INCIDENCE OF OCULAR CONGENITAL ANOMALIES IN A NIGERIAN TEACHING HOSPITAL

By

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SUMMARY

Objective: The aim of this study was to report the pattern and incidence of congenital eye defects among patients seen in a semi-urban academic tertiary referral institution.

Methodology: We reviewed all consecutive new patients with congenital abnormality seen between January 1998 and December 2003 at Obafemi Awolowo University Teaching Hospital, Ile-Ife, Nigeria Data was analyzed using simple descriptive statistics on SPSS statistical package version 10.

Results: A total of 189 patients with congenital defects were seen during the study period, of which 31(16.4%) had congenital eye defects {male 16(51.6%), female 15 (48.4%)} with a male to female ratio of 1:1. Their ages at presentation ranged from 1 day to 23 years with mean age+/- SD at 1.6 years +/-0.5 years. Congenital eye defects seen were congenital cataracts 6(19.4%), microphthalmia 5(16.1%), nasolacrimal duct obstruction 4(12.9%), congenital glaucoma 3(9.7%), lid coloboma 3(9.7%), congenital ptosis 2(6.5%), pseudo-proptosis 2(6.5%), and lid haemangioma 2(6.5%). Prevalence of blindness was 9.8%.

Conclusion: Congenital eye defects constitute a significant cause of morbidity (16.4%) and blindness (1.6%) among cases of congenital defects seen in the teaching hospital.

Key Words: *Congenital, Eye defects*

INTRODUCTION

Congenital defects are abnormalities of structure or function present at birth. They may be caused by genetic or environmental factors, or a combination of both. The causes of many defects remain unknown. Developmental defects may be lethal, semi-lethal, or compatible with life, causing very little effect or only aesthetic effect.

Congenital eye defects are among the most common birth defects and are the leading cause of childhood visual impairment and blindness.[1-3] Congenital and developmental anomalies of the

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eye could be as a result of defects in formation of optic vesicle (anophthalmos, microphthalmos, synophthalmos), defective invagination of optic vesicle (non attachment of retina, coloboma), neuroectodermal dysplasia (retinal dysplasia, hereditary retinal atrophies, retinochoroidal hypoplasia), ectodermal dysplasia (cataracts, spherophakia, lenticonus, microphakia, coloboma), corneal dystrophies, choristomas (dermoid, dermolipoma), and eyelid abnormalities (ankyloblepharon, micropalpebra, eyelid coloboma, entropion, ectropion, trichiasis).

Aetiologies of congenital ocular abnormalities include genetic, physical, chemical, infectious agents, irradiation, nutritional or metabolic problems[4-9].

There had been reports of congenital eye defects in animals and series of reports on some rare types of congenital eye defects especially in association with other lesions[1-5]. Two cases of Waadenburg's syndrome associated with heterochromia iridis with retinal pigmentary changes and blue iris were reported earlier in Nigeria[6]. Also a few numbers of congenital cataracts were reported in Ibadan during a review of all cataract cases seen over a period of time[7]. However there is paucity of literature on the epidemiology of congenital eye defects in any Nigerian tertiary hospital.

We decided to report the pattern and incidence of congenital eye defects seen in a tertiary health institution in Nigeria with a view to planning an eye care programme for affected patients in the health institution.

PATIENTS AND METHODS

We reviewed all consecutive new patients with a diagnosis of congenital abnormality seen at the neurosurgical unit, postnatal and paediatrics' clinics, paediatric surgical unit, out patients' department and orthopaedics clinics of Obafemi Awolowo University Teaching Hospital, Ile-Ife, Nigeria between January 1998 and December 2003. The objective was to quantify the proportion of children who have congenital ocular defects among those with congenital defects and to evaluate the pattern of other associated defects.

Information and clinical evaluation was documented on all cases of congenital defects seen by the neurosurgeon, otolaryngologist and paediatric surgeons, paediatricians and orthopaedic surgeons. Those who had associated ocular defects were referred to the ophthalmologist were further studied. Informed consent was obtained from patients, parents, and other accompanying adult relatives. Institutional Ethical Committee approval was obtained before the study was embarked upon.

Data obtained included patients' demographics, presenting problems, diagnosis, source of referral, age of both parents, history of exposure to teratogens, associated facial or systemic defects and outcome of management with regards to structural, visual and functional improvement were evaluated. All the patients were thoroughly examined, investigated and referred to the paediatricians for further evaluation.

Clinical diagnoses were made based on the congenital defects detected, and in some others, investigations like radiographs, ocular ultrasound, echocardiogram, blood studies (PCV, ESR, WBC, and Genotype), examination under anaesthesia, excisional biopsy and contrast studies were utilized. Joint surgical management was employed by the ophthalmologists and other colleagues where necessary.

