GENETICS AND RACE

PART II

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GENETIC MARKERS AND RACE HISTORY

One of the uses to which genetic markers can be put is the elucidation of problems of race history. To illustrate this, two examples have been chosen: the relatively simple problem of the origin of the Icelanders and the more complex matter of the racial history of the Jews.

Iceland and the Vikings

For a long time, historians have debated the origin of the Icelanders. Was Iceland colonized by the Vikings from Norway or by settlers who came from Ireland? The evidence of the ABO blood group gene frequencies may shed some light on this problem (Table IV).

TABLE IV. FREQUENCY OF ABO BLOOD GROUP GENES IN NORWAY, ICELAND AND THE BRITISH ISLES (PERCENT)

	A	*B*	*O*
Norway	31	6	63
Iceland	18	7	76
Ireland	17	7	76
England	25	5	69

It will be seen that the gene frequencies of Icelanders are very similar to those in Ireland but quite different from those in Norway. The genetic data supports the hypothesis that only a small number of Icelanders came from Norway—probably just the rulers—and that the majority are descended from Irish immigrants.

The Racial History of the Jews

There were 3 main waves of emigration of Jews from Israel in antiquity:

1. In 586 B.C.E., after the destruction of the first temple by Nebuchadrezzar, large numbers of Jews were exiled to Babylon. Many returned after the Babylonian empire was overthrown by the Persians. However, large numbers remained in Mesopotamia. The modern Jewish communities of Iraq and Persia are the descendants of these early exiles. They are referred to as the *Oriental Jews.*

2. In about 334 B.C.E., after the Alexandrian conquest, many Jews emigrated to other parts of the Hellenic empire, particularly to North Africa and the Balkans. The descendants of these emigrants are known as *Sephardi Jews.*

3. In about 70 C.E., after the destruction of the second temple by Titus's Romans, another large-scale exodus of Jews from Israel was enforced. These exiles migrated to various parts of the Roman empire—chiefly to Italy and to Spain. The descendants of those who went to Spain were expelled in 1492 and the majority joined the Sephardic communities of North Africa and the Balkans. The Jews of Italy spread with the conquering Romans all over North-Western Europe—to Germany and France and later to Poland and Russia. Subsequent migrations led to their still further dissemination and the modern Jewish communities of America, Britain and South Africa are their descendants. They are referred to as the *Ashkenazi Jews.*

During these 2,500 years of dispersion, the various Jewish communities tended to lose contact with each other. Quite substantial differences in language and culture were inevitable, as was a certain degree of outbreeding with their Gentile neighbours. Particularly in North Africa and in the Orient Jews became almost indistinguishable physically from the Gentiles among whom they lived.

Then, during the last two decades, considerable numbers of Oriental, Sephardi and Ashkenazi Jews have been returning to Israel. Geneticists have examined many hundreds of them and have made full use of this unique opportunity for testing the effect of more than 2,000 years of dispersion on their genotype. Some of the findings are presented in Table V.

TABLE V. THE DISTRIBUTION OF CERTAIN GENETIC MARKERS IN 3 JEWISH COMMUNITIES IN ISRAEL AND IN NON-JEWS FROM NORTH-WEST EUROPE (PERCENT)

	G6PD deficient	*Rhesus positive*	*B gene*	*PTC tasters*	*Hp^1 gene*	*Finger-print pattern index*
Oriental	25	95	18	84	29	14·0
Sephardi	2	90	16	85	28	13·7
Ashkenazi	0·2	91	12	79	30	13·7
Non-Jews from N.W. Europe	0·2	85	6	70	40	12·2

It will be seen that in respect of certain traits—particularly G6PD deficiency—there are considerable differences between the three Jewish communities. The high frequency of G6PD deficiency in Oriental Jews is typical of Eastern Mediterranean communities; the Sephardi Jews are only slightly affected; the Ashkenazi Jews resemble their Gentile neighbours in being almost free from G6PD deficiency.

In other respects—rhesus positivity, B gene frequency and PTC tasting—the three Jewish communities are more similar. The Ashkenazi Jews still show some resemblance to their Gentile neighbours, but the Oriental character of their gene frequencies is quite obvious.

