CONGENITAL EYE DISEASES IN ABUJA, NIGERIA

*OE BABALOLA *Mb Chb, FWACS, FMCOphth*

BI BABALOLA, RN, RM, BSc.

Rachel Eye Center, P.O. Box 4108, Garki, Abuja
Rachel@alpha.linkserve.com

SUMMARY

Aim: To look at the incidence of congenital eye disease in Rachel Eye Center, Abuja, in the hope that some of the more common problems may be highlighted and strategies for a more effective service may evolve.

Materials and Methods: The case notes of all new patients aged one year or less who presented at the Rachel Eye Center, Abuja, where between 150-250 new cases are seen every month, over a ten-year period (1990 and 2000) were reviewed.

Results: One hundred and thirty seven patients with a definitive diagnosis were selected for the study, of which 30.5% had congenital eye disease. The commonest causes were nasolacrimal duct obstruction (12.4%), cataract with or without microphthalmos (5.1%), strabismus (2.9%), and buphthalmos (2.9%).

Conclusion: Further research is needed to determine what proportion of congenital cataracts is associated with rubella in Nigeria, even though the association is well established elsewhere, so that the MMR (measles, mumps, and rubella) vaccine may be included in the Expanded Programme on Immunization, if it is found to be a problem of public health significance.

There is also a need to strengthen the institution of social welfare in the country so that those born incurably blind can be better assisted.

Key words: congenital blindness, cataract, rubella, mumps, measles

INTRODUCTION

The eye is one of the most embryologically complex organs in the body with epidermal, mesodermal and endodermal components.[1] There is a racial heterogeneity in the incidence and prevalence of congenital eye disease, which results from the complex interaction of genetic and environmental factors. It is estimated that 3% of all live births have congenital defects[1] and about 1.6 in 1000 live births are born with ocular malformations.[2] There is also some evidence that there are racial differences in the prevalence of these defects. For instance, Chumbley[3] noted that there was a low prevalence of concomitant squints and angle closure glaucoma in black Mashona populations in Zimbabwe, and Chew et al.[4] found that Caucasians are generally predisposed to develop squints, especially esotropia.

Other researchers in Nigeria, including Akinsola, and Ajaiyeoba,[5] Nwosu,[6] Majekodunmi,[7] Soyannwo and Baiyeroju,[8] Ukponwan,[9] and Dawodu,[10] have variously addressed the problem of congenital eye disease in Nigeria.

These racial differences prompted us to look at the incidence of congenital eye disease at Rachel Eye Center in Abuja, in the hope that some of the more common problems may be highlighted and strategies for a more effective service may evolve.

MATERIALS AND METHODS

The Rachel Eye Center is an urban practice located in the Federal Capital Territory of Nigeria. The clinic has two main branches in Kubwa and Abuja; it also carries out community outreach from time to time. The catchment area of the clinic extends beyond the Federal Capital Territory, and between 150 - 250 new cases are seen every month. The case notes of all new patients aged less than one year at presentation, seen in the clinic between 1990 and 2000, were retrieved from the computerized databank and analysed for ocular pathology in the worst affected eye. All cases in which the diagnosis was not explicit were excluded. The various pathologies were then classified into 'congenital' and 'non'congenital'. The functional definition of 'congenital' adopted was: 'a problem present at birth

* *Author for correspondence*

which is attributable to genetic, hereditary or intra-uterine factors'. This definition excluded conditions which, though present at birth, were manifestly acquired during parturition, such as ophthalmia neonatorum and forceps injury. The data was analysed using Microsoft Excel.

RESULTS

One hundred and thirty seven new patients, aged less than one year at presentation, were selected for the study. The frequency distribution associated with various diagnoses is shown in table 1, while table 2 shows the distribution of pathologies deemed congenital. Of the 137 cases identified with pathology, 42 were congenital (30.5% of all cases). The commonest cause of ocular morbidity (congenital and otherwise) among the group was vernal conjunctivitis, accounting for a little less than one in three cases, followed by conjunctivitis and ophthalmia neonatorum accounting for 16% of cases. However, the commonest cause of congenital ocular morbidity in this series was nasolacrimal duct obstruction (17 cases), which accounted for 12% of all morbidity and 40% of all congenital eye morbidity. This was followed by cataracts with or without microphthalmos (7 cases), strabismus (4 cases), buphthalmos (4), and two cases each of cortical blindness and cerebral palsy. One case each was seen of heterochromia iridis, cloudy cornea of unspecified aetiology, optic atrophy, ocular albinism, dermoid cyst and retinoblastoma.