Patients whose visual acuity is less than 3/60 or visual field of less than 10^0 from fixation in the better eye were

classified as blind while those who have NPL vision in one eye had uniocular blindness (WHO classification of blindness and visual impairment, 1999).

The data was analyzed using descriptive statistics on SPSS statistical package version 10 using both descriptive and inferential methods.

RESULTS

A total of 189 patients with congenital defects were seen during the study period, of these 31(16.4%) had congenital eye defects.

Sixteen of them were males (51.6%) while 15 (48.4%) were females with a male to female ratio of 1:1. Their ages ranged from 1 day to 23 years with mean age +/- SD at 1.6 +/-0.5 years.

The distribution of congenital ocular defects across gender is shown in table 1.There was no statistically significant difference in sex of the study population (P=0.108).

Common congenital eye defects found were congenital cataracts 6(19.4%), micro-ophthalmia 5(16.1%), nasolacrimal duct obstruction 4(12.9%), congenital glaucoma 3(9.7%), lid coloboma 3(9.7%), congenital ptosis 2(6.5%), pseudo-proptosis 2(6.5%), and lid haemangioma 2(6.5). Others were limbal dermoid (Goldenhar's syndrome), entropion, squint, anophthalmos representing 1(3.1%) each, table 2.

Other associated congenital defects encountered were meningoencephalocoele 4(44.4%), congenital rubella syndrome 2(22.2%), cleft lips 2(22.2%), auriculovertebral dysplasia 1(11.1%) and Aperts' syndrome consisting of craniosynostosis, syndactyl of the toes and mental retardation 2(22.2%), table 3.

Five patients presented with bilateral micro-ophthalmia, two (40%) were blind in both eyes while another two (40%) had uniocular blindness.

A 3months old boy who had bilateral micro-ophthalmia with bilateral congenital cataract was irreversibly blind.

Blindness prevalence rate among patients with congenital eye defects was 9.8% and 1.6%among all cases of congenital defects recorded during the study period.

Five out of six patients with congenital cataract (83.3%); and all of those with

nasolacrimal duct obstruction, coloboma, congenital ptosis and limbal dermoid were successfully treated. Two out of the three patients with congenital glaucoma had trabeculectomy and only one of these was followed up for up to twelve months. The others were lost to follow up. No treatment was offered to those with anophthalmos, microphthalmos and proptosis.

Only 2 patients (6.7%) had a positive history of likely aetiologic predisposition to the teratogenic effect of X-irradiation in either of their parents.

The mean age+/- SD of the parents was 31+/- 2.3 for mothers and 36 +/- 1.8 for fathers. There was no statistical significance association between the parents age and presence of congenital ocular defects (P=0.082).

DISCUSSION

The mature eye is a complex organ that develops through highly organized process during embryogenesis. Alteration in genetic programming can lead to severe disorders that become apparent at birth or shortly afterwards, for example, half of the cases of blindness in children was said to have a genetic cause[4].

There have been reports on different types of congenital eye defects in literature[2, 3]. Most of these also reported outcomes of management of the cases presented[8-14]. Congenital

malformation of the eye can cause blindness in children. They occur throughout the world and in most cases the aetiology is unknown

It has been reported that up to 50% of cases referred to the genetic services have ophthalmologic abnormalities[5, 8]. Conditions including chromosomal abnormalities, metabolic disorders, Mendelian syndromes and environmental factors are associated with ocular abnormalities. Anatomically, positional and adnexal abnormalities are the commonest[8].

Linkages studies have largely been unsuccessful and the risk of siblings is generally low[5]. Epidemiologic and laboratory evidence support a hypothesis that there may be genetic (recessive) predisposition to the teratogenic effect of mild to moderate maternal vitamin A deficiency (VAD) during pregnancy. This may explain high prevalence of congenital eye anomalies in a part of Asian countries, where maternal VAD is common and consanguineous marriages are common[9].

Epidemiologic evidence supports a hypothesis that chromosomal abnormalities induced by environmental factors are associated with ocular abnormalities seen. The aetiologic basis of the congenital eye defects could only be inferred from available information in our study .Lack of appropriate genetic diagnostic facilities made laboratory diagnostic confirmatory evidence impossible.

History of exposure to irradiation was obtained in two parents. One was a father who worked in the teaching hospital as a radiologist while the other was a mother who works as a technologist in the radiography department of another hospital.

None of the patients had a family history of ocular or non-ocular congenital defects. Parental age had no significant association with the presence or type of ocular defects.

Associated systemic defects recorded in our study were congenital rubella syndrome (CRS), Goldenhar's syndrome, Apert's syndrome, Downs's syndrome, cleft palate, and meningoencephalocoele and auriculovertebral dysplasia.

