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# Interpretation of variability within and between population.

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**SUMMARY**

To help interpret the biological variability that occurs among human populations, the geographical patterns shown by different biological features are examined. The first group of such features shows gradients that suggest adaptation, thought to be the result of natural selection acting on gene pools. A second group shows great geographical gradients unrelated to any identifiable selective agent, and these seem largely attributable gene flow, especially by demic diffusion. The explanation of the extremes at the terminations of the gradients is complex, and factors involved are likely to have been the fundamental stability of gene frequencies, the action of selection, and random events.

When the great age of discovery first drew to attention the many differences in appearance of mankind throughout the world, the curiosity that was aroused resulted in further voyages of exploration and eventually to the accumulation of the vast mass of descriptive information that exists in the early literature. Though the attribution of these differences to the Creation was rejected as a result of the changed opinion that followed Darwin's work, yet we are only today beginning to understand the processes, the complexity of the mechanisms by which human variability has been brought about; Much of our knowledge derives principally from two empirical sources: (a) the patterns of geographical variation that can be observed on a continental scale; (b) the detailed study of local populations isolated from the mainstream, and particularly in an historical context. Then there is a third source, (c) the experimental validation of the hypotheses based on the other two, either by direct planned investigation or by «nature's» experiments in particular situations, from which, e.g. morbidity or mortality data can be examined.

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## PATTERNS OF GEOGRAPHICAL VARIATION ON A WORLD SCALE

### 1. Appearance

a) *Nasal shape*

Though there was considerable early speculation, the first morphological character to be seriously examined was the shape of the nose, measured by its index, breadth relative to its height. Arthur Thornson was impressed by its great variation from people to people, and the geographical pattern it showed. Thomson & Buxton (1923) put this pattern on a statistical footing, establishing that mean nasal index in indigenous peoples was closely related with climate, with a correlation coefficient of +0.629 with mean annual temperature and +0.419 with relative humidity, taken separately, and +0.724 when taken together. It was suggested that the modifications of nose shape might be related to the need for moistening inspired air or might be related to heat loss from the respiratory tract. Further evidence comes from physiological studies. A prime function of the nasal epithelium appears to be moistening of the air. The cilia are more susceptible to changes in the consistency of the film in which they work, and therefore to drying out, than they are to quite large degrees of heating and cooling. The cilia move toward the exterior mucus and any dust that enters the

nose, and so clean the inspired air, which is broken up into shallower streams passing through the cleft between the convoluted internal surfaces. A strong influence of relative humidity on mucous flow rate has been demonstrated. Consequently, the critical physical character primarily concerned in the exchange of water from the internal epithelium to inspired air is the absolute humidity. There is indeed a closer association between the nasal index and absolute humidity, for the initial data showed a correlation coefficient with wet bulb temperature of 0.77 and with vapour pressure of 0.82, a significant increase over the correlations initially observed (WEINER, 1954).

Clinical work also shows the importance of humidity. In patients with tracheotomy, the nasal passages are by-passed. The air reaching the lungs is little different in temperature and humidity from room air, it takes up moisture in the bronchi from the mucous membranes so that bronchial secretions become dry. This is not a serious handicap to anyone who can cough them up, but it may be disastrous in patients whose power to cough is impaired and whose bronchi do become blocked. This became very obvious in patients with various forms of paralysis necessitating tracheotomy and artificial respiration, as in the Danish poliomyelitis epidemic of 1952 before the need for humidification was recognised. If in these cases inspired air is properly humidified, there are no sinister complications of encrusted secretions.