Table 1. Ocular Pathology in Patients < 1 year old seen at Rachel Eye Center, Abuja 1990-2000

S/No	Pathology	Frequency	%	Congenital?
1	Vernal conjunctivitis	41	29.9	No
2	Conjunctivitis	22	16.1	No
3	Nasolacrimal duct obstruction	17	12.4	Yes
4	Ophthalmia neonatorum	10	7.3	No
5	Cataracts +- microphthalmos	7	5.1	Yes
6	Keratitis	7	5.1	No
7	Trauma	5	3.6	No
8	Strabismus	4	2.9	Yes
9	Buphthalmos	4	2.9	Yes
10	Cortical blindness	2	1.5	Yes
11	Anterior staphyloma	2	1.5	No
12	Episcleritis	2	0.15	No
13	Cerebral palsy	2	1.5	Yes
14	Orbital cellulitis	2	1.5	No
15	Proptosis	1	0.7	No
16	Heterochromia iridis	1	0.7	Yes
17	Cloudy cornea? Cause	1	0.7	Yes
18	Retrobulbar neuritis	1	0.7	No
19	Optic atrophy	1	0.7	Yes
20	Mucocutaneous lesion	1	0.7	No
21	Corneal foreign body	1	0.7	No
22	Ocular albinism	1	0.7	Yes
23	Dermoid cyst	1	0.7	Yes
24	Retinoblastoma	1	0.7	Yes
	Total	137	100	

Table 2. Congenital Eye Diseases seen at REC 1990-2000

S/No	Pathology	Frequency	Percent of all cases	Percent of Congenital cases
1	Nasolacrimal duct obstruction	17	12.4	40.4
2	Cataracts +- microphthalmos	7	5.1	16.6
3	Strabismus	4	2.9	9.5
4	Buphthalmos	4	2.9	9.5
5	Cortical blindness	2	1.5	4.7
6	Cerebral palsy	2	1.5	4.7
7	Heterochromia iridis	1	0.7	2.3
8	Cloudy cornea? Cause	1	0.7	2.3
9	Optic atrophy	1	0.7	2.3
10	Ocular albinism	1	0.7	2.3
11	Dermoid cyst	1	0.7	2.3
12	Retinoblastoma	1	0.7	2.3
	Total	**42**	**30.5**	**100**

DISCUSSION

According to Shephard,[11] the cause of congenital malformations is unknown in 40-65% of instances. It is associated with a specific teratogenic agent in 8-10% of

cases, is monogenic in 15-25% of cases, and is due to chromosomal abnormalities in 15-28% of cases. It is, therefore, important to identify the causes of congenital eye malformations in our environment with a view to mapping out preventive strategies wherever possible. The commonest cause of congenital eye disease in our series is nasolacrimal duct obstruction. This is said to be due to delayed canalization near the valve of Hasner,[12] and is characterized by epiphora and watering of the eyes in infants. The fluorescent dye clearance test is useful for diagnosis, especially if it is unilateral. It is often necessary, however, to rule out congenital glaucoma. We recommend massage initially, followed by probing and syringing and then dacryo-cystorhinostomy (DCR), if these measures fail. About 98% of our cases have required probing and syringing and this may have to be repeated in about 5% of the cases. Only very few require DCR. This procedure is best undertaken by trained paediatric ophthalmologists which are unfortunately few and far between in Nigeria.

