

Family Studies on Ocular Refraction and Its Components

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A study on 78 pairs of uniovular twins contrasted with 40 pairs of binovular twins and 48 pairs of unrelated subjects has established that refraction as a whole and its major individual components—the axial length and the powers of the cornea and lens—are genetically determined both for emmetropia and the various types of refractive errors, including those of a high degree (Sorsby, Sheridan, and Leary, 1962; Sorsby and Fraser, 1964). The coefficients of correlation in these different refractive states for refraction and its components approached unity in the uniovular twins, 0.5 in the binovular twins, and zero in the unrelated controls. It is likely that more than one mode of inheritance comes into play, and family studies have been undertaken to evaluate this aspect.

Material and Methods

Twenty-eight families with a total of 106 subjects were studied. To eliminate disturbances arising from senescent changes in the parents, and from uncompleted growth in the offspring, only families in which the parents were under 63 years and the offspring studied were 16 years and over were considered. Any family in which either of the parents showed a pathological eye trait (other than an unusual refraction) was excluded. In all other respects the families were an unselected and probably a representative sample of the general population. The detailed findings are recorded in the Appendix.

In addition to these families, the records were also studied of a group of 21 families selected for unusual refraction in either one of the parents or one of the offspring. This group has been reported in a previous study (Sorsby, Benjamin, Davey, Sheridan, and Tanner, 1957), and a further statistical assessment is attempted here.

The methods employed have been recorded in the two previous studies and are discussed more fully elsewhere (Sorsby, Benjamin, and Sheridan, 1961).

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Analysis

The coefficients of correlation in the 28 unselected families are set out in Table I. The over-all pattern is clear enough: in contrast to the lack of correlation between the parents, there is a substantial degree of correlation for parent/child and for mid-parent/offspring coefficients, though these coefficients fall short of the values of 0.50 and 0.71 expected under the hypothesis of the additive effects of a number of genes without dominance. The coefficients for the relatively small number of sibs available approach the expected value of 0.5 for refraction as a whole and for axial length and the power of the lens.

TABLE I

COEFFICIENTS OF CORRELATION FOR REFRACTION AND ITS COMPONENTS IN 28 UNSELECTED FAMILIES

	Father/ Mother 28 Pairs	Parent/ Child 100	Mid- parent/ Offspring 50	Sib/Sib 24/22
Refraction	-0.119	0.227	0.348	0.358
Axial length	0.108	0.314	0.434	0.492
Corneal power	0.138	0.378	0.542	0.093
Lens power	-0.288	0.155	0.278	0.553
Anterior chamber depth	-0.098	0.265	0.388	0.019

In the previously reported families, selected for unusual refraction in the parents or offspring, the coefficients of correlation for the refraction and its components set out in Table II (recalculated on the same basis as the material recorded here) show that with a high degree of correlation for refraction and axial length for the two parents, there is a high degree of correlation for refraction and all components except axial length between parents and children. In this series the small number of sibs precludes any worth-while analysis of relevant coefficients.

TABLE II

COEFFICIENTS OF CORRELATION FOR REFRACTION AND ITS COMPONENTS IN 21 FAMILIES WITH UNUSUAL REFRACTION IN EITHER THE PARENTS OR THE OFFSPRING

	Father/ Mother 21 pairs	Parent/ Child 56	Mid- Parent/ Offspring 28	Sib/Sib 7
Refraction	0.339	0.285	0.342	-0.104
Axial length	0.321	0.062	0.076	-0.122
Corneal power	-0.039	0.271	0.401	-0.289
Lens power	-0.099	0.416	0.580	0.349
Anterior chamber	0.111	0.323	0.412	0.615

Note: In this Table the second of each of two pairs of identical twins (families 2 and 8) have been omitted.

Discussion

The 28 unselected families included six families in which one or both parents had a refractive error outside the range of +4.00 to -4.00 D. With a larger number of families, these two groups of refraction might with advantage have been considered separately, but even with the present small numbers the only unexpected findings are the relatively low coefficients for the cornea and the anterior chamber in sibs, and for the lens in parent/child relationship. The available evidence, therefore, points to the strong probability that with larger numbers inheritance of refraction and its components in the general population is likely to follow the pattern set by a number of genes with additive effect.