b) *Skin pigmentation*

The second example is skin pigmentation which shows striking variation from population to population. Its early analysis was unsophisticated because of the difficulty of measuring it, and it was only with coming of the measurement of colour by reflectometry that any advance was made. Skin colour is due to the presence of several pigments, and to an additional optical effect, scattering. Melanin, the most important pigment, is located as discrete particles mainly in the deepest layers of the epidermis in lightskinned people, but in dark-skinned they are more abundant, spreading right up to the superficial layers. The general association of dark skin and tropical habitat has long been known. Over 80% of the variance in population mean reflectance at each wavelength measured is attributable to climatic variables (ROBERTS, 1977). Parallel gradients are observed in different continental groups, suggesting that darkness of the skin may have evolved several times in man. The geographical distribution suggests strong selective advantages to dark skin colour where there is increase in the amount and intensity of insolation. The ultraviolet parts of the spectrum in particular have some beneficial effect at minimal exposures, but at greater doses promote injury in the form of sunburn and carcinogenesis. The carcinogenic wavelengths lie between 2537 and 3341 A units, while erythema is induced increasingly rapidly at wavelengths of less than 3,200, reaching a maximum at 2,800 A. Man is, of course, protected by the earth's atmosphere from much ultraviolet radiation, and much of the radiation reaching the surface of the body does not penetrate to the deeper layers of the skin, but some does so. When sheets of the horny layers of skin from Africans and Europeans in Nigeria, shown to be similar in thickness to each other, were placed on a photographic plate and exposed to ultraviolet irradiation, the European specimen showed much more pronunced penetration than did the dark African skin. The results of ultraviolet therapy in Whites with rickets show that active rays do penetrate deeply enough to affect at least the superficial capillary circulation or to stimulate the nerve endings. Besides this and other experimental evidence, there are empirical observations. The response of albinos in the tropics who have little or no melanin is to develop a thickening of the horny layer, which though less efficient at least provides some protection. Cutaneous cancers are rare in dark-skinned as compared with light-skinned subjects in the tropics.

While there is little doubt that skin colour variations have been produced by selection, quite rapid and quite intense, the precise advantages of a dark skin in the tropical environment are obviously complex and still not fully clear. But also, selection for fair skin at higher latitudes may have occurred. In the conditions of coolnes, heavy cloud, and relatively little radiation stress of north-west Europe, there would be no advantage in dark skin. The occurrence of the lightest skin here may perhaps be due to the effects of chance on Europe's ancestors, implying selective neutrality of lightness. However, the suggestion from genetic studies (HARRISON & OWEN, 1964) of overall dominance of the genes from the white parent over those from the black, carries the implication that selection for depigmentation may have been strong. Ultraviolet rays penetrating the skin cause a rearrangement of atoms in the sterol molecules, endowing them with the properties characteristic of vitamin D, and this anti-rachitic potency is most effective in the range 2,500-3,130 A. In conditions of minimal sunshine where dietary vitamin D is low, increased access of these rays to the deeper skin layers could well be selectively advantageous in preventing the development of rickets. Black children dwelling in the great cities in the northern part of America, and Pakistani children in Glasgow, are more susceptible to rickets than are White children, and it is possible that this may indicate something more than a dietary deficiency. It may well be due to the fact that, of the lessened amount of

ultraviolet light reaching the skin in temperate latitudes, again diminished by atmospheric pollution and indoor living, what remains after absorption by the melanin of the black skin is insufficient to stimulate the production of vitamin D in the dermal layers.

c) *Body shape*

A further feature of interest is body shape. A zoological rule - ALLEN'S (1877) rule - states that in warm-blooded species the relative size of exposed portions of the body decreases with decreasing mean temperature. The physiological mechanism is the loss or gain of body heat in relation to the amount produced. The amount of heat produced by the body depends largely on its mass. The amount lost or gained by convection, radiation and evaporation depends on its surface area. Increase in limb length relative to trunk and increased linearity of form increases the surface area relative to mass.

Mean sitting height, expressed as a percentage of stature (the relative sitting height) in male indigenous groups, when plotted against the mean annual temperature of their habitat, shows a highly significant linear association ($r = -0.619, b = -0.355$/ºC), values indicating that nearly two-fifths of the total variance is ascribable to mean temperature. The same relationship also appears in female samples ($r = -0.654$, $b = -0.389$/ºC). The temperature relationship within the major racial groups remains highly significant and, particularly interesting, it seems that not only do parallel temperature relationships exist within each continental group, but each continental group is itself adjusted to a particular habitat range. Indeed, after allowing for the effect of temperature there are highly significant differences amongst such groups. Lower values than the overall adjusted mean occur in Australian aborigines, African negroes, Khoisan and Melanesian groups, i.e. those which are generally thought to have occupied a tropical habitat far a considerable period. Other measurements of the lower limb are also available —symphyseal height, navel height to anterior superior iliac spine, trochanteric height— and each expressed in proportion to stature shows increase with increase in mean temperature. Moreover, the lower limb, besides increasing in length relative to stature, seems to change its shape and become more slender with increase in temperature; there is a decrease in relative calf circumference, thigh circumference, and an increase in relative thigh length and calf length, and also in the calf/thigh index in which the length of calf is expressed relative to that of the thigh. It seems that the distal portion of the limb increases in linearity more markedly than does the proximal, giving the characteristic elongation of the lower segment so frequently noted in dwellers in hot climates.

