Prevalence of Congenital Ocular Anomalies in Paediatric Age Group (0 - 14 Years) in a Tertiary Care Hospital at Kancheepuram, South India

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ABSTRACT

BACKGROUND

Congenital ocular anomalies are one of the important causes of childhood blindness. Blindness is a serious public health issue, and also a social and economic problem worldwide. Majority of global blindness is avoidable and curable. Most of global blindness is in developing countries. The objectives of this study were to determine the prevalence of congenital ocular anomalies in the paediatric age group and its associated risk factors in a tertiary care hospital in Kancheepuram.

METHODS

This cross-sectional study was conducted among children of 0 – 14 years age group attending the ophthalmology department of SRM Medical College and Hospital, Kancheepuram from December 2018 to November 2020. Congenital ocular anomalies were detected by detailed ocular examination and the type of congenital ocular anomaly was assessed. Associated risk factors like family history of congenital ocular anomalies, family history of consanguinity and significant antenatal history of the mother of the children were collected.

RESULTS

Out of 9865 cases, we found that 61 cases had congenital ocular anomalies. There was a male preponderance. Majority of the cases were in the age group 0 - 5 years. A positive history of consanguinity was present in 10 % of cases. The most commonly found cases were congenital cataract and congenital dacryocystitis.

CONCLUSIONS

Children with congenital ocular anomalies and functional vision should be given glasses and low vision aid. There is a great need for early screening, detection and treatment by paediatric or trained ophthalmologists in referral hospitals.

KEYWORDS

Congenital Ocular Anomalies, Paediatric Age Group, Prevalence

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DOI: 10.18410/jebmh/2021/175

How to Cite This Article:
Wari SAN, Huda R. Prevalence of congenital ocular anomalies in paediatric age group (0 - 14 years) in a tertiary care hospital at Kancheepuram, South India. J Evid Based Med Healthc 2021;8(14):899-903. DOI: 10.18410/jebmh/2021/175

Submission 04-12-2020,
Peer Review 10-12-2020,
Acceptance 09-02-2021,
Published 05-04-2021.

BACKGROUND

Congenital anomalies are present at birth, and may be diagnosed later in life. It includes any behavioural, structural, functional and even metabolic disorders which are present at birth. It is caused due to defective development of eye tissue during its intrauterine life. Congenital anomaly of the eye may occur because of genetic effect, environmental factors,[1] various teratogens or chromosomal anomalies in the developing embryo during intrauterine life.

In a study in Italy BY Stoll C et al. chromosomal abnormalities, especially trisomy 13, were reported to be an important factor for clinical anophthalmos. In few epidemiological studies, consanguinity has been accepted as an important cause for congenital anomalies.[2] Intrauterine viral infections, such as cytomegalovirus or rubella can cause significant number of congenital ocular abnormalities.[3]

People have reported cases of anophthalmos in children born to women with severe vitamin A deficiency during pregnancy.[4] Besides maternal infections, genetic factors, drugs taken during pregnancy and radiation also play a role in causing congenital anomalies.[5] Congenital ocular anomalies are one of the important causes of childhood blindness.[6] Congenital cataract is a major cause of visual impairment and blindness in childhood throughout the world.[6] Maternal infection with rubella in the first trimester of pregnancy results in congenital rubella syndrome. It is an important cause of blindness in developing countries. Females of child bearing age are quite susceptible to rubella infections. It is one of the few diseases which can be prevented by rubella immunization program.

Congenital ocular anomalies are one of the important causes of childhood blindness.[7] Many children with congenital ocular anomalies have low vision. Visual rehabilitation and low vision aids can actually be very useful to these children. The use of these low vision devices should be discussed with child and parents in low vision clinic. Therefore, there is growing need for early screening, detection, treatment by pediatric or trained ophthalmologist and rehabilitation of these cases.[8] With the advent of newer technologies, telemedicine should be encouraged and utilised for remote areas. It can prove in future to be a good screening tool for early detection of these congenital ocular anomalies.