Finally, in respect of the Hp^1 gene frequency and the finger-print pattern index, the three Jewish communities show a remarkable degree of homogeneity. Even the Ashkenazi Jews show no sign of their long sojourn in the West.

The data from this remarkable natural experiment in population genetics are not yet complete, and at this stage only tentative conclusions can be drawn. From the sociological and anthropological points of view it seemed that the dispersion of the different Jewish communities had resulted in such a great deal of variability that it was no longer possible to group all the Jews into a single race. The genetical studies, however, have shown that this is not so. Widely separated Jewish communities still resemble each other so closely in the frequency of several of their genes that from the biological point of view they can be regarded as constituting a single racial unit.

THE PROCESS OF RACE FORMATION

Until quite recently, the process of the formation of the races of man could only be studied rather inadequately by the methods of social and physical anthropology, or by theoretical analogy from observations made on experimental plant and animal populations. But, as more and more data are being assembled on gene frequencies in human populations, it is becoming possible to approach the problem of the formation of human races more directly. What follows should be regarded as a tentative and highly simplified attempt at a synthesis from the data already available.

Phase I

(*a*) *Migration.* A number of families or individuals leave the parent population and migrate to a new territory.

(*b*) *Isolation.* The migrant group becomes cut off from the parent population by geographical barriers—mountain ranges, sea, etc. The migrant group will no longer be subjected to whatever influences are changing the genetic pattern of the parent group.

For example, we may consider the Lapps (Table VI). The progenitors of the present-day Lapps were presumably

TABLE VI. THE FREQUENCY OF SOME GENETIC MARKERS IN NORTH-WEST EUROPEANS AND IN LAPPS (PERCENT)

Blood group genes	*N.W. Europeans*	*Lapps*
O	64	61
A_1	22	4
A_2	7	32
B	7	2
Rh-	15	7
M	55	39
Fy^a	41	55
PTC non-tasters	32	6

genetically typical members of the North-Western European community who migrated to the remote North of the continent in antiquity. Since then they have been more or less unaffected by the biological processes which have modified the genetic constitution of the parent stock. Two major genetic changes have occurred in the North-Western Europeans since the migration of the Lapps. Firstly, there has been a spread of the rhesus negative gene from its origin in the Basque territory of Northern Spain. The Lapps being farthest North have been less influenced by this spread: only 7% are rhesus negative compared with 15% in other North-West European populations. Secondly, the Mongolian invasions from the East during the 12th and 13th centuries probably introduced the blood group B gene into Europe. The frequency of this gene is highest in Central Asia (25 - 30%) but becomes progressively less as one moves westward across Europe. The average frequency of the B gene in North-West Europe is now about 7%. Again, the intrusion of this gene has hardly affected the Lapps, only 2% of whom carry the B gene. The data in Table VI show how, as a result of their isolation, the Lapps now present a pattern of gene frequencies which in several respects is strikingly different from that of other North-Western European peoples.

Phase II

(*a*) *Selection*

Some members of the migrant group may carry genes which are better suited to their new homeland. Those who carry these genes will adapt more readily than their fellows to the new environment. Hence, they will have an inherent biological advantage which will be transmitted to their offspring. Those who carry these valuable genes will breed more prolifically than those who are at a biological disadvantage. Eventually, the disadvantageous genes will die out more or less completely, and a new 'race' will emerge characterized by a high frequency of the advantageous genes.

A familiar example of the effects of selection is *skin colour*. Pigmented skin resists the penetration of 'actinic rays' (ultraviolet light and heat). In a tropical climate, this is an advantage because blistering of the skin is prevented, skin cancer is rare and the body temperature is kept at a healthy level. Even in the subtropical climate of South Africa, skin pigmentation ensures a remarkable degree of protection against skin cancer (Table VII). Before the introduction of suitable tropical clothing for white-skinned people, pigmented individuals enjoyed a considerable biological advantage in hot climates. On the other hand, in temperate climates pigmented skin is a disadvantage: heat is lost and rickets is common because ultraviolet light cannot penetrate.

