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Twin Study on Myopia

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Abstract. In order to reassess the relative importance of genetic and environmental factors in the development of myopia in Chinese schoolchildren, 90 pairs of MZ and 36 pairs of like-sex DZ twins were enrolled for detailed ophthalmological examination about their ocular refractions. Corneal curvatures and axial lengths were also measured. With equivalent settings of the range for concordance, corneal curvatures showed higher concordance rate (84%) than ocular refractions (65%) and axial lenghts (59%) in MZ twins. F-test on the intrapair variances between MZ and DZ twins revealed a significant hereditary role in determining the ocular refraction and its optical components. The degree of genetic determination was expressed by heritability indices, which were derived from intraclass correlation coefficients. The diversity of sample ages and refractions, while making the obtained data difficult to further explore the gene-environment interaction, led to the observation of more intrapair differences with age and myopic progression in MZ twins.

Key words: Myopia, Heritability, Twin concordance

INTRODUCTION

Recent epidemiological studies [2,4] have shown that the prevalence of myopia among schoolchildren in Taiwan is as high as 80% at the age of 18. Biometric analyses [4,5] pointed to the importance of axial length elongation in the production and progression of myopia.

While traditional concepts, based on Sorsby's biological theory [9], hold that most ocular refractions are determined by polygenic inheritance, the distribution of ocular refractions among students in Taiwan is too much skewed toward myopia. The 90% prevalence rate of near-sightedness among medical students in two medical colleges [3,7]

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cannot be fully explained by genetic theory alone, nor can their "mean" refractive status of –4 diopters be normalized.

In Sorsby's twin study [8], they set 0.5 mm difference in axial lengths, equivalent to 1.5 diopters' discrepancy in ocular refractions, as the range for concordance, resulting in an 83% concordance rate for axial lengths with uniovular twins instead of the actual 60%. This erroneously high concordance rate was then interpreted as "hereditary".

In order to elucidate the genetic and/or environmental components in the determination of ocular refractions, the present twin study was carried out.

MATERIALS AND METHODS

Same-sex twin schoolchildren were recruited for detailed ophthalmological examinations. Zygosity was determined mainly on the basis of red-cell antigen systems (ABO, MNSs and Rh), birth membranes, and similarity index, together with dermatoglyphic analysis. Thus, 43 male and 47 female MZ and 19 male and 17 female DZ twin pairs aged 7 to 23 years (mean = 15.9 ± 3.4) were enrolled in this study.

After subjective method of visual acuity testing with spectacle corrections, three drops of 1% tropicamide were instilled into each eye at intervals of five minutes. Retinoscopy was performed 20 to 40 minutes following instillations to ensure maximal cycloplegia. And the results were checked with the automated refractometer to minimize the bias. For eyes in which astigmatism was present, the spherical equivalent was taken into account.

Keratometry was measured with an ophthalmometer (Topcon, OM3). Corneal curvatures of both horizontal and vertical meridians were recorded and averaged. The axial lengths of eyeballs were determined ultrasonographically with a digital biometric ruler (Sonometrics, Model 310).

RESULTS

The data obtained were analyzed as follows.

Incidence of concordance. The average intra- and interexamination errors were 0.25 to 0.5 diopters for ocular refractions, 0.05 mm for keratometry and 0.1 to 0.2 mm for axial length measurements with ultrasound. So, for the incidence of concordance, differences of 0.5 D in ocular refractions, 0.1 mm in corneal curvature and 0.2 mm in axial lengths were set as the range for concordance. In fact, the latter two values also are roughly equivalent to 0.5 D according to Gullstrand's equation of schematic eyes. Table 1 shows the intrapair concordance rates for ocular refraction, corneal curvature and axial length in MZ and DZ twins.

The difference of concordance rates of ocular refraction between MZ (65%) and DZ (46%) is significant ($P < 0.01$). The difference for corneal curvatures (84% vs 72%) is also significant ($P < 0.05$). But there is no significant difference of concordance rates for axial length between MZ (59%) and DZ (47%) twins.

Table 1 - Intrapair concordance

	Ocular refraction		Corneal curvature		Axial length	
	R	L	R	L	R	L
MZ twins	62/90 (69%)	55/90 (61%)	78/90 (87%)	74/90 (82%)	48/74 (65%)	39/74 (53%)
DZ twins	17/36 (47%)	16/36 (44%)	25/36 (69%)	27/36 (75%)	15/31 (48%)	14/31 (45%)
	$\chi^2 = 8.0$ P < 0.01		$\chi^2 = 5.0$ P < 0.05		$\chi^2 = 2.5$ P < 0.01	

Genetic variance. Intra- and interpair variances may be compared by dividing the larger one by the smaller one to give F value. Table 2 showed the intra- and interpair F values for ocular refraction, corneal curvature and axial lengths. A significant intrapair F value with an insignificant interpair F value implies that hereditary factors play a role in the determination of ocular refraction and its optical components.

