Laboratório de Genética Humana, Faculdade de Medicina
Universidade de São Paulo, Brazil

The Genetic effects of Immigration in a Rural Community of São Paulo, Brazil

P. H. Saldanha

I. Preface

The present work is an attempt to apply modern theoretical concepts of population genetics in the investigation of a human community. The theory of population genetics has grown rapidly through deductive-mathematical analysis, especially in consequence of the work of Fisher, Wright, Haldane, and Dahlberg. However, their theoretical conclusions have only recently begun to be checked by empirical data, mostly from human populations. The results obtained show that man is especially suitable for this kind of investigation.

Human populations are not only suitable for general studies of heredity, they also illustrate some specific evolutionary problems not found in other species. One of these, undoubtedly related to culture, is the evolutionary problem of migration and intermixture between populations previously isolated one from another. The great interpopulational variability of the human species in pre-historic periods is changing toward an intrapopulational polymorphism, caused by the increasing geographical and social mobility of man. The previously existing geographical and ecological barriers are being replaced by cultural and social ones. Migration as an evolutionary factor shows a more important role than in any other species.

In recent generations human migratory movements have become both larger and more generalized. Small villages and rural communities are increasing in and overlapping their new boundaries, as can be seen especially in Europe. Moreover, the past three generations have been characterized by " organized " migratory movements between remote regions, and between genetically different people, as seen especially in America. The breakdown of previously isolated communities under strong immigration pressure has not been analysed from a genetical point of view. However, these immigratory movements should be changing markedly the genetical structure of the populations. In this situation, there is likely to occur a decrease in the mean coefficient of inbreeding of these populations which will result in a relatively lower frequency of recessive abnormalities and diseases. Besides this expected eugenic effect, new gene combinations are produced and " tested ", as a result of

Ricevuto il 20 II. 1962.

the relatively greater heterozygosity of the new populations. The consequences of this phenomenon can be extremely important to human evolution. These studies seem also to be important for the understanding of some specific problems related to the dynamics of gene frequency and interaction, as for example, in the case of polygenic traits, viz. stature (that seems to be increasing in the last generations).

This investigation tries to analyse a typical southern rural Brazilian community that received a great number of European immigrants. In spite of some interesting conclusions drawn from this study, the author is himself advised of the limitations related to both the data and the methods used in such a kind of investigation. The major part of the data presented herewith were used in a doctoral thesis, submitted to the Universidade de São Paulo, Brazil.

The author wishes to express his thanks to several persons who helped him, in different ways, in the elaboration of the investigation. First, he is very thankful to Dr. Harry M. Miller, Jr., as the Rockefeller Foundation adviser, for the support of an initial plan on research of human genetics in São Paulo, that made possible the largest part of this investigation. To Professor C. Pavan, Director of the Departamento de Biologia da Universidade de São Paulo, in which the bulk of the work was conducted, the author is deeply indebted for the suitable working conditions and invaluable assistance. The author must also thank to Dr. O. Frota Pessoa, Professor F. G. Brieger, Dr. A. Brito da Cunha, Dr. L. E. Magalhães for criticism and important suggestions, and he is particularly thankful to the late Professor G. Dahlberg, pioneer of modern genetical investigation of human populations, for suggesting this kind of research.

In the collection of the data the author had valuable aid from different persons what must be acknowledged. Thanks are due to Professor P. Sawaya, Reverendissimo D. Paulo Rolim Loureiro, Reverendissimo D. Francisco Borja do Amaral, Father Luiz G. A. Cavalheiro, Mr. Omero Cordeiro, Mr. João Gnaccarini, Te. Abilio M. de Almeida, Dr. Walter Rizzo, Mr. José Cesar Gnaccarini, Mr. Johann Becker, Miss Natalia Grabusewicz, Mr. Heni Sauaya, Miss Candida P. Gomes, and Miss Therezinha Ungaretti.

Finally the author is deeply indebted to his wife, Sonia Guinsburg Saldanha, for constant stimulus, criticism, suggestions as well as for having helped him actively in the collection and analysis of the data.

However, the opinions and conclusions presented herewith are entirely of the author's responsibility.

* * *

The English version of this manuscript was accomplished in the Department of Human Genetics, University of Michigan. The author thanks Dr. James V. Neel and Dr. Richard H. Post for reading the English draft of the manuscript.

II. Theoretical introduction

Actually the methodological approaches for the investigation of the structure of human populations belong to two extreme categories: the island model and the model of isolation by distance. Both models can only give the information on the probable size of human reproductive units. The isolate model first proposed by Wahlund (1928) and developed by Dahlberg (1929, 1938, 1947, 1948) assumes completely isolated populations within which random mating (panmixia) is postulated. These breeding units were designated as genetic isolates. Since the isolate size is bounded by geographical and social barriers between the individuals, at random, some of the marriage in the isolate are expected to be consanguineous. This means, in other words, that the frequency of cousin marriages reflects the size of isolates. Postulating that the isolate size and the average number of fertile children are known, the frequencies with which cousin marriages of different types are to be expected on the hypothesis of random mating can be estimated. In human populations, relatives closer than uncles and nieces or aunts and nephews do not marry, and even this last type of consanguineous marriage seems often to be avoided. Therefore, the most inportant type of close relative marriage is that between first cousins. In an isolate of n individuals with the average number of fertile children equal to b, any individual, taken at random, from the isolate will have $b(b-1)$ first cousins among $n/2$ individuals of opposite sex. The probability of first cousin marriage then will be (cf. Dahlberg, 1947):

$$c = 2b(b-1)/n, \text{ hence } n = 2b(b-1)/c \qquad \text{II. 1. (a, b)}$$

Using similar reasoning the frequencies of other types of cousin marriage can be estimated provided that mating is entirely at random (Dahlberg, 1929). Assuming the relationship degree (r) through collateral lines to be symmetrical (cousins belonging to the same generation) or asymmetrical (cousins belonging to different generations), the frequency of any type of cousin marriage can be obtained by (cf., also Morton, 1955):

$$c(\text{sym}) = 2^r b^r(b-1)/n, \text{ and } c(\text{asym}) = 2^{r+2} b^{r+1}(b-1)/(2+b)n \qquad \text{II. 2. (a, b)}$$

However, Frota-Pessoa (1957) has shown that Dahlberg's formulae produce figures not completely exact, mainly when $b > 2$, as in the non-European populations. The estimates of isolate sizes by using the corrected formulae are about 50% higher than those obtained by Dahlberg's formulae.

The neighbourhood model developed by Wright (1943, 1946, 1951) assumes that the individuals are continuously and uniformly spread out on a bidimensional area, being isolated one from another only by distance. From the dispersion rate of the individuals, which is inversely related to the frequency of cousin marriage, an estimate of the breeding size can be obtained. If the parents (the couples considered) of the individuals come from a circular area with radius R, the neighbourhood size, N, is directly proportional to that area, and the grand-parents should come

from a larger area equal to $\sqrt{2}$ R, with a neighbourhood size equal to $2\mathcal{N}$. Similarly the ascendants of n^{th} generation of each individual should occupy an area $\sqrt{n}$ R with a neighborhood equal to $n\mathcal{N}$. Wright (1946) has shown that the distances between parent's and child's birth place (x) should be considered to exhibit a normal distribution with variance equal to $\sigma^2_{(x)}$. For the case of bisexual species with bidimensional distribution, as the human, the neighbourhood size can be estimated by:

$$\mathcal{N} = 4\pi\sigma^2 d = 12.6\ \sigma^2 d \qquad \text{II. 3.}$$

provided that the populational density, d, is homogeneous all over the area. This expression represents a circle with a radius equal to 2σ, having 86.5% of the parents of the individuals in its center. Similarly, the neighbourhood size can be estimated for the case of lineal distribution.

Both the isolate and neighbourhood approaches are based on several oversimplified assumptions that are difficult to apply to actual human populations. Because of the complexity of the demographic, cultural and social factors underlying the dynamics of human populations, a more exact approach must consider the semi-isolated distribution of human populations constituted by breeding units with variable rates of intermigration. The intermigration rates between two breeding units should be directly proportional to their distance. The main difficulties of the isolate concept rest on the non-randomness of cousin marriage, familial correlation in fertility and actual intermigration between human breeding units which makes it difficult to envisage the isolate boundaries. Similarly the difficulties related to the neighbourhood concept are the assumptions that isolation is maintained exclusively by distance and demographic density is homogeneous in the human species. The relative importance of each theoretical simplification can be evaluated only for the actual population studies. However, those models are based on inversely related parameters, the frequency of cousin marriage and migration (dispersion) rates, so that one must counterbalance another in the actual investigation of human populations.

Since this investigation is concerned with a previously isolated community, further consideration of the isolate concept must be discussed. The effect of subdivision of the population into small breeding units was first considered by Wahlund (1928). The subdivision effect on the genotypical distribution is similar but independent to inbreeding. Wahlund has shown that the homozygote frequency expected in a subdivided population is higher than that expected in a panmictic population. If the gene frequency in each of m equal isolates is q_i, and in the whole population is $\bar{q}=\Sigma q_i/m$, with a corresponding variance, $\sigma_q{}^2=(\bar{q} - q_i)^2/m$, the homozygote frequency in the whole population will be $q^2+\sigma_q{}^2$ instead of q^2 (see also Li, 1955, p. 297). The fraction $\sigma_q{}^2$ represents the increased homozygosis at the expense of heterozygosis. The greater the difference between gene frequencies in the isolates, the greater will be the variance value. The increase of homozygote frequency due to subdivision of the population is known as Wahlund's effect and it is likely to occur more frequently in populations sub-divided into small breeding units because of the sampling

variation. Table 1 shows a simple case of a population divided into two equal-sized isolates with variable differences in gene frequencies between them. Since these differences disappear after complete panmixia has occurred, genetical homogeneity is to be expected as an effect of increased dispersion.

The subdivision of the population into small breeding units is also important because the frequency of cousin marriage randomly expected is increased. Although

Tab. 1. Increase of homozygote frequency expected in a population divided into two equally sized isolates with variable difference in gene frequencies (see text)

Gene frequency				Homozygote frequency expected in the whole panmictic	Gene frequency variance variance	Homozygote frequency expected in subdivide population
In isolates			In the whole population			
I (q_1)	II (q_2)	Differ. (q_1-q_2)	$\bar{q} = \Sigma q_i/2$	($\bar{q}^2$)	$\sigma_q^2 = \frac{\Sigma(q_i - \bar{q})^2}{2}$	($\bar{q}^2 + \sigma_q^2$)
1	0	1	0.5	0.25	0.25	0.50
0.9	0.1	0.8	0.5	0.25	0.16	0.41
0.8	0.2	0.6	0.5	0.25	0.09	0.34
0.7	0.3	0.4	0.5	0.25	0.04	0.29
0.6	0.4	0.2	0.5	0.25	0.01	0.26
0.5	0.5	0	0.5	0.25	0	0.25

population as a whole can be considered endogamous, this is not true for each breeding unit. In a population of 1,000 individuals, for example, where the average number of fertile children is 2, the frequency of random cousin marriage is very low but it can be appreciable if the population is divided in 2, 5 or 10 breeding isolates of equal size (Table 2).

Tab. 2. Expected frequency of random first cousin marriage in a population of size 1,000 subdivided into 2, 5 and 10 equal-sized isolates, or breeding units, with the average number of fertile offspring equal to 2

Number of breeding units	Isolate size (n)	Expected frequency of first cousin marriage (%) [c=2b(b-1)/n]
1	1,000	0.4
2	500	0.8
5	200	2.0
10	100	4.0

Since homozygosis is increased by cousin marriage according to its particular coefficient of inbreeding (Wright, 1922) the frequency of recessive traits is likely to be higher in a population divided into small breeding units. The effect of inbreeding on the homozygote frequency can be measured as a weighted mean of all consan-

guineous marriages, according to their particular inbreeding coefficient, in relation to all marriages occurring in the population (cf. Haldane and Moshinsky, 1939). Postulating that $f_1, f_2, \ldots, f_n$ are the inbreeding coefficients of different types of cousin marriages, occurring with frequencies $n_1, n_2, \ldots, n_n$, the mean coefficient of inbreeding of the population is obtained by:

$$\alpha = (n_1f_1+n_2f_2+ \ldots +n_nf_n) / N=\Sigma_i(f_in_i) / \Sigma_i(n_i), \qquad \text{II. 4}$$

where N is the total number of marriages in the population.

Now, suppose that all types of cousin marriage occur at random in a small isolate. In this situation it is possible to calculate the α value expected as well as the absolute and relative increase of homozygote frequency for recessive genes. Assuming the frequency of a recessive gene is q and that of its corresponding allele is p, the increased homozygotic fraction will be αpq, so that the total frequency of homozygotes in the population wil be:

$$f(_{aa}) = q^2+\alpha pq = (1-\alpha)q^2+\alpha q \qquad \text{II. 5}$$

The homozygote frequency depends on the gene frequency and the inbreeding rates what means that it depends on the isolate size. The expected mean coefficient of inbreeding determined by random consanguineous marriages up to second cousins for isolates of different sizes can be calculated, by using Dahlberg's formulae, as well as its effect on the absolute and relative increase of recessive homozygotes. Table 3 shows that the homozygote increase is appreciable only for isolates of small sizes, assuming gene frequency is 0.1.

Tab. 3. Absolute and relative increase of recessive homozygotes expected from the random inbreeding rate (up to second cousins) in isolates of variable size, average number of fertile offspring equal to 2 and gene frequency equal to 0.1

Isolate size (n)	Mean coefficient of inbreeding expected in panmixia (α)	Increase of frequency of recessive homozygotes (αpq) (*)	Total frequency of recessive homozygotes ($q^2+\alpha pq$)	Relative increase of recessive homozygotes ($\alpha pq/q^2$)
—	(%)	(%)	(%)	(%)
100	2	0.18	1.18	18
200	1	0.09	1.09	9
500	0.4	0.036	1.036	3.6
1,000	0.2	0.018	1.018	1.8
5,000	0.04	0.0036	1.0036	0.36
10,000	0.02	0.0018	1.0018	0.18
100,000	0.002	0.00018	1.00018	0.018
1,000,000	0.0002	0.000018	1.000018	0.0018

* frequency of the recessive gene, q=0.1.

https://doi.org/10.1017/S1120962300020321 Published online by Cambridge University Press

It can also be shown that a relatively appreciable homozygote increase is only to be expected for gene frequencies equal to or below l.c.1. Table IV shows the absolute and relative increase of recessive homozygotes caused by random inbreeding rates expected in an isolate of 100 individuals with the average number of fertile children equal to 2. The α value expected in this situation is 0.02. However, lower

Tab. 4. Absolute and relative increase of recessive homozygotes expected from inbreeding rates (up to second cousins) in an isolate of size equal to 100, average number of fertile offspring equal to 2 and variable gene frequency

Gene frequency (q)	Frequency of recessive homozygotes (q^2)	Increase of frequency of recessive homozygotes (αpq) (*)	Total frequency of recessive homozygotes ($q^2+\alpha pq$)	Relative increase of recessive homozygotes ($\alpha pq/q^2$)
(%)	(%)	(%)	(%)	(%)
90	81	0.18	81.18	0.222
80	64	0.32	64.32	0.500
70	49	0.42	49.42	0.857
60	36	0.48	36.48	1.333
50	25	0.50	25.50	2.000
40	16	0.48	16.48	3.000
30	9	0.42	9.42	4.666
20	4	0.32	4.32	8.000
10	1	0.18	1.18	18.000

* Mean coefficient of inbreeding expected at random, $\alpha=0.02$

gene frequencies depend on the isolate size (n), so that the lowest gene frequencies cannot reach values below $1/2n$. Then, a very appreciable isolate effect on the homozygote frequency cannot be expected separately for each small breeding unit in the population, but it can be expected for the whole population. This situation seems to prevail for amaurotic idiocy in Sweden (Dunn, 1947).

The problem of isolate effect on the frequency of recessive homozygotes can be considered in a different way. Homozygotes for a rare recessive gene in the population with frequency q are produced by two different types of matings: marriages between unrelated heterozygotic subjects, with frequency q^2 [more exactly $q^2(1-\alpha)$]; and by consanguineous marriages. Assuming that the particular coefficient of inbreeding related to an undetermined cousin marriage is f, recessive homozygotes are produced by genes inherited from one common ascendant, with frequency fq, and by genes inherited from non-common ascendants, with frequency $(1-f)q^2$. Since the frequency of that type of cousin marriage is c, the frequency of recessive homozygotes produced

by them in the population is $c[fq+(1-f)q^2]$ and the proportion of consanguinity among parents of recessive homozygotic individuals is:

$$k = c[fq+(1-f)q^2]/q^2 \quad (1)$$

For the particular case of first cousin marriage, the formula is:

$$k = c(1+15q)/16q$$

The Table 5 shows the proportion of first cousin marriages among parents of affected for recessive traits expected from different gene and first cousin marriage frequencies in the population.

