table 5: Systemic involvement.

Total no. Of patients	Patients with systemic involvement	%
60	4	6.06

DISCUSSION

We shall discuss the anomalies in relation to their embryological development in details starting from the most frequent one.

Nasolacrimal Duct Anomalies: It may exist at any level from the punctum to the opening of the nasolacrimal duct in the inferior meatus. The ectasia of the lacrimal passage occurs due to the failure of fusion of the nasal and maxillary processes. The deformities of the canaliculi and puncta are consequent upon abnormal buddings from the upper end of the ectodermal rod. A failure of canalization of the lacrimal passage is more frequent which is also found in our study. More often mechanical pressure from the amniotic bands is incriminated in the etiology. The atresia of the nasolacrimal duct may be familial and is inherited as an autosomal dominant character [6].

We found nasolacrimal duct anomaly as the most frequent anomaly accounting for 33.3% of our cases. However, Bodunde et al in their study found this anomaly in 10.5% of cases [2].

Congenital Cataract: This is usually present at birth or develops just after birth. The hereditary predisposition, rubella syndrome, toxic agents, and prematurity are important causes responsible for this anomaly. Vitamin deficiency, ionizing radiations, endocrine dysfunction, inborn errors of metabolism are also included in its etiology. The most critical period for congenital cataract development lies between 5^{th} -8^{th} weeks when cellular activity is maximal. Interference during this period results in abnormal primary lens fibers leading to the development of central cataract. The involvement of secondary lens fibers during 8^{th}-16^{th} weeks produces zonular cataract [6].

29.3% of cases were found to be associated with congenital cataract in our study in contrast to the study by Chuka-Okosa et al in 2005 & Bodunde et al in 2006 where they found it to be 42.6% &47.6 % respectively [2,7].

Coloboma of the uveal tract: This includes coloboma of the iris, ciliary body, and choroid. The Coloboma of the uveal tract can be either typical or atypical. The typical coloboma often affects both iris and ciliary body. Generally, this anomaly occurs due to non-closure of the embryonic fissure. The atypical coloboma of the ciliary body or iris occurs due to the persistence of the embryonic vascular system of the lens preventing the forward growth of the neuroectoderm [6].

And this is not related to non- closure of the embryonic /optic fissure. The typical coloboma classically located in the inferior sector.

The incidence of coloboma depends on the type

of population studied. The worldwide incidence of ocular coloboma ranges from 3.2% to 11.2% [8]. However the incidence of coloboma is quite high in the present study(20%).

Microphthalmos: When one or both eyes are markedly smaller than normal, the condition is called as microphthalmos. Pure microphthalmos (Nanophthalmos) is due to arrest in development of the eyeball in all dimensions after the closure of optic fissure. It is usually transmitted as autosomal recessive and occasionally as autosomal dominant when associated with colobomatous defects [6].

Anophthalmos: This is an uncommon condition where the eyeball is totally absent. This is of 3 types-

1. Primary- due to suppression of the optic analage after the formation of the rudiment of forebrain.

2. Secondary-due to gross suppression and malformation of the anterior part of neural tube.

3. Degenerative or consecutive-due to degeneration of optic vesicle after its formation.

The birth prevalence of microphthalmos and anophthalmos ranges from 0.6 to 4.2 per 100000 birth and 2 to 17 per 100000 birth in different literatures from western countries [9-16].

In most of this studies the prevalence of anophthalmos is higher than microphthalmos but in our study both microphthalmos and anophthalmos found in 4% of our total cases. Ethnic minorities (in particular children of Pakistani and Bangladeshi ethnicity) appear to have a higher incidence of anophthalmos and microphthalmos than do white children. Our finding is consistent with data from other epidemiologic studies undertaken in South Asian countries, which are estimated to have the highest prevalence of severe visual loss from congenital ocular anomalies [17,18].