TABLE VII. SKIN CANCER : RACIAL INCIDENCE

	Total cases	*Whites*	*Cape Coloureds*	*Ratio Whites : Coloureds*
Rodent ulcer	857	804	29	27:1
Squamous carcinoma	394	362	32	11:1

[From: T. Schrire—Groote Schuur Hospital, 1949 - 1957]

Another important example of selection is provided by the *sickle-cell trait*. Those who are homozygous for the gene for the abnormal haemoglobin S (HbS) suffer from a severe haemolytic anaemia (sickle-cell anaemia) and seldom survive to produce offspring. Yet in several African, Mediterranean and Asiatic populations there are a great many apparently healthy heterozygote carriers of HbS. The gene frequency of HbS in some of these populations may be as high as 30%. Why has this harmful gene not died out? The only reasonable explanation is that there must be something useful about the HbS gene which has ensured its persistence in these communities: there must be some advantage to the heterozygote carriers of the HbS gene which balances the disadvantage of often producing children with sickle-cell anaemia. But what is this advantage?

The solution of this problem began with the observation that the geographical distribution of the HbS gene was similar to that of the distribution of malaria in the early part of this century. Next, it was shown that in malarious areas, the parasites are rarely found in the blood of HbS carriers. It was postulated that the presence of the HbS gene provided a high degree of resistance to malarial infection. This has been confirmed both experimentally and clinically. If HbS carriers and controls are deliberately exposed to the bites of malaria-bearing mosquitoes, the carriers are rarely infected but the controls usually are. In the hospitals of tropical Africa the severe forms of malaria (cerebral malaria and blackwater fever) are relatively common in the general population, but are rarely encountered in HbS carriers. In several African territories the death rate from malaria in known HbS carriers is less than 5% of the malaria death rate in the general population.

Thus, the gene for HbS provides a selective advantage over the gene for normal haemoglobin (HbA) in malarious areas, and despite its other disadvantageous properties it manages to persist at a high frequency. This phenomenon is an example of a 'balanced polymorphism.'

(*b*) *Mutation*

From time to time new genes will appear in the migrant group as a result of spontaneous mutation. Most mutations are harmful and will quickly die out. However, a mutation occasionally arises which will be useful in the new environment and confers a selective advantage on its carriers. In the course of many generations this mutation may become widely established, and it will then be a characteristic feature of the new race.

For example, *steatopygia* in Bushwomen is regarded by many to have developed in this way. Some believe that the fatty deposits in the buttocks provide a valuable store of food from which Bushwomen can draw nourishment in times of drought or to satisfy their increased nutritional requirements in pregnancy. Others believe that the selective advantage is aesthetic: Bushmen are attracted to women with steatopygia and the bigger her buttocks the more sought after will a Bushwoman be as a mate.

Another genetic trait which seems to have arisen by mutation and for which more quantitative data are available is the rhesus blood group variant *cDe* (*Ro*). It will be seen from Table VIII that this variant occurs chiefly in

TABLE VIII. THE FREQUENCY OF THE RHESUS BLOOD GROUP VARIANT cDe (Ro) IN VARIOUS POPULATION SAMPLES (PERCENT)

Africa	
Nilotes (Luo)	87
Eastern Bantu (Kikuyu)	71
Southern Bantu (Johannesburg)	65
Negroes (Ashanti)	62
Hottentots (South West Africa)	68
Bushmen (Kalahari)	84
Pygmies (Tswa)	63
Ethiopians (Amhara)	53
Egyptians	20
America	
N. American Negroes (Baltimore)	44
N. American Whites (New York)	4
Europe	
English	2
Italians	2
Asia	
Indians (South India)	4
Chinese (Canton)	3

Negroid populations, in the majority of whom the frequency is above 60%. It is interesting that the highest frequencies are in the tall Nilotes and in the rather stunted Bushmen. Populations in close contact with the Negroids, for example, the Egyptians and Ethiopians, also have relatively high cDe frequencies. Outside of Africa this variant is relatively uncommon except in peoples who are descended from African stock: for example, the Negroes of North America. No selective advantage for cDe has yet been identified.

(c) *Genetic Drift*

The genetic pattern of the migrant group can also be modified by the processes of genetic drift, which may be positive or negative.