For the better known recessive genes, the *k* values range from 10% to 80%. This should indicate the isolate effects due to the subdivision of the population, since

Tab. 5. Percent of first cousin marriage, among parents of rare recessive homozygotes, expected for three different gene frequencies and four different rates of first cousin marriage in the population

Frequency of recessive homozygotes (q^2)	Gene frequency (q)	k* when frequency of first cousin marriage in the population (c) is:			
		0.005	0.01	0.02	0.04
0.001	0.032	1.45	2.89	5.78	11.56
0.0001	0.01	3.59	7.19	14.37	28.74
0.00001	0.0032	10.23	20.47	40.94	81.88

* $k = [c(1+15q)/16q]$.

the *k* values increase in proportion to the frequency of first cousin marriages in the population. The mean frequency for several abnormal recessive genes should be taken as about 0.01 (as for example albinism) so that the breakdown of isolates into an unique panmictic population should reduce the frequency of abnormal recessive traits by about 15% (probable range between 4% and 30%). Even so, the figures can actually correspond to a small absolute number of affected.

Now, it must be considered that the isolate concept was introduced as statistical one, probably because it was first envisaged to be applied to stable European populations. However, in its historical process, either the isolates shall be relatively stable in size, among undeveloped rural and primitive areas, or they can increase rapidly as a result of demographic and social changes. The increase of the isolate size was not considered enough by previous investigators but it is an important problem among American populations. The size increase of the isolates should be within their boundaries, caused by the relative increase of birth rates or that effect should be deter-

(1) A more exact formula must have q^2+apq as the denominator.

mined by immigration into the previously isolated breeding units. Both phenomena seem at present to be occurring rapidly in America.

The increase in birth rate, without regard to differential fertility, does not change the genetic makeup of the isolates, while this is not the case for differential immigration. If the size of a breeding isolate, n_1, changes to n_2, the absolute number of each genotype increases but its relative frequency is not changed. Assuming that the average number of fertile offspring in the population changes from $b_1=2$ to $b_2=4$, the isolate size increases from n to 2n. The average number of first cousins in the population will be $[2b_2(b_1\text{-}1)]$ because uncles and aunts occurring with the frequency $2(b_1\text{-}1)$, belong to the previous generation when $b = b_1$, however having an average of $b = b_2$ fertile offspring (first cousins). As it can be seen in Table 6, the frequency of first cousin marriages, estimated by the formula $c = 2b_2(b_1\text{-}1)/n_2$

Tab. 6. Frequencies in successive generations of first cousin marriages expected where the initial number of fertile offspring, 2, in an isolate of initial size, 100, increases by 100% in the first generation

	Initial generation	First generation	Second generation	Third generation	Fourth generation
Average number of fertile offspring (b)	2	4	4	4	4
Average number of first cousins [2b(b-1)]	4	8	24	24	24
Isolate size (n)	100	200	400	800	1600
Frequency of first cousin marriage [c = 2b(b-1)/n]	4%	4%	6%	3%	1.5%

does not change in the first generation following the increase in birth rate. However, in the second generation, the average number of uncles and aunts increases, being $2(b_2\text{-}1)$, so that the frequency of cousin marriages, $c = 2b_2(b_2\text{-}1)/n_3$ also increases in this generation (2). The greater is the increase of *b*, the greater will be the increase of the frequency of first cousin marriages. For example, if $b_1 = 2$ and $b_2 = 6$, *c* changes from 4% in the first generation to about 7% in the second generation. From this latter generation the frequency of first cousin reaches lower and lower levels, as a result of constant increase of the population. The Table 6 shows the variation of the frequency of first cousin marriage in an isolate of initial size 100, when the initial average number of fertile offspring 2 increases to the level of 100%. As a general conclusion the increase in birth rate in the isolate can determine a fairly higher frequency of first cousin marriage in the generation just following the birth

(2) Using the formula corrected by Frota-Pessoa (1957) the increase probably should be higher. The writer is indebted to him for the discussion that made clearer this problem.

rate increase, but soon this effect changes in the opposite direction because of the constant increase of the isolate.

The increase of dispersion rates of human beings in successive generations is, however, of relatively greater theoretical and practical importance. The removal of geographical and social barriers, permitting a higher migratory flow between previously isolated breeding units is a relatively recent phenomenon determined by the improvement of economic conditions and deep change in the available means of communications of human populations in recent generations. From a genetic point of view, this process was first considered by Dahlberg (1947 and previous papers) that named it the *break down of isolates.*

The changes underlying the breadkown of isolates are multiple. Firstly, the removal of barriers between two isolates can be considered only with respect to the exchange of genes. Wright (1949) defines immigration pressure as the frequency of immigrants who, upon marrying, are able to reproduce. The effect of immigration on the gene frequency of a breeding population is represented by: $d_q = m(q-q_i)$, q_i being the gene frequency among the immigrants and q, in the population and m, the immigration rate. As it can be seen in Table 7, the intensity of d_q depends

Tab. 7. Variation in gene frequency (d_q) expected for variable immigration rates and differences in gene frequency between the population and immigrants

Immigration rate	Difference between gene frequencies in the population and among the immigrants ($q-q_i$)			
(m)	0.10	0.20	0.50	1.00
0.05	0.005	0.01	0.025	0.05
0.10	0.01	0.02	0.05	0.10
0.20	0.02	0.04	0.10	0.20
0.50	0.05	0.10	0.25	0.50

on both the difference ($q-q_i$) and the relative numbers of immigrants. It is likely that distance between populations is correlated with genetical divergence, so that the effect is to be higher when gene exchange occurs between remote populations.

From the eugenical point of view, the effect of the immigratory flow between previously isolated breeding populations on the frequency of rare recessive genes seems to be an important result of the breakdown of isolates (Dahlberg, 1939). If a large population is divided into several smaller breeding units, it is expected that mutations for rare recessive genes occur only in some isolates, since mutation is rare event. In this situation, the gene will have a greater chance for homozygosis. Similarly, the breakdown of isolates is more likely to occur between breeding units that differ in the frequency of rare recessive genes. Moreover, most mutants should exist in only a few isolates. Considering separately an isolate where a mutation occurred, the breakdown of isolates decreases the frequency of a rare gene according to the

expression, $q_1 = q_0/(1+a)$, (cf. Dahlberg, 1947), q_0 being the initial gene frequency and *a* the relative size increase of the isolate determined by removal of previous barriers.

The breakdown of isolates probably commenced a long time ago, even when human populations were spread out as very small breeding units in pre-historical times. However, the intense migratory movements between populations belonging to remote regions are deliberate recent events. As a matter of fact, these movements are causing profound changes in the genetical structure of modern human populations. Among the main genetical effects that should be accounted for the breakdown of isolates are the following:

a) Decrease of random inbreeding rates expected as a consequence of the increase of population size.

b) Decrease of variance of gene frequency (Wahlund's effect).

c) Decrease of homozygote frequency expected as a result from *a* and *b*.

d) Increase of heterozygote frequency for both rare and common genes, permitting new genotypic combinations of monogenic and polygenic systems. This effect is marked in the case of heterotic loci.

e) Decrease in the probability of random drift as a consequence of the increased population size.

It should be emphasized that the breakdown of isolates is particularly important for rare recessive genes. Since the homozygote frequency depends on the inbreeding rates of the population (i. e., on the breeding isolate size) the breakdown of isolates should decrease the frequency of affected persons for rare recessive abnormalities and diseases. This means that the equilibrium between mutation and selective elimination rates is disturbed. Haldane (1939) suggested that it can account for the fact that human recessive mutants are rarer than dominant ones (Levit, 1936). In a population for a long time divided into small breeding units the homozygote frequency to be expected is $x = q^2+\alpha pq$, with q the frequency of a rare recessive gene, p that of its allele, and α the mean coefficient of inbreeding in the population. In this situation, frequency of a recessive gene is approximately obtained by $q = \sqrt{[(\alpha^2+4x)-\alpha]}/2$, instead of $\sqrt{q^2}$. When the equilibrium is disturbed by the decrease of inbreeding rate the relationship $x = \mu/s$, with μ the mutation rate, and s the selective coefficient of the gene, no longer holds. As selective elimination depends on the inbreeding rate, that is decreased, gene frequency increases steadily to the equilibrium for the new lower level of inbreeding rate. However, Haldane has shown, on theoretical grounds, the new equilibrium is reached only, by the better known human abnormal recessive genes, after hundreds of generations. As a matter of fact the decrease of inbreeding rates is a relatively recent and generalized event, occurring throughout the world, particularly in western countries (cf. Saldanha, 1960). Therefore it is unrealistic to postulate equilibrium for human recessive abnormal genes.

III. The community investigated

Demographic Data and The Origin of The Present Population

Differential immigration is becoming one of the most effective agents for human evolution in recent generations. America is a suitable area for this kind of research since immigrants of different genetic origin as Negroes and Europeans of several nationalities, have arrived at that part of the world. In Brazil, immigration has been very active since 1880 and the bulk of migratory movement from Europe has occurred from 1900 to 1930, as consequence of the slave freedom law, promulgated in 1888. The total number of immigrants arriving at Brazil from 1819 to 1954 is about 5 million (Carneiro, 1950). They are mainly represented by Italians (30%), Portuguese (29%), Spanish (12%), Germans (5%), Japanese (4%), Russians (2.5%), Austrians (2%), Syrian-Lebanese (1.5%), Polish (1%), and other nationalities less than 1%. These immigrants have settled to a greater extent in the southern states. Italians, Spanish, and Japanese have mainly settled at the State of São Paulo. Russians, Germans and people of teutonic derivation have chosen the most southern states such as Paraná, Santa Catarina and Rio Grande do Sul. Portuguese have mainly come to the States of Rio de Janeiro and São Paulo.

It is of interest to know the effect of those immigratory movements on the genetical structure of previously settled populations. From a previous investigation on inbreeding rates of communities in areas under different demographic and social conditions (Saldanha, 1960), the parish of Capivari was selected because it has received a very large amount of non-Brazilian immigrants during the last two generations.

The geographical location of the parish of Capivari is shown in Figure I. This parish belongs to the Diocese of Piracicaba, in which the immigratory movement was quite active. The community is 165 km, from São Paulo City, about 22°59'56" S. latitude and 47°20'19" W. Gr. longitude, and it is 512 meters high on the inner plateau of the State of São Paulo.

The Capivari village was founded in 1832, and since its foundation, the main economical activity has been the sugar cane culture (Campos, 1952). The examination of catholic marriages records for the period of 1822-26 showed that among 272 individuals (136 marriages) about 50% were born in the community, 40% in neighbour villages, and about 10% birth places could not be determined. The parish chapel from which these records were taken was built in 1820.

According to General Census of 1950, the district that corresponds to the parish of Capivari has 14,052 habitants and its area is 319 Km^2, resulting in ca. 44 individuals per km^2. The rural population is relatively high (49.8%) and the ethnical distribution in the community is ca. 89% whites, 2% Mulattos, and 9% Negroes. The frequency of Mullatos is relatively low as compared to that in the state (3%) and nation (26.5%). This fact is likely to be due to complete segregation of Negroes by cultural, social, and economical factors.

Immigration to the community was highest from 1880 to 1920. Most large land

properties that belonged to Brazilian owners became small sugar cane farms acquired by the immigrants. The relative numbers of non-Brazilian immigrants (estrangers) who entered the community can be obtained from demographic censuses of the municipality. The percent of estrangers (in relation to the whole population) was nil in 1836 (non-official census), 0.7% in 1872, 8.2% in 1890, 16.7% in 1920 and 3.5%

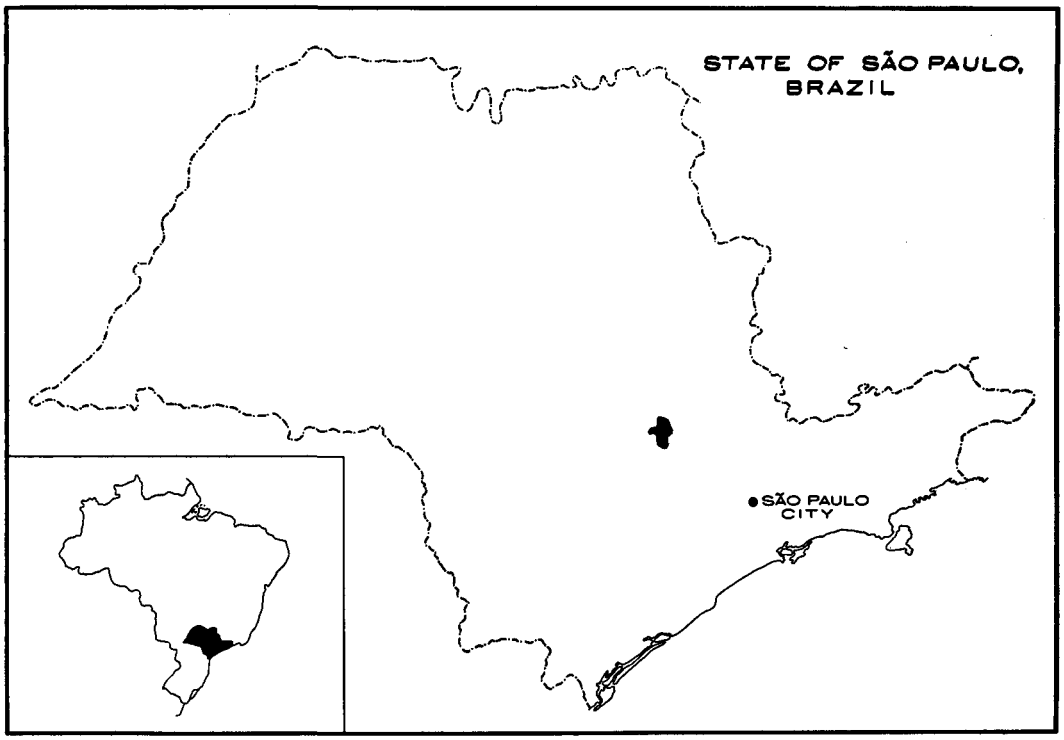

Fig. I. Geographical location of the parish of Capivari

in 1950. It is of interest to know the national derivation of these immigrants, mainly those who married in the community. Table 8 shows the results of examination of Roman Catholic marriage records of different periods in the community. As it can be seen in Table 8, non-Brazilians were practically absent up to 1890 when the total percent of estrangers in the community was about 27%. Among them ca. 89% were of Italian derivation. Another source of information can be taken from the General Census of 1950. In that year, the frequency of Italians in relation to all aliens living in the community was about 67%. This figure is very high as compared to that for the state (23%) and nation (18%).

An investigation of the origin of 3,253 young men recorded in the office for Army Obligatory Recruitment in the community for a twelve-year period (1945-1956)

showed that the Italian contribution to the present population was even higher. National origin as recognized by parental name (3), is shown in Table 9. It should be concluded that 50% of the individuals belonging to the present generation has at least one parent of Italian derivation.

Tab. 8. Frequency of non-Brazilians (immigrants) marrying into the community during different periods, according to their Nationality

Period	N [1]	Italian (%)	Spanish (%)	Portuguese (%)	Other Nationalities [2] (%)	Total (%)
1830-35	140	0	0	0	0	0
1860-65	306	0	0	1.96	0	1.96
1890-95	1110	22.70	1.08	1.26	2.17	27.21
1920-25	1262	5.70	2.22	0.87	0.47	9.27
1950-55	1618	0.12	0	0.06	0.25	0.43

[1] Number of individuals married in the community.
[2] Syrian, Austrian, Swiss, German and French.

Tab. 9. Origin of the present population according to parental ancestry of 3253 young men recorded for obligatory military service in the community of Capivari (see text)

Parental ancestry	Percent (approx.)
Brazilian	23
Italian	41
Brazilian and Italian	16
Other whites	9
Negroes and Mulattoes	11
	100

An attempt to identify the birth places of the Italian immigrants was made. Examination of Roman Catholic marriage records in the community for the periods 1890-95, 1920-25, and 1950-55, showed that practically all Italians arriving in the community were born in northeastern Italy. Figure II shows the main Italian regions from which they emmigrated, were: Veneto (provinces: Venezia, Padua, Treviso, Rovigo, and Verona), Lombardia (Provinces: Mantua and Milan) and Emilia-

(3) In the community women keep the maiden name after marriage.

