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CONGENITAL CAUSES OF VISUAL IMPAIRMENT IN PERSONS SEEKING VISUAL DISABILITY CERTIFICATE – A RETROSPECTIVE STUDY

Ophthalomology

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ABSTRACT

Background: Despite being rare,Congenital eye disorders, in isolation or as in combination are important causes of childhood blindness. This retrospective study aims at documenting different congenital causes of visual impairment in Department of Ophthalmology, Government Coimbatore medical college,Coimbatore. **Methodology:** This retrospective study involves review of case notes and medical records in the disability register of patients who attended the tertiary care centre for disability certificate from January 2022 – December 2022. **Results:** The leading congenital cause of visual impairment according to our study was retinitis pigmentosa which accounts for 35% ,followed by macular dystrophies (14%),microphthalmia(9%), congenital corneal opacity(8%), idiopathic nystagmus (6%),congenital ptosis (5%), microcornea (5%), congenital cataract(4%),uveal coloboma(4%), retinoblastoma (2%),anophthalmos(2%),optic nerve hypoplasia(2%) and congenital strabismus (2%).Other miscellaneous causes were lens coloboma, congenital glaucoma each contributes 1%. **Conclusion:** Congenital eye disorders are important causes of visual morbidity and mortality in children.This study mainly emphasize the importance of screening programmes for early detection and treatment of congenital causes of avoidable blindness and regular maintainence of blindness register and education about consanguinety and premarital counselling.

KEYWORDS

Congenital eye disorders; Childhood blindness; Screening; Genetic counselling

INTRODUCTION :

Despite being rare,Congenital eye disorders, in isolation or in combination, or as part of a syndrome are important causes of childhood blindness. The eyes are formed by a delicate and complex process in human embryo. Any disturbance in this process can lead to congenital eye malformations.

Though they are relatively rare, they account for approximately five per 10,000 live births.Genetic factors play a major role in many kinds of eye disease, including those diseases that are the leading cause of blindness among infants, children and adults. Among them ,the commonest cause of blindness is inherited eye diseases such as congenital glaucoma, congenital cataracts, optic atrophy and retinal degeneration.

Vision loss in early childhood will adversely affect the development, mobility, education, and social and employment opportunities.The prevalence of blindness in children accounts for approximately 0.3/1000 (of total population) in affluent countries to 1.5/1000 in the poorest. But it is difficult to obtain reliable population-based data on the causes of blindness in children particularly in developing countries. This retrospective study was carried out in Department of Ophthalmology, Coimbatore medical College and hospital aimed at documenting the causes of congenital eye diseases by including the patients who approach for visual impairment certificate.

Aim of the study :

This study mainly aims at documenting the different congenital causes of visual impairment in persons seeking visual disability certificate in Department of Ophthalmology, Government Coimbatore medical College, Coimbatore from January 2022-December 2022.

METHODS:

This is a retrospective study which involves reviewing the case notes and medical records in the disability register (secondary data analysis) of patients who attended the tertiary care centre for disability certificate from January 2022 – December 2022. The study included patients who had been diagnosed by a medical board as having a visual impairment.

In the out-patient department, patients who were referred from the medical board were examined. The data collected includes age, gender, the underlying cause of the condition, and the reason for getting a certificate of visual impairment Complete ophthalmological examination including visual acuity, tonometry,fields assessment by bjerrum screen , anterior segment examination by torch light and slit lamp biomicroscopy and posterior segment examination by fundus examination and OCT and B scan wherever required. Certificate issued based on the final diagnosis and degree of visual impairment and field restriction.

OBSERVATION AND RESULTS :

The total number of persons in our study were 100.

Gender distribution:

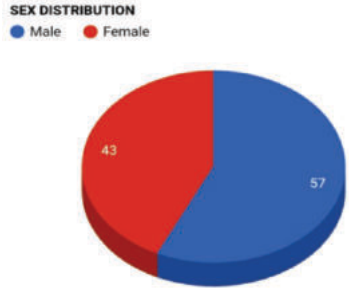


Figure 1- Sex distribution

Among 100 cases, 53 were males and 47 were females with a male:female ratio of 1.3:1.

