Populations of Hybrid Origin as Source Material for the Detection of Linkage

DAVID C. RIFE

Institute of Genetics and Department of Zoology, Ohio State University, Columbus 10, Ohio

A CORRELATION in the occurrence of two genetic traits within a population is usually interpreted as evidence against, rather than for linkage. Linkage results in correlations within families, but in opposite types of correlation from one family to another, the type of correlation within a particular family depending upon whether most of the chromosomes are in the coupling or repulsion phase. Thus opposite types of correlation within different families cancel correlations within the population as a whole.

This corollary is derived from the observation that mutations are recurrent and that they appear to occur independently. Regardless of whether linked or not, genes at different loci tend to occur independently of each other within panmictic populations. The fact that the Rh antigens, C, D, and E do not occur independently of each other (Rife, 1948) casts doubt on Fisher's belief that they are linked (Fisher, 1947) unless they are so closely linked that crossing over never occurs. Pleiotropy provides a reasonable interpretation of consistent correlations between genetic traits in all population. If Wiener (1946) is correct in assuming that a single series of multiple alleles is responsible for the Rh variations, some members of the series are pleiotropic, as they produce two or more antigens.

Relatively new populations of hybrid origin provide an exception to the foregoing rule. Under specific circumstances correlations within them may be indicative of linkage. This paper is concerned with the nature of these circumstances, and the report of an investigation of two hybrid populations which revealed associations suggestive of autosomal linkage.

CORRELATIONS WITHIN HYBRID POPULATIONS

Members of an allelic series attain genetic equilibrium with only a single generation of random mating. (Hardy, 1908, Weinberg, 1908). This does not hold true for the relationships between the members of different sets of alleles. (Robins, 1918, Haldane, 1925, Li, 1948). The rate at which equilibrium is approached depends solely upon the percentage of crossing over. Within a hybrid population the deviation from equilibrium is reduced by half each generation if the loci are not linked, or if linked by the amount of crossing over between the two loci. The initial degree of deviation from equilibrium depends

Received July 25, 1953.

upon relative sizes of the parent populations, and the differences in frequencies of the genes under consideration. In situations where the parent populations are of opposite homozygous genotypes ($AABB \times aabb$, or $AAbb \times aaBB$) the rate of approach to equilibrium may be readily calculated. The proportion of recombination gametes in any particular generation $= 1 - [x^2 + y^2 + 2xy(1-c)^n]$ where x and y represent the relative proportions of the two parent populations, c = percentage of crossovers, and n = number of generations since hybridization. Figure 1 illustrates the rate of equilibration for six generations where x and y each = .5; for no linkage (50% crossovers), 20, 10, and 5% crossovers. The deviations from equilibrium are represented by the heights of the lines above the base.

Non-linked genes approximate equilibrium after six generations, whereas those linked with 5% crossovers show only about 10% recombinations over the same period. Over a hundred generations would be required for genes linked with 1% crossovers to approximate equilibrium. It is evident that closely linked genes may manifest correlations within hybrid population, many generations after non-linked genes have attained approximate equilibrium. Thus if a marked correlation is apparent between two genetic traits within a hybrid population, whereas no correlation exists between a third genetic trait and either of the other two, the correlation strongly suggests that the first two traits are conditioned by linked genes, while the third is probably due to genes

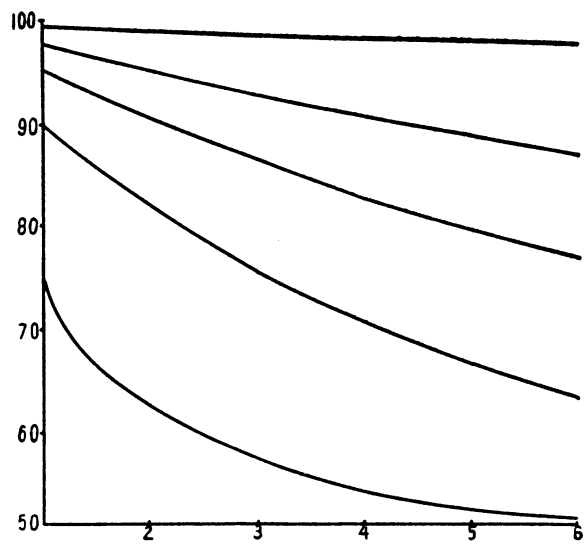

FIG. 1. Graphic representation of the rate at which two pairs of alleles approach genetic equilibrium over a period of six generations, in a population of hybrid origin. The percentages of non-recombination gametes are represented on the vertical axis, the number of generations on the horizontal axis. Rates of equilibration are shown for no linkage, and linkage with 20%, 10%, 5%, and 1% crossovers. The parent populations were of equal size (x = .5, y = .5).

on another chromosome. Pleiotropy may also result in a correlation in the incidence of genetic traits, but if this be the cause of the association, we should expect similar correlations in all populations, whereas linkage would result in no correlation within stable populations, or perhaps the opposite type of correlation within other hybrid populations.