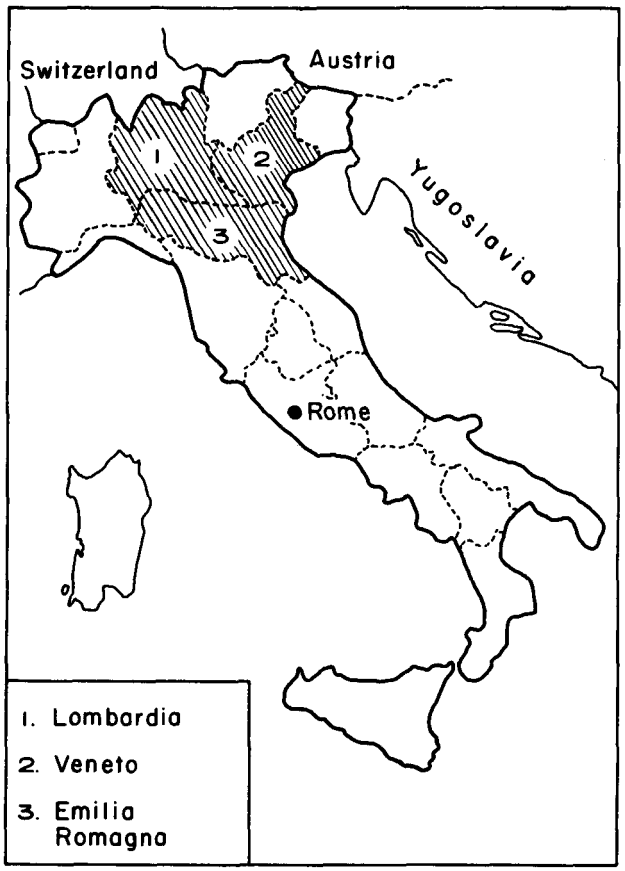

Fig. II. Locations of regions in Italy which furnished most of migrants marrying in the parish of Capivari

Romagna (provinces: Ferrara, Bologna, Parma, and Modena). Invariably they came from rural places and exhibit very distinct physical features such as: tall stature, slender body, brachycephaly, light eyes, and light skin, that indicate they are " nordic " with alpine and dinaric components (Coon, 1939).

Another point of genetic interest is the fecundity in the community. In spite of the difficulties of obtaining reliable data on this subject, this information must be obtained for further genetical considerations. According to the General Census of 1940, the community had about 1800 fertile women between 15 and 39 years of age. This suggests that the parents living during a twenty-five year period number about 3,600. The relation between the number of fertile women and the total number of children born during a generation corresponds roughly to the average number of children per couple, that is 3.95. However, this figure should be lower, provided that it does not take into account the infertile marriages (the infertile adult women). The relation between the number of fertile women and the total number (fertile plus infertile) of women above the age of 45 years suggests that the rate of infertile couples is not higher than 10%. These findings are consistent with the average number of children per family, estimated from the female reproduction rate ($r=1.75$) for the whole State of São Paulo (Mortara, 1955). As the sex-ratio prevailing in the State of São Paulo is 1.05, the average number of children per family is 3.59 (2.05×1.75). Probably the actual value to be found in the community is somewhat higher. A sample of 247 families taken from the population showed that the average number of children per family should be variable according to the origin of the parents. Among 60 families of pure Brazilian derivation the mean is to 3.88, among 72 families of pure Italian ancestry the mean is 4.26,

https://doi.org/10.1017/S1120962300020321 Published online by Cambridge University Press

among 59 " hybrid " families the mean is 4.26, among 59 " hybrid " families the mean is 3.53 and among the 56 remaining families the mean is 4.87. However, the sample is not large enough to make the differences statistically significant. The overall mean for the 247 families, 4.13 ± 0.14, is, however high as compared to that for the whole State. The average number of children per family, does not fit a Poisson distribution even making the frequency of infertile couples equal to 5% ($\chi^2_{(7)} = 34.3$ $P < 0.01$). The variance (4.99) is slightly higher than the mean.

It must be observed that there are no deep cultural, social, and economical differences between individuals of " pure " Brazilian origin and those having at least one Italian ascendant. All share the same privileges in the community. Religion is another factor making for social homogeneity. About 97% of the individuals are Roman Catholic. This is a relatively high frequency as compared to ca. 91% in the state and 93.5% in all Brazil, according to the General Census of 1950.

IV. The analysis of the breakdown of the isolate

Immigration Rates and Frequencies of Consanguineous Marriages

The breakdown of isolates seems to be a generalized process, but the recent and sudden increase of dispersion rates of man in western countries so marked, is a modern phenomenon determined by the impact of civilization. However, in Europe the breakdown of isolates has occurred as a steady process, and differs in intensity from the " organized " immigratory movements to America. In the community investigated it was possible to detect the process by using two sources of data: the dispersion rates, and frequencies of cousin marriages in different periods.

The Immigration Rates

Immigration rates are a direct indication of the isolation of the population. Data related to the rates of immigrants marrying into the community are more reliable than crude immigration rates and they can be obtained from Roman Catholic marriage registers. The birth places of the individuals marrying into the community were identified for the following periods: 1830-35 (including part of 1839), 1860-65, 1890-95, 1920-25 and 1950-55. Frequencies of the following classes were obtained: marriages between Brazilians (BxB), marriages between foreigners (FxF), marriages between Brazilians and foreigners (BxF), and total frequency of non-Brazilian immigrants (foreigners, F). It was also possible to classify birth places of grooms and brides marrying into the parish of Capivari as being in or out of the community. The following combinations were obtained: groom and bride born in the parish of Capivari (ii), only the groom born in the parish of Capivari (io), only the bride born in the parish of Capivari (oi), both born out of the parish of Capivari but in different parishes (oo), and both born out of the parish of Capivari but in the same parish ($\overline{oo}$). Using

this information the overall (Brazilian and non-Brazilian) rate of immigrants marrying within the community was obtained by the expression: $m = [(io)+(oi)+2[(oo)+(\overline{oo})]]/2N$, with N representing the total number of marriage registers examined. The exogamy index (cf. Freire-Maia, 1952) was estimated by the expression, $e = [(oi)+(io)+(oo)]/[(ii)+(\overline{oo})]$. All results, obtained for different periods, are shown in table 10. As can be seen in this table, the immigration rates reached the

Tab. 10. Immigration rates into the parish of Capivari during five different periods as shown by national origin and in-parish-out-parish origin of spouses

Period	N.	Marriage classes (%)			F	Marriage combinations (%)					m	e
		B+B	F+F	B+F	(%)	ii	oi	io	oo	$\overline{oo}$	(%)	
1830-35	70	100.0	0	0	0	68.6	11.4	7.1	8.6	4.3	22.1	0.37
1860-65	153	97.4	1.3	1.3	2.0	48.4	26.1	9.8	11.1	4.6	33.7	0.89
1890-95	555	69.7	24.2	6.1	27.2	36.0	22.7	7.4	21.8	12.1	48.9	1.08
1920-25	631	84.8	3.3	11.9	9.3	50.9	23.6	11.6	11.1	2.9	31.5	0.86
1950-55	809	99.1	0	0.9	0.4	51.2	22.4	12.4	10.6	3.4	31.5	0.83

N = Number of marriage registrations examined.
B = Brazilian.
F = Foreigner
ii = Both groom and bride born in the community.
oi = Bride only born in the community.
io = Groom only born in the community.
oo = Both groom and bride born outside the community but in different parishes.
$\overline{oo}$ = Both groom and bride born outside the community but in the same parish.

Total immigration rate, $m = \frac{(io)+(oi)+2[(oo)+(\overline{oo})]}{2N}$

Exogamy index, $e = \frac{(oi)+(io)+(oo)}{(ii)+(\overline{oo})}$

highest levels about 1890-95. Since then there has been general and uniform decrease of immigration rates. Therefore the highest frequency of marriages between Brazilians and foreigners is found in the subsequent generation (1920-25). In spite of the decrease in frequency of non Brasilians since 1890, the number of their descendants should be increased more and more after that time, as can be concluded from table 9.

Before the immigratory movement of 1830-35 the community displayed a very low exogamy index, because about 80% of those marrying in the parish of Capivari had been born there. From 1920, the figures seem to indicate a general return to stability. It is interesting to note that the frequency of (io) marriages was substantially lower than the frequency of (oi) marriages. This indicates that female immigrants were rarer than males. It must be pointed out that the increased frequency of non-Brazilian immigrants is followed by a similar increase of frequency of Bra-

zilian immigrants into the community. Of course, this fact should be due to social and economical changes of the community life after the non-Brazilian immigration.

Similar data were obtained for parishes under different demographic and social conditions in the state of São Paulo as will be presented elsewhere. Among the communities investigated, the parish of Natividade was selected because it shows different features in contrast with Capivari. Table 11 shows a comparison of immi-

Tab. 11. Comparison between the immigration rates in the parishes of Capivari and Natividade for corresponding periods (for explanation see text and Table 10)

Parish	N.	Marriage classes (%)			F (%)	Marriage combinations (%)					m (%)	e
		B+B	F+F	B+F		ii	oi	io	oo	$\overline{oo}$		
Capivari	2148	87.2	7.3	5.5	10.1	47.0	23.1	10.7	13.7	5.6	36.2	0.90
Natividade	1128	99.2	0	0.8	0.4	61.4	18.0	8.2	5.0	7.4	25.5	0.45

gration rates between Capivari, during the periods of 1860-65, 1890-95, 1920-25, 1950-55, and Natividade, during four similar periods. As can be seen in that table, Capivari exhibits quite different immigratory features in comparison with Natividade, a relatively isolated community on the mountains (Serra do Mar) of northeastern São Paulo.

Application of the "Neighbourhood" Concept to the Present Data

As discussed in the introduction, an estimate of the probable neighbourhood size (N) can be obtained (Wright, 1946) provided that the variance (σ^2) of the distances (x) between parent's and child's birth places as well as the demographic density (d) of the population are known, by using the formulae $N = 4\pi\sigma^2 d$ for areal continuity and $N = \sqrt{\pi\sigma d}$ for lineal continuity. Since "neighbourhood size" is a circular area of radius 2σ (in the case of areal continuity), the increase of variance of dispersion, that is inveresly related to the inbreeding coefficient of the population, is an important indication of the increase of genetic homogeneity of the population. In other words, the distance between individuals is correlated with the genetic diversity because, as the distance increases the choice of mates decreases. However, to make use of the neighbourhood model it is necessary to assume that the dispersion of individuals, that is, the distance between parent's and child's birthplaces, shows a normal distribution. Departures from this assumption as well as from the assumption of homogeneous demographical density are found in human populations since they are stratified by geographical, ecological, cultural and social factors.

Recently some investigators have attempted to obtain information on the dispersion patterns of human populations in different regions (Sutter and Tran-Ngoc-Toan, 1957; Cavalli-Sforza, 1958; Alström, 1958; Fraccaro, 1959). From these studies it

is apparent that human dispersion shows a clearly leptokurtic distribution. It was possible to collect data on dispersion rates for some parishes of the state of São Paulo, which will be reported elsewhere. In the present investigation an analysis of distance is shown to evidence that the breakdown of the isolate can be investigated in terms of dispersion rates.

The birthplace of 2200 persons marrying in the community were collected from Roman Catholic marriage registers, covering 5 different periods: 1830-35, 1860-65, 1890-95 1920-25, and 1950-55. It is assumed that the marriage place corresponds to the birthplace of children born to those marriages. The mean distance for male ($\bar{x}_m$), female ($\bar{x}_f$) and both sexes ($\bar{x}_t$), total variance of distance (σ^2_x) were estimated separately for native born Brazilians and total population because non-Brazilian immigration is a more complex process. To estimate the distance between birth and marriage places for each individual, the direct distance in km. was measured on maps. Table 12 shows the results obtained for each period investigated. As can be seen in that table, mean distance and variance of distance increased from 1830 to 1895. In later periods the figures show a relative stability but on a level higher

Tab. 12. Mean distance and variance of distance between birth and marriage places of the individuals marrying in parish of Capivari in different period

Period	sample	$\bar{x}_m$	$\bar{x}_f$	$\bar{x}_t$	σ^2_t
1830-35	A	12	10	11	411
	B	12	10	11	411
1860-65	A	24	12	18	1,062
	B	190	177	183	1,297,156
1890-95	A	148	36	91	127,539
	B	2,971	2,603	2,787	18,607,930
1920-25	A	31	18	24	17,503
	B	1,237	553	895	7,720,820
1950-55	A	36	25	30	22,956
	B	114	25	69	429,547
Total	A	56	24	39	41,241
	B	1,163	838	1,000	8,372,511

A = not including foreign-born. B = including foreign-born.
$\bar{x}_m$ = mean distance between birth and marriage places for males.
$\bar{x}_f$ = mean distance between birth and marriage places for females.
$\bar{x}_t$ = mean distance between birth and marriage places for both sexes.
σ^2_t = variance of distance between birth and marriage places for both sexes.

than before 1860. The increase is extremely high when non-Brazilians are taken into account but they present a very artificial situation. From these results, it can be concluded that the increase of non-Brazilian immigration was followed by a corresponding increase in dispersion rates of the Brazilians.

It is of interest to compare the figures for the parish of Capivari with those of communities in different situations. The parish of Natividade displays very distinct features and it is represented here for comparative purposes. While it seems to be hard that the neighbourhood size calculated from variance found for the parish of Capivari has a sound sense, that estimate was obtained by assuming that demographic density is uniform and arbitrarily taken as unity. To estimate the mean distance, variance of distance and neighbourhood size, the non-Brazilian individuals were not taken into account. Table 13 shows the results obtained for the parishes of Capivari and Natividade in the period 1905-1955. It must be observed that the total frequen-

Tab. 13. Comparison of means and variances of distance between birth and marriage place of Brazilians marrying in the parishes of Capivari and Natividade, 1905-55 (for explanation see text and table 12)

Parish	$\bar{x}_m$	$\bar{x}_f$	$\bar{x}_t$	σ_t^2	Neighbourhood size *
Capivari	57	24	40	42,426	534,568
Natividade	13	7	10	1,126	14,188

* Assuming areal continuity and homogenous density equal to 1, by the formula, $N = 12.6\sigma^2 d$

cies of cousin marriages for the same period were 6.73% and 1.42%, respectively, in those parishes.

From table 13 it appears that neighbourhood concept as applied to civilized populations, particularly those under complex demographic cultural and social conditions may not produce sound results. Probably the variance of distance is strongly influenced by non-randomness of dispersion rates. Another important factor distorting the estimates of neighbourhoos size is the non-homogeneity of density among human populations. The estimates of neighbourhoos size based on variances obtained for primitive (Roberts, 1956-1957) and civilized populations (Sutter and Tran-Ngoc-Toan, 1957; Alström, 1958) are, nevertheless, high as indicated by the frequencies of cousin marriage. The figures obtained by Sutter and Tran-Ngoc-Toan (1957) for French parishes should display even greater discrepancies since the dispersion rates were measured from consorts' dwelling places instead of their birthplaces.

However, the distance between birth and marriage places should be valuable as a parameter for dispersion, provided that the limitations involved in this information are taken into consideration. It is likely, for example, that mean parental distance

is lower for consanguineous partners than for unrelated ones. Table 14 shows the mean distances calculated for different types of consanguineous marriages in the parish of Capivari in the past and present centuries. The overall mean distance for consanguineous partners is 25 km. This value is positively lower than that found for all marriages occurred in the community, in both cases with (ca. 1000 km) or without (ca. 40 km) the non-Brazilians being included in the calculations. (cf. Table 12).

Another problem brought up by figures in table 14 is concerning to randomness of frequencies of different types of cousin marriages. Assuming complete randomness in choice of mates, it might be expected that all types of consanguineous marriage presented similar mean distance between birth and marriage place but instead, this is not the situation found. As consanguineous individuals of more distant degree are related to a common ancestor many generations ago, they should be more

Tab. 14. Mean distance in kilometers between birth and marriage place of Brazilian partners of six types of consanguinity marrying in the parish of Capivari during the XIX and XX centuries

Period	Type of consanguinity						
	½ C	1 C	1½ C	2 C	2½ C	3 C	TC
XIX century	5	14	12	13	16	16	14
XX century	0	61	8	7	—	—	36
Total	4	42	9	10	16	16	25

scattered than close consanguineous individuals would be. If this is true, it is likely to find a correlation between mean distance and degree of consanguinity of the couples, as it seems to be the situation in the XIX century. However, because the intense mobility of the individuals in the XX century distance between consanguineous individuals of distant degree became so large that only those living relatively near can marry. Data in Table 14 could be interpreted this way.

At present, extensive data are not available for more definite conclusions on the applicability of the theoretical models for treating human populations from a genetic point of view. Therefore elaboration of a more appropriate general theory has to await more complete and accurate empirical data from actual populations under different geographical and social conditions.