The upper limb, too, shows similar correlations with temperature. Relative span increases with mean annual temperature ($r = +0.470$, $b = +0.391$/ºC), and once again multiple covariance analysis indicates highly significant differences between continental groups. Direct measurement of arm length shows a similar increase, and this appears to be due to tendencies to elongation in each of the segments of the arm (upper arm, forearm and hand) relative to stature. The antibrachial index (forearm length expressed as a percentage of upper arm length) also increases with temperature, so that it appears that greater increase occurs in the lower arm than in the upper. Since the forearm is generally more slender than the upper arm, a given increase in length produces less increase in volume. In other words, greater modification in mass/surface area ratio is effected by increase in the length of the lower end of the arm.

The trunk, too, participates in these tendencies. Mean chest girth of indigenous groups shows a coefficient of correlation $r = -0.575$, and of regression $b = -0.086$/ºC, highly significant, and the other trunk measures — bicristal breadth and biacromial breadth— both show highly significant decreases with increase in temperature (Figure 5). In warmer climates, then, the pelvis, the chest ant the shoulders are more slender than in cool regions.

No matter which measure one takes, all variation in relationship to mean environmental temperature is highly significant and is such as to increase the ratio of body surface area to body mass at higher temperatures, and to decrease it at lower (ROBERTS, 1978). Thus, the apparent conflict in finding both pygmies and long lanky Nilotics as adaptations to high temperatures, can be resolved. In the pygmy, by reduction of size the body mass is reduced, thereby reducing the ratio of weight per surface area. In the linear Nilotic, the great increase in slender limb length increases the surface area per unit weight, achieving the same overall effect of reduced weight per surface area ratio.

Experimental data support this empirical evidence of climatic relationships, show the physiological changes that occur under extreme climatic conditions, and indicate the functional reasons for the morphological association. When a person enters a hot environment, the skin temperature rises, the surface blood vessels dilate, there is a slight increase in blood and plasma volume, and more blood flows through the skin, carrying more heat to the periphery to be dissipated. The blood pressure drops, with the result that the heart output increases to maintain blood pressure, and the pulse rate increases. When the ambient temperature rises above about 27ºC, less heat is lost by radiation and convection, and one begins to lose more and more heat by eva-

poration of sweat, until, when the ambient temperature rises above the body temperature, the body is actually gaining heat by radiation and convection, and the only means of heat loss is by sweating. As the environment becomes more severe, or if the amount of heat one produces internally through increased workload markedly increases, there is first discomfort, then distress, and eventually there comes a point when the sweat glands break down, one can only store heat, and collapse occurs. With recurrent exposure, one becomes acclimatized so that this process is deferred, and it happens at a higher stress. Under extreme conditions, particularly evaporative loss is difficult by reason of high humidity, the importance of a slight increase in area over which sweating occurs is clear.

When a person is exposed to cold, the body loses heat primarily by conduction, convection and radiation, the rate depending on the difference between body surface and environmental temperature, and the amount depending on the rate and the extent of body surface. Heat is also lost by evaporation from the lungs and by insensible perspiration. The skin temperature drops, and often vasoconstriction diminishes the conductivity of the peripheral tissues. But the limit of its effect is soon reached, after which skin cooling continues to occur. Heat continues to flow from the deep tissues to the surface, so that the deep body temperature begins to fall. To maintain thermal equilibrium one increases heat production. But if the deep body temperature continues to drop, coma, muscular rigidity and death ensue. In cold conditions, the diminished surface area/body mass ratio clearly is important.

There is now extensive literature on heat and cold tolerance. Generally, the experimental evidence is consistent and indicates the importance of morphological differences in climatic stress and supports the generalisations from the empirical analysis. Variation in ability to cope with climatic extremes appears to be a major selective mechanism..

d) *Conclusion*

These three examples all illustrate adaptive variation. They are all characters where the process by which the geographical pattern comes about is complex, and is affected by buffering factors. First there are the cultural factors that intervene between the individual and the environment, but these are of limited effect only, and have been with us only in our most recent evolution. A second buffer is provided by man's physiology. Thirdly, there is plasticity in development and in the response of the growing individual to the environment in which he finds himself. Yet no pygmy child will grow into a Nilotic negro, no matter how excellent his environment; no north-west European will develop a dark skin to match that of the Melanesian, no matter how exposed to sunlight he may be; no Nilotic will develop the proportions of the Eskimo, no matter how much blubber he consumes. The conclusion is that the basis of man's bodily variation discussed here is genetic in origin, representing the accumulated contributions over countles generations of selection, to give improved adaptation to climate.