METHODS

This was a cross sectional study conducted in the ophthalmology department of SRM Medical College and Hospital for a period of two years from December 2018 to November 2020 among the children belonging to the paediatric age group from 0 - 14 years presenting to our ophthalmology department. Children with congenital ocular anomalies between ages 0 - 14 years were identified. Patients with history of trauma to eye, retinopathy of prematurity and retinoblastoma were not considered as congenital ocular anomalies. Totally 9865 children in the age group 0 – 14 years attended the Ophthalmology Outpatient Department (OPD) during the study period and all were included in the study.

Study Procedure

After obtaining informed consent from the patient's parents, a detailed history was taken. History includes illness, medication, radiation or infection during pregnancy, family history of congenital anomalies and consanguinity was taken. Detail ocular and systemic examination was done. In ocular examination, visual acuity, refraction, squint evaluation, retinoscopy, tonometry, pupillary reflex, anterior segment examination and fundus examination was done. Examination under anaesthesia was done, where required. Visual acuity was tested with Snellen chart, E chart. In small kids, optokinetic nystagmus, preferential looking test and finger counting techniques were used. Anterior segment was examined using slit lamp biomicroscopy. Fundus was examined using direct and indirect ophthalmoscope.

Statistical Analysis

Data collected was entered in Microsoft (MS) Excel sheet and analysed using Statistical Package for the Social Sciences (SPSS) software version 21. Categorical variables were represented in frequencies and percentages. Pictorial representations (bar charts, pie diagrams, line diagrams and scatter plots) were used as appropriate. Chi-square test was used to determine the significance of the association between two or more proportions. Fisher's exact test was used when more than 20 % of the cell values had expected cell value less than 5. P-values less than 0.05 were considered statistically significant.

RESULTS

Totally, 9865 children in the age group 0 – 14 years attended the Ophthalmology OPD during the study period and all were included in the study. Majority of the patients were from rural area (77 %) and 23 % of patients were from urban area. Out of the 9865 children, 61 children were diagnosed to have congenital ocular anomalies. Out of the 61 congenital anomaly cases, 25 (41 %) were in 0 - 5 years of age, followed by 19 (31 %) cases in 5 - 10 years of age group and 17 (28 %) in 10 - 14 years. 40 (65.6 %) children were males and 21 (34.4 %) were females. Male and female distribution were almost equal in all age groups. (Table 1)

Among the risk factors, 10 % of the cases had a positive history of consanguineous marriage, out of which half of the cases had second degree of consanguinity and 20 % had family history of ocular congenital anomalies. Regarding the antenatal history of the mothers of the affected children, 28 % cases had a history of bleeding per vagina during gestation of mother of the affected individual and 21 % had a history of taking some medication during their gestational period of affected child. (Table 2)

Regarding the distribution of type of congenital ocular anomalies, 31 % had congenital cataract followed by 21 % of whole globe anomalies and 21 % of congenital

nasolacrimal duct obstruction (NLDO), 11.5 % of coloboma of uveal tract and 6 % each of congenital corneal involvement & congenital glaucoma. Among the congenital cataract cases, 50 % had nuclear cataract and 40 % lamellar variety. Among the whole globe anomalies, 46 % cases had microphthalmos, 30 % cases had microphthalmos with cyst and 24 % cases had anophthalmos. Among 2 cases of corneal involvement, there were 2 cases of microcornea, 1 case of sclera cornea and 1 case had corneal opacity. In our study, 3 out of 19 cases of congenital cataract, 1 out of 7 cases of uveal coloboma, 1 out of 4 cases of congenital glaucoma and congenital corneal involvement had a positive family history. (Table 3).

Age Group	Gender		Total
	Male	Female	
0 - 5 years	16 (40 %)	9 (42.9 %)	25 (41 %)
5 - 10 years	12 (30 %)	7 (33.3 %)	19 (31.1 %)
10 - 14 years	12 (30 %)	5 (23.8 %)	17 (27.9 %)
Total	**40 (100 %)**	**21 (100 %)**	**61 (100 %)**

Table 1. Age and Sex Wise Distribution of the Congenital Ocular Anomaly Subjects

	Risk Factors	Frequency (N = 61)	(%)
Family history	Yes	12	19.7 %
	No	49	80.3 %
History of consanguinity	Yes	6	9.8 %
	No	55	90.2 %
Degree of consanguinity	First	2	3.3 %
	Second	3	4.9 %
	Third	1	1.6 %
History of bleeding P.V	Yes	17	27.9 %
	No	44	72.1 %
History of medication during pregnancy	Yes	13	21.3 %
	No	48	78.7 %

