

Heredo-Familial Expression of Ametropia among Siblings of a Nigerian Family

Emmanuel Olu Megbelayin^{1,2}, Sylvia Iquo-Abasi Akpan^{1,2}

¹Department of Ophthalmology, University of Uyo Teaching Hospital, Uyo, Nigeria

²Faculty of Clinical Sciences, University of Uyo, Uyo, Nigeria

Email: favouredolu@yahoo.com

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Abstract

A case series of 4 siblings who had auto-refraction and subjective refraction, refinement was with 0.25 Jackson Cross Cylinder. Presenting complaint was either blurring of distant vision or missing eye glasses. The eldest sibling started using eye glasses at 15 years of age while the other siblings commenced much earlier. Presenting visual acuities in 7 eyes ranged from 6/9 to 6/18, one eye of the eldest sibling had visual acuity of 6/36. Spherical errors had gender bias with the males more likely to be hypermetropic and the females more likely to be myopic. However, cylinder powers and axes were closely related in all siblings. It was concluded that heredo-familial traits could partly account for some of the striking similarities noted in the outcomes of refraction among the siblings.

Keywords

Heredofamilial, Siblings, Ametropia

1. Introduction

Many studies have corroborated the familial and heritability of refractive state of the eye [1] [2] [3]. The chief contributors to ocular refraction and dioptric power are cornea, lens, anterior chamber depth and axial length. Biio *et al.* [4] in 2005 reported that estimates of heritability for axial length range from 40% - 94% and anterior chamber depth from 70% - 94% with linkage to chromosomes 2p24 and 1P32.2 respectively. Heritability estimates for corneal curvature, in the same study, was 60% - 92% with linkage to chromosomes 2p25, 3P26 and 7q22. Lyhne *et al.* [5] in an earlier study in 2001 among twins aged 20 - 45 years reported 90% - 93% heritability for crystalline lens thickness. Different modes of Mendelian inheritance are associated with refractive errors including autosomal dominant (AD) and sex-linked (X-linked). Loci for autosomal do-

minant high myopia are located on chromosomes Xq28 18p11.31 2q37, Xq23-25 and 4q.

To date, almost 100% of identified loci for non-syndromic high myopia are either AD or X-linked with high penetrance [6]. In a dizygotic twin study, Hammond *et al.* [2] found that Paired box gene 6 (PAX 6) is strongly linked with refractive errors. Myopia has the strongest evidence for genetic susceptibility, although studies have shown different loci between juvenile-onset myopia (low to moderate myopia) and high myopia. Conversely, hypermetropia and astigmatism have weaker and less consistent linkage with inheritance [7] [8].

In the current report, we undertook a classic comparative study to assess similarities and differences in the outcomes of refraction among 4 siblings of an African family.

2. History

Four siblings from a monogamous setting presented on the same clinic day with history of either missing or broken eye glasses. The youngest of the sibling was 9 years old while others were 14, 18, 21 years old. The first two siblings were males while the last two were females. The onsets of eye glasses use were 6, 13, 12, 15 years from the youngest to the eldest sibling respectively, inability to see distant objects being the reason for their use. Both parents use glasses for sight and reading. Other parts of the history were not contributory.

Clinical examination revealed normal anterior and posterior segments. Auto refraction was carried out with POTEK CO., LTD, (PRK) 2000, Korea followed by a subjective refraction. Axis was refined with Jackson Cross Cylinder (-0.25 type). The outcomes of refraction were satisfactory precluding the need for cycloplegic refraction.

3. Results

There were 4 siblings, 2 males and 2 females. Presenting visual acuities (VA) are shown in **Table 1**. VA improved to 6/6 in both eyes in all siblings after refraction except the last sibling who had a VA of 6/9 (OU). Details of the refraction of the patients are shown in **Table 2**.