## 2. Single gene characters

When the simpler Mendelian characters are examined, where there is no such buffering, the phenotype is the direct expression of the genes. It was this type of feature that emancipated us from typology, that showed how to characterize a population in terms of its array of gene frequencies, and how to understand evolution in terms of gene frequency change. Moreover, the gene frequencies in a large population tend to remain constant from generation to generation. The geographical patterns are quite different. The features that are polymorphic, with sufficient variation in gene frequency, show two types of pattern. There are the strong regional variations in gene frequency associated with particular selective pressures, e.g. of the abnormal haemoglobin S genes and the Duffy $Fy^4$ gene in relation to malaria. But for the majority of the bloodgroup, enzyme and serum protein types, the very clear geographical gradients in gene frequency (allowing for sampling variation) appear regular and unrelated to any identifiable selective agent. Sometimes the gradients are stepped, with breaks of slope representing the occurrence of barriers to gene flow. Where such breaks of slope coincide in many features, this may be indicative of zones of transition between races. But whether or not these occur, the gradients are suficiently strong for geographical distance between populations to be a major determinant of their gene frequencies. The terminations of the gradients, i.e. the maximal and minimal frequencies reported, differ from character to character, so that for example the highest frequency of bloodgroup B occurs in central Asia, that of gene *M* in Central America, that of gene C in the New Guinea region, that of gene d in the Basques. Hence no one human population can be picked out as being conspicuously different from all others in possessing a concentration of extreme frequencies. This implies that all have advanced similar distances along the evolutionary pathway, but in slightly different directions. The question then is how have these extreme frequencies at the terminations of the gradients come about, and what is the mechanism by which the gradients have evolved?

a) *Gene frequency gradients*

Taking these questions in reverse order, a gradient may be most reasonably interpreted either as

adaptive to increasing selection pressure along it, or as representing the effect of gene flow. The fact that the great gradients in Mendelian gene frequencies in man have few parallels in any identifiable selective factor is not to say that these characters have no selective value; for selection here may well be a function not of geographically varying factors but of differences within the population, i.e. between individuals sharing a common geographical environment but differing in biological makeup and personal environment. But the alternative explanation, gene flow, seems much more reasonable. The gene frequency maps (MOURANT, 1954) with their succession of isogenes suggest waves of advance, e.g.of the *D* and *B* genes westwards across Europe. But what form did that advance take?

It is not necessary to invoke massive population movements, transferring massive numbers of new genes over large distances. Certainly these occurred in the 17th, 18th and 19th centuries, e.g. into the Americas from Europe and Africa, but these were very much a product of history, of the new technologies and skills. They are unlikely to have occurred earlier. Certainly there were the historic movements of peoples, e.g. into Europe, but their genetic effect may well be overestimated. First, the areas into which they moved were already populated, movement was slow, and in the time taken by the migrants there was ample opportunity for them to acquire partners, and hence genes, from the peoples through whom they passed, and at the same time leave some of their own. Intermixture, either peaceful or violent, undoubtedly occurred, diluting the new comers' gene pool. The gene pool of the Visigoths who entered Spain is unlikely to have remained the same as that of their ancestors when they entered Thrace. Secondly, it seems likely that history and folklore magnified the numbers involved in the movements. In many cases, what probably happened was movement of a small number of people who brought their genes as well as their ideas to the new areas, intermixed with the inhabitants already there, who accepted or rejected to varying degrees the values, customs, language of the invaders, and because of their prestige later generations came to claim descent from the newcomers. Moreover, the distance travelled by the majority participating in, or fleeing from, invasion, is not likely to have been enormous. The indigenous families who fled before an invding group are unlikely to have journeyed far, for example the escape of some south Slavs from the advancing Turks took them only as far as the Dalmatian Islands from the mainland. Most of those in the Roman armies who conquered Britain came not from Rome but from Gaul and Britain itself. In brief, the interpretation of the gene frequency gradients does not call for population movement that was massive in amount or distance covered.