Age distribution :

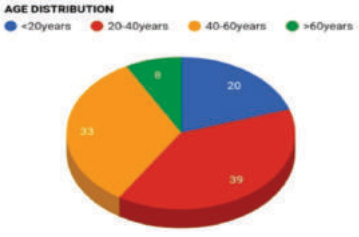


Figure 2- Age distribution

Wide range of age group were taken from below 20 years to above 70 years.Of which maximum number of cases were between 20-40 years of age(39%)followed by 40-60 years(33%) , below 20 years(20%) and above 60years of age(8%)

Amount of visual disability:

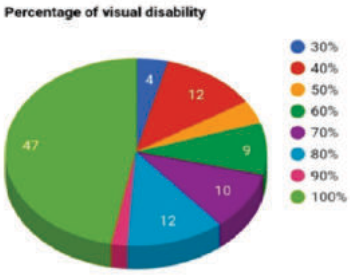
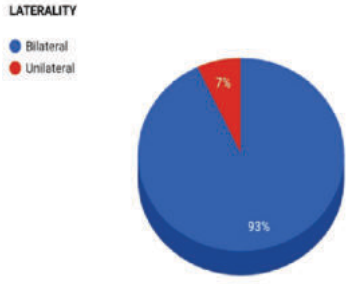


Figure 3- Percentage of visual disability
Visual ability of 100% noted in 47persons which was the maximum in our study followed by 40% in 12 cases,80% in 12 cases,70% in 10 cases,60% in 9 cases,30% in 4 cases,50% in 4cases and 90% in 2 cases.



Laterality:
Figure 4- Percentage of laterality
About 93% cases were bilateral and 7% were unilateral.

Congenital causes of visual impairment :
Table 1- Different congenital causes of visual impairment

CAUSES	PERCENTAGE
1.ANOPHTHALMOS	2
2.MICROPHTHALMOS	9
DISEASES OF CORNEA	
1.MICROCORNEA	5
2.CONGENITAL CORNEAL OPACITY	8
DISEASES OF LENS	
1.CONGENITAL CATARACT	4
2.LENS COLOBOMA	1
DISORDERS OF UVEA	
1.COLOBOMA	4
DISEASES OF RETINA	
1.RETINITIS PIGMENTOSA	35
2.OTHER RETINAL DYSTROPIES	14
3.RETINOBLASTOMA	2
DISEASES OF OPTIC NERVE	
1.OPTIC NERVE HYPOPLASIA	2
2.CONGENITAL GLAUCOMA	1
OTHER CAUSES	
1.IDIOPATHIC NYSTAGMUS	6
2.CONGENITAL PTOSIS	5
3.CONGENITAL STRABISMUS	2

The leading congenital cause of visual impairment according to our study was retinitis pigmentosa which accounts for 35% ,second leading cause was Macular dystrophies (14%) followed by microphthalmia(9%), congenital corneal opacity(8%), idiopathic nystagmus (6%), Congenital ptosis (5%), microcornea (5%), Congenital cataract(4%),uveal coloboma(4%), Retinoblastoma(2%), anophthalmos(2%),optic nerve hypoplasia(2%) and Congenital strabismus (2%).Other miscellaneous causes were lens coloboma, congenital glaucoma each of which accounts for 1%.

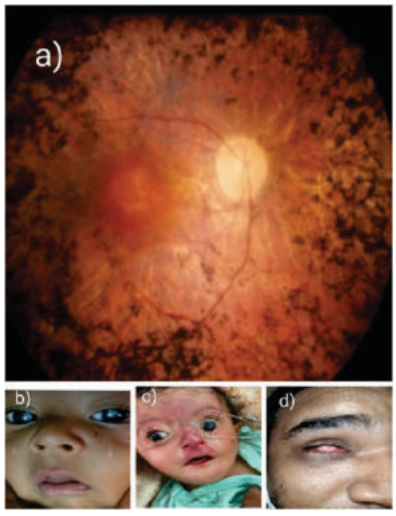


Figure5- a)Retinitis pigmentosa; b)Congenital cataract;c) Congenital glaucoma ;d)Anophthalmos

DISCUSSION:
Though it is rare, Childhood blindness prevails as a significant global health problem. The main priority of 'VISION 2020—The Right to Sight' programme -control of childhood blindness which is a global initiative that aims to eliminate avoidable blindness by the year 2020.So as to adapt some control programs to prevent childhood blindness in our country it is necessary to emphasize the causes of visual impairment in childhood of which congenital causes plays a major role.