As an instance of *negative genetic drift*, consider a certain gene (e.g. blood group B gene) which occurs in the parent population with a frequency of 10%. Twenty persons leave the parent population to establish a new colony in a remote territory. It is quite possible that by chance none of the migrants carries the B gene; or, perhaps just one of them carries the gene, and he fails to produce offspring. As a result, as the new race develops it will differ from its parent population in not having any B genes. This process of negative genetic drift can be defined as *the loss of a gene by random fluctuation in an initially small, isolated community*. It is possible that the almost complete absence of the B gene in the native populations of America arose in this way. It is believed that these populations have descended from groups of migrant Mongoloids who, in ancient times, crossed from Asia to North America via the land bridge at what is now the Behring Strait. It will be seen in Table IX that

TABLE IX. THE FREQUENCY OF BLOOD GROUP B GENE IN VARIOUS POPULATION SAMPLES (PERCENT)

Far East	1·5 - 30
Eskimo	0 - 1
N. American Indian	0 - 2
S. American Indian	0 - 5
Europeans	7 - 10

although the B gene frequency is high in Far East Asia, it is almost absent in the native American populations. Some B genes have been introduced into the American peoples by contact with White settlers from Europe who, in turn, had acquired the gene from the Mongol invaders of Europe in mediaeval times.

Positive genetic drift can be defined as *the establishment in high frequency of a formerly rare gene by random fluctuation in an initially small, isolated community*. As an example, consider a certain gene, say, the gene for porphyria, which exists as a mutation in only one member of the parent population. Forty settlers leave the parent population to establish a new colony 6,000 miles away; by chance, one of the colonists happens to be the carrier of the porphyria gene. The new settlers thrive in their new environment and produce a numerous progeny: within 300 years they have produced 1,000,000 offspring—many of whom carry the porphyria gene.

As many readers familiar with the work of Dr. Geoffrey Dean, from Port Elizabeth, will know, this example is a true one. In 1685 Gerrit Jansz van Deventer left Holland to settle at the Cape as one of the first free burghers. In 1688 he married Ariaantje Jacobs who came from a Rotterdam orphanage. One million of the 2 million Afrikaners in South Africa today are descended from the original 40 free burghers. About 8,000 - 10,000 of these descendants (about 1%) carry the porphyria gene; about 500 Cape Coloureds are also affected: all of these can trace their ancestry back to Gerrit Jansz van Deventer and Ariaantje Jacobs, one of whom must have carried the porphyria gene. Purely by chance the gene for porphyria is common in the Afrikaners; it is at present almost unknown in Holland where it originated.

Phase III

Mixture (*hybridization or heterosis*)*

In the course of time some members of the newly formed race may migrate again and come into contact with members of another race. Alternatively, members of a foreign race may invade the territory of the new race. Mating will take place between members of the two races and a new population will result whose gene frequencies

*The term *miscegenation* has been avoided in this essay since it is often misunderstood. Because the word begins with *mis-*, it is widely assumed that the term implies something unfavourable. In fact, *mis-* here is not a prefix at all. The term miscegenation is derived from two Latin words, *miscere* (mix) + *genus* (race) and simply means racial mixture without any unfavourable implications.

will be quite different from those of the mixing races and from those of the parent populations from which the latter are derived.

In fact, there are virtually no 'pure races.' All the races of modern man are more or less complex hybrids of other races. It has already been pointed out that the 5 - 10% B gene frequency in European races represents a degree of Mongoloid admixture. The 'African' rhesus variant cDe is found in most other races as well. The typically 'European' rhesus-negative trait is also found in many other parts of the world. One can extend the argument down to the individual level: in the final analysis every human being is a hybrid, the result of the association of two different sets of genes each of which is derived from one parent. Except in the case of identical twins, no two individuals have the same set of genes; the two parents are therefore genetically dissimilar and their offspring—all of us—are hybrids.

Accordingly, to a geneticist, the popular concept of a 'hybrid race' is nonsense. When laymen use this expression they really mean the result of the *recent* hybridization of two older hybrid races.

Viewed in this light, much of the argument about the alleged inferiority of 'hybrid races' becomes meaningless. One need only add that even as far as the relatively new hybrid races are concerned, there is no scientific evidence whatsoever for believing that racial mixture leads to any degree of inferiority, or, for that matter, superiority, in the offspring. As soon as there are objective criteria available for 'superiority' and 'inferiority' as applied to races and as soon as there are adequate methods for measuring them, it will be possible to investigate the effects of recent racial hybridization on these qualities. Until then there is only one conclusion: we do not know.