Persistent pupillary membrane: This condition occurs due to the persistence of a part of the anterior vascular sheath of the lens It may be attached to the anterior surface of the iris or posterior surface of the cornea. Heredity has little influence on the incidence of this anomaly. Remnants of pupillary membrane are very common occurring in 95% of normal newborn babies.[19] However most of the time this remnants are very small and needs no treatment, These remnants usually disappear or shrink by first years of life [20].

In the present study, we found association of persistent pupillary membrane in 2.7% of the anomalies.

Congenital glaucoma: This may be of two types:

1. Developmental

2. Secondary.

1. Developmental glaucoma can be of either simple or complicated. The simple type is not associated with any other congenital ocular anomalies. The trabecular blockage occurs due to the failure of absorption of embryonic mesodermal tissue and improper cleavage between corneal trabeculum and iris. The complicated type is associated with other ocular anomalies.

2. Secondary type is caused by rubella and other intrauterine infections. Most cases of congenital glaucoma are sporadic. However, if it shows a hereditary tendency, occur as an autosomal recessive trait [6].

Primary Congenital Glaucoma(PCG) is a rare eye disorder which accounts for 0.01–0.04% of total blindness [21]. The incidence of PCG is different in different populations. The incidence is 1 in 3300 in Andhra Pradesh, India. In Andhra Pradesh, the disease accounts for 4.2% of all childhood blindness [22]. The incidence of PCG is increased when “founder effect” or a high rate of consanguinity are found in a population. In our study, we found 1.3% of our cases as congenital glaucoma. We found the association of consanguinity with all the congenital ocular abnormalities to be 13.13%.

Congenital Ptosis: Congenital drooping of upper lid is called as congenital ptosis.It is due to lack of proper differentiation of levator palpebrae superioris(LPS). In this patients the LPS muscle may be underdeveloped and completely absent.It is often inherited as autosomal dominant trait [6].

The incidence of congenital ptosis worldwide has not been reported officially. However in a major study on prevalence and inheritance on genetic eye diseses,it has been reported to be 0.18% in China [23].

In our study we found only one case (1.3%) of congenital ptosis during the study period of two years.

Heterochromia iridis: This is a condition wherein two irides have different colors or segment of an iris has a different color from the remainder. The etiology is not clear. Autonomic or metabolic disturbances, intrauterine toxins or infections, may be implicated. Defective closure of the neural tube and deformities of the cervical spine is associated with this anomaly [6].

We found 1.4% of cases as heterochromia iridis in our study which is consistent with Waardenburg et al in 1951 [22].

Coloboma of Lid: This is relatively common and may vary from small notch at lid margin to complete absence of lid. It is commonly inherited as autosomal dominant trait.Though the etiology is disputed,the defect may be caused either by the pressure from amniotic band or by a localized failure of union of lid fold.[6]We found two case of coloboma of eye lids in one patient which is bilateral in nature and was not associated with any other congenital anomaly.

CONCLUSION

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.