Table 2. Distribution of the Congenital Ocular Anomaly Subjects Based on Risk Factors

Congenital Ocular Anomalies	Total Cases (N = 61) N (%)	Male (N = 40) N (%)	Female (N = 21) N (%)	Positive Family History (N = 8) N (%)
Congenital cataract	19	12 (63.2 %)	7 (36.8 %)	3 (15.8 %)
Whole globe anomalies	13	9 (69.2 %)	4 (30.8 %)	0 (0 %)
Coloboma of uveal tract	7	5 (71.4 %)	2 (28.6 %)	1 (14.3 %)
Congenital corneal involvement	4	3 (75 %)	1 (25 %)	1 (25 %)
Congenital glaucoma	4	2 (50 %)	2 (50 %)	1 (25 %)
Congenital NLDO	13	8 (61.5 %)	5 (38.5 %)	2 (15.4 %)
Congenital lid abnormalities	1	1 (100 %)	0 (0 %)	0 (0 %)

Table 3. Distribution of Sex and Family History among Subtypes of Congenital Ocular Anomalies

	Risk Factors	Congenital Ocular Anomalies		χ2 Test (P-Value)
		Yes	No	
Age group	0 - 5	25 (41 %)	3131 (31.9 %)	0.281
	5 - 10	19 (31.1 %)	3819 (39 %)	
	10 - 14	17 (27.9 %)	2854 (29.1 %)	
Gender	Boys	40 (65.6 %)	40 (0.4 %)	0.243
	Girls	21 (34.4 %)	21 (0.2 %)	
Family history	Yes	12 (19.7 %)	529 (5.4 %)	< 0.001*
	No	49 (80.3 %)	9275 (94.6 %)	
History of consanguinity	Yes	6 (9.8 %)	346 (3.5 %)	0.008*
	No	55 (90.2 %)	9458 (96.5 %)	
History of bleeding P.V.	Yes	17 (27.9 %)	1267 (12.9 %)	0.001*
	No	44 (72.1 %)	8537 (87.1 %)	
History of Medication During Pregnancy	Yes	13 (21.3 %)	2389 (24.4 %)	0.579
	No	48 (78.7 %)	7415 (75.6 %)	

Table 4. Association of different Risk Factors with Congenital Ocular Anomalies

*indicates statistically significant association at P < 0.05

Regarding the association of risk factors with congenital ocular anomalies, age and sex had no difference in their distribution. Family history, history of consanguinity among their parents and history of bleeding per vagina (PV) in the mother during antenatal period had a significant association with presence of congenital ocular anomalies among the children (Table 4).

DISCUSSION

In our study, the prevalence of congenital ocular anomalies in paediatrics age group was 6.1 / 1000 cases. Almost a similar prevalence of 7.5 per 1000 cases of congenital ocular anomalies, was reported by Stoll et al. in their study.[2] The incidence of congenital anomalies in a study by Singh et al. was reported to be 10.5 per 1000 live birth.[9] Dandona did a study of epidemiology of childhood blindness in Andhra Pradesh and found out that 20 % of blindness was because of congenital ocular anomalies.[6]

In our study, 19.7 % had some positive history either in parents or in siblings. Singh et al. in their study reported that in 14.3 % cases of congenital ocular anomalies had evidence of positive family history.[9] Out of 19 congenital cataract cases in our study, 16 % had positive family history whereas out of 4 congenital glaucoma cases 25 % of patients had a positive family history. Rao G.N et al. found out that 15.3 % of childhood blindness is due to congenital cataract.[10] In our study, we saw that 50 % of congenital cataract was nuclear cataract followed by lamellar variety. The percentage of lamellar cataract was found to be 40 %. Postnatally, the lens became a reservoir for the rubella virus leading to persistent kind of infection due to which the nuclear cataract became a total cataract. In a study at Aravind Eye Hospital, Madurai found that rubella cataract was present in nearly 93 percentage of eyes.[11] They noted that nuclear cataract was the commonest subtype found in their study. According to Givens et al. this incidence was 27 % in their series.[12] There was another Aravind Eye Hospital study, which discussed the aetiology of congenital cataract in 1994.[13] It said that nearly 25 % of it were because of rubella infection in infants below one year of age, all of which were of nuclear subtype. If we recognise early and treat this disease, the quality of life can be improved for these infants.