If certain conditions are met a correlation between two out of three genetic traits may be considered as evidence for autosomal linkage. They are as follows: the population in which the correlation occurs must be of hybrid origin, the parent populations must have been characterized by marked differences in the frequencies of all of the genes under consideration, at least one of the traits showing the correlation must be of no selective importance; and the correlation must be absent in old established populations. It is difficult to conceive of any cause, aside from linkage, which would satisfy these conditions.

THE HYBRID POPULATIONS AND THEIR ORIGINS

Two populations of hybrid origin were investigated, both of which are of Negro-Caucasian descent. One population consisted of 100 northern Sudanese, the other of American Negroes.

The northern Sudanese are principally a mixture of Arabs and Negroes with some admixture of Hamitic peoples. Arabs have migrated to the northern Sudan from Arabia across the Red Sea and from Egypt as far back as the seventh century A.D. Until recent times they captured Negroes from the southern Sudan and other regions for slaves, and frequently married them. Although the northern Sudanese of today are Mohammedans and are Arabic in culture, evidences of considerable Negro ancestry are readily apparent.

Data were obtained from students at the Egyptian secondary school in Khartoum.[1] These students come from all over the northern Sudan, and would seem to provide a wide sample of unrelated northern Sudanese. All of the students are male.

The data on American Negroes were obtained over a period of years from students at the Ohio State University. Although 99 individuals have been included in the study, complete data for the present investigation were available for only 35 of them. Both males and females are included. American Negroes are essentially of West African Negro and northwestern European or British descent.

Although both northern Sudanese and American Negroes are of mixed Negro-Caucasian descent, the Negro and White ancestors of each are somewhat different. The Negro ancestors of the northern Sudanese were presumably

[1] The data were collected during February, 1952, by an expedition consisting of Mr. Martin Johnson, Mr. Robert Murphy, Dr. and Mrs. E. T. Bullard, Mr. Rafia Bouymi, and the author. Grateful acknowledgement is due Dr. Roha, supervisor of Egyptian education in the Sudan, for his cooperation in the project.

Nilotes and other Sudanese for the most part, whereas the ancestors of the American Negroes were largely West African. The White ancestors of the Sudanese were mostly semitic (Arab), whereas those of the American Negroes were northwestern European and British.

TYPES AND COLLECTION OF DATA

Data collected from the northern Sudanese include kodachrome photographs of heads, taste reaction to phenylthio-carbamide, and hand prints.

The photographs were taken out-of-doors in the shade, over a two hour period. Conditions were maintained as uniform as possible throughout the procedure. One member of the expedition recorded his judgment as to shade of pigmentation for each individual. The author classified the photographs as to "light" and "dark" according to his judgment, before referring to the observations noted by the observer at the time the photographs were taken. Agreement in judgment was noted in 95% of cases. The observer who recorded his judgments used 5 shades, and the author found that his "darks" correspond to the two darkest shades, and his "lights" to the three lightest shades recorded by the observer. The observer obtained the following distribution beginning with the darkest shade: 23, 17, 53, 6, 1. None of the darkest shade are as dark as the pictures of over 300 Nilotic Negroes taken at a later date, and only the one in the extreme light class could be assumed to be of unmixed White ancestry with reasonable assurance. All of the subjects possessed black or very dark brown hair, ranging from curly to woolly in form. The "dark" northern Sudanese referred to henceforth include the two darkest classes of the observer, the "light" Sudanese to the three lightest classes, a total 40 "dark", and 60 "light" Sudanese.

Taste reaction to phenylthio-carbamide was determined by placing a small amount of the substance on the back of the tongue and noting whether the subject detected a distinct taste after a minute. Prints were obtained of entire palms and the fingertips. The taste reactions and hand prints were also obtained for American Negroes.

Black and white photographs of both front and profile head views were available for 44 American Negroes. These were taken under uniform conditions by the Department of Photography of the Ohio State University. The photographs were arranged independently into "dark," "light," and "doubtful" by four individuals who were unfamiliar with the people who had been photographed. Agreement was reached on 19 "dark," 16 "light," and 9 "doubtful."

Skin pigmentation, dermatoglyphics, and tasting ability fulfill the requirements for testing for linkage in hybrid populations. Each show marked differences in their frequencies in the parent populations, and neither dermatoglyphics or tasting ability are of any selective importance. The three traits occur independently of each other in panmictic populations. Tasting ability

appears to be determined by a single pair of alleles, dermatoglyphics include several types of variations due to multiple genes, and the differences in pigmentation between Negroes and Caucasians appear to be due to at least two pairs of alleles (Davenport, 1913).