Conflicts of Interests: **None**

REFERENCES

[1]. Tamm ER, Ohlmann A.Development of the human eye.Ophthalmologe. 2012 Sep;109(9):911-28.
[2]. Bodunde OT, Ajibode HA.Congenital eye diseases at Olabisi Onabanjo University Teaching Hospital, Sagamu, Nigeria.Niger J Med. 2006 Jul-Sep;15(3):291-4.
[3]. Kainbo WA, Kaimbo D, MiWilambiwe WA. Congenita Malformations of one eyeball and its appendices in Zaire. Bull Soc.Belge Ophthalmol 1994; 254:165 - 70.
[4]. Bermejo E, Martinez-friab ML . Cong. Eye malformations: Clinical epidemiological analysis of 1,124,654 consecutive births in Spain. Is J Med Genetics 1998; 75(5): 497 - 504.
[5]. el-Gilary AH, el-fedaway S, Tharwat M. Causes of blindness and needs of the blind in Mansourg Egypt.East Mediterr Health J 2002; 8 (1): 6 - 17.
[6]. HV Nema, VP Sing, Nitin Nema.Anatomy of the Eye and its Adenexa, Third Edition.
[7]. C.M.Chuka-Okosa,N.O.Magulike2 and G.C.Onyekonwu. Congenital eye anomalies in Enugu, South-eastern Nigeria.WAJM Vol-24 No.2.April-June 2005.
[8]. Fujiki K, Nakajima A, Yasuda N, Tanabe U, Kabaswa K.Genetic analysis of microphthalmos.Ophthalmic Paediatr Genet 1982;1:139-49.
[9]. Bermejo E Martinez-Frias ML. Congenital eye malformations: clinical-epidemiological analysis of 1,124,654 consecutive births in Spain. Am J Med Genet. 1998;75(5):497–504.
[10]. Clementi M Tenconi R Bianchi F . Congenital eye malformations: a descriptive epidemiologic study in about one million newborns in Italy. Birth Defects Orig Artic Ser. 1996;30(1):413–424.
[11]. Stoll C Alembik Y Dott B Roth MP. Congenital eye malformations in 212,479 consecutive births. Ann Genet. 1997;40(2):122–128.
[12]. Spagnolo A Bianchi F Calabro A. Anophthalmia and benomyl in Italy: a multicenter study based on 940,615 newborns. Reprod Toxicol. 1994;8(5):397–403.
[13]. Kallen B Tornqvist K The epidemiology of anophthalmia and microphthalmia in Sweden. Eur J Epidemiol. 2005;20(4):345–350.
[14]. International Clearinghouse for Birth Defects Surveillance and Research. Annual Report. Rome, Italy: ICBDSR; 2005.
[15]. Shaw GM Carmichael SL Yang W Harris JA Finnell RH Lammer EJ. Epidemiologic characteristics of anophthalmia and bilateral microphthalmia among 2.5 million births in California, 1989-1997. Am J Med Genet A. 2005;137(1):36–40.
[16]. Hu DN. Prevalence and mode of inheritance of major genetic eye diseases in China. J Med Genet. 1987;24(10):584–588.
[17]. Hornby SJ Gilbert CE Rahi JK. Regional variation in blindness in children due to microphthalmos, anophthalmos, and coloboma. Ophthalmic Epidemiol. 2000;7(2):127–138.
[18]. Shah SP, Taylor AE, Sowden JC, Ragge NK, Russell-Eggitt I, Rahi JS, Gilbert CE; Surveillance of Eye Anomalies (SEA-UK) Special Interest Group. Anophthalmos, microphthalmos, and typical coloboma in the United Kingdom: a prospective study of incidence and risk. Invest Ophthalmol Vis Sci. 2011 Feb 1;52(1):558-64.
[19]. Tanzer DJ, McClatchey SK. Spontaneous hyphema secondary to a prominent pupillary membrane in a neonate. J Pediatr Ophthalmol Strabismus 1997;34:318–320.
[20]. Burton bjl, adams ggwPersistent pupillary membranes.British Journal of Ophthalmology 1998;82:709.
[21]. Mandal AK, Chakrabarti D.Update on congenital glaucoma.Indian J Ophthalmol. 2011 Jan;59 Suppl: S148-57.

[22]. Waardenburg, P.J.: A New Syndrome Combining Developmental Anomalies of the Eyelids, Eyebrows and Nose Root With Pigmentary Defects of the Iris and Head Hair and With Congenital Deafness, Amer J Hum Genet 3:195-253 (Sept) 1951.

[23].Hu DN.Prevalence and mode of inheritance of major genetic eye diseases in China.J Med Genet.1987;24(10):584-588.

How to cite this article:

Sarita Behera, Ravindra Kumar Chowdhury, Mamata Sar. A CLINICO-EMBRYOLOGICAL STUDY OF CONGENITAL OCULAR AND ITS ADNEXAL ANOMALIES IN A TERTIARY CARE HOSPITAL. Int J Anat Res 2019;7(1.2):6138-6143. **DOI:** 10.16965/ijar.2018.422