The Frequency of Cousin Marriage

Since the inbreeding rate should reflect the isolate size it is important to appreciate the temporal trends in frequencies of cousin marriage in the population. For this purpose data from Roman Catholic and civil marriage records in the community were collected. Unfortunately this kind of information was not available for the

whole XIX century. Parish archives were examined for the periods: 1830-35, 1860-65, and 1890-1955, and a total of 8026 Roman Catholic marriage registrations were obtained. Cousins closer than third degree are not permitted to marry by cannon law, unless the bishop's dispensation is obtained. The types of consanguinity that can be identified by means of such dispensation are designated as follows: 1/2 C, marriage between uncles and nieces or between aunts and nephews; 1 C, first cousins; 1 1/2 C, first cousins once removed; 2 C, second cousins; 2 1/2 C, second cousins once removed; 3 C, third cousins; TC, total consanguinity; and N, the total number of marriage registers examined. Multiple cousin marriages were considered as respective separate single marriages and counted accordingly. By using the frequencies of different types of cousin marriages the mean coefficient of inbreeding of the population (α) was calculated.

Comparative data on frequencies of cousin marriages from civil records were obtained for the following periods; 1890-96, 1920-26, and 1950-56. Until 1917 consanguinity was assigned in register books. After that time, the identification of consanguineous marriages was only possible for first cousins because they share one pair of grandparents.

Table 15 shows the frequencies of different types of cousin marriages, the mean coefficient of inbreeding, and the isolate sizes estimated from the frequencies of first

Tab. 15. Frequencies of different types of cousin marriage, mean coefficient of inbreeding and isolate size estimated from the frequencies of first cousins in different periods

Period	N.	Type of consanguineous marriage (%)							α ($x*10^5$)	Isolate size *
		½ C	1 C	1½ C	2 C	2½ C	3 C	Total		
1830-35	70	1.4	4.3	—	—	—	4.3	10.0	463	558
1860-65	164	—	9.8	2.4	2.4	3.0	9.8	27.4	786	245
1890-95	563	0.9	2.3	0.7	1.6	0.9	1.4	7.8	315	1043
1920-25	631	—	0.8	0.5	—	—	—	1.3	64	3000
1950-55	810	—	0.7	0.5	1.4	—	—	2.6	83	3428

* Assuming b = 4.

cousin marriages in different periods. To calculate the isolate size the average number of fertile offspring was considered equal to 4. While this figure should not have been constant for the whole period analysed, the estimates of isolate sizes can give a rough idea of the relative isolation of the community.

The frequencies of cousin marriage in different periods are extremely heterogeneous ($\chi^2_{(4)} = 66.3$, $P<0.0001$). The highest frequencies of first cousin marriages occurred in the period 1860-65, indicating that the community was relatively small and isolated before the non-Brazilian immigratory movement started.

Table 16 shows more detailed temporal trends of the frequencies of first cousin

marriages, total consanguinity and α values, from 1890-1955. As it can be seen there was a steadily uniform decrease of cousin marriage rates from 1900, just when immigration into the community became heavier. Since 1920, the inbreeding rates seem to have been stable on a very low level. The 20 year period (1900-20) of rapid decrease

Tab. 16. Frequencies of first cousin marriage, total consanguinity and mean coefficient of inbreeding for five year periods from 1890 to 1955 in the parish of Capivari

Period	1890 -94	1895 -99	1900 -04	1905 -09	1910 -14	1915 -19	1920 -24	1925 -29	1930 -34	1935 -39	1940 -44	1945 -49	1950 -54	1955	1890 1919	1920 1949	Total*
N.	474	486	495	586	614	711	561	440	544	725	680	666	663	147	3366	3616	8026
1 C (%):	2.3	2.9	3.0	1.2	1.1	0.8	0.7	0.7	0.2	0.3	0.4	0.2	0.8	0.7	1.8	0.4	1.2
TC (%) :	8.4	6.4	4.4	3.2	2.1	1.3	1.2	0.7	0.2	0.8	1.0	0.6	2.6	2.7	4.0	0.8	2.9
α (x 10^5) :	338	228	225	122	97	64	61	43	11	30	37	35	80	96	166	36	114

* It also includes data from the periods 1830-35 and 1860-65.

in frequencies of cousin marriage can be related to the time of most active non-Brazilian immigration (see above).

The decrease of inbreeding rates can be evaluated very clearly in Figure III.

According to Freire-Maia (1957) the lowest inbreeding rates in Brazil were found in the State of São Paulo. Table 17 shows, however, that the frequencies of cousin

Tab. 17. Comparison of frequencies of cousin marriage, mean coefficient of inbreeding and isolate size estimated from the frequency of first cousin marriage, observed in the whole state of São Paulo (samples from different dioceses) and those found in the parish of Capivari, for a corresponding period

Region	Period	N.	Type of consanguineous marriage (%)					α (x*10^5)	Isolate size *
			½C	1 C	1½C	2 C	TC		
State of São Paulo **	1939-55	—	0.02	0.85	0.20	0.30	1.37	67	2188
Parish of Capivari	1920-55	4426	0.02	0.45	0.23	0.41	1.11	44	4132

* Assuming b = 3.59.

** Data from Freire-Maia (1957).

marriages in the parish of Capivari are substantially lower than those found for some dioceses in the State of São Paulo. The mean isolate size for the whole state is one half of that estimated for the parish of Capivari, assuming that the average number of fertile offspring is the same.

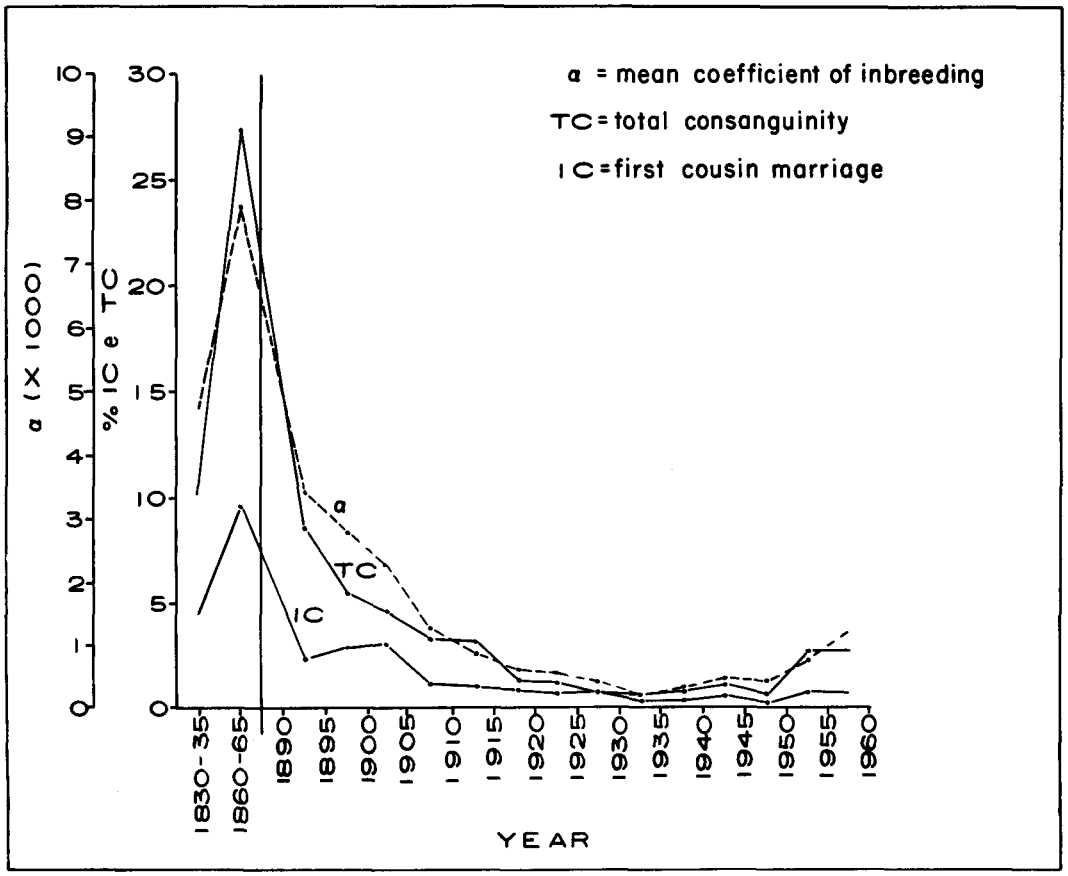

Fig. III. Temporal trends of inbreeding rates in the parish of Capivari

The present writer has investigated the inbreeding rates of regions of the State of São Paulo with different frequencies of non-Brazilian immigrants (Saldanha, 1960). Among the communities investigated, the parish of Natividade, a relatively isolated one located on the mountains (Serra do Mar) of northeast of São Paulo, showed a negligible crude rate of non-Brazilian immigrants (0.28%), according to the General Census of 1950. Table 18 shows a comparison between inbreeding rates observed in the parishes of Capivari and Natividade for the period between 1905 and 1955. As in both parishes all Roman Catholic marriage registrations in that period were examined, it is possible to follow the temporal trends in frequencies of first cousin marriage. Figure IV shows that the frequency of first cousin marriage for the parish of Natividade has remained on a relatively high level, discarding the variations due to sampling. In the parish of Capivari the decrease in frequency of first cousin marriage was quite uniform.

Tab. 18. Comparison of inbreeding rates found in the parishes of Capivari and Natividade during a fifty-one year period (1905-55)

Parish	N.	Frequency of 1 C	$\chi^2_{(1)}$	P	Frequency of all types of cousin marriage (to 3 C)	$\chi^2_{(1)}$	P
Capivari	6337	40 (0.63%)	55.0	< 0.0001	90 (1.42%)	185.3	< 0.0001
Natividade	2810	69 (2.46%)			189 (6.73%)		

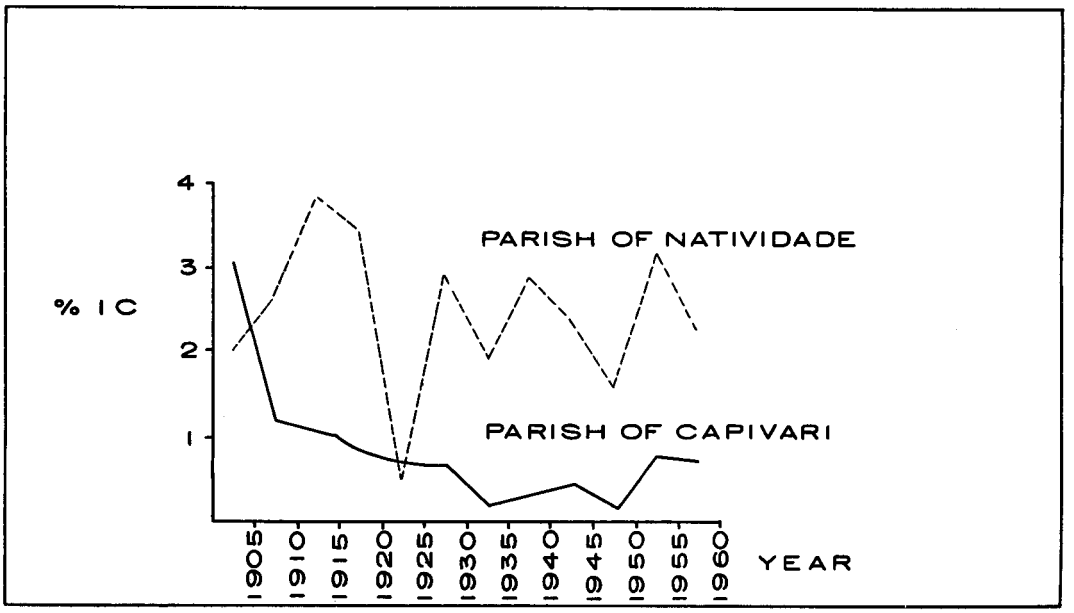

Fig. IV. Frequencies of cousin marriage in the parishes of Capivari and Natividade during a fifty-one year period (for explanation see text)

Critical Consideration of the Available Consanguinity Data

From the figures presented above, the frequencies of cousin marriage can give a very clear idea about the dynamics of the population, provided that supplementary information is available. However, the critical point is the extent that inbreeding rates reflect the isolation of the population, in agrement with the genetic theory. Recently some investigators, particularly Morton (1955) have pointed out that the isolate concept applies poorly to actual situations, because of the non-randomness of consanguineous marriages. Morton's opinion is based on the estimation of isolate

size from frequencies of different types of cousin marriage obtained for several large regions. More reliable information is available in the investigation of a relatively limited breeding population, as for example, an individual parish.

Since Brazil's population is about 95% Roman Catholic, parish marriage registers seem to be a very important source of information on inbreeding rates. However, it is necessary to examine their reliability. In the community studied it was possible to obtain information on the frequency of first cousin marriage from the civil registers for the following periods: 1890-96, 1920-26, and 1950-56 (see page 179). Table 19 shows a comparison between the frequencies of first cousin marriage estimated from the civil and Roman Catholic marriage registers for corresponding periods. As it

Tab. 19. Comparison between frequencies of first cousin marriage estimated from the civil and Roman Catholic registers for corresponding periods

Register	Period	N.	Frequency of first cousin marriages	$\chi^2_{(1)}$	P
Civil	1890-96	351	18 (5.13%)		
				5.2	=0.02
Roman Catholic	1890-95	563	13 (2.34%)		
Civil	1920-26	1205	5 (0.41%)		
				1.1	=0.30
Roman Catholic	1920-25	631	5 (0.79%)		
Civil	1950-56	923	3 (0.32%)		
				1.4	=0.23
Roman Catholic	1950-55	810	6 (0.74%)		

can be seen in Table 19 there are differences between the two sets of figures, being significant at the 5% level of probability for the period 1890-96.

The isolate sizes estimated from the frequencies of first cousin marriages observed in civil registers are appreciably lower for the period 1890-96 and higher for the periods 1920-26, and 1950-56. The decrease of inbreeding rates was marked. However, there are several reasons for doubting the values obtained from civil registers. After the establishment of civil marriage registers (about 1890) some cousin couples possibly avoided the religious ceremony because they were embarrassed in asking for dispensation; others, possibly, did not admit their blood relationship. However, in rural parishes the priests usually know the consanguineous relationships of the parishioners. A more realistic possibility is that Roman Catholics in the past generations may have obtained civil marriage registration some time after their church marriage. In fact, the Roman Catholic and civil resistrations of consanguineous marriages show a time difference of about 10 years in some cases. Thus the Roman Catholic

marriage registers may be more reliable for genetic purposes since they were not delayed.

Haldane and Moshinsky (1939) noted that the frequencies of first cousin marriages are about 55% of all consanguineous marriages (ca. 70% of α value) in western European populations. In the parish of Capivari the relative frequency of first cousin varied according to date. The following values were found: 42.8% (1830-35), 35.6% (1860-65), 29.6% (1890-95), 62.5% (1920-25), and 28.6% (1950-55). This could indicate that immigration affected the frequency of each type of consanguinity in various ways. However, sampling variation could account for the variation found since they showed no systematic trend.

Non-randomness in observed frequency of first cousin marriage can be checked provided it is possible to calculate the isolate size by independent data. It was stated on page 19 that the number of fertile women between 15 and 39 years was about 1800 according to the General Census of 1940. This meains that the isolate size for a 25 year generation has been about 3600 in more recent generations. Considering that the average number of children per family in the community is about 4, it is possible to estimate the expected frequency of first cousin marriage, by substituting the values $b = 4$ and $N = 3600$ in Dahlberg's formula $C = 2b(b-1)/N$. The expected values are in very good agreement with the observed ones in the 1920 and 1950 data, as shown below:

Frequency of first cousin marriage			
Period	Observed in		Expected
	Civil register (%)	Roman Catholic Register (%)	
1920	0.41	0.79	0.67
1950	0.32	0.74	

The expected frequency of first cousin marriage suggests that the figures observed in Roman Catholic registers are nearly random.

Now, assuming randomness in frequency of first cousin marriage, it is tentatively possible to calculate to what extent the observed frequencies of other types of cousin marriage depart from the values expected by the isolate theory. By using Dahlberg's (1929) formulae it is expected that each individual taken at random from the population should have, on the average, $[4b(b-1)/(2+b)]$ uncles or aunts, $[2b(b-1)]$ cousins, $[8b^2(b-1)/(2+b)]$ cousins once removed, $[4b^2(b-1)]$ second cousins, $[16b^3(b-1)/(2+b)]$ second cousins once removed and $[8b^3(b-1)]$ third cousins. Assuming that the average number of fertile children has been equal to 4, for each first cousin there should be 0.33 uncles or aunts, 2.66 first cousins once removed, 8 second cousins, 21.33 second cousins once removed and 64 third cousins. Table 20 shows the comparison between the observed frequencies of different types of cousin marriage and

those randomly expected in relation to the frequency of first cousin marriage taken as unity in different periods. The overall expected and observed figures are significantly different. The lower is the degree of relationship, the higher is the discrepancy.

