Congenital eye anomalies in Enugu, South-eastern Nigeria

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Summary

Objectives: **To determine the types of congenital ocular anomalies seen in the eye clinic of the University of Nigeria Teaching Hospital Enugu, over an 8-year period from January 1992 to December 1999.**

Method: **In a retrospective non-comparative case-series study, the records of all patients with congenital ocular anomalies seen between January 1992 and December 1999, were analysed for types of anomalies, aetiology, sex and ages at presentation of all patients.**

Results: **A total of 54 cases were analysed: 25 females; 29 males (M:F = 1: 1. 2).**

Congenital cataract was the most frequently occurring congenital ocular anomaly (42.6%); followed by congenital glaucoma (22.2%) and anophthalmia/microphthalmia and congenital esotropia (9.3%) each.

Conclusion: **In spite of the limitations of this study our findings are similar to those in other studies.**

Key-words: *Congenital, Anomalies, Eye.*

Résumé

L'objectif: Determiner les types des anomalies congénitales oculaires vues dans la clinique oculaire du centre hospitalier universitaire du Nigeria, Enugu au cours d'une période de 8 ans du janvier 1992 au décembre 1999.

Méthodes: A travers une étude de séries des cas non comparatif retrospectif, les dossiers de tous les patients avec des anomalies congenitales oculaires vues entre janvier 1992 et decembre 1999 ont été analysés pour savoir les types des anomalies, étiologies, sexe et âge pendant présentation de tous les patients.

Résultats: Un nombre total de 54 cas ont été analysés 25 du sexe féminnin, 29 sexe masculins (M:F = 1,1:2). La cataracte congénitale était une anomalie avec une fréquence la plus remarquable 42,6%, Suivie par glaucome congénital 22,2% et anophtalmie/microphtalmie et esotropie congénitale 9,3% chacun.

Conclusion: En dépit des limitations de cette étude, nos résultats sont pareils avec ceux des autres études.

Introduction

There is scarcely any data from Africa on the public health importance of blindness or visual morbidity from congenital ocular anomalies. Most emphasis is on cataract-blindness and in some areas on trachoma and onchocerciasis.

Yet, even though rare and not of much impact when considered singly, collectively the impact of congenital eye malformation on vision is significant[1].

It is estimated that the major structural ocular malformations anophthalmos/microphthalmos and coloboma account for 16.7% of childhood blindness globally[2].

Childhood blindness is one of the diseases that have been given priority in the ongoing WHO Vision programme 'The Right to Sight' because of the higher number of blind-years a blind child has[3].

Additionally, visual loss in childhood has implications for all aspects of the child's development. It poses educational, occupational and social challenges with affected children being at risk of behavioural, psychological and emotional difficulties, impaired self-esteem and poorer social integration[4-5].

In developing countries where a much higher proportion of the population are children than in affluent regions, it is estimated that the number of blind children is about 600/million[2].

This study has been conducted in the eye department of the University of Nigeria Teaching Hospital, Enugu to determine the types of congenital ocular anomalies seen in the major and oldest tertiary hospital in South-Eastern Nigeria.

Methods

The records of all cases of congenital malformations of the eye seen at the eye clinic of University of Nigeria Teaching Hospital (U.N.T.H), Enugu over an 8-year period (January 1992 – December, 1999) were reviewed. They were analysed for types, aetiology sex and ages at presentation of patients.

Results

Information on types of anomalies and sex of patients is summarised in table 1.

There were 29 males and 25 females (M:F = 1.2:1). Congenital cataract was the most common anomaly accounting for 42.6% of all the cases seen.

Anophthalmos/microphthalmos accounted for 9.3% of the cases seen being the third most frequently occurring anomaly together with congenital esotropia.

The aetiology was not determined in 51 cases. Congenital rubella syndrome was presumed to be the cause in 2 cases of cataract, and maternal ingestion of steroid contraceptives suspected to be the aetiology in one.

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Table 1 Types of congenital anomalies of the eye seen at UNTH, Enugu (1992– 1999)

Type of malformation	Sex M	Sex F	Total	Frequency (%)
1. Congenital Cataract	13	10	23	42.6
2. Congenital glaucoma	9	3	12	22.2
3. Congenital esotropia	2	3	5	9.3
4. Microphthalmos/anophthalmos	2	3	5	9.3
5. Congenital nystagmus	0	2	2	3.7
6. Congenital myogenic ptosis	0	2	2	3.7
7. Goldenhar's syndrome	1	0	1	1.9
8. Albinism (Oculocutaneous)	1	0	1	1.9
9. Iris coloboma	0	1	1	1.9
10. Limbal dermoid	0	1	1	1.9
11. Usher's syndrome	1	0	1	1.9
Total	**29**	**25**	**54**	**100%**

Table 2 Ages at presentation of patients with congenital anomalies seen at UNTH, Enugu (1992 – 1999)

Age	Number	Frequency (%)
</=1yr	19	35.1
>1 yr–5 yrs	16	30.5
>5yrs	19	35.1
	54	**100%**

More than 50% of the patients presented above the age of 1 year (Table 2).

Discussion

Congenital anomalies include all forms of developmental defects present at birth, whether caused by genetic, chromosomal or environmental aetiologies[6].