The Calculation of Racial Mixtures

Returning to the process of race formation, we have still to consider how genetic markers enable us to calculate the relative contributions of two parent races to the gene pool of their hybrid offspring. For example, if a contingent of new immigrants arrives in a territory and its members intermarry with the indigenous race and so produce a third, hybrid, race; then, provided that gene frequencies are known for the 3 races, the relative contributions of the immigrant and the original races to the hybrid race can be calculated. The statistical method is quite elementary: if q_1 is the frequency of a given gene in the indigenous race, q_2 its frequency in the immigrant race, and q_k its frequency in the hybrid race, then the genetic contribution of the indigenous race can be calculated from the formula:

$$\frac{q_k - q_2}{q_1 - q_2}$$

In practice, certain assumptions have to be made. It has to be assumed that the gene frequencies of the non-hybrid descendants of the indigenous and immigrant races are the same now as they were when hybridization first occurred. For example, if one wished to calculate the relative contribution of African and White genes to the gene pool of the modern American Negro, one would have to assume that the gene frequencies of the West African Negro is the same today as it was 300 years ago, and similarly in respect of the Whites.

THE GENETIC PATTERN OF CAPE TOWN'S RACIAL GROUPS

It is against this background of the genetics of race that I should like to present some of the preliminary results of an investigation into the genetical characteristics of Cape Town's racial groups. Altogether 1,200 individuals are being studied by a variety of genetic marker techniques (Table I). However, the investigation is not yet complete and the data in Table X refer to only 400 individuals:

TABLE X. THE FREQUENCIES OF SOME GENETIC MARKERS IN 3 OF CAPE TOWN'S RACIAL GROUPS AND THE PROPORTION OF 'AFRICAN' GENES IN THE CAPE COLOURED GENE POOL*

	Frequencies			*Proportion of 'African' genes in the Cape Coloured gene pool*
	White	*Coloured*	*African*	
Genes				
B	·08	·12	·16	·50
r (cde)	·40	·28	·17	·52
Ro (cDe)	0	·23	·74	·30
R_1 (CDe)	·48	·42	0	·13
M	·52	·56	·63	·36
S	·30	·26	·20	·40
Henshaw	0	·01	·04	·25
K	·02	·01	0	·33
Fy^a	·41	·36	·10	·16
P_1	·54	·53	·63	—
Hp^1	·38	·41	·56	·17
ptc	·55	·45	·25	·33
Phenotypes				
Atypical cholinesterase	·07	·03	0	—
Hp 2:1 M	0	·03	·06	—
Tf CD_1	0	·04	·08	—

*The gene frequencies in this and the subsequent Table are given in the conventional manner of genetical statistics, in which 100% is represented by 1·0, 50% by ·5, 2% by ·02, etc.

200 Cape Coloureds, 100 Africans and 100 Whites. The 3 groups are closely matched in respect of age and sex. Jews have not been included in the White series; the Cape Malays are also being analysed separately. Only those markers are listed which showed significant variation between the 3 races. So far we have found no significant variability in respect of the A and O genes; the Diego gene (which we have not yet encountered in Cape Town); haemoglobin types (almost all our subjects have the normal haemoglobin A); G6PD deficiency; and colour vision. The analyses of the finger-print pattern indices and of the Gm and Inv groups are not yet complete.

It will be seen in Table X that except in the case of the P_1 gene of the P blood group system, the frequencies of the genetic markers in the Cape Coloured race are always intermediate between those of the Whites and the Africans. The Cape Coloured community of Cape Town and its environs are generally regarded as a hybrid of African (Bantu and Hottentot), Cape Malay and White stocks. If

for the moment we ignore the influence of the Cape Malay genes, we can assume that the Cape Coloured people are the hybrid descendants of the indigenous Africans and the immigrant Whites. It is then possible to apply the above formula to estimate the relative contribution of the African genes (q_1) and the White genes (q_2) to the gene pool of the Cape Coloureds (q_k). The results for eleven gene frequencies are shown in the last column of Table IX. It will be seen that the African contribution to the Cape Coloured gene pool is almost always less than 50%. Taking an average over the whole series of gene frequencies, it can be estimated that the African contribution to the gene pool of the Cape Coloureds is about 31%. It must be emphasized again that the data are incomplete; but, allowing for this and for other reservations, it is reasonable to conclude that from the genetical point, the Cape Coloureds are more closely related to the Whites than to the Africans.