It can be concluded that the isolate concept, from a formal point of view, can represent a situation prevailing only in stable and small isolated populations. Since cousins of low degree are related by common ascendants of many generations ago,

Tab. 20. Comparison between frequencies of different types of consanguineous marriage (up to third cousin) observed in the parish of Capivari and those expected by assuming randomness in frequency of first cousin marriage taken as unity

Type of consanguinity	Expected relative frequency (b = 4)	Observed absolute frequency in						Expected total (e)	Dif. (d)	d^2/e	P
		1830-35	1860-65	1890-95	1920-25	1950-55	Total				
½ C	0.33	1	—	5	—	—	6	14	8	4.6	=0.04
1 C	1	3	16	13	5	6	43	43	—	—	—
1½ C	2.66	—	4	4	3	4	15	114	99	86.0	<0.0001
2 C	8	—	4	9	—	11	24	344	320	297.7	<0.0001
2½ C	21.33	—	5	5	—	—	10	917	907	897.1	<0.0001
3 C	64	3	16	8	—	—	27	2752	2725	2698.3	<0.0001
Total	—	7	45	44	8	21	125	4184	4059	3937.7	<0.0001

several factors can change their expected frequencies. In small populations within which all marriages occur at random, the equilibrium frequencies of cousin marriages are found only if the average number of children is stable and the parents contributing to the next generation had been born within the isolate. Even so, population stratification of any extent should affect the frequencies of cousin marriage of lower degree more strongly. As a matter of fact the dispersion rate is not equal for each individual of the population so that the dispersion areas for cousins of lower degree should be larger than those for cousins of higher degree. Consequently the probability of the former intermarrying is less than that for the latter. Actually, it is not possible to establish any constant relationship between the frequencies of different types of cousin marriage because they also depend on complex factors not considered by the theory.

Morton (1955) has noted that the isolate sizes estimated from frequencies of different types of cousin marriage usually show marked discrepancies. This kind of discrepancy, previously referred to by Dahlberg (1929) himself, does not invalidate the isolate concept as a general tool to be employed in human population genetics. Comparison of isolate sizes estimated from frequencies of different types of cousin marriage does not make sense. However, the inter-regional comparisons of isolate size estimated from the frequencies of the same type of cousin marriage seem to give a good indication of genetical isolation of the individuals in different places.

V. The effect of immigration on the frequency of some congenital abnormalities

General Consideration

As mentioned in the introduction, immigratory flow can be of eugenic importance. It should be expected that the breakdown of isolates determined by immigration decreases the frequency of rare hereditary abnormalities. However, this effect should be higher for recessive anomalies because of the expected decrease of the inbreeding rates. In the parish of Capivari, the mean coefficient of inbreeding decreased from 0.00783 in 1860-65, to 0.00083 in 1950-55, a value about ten times lower.

This effect may be considered further. If the frequency of a recessive trait (x) in the population were 0.0005 (as in the case of hare lip), the present gene frequency, at equilibrium conditions, would be:

$$q = \sqrt{(\alpha^2+4x)}-\alpha/2 = 0.017$$

and the homozygote frequency, in the absence of consanguineous marriages, $q^2 = 0.000289$.

Assuming, further, that the gene frequency was maintained constant during several generations, (4) the effect of inbreeding on the homozygote frequency should be appreciable in the past generations (Table 21).

As shown in Table 21, the consequences of the decrease of mean coefficient of inbreeding are substantial. In the period 1860-65, the relative increase in the homozygote frequency, assuming panmixia, should be ca. 45%, as compared with the same population in which α was zero. Assuming 1000 different recessive dele-

Tab. 21. Expected effects of the inbreeding rate observed for each of five different periods of time in the parish of Capivari on the frequency of recessive homozygotes

Period	Observed mean coefficient of inbreeding (α)	Increase of frequency of rec. homozygotes (αpq)*	Total frequency of recessive homozygotes ($q^2+\alpha pq$)	Relative increase of rec. homozygotes ($\alpha pq/q^2$)%
1930-35	0.00463	0.000077	0.000366	26.6
1860-65	0.00786	0.000131	0.000420	45.3
1890-95	0.00315	0.000053	0.000342	18.3
1920-25	0.00064	0.000011	0.000300	3.8
1950-55	0.00083	0.000014	0.000303	4.8

* $q = 0.017$ and $q^2 = 0.000289$.

(4) Of course, this should not be the case for the population investigated.

terious genes with the same frequency (0.017) and the isolate size 500, the total increase in homozygote frequency (0.131) for the period 1860-65 should correspond to an excess of 65 affected individuals due to the inbreeding effect. (5)

The decrease in the observed a values was determined mainly by the lowering in the frequency of first cousin marriages. The effect expected from the decrease of the frequency of first cousin marriages can be appreciated in a different way. Assuming constant gene frequency, the proportion of individuals affected with recessive abnormal traits born from first cousin marriages in 1860-65 should be markedly higher than that expected in the period 1950-55. Table 22 shows the proportion of parental consanguinity expected in the periods, 1860-65 and 1950-55 in the cases

Tab. 22. Proportion of first cousin marriage expected among individuals affected with recessive abnormalities in the parish of Capivari during the 1860-65 and 1950-55 periods, assuming variable gene frequency

Gene frequency	Expected parental consanguinity (%)*	
	in 1860-65**	in 1950-55**
1/32 (q^2=0.001)	28.5	2.2
1/100 (q^2=0.0001)	70.1	5.3

* k = c(1+15q) / 16q.

** Frequency of first cousin marriages: in 1860-65 = 0.0976 and in 1950-55 = 0.0074.

of gene frequencies of 1/32 and 1/100. If the lower gene frequency prevailed, the production of individuals affected with recessive abnormalities through first cousin marriage should be extremely high for the period 1860-65, just before the immigratory flow.

The Incidence of Seven Congenital Abnormalities in the Present Population

A Public Health Post for Children was founded in the community, which started in 1951 to attend out-patients from the general population. From 1951 to 1958 (July) 7746 different children between 0 and 12 years were examined by a pediatrician. The results of the examination of each child were recorded in files revealing both the father and the mother's name. Since married women keep their maiden name, the children were classified according to both parents' origin (see page 181)

(5) Discarding the possibility of individuals simultaneously homozygote for different deleterious genes.

as follows: both parents of Brazilian descent (B); both parents of Italian descent (I); one parent of Brazilian descent, and one of Italian descent (H); parents descended from other nationalities, " pure " or mixed (R); and Negroes and Mulattoes (N). The frequencies of these groups in the Public Health Post are not markedly different from those found in the records of the Office for Army Obligatory Recruitment (cf. Table 9), as shown below:

Records from	Groups (%)				
	B	I	H	R	N
Public Health Service for Children	27	31	21	7	13
Office for Army Obligatory Recruitment	23	41	16	9	11
Difference	+4	−10	+5	−2	+2

From the above comparison it seems that there is an under-representation of the I and R groups and a slight over-representation of the B, H, and N groups. Since the I and R groups are predominantly more rural, whereas the Public Health Post is located in the town, it is likely that they look for medical care in only the more urgent situations.

In this investigation seven relatively frequent types of congenital abnormalities were selected, namely harelip and cleft palate, malformations of upper and lower extremities, congenital heart disease, mongolism, epilepsy, inguinal hernia, and gross mental retardation. These abnormalities were chosen for the following reasons: *a*) their clinical diagnosis is relatively simple by medical inspection; *b*) they usually have a hereditary background; *c*) their frequency is usually high enough to be significant in relatively small samples. Since the same criteria were employed to detect these abnormalities among the groups of different parental origin, it seems that no systematic bias could influence the inter-group comparison for the community, but this is not the case for comparisons with other populations. The frequency of most human congenital abnormalities decreases with the age of the population investigated (c. United Nations, 1958, p. 196) inasmuch as selection should be operating by decreasing the viability of the affected. Thus, the comparison between the present 0-12 year material and soon after birth samples should be biased, regardless of the differences in ascertainment. It is also likely that the present sample is at some extent over-represented because the congenitally defective children might be expected in public health services more frequently than the unaffected, particularly among older children. This bias is in the opposite direction from that caused by the decreased viability of the affected children.

Table 23 shows the frequencies of the seven congenital abnormalities among 6742 white individuals, classified according to parental origin. A group of 1024 Negroes was excluded because they belong to a distinct population.

The total frequencies of the seven congenital abnormalities among the different groups are significantly heterogeneous ($\chi^2_{(3)} = 7.5$, $P = 0.025$). Significant heterogeneity at the 5% level of probability is also found for the total frequencies observed among B, I, and H groups, because only one case of congenital abnormality (mongolism) was found in the " hybrid " group. A comparison between the overall frequencies found among the " pure " groups (B+I) and that observed in the " hybrid " group (H) showed that the difference is significant ($\chi^2_{(1)} = 3.9$, $P<0.05$; Yate's correction being applied). On the basis of polygenic additive systems are responsible for the effects investigated, it should be expected that the " hybrid " group might show a frequency of congenital abnormalities intermediate between those found among the " pure " groups, but instead it displays a comparatively low figure.

Before interpreting these findings, a brief note should be made on the hereditary basis of these seven abnormalities. According to Fogh-Andersen (1943) harelip with or without cleft palate and isolated cleft palate represent two distinct genetic entities. The relatively high concordance of harelip in MZ twins in comparison with DZ twins suggests a strong hereditary component and the high incidence of affected sibs of propositi born to normal parents, suggest that recessive genes are involved. Isolated cleft palate seems, however, to be a dominant trait with very low penetrance or determined by non-genetic factors. Because harelip with or without cleft palate and isolated cleft palate are readily diagnosed through medical examination, their frequencies are relatively reliable for comparative studies. Table 24 shows the frequencies of harelip with or without cleft palate and isolated cleft palate at birth among different white populations and in the parish of Capivari. Despite the fact that most individuals examined in the parish of Capivari were under one year old, the sample includes individuals between some days and 12 years old. Therefore differential elimination should account for part of the difference found between the frequency of the deformity in the community investigated and the frequencies, at birth, in other white populations. However, the total frequency found in the parish of Capivari is even lower than that observed in the general (adult) population of Denmark (see Table 24).

Malformations of the extremities include different types of defect, many of them being distinct genetic entities. Some malformations such as polydactily and syndactily are usually dominant while others such as ectrodactily, club foot and lobster claw have a more complex genetic basis. Among the abnormalities of the extremities observed in the population investigated are three cases of club foot (talipes equinovarus) (two in the B group and one in the I group), one case of congenital malformation of the right arm and left hand (in the B group) and one case of polydactily observed in a Negro individual, not included in the analysis. Also observed, and excluded from the present analysis was a Negro with bilateral club feet (talipes equinovarus) and ankylosis of the interphalangeal joints in the three posterior fingers of both hands. The total incidence of malformations of the extremities (0.0074) in the community investigated is appreciably lower than found in other white popula-

Tab. 23. Incidence (per thousand) of seven congenital abnormalities in the present population of the parish of Capivari, according to the parental origin of the individuals (for significance of differences see text)

Group (according to parental origin)	Total No. of individuals	Hare lip and cleft palate	Malformations of extremities	Congenital heart disease	Mongolism	Epilepsy	Inguinal hernia	Mental retardation	Total
Descendants of Brazilians	2405	1.3	1.7	0.8	0.4	0.4	0.4	0	5.0 (12)*
Descendants of Italians	2103	0.5	0.5	0.5	0	0	1.0	1.0	3.3 (7)
Descendants of Brazilians and Italians	1658	0	0	0	0.6	0	0	0	0.6 (1)
Descendants of other nationalities (pure or mixed)	576	0	0	1.7	0	3.5	1.7	0	6.9 (4)
Total	6742	0.6	0.7	0.6	0.3	0.4	0.6	0.3	3.6 (24)

* Actual number of individuals observed.

https://doi.org/10.1017/S1120962300020321 Published online by Cambridge University Press

tions. It is not likely that decreased viability of the affected with such types of malformations can account for the difference.

Congenital heart disease is also a general name including several different clinical and genetic entities. Congenital heart disease is usually recognized at birth, but by no means is this true for every case (cf. Neel, 1958; McKeown, and Record, 1960). Since the incidence of congenital heart disease among sibs of propositi is six times higher than the frequency in the general population (McKeown, MacMahon,

Tab. 24. Frequency (per thousand) of hare lip with or without cleft palate and isolated cleft palate obtained by different investigators among some Caucasian populations and in the parish of Capivari

Country	Sample size	Hare lip ± clef palate	Isolated cleft palate	Hare lip + cleft palate	Reference
U.S.A. (Baltimore)	15,565	0.90	0.19	1.09	Davis, 1924
France	100,889	0.91	0.14	1.05	Peron, 1929*
Germany	102,873	0.78	0.21	0.99	Günther, 1931*
Holland	15,270	—	—	1.05	Sanders, 1934
Denmark	128,306	1.16 (0.70)**	0.34 (0.30)**	1.50	Fogh-Andersen, 1943
Switzerland	50,147	—	—	1.48	Ehrat, 1948
Sweden	44,109	1.36	0.39	1.75	Böök, 1951
England	218,693	0.78	0.52	1.30	MacMahon and McKeown, 1953
Brazil (São Paulo)	13,024	1.30	0.31	1.61	Saldanha et al. 1960
Average	688,876	—	—	1.29	—
Parish of Capivari:					
Descendants of Brazilians	2,405	0.83	0.41	1.25	Present paper
Descendants of Italians	2,103	0.48	0	0.48	» »
All groups	6,742	0.29	0.29	0.59	» »

* According to Fogh-Andersen (1943).
** Incidence in the general population cf. Kemp (1950).

and Parsons, 1953), it can be assumed that its causation involves genetic factors. While the mechanism of hereditary transmission is not known, congenital heart disease is often associated with other hereditary abnormalities and syndromes. The total frequency of congenital heart disease in the community investigated was 0.0059, a lower frequency than in other white populations (MacMahon, McKeown and Record, 1953; Böök, 1951).

Mongolism is an easily diagnosable syndrome and has a relatively high frequency. The etiology was not clear until two years ago when Lejeune et al., (1959) showed that a mongolian caryotype exhibited an extra autosomic chromosome. The mongolian trissomy has been confirmed extensively and it probably depends on the genetic

constitution of the mongol's mother since the frequency of mongolism births increases with the maternal age (Carter and McCarthy, 1951; Öster, 1953; Penrose, 1955a). It is curious that the unique case of congenital abnormality found among the " hybrid " group of the population investigated is a mongolian. The total incidence of mongolism in the parish of Capivari was very low (0.0030) compared to other white populations (cf., Penrose, 1954).

Idiopathic epilepsy shows about 25% familial incidence. Although the trait associated with various kinds of mental retardation, it seems to be an independent genetic entity (Penrose, 1954). The difference between concordance figures among MZ twins (66%) and DZ (3%) suggest that the anomaly is undoubtedly inherited (Conrad, 1940). Reliable frequency estimates of idiopathic epilepsy in the population are hard to obtain but figures as high as 0.005 are to be expected. Therefore the frequency of epilepsy in the population invertigated (0.0044) does not seem to be different from those to be found in other white populations.

Inguinal hernia may be influenced by environmental factors such as post-natal care, birth traumas, etc., but the defect depends on a hereditary predisposition (Weimer, 1949). There is a relatively higher incidence of inguinal hernia in males, and in some families the anomaly is inherited from father to son. The incidence of inguinal hernia in the population studied (0.0059) is appreciably higher than that observed in other white populations (cf., Böök, 1951).

Severe mental retardation is frequently associated with gross physical defect and usually seems to have a hereditary basis (Penrose, 1954). Typical idiots are usually born to normally intelligent parents, but this is not generally true of sub-normals, i. e., feeble-minded. Twin studies suggest that mental retardation is strongly determined by heredity. The frequency of idiocy is about 0.025 (all types of oligophrenia) in the white populations, being higher among males (Penrose, 1954). The frequency of gross mental retardation observed in the population investigated (0.0030) is appreciably low.

Conclusion

From the results presented in the Table 23 it seems that the overall frequency of the seven abnormalities in the populations studied is appreciably low but the comparison with other white populations can hardly be taken too seriously because of the differences in the ascertainment of the data. However, the eugenic effect of immigration is suggested by inter-group comparisons of total frequency of the seven congenital abnormalities. The " mixed " (H) group showed practically complete absence of congenital anomalies (excepting one mongolian idiot). The immediate consequence of the breakdown of an isolate by immigration is the increase in heterozygote frequency of rare and common genes (see introduction). The frequency of rare recessive traits is more markedly decreased than that of dominants because of the decrease in the inbreeding rate. However, a very marked effect is likely to be expected only for heterotic loci. Since the seven abnormalities here studies include cases of complex genetic causation, it seems to be permissible to explain the

" eugenic " situation of the " mixed " group of the population by means of a increased frequency of heterotic combinations. Neel (1958) has commented that 80% of human major congenital abnormalities have no clear hereditary causation and, on the basis of Lerner's (1954) theory of genetic homeostasis, suggested that such congenital defects should represent " phenodeviants " of polygenic homeostatic systems. According to this idea, mixed population displaying a relatively greater heterozygote frequency, should present a lower incidence of congenital defects. The present data seem to be in agreement with that hypothesis.