4. Discussion

In our practice, it is an uncommon occurrence for 4 siblings to present on the same clinic day for a non-infectious ocular condition like epidemic haemorrhagic conjunctivitis. This occasion was rare and the striking outcomes of refractions were easily noticed. Studies on refractive errors are common in our clime; however, analytical studies of cases series are sparse. Studies in support of genetics as one of the risk factors of refractive errors have focused on molecular theories [2] [4]. The strength of the current study is the comparative analysis involved in which refraction results of first degree relatives are juxtaposed to allow for logical inferences.

Presenting VA in the siblings ranged from 6/9⁺¹ to 6/36. Except for the eldest, there was clustering of presenting vision between 6/9 and 6/18. This falls within a normal range according to World Health Organization [WHO] classification while the 6/36

Table 1. Presenting visual acuity.

Sibling	Visual acuity	
	OD	OS
1	6/9 ⁺¹	6/12
2	6/12	6/12
3	6/12	6/18 ⁺²
4	6/36	6/18

Table 2. Analysis of refraction results in all siblings.

Sibling	Age (years)	Sex	Post-refraction									
			Spheres		Cylinders		Axes		Spherical equivalents		Astigmatism type	
			OD	OS	OD	OS	OD	OS	OD	OS	OD	OS
1	9	Male	+0.50	+0.50	-3.00	-3.50	30	160	-1.00	-1.25	With the rule	With the rule
2	14	Male	0.00	0.00	-2.00	-2.00	180	180	-1.00	-1.00	With the rule	With the rule
3	18	Female	-2.50	-3.00	-2.00	-2.50	180	180	-3.50	-4.25	With the rule	With the rule
4	21	Female	-3.50	-2.00	-3.00	-2.50	20	170	-5.00	-3.25	With the rule	With the rule

presenting VA in the right eye of the eldest patient is considered to be a form of visual impairment. This can be attributable, at least in part, to a delay in use of glasses until 15 years. Stimulus deprivation amblyopia is a likely outcome of an eye deprived of quality retinal images over a prolonged period as the brain has learnt to cope with blurred images.

Gender as a determinant of refractive status has not been firmly established in available epidemiological studies. In a study in Szczecin, Poland among school children aged 6 - 18 years, Czepita *et al.* [9] reported that myopia occurred more frequently in girls (7.4%) than in boys (5.1%)— $p < 0.001$. Hyperopia occurred more frequently in boys (19.6%) than in girls (18.2%)— $p < 0.001$. A slightly higher prevalence of astigmatism in girls (1.9%) than in boys (1.5%) was also observed ($p > 0.05$). In that study, whereas spherical errors reached statistical significance, cylindrical error was not statistically significant. This outlook is similar to the findings in the current study where the males have a close range spherical error; plano to +0.50. On the other hand, the females share common myopic characteristics. This presents a bipolar spectrum where males lie on the hypermetropic end and females on the myopic end. Similar to the finding by Czepita *et al.*, these siblings have cylindrical errors that had no gender bias.

There is the possibility that the non-gender disposition of astigmatism in these siblings may have been decided by inheritance as all astigmatic powers clustered around -2.00 and -3.50 and the axes in all hovered around 180 degrees. Additionally, all had

with-the-rule astigmatism. In a twin study, Hammond *et al.* [1] reported dominant genetic effects accounting for between 42% and 61% of the variance of astigmatism. In another twin study, Dirani *et al.* [10] reported a similar proportion of the variance (50%) of dominant genetic effects in astigmatism. Although, research on the genetics of astigmatism is limited, these studies [1] [11] [12] and the current observation among siblings suggest that dominant genes may be linked with astigmatism.

With spherical equivalents that ranged from -1.00 to -5.00 , all the siblings were myopic. This agrees with previous studies [13] [14] that myopia has a strong genetic mode of inheritance. To date, several genetic loci for non-syndromic myopia have been mapped, including 12 loci linked to high myopia. Moreover, two recent independent genome-wide association studies involving large cohorts of refractive error patients identified loci at chromosome 15q14 and 15q25 [15] [16] [17].

From the observation of refraction outcomes of the siblings under study, it is concluded that genetics continues to play significant role in emmetropization much as previously established environmental factors like near-work and urbanization [18] [19]. It is hoped that further studies would expand the scope of the current study to accommodate more families.

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