For the greater part of human history, and particularly when the skeleton of the genetic maps was laid down, mankind was primitive. Like the animals, it was essential for him to know his territory, to know where to find his food and other requirements. There are many studies of animals showing site fidelity, the tendency of individuals to remain in or near the area of their birth. There must have been strong contraints on migration, such as knowledge of the location, movement and availability of food sources, and it could not have been common for whole groups to have left their natal range on long one-way treks into the unknown. Instead, it seems much more likely that groups usually remained within a known territorial range, from which individuals or families gradually moved into adjacent territories —the process of demic diffusion. Certainly massive movement could not have ocurred between continents, otherwise the characteristic gene frequency differences would have disappeared. Obviously there was sufficient gene flow worldwide to maintain species homogeneity. For even a slow trickle of genes by way of mate exchange can be sufficient to keep continuity over a long period. If in each generation there are one or two individuals who move in a certain direction to a neighbouring locality, the movement of genes that this produces though not great is regular, and in the long run will have the same effect as a more massive population movement. This process is likely to have been fundamental in establishing the gene frequency gradients. It is not necessary to regard them as indications of massive waves of migrants, but as the outcome of currents of genetic diffusion.

In brief, the interpretation of the gene frequency gradients does not call for population movement that was massive in amount or distance covered. Instead, it is more likely to have consisted of long-continuing, steady demic diffusion, and occasionally where invasions are thought to have ocurred, most of the incoming genes are not likely to have been brought from great distances.

b) *The extremes*

Turning now to the second question, how did the gene frequencies at the extremes of the gradients come about? One posibility is that their features are the result of local selection, as perhaps is the case with the high frequency of haemoglobin C in West Africa, centred on the Upper Volta, from which focus the frequency declines outwards in all directions. It seems likely that an original mutant there proved advantageous, increased in frequency under the influence of natural selection, to its present level. Such a process may refer to selection pressures that are continuing, or that operated in the past an now do so no longer, so that one is left with a semi-fossil gene frequency. Another possibility is illustrated by

the high frequency of blood group gene N in the Ainu; or the slight increase in the B gene frequency along the Celtic fringe of the British Isles; or the very high HLA A2 frequency and the very low NS haplotype frequency in Orkney; these seem to be due to the continuing survival there of relict populations from an earlier time, in what proved to be regions of refuge. This is not to say that the present populations are the same as, or genetically identical to, those ancestors. What it means is that the present-day gene pools contain major contributions from those of the original peoples. Perhaps this is the explanation of the Basque uniqueness.

One has only to travel in the countryside around Vitoria to realise how different are the Basques from other European peoples. At the period when to establish origins was a major preoccupation, two apparently contradictory opinions on Basque origins were advanced. According to COLLIGNON (1894), the Basques constitute the only present survivor of an ancient population of prehistoric western Europe — the Cromagnons. According to ARANZADI (1905) on the other hand, the Basques, despite their different characteristics, represent only a local population, a local variant among others in south-west Europe. When evdence from genetic characters became available, Collignon's hypotesis appeared established, for the first bloodgroup study in 1937 and all subsequent surveys showed that the frequency of bloodgroup B in the Basques is the lowest in Europe, and indeed is zero in a proportion of studies. Subsequent work showed that they also have the highest frequency of any known population of the Rhesus gene d, and the Rhesus haplotype *cDE* is at the lowest frequency in Europe. Neighbouring populations show a tendency towards these extremes, explicable on the Collignon hypothesis in terms of some Basque admixture of unknown antiquity. Studies of other more recently discovered genetic characters point in the same dierection. But, to reiterate, neither of these hypotheses takes cognisance of the fluid and dynamic nature of genetic evolution. Thes Basques are not a .«fossil» population. They are, and have been, in process of evolutionary change as are all others. In their genepool, however, is a greater proportion of the early European genes, not in terms only of gene frequency but also of linkage groups.

## THE EVIDENCE OF ISOLATED POPULATIONS

To understand the complexity of the processes involved, it is necessary to turn to the second principal type of study, that of isolated populations. There is one that provides a particularly good ilustration, that of the tiny, remote and inhospitable island of Tristan da Cunha in the middle of the South Atlantic ocean, one of the most remote islands in the world, 1800 miles from Cape Town, 2000 miles from Rio de Janeiro, 1500 miles from St. Helena.