VI. The effect of immigration on the distribution of stature and other normal genetic traits

MATERIAL

The investigation of common " non-pathological " traits is of interest in population genetics not only to obtain estimates of population composition but also for the solution of specific genetic problems. In this sense the so-called " normal " or " non-pathological " genetic traits have a misleading connotation because, in the case of both monogenic and polygenic hereditary characteristics, they can be under selective forces the intensity of which is hardly detectable by ordinary statistical methods.

Seven genetic traits were investigated in the present work: stature, eye colour, taste sensitivity to phenylthiourea, middle phalangial hair, ear lobe attachment and colour-blindness. The investigation of stature and eye colour was based on the files recorded in the Office for Army Obligatory Recruitment in the parish of Capivari. The register includes other anthropological traits but stature and eye colour were selected because they seem to be more reliable. Stature was measured by means of a wooden anthropometer and eye color by simple inspection. Eye colour was classified in four categories: black, brown, green, and blue. In the present analysis, black and brown were merged into a dark class and, similarly, green and blue into a light class. Since eye colour registers for a few files are not available, the number of individuals investigated for stature is slightly larger than that for eye colour. The material includes records of 2882 males, mostly 17 years old and the remainder 18 years old. The files were recorded during 1945-1956. Therefore they refer to individuals born between 1927 and 1940.

The study of the remaining traits is based on examinations of 291 individuals, 131 males and 190 females, between 10 and 24 years (mean age: 16) in the public local High School.

DISTRIBUTION OF STATURE AND EYE COLOUR

Stature

The distribution of stature among human populations raises some interesting and complex problems. The dynamics of stature among " mixed " populations and the increase in average height during recent generations among western Europeans are of outstanding concern. However, the analysis of these problems depends on the more precise understanding of the genetic background of stature and more complete information about the structure of the populations in which the trait is investigated. Before analysing the present data, some considerations of the inheritance of human height must be given.

The population distribution of human stature is approximately normal, provided that the sexes are taken apart. This situation prompted early investigators (Blakeslee, 1914; Davenport and Love, 1921) to suggest the hypothesis of polygenic inheritance. However, critical information of the additive nature and the number of genic loci envolved as well as the role of environmental factors on adult stature are so far very scarce. The simplest model accounting for human stature inheritance assumes several two-allele independent loci, the effects of which are absolutely additive. Since the distribution range of individual heights is finite, by assuming equal gene frequency for each locus, the genotypic distribution is obtained according to the binomial rule: $(p+q)^{2n} = (p^2+2pq+q^2)^n$, where n stands for the number of loci and p and q, the frequencies of effective and non-effective allelic genes. By assuming further no environmental effect, the difference between higher and lower limits of individual height, that is, the range i can be defined as $i = 2en$, so that the minimum phenotypic effect per locus is: $e = i/2n$. Under the above assumptions, alternative hypotheses cannot be rejected because only i can be measured directly.

A more direct approach is that based on the correlation between consanguineous individuals. Fisher (1918) has shown that the inheritance of metric traits can be explained by Mendelian theory and put forward the concept of polygenic inheritance. Total variance (σ_t^2) of continuously distributed characters can be partioned into genetic (σ_g^2) and non-genetic (σ_n^2), so that $\sigma_t^2 = \sigma_g^2+\sigma_n^2$. Furthermore, genetic variance can be assumed to be due to the action of additive (σ_a^2) and non-additive or dominant (σ_d^2) genes, or, further caused by the interaction of both kinds of genes, but it is independent of gene frequencies. So, the phenotypic correlation to be found between relative of coefficient of relationship f, can be obtained by:

$$r = f.\ (\sigma_g^2-\sigma_n^2)/\sigma_g^2, \qquad \text{VI. 1.}$$

assuming that all loci contributing to the genetic variance are additive in nature. Furthermore, discarding the effects due to environmental factors (σ_n^2), the genotypic correlation is taken to be equal to the coefficient of relationship. It should be 1 for monozygotic twins, 1/2 for full sibs or parents and children, 1/8 for first cousins, etc. However, the greatest limitation of this methods rests on the difficulty of evaluating

the variance due to non-genetic factors, and the relative contributions of additive and dominant loci, using human material. Both dissimilar environmental conditions to be found among relatives and dominant loci should decrease the estimate value of phenotypic correlation. Positive assortative mating, as has been observed for for several human metric traits (cf. Spuhler, 1959), should cause a reverse effect (Hogben, 1939, pp. 106-114). Similarly an increased correlation estimate is expected between like-sexed relatives on the assumption that sex-linked loci are envolved in determining human stature (Brues, 1950). Probably because the biases considered above, figures estimated from different types of relationship, are usually higher than theoretical ones on the grounds of additive loci. Most results based on sib correlation are higher than the expected value of 0.5 (Bowles, 1932; Hogben, 1939, p. 103; Howells, 1948). While the results indicate the importance of hereditary factors, no precise answer could be drawn from those investigations. Fisher (1918) suggested that no more than 5% of the phenotypic variance in stature can be attributed to non-genetic factors.

Twin data have supported additional information on the inheritance of stature in man. Comparison between identical and fraternal twins could be even more suitable to evaluate the role of non-genetic factors. Heritability (h) of a metric can be measured from the variances of intra-pair differences among monozygotic (MZ) and dizygotic (DZ) twins, by the formula;

$$h = (\sigma^2_m - \sigma^2_d)/\sigma^2_d \quad (6), \qquad \text{VI. 2}$$

assuming again that intra-pair differences are determined by heredity. Since MZ twins have, on the average, more similar environmental conditions than DZ controls, an estimate of heritability is not a precise measure of hereditary factors. However, twin investigations are relevant as a general approach. The pioneer work of Dahlberg (1926) showed that intra-pair differences in stature (in cm.) were: 1.63 ± 0.13 for 96 MZ twins, 5.07 ± 0.40 for 91 like-sexed DZ twins, 5.76 ± 0.64 for 46 unlike-sexed DZ twins, and 4.68 ± 0.33 for 117 sibs. The results obtained by Newman, et al. (1937) are similar. The differences found among the American material were: 1.7 for 50 MZ twins reared together, 1.8 for 19 MZ twins reared apart, 4.4 for 50 DZ twins and 4.5 for 52 sibs. Undoubtedly twin studies suggest that hereditary factors should account for the major part of phenotypic variation in stature. The estimation of heritability from the above data (see also Clark, 1956) are about 80-90% but several biases inherent in the twin method must be kept in mind (Price, 1950).

Other complications in the genetic analysis of human stature are brought out by sex differences. Mean height is invariably greater in males, thus the overall distribution is bimodal. Factors causing sex differences should be very complex, however, and mainly resulting from hormone differences between the sexes rather than " direct " gene action. The possibility of sex-linked genes determining human stature

(6) Significance is obtained from $F = \sigma^2_d/\sigma^2_m$.

was suggested by Finney's (1939) analysis but it should not be a simple relationship (Brues, 1950; Tanner and Healey, 1956).

Pertinent to the genetic analysis of human stature is the general trend of mean height among different generations. Boas' (1912) classical investigation of the descendants of immigrants from Europe (Bohemian, Hungarian, Polish, Jewish, Sicilian, Napolitan) in America showed that those groups were, on the average, higher than the values observed among the populations from which their parents came. Similar results were found among descendants of Japanese immigrants in continental U.S.A. and Hawaii (Spier, 1929; Ito, 1936; Shapiro and Hulse, 1939). Differences were also found among descendants of Chinese (Lasker, 1946) and Mexicans (Goldstein, 1943) in the United States. Such changes in body size have been explained by most anthropologists on the grounds of environmental improvement. However, practically no emphasis has been placed on the genetic recombination due to marriages between migrants born in different communities. Of course, most of such marriages are exogamous while those in the original countries were more often " endogamous " (7) and the relative role of environmental improvement and genetic recombination can hardly be evaluated from these investigations. Furthermore, stature increase has been known in several European countries over the past generations. The phenomenon has been documented quite well for Scandinavian countries. In Denmark the average increase in mean height was about 9-10 cm. from 1815 to 1949 (Kemp, 1951, p. 144). Similar results were obtained for Sweden (Hölmgren, 1952). Differences were recorded in Norway (Mohr, 1934, p. 225) even for a ten-year period. In Italy, vital statistics have shown that, in the last 150 years, the mean stature increased by 1/10 cm. per year (cf. Conterio and Cavalli-Sforza, 1959) and in the United States the university student generation seems to be taller at present (Hooton, 1946, p. 248).

Interesting conclusions can be drawn from the investigation on height increase in different ages. Broman, et al. (1942) showed that in Sweden during a 60-year period the average stature increased for all ages, the increase being greatest for 14-year old males and 12-year old females. While the average increase was 15 cm. for the 10-18 year group, it was only 5 cm. for adults.

Lundman (1940), based on measurements of over a million men, showed that stature increased 7-8 cm. during the last century, but no correlation could be found between stature changes and differences in economic conditions of the district. These results indicate that environmental factors influence growth speed but that adult stature may be to a large extent determined by inheritance. This conclusion is supported by differences in stature between adolescent twin pairs (Dahlberg, 1926). Among MZ twins, the differences were: 1.33 ± 0.28 for ages below 10.5 years, 2.26 ± 0.29 for ages between 10.5 and 15.5 years, and 1.15 ± 0.16 for ages above 15.5 years. The differences for DZ twins were; respectively, 4.18 ± 0.60, 5.80 ± 0.71 and $4.94 \pm$

(7) Endogamous and exogamous marriages are referred to in the sense the partners were or were not born in the same community.

0.75. Therefore changes in growth speed should be expected without a corresponding increase of average adult stature. Morant (1950) has shown that, although British mean stature has not changed during the past 60 years, average stature was reached in 1880 at 28 years but in 1945 it was reached at 21.5 years. For example, the increase for 17 year olds was 2.03 cm.

Differences in average stature between regions and social classes (Broman, et al., 1942) bring similar difficulties for explaining the relative roles of environmental and hereditary factors.

Since an appreciable part of stature variance seems to be due to heredity, efforts to understand the trends of this metric trait among human populations seem to be relevant. Dahlberg (1938, 1947) has pointed out that while increased stature has been quite uniform and constant, the same is not true for the improvement of living conditions in Sweden, therefore the breakdown of isolates which has progressively taken place in that country can furnish the basis for a genetic explanation of the phenomenon. " If stature is determined by a number of genes which are mainly dominant ", the increase of areal and social mobility of individuals, starting with the industrialization age, should also have determined an increased heterozygosity for such loci. Consequently, it should be expected that, until world population reaches the " ideal " situation of complete random mating, average stature would continue to increase.

Hulse (1957) has presented some relevant data concerning that effect. Mean stature of individuals produced by couples born in the same and in different small Swills villages was compared. The material included persons falling in three categories: subjects still living in Switzerland, Swiss immigrants in California and subjects born to these immigrants. The mean height of these three groups were different but, for any category, the mean height of the individuals born to exogamous marriages was about 2 cm. higher than the figure obtained for those born to endogamous ones.

Gini (1949-1950) showed that height distribution in representative European populations is leptokurtic. Lenz (1952) suggested that leptokurtosis should be caused by assortative mating and/or isolate effects. The trend towards a mesocurtic shape of normal distribution has been noticed particularly among " mixed " populations, resulting from the breakdown of previously isolated communities.

Fisher and Gray (1937), using the regression between father-son heights, tested the possibility of dominant genes determing human stature but their results were doubtful. A similar conclusion was obtained by Tanner and Healey (1956) using the same approach. On the other hand Penrose (1954, 1955 b) has envisaged a model to explain the population distribution of such human metric traits as intelligence and stature. According to that investigator, human metric characteristics should display heterotic effects, expressed by differential fertility. Correlation between stature and number of children is —.20 (Maxwell, 1953). A higher correlation was obtained for intelligence (—.28, according to Thompson, 1949). If the increase in fertility is associated with a corresponding heterozygosity increase for the genes determining stature the character can be maintained at genetical equilibrium, provided

that selection against extreme phenotypes is counterbalanced by segregation among heterozygotic and more fertile phenotypes. So, the height variance of the fertile individuals should be smaller than that found for the general population. Such a model would be more efficient on the assumption of assortative mating, a common situation for human metric traits.

The assumption that extreme phenotypes are more frequently killed by disease would, at least partly, account for differential fertility in human height and furnish an alternative explanation for the secular trend in average stature. However, by comparing the mean heights between the sibs of propositi killed by different diseases at pre-reproductive ages and controls from the general population, Conterio and Cavalli-Sforza (1959) have suggested that the effect observed for mortality by some specific infectious disease (pertussis + diphtheria) and by prematurity can account for only ca. 4% of the trend of average stature in Italy.

Since the sample investigated resulted from the intermixture of two non-related populations, data on the distribution of stature seems to be of interest. Individuals were classified according to parental origin in four groups: descendants of Brazilians, descendants of Italians, descendants of Brazilians and Italians, and descendants "pure" or "mixed" of other nationalities (remaining group). Table 25 shows

Tab. 25. Means and variances for the distribution of stature among males in the parish of Capivari, according to parental origin

Group *	Number	Range	Mean and standard error	Variance	F
Brazilian	760	141-186	162.94±0.28	61.81	
					1.26
Brazilian and Italian	501	140-190	166.08±0.31	49.02	
					1.25
Italian	1318	137-185	166.47±0.21	61.14	
Sub total	2579	137-190	165.35±0.15	61.42	
Remainder	303	140-189	165.04±0.47	65.38	
Total	2882	137-190	165.32±0.15	61.84	

* According to parental origin.

the means and variances of stature distribution in the parish of Capivari according to parental origin.

The analysis of variance for the mean among groups is highly significant, as it is shown in Table 26. If the analysis is restricted to the three relevant groups (B-I-H) the significance is even greater (F = 53.16).

The average stature of the " hybrid " group is quite similar to that of the tallest (group of Italian derivation) ($t = 1.00$, $P = 0.28$). However, its variance (49.02) is significantly less than that of " pure " groups (61.81 and 61. 14, for descendants of Brazilians and descendants of Italians, respectively). Figure V shows the distribution of observed height frequencies among the three groups of relevant ancestry.

Tab. 26. Analysis of variance of stature of the four groups classified according to parental origin (see text)

Sources of variation	Sum of squares	d. f.	mean square	F
Between groups	6,302	3	2,100.67	33·95
Within groups	171,926	2,779	61.87	
Total	178,228	2,882	61.84	

As can be seen in Figure V the distribution pattern of the Brazilian group is a somewhat platikurtic while those of Italian and " hybrid " groups are slightly leptokurtic and negatively skewed.

Since the individuals belonging to the three relevant groups have lived under the same ecological (8) and social conditions, a genetic explanation of the results must be attempted. If human stature is determined only by additive genes, the mean height of the " hybrid " group would be expected to be intermediate between those of the " pure " groups. However, to explain the results in the present work, it is difficult to separate the possible effects caused by: (a) additive genes, $a(F_{1di} \langle P_t)$; (b) *dominant genes*, $b(F_{1dd} = P_t)$ and (c) *overdominant genes*, $c(F_{1ds} \rangle P_t)$, where F_1 and P_t stand for filial and the tallest paternal groups, respectively. The total effect can be represented by:

$$E = a(F_{1di} \langle P_t) + b(F_{1dd} = P_t) + c(F_{1ds} \rangle P_t)$$

The results obtained in the present investigation should be explained by: (1) b is finite and $a = 0$ and $c = 0$; (2) a and c are finite and $b = 0$; and (3) a and b are finite and $c = 0$. (9) There are some reasons to consider (3) the most probable explanation. First, other direct evidences are in support of the simpler explanation (3). Second, the mean height of the " hybrid " group does not differ from that of taller " paternal " one but its variance is significantly smaller as compared with other groups. This situation is expected from the operation of dominant genes. It is hard to believe that such large differences in sample variances are due only to genetic

(8) The " hybrid " group and that of Italian derivation are somewhat more rural than the Brazilian group.
(9) More complex explanations are not excluded, as, for example, a, b and c being finite.

factors. Environmental factors could also account for the results but it is necessary to assume that the living conditions of the " hybrid " group were more homogeneous than those of the " pure " ones. However, such environmental differences among the groups investigated could hardly be detected.

It is of interest to compare the figures found for the parish of Capivari with those obtained for northeastern Italy. Gini (1935) has published data on distribution of stature among 25-year-old individuals examined during 1884-1886 in Italy. The weighted mean for the regions: Lombardia, Veneto and Emilia (n = 498) is 165.7. This figure is quite similar to that found for the group of Italian extraction in the parish of Capivari. However, the individuals examined in the latter community are appreciably younger. Cavalli-Sforza (1960) has provided data from 7687 20-year males examined for military service in the region of Parma. Since the sample includes only farm occupied individuals, it seems to be better material for comparison. The mean stature for the latter group (164.47 ± 0.07) (10) is significantly lower than the figure (166.46 ± 0.21) obtained for the 2-year-younger group of Italian derivation in the parish of Capivari.