In our study, the commonest congenital disorder causing blindness is retinitis pigmentosa (39%) and other retinal disorders like macular dystrophies accounts for 14% followed by retinoblastoma (2%).

S. Krishnaiah et al. (2012) concluded that retinal blindness was the second most common cause of severe visual impairment/blindness (SVI/BL) (18.9%) identified in this population.The major cause was retinal dystrophy (17.1%), the finding approximately close to the estimate reported previously from the same population by Hornby SJ etal (2008)(1)

Congenital Eye Abnormalities By Simeon A. Boyadjiev Boyd , MD, University of California, DavisReviewed/Revised Sep 2022 found that retinal causes were responsible for SVI/BL in 282 (24.9%) children. The majority were retinal dystrophies (179, 15.8%) and albinism (15, 1.3%). Retinopathy of prematurity was diagnosed in 22 cases (1.9%). Other retinal disorders, including retinal detachment and retinoblastoma, accounted for the remainder (5.9%) (2).

In our study , microphthalmia accounts for 9%, microcornea 5%,anophthalmos 2% and lens and iris coloboma 2% each. Thus these are the disorders which mostly favours autosomal recessive inheritance thus signifying the importance of effects of consanguinous marriages.

Krishnaiah et al in a study (2012) concluded that 23 % of children had SVI/BL definitely attributable to genetic disease. The mode of inheritance in the majority of these children was autosomal recessive. Cataract and optic nerve disease were also important and accounted for approximately half of the autosomally dominant inherited diseases. Consanguineous marriages, which increase the likelihood of autosomal recessive diseases, are common in certain communities in India. First-cousin marriages occur throughout the country and maternal uncle/niece marriages are common in South India. Congenital ocular anomalies(anophthalmos, microphthalmos, microcornea, coloboma and aniridia) accounted for 22% of children with SVI/BL. These conditions appear to be relatively more important in India than in other developing countries(1).

The finding of higher rates of microphthalmos and anophthalmos has been found to be a common finding in many studies conducted earlier on blind children in India,congenital ocular anomalies were responsible for major causes of blindness in 41.3% and 27.4% of children in Maharashtra and Delhi, respectively.

In our study , congenital cataract accounts for 4% and this comes under the main cause of preventable blindness before the child starts developing amblyopia so here early diagnosis and treatment needs a prompt role. Other preventable causes were congenital corneal opacity (8%), congenital ptosis(5%),congenital strabismus(2%) and congenital glaucoma(1%).Other miscellaneous causes were idiopathic nystagmus(6%), uveal coloboma(4%) and optic nerve hypoplasia (2%).

Rogers NK etal in (2013) found congenital cataract is the leading cause of surgically correctable blindness in most developing countries (7).

In a study in China (2010),it was found that 19% of children had visual impairment from disorders of the lens. Of these 65% were aphakic and/or amblyopic probably as a result of late surgery or inadequate refractive correction with 32% having unoperated cataracts(3)

CONCLUSION :

Congenital eye disorders are important causes of visual morbidity and mortality in children.But large proportion of the childhood blindness is avoidable.Hence early detection of the causes of childhood blindness allow for early action against avoidable blindness .This study mainly focuses on the need for institution of screening programmes at maternity centres before the babies are discharged for early detection of congenital causes of avoidable blindness and treatment for the same.It also emphasize the need for maintaining blindness register and education about consanguinous marriage and premarital counselling to avoid congenital ophthalmic disorders.

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Conflict of interest - None.

Ethical approval - approved by Institutional ethics committee.

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