The island is almost unique in that it is possible to trace the evolution of the population from its beginning, from the data on births, deaths, arrivals and departures. The present population takes its origin from the garrison which was landed there in 1816 to forestall any attempt at rescue of Napoleon from his exile on St. Helena, for when the garrison was withdrawn the following year three members, one with his wife and two children, remained. During the first few years there came others, and according to the literature, fifteen individuals are the ancestors of the present population of some 300, and these fifteen included two pairs of sisters and one mother-daughter pair. There were several others who came for a time and departed without leaving any descendants on the island, and occasionally also others who, for want of a better term, may be called «hidden ancestors», who also came for a time and then departed leaving a descendant on the island.

Living was at first adequate if not easy for this little community, for during the days of sail they could trade with the ships that called in for fresh water, the islanders providing potatoes and fresh meat in exchange for flour, sugar, wood, paint and hardware. But with the coming of steam, boats no longer called; the islanders were just able to support themselves by their vegetable plots, their animals and their fishing. The standard of life seems to have steadily declined. A number of the community emigrated, either as individuals dissatisfied with the difficulties of existence, or as a group; for example, after a particularly severe tragedy in 1885 when 15 adult males were drowned in an accident at sea endeavouring to make contact with a passing vessel as was usual, the departures of their widows and children diminished the population to 59, whereas in 1884 it had been 106. Others remained; after each major exodus the numbers grew again, although with increasing isolation the islanders depended increasingly on charity supplied through missionaries.

In 1940 a British naval garrison was stationed there, and the islanders became acquainted with money, employment, and the goods that money could buy. After the War, the Tristan da Cunha Development Company was established to fish for crayfish and market them via Cape Town, thus providing contact with the outside world. The enterprise was successful, sufficient to provide funds for the installation in 1960 of running water in each house, modern sanitation, and the prospect that at last the island could become self-supporting. Then on 10 October 1961 with very little warning, the volcano erupted just behind the settlement. The folk left their houses and subsequently went to Cape Town and then to England, finally returning to the island when all was once more safe.

It is against this historical background that the effects of their isolation on their evolving genetic structure can be appreciated. With the small population size, and the great reductions in it that occurred, there was little choice of spouse for those wishing to marry —theycould only seek mates amongst their relatives (ROBERTS, 1967). Consequently there occurred the development of inbreeding in the population, inbreeding being measured by the inbreeding coefficient, the probability that the two genes that an individual possesses at a locus are identical, being replicates of a single gene in the common ancestor of his father and mother.

*Inbreeding*

The inbreeding coefficients for all members of the population since its inception show that the mean autosomal inbreeding coefficient (F) has steadily increased with the passage of time. In this secular increase, the first reduction in population size of 1855-57 was obviously important in initiating the development of inbreeding, though it would certainly have occurred at a later date if there had been no such reduction in numbers. In the second population reduction, the deaths at sea in 1885 slightly reduced the mean level, but it was the subsequent emigration of the families of the drowned men that accelerated the increase in the inbreeding level. However, the effects of neither event are unduly great by comparison with the overall increase in the intervening periods. The general increase during the history of the population is of course to be expected, since in a closed population of limited size the later the generation an individual belongs to, the more common ancestors are available to him.

For loci on the X chromosome the mean X-linked inbreeding coefficient $F_x$ shows a somewhat different pattern of increase. First the mean today is at a rather higher level than the autosomal, and has indeed been so throughout most of the history. Secondly, the effects of the population size reductions are much more pronounced. The first produced an increase some four times as great as in the autosomal, and the second produced a remarkable decrease instead of an increase, with the mean $F_x$ diminishing to one-third of its previous value in a five-year period. In the intervening period the rate of increase of $F_x$ was more rapid than for the autosomal.

All in all, however, the present mean levels indicate that the inbreeding that has so far occurred is not excessive. The average for the 1961 population was .040 (i.e. the probability that the average individual possesses two identical genes at a given locus is only 4%), a figure which is only two-thirds that of the offspring of a marriage between two first cousins, and is not at all high.

*Kinship*

A second highly useful measure of genetic structure is the coefficient of kinship (Ø) between two individuals which measures the probability that two homologous genes drawn at random, one from each individual, will be identical. Again the kinship coefficient between all members of the population, and the mean at the end of each year, has been calculated.