Congenital cataracts are associated with a multiplicity of causes. These include intrauterine infections, especially rubella. Viruses were isolated in 10% of congenital cataract lens aspirates and rubella antibodies were detected in a further 24.5% of cases in an Indian study, raising the possibility of an association in about 35% of cases of congenital cataract.[13] Other intrauterine causes are toxoplasmosis and cytomegalovirus. There is still a debate as to whether or not it is necessary to introduce rubella vaccination in Nigeria. That would likely be the measles, mumps, and rubella (MMR) vaccine in infants, and prenatal vaccination for young women. The major argument for introducing the vaccine is the fact that a significant proportion of not only congenital eye disease but also congenital heart and other systemic diseases may be prevented. Arguments against are that: (a) the seroprevalence of rubella is already high in our community,[14] ranging in many studies above 80%, thus rendering any mass vaccination campaign potentially unnecessary with the presumption that immunity is already widespread, and (b) the vaunted possible association with autism following the vaccination,[15] although this is now hotly disputed.[16,17] These arguments notwithstanding, studies in France, where the vaccination has been administered for the past 35 years, have quantified the benefits of MMR vaccination in that country and concluded, inter alia, that it has led to huge benefits in terms of public health. Specifically, among other things, it has contributed to the avoidance of an estimated 3000 cases of rubella infection occurring during pregnancy with its well-known sequelae, and to the avoidance of 12,000 deaths. A study carried out in Maiduguri[14] indicated that 46% of pregnant women have no immunity against rubella and are therefore at risk. The study strongly recommended that MMR vaccination should be introduced in Nigeria.

Congenital cataracts are said to be hereditary, as an autosomal dominant trait in 33% of cases.[18] Babalola et al[19] have described family pedigrees of hereditary cataract in Nigeria. The importance of genetic counselling in this regard cannot be over- emphasized. Probands and parents of cases of congenital cataract ought always to be examined for signs of cataract; dominantly inherited cataract has highly variable penetrance and expressivity. The other important causative factor to be ruled out is of course galactosemia, either due to galactose-1-phosphate uridyl transferace (GIPUT) deficiency, which tends to have more widespread systemic manifestations, or due to galactokinase deficiency, which may have solely ocular manifestation in the form of cataract. A test for reducing substance after ingestion of milk products is mandatory for all cases of congenital cataract, especially since the cataract is potentially reversible if milk products are withheld.

The relative rarity of strabismus in African populations has been commented upon by other workers, especially those in multiracial societies.[3] It is still necessary, however, for the average resident to master the techniques of managing especially simple horizontal comitant squints, as well as amblyopia.

Buphthalmos is probably more common in Nigeria than our limited series would tend to suggest. Late presentation is a common problem, thus the only option open to a surgeon is trabeculectomy, rather than goniotomy, because corneal clarity has often been lost.

Cerebral palsy is not unusual in the newborn in our practice. In England and Scotland, Pharaoh et al.[20] observed a cerebral palsy prevalence of 2.1 per 1000 neonatal survivors, of which 8.9% had severe visual disability. Work needs to be done to improve social welfare mechanisms, which are weak in Nigeria; they offer the only means of adequate care for these unfortunate babies.

Neonatal/childhood cortical blindness (retrogeniculate cortical, i.e. grey matter, and subcortical, white matter, visual loss) is not unusual in the literature.[21] This damage is often related to peri-ventricular leucomalacia.

Cloudy cornea of uncertain aetiology may be associated with mucopolysaccharidosis, especially Scheie's (or Hurler's) syndrome. There are no corneal deposits in the Hunter and Sanfillippo variants.

Albinism is a problem frequently encountered in clinical practice in Nigeria. Tyrosinase positive and

tyrosinase negative as well as the X-linked ocular variant are encountered.[22] Unfortunately, foveal hypoplasia limits the effectiveness of optical corrections.

The management of retinoblastoma is problematic in Nigeria. This is largely owing to late presentation, by which time only enucleation or exenteration may be offered. The clinical choice becomes more of a dilemma when both eyes are affected, such as in the Ibadan study[23] where 18% of histologically confirmed cases were bilateral. This is compounded by the fact that many esoteric means of treatment such as radioactive plaques, external cobalt radiation, endo-laser photocoagulation and even cryo-therapy are simply not available in many centres in the country.

CONCLUSION

About one third of all patients less than one year old presenting with eye problems in our practice appear to have congenital eye disease. Nasolacrimal duct obstruction, cataracts, strabismus and buphthalmos were the commonest identified causes. Research is needed to determined what proportion of cataracts is associated with rubella in Nigeria, even though the association is well established elsewhere. It is recommended that the MMR vaccination should be included in the expanded programme of immunization in Nigeria. There is also a need to strengthen the institution of social welfare in the country so that those born incurably blind may be better assisted.