Data from various investigators indicate that most African Negroes have significantly higher frequencies of tasters than do Caucasians (Lee, 1934, Parr 1934, Rife, 1953a).

Palmar dermatoglyphics likewise occur with different frequencies among Negroes and Caucasians, Negroes being characterized by higher frequencies of patterns in the second and fourth interdigital areas, and lower frequencies in the third interdigital and hypothenar areas (Pons, 1952).

The incidences of whorls on fingertips, however, do not manifest as clear cut differences between Negroes and Caucasians in general, as they do among both Negro and Caucasian populations. Some West African Negroes have approximately 40% whorls, whereas Nilotic Negroes possess between 25% and 30% whorls. Among Caucasians northwestern Europeans show 25% to 30% whorls, whereas Middle Eastern peoples show approximately 40% whorls (Rife, 1953b).

The ABO blood groups occur with somewhat similar frequencies among African Negroes, northern Sudanese, Ethiopians, and Bedouins (Rife, 1953a). Data on associations between blood groups and pigmentation in northern Sudanese would thus shed little light on linkage relationships and were not included in this investigation. The situation is different among American Negroes, as West Africans and White Americans show highly significant differences (table 5). Group A occurs approximately twice as frequently among Whites as among Negroes, whereas B occurs approximately twice as frequently among African Negroes as among American Whites.

RESULTS

Tests were made for associations between the occurrence of patterns on each of the five palmar areas with pigmentation, with ability to taste, and of pigmentation with tasting ability. The incidence of patterns in the second interdigital area revealed a highly significant association with pigmentation the "dark" showing much higher frequency of patterns than "light" (table 1).

TABLE 1. ASSOCIATIONS BETWEEN PIGMENTATION AND PALM PATTERNS IN SECOND INTERDIGITAL AREA

	DARK		LIGHT	
	Pattern	No pattern	Pattern	No pattern
100 Northern Sudanese	11	29	4	56
35 American Negroes	3	16	0	16
Total	14	45	4	72

$X^2 = 9.46$, df = 1, $p < .01$

TABLE 2. ASSOCIATIONS BETWEEN PIGMENTATION AND ABILITY TO TASTE PHENYLTHIOCARBAMIDE

	DARK		LIGHT	
	Tasters	Non-tasters	Tasters	Non-tasters
100 Northern Sudanese	38	2	56	4
27 American Negroes	12	5	10	0
Total	50	7	66	4

$X^2 = 1.93$, df = 1, p between .70 and .50

The higher incidence of patterns among "dark" is apparent among both Sudanese and American Negroes. No significant associations were found between pattern occurrence and tasting ability, or between tasting ability and pigmentation (tables 2 and 3). The high association between patterns in the second interdigital area and dark pigmentation, and the absence of associations between taste and either pattern of pigmentation strongly suggests linkage between genes responsible for patterns and pigmentation. Pleiotropy seems to be ruled out, as patterns occur among both parent populations, although with greater frequencies among Negroes.

No significant correlations were found between pigmentation or tasting ability and the occurence of patterns in any of the other areas. Whorls on fingertips, however, show a highly significant correlation with pigmentation (table 4). The reason for this association is a bit puzzling, especially among northern Sudanese, for if it is due to linkage one might expect "light" to have higher frequencies of whorls than "darks" in view of the fact that southern Sudanese Negroes (Nilotes) have much lower whorl frequencies than do Middle Eastern peoples, or even northern Sudanese. There are two possible explanations for the association. First, perhaps Sudanese Negroes in the south western Sudan, whose dermatoglyphics have not been investigated, may possess a high frequency of whorls. They doubtless have constituted a sizable proportion of the Negro parent population. Second, there is a positive correlation between the occurrence of whorls on fingertips and patterns in the second interdigital area in various populations, suggesting one or more pleiotropic genes affecting

TABLE 3. ASSOCIATIONS BETWEEN ABILITY TO TASTE PHENYLTHIOCARBAMIDE AND PATTERNS IN SECOND INTERDIGITAL AREA

	PATTERNS		NO PATTERNS	
	Tasters	Non-tasters	Tasters	Non-tasters
100 Northern Sudanese	15	0	79	6
77 American Negroes	9	2	57	9
Total	24	2	136	15

$X^2 = 0.11$, df = 1, p between .95 and .70

TABLE 4. ASSOCIATIONS BETWEEN WHORLS AND PIGMENTATION

	DARK		LIGHT	
	Whorls	No whorls	Whorls	No whorls
34 American Negroes	70	120	35	115
100 Northern Sudanese	211	189	276	324
Total	281	309	284	466

$X^2 = 11.77$, df = 1, $p < .01$

both whorls on fingertips and patterns in the second interdigital area (Rife, 1943). We should expect whorls to be associated with the same traits as second interdigital patterns, if this be true. The association between whorls and patterns among American Negroes is in the direction one might expect, as many West African Negro populations are characterized by higher whorl frequencies than are North American Whites.