In the evolution of kinship the pattern is different from that of inbreeding. Whereas inbreeding has risen steadily and is continuing to do so, the mean kinship coefficient has reached a plateau. It was appreciably elevated by the first population size reduction, much less so by the second, and its increase in the last fifty years has been quite slight. Much more striking, however, is the number of pairs of individuals out of all possible pairs who were not related at all. The percentage of zero coefficients declined rapidly, again particularly in the first period of population reduction, but not conspicuously so, and there was virtually no effect of the second population reduction. However, the decline continued until in 1970 the position was reached where there were no zero coefficients, that is to say everybody in the population was related to everybody else.

*The distribution of genes in the population.*

The distribution of genes in the population depends on two factors: the first is which of the founding ancestors introduced which genes. The founders themselves may not carry a representative sample of the genepools of the populations from which they derived, and it may well be that there was in them an elevated incidence of a particular gene - the founder effect. In a number of characters it has been possible to identify the ancestor responsible for introducing a particular gene to the Tristan population and in some cases the answer is quite unequivocal. For serum cholinesterase $C_5$ the gene can only have been introduced by one of the original St. Helena women, who alone is ancestral to all carriers of the allele. Similarly, she was responsible for introducing esterase $D^2$. The rhesus haplotype Cde was introduced by one of the Italians. For other features it is not possible to be precise, but a probability that a given ancestor introduced the gene can be calculated (THOMPSON, 1978).

The second factor affecting the distribution of genes is how much each founding ancestor contributes to the genetic constitution of the present po-

pulation. Its importance can be demonstrated by reference to the two major episodes of population decline, due to large-scale emigration. The first was due to a combination of chance circumstances, the death of the founder member at the time when a missionary was present on the island who made no secret of his desire to get away, and under his influence two-thirds of the population left; the second emigration occurred after the boat disaster in which the majority of the adult male population were lost, and their widows and children subsequently left the island. There must have been a large chance element in which individuals emigrated and which did not; this can be identified as random effects of a bottleneck situation. But there must also have been a large non-accidental component to the actual emigration itself. In both instances it was family groups that emigrated; this brought about what may be termed a «booster» effect, whereby although the genes lost by accident were a random sample, the loss of some of them was exaggerated by the consequent deliberate emigration of families. Hence which genes and what proportions of them were lost from the population's genepool were a partly random, partly non-random array. The families that departed obviously included individuals who felt that they could no longer accept the conditions on the island or its prospects for them, so the non-random component may be, in part, identified as selective.

Examining the effects of such bottlenecks on the genetic constitution of the population by the PACK method of analysis, i.e. the Probable Ancestral Contributions to each individual, the calculation of these probable contributions at a series of points in time shows how the genetic constitution of the population varies during the period. The effects of the population reductions are quite clear. In the first bottleneck the primary effect was deprivation of the population of eight of its founder ancestors and a recent arrival, so that instead of the genes from twenty ancestors that were present in the genepool in 1855 there were contributions from only 11 ancestors. Secondly, there was a change in their relative contributions. Genes from two of the principal contributors were among those that completely disappeared. The contribution of ancestor 1 was halved, and that of ancestors 3 and 4 more than doubled. The profile of the ancestral contributions altered appreciably as a result of this first bottleneck. Then from the end of 1857 to 1884 the profile changed very little, though to it had been added contributions from six more arrivals. The boat disaster of 1885 had relatively little effect, but the subsequent emigration was much more pronounced. Again, all the contributions (totalling some 8%) were lost from four relatively recent arrivals, and there was a change in the relative contributions of the remainder.

But the PACK method can also be applied to monitor the ongoing evolution of a population on a year-to-year basis, or in whatever time units are required; for whenever a member of the population dies or emigrates, or there is a new arrival by birth or immigration, there is a slight change in the genetic constitution. In Tristan differential fertility is shown to play a very important part in the genetic evolution of the population. Some 75% of the total change in genetic constitution in the first period of population growth before the first exodus, 72% in the period between the two bottlenecks, and 53% in the period after the second bottleneck to 1961, were due to differential fertility. Over the total history of the Tristan population, differential fertility accounts for approximately 47% of the accumulated change in genetic constitution.