The findings for the selected cases suggest that this also applies to the different components of refraction, other than the axial length which has characteristic features of its own. The study on twins has shown axial length to be genetically determined, while the high correlation coefficients

for the cornea and lens seen in the present study emphasize that the compensating mechanism of reduction in the powers of the cornea and lens are effective for variations in axial length within the range seen in emmetropia but not beyond. It is likely that—in contrast to the other components of refraction including axial lengths within the emmetropic range—abnormally long or short axial lengths are determined monofactorially, and it is fairly certain that there is more than one mode of inheritance of such abnormal axes. These are possibilities that need further exploration.

Summary

Twenty-eight unselected families with a total of 106 subjects were studied for ocular refraction and its components. In contrast to the lack of correlation between the parents, there was a substantial degree of correlation for parent/child and for mid-parent/offspring coefficients.

In 21 previously reported families selected for unusual refraction and recalculated on the same basis as the material recorded here, there was a high degree of correlation between parents and children for refraction and all components except axial length.

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REFERENCES

- Sorsby, A., Benjamin, B., Davey, J. B., Sheridan, M., and Tanner, J. M. (1957). Emmetropia and its aberrations. *Spec. Rep. Ser. med. Res. Coun. (Lond.)*, No. 293.
 —, —, and Sheridan, M. (1961). Refraction and its components during the growth of the eye. *ibid.*, No. 301.
 —, and Fraser, G. R. (1964). Statistical note on the components of ocular refraction in twins. *J. med. Genet.*, 1, 47.
 —, Sheridan, M., and Leary, G. A. (1962). Refraction and its components in twins. *Spec. Rep. Ser. med. Res. Coun. (Lond.)*, No. 303.

Appendix

Refraction and its components in 28 families with a total of 106 subjects

Family No.	Member	Age (yr.)	Ocular Refraction in Vertical Meridian (D)	Corneal Power in Vertical Meridian (D)	Anterior Chamber Depth (mm.)	Lens Equivalent Power (D)	Lens Power at Corneal Vertex (D)	Axial Length (mm.)
1	Father	56	R -1.48	43.7	3.2	21.7	12.8	24.2
			L -1.72	44.1	3.5	22.6	12.9	24.1
	Mother	55	R +1.26	41.4	3.5	19.5	11.9	24.5
			L +1.56	41.3	3.5	19.1	11.5	24.5
	Son	25	R -3.59	41.7	3.6	21.8	13.7	25.7
			L -4.27	41.7	3.6	22.8	14.4	25.7
	Daughter	24	R +0.25	42.3	3.9	19.7	11.6	24.6
			L +0.50	42.1	3.7	19.2	11.5	24.7
	Daughter	16	R +0.26	41.8	3.6	20.8	12.8	24.3
			L +0.50	41.7	3.6	20.8	12.7	24.3

