

GENETIC FACTORS IN MYOPIA

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A school population has been screened to locate same-sexed twins with myopia and also to compare intelligence test performance of myopic and nonmyopic individuals. Augmentation of the twin data by a survey of the world literature has led to the identification of a total of 106 MZ twin pairs, 100 of them concordant for myopia, as well as 41 DZ pairs, 12 concordant. Myopic students score eight points higher on IQ tests than nonmyopes, the entire bell shaped distribution curve being shifted to a higher range. The intellectual gain precedes in time the development of nearsightedness.

In the course of an investigation into the nature of the well documented relationship between myopia and giftedness (Karlsson 1973), the question of the evidence for genetic factors has come up for reevaluation. There has always been considerable interest in possible external causes of myopia, the condition being attributed to excessive near work, eye strain, etc. (cf. Waardenburg 1963). Recent reports of an apparent increase in nearsightedness in the younger generations of Eskimos and North-Canadian Indians (Morgan and Munro 1973) have given renewed impetus to efforts to unravel the underlying mechanisms. Some investigators are attempting to produce myopia in monkeys through environmental manipulations (Young 1961).

A tendency for family concentration of nearsightedness is recognized, and even in the studies of Eskimos this is quite apparent. Differences are also reported between ethnic groups, e.g., between Negroes and Caucasians in the United States. Some industrialized nations have very high rates, the frequency of myopia being in the order of 25% among those of Jewish, Japanese, or West-European origin, while primitive populations show low rates.

The present investigation was undertaken to augment the data on myopic twins and make a more definitive comparison of the performance of myopic and nonmyopic individuals on intelligence tests.

METHODS AND RESULTS

This study was conducted at the high school in Napa, California, where approximately 800 students matriculate each year, representing the majority of 18-year-old individuals in a community of 40,000 inhabitants. The student health service has sufficient information to make it possible to identify most of the myopic pupils. Scores on IQ tests administered at different ages are also available.

Same-sexed twins with myopia were located by the use of the collection of individual photographs printed annually in the school year-book. Students wearing eye glasses who shared a surname with a classmate were identified, and to ascertain whether they were twins a check was made of their birth dates and addresses. The school health records were consulted to determine whether myopia was present, and the parents were contacted where possible to obtain their opinion about the degree of resemblance of the twins. The 9 pairs identified in this manner were classified into 7 MZ sets, all concordant for myopia, and 2 DZ sets, both discordant. Those considered uniovular showed very striking similarities, while the biovular pairs exhibited marked differences. The twin data were augmented by a survey of the world literature which led to the identification of additional 99 MZ pairs with myopia as well as 39 DZ pairs.

The overall results are summarized in Table 1, further details and references having been published elsewhere (Karlsson 1974). The twins included in the table have an index case requiring a bilateral correction of one diopter or above, a pair being considered concordant if any degree of myopia is also present in the cotwin.

Table 1. *Concordance for myopia in twins as reported by various investigators*

| | MZ pairs | | DZ pairs | |
|------------------------------------|----------|------------|----------|------------|
| | N | Concordant | N | Concordant |
| Individual case reports | 21 | 19 | 2 | 2 |
| Series with 1-3 myopic cases each | 28 | 27 | 4 | 1 |
| Series with 7-17 myopic cases each | 50 | 47 | 33 | 9 |
| Present series | 7 | 7 | 2 | 0 |
| Total | 106 | 100 | 41 | 12 |

In the second phase of the study, recent intelligence test scores were obtained on all graduating seniors for the years 1970, 1971, and 1972. A breakdown of the data is presented in Table 2. In the population under study the average IQ is 106, being 105 for all nonmyopes and 113 for the myopic students. The rate of myopia in different IQ groups was also ascertained, showing the condition to be absent in the 30 students in the lowest range, IQ 55-74. In successive ranges the rate of the disorder rises systematically, the highest rate, 44.4%, being encountered in the group with IQ above 134, comprising 45 students. This contrasts with the average myopia rate of 14.9 for the entire population of 2527 students.

Since the school records contain IQ test results obtained at different age levels, the scores achieved by the myopes and nonmyopes ten years earlier were compared to the result at age 17-18 years. The distributions were found to be essentially identical, the myope group having at age approximately eight years already attained elevated scores.

Table 2. *Scores attained by 17-18 year-old pupils on the Lorge-Thorndike group IQ test administered at the Napa High School in 1970-1972*

| | N | Mean IQ | SD of IQ curve | SE of mean IQ |
|--------------------|------|---------|----------------|---------------|
| All students | 2527 | 106 | 14.1 | 0.3 |
| Males | 1272 | 107 | 14.6 | 0.4 |
| Females | 1255 | 105 | 13.5 | 0.4 |
| Non-myopic males | 1095 | 106 | 14.4 | 0.4 |
| Myopic males | 177 | 114 | 13.8 | 1.0 |
| Non-myopic females | 1055 | 104 | 13.2 | 0.4 |
| Myopic females | 200 | 111 | 13.9 | 1.0 |
| All non-myopes | 2150 | 105 | 13.8 | 0.3 |
| All myopes | 377 | 113 | 13.2 | 0.7 |

DISCUSSION

It is apparent from the data in Table 1 that MZ twins generally tend to be concordant with respect to myopia. The actual concordance rate is 94% according to the pair method and 97% if the proband method of computation is employed. Most of the discordant MZ pairs are composed of mildly myopic index cases (below two diopters) with essentially emmetropic partners. If one limits the consideration to the 24 twin pairs from large and carefully documented series in which the myopia of the index case

is at least two diopters, all are concordant. One thoroughly investigated individual case report of MZ twins shows definite discordance for severe myopia (Orth 1954). The overall findings, however, give strong support to hereditary factors.

While twin data basically provide little information about genetic mechanisms, the rate observed in the DZ twins in the present study is consistent with the expectation on the basis of recessive inheritance, this being the favored genetic system for myopia (Furusho 1957, Karlsson 1975). Reports on the offspring of two myopes also show very high rates of nearsightedness. Theoretically that rate should approximate 100% if the recessive hypothesis is correct, but the available reports often include young children, and in some families one parent has a mild form of myopia. It has therefore not been fully established whether essentially all children of two definite myopes (— 2D or above bilaterally) eventually become themselves myopic, although some evidence supports this view (Karlsson 1975). The possibility of somewhat incomplete penetrance thus remains an open question, and certainly it appears that there is an overlap in the region of mild myopia between the emmetropic and myopic populations (Furusho 1957).

The main interest in the present study is in the relationship of myopia to giftedness, which in reality has been quite well documented (Karlsson 1973). Since the twin evidence gives further weight to heredity as the main cause of myopia, it lends support to the view that there is a myopia gene which exerts its principal effect on the brain. The eye effect may then be viewed as a secondary complication, which however reveals the presence of the gene. The present study indicates that myopes score approximately eight points higher than nonmyopes on IQ tests, the configuration of the distribution curves for the two groups being essentially identical, except that the entire bell shaped curve for myopes is shifted to a higher range.

Of special interest is the fact that the IQ scores of the two populations are already different on tests administered ten years earlier, i.e., before nearsightedness generally becomes manifest. If the intelligence gain precedes in time the development of nearsightedness, the hypothesis is untenable that test advantages are derived from myopic vision or from confinement of a handicapped person to reading activities.

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