*Evolutionary progress*

The evolutionary progress of the Tristan population can be summarised in terms of its genetic distance from the present population (ROBERTS & BEAR, 1980). Identifying genetic constitution in terms of the probable ancestral contributions, genetic distance travelled by an evolving population in the period between two given years may be measured, for example, by the Z statistic of Jacquard (1974) or the $\Theta$ statistic of CAVALLI-SFORZA and EDWARDS (1967). There was quite rapid initial change in the period to 1840, due mainly to the varying rates of reproduction of the founders (ROBERTS, 1967), then little until the first population decline in 1855-57 which had an appreciable effect, then little until the second population decline which again brought increase in similarity to the present constitution. Thereafter there occurred a slight but steady progression towards the present. Both the Z and $\Theta$ measures agree in giving the general impression of a population steadily changing in its genetic constitution, emphatically so at the two periods of population decline, and with increasing stability in later decades with larger population numbers.

The impression is given that the islanders were in the grip of fate, that the inexorable processes of increasing inbreeding and increasing kinship were progressing slowly but surely, and that this was the price paid for their isolation, for continuing to live on the island. Those who emigrated freed themselves from these processes but at the cost of enhancing their effects on the folk who remained behind.

*Conclusión*

One can now identify the types of process responsible for the unique genetic features of the Tristan population.

(1) The founder effect -the genes that were brought by the ancestors at frequencies that were quite unrepresentative of those in the populations from which they derived.

(2) The inbreeding that occurred, that caused a number of recessive genes to manifest in the homozygotes.

(3) The close web of kinship illustrated in the frequency of a number of dominant features.,

(4) The random changes due to the chance element in the population reductions in the bottlenecks.

(5) The «booster» effect of the departure of close relatives of those who were lost to the population, either following their deaths or accompanying those who chose to leave.

(6) The effects of selection; positive selection encouraged those who felt at an advantage on Tristan to settle and remain there, and negative discouraged those who could not stand the isolation and difficulties of the Tristan way of life from remaining there.

Through these the effect of isolation is allpervasive. It protected the initial genepool from dilution. It led to the development of the inbreeding and the kinship levels. It prompted the major periods of exodus and the occasional individual departures, as well as the positive and negative aspects of selection that have been picked out. Certainly, the alteration of the genetic constitution by the numerical bottlenecks of the population would have had less enduring genetic effect had there been more population movement and gene flow in the intervening periods.

The example of Tristan da Cunha discussed here, small and remote though the population is, has for academic biology an importance quite out of proportion to its numbers. Their isolation affected their genetic constitution in numerous ways. The study illustrates the type of problem that must have been all too common in the very earliest days of human evolution when populations were small, as well as in other more recent isolated small groups. The processes identified are likely to have operated, though to a lesser degree, in other isolated populations. It is therefore possible that they also applied to the Basques or their ancestors in their earliest days. In the early periods of settlement, penetration of small groups into the valleys, after the retreat of the ice permitted it, and the operation of such processes is likely to have led each to become its own Tristan. Such processes are certainly unlikely to be pronouncedly affecting the Basque population today, with the size it is. But this line of thought leads one to wonder whether today the Basques are a homogeneous entity, or if local subpopulations differ? And if so, whether local variation derives from early evolution or more recent processes and especially admixture from immigrants. For if there are local isolates within the Basques, then the pattern of ongoing evolution is likely to be quite different. What is urgently needed is a thorough genetic survey to establish the extent of local variation among the Basques, and the effect that recent population movements are having on it.

## CONCLUSION

To help interpret the biological variability that occurs among human populations, this study examines the geographical patterns shown by different features. It distinguishes first those that show adaptation, with particular reference to climate, thought to be the result of natural selection acting on the gene pools but buffered by culture, physiological responses, and developmental plasticity. Secondly are distinguished the simple genetic characters; some of these show regional frequency patterns associated with particular selective factors; the frequencies of all considered simultaneously differentiate the great continental groups or races of mankind; the majority show great geographical gradients unrelated to any identifiable selective agent. These gradients seem largely attributable to gene flow, especially by demic diffusion - small scale movements over relatively short distances by a few individuals over protracted periods. The extremes at the terminations of the gradients have all become stabilized, no matter what the originating processes, by increased population size. But certainly for these the explanation is complex. This complexity is illustrated by reference to the population of Tristan da Cunha, which shows how the fundamental stability of gene frequencies from generation to generation is affected by different types of selection, the mating pattern, and especially by random events and the founder principle. The extent to which such processes operate on other populations depends on their size and whether they form a homogeneous entity or are divided into a number of subpopulations. The Basques in their early days are likely to have been influenced by such processes. The extent to which they have operated recently depends on whether the Basques are homogeneous.

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