Unfortunately, the number of individuals tested for associations between blood groups and pigmentation among American Negroes is too small to be of any significance. An association is suggested, however, by the absence of any group B among "lights," and the similar percentages of A and B among "darks." It should be kept in mind that American Negroes are a relatively newer hybrid population than northern Sudanese, and that scarcely enough time has elapsed to eliminate all associations even between non-linked genes.

If linkage is indeed the reason for the association between second interdigital patterns and pigmentation, it suggests that two pairs of genes may be responsible for differences between Negroes and Whites, as proposed by Davenport (1913). If three or more independent pairs are concerned, it is difficult to see how linkage between only one pair with patterns could produce such an obvious association. There is some evidence that the presence of a pattern in the second interdigital area may be due to a simple dominant gene, lacking complete penetrance.

A collection of the palmar dermatoglyphics of 20 White American families assembled by the author reveals that among 15 families in which both parents lacked a pattern, only 3 out of 87 children possessed the pattern. All three occurred in the same family, in which 6 other children lacked the pattern.

TABLE 5. ABO BLOOD DISTRIBUTIONS

	%O	%A	%B	%AB
325 West African Negroes	52.30	21.50	23.00	3.20
20,000 Whites, U.S.A.	45.00	41.00	10.00	4.00
87 American Negroes	54.02	26.43	17.24	2.29
19 "dark" Negroes	52.63	21.05	26.31	0.00
16 "light" Negroes	56.25	42.75	00.00	0.00

Among 5 families in which one parent possesses the pattern, 11 out of 29 children manifest it. It occurs among the children in 3 of the families.

I wish to emphasize that the association between hand patterns and skin pigmentation suggests linkage, but does not necessarily prove it. Data from mixed Negro-White families should enable one to establish or reject linkage as the reason for the association. Linkage appears to be the most likely explanation, however.

Hybrid populations provide a virtually untapped reservoir of material which could be used to good advantage for the detection of linkage. They could be of special importance in tackling the problem as to whether or not ethnic groups differ in mental capacities, providing valid tests are used and emotional biases are not allowed to interfere with objective evaluations.

SUMMARY

1. Association between two genetic traits, and the absence of associations of either with a third genetic trait within hybrid populations, may under specified conditions be indicative of linkage.

2. Associations between hand patterns and skin pigmentation indicative of linkage were observed among northern Sudanese and American Negroes.

REFERENCES

DAVENPORT, C. B. 1913. *Heredity of skin color in negro-white crosses.* Washington: Carnegie Inst. Pub. 188.

FISHER, R. A. 1947. The Rhesus factor. *American Scientist* 35: 92–100, 113.

HALDANE, J. B. S. 1925. A mathematical theory of natural and artificial selection. Part III. *Proc. Comb. Phil. Soc.* 23: 363–372.

HARDY, G. H. 1908. Mendelian proportions in a mixed population. *Sc.* 28: 49–50.

LEE, B. F. 1934. A genetic analysis of taste deficiency in the American negro. *Ohio Jour. Sc.* 34: 337–342.

LI, C. C. 1948. An introduction to population genetics. *National Peking University Press.* Peiping, China.

PARR, L. W. 1934. Taste blindness and race. *Jour. Hered.* 25: 187–190.

PONS, JOSE. 1952. Impresiones dermapapilares en estudiantes Universitarios Barceloneses. Trabajos del Instituto Bernardina de Shagun de anthropologia y etnologia. XIII No. 2: 7–131.

RIFE, D. C. 1953a. An investigation of genetic variability among Sudanese. *Am. Jour. Phys. Anthrop.*, n.s. V. 11, No. 2.

————. 1953b. Fingerprints as criteria of ethnic relationship. *Am. Jour. Hum. Gen.* 5: No. 4, 389–399.

————. 1943. Genetic interrelationships of dermatoglyphics and functional handedness, *Genetics* 28: 41–48.

————. 1948. A chromosome frequency test for linkage of the Rhesus blood factor. *Ohio Jour. Sc.* XLVIII. No. 3, pp. 116–118.

ROBBINS, R. R. 1918. Some applications of mathematics to breeding problems III. *Genetics* 3: 375–389.

WIENER, A. S. AND E. B. SOHN. 1946. *Annals of the New York Academy of Sciences* XLVI; 883–892.