Two assumptions made in reaching this conclusion must be considered in more detail. Firstly, the Africans whom we have studied are all members of the Xhosa and Fingo tribes of the Southern Nguni, all of whom belong to the Southern Bantu-speaking group. No Hottentots (Khoisani) are included. Historically, it is known that the original progenitors of the present Cape Coloureds were the local Hottentots, White settlers or visitors, and Malay slaves. The Bantu-speaking Africans only appeared in the Western Cape much later. The gene frequencies of the Hottentots have not been included in our analysis of the Cape Coloured gene pool. This may seem a serious defect, but the available data on the gene frequencies in Hottentots suggest that, from the genetical point of view, the latter do not differ significantly from the Southern Bantu. However, much more data on these two African groups is required before they can confidently be regarded as genetically similar.

Secondly, no allowance has been made for the Cape Malay contribution to the Cape Coloured gene pool. It has been suggested that for several generations contact between these two groups has been so close that apart from their religion they are virtually indistinguishable. Others have argued that while some interbreeding has occurred, the Cape Malays have remained a closely-knit community and should be regarded as racially distinct. To elucidate this problem, we have studied the genetic markers of 100 Cape Malays of the same age and sex distribution as the other 3 groups. Only individuals whose parents and grandparents were also Cape Malays were included in this group. Their gene frequencies, in contrast with those of the 200 Cape Coloureds, are shown in Table XI.

It will be seen that in respect of several genetic markers there are substantial differences between the frequencies of the Cape Coloureds and those of the Cape Malays. At this stage of the investigation it may tentatively be concluded that because the Cape Malays differ from the Cape Coloureds in the frequency of some of their genes, they should be regarded as a genetically distinct race. It is interesting to see that where there are substantial differences between the gene frequencies in these 2 groups (those marked with an asterisk in Table XI), the trend in the Cape Malays is toward the 'Mongoloid' distribution (*cf.* Table II). In other words, after about 200 years at the Cape, the Eastern heritage of the Cape Malays can still be demonstrated.

TABLE XI. THE FREQUENCIES OF SOME GENETIC MARKERS IN THE CAPE COLOURED AND CAPE MALAY PEOPLES LIVING IN CAPE TOWN AND ITS VICINITY

	Cape Coloured	*Cape Malay*
Genes		
B	·12	·13
r (cde)	·28	·22
Ro (cDe)	·23	·14*
R_1 (CDe)	·42	·52*
M	·56	·56
S	·26	·25
Henshaw	·01	·02
K	·01	·01
Fy^a	·36	·52*
P_1	·53	·38*
Hp^1	·41	·33*
ptc	·45	·44
Phenotypes		
Atypical cholinesterase	·03	0
Hp 2:1 M	·03	·02
Tf CD_1	·04	·02

*See text.

CONCLUSIONS

The genetical approach to the classification of the races of man has been discussed. The use of genetic markers in studying the variability and the affinities of different peoples has been reviewed and an outline of the processes of race formation has been presented. It is hoped that when the objective and calmly scientific approach of the geneticist to race is more widely adopted, much of the prejudice and confusion with which the subject is now bedevilled will quickly disappear. This may prove to be one of the greatest contributions of genetics to the benefit of all mankind.

Some of the preliminary findings in a genetical survey of the peoples of Cape Town have been presented. Two very tentative conclusions have been derived from these data: firstly, that the Cape Coloured people are genetically more closely related to the Whites than to the Africans; and secondly, that the Cape Malays should be regarded as a distinct racial group who still retain features of their Eastern origin.

It is hoped soon to publish the results of this survey in more detail with all the necessary technical and statistical apparatus. The opportunity will then be taken personally to thank my colleagues in the Comprehensive Medicine Group and the many citizens of Cape Town who have made this study possible.

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