Family No.	Member	Age (yr.)	Ocular Refraction in Vertical Meridian (D)	Corneal Power in Vertical Meridian (D)	Anterior Chamber Depth (mm.)	Lens Equivalent Power (D)	Lens Power at Corneal Vertex (D)	Axial Length (mm.)
2	Father	45	R +1.55	41.0	3.2	21.6	12.8	24.1
			L +1.55	41.1	3.3	20.9	12.5	24.2
	Mother	44	R +1.01	45.7	3.2	20.4	11.7	22.8
			L +0.76	44.8	3.3	20.5	11.7	23.3
	Son	24	R -0.50	41.6	4.0	18.3	10.6	25.8
L -0.74			41.7	4.1	18.5	10.0	26.2	
Son	17	R +0.25	43.8	3.8	17.5	10.2	24.6	
		L 0.00	44.1	3.8	18.3	10.7	24.3	
3	Father	50	R +2.24	43.6	3.0	23.0	13.4	22.5
			L +2.01	43.6	2.8	21.1	12.6	22.9
	Mother	46	R +3.66	44.5	2.4	22.9	14.0	21.5
			L +3.69	44.1	2.5	21.5	13.1	21.9
	Daughter	23	R -0.75	44.0	3.6	19.9	11.7	24.3
L -1.23			43.3	3.6	18.5	11.1	25.1	
Son	16	R +1.27	43.5	3.4	20.2	11.6	23.7	
		L +1.52	43.5	3.4	19.5	11.2	23.7	
4	Father	56	R +1.27	42.0	2.7	23.3	14.1	23.2
			L +0.70	42.2	2.9	22.2	13.6	23.6
	Mother	46	R +1.77	44.3	3.2	19.4	11.3	23.2
			L +1.52	44.2	3.2	21.2	12.1	23.0
	Daughter	17	R +4.22	43.7	4.0	17.7	10.0	23.0
L +5.39			43.7	3.7	17.8	10.0	22.6	
Son	16	R -0.25	42.3	3.8	19.3	11.4	24.9	
		L -0.50	42.4	3.5	18.2	11.0	25.2	
5	Father	42	R -3.61	41.6	2.9	19.4	12.6	26.4
			L -2.68	41.9	3.2	19.8	12.5	25.8
	Mother	43	R -1.25	43.3	3.6	22.6	12.9	24.3
			L -1.45	43.2	3.3	22.8	13.3	24.2
	Son	17	R +0.32	40.9	3.8	22.0	12.6	24.8
L +0.76			41.0	3.8	21.3	12.3	24.7	
6	Father	51	R -0.75	45.1	3.1	21.0	12.2	23.6
			L -0.75	44.7	3.1	21.4	12.4	23.7
	Mother	51	R +0.63	46.5	3.3	21.5	11.2	22.9
			L +0.54	47.0	3.6	20.7	10.7	22.9
	Daughter	17	R +0.50	46.3	3.5	19.2	10.5	23.3
L +0.76			46.2	3.4	20.2	11.4	22.9	
Son	16	R +1.01	44.3	3.5	19.4	10.9	23.7	
		L +1.03	44.8	3.4	20.4	11.6	23.2	
7	Father	50	R +0.82	43.4	3.6	22.7	12.8	23.4
			L +0.76	43.4	3.6	22.1	12.5	23.5
	Mother	51	R +8.07	43.2	2.0	25.0	15.5	20.0
			L +	—	—	—	—	—
	Daughter	21	R +3.65	43.4	3.5	23.4	13.1	22.2
L +1.27			43.3	3.5	23.2	13.5	23.0	
8	Father	50	R +0.76	43.0	3.0	19.5	11.2	24.3
			L +0.76	42.9	2.9	20.3	11.7	24.1
	Mother	44	R +0.01	41.4	3.4	20.3	12.0	24.5
			L +0.75	41.9	3.3	19.5	11.7	24.6
	Daughter	19	R +0.53	41.7	4.1	18.3	10.5	25.3
L +0.76			41.7	4.1	17.9	10.1	25.4	
Son	17	R +0.75	41.2	3.0	17.6	10.7	25.3	
		L +0.75	40.5	3.1	17.7	11.1	25.5	
9	Father	56	R	Traumatic cataract		—	—	—
			L +0.25	44.2	3.1	23.8	13.7	23.0
	Mother	45	R +0.78	42.3	3.0	20.5	12.0	24.2
			L +1.03	42.3	—	—	—	—
	Son	18	R +1.27	44.0	3.2	19.1	11.3	23.6
L +0.97			44.0	3.6	20.6	11.9	23.4	
Daughter	17	R +1.24	43.3	4.0	20.0	11.1	24.0	
		L +0.82	44.2	—	—	—	—	
10	Father	52	R +5.38	42.2	2.5	20.4	12.1	22.3
			L +6.55	41.9	—	—	—	—
	Mother	52	R -0.74	41.4	3.1	23.3	13.7	24.5
			L -0.74	41.7	3.2	24.1	13.9	24.3
	Son	26	R 0.00	42.5	3.5	22.3	12.6	24.2
L +0.25			42.7	3.5	21.4	12.3	24.1	
Son	24	R +3.09	41.9	3.9	22.5	12.4	23.2	
		L +3.11	41.9	3.5	21.1	12.0	23.4	
Daughter	16	R +3.65	43.3	3.1	25.7	14.5	21.7	
		L +3.65	44.1	3.2	26.4	14.7	21.4	