Out of the 1.5 million children estimated to be blind globally 16.7% (250,000) are due to major congenital malformations of the eye namely anophthalmos (absent eyes); microphthalmos (small eyes) and colobomas (incomplete eyes)[1].

In this study we analysed the records of all patients with congenital ocular anomalies who presented in the eye clinic of UNTH, Enugu over an 8 year-period (1992 – 1999).

As with all retrospective studies, our results must be interpreted cautiously.

Frequency of anomalies

In our series congenital cataract was the most common anomaly (42.6%) followed by congenital glaucoma (22.2%). Anophthalmos/microphthalmos and congenital estropia each accounting for 9.3% of the cases seen were the third most common occurring anomalies. Similar to our findings, Stoll et al[7] in France also reported congenital cataract as the most frequently occurring anomaly in their series. Contrarily however, anophthalmos/microphthalmos and coloboma were the second and third most common anomalies in Stoll's series[7].

Anophthalmos/microphthalmos only accounted for 3.9% of ocular anomalies seen in the latter's series and was the seventh most common anomaly. While Bermejo et al[8] found anophthalmos/microphthalmos to be the most common occurring anomaly in their series congenital cataract was the second most common and coloboma the third.

These studies are, however, not comparable in type of study, study population, and sample size.

Stoll et al[7] and Bermejo et al[8] prospectively studied congenital eye malformations in consecutive births in geographical areas while we studied cases that presented in the eye clinic of a tertiary hospital over an 8 year-period retrospectively.

Again, while we studied only 54 cases Stoll[7] and Bermejo[8] et al studied 78 and 414 cases respectively.

Onwasigwe's[9] study like ours was hospital-based. Similar to our findings, cataract was the most common occurring anomaly (30.7%) in his series. Contrary to our study this was followed by squint (19.8%) and buphthalmos (17.9%) respectively.

Cases of tarsal plate eversion and cryptophthalmos which were reported in his series were not found in ours.

Additionally, we found cases of nystagmus and simple myogenic ptosis which did not occur in his.

The differences in findings in the two studies could be accounted for by differences in the patient selection, sample size and duration of study. While Onwasigwe's cohort consisted of patients who were seen both prospectively and retrospectively over the 5 year-period of study we retrospectively studied 54 patients seen in 8 years.

Again, Onwasigwe's sample size was almost twice ours (101).

Ages at presentation

More than 50% of our patients first presented above the age of one year. This is contrary to the results from Onwasigwe's[9] series where only 18.8% of the patients presented above the age of 6 months. The rest presented between one and six months of age.

It is possible that most of our patients first sought help from spiritual sources such as the traditional healers, spiritual churches etc based on the belief that this unsightly appearance of their babies could not be from any other cause than evil spirits.

It is also likely that they were ignorant of whom to take their baby to on seeing this anomaly at birth.

Another factor that could determine how early a patient with congenital ocular malformation presents is the degree of anomaly e.g. a dot cataract could go unnoticed, or being seen as insignificant could be ignored by the parents.

However, a very hazy cornea from buphthalmos is more likely to present shortly after the child is born. On the other hand, a parent who has had a child with congenital eye malformation is more likely to notice a defect in any other child and present earlier irrespective of the degree of malformation.

Aetiology

This was not determined in 51 of the cases. However, Congenital rubella syndrome was presumed to be responsible for the congenital cataract in 2 patients, and maternal ingestion of steroids the suspected aetiology in one.

Two cases of congenital cataract had other associated ocular abnormalities, one of these in addition had a hearing loss.

Congenital cataract may be unilateral or bilateral, isolated or part of a long list of systemic diseases[10]. Galactosemia, intrauterine infections (TORCH), and heredity are some of the causes of congenital cataract.

Similar to our findings Onwasigwe[9] in his series also found congenital rubella syndrome to be the most common known cause responsible for 7.9% of the causes of congenital anomalies with congenital cataract.

The determination of aetiology in our study was based only on history and findings on ocular and systemic evaluation of the patients.

It is possible that obvious familial causes were missed as tests on parents and siblings for galactosemia were also not conducted in any of the patients.

In spite of its limitations the frequency of occurrence of the congenital ocular anomalies found in this study is similar to those reported in other studies, and the common types of the anomalies seen are amblyogenic.

Other associated anomalies

Five (9.3%) causes in our series had associated ocular and/or systemic malformations.

Stoll[7] and Bermejo et al[8] in theirs found 53.8% and 79% respectively to have other congenital anomalies.

Again, while the systemic anomalies reported in our series were deafness and cleft lip, Bermejo[8] found limb anomalies to be the most frequently occurring (59.3%) followed by auricular/facial anomalies (47.1%). Oral clefts occurred in only 29.4% of the patients.

Clubfeet was also reported to be one of the more common systemic anomalies in Stoll's series[7]. Others included microcephaly, hydrocephaly and facial dysmorphia.

Malformations of the eye and its surrounding tissues may occur in isolation, in combination, or as part of a systemic malformation syndrome[10].

Therefore, whenever a congenital ocular anomaly is identified, other associated abnormalities must be sought for by a detailed ocular and systemic examination.

Conclusion

In spite of its limitations, the frequency of occurrence of the ocular anomalies found in this study is similar to those reported in other studies, and the common types of the anomalies seen are amblyogenic.

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