In addition to the defects recorded in our study, other researchers have reported congenital absence of inferior rectus muscle[15], congenital diaphragmatic hernia (CDH) in association with a wide range of ocular malformations[16], Para macular coloboma[17], congenital reduplication of the lacrimal punctum and canaliculus [16] among others.

Although congenital anophthalmia and microphthalmia have been successfully treated with the use of non-expansible conformers[19]we were unable to manage such cases largely because our centre lacks the required facilities.

Congenital cataracts and glaucoma have been successfully treated as well[8, 16, 20]. Surgical procedure for congenital glaucoma included trabeculectomy, trabeculotomy, goniotomy, Molteno shunt implantation, cyclodialysis, and cyclocryotherapy[20].

Five out of six cases of congenital cataract in our study were successfully managed with lensectomy and spectacle correction of aphakia, and we recorded good visual recovery i.e. visual acuity was better than 6/18 in the better eye with correction. Follow up was still difficult due to high rate of default (67.7%) among our patients.

Two out of the three patients with congenital glaucoma had surgery and only one of this was followed up for more than six months. This patient had an IOP of 18mmHg a year post surgery. Overall success rate in patients with congenital glaucoma could not be determined due to high default rate. The high default rate is an indication for early surgery in patients with congenital glaucoma in Nigeria.

Lesions like lid coloboma, haemangiomas, and limbal dermoid, ptosis, and nasolacrimal ducts obstruction

were successfully managed with appropriate surgical procedures i.e. Flap rotation surgery for coloboma, tumour resection with reconstructions for lid haemangioma and tumour resection with conjunctiva auto graft for limbal dermoid. We recorded good structural, functional and visual outcomes (VA better than 6/18).

The patient who had frontonasal meningoencephalocoele was jointly managed by a team of neurosurgeon, plastic surgeon, orthopaedic surgeon, otolaryngologist and ophthalmologist. Plastic surgeons were involved in the management of the patient with frontonasal haemangioma.

Five patients presented with bilateral micro-ophthalmia and two of them were blind in both eyes at presentation while the other two had uniocular blindness (VA=NPL). No definitive treatment could be offered for the 3-months old boy who had bilateral micro-ophthalmia and congenital cataract since he was irreversibly blind.

Our study was designed to determine the incidence and pattern of presentation of congenital and developmental anomalies of the eye in our community. We studied patients who attended clinics at the teaching hospital in Ile-Ife.

The figures reported may not be truly representative of the picture in our community since not all the patients with birth defects will eventually present in the teaching hospital.

Congenital eye defects constitute a significant cause of morbidity and blindness (1.6%) among cases of congenital defects seen in the teaching hospital. There are public health implications for the prevention of blindness due to congenital eye malformations.

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Table1
DEMOGRAPHIC CHARACTERISTICS OF THE STUDY POPULATION

Age Range (yrs)	Male (%)	Female (%)	Total (%)
<1	9 (29.0)	14 (45.2)	23 (74.2)
1-9	6 (19.4)	1 (3.2)	7 (22.6)
10-19	-	-	-
20 - 29	1 (3.2)	-	1 (3.2)
Total.	16 (51.6)	15 (48.4)	31 (100.0)

Table2.
DISTRIBUTION OF CONGENITAL OCULAR DEFECTS IN THE STUDY POPULATION

Type of Defect	Male (%)	Female (%)	Total (%)
Congenital cataract	4 (12.9)	2 (6.5)	6 (19.4)
Microphthalmia	2 (6.5)	3 (9.7)	5 (16.1)
Nasolacrimal duct obstruction	3 (9.7)	1 (3.2)	4 (12.9)
Congenital glaucoma	1 (3.2)	2 (6.5)	3 (9.7)
Lid coloboma	-	3 (9.7)	3 (9.7)
Ptosis	2 (6.5)	-	2 (6.5)
Proptosis	1 (3.2)	1 (3.2)	2 (6.5)
Limbal dermoid	-	1 (3.2)	1 (3.2)
Entropion	-	1 (3.2)	1 (3.2)
Anophthalmos	1 (3.2)	-	1 (3.2)
Squint	-	1 (3.2)	1 (3.2)
Lid haemangioma	2 (6.5)	-	2 (6.5)
Total	16 (51.6)	15 (48.4)	31 (100.0)

Table3
ASSOCIATED NON-OCULAR CONGENITAL DEFECTS IN THE STUDY POPULATION

Non-Ocular Defect	Male (%)	Female (%)	Total (%)
Meningoencephalo coele	3 (33.3)	1 (11.1)	4 (44.4)
Congenital Rubella Syndrome	2 (22.2)	-	2 (22.2)
Aperts Syndrome	1 (11.1)	1 (11.1)	2 (22.2)
Auriculo-vertebral dysplasia	-	1 (11.1)	1 (11.1)
Total	7 (55.6)	4 (44.4)	11 (100.0