Family No.	Member	Age (yr.)	Ocular Refraction in Vertical Meridian (D)	Corneal Power in Vertical Meridian (D)	Anterior Chamber Depth (mm.)	Lens Equivalent Power (D)	Lens Power at Corneal Vertex (D)	Axial Length (mm.)
21	Father	49	R +6.14	41.2	2.8	21.3	12.0	22.5
	L +6.44		41.0	2.9	22.3	12.2	22.3	
	Mother	49	R +0.25	44.1	3.2	22.5	12.9	23.3
	L +0.26		45.2	3.2	23.6	13.5	22.6	
Son	21	R +1.27	42.0	3.4	19.1	11.3	24.4	
		L +1.78	42.1	3.4	19.1	11.4	24.1	
Daughter	17	R +1.78	44.1	3.5	19.5	11.2	23.4	
		L +2.04	44.3	3.6	19.6	11.2	23.2	
22	Father	49	R -0.50	43.1	2.6	21.9	13.4	23.8
	L -0.99		42.9	3.1	21.4	13.0	24.3	
	Mother	46	R +2.03	43.4	—	—	—	—
	L +1.77		44.1	3.2	20.7	12.1	23.0	
Son	18	R -1.95	42.3	3.7	18.4	11.2	25.9	
		L -1.95	41.9	3.8	18.6	11.4	26.0	
23	Father	49	R +1.00	42.0	2.9	23.2	14.1	23.4
	L +1.01		41.7	—	—	—	—	
	Mother	38	R -3.20	44.0	3.8	15.9	9.4	26.6
	L -3.20		44.4	3.6	16.3	9.7	26.2	
Daughter	18	R +0.02	44.4	3.5	20.3	11.6	23.8	
		L +0.53	45.6	3.2	19.9	11.5	23.1	
Son	16	R +1.01	43.7	3.5	19.2	11.2	23.9	
		L +0.75	43.7	3.3	18.0	10.9	24.1	
24	Father	44	R +0.51	41.2	3.0	20.5	12.4	24.7
	L +0.77		40.0	2.8	20.5	12.9	24.9	
	Mother	45	R +0.76	41.1	2.7	19.5	12.2	24.7
	L +0.76		41.1	2.7	19.9	12.5	24.5	
Son	19	R +1.27	40.8	3.5	20.9	12.4	24.5	
		L +1.01	40.8	3.5	19.9	12.0	24.8	
25	Father	53	R -0.99	43.1	3.0	21.8	13.0	24.2
	L -1.68		44.1	3.0	22.4	13.4	23.9	
	Mother	53	R +1.27	46.2	3.1	21.3	11.8	22.5
	L +1.01		46.4	3.0	21.5	11.9	22.5	
Daughter	19	R -1.46	44.8	3.5	18.4	11.0	24.6	
		L -1.45	44.7	3.5	19.5	11.6	24.3	
Son	18	R +0.76	42.0	3.5	20.4	12.0	24.4	
		L +1.01	42.2	3.5	20.3	12.0	24.1	
26	Father	48	R -1.22	42.8	2.8	22.0	13.8	24.1
	L -0.48		42.8	2.8	21.4	13.2	24.0	
	Mother	49	R +1.27	44.8	3.2	19.0	11.0	23.4
	L +1.27		44.8	3.3	19.7	11.0	23.4	
Son	18	R -4.06	42.1	3.5	21.9	13.7	25.8	
		L -3.10	41.7	3.5	21.3	13.2	25.7	
Son	16	R +1.03	45.2	3.6	20.1	11.3	23.2	
		L +1.29	45.2	—	—	—	—	
27	Father	50	R +1.27	44.6	2.7	21.1	13.0	22.6
	L +1.52		44.8	2.6	20.8	13.1	22.4	
	Mother	47	R +1.01	46.1	3.0	19.6	11.5	22.8
	L +1.01		46.3	3.0	19.7	11.6	22.6	
Daughter	19	R +1.01	46.0	3.2	21.4	12.2	22.5	
		L +1.01	46.0	3.2	21.3	12.3	22.5	
Son	16	R +1.01	45.6	3.8	21.2	11.4	23.0	
		L +1.01	46.3	3.7	21.8	12.0	22.5	
28	Father	49	R +0.30	43.0	2.9	19.9	11.9	24.1
	L +1.29		42.8	2.8	19.3	11.3	24.1	
	Mother	40	R -2.84	42.7	3.1	21.2	12.8	25.3
	L -2.18		42.4	3.4	19.9	12.0	25.5	
Daughter	18	R -1.23	44.2	3.5	19.3	11.4	24.5	
		L -0.74	44.2	3.6	20.1	11.7	24.2	