In our study, we found more number of cases of congenital ocular anomaly in children whose parents had a positive history of consanguineous marriages and this was supported by many research articles.[14,15,16] In an Egyptian study of 2500 cases presenting with genetic disorders, Reham H et al. found that 2.4 % suffered from one or more ocular anomalies. They in their study reported a very high consanguinity as 76.7 % and a positive family history of 35 % of ocular cases. According to their study, congenital cataract was the most common congenital ocular anomaly.

In our study, 17 out of 61 (27.9 %) cases had a history of bleeding PV during pregnancy period, with or without history of fever. In our study, there was 21 % (13 cases out of 61 cases) positive history of taking some form of medication during gestation. Nishimura et al.[17] found out that there might be some positive association between congenital ocular anomalies and genital bleeding or any history of abortion.[17]

We found that the prevalence of globe anomalies was 21 %. Rahi and Allen Foster in their study, had reported a similar prevalence of globe anomalies of 25 %.[18,19] Out of the thirteen cases of whole globe anomalies, we found 46% cases had microphthalmos, 30 % cases had microphthalmos with cyst and 24 % cases had anophthalmos. Roy & Deutch had reported that maternal infection and medication during gestation produces microphthalmos. In a study by Roa G.N et al. reported that due to whole globe anomalies, the percentage of blind child was 29 %.[10]

In our study, out of 61 cases, 7 cases were (11 %) found to have some form of coloboma of uveal tract. Clark E et al. reported 2.4 % of typical coloboma out of 5000 cases of congenital anomalies.[20] According to them, the typical colobomatous defects were the commonest. They found anophthalmos to be rare. They also reported that it was more common in South Asian population than the white population. In a study by Dutta et al. 10.47 % of congenital ocular anomalies were due to uveal tract colobomas.[21]

In our study, 7 % of total cases of ocular congenital anomalies had congenital glaucoma. Age of presentation was three months to ten months. Primary congenital glaucoma had a familial inheritance pattern in about 10 - 40 % of cases.[22] In Andhra Pradesh, study done by Rao GN et al. it contributed to 4.2 % of all childhood blindness. Some studies also suggested that if a high percentage of consanguinity was found, then there was an increased incidence of primary congenital glaucoma in that population. Children with congenital glaucoma usually have severe visual impairment. Glasses should be prescribed after refraction and patching should be done to treat amblyopia. Visual rehabilitation and low vision aid should be used to help these children lead a near normal life.[23] In our study, 21 % of cases had congenital nasolacrimal duct obstruction. Various studies report the prevalence of congenital NLDO varies from 5 % to 20 % in early childhood. Mac Ewen et al. reported that epiphora due to congenital NLDO was around 20 % in the first year of life.[24]

We believe that although there were different causes of blindness, that blindness caused by congenital anomalies should be addressed early and treated promptly. We want to mention that consanguineous marriage is one of the contributing factor. Such marriages are common in India, especially in the area where we conducted our study. So creating a general awareness and considering premarital counselling as an important tool in our attempt to prevent congenital ocular anomalies.

CONCLUSIONS

We want to emphasize over the fact that those congenital anomalies which can be treated, like congenital cataract, congenital glaucoma and congenital lid anomalies should be diagnosed early and managed at the appropriate time by trained pediatric ophthalmologist or an experienced ophthalmologist. We should implement programs which gives utmost importance to maternal nutritional status and maternal health during pregnancy. Mobile ophthalmic units or telemedicine should be brought into use for screening and early detection of cases. More ophthalmologists should be trained in paediatric eye disease. This will help in avoiding blindness in children. Premarital counselling on consanguinity should be encouraged. Since congenital ocular anomalies may lead to major visual disability. We would recommend that children with ocular anomalies and functional vision should be given glasses and low vision aid services. We should encourage more research in this field which will assist to further help the preservation and restoration of vision in children suffering from congenital ocular anomalies.

Data sharing statement provided by the authors is available with the full text of this article at jebmh.com.
Financial or other competing interests: None.
Disclosure forms provided by the authors are available with the full text of this article at jebmh.com.

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