As a general conclusion, the results presented above can be explained at least in part by the action of dominant genes determining human stature. The data are also consistent with the expected effects caused by the breakdown of isolates and provide some additional evidence to explain the trend of average stature among human populations. However, precise answers to these problems depend upon the use of more refined methods and more informative data.

Eye Colour

Eye colour is a trait commonly studied in anthropological investigations. Boyd (1950, p. 312) shows that the distribution of eye colour among human populations varies within broad limits. Eye colour does not seem to follow a simple scheme of inheritance. It is apparent by simple inspection of pedigrees that darker eyes are dominant over lighter shades. (Davenport, 1927). However, Brues (1946) showed that eye colour is probably determined by the interaction of several autosomal and sex-linked gene pairs.

Table 27 shows the distribution of dark and light eyes among males in the parish of Capivari, according to their parental origin. Frequencies of light eyes are significantly heterogeneous among the four groups investigated ($\chi^2_{(3)} = 85.4$, $P < 0.0001$). According to Coon (1939, p. 558) the frequency of light eyes in northeastern Italian populations is about 27%. The figures presented in the Table 9 indicate that the classification of the individuals by means of parental names should be a reliable procedure.

(10) The value presented in Cavalli-Sforza's paper was 164.47 ± 0.70 but it should be a misprinting.

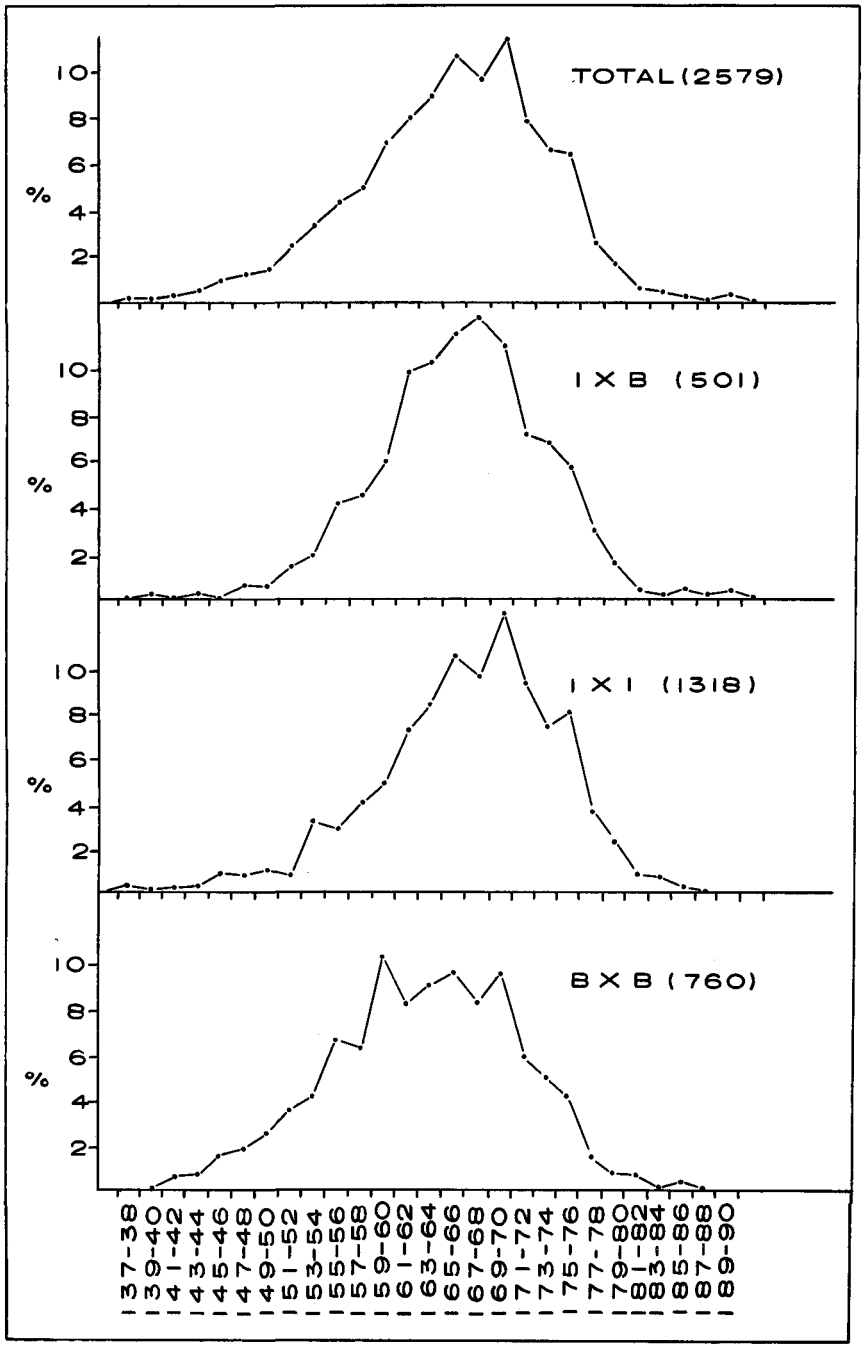

Fig. V. Distribution of observed frequencies of stature in three groups in the parish of Capivari, classified according to their ancestry

https://doi.org/10.1017/S1120962300020321 Published online by Cambridge University Press

Correlation Between Stature and Eye Colour

When intermixture occurs among populations differing in the frequency of genetic traits, genetic equilibrium (11) is reached after a variable number of panmitic generations. In the case of traits determined by allelic genes, it follows just over one generation under panmixia, but this is not true for the traits produced by non-allelic genes. In the latter situation, the number of generations removed until equilibrium is reached

Tab. 27. Distribution of eye colour among males in the parish of Capivari, classified by parental origin

Group*	Number	Dark eyes** (no.)	Light eyes** (no.)	(%)	Heterogeneity
Brazilian	744	692	52	6.99±0.93	$\chi^2_{(3)} = 85.4$
Brazilian and Italian	497	437	60	12.07±1.46	
Italian	1297	1016	281	21.67±1.14	$P < 0.0001$
Remainder	303	241	62	20.46±2.32	
Total	2841	2386	455	16.02±0.69	—

* According to parental origin. ** Dark = black + brown — Light = green + blue.

depends upon the differences in gene frequencies between parent populations and if the traits are determined by linked or independent gene pairs. For linked traits equilibrium also depends upon the frequency of crossing over between the corresponding loci (for discussion see Rife, 1954). Consequently, correlation between traits determined by non-allelic genes found in mixed populations means that genetic equilibrium has not been reached. Table 28 shows the comparison of means and

Tab. 28. Means and variances of stature among males in the parish of Capivari, classified by eye colour

Group*	Number	Range	Mean and standard error	Variance	F	t (**)	P
Light eyes	427	140-184	165.61±0.40	68.26	1.12	1.58	= 0.11
Dark eyes	2253	137-189	164.92±0.17	61.13			

* Light = green + blue — Dark = black + brown. ** for 580 d. f.

(11) In this context, genetic equilibrium means that no correlation between the hereditary traits is found.

variances of stature between light-eyed and dark-eyed individuals. The mean stature observed in the former group is slightly but not significantly higher than that found in latter one ($t = 1.58$, $P = 0.11$). Similarly, the variances among the two groups are not significantly heterogeneous (Table 28). This situation seems to indicate the present population is fairly panmictic and not far from genetic equilibrium.

Tab. 29. Distribution of taste thresholds for phenylthiourea and frequency of non-tasters in the parish of Capivari, according to sex and parental origin

Group	Number	Taste threshold for phenylthiourea								Frequency of non-tasters %	Heterogeneity
		0	1	2	3	4	5	6	7		
Desc. of Brazilians	57	5	2	2	2	2	3	4	37	28.07	$\chi^2_{(3)} = 7.97$
Desc. of Italians	91	2	3	3	4	2	2	6	69	17.58	$P < 0.05$
Desc. of Brazilians and Italians	70	5	5	1	4	2	1	8	44	25.71	
Remainder	73	12	6	1	—	3	5	6	40	36.99	
Total males	131	10	5	1	5	2	4	11	93	20.61±3.53	$\chi^2_{(1)} = 4.19$
females	160	14	11	6	5	7	7	13	97	31.25±3.66	$P = 0.04$
both sexes	291	24	16	7	10	9	11	24	190	26.46±2.59	

Distribution of Other "Normal" Common Traits

Taste Sensitivity to Phenylthiourea

Variation into the ability to taste phenylthiourea is believed to depend upon a main pair of allelic genes (Snyder, 1932, Merton, 1958, and Das, 1958). If series of gradually concentrated solutions are used in taste discrimination, the populational distribution of taste thresholds is bimodal, and the antimode should separate tasters from non-tasters. Racial variation in the frequencies on non-tasters has extensively been shown (cf. Saldanha, 1958).

The taste thresholds of 131 males and 160 females were measured. Seven solutions of phenylthiourea were employed. The most concentrated contained 1300 mg/l was numbered 1. The solution 2 was as concentrated as one half of solution 1, and so forth. To separate tasters from non-tasters, the antimodal value was considered falling between 5 and 6 thresholds. Table 29 shows the distribution of taste thresholds and the frequency of non-tasters, according to sex and parental origin.

The frequencies of non-tasters among four groups were significantly heterogeneous ($\chi^2_{(3)} = 7.97$, $P<0.05$). Sex differences were also statistically significant

($\chi^2_{(1)} = 4.19$, $P = 0.04$). Similar differences were reported among white Brazilians of Rio de Janeiro (Saldanha and Guinsburg, 1954) and Portuguese (Cunha and Abreu, 1956).

The overall frequency of non-tasters (26.46 ± 2.59) is intermediate between those found among Brazilian whites of Portuguese derivation (30.49 ± 3.59, cf. Saldanha and Guinsburg, 1954) and northeastern Italians (23.60 ± 0.80, cf. Silvestroni and Bianco, 1950), but the differences, are not significant. It is interesting to point out that, in an investigation of phenylthiourea sensitivity (Kalmus, 1957) carried out in the same region (Piracicaba) also under strong Italian immigration, the frequency of non-tasters was comparatively low (24.32 ± 4.99). Since Indian and Negro admixture in preponderantly white southern Brazilian is negligible (cf. Saldanha, 1957), the result can be explained by a similarly large Italian component in those populations.

Middle Phalangeal Hair

Absence of hair in the middle segment of the fingers seems to be under genetic control. Danforth (1921) suggested that the absence of middle phalangeal hair should be recessive over its presence and Bernstein and Burks (1942) found some evidence for polyallelic inheritance. Genes determining the presence of middle phalangeal hair on a larger number of fingers would be dominant over those responsible for the occurrence on a lesser number or on no fingers. Populational variation of the trait has been shown by Boyd (1950, p. 285) so that the presence of middle phalangeal hair seems to be a northern European characteristic (Bernstein, 1951).

Fingers of 131 males and 158 females were observed by means of a pocket lens. Table 30 shows the distribution and frequencies of individuals without middle phalangeal hair, according to sex and parental origin.

Sex differences in frequency of individuals without middle phalangeal hair is highly significant ($\chi^2_{(1)} = 5.60$, $P = 0.02$) but among the four different groups of the population, it was not heterogeneous ($\chi^2_{(3)} = 5.17$, $P = 0.15$). The frequency (26.70 ± 3.18) of individuals without middle phalangeal hair among Dutch immigrants (Saldanha et al., 1960) and that (50.00 ± 3.54) among mixed northeastern Brazilian populations (unpublished) can be taken as extreme limits in Brazil. The frequency (ca. 45%) among 52 individuals of Italian derivation in the United States (Bernstein and Burks, 1942) agrees with that observed in the parish of Capivari (48.10 ± 2.94) but it differs from the figure (26.99 ± 1.91) among whites of Baltimore (Glass et al., 1952).

Ear Lobe Attachment

Studies on pedigrees by Powell and Whitney (1937) suggested that free ear lobe is dominant over the attached. Wiener (1937) published family data which do not confirm that assumption because of the difficulty of classifying the several intermediate grades of ear lobes observed. On the basis of family studies (Kloepfer, 1946) has

shown that the variation in ear lobe pattern is genetic. However, Suzuki's (1950) analysis of 1130 families could not support the statement that free lobe is a dominant trait. Data so far published indicate that the distribution of ear lobe types exhibits racial differences and attached ear lobes seem to be an African characteristic (Gates, 1954).

The ear lobe attachement of 130 males and 158 females were examined and classified in three types: (a) completely attached lobe (attached), (b) completely free

Tab. 30. Distribution of middle phalangeal hair and frequency of subjects without the trait in the parish of Capivari, according to sex and parental origin

Group	Number	No. of fingers with phalangeal hair					Per cent of subjects without phalangeal hair	Heterogeneity
		0	1	2	3	4		
Desc. of Brazilians	57	26	8	9	8	6	45.61	
								$\chi^2_{(3)} = 5.17$
Desc. of Italians	91	38	21	16	12	4	41.76	
								P = 0.15
Desc. of Brazilians and Italians	70	33	12	12	9	4	47.14	
Remainder	71	42	13	5	7	4	59.15	
Total males	131	53	26	24	19	9	40.46±4.29	
								$\chi^2_{(1)} = 5.60$
females	158	86	28	18	17	9	54.43±3.96	
								P = 0.02
both sexes	289	139	54	42	36	18	48.10±2.94	

lobe (free), and (c) intermediate types of attachment (intermediate). Table 31 shows the distribution and frequencies of individuals with free ear lobes, classified according to sex and parental origin.

The frequency of individuals with free ear lobes among males and females is significantly different ($\chi^2_{(1)} = 4.32$, P = 0.04) but the heterogeneity among the groups classified according to parental origin is not significant ($\chi^2_{(3)} = 2.24$, P = 0.55). Figures for Italian populations are not available. The overall frequency (22.57±2.46) observed in the parish of Capivari differs appreciably from those among whites in the United States (40.53±2.52; cf. Glass et al., 1952), Dutch immigrants in Brazil (11.40±2.29; cf. Saldanha, et al., 1960) and northeastern Brazilian populations (16.00±2.50; unpublished).

Colour Blindness

Normal colour vision is trichromatic but some individuals can be dichromatic or even monochromatic. The commonest colour vision anomaly in man is sex-linked red-green blindness. Frequencies of colour-blind individuals varies fairly in different populations (cf. Boyd, 1950, p. 289) and the condition is probably under the action of selective factors.

Colour vision of 131 males and 158 females was tested by means of Ishihara's tables. The following types of colour vision defects were identified: deuteranopia (D),

Tab. 31. Distribution of type of ear lobes and frequency of subjects with attached lobes in the parish of Capivari, according to sex and parental origin

Group	Number	Type of ear lobe			Per cent of subjects with attached ear lobes	Heterogeneity
		free	interme-diate	attached		
Desc. of Brazilians	56	33	9	14	25.00	
						$\chi^2_{(3)} = 2.24$
Desc. of Italians	91	40	30	21	23.08	
						P = 0.55
Desc. of Brazilians and Italians	70	30	22	18	25.71	
Remainder	71	46	13	12	16.90	
Total males	130	73	35	22	16.92±3.29	
						$\chi^2_{(1)} = 4.32$
females	158	76	39	43	27.22±3.54	
						P = 0.04
both sexes	288	149	74	65	22.57±2.46	

deutero-protanopia (DP), incomplete redgreen blindness (IRG), and incomplete green blindness (IG). Table 32 shows the distribution of colour vision anomalies in males and females. Males were further classified according to parental origin.

According to the theory of sex-linked colour blindness, the square of the frequency of colour-blind males should be equal to the frequency of colour vision defects to be found among females, provided that the population approached genetic equilibrium. A test for checking this situation (cf. Snyder, 1951) is obtained by:

$$\sqrt{\text{affected females}} - \sqrt{\text{affected males}} = 0.69 \pm 0.043,$$

https://doi.org/10.1017/S1120962300020321 Published online by Cambridge University Press

Since the deviation differs significantly from 0 (d/s.e. = 16, P<0.001), the results suggest that the population for this sex-linked trait is far from genetic equilibrium.