References

1. Kerstin Stromland and Marilyn Miller. Ocular teratology. In: *Duane's Foundations of Clinical Ophthalmology*. Philadelphia: Lippincott-Raven, 1997; chapter 39, p. 1.
2. Baird PA, Anderson TW, Newcombe HD, Lowry RB. Genetic disorders in children and young adults: A population study. *Am J Hum Genet* 1988; **42:** 677.
3. Chumbley LC. Impressions of eye diseases among Rhodesian blacks in Mashonaland. *S Afr Med J* 1977; **52(8):** 316-8.
4. Chew E, Remaley NA, Tamboli A,. Zhao J, Podgor MJ, Klebanoff M. Risk factors for esotropia and exotropia. *Arch Ophthalmol* 1994; **112(10):** 1349-55.
5. Akinsola FB and Ajaiyeoba AI. Causes of low vision and blindness in children in a blind school in Lagos, Nigeria. *West Afr J Med* 2002; **21(1):** 63-5.
6. Nwosu SN. Ocular problems of young adults in rural Nigeria. *Int Ophthalmol* 1998; **22(5):** 259-63.
7. Majekodunmi AA. Bilateral congenital anophthalmos. Report of two cases. *J Pediatr Ophthalmol* 1977; **14(1):** 42-3.
8. Soyannwo OA and Baiyeroju AM. Non-cardiac surgery and anaesthesia in children with congenital heart disease. *West Afr J Med* 1999; **18(2):** 87-90.
9. Ukponwan CO. Congenital anophthalmos in Benin city, Nigeria. *West Afr J Med* 1999; **18(2):** 141-3.
10. Dawodu OA. Total eversion of the upper eyelids in a newborn. *Niger Postgrad Med J* 2001; **8(3):** 145-7.
11. Shephard TH. Human teratogenicity. *Adv Peditr* 1986; **33:** 225.
12. Katowitz JA and Joanne EL. Lacrimal drainage surgery. *Duane's Ophthalmology* Vol. 5, 78, pp 4-5 Philadelphia: Lippincott-Raven. 1997.
13. Malathi J, Therese KL and Madhavan HN. The association of rubella virus in congenital cataract - a hospital based study in India. *J Clin Virol* 2001; **23(1-2):** 25-9.
14. Bukbuk DN, el Nafaty AU and Obed JY. Prevalence of rubella specific IgG antibody in non-immunized pregnant women in Maiduguri, north eastern Nigeria. *Centr Eur J Public Health* 2002; **10(1-2):** 21-23.
15. Wakefield AJ, Murch SH, Anthony A et al. Ileal-lymphoid-nodular hyperplasia, non-specific colitis, and pervasive developmental disorder in children. *Lancet* 1998; **351(9103):** 637-41.
16. DeStefano F and Thompson WW. MMR vaccination and autism: Is there a link? *Expert Opin. Drug Saf.* 2002; **1(2):** 115-20.
17. Miller, E. Measles-mumps-rubella vaccine and the development of autism. *Semin Pediatr Infect Dis* 2003; **14(3):** 199-206.
18. Beby F, Morle L, Michon L, Edery P, Burillon C, Denis P. The genetics of hereditary cataract. *J Fr Ophthalmol* 2003; **26(4):** 400-8.
19. Babalola OE, Danboyi P and Abiose AA. Hereditary congenital cataracts associated with sickle cell anemia in a Nigerian family. *Tropical Doctor* 1996; **30(1):** 12-14.
20. Pharaoh PO, Cooke T, Johnson MA, King R and Mutch L. Epidemiology of cerebral palsy in England and Scotland, 1984-9. *Arch Dis Child Fetal Neonatal Ed* 1999; **80(2):** F158.
21. Brodsky MC, Fray KJ and Glasier CM. Perinatal cortical and subcortical visual loss: Mechanisms of injury and associated ophthalmological signs. *Ophthalmology*. 2002; **109(1):** 85-94.
22. Okoro AN. Albinism in Nigeria. A clinical and social study. *Br J Dermatol* 1975; **92(5):** 485-92.
23. Akang EE, Ajaiyeoba IA, Campbell OB, Olurin IO, and Aghadiuno PU. Retinoblastomas in Ibadan, Nigeria: II - Clinicopathologic features. *West Afr J Med* 2000; **19(1):** 6-11.