Correlations coefficient. The intraclass correlation coefficient is derived from the difference between inter- and intrapair variances, divided by the sum of inter- and intrapair variances. The correlation coefficients with ocular refraction, corneal curvature and axial length in MZ and DZ twins are shown in Table 3.

Heritability estimation. According to Falconer (1960), heritability is defined as $h^2 = 2 (r_{MZ} - r_{DZ})$, where r is the correlation coefficient. Table 4 shows heritability indices for ocular refraction ($h^2 = 0.24$) corneal curvature ($h^2 = 0.25$) and axial length ($h^2 = 0.27$).

Table 2 - F values for genetic variances

	Ocular refraction		Corneal curvature		Axial length	
	R	L	R	L	R	L
Intrapair	2.2	1.5	2.1	1.7	2.9	2.2
Interpair	0.9	0.8	1.1	1.1	1.0	1,1

Table 3 - Intraclass correlation coefficients

	Ocular refraction		Corneal curvature		Axial length	
	R	L	R	L	R	L
MZ pairs	0.88	0.83	0.89	0.86	0.92	0.88
DZ pairs	0.76	0.79	0.77	0.75	0.79	0.72

Table 4 - Heritability indices

	Ocular refraction	Corneal curvature	Axial length
R	0.24	0.25	0.27
L	0.07	0.22	0.31

DISCUSSION

Our results show that heredity plays a role in the determination of ocular refraction and its optical components, especially the anterior corneal curvature, which has 85% concordance rate in MZ twins, the heritability index being estimated as $h^2 = 0.25$. The axial length, though heritability is 0.27, has a relatively low concordance rate: only 59% in MZ twins.

The ranges for concordance can be arbitrarily chosen with varied incidences of concordance. While the settings of difference we used seem quite reasonable, they would result in too high concordance rates for both MZ and DZ twins, if applied in studying the genetic or environmental nature of astigmatism, either ocular or corneal.

However, other approaches have their limitations. The diversity of ocular refractions with age could have increased the interpair variances to inflate the correlation coefficients. Genetic variances and heritability estimations were thus disturbed, making the analysis more difficult.

Table 5 - Intrapair differences among MZ twins with ages

	0	± 0.5	± 1.0	± 1.5	± 2.0	± 2.5	± 3.0	Total
7	1	1						2
8								0
9	2			2				4
10	16	4						20
11		3	1					4
12	3	1	1	1				6
13				2				2
14	12	2	1	1	2			18
15	9	4	5	3	1	1	1	24
16	15	10	4	2	2	1		34
17	2	2	4	4				12
18	7	5	2					14
19		4	2	1		1		8
20	6	7	3	2	1	1	2	22
21			2	2		1	1	6
22	1		1					2
23					2			2
Total	74	43	26	20	8	5	4	180

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Table 6 - Intrapair differences among MZ twins with refractions

	± 0	± 0.5	± 1.0	± 1.5	± 2.0	± 2.5	± 3.0	Total
+ 2.0		1			1			2
+ 1.5								0
+ 1.0	6	2						8
+ 0.5	18	4	1					23
E	20	5	2	2				29
– 0.5	8	4	1					13
– 1.0	2	4	2	2				10
– 1.5	3	5	4	2				14
– 2.0	2	4	3	3	2		1	15
– 2.5		1	2	6	1	1		11
– 3.0	6	1	2		1	1		11
– 3.5	1	1	2	1	1	1	1	8
– 4.0	3	4	1		1			9
– 4.5	1	1	1		1			4
– 5.0	1	1	1	1		1	1	6
– 5.5		1	2	2			1	6
– 6.0	1	2		1				4
– 6.5		1	1					2
– 7.0	1							1
– 8.0	1	1	1			1		4
Total	74	43	26	20	8	5	4	180

As a matter of fact, intrapair differences of ocular refraction in MZ twins increased with age too (Table 5, $R = 0.33$, $P < 0.01$). Table 6 indicates that the intrapair differences also increased with the progression of myopia ($R = 0.19$, $P < 0.05$). The variegated ocular refractions with age and myopic progression implied that the environmental factors might affect the eye in the way of "myopization". A previous study [1] also suggested that the impact of environmental factors, like the habit of studying and reading, could interact with genotype in the development of myopia.

From the standpoint of epidemiology, if the population studied has low prevalence of myopia, it is relatively difficult to find the environmental components in the determination of ocular refraction. This happened with Sorsby's series [8], in which only 20% were myopic. In contrast, over 60% of twins in our series had myopia. Hence, it will provide us with a better understanding of the gene-environment interaction on myopia, when longitudinal follow-ups are carried on [6].

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