Frequencies of colour-blind males among groups classified by parental origin are not heterogeneous ($\chi^2_{(3)} = 3.40$, $P = 0.35$) but, since colourblindness is not present in the " hybrid " group, the non-significant result should be due to the small-

Tab. 32. Distribution of colour vision anomalies and frequency of colour-blind subjects in the parish of Capivari among males and females. Males were also classified according to parental origin

Group	Number	Type of colour-blindness					Per cent of colour-blind subjects	Heterogeneity
		D	DP	IRG	IG	Tot.		
Desc. of Brazilians (l. c.)	27	1	—	1	—	2	7.41	$\chi^2_{(3)} = 3.40$
Desc. of Italians (l. c.)	43	2	1	—	—	3	6.98	$P = 0.35$
Desc. of Brazilians and Italians (l. c.)	27	—	—	—	—	—	—	
Remainder (l. c.)	34	2	1	1	—	4	11.76	
Total males	131	5	2	2	—	9	6.87±2.21*	
females	158	—	—	2	1	3	1.90±1.09	

* Gene frequency estimated by means of Maximum likelihood method = 0.0920±0.213.
D – deuteranopia; DP – Deutero-protanopia; IRG – incomplete red-green blindness;
IG – incomplete green blindness.

ness of sample sizes. The gene frequency estimated by means of the maximum likehood method is 0.0920±0.0213. The overall frequency of colour vision defects among males in the parish of Capivari is not appreciably different from those observed among other white populations.

Correlation Between Common Traits

Correlation tests for the distribution of taste sensitivity to phenylthiourea, middle phalangeal hair and ear lobe attachment can provide further information about the " dynamics " of the present population. As was considered above, such information depends on the differences in frequencies of the traits among the original populations and also the sample sizes on which the calculations are based. Table 33 shows

Tab. 33. Estimates of correlation between taste sensitivity to phenylthiourea, middle phalangeal hair and ear lobe attachment

Traits		Middle phalangeal hair		Ear lobe type		χ^2 P (1 d. f.)		Q ± s.e.
		with	without	non-attach.	attached			
Taste sensitivity to phenylthiourea	tasters	117 (112)	96 (101)	—	—	1.77	0.16	0.176±0.130
	non-tasters	35 (40)	41 (36)	—	—			
Taste sensitivity to phenylthiourea	tasters	—	—	167 (164)	45 (48)	0.83	0.35	0.140±0.152
	non-tasters	—	—	56 (59)	20 (17)			
Middle phalangeal hair	with	—	—	115 (117)	35 (33)	0.39	0.55	0.089±0.142
	without	—	—	110 (108)	28 (30)			

The figures between brackets stand for the expected numbers.

the estimates for the three possible correlations, calculated by means of Chi-square test and Yule's coefficient of association (Q).

No correlation was significant at the 5% level of probability and the coefficients of association are quite small. These results suggest that the frequencies of the traits considered in the calculations are close to the equilibrium.

VII. The relative effectiveness of certain evolutionary factors on the genetic structure of the present population

General consideration

In this section, an attempt will be made to evaluate some genetic and demographic data obtained for the population studied, according to the general principles of population genetics, mainly those worked out by Wright (1931, 1938, 1939, 1940, 1942, 1946, 1948 and 1951). However, it must be emphasized that the actual circumstances under which the data were obtained hardly meet the necessary theoretical over-simplified requirements. Even so, it seems to the present writer that such considerations are of some practical value in suggesting relevant points to be considered in further theoretical elaborations.

In this context, an important parameter to be estimated is the effective size of the population. While both isolate and neighbourhood sizes are probabilistic estimates, the effective population size should be the actual number of parents contributing to the next generation in a breeding unit. This number should be less than the total number of adult individuals in the population. Some breeding populations are stable and this situation means that the average couple produces, in each generation, the necessary number of children to substitute for them in the following generation. Furthermore, the breeding population size can vary from generation to generation. In the case of cyclic variation, the effective population size is more closely related to the minimum than to the maximum number (Wright, 1942). For the specifical case of human populations with sex-ratio approximately equal to unity, the effective population size can promptly be obtained by:

$$N = \frac{N_p\bar{k}(N_p\bar{k} - 1)}{(N_p - 1)\ \sigma_k^2 + N_p\bar{k}(\bar{k} - 1)}, \qquad \text{VII. 1}$$

provided that the number of parents (N_p) and mean ($\bar{k}$) and variance (σ_k^2) of the number of children per couple are known.

Since a stable population should indicate that $\bar{k} = 2$, in this situation the effective population size can be obtained by:

$$N = \frac{4\ N_p - 2}{\sigma_k^2 + 2}, \qquad \text{VII. 2}$$

https://doi.org/10.1017/S1120962300020321 Published online by Cambridge University Press

assuming that family size has a Poisson distribution ($\bar{k} = \sigma_k^2$). Even for $\bar{k}$ values slightly larger than 2, the two formulae should produce differences which can lead to erroneous conclusions. If the family size departs from Poisson distribution, effective population size should be different from the actual number of parents in the population. This situation can be defined as an index of variability in progency ($i = \sigma_k^2/\bar{k}$, cf. Crow and Morton, 1955). When *i* value is greater than unity, effective population size should be less than the actual number while the opposite result is to be expected for *i* values less than unity.

Human populations, similarly to many other animal species, are often scattered as relatively small breeding groups. This situation is more likely to be found among primitive and rural communities. Wright (1931, 1939, 1940, and 1948), on theoretical basis, showed that, for a population of effective size equal to N, when $4N\mu$, $4N\delta$ or 4Nm (where *u*, *s* and *m* stand for mutation rate, selective coefficient and migration rate, respectively) are larger than unity, evolutionary changes should be caused by systematic factors (mutation, selection and migration); but for values less than 1, genetic differentiation should be determined mainly by random fluctuation in gene frequency. So, evolutionary process among small isolated human communities is supposed to depend strongly on the effective population size. In this situation, the heterozygosis for every locus is decreased by 1/(2N) in each generation (for discussion, Kimura, 1955) and the variance of gene frequency (q) for a particular locus, is obtained, according to the binomial theorem, by:

$$\sigma^2_{\delta q} = \frac{q\ (1\text{-}q)}{2N} \quad (12) \qquad \text{VII. 3}$$

However, human populations hardly ever meet the assumption of complete (or nearly complete) isolation, so that the application of formula VII.3 to human populations will often be misleading.

A more realistic assumption must consider human populations as being made up of semi-isolated breeding units among which intermixture is occurring (cf. chapter II). The rate of admixture between two breeding isolates should be proportional to their distance. In a population subdivided into small breeding groups of size N, the homozygote frequency is increased by the occurrence of consanguineous matings of different types. If the net effect of consanguinity is measured by $F(=\alpha)$, the inbreeding coefficient of the population, the homozygote proportion for a particular gene with frequency *q* is $q^2+Fq(1\text{-}q)$ (cf. p. 163). The inbreeding effect is similar to that due to variation of gene frequency among the breeding units (Wahlund's effect, cf. p. 162). Comparing the homozygote frequencies prevailing for the two situations (cf. Wright, 1943; see also Li, 1955, p. 299), the following relation is obtained:

$$\sigma^2_{\bar{q}} = F\bar{q}(1\text{-}\bar{q}) \text{ or } F = \frac{\sigma^2_{\bar{q}}}{\bar{q}(1\text{-}\bar{q})}, \qquad \text{VII. 4 (a, b)}$$

where $\bar{q}$ is the mean gene frequency in the whole population.

(12) The exact value for the *nth* generation is obtained by: $\sigma^2 = q(1\text{-}q)\ [1\text{-}(1\text{-}1/2N)^n]$ (Wright, 1942, p. 231).

Intermixture between a particular breeding group and the rest of the population tends to level gene frequencies and prevent the breeding unit from approaching homozygosis. Thus, the variance σ_q^2 is reduced to $(1\text{-}m)^2\sigma_q^2$ in each generation (for discussion see Li, 1955, p. 302). When equilibrium is reached, the mean value of F remains constant from generation to generation, according to the relation (Wright, 1951):

$$F = (1\text{-}m)^2 \left[\frac{1}{2N} + \frac{2N\text{-}1}{2N}.F'\right],$$

where F' stands for the mean coefficient of inbreeding in the previous generation. From the considerations above (cf. Wright, 1931, 1943; also Li, 1955, p. 320), F and σ_q^2 are obtained by the following formulae:

$$F = \frac{(1\text{-}m)^2}{2N\text{-}(2N\text{-}1)\ (1\text{-}m)^2} \text{ and } \sigma_q^2 = \frac{\bar{q}(1\text{-}\bar{q})}{2N\text{-}(2N\text{-}1)\ (1\text{-}m)^2} \qquad \text{VII. 5 (a,b)}$$

If the migration rate is small, the above expressions can be re-stated as: (cf. Li, 1955, p. 305):

$$F = \frac{1}{4Nm+1} \text{ and } \sigma_q^2 = \frac{\bar{q}(1\text{-}\bar{q})}{4Nm+1} \qquad \text{VII. 6 (a, b)}$$

As a general statement, random differentiation of sub-populations is expected to be appreciable only when F approaches 0.05 (Wright, 1951). Since F is inversely proportional to m, the two parameters can be used to check one another, and the above consideration can be put in a different way (cf. Li, 1955, p. 308). According to formula VII.6.a, random fluctuations in gene frequency can be of evolutionary importance only when the value Nm (varying inversely to F), become less than 5($4Nm = 20$). The amount of genetic differentiation measured by Nm and referred to as an " isolation index ", has been emphasized by Lasker (1960). However, the theoretical concepts of population genetics are useful only if practical limitations concerned in the study of actual human populations are considered.

Effective population size, genetic drift and inbreeding coefficient of the present population

The effective population size (N) for rapidly increasing populations is very close to the number of parents in a particular generation (for discussion see: Crow and Morton, 1955). For relatively stable populations, N does not differ appreciably from Dahlberg's isolate size (n). In practical investigations, however, no clear distinction between N_p, N and n has been made (Glass et al., 1952, Lasker, 1954; Böök, 1956) and N has been roughly taken as one fifth of the total population (Lasker, 1954).

Assuming that the sex-ratio is not appreciably different from unity and that matings are permanent, the number of women between 15-39 (a generation) (13) multiplied by 2 (to include males) should represent reasonably the average number of parents in the present generation (N_p). According to the General Census of Brazil (1940) there were about 1800 women belonging to that age range in the parish of Capivari, so that $N_p = 3600$. This figure is not very much different from isolate size 3428 (cf. Table 15) for the present population. The mean and variance of the number of children per couple were respectively 4.13 and 4.99 which must be regarded as upper limits (see p. 15). (14) By applying formula VII.1 to these data, the effective population size is: Ne = 3427.

It is interesting to notice that the N figure obtained by means of formula VII.2 is appreciably lower (ca. 2000). In a previous consideration of these data (Saldanha, 1959) all conclusions were based on the latter estimate. However, it seems that the N value based on formula VII.1 is more appropriate and it is not different from the isolate size.

Now, suppose that mutation rate and selective pressure are negligible enough to be considered ineffective in an unique generation, so that changes in gene frequencies in the present population depend only on migration rates and random gene fluctuations. Suppose, further, that the population consists of several breeding units like that of the parish of Capivari. In this situation the variance of the gene responsible for taste insensitivity to phenylthiourea (for example) can be calculated by means of formula VII.5.b as $\sigma_g^2 = 0.000067$, with standard deviation: 0.515+0.08. This figure is not appreciably different from that (0.515+0.06) estimated by formula VII.3. It can be calculated that fluctuations in gene frequency within the limits 0.499 and 0.531 should have a probability of 95% (2.sd). Under such circumstances, it is apparent that genetic drift is absolutely ineffective. In the absence of systematic evolutionary factors, the loss of heterozygosis in the population studied should be $1/(2N) = 0.015\%$. Assuming that human genetic diversity is due to 2000 " not fixed " loci (ca. 10% of total human loci), that figure means that no measurable genetic change can be attributed to random fluctuations. The same conclusion is indicated by the product Nm obtained for the parish of Capivari. The figure Nm = 1080 is quite large, so that genetic differentiation cannot be explained by random genetic drift.

From the frequencies of different types of cousin marriage (cf. Table 17) the inbreeding coefficient of the present population was estimated as $\alpha = 0.00044$. Estimates of mean inbreeding coefficient can also be obtained indirectly. Assuming that the present population was derived from a " virtual " population in which the frequency of non-tasting gene for phenylthiourea would be the mean (0.599) between Brazilian whites and northeastern Italians (cf. p. 204), the coefficient of inbreeding

(13) Since women used to marry relatively young in the community investigated, 25 years is a good estimate of the average generation period.

(14) Index of variability in progeny for the population investigated is, $i = \sigma_k^2/\bar{k} = 1.21$.

is estimated as $F_{\sigma q} = 0.00028$ by means of formula VII.4.b. Similarly an estimate of the mean inbreeding coefficient can be based on the migration rate ($m = 0.315$). By using formula VII.5.a it is calculated as $F_m = 0.00014$, a figure appreciably lower. The indirect estimates of F should indicate that the overall frequency of cousin marriage in the present generation of the parish of Capivari is somewhat higher than that expected at random. However, indirect estimates of F based on effective population size obtained by formula VII.2 are appreciably similar. These figures were $F_{\sigma q} = 0.00041$ and $F_m = 0.00040$ (Saldanha, 1959). No conclusion can be taken too seriously because of the oversimplifications supporting the estimates.

Gene flow into the present population

The effect of immigration into the present population of the parish of Capivari can be evaluated another way. It is apparent from table 10 that immigration rates into the population investigated has varied from time to time. The community, since its foundation, has interchanged migrants with other communities but the immigration rates became appreciably higher following non-Brazilian migratory movements into the community after 1890. As discussed in chapter II, migration is relevant to genetic problems particularly when there are differences between the populations concerned. Since the alien immigrants came mainly from northeastern Italy (fig. II) the genetic changes in the population studied are expected to be due mostly to the genetic differences between northeastern Italians and the descendants of Brazilian settlers of the parish of Capivari. By considering only gene frequencies among northeastern Italians (q_x), " pure " Brazilian whites (q_o) and the present population in the parish of Capivari (q_k), the relative Italian and non-Italian (mostly Brazilian) components can be calculated by means of Ottensooser's (1944) formula, extended by Glass and Li (1953) to estimate the average gene flow per generation (m):

$$(1-m)^k = \frac{q_x - q_k}{q_x - q_o} \qquad \text{VII. 7}$$

Using the frequency of the non-tasting gene for phenylthiourea among northeastern Italians (see p. 204), Brazilian whites in Rio de Janeiro (Saldanha and Guinsburg, 1954) as representative of the parent populations and assuming two generations of immigration contact ($k=2$), the percent of Italian and non-Italian admixture and the average gene flow per generation from the populations in northeastern Italy into the community of Capivari were calculated by means of formula VII.7. The results are:

q_x	q_o	q_k	k	Italian admixture (%)	non-Italian admixture (%)	m
0.486 ± 0.007	0.552 ± 0.027	0.515 ± 0.021	2	56.1	43.9	0.337

Table 9 shows direct evidence that among the individuals living in the present population of the parish of Capivari 55% are of Italian origin, that is, 46% have both and 18% one parent of Italian descent, while 45% are non-Italian (including about 35% of Brazilian ancestry), in the sense that both or at least one parent was of non-Italian origin. These figures are in complete agreement with the indirect estimate shown above.

The gene flow from white into Negro populations has been investivated in the United States (Roberts, 1955; Glass, 1955) and in Brazil (Saldanha, 1957). In these investigations, figures obtained do not exceed 5%. The value obtained in the present study (33%) is appreciably higher. However, it is not very reliable because the difference in gene frequency between the basic populations is not very large.

Conclusion

It is of some interest to compare the figures obtained in the investigation of the parish of Capivari with those for communities under different ecological and social situations. Table 34 shows the estimates of some parameters relevant to population genetics obtained for American aborigenes, African tribes and white populations by different investigators. In order to make the figures comparable, most of the estimates presented in table 34 were re-calculated by means of more appropriate formulae. Since the major part of the papers did not indicate the demographic data from which N was estimated, no attempt has been made to check these figures.

From table 34 it can be seen that random fluctuations in gene frequency are expected to be important as potential evolutionary factor only among communities under very particular situations. This can be the case for some Brazilian groups (Lasker, 1954) but they represent an extreme situation. In very isolated communities as that formed by " Dunkers ", a religious isolate (Glass et al., 1952) or in the population now living in Susak Island (Dolinar, 1960), random genetic drift can account for some changes in gene frequencies. However, the critical (actual) evaluation of the operation of that evolutionary factor is only possible by comparing gene frequencies in successive generations. While differences were found for the MN blood group system in the " Dunker " isolate (Glass, 1956) no discrepancy was detected in two successive generations for ABO (the only blood group investigated) in Susak Island (Dolinar, 1960, p. 19). Differences in frequency of genetic traits between geographical or social isolates and the populations from which they have been derived have been often attributed to random genetic drift and/or selection (Sanghvi and Khanolkar, 1949; Birdsell, 1950; Laughlin, 1950; Glass, 1954; Sirsat, 1956; Kraus and White, 1956; etc.). Such isolates should spring from relatively small numbers of migrant settlers so that random or selective differential emmigration, under such circumstances, is more likely to account for the differences in genetic traits. Although the genetic effect of differential migration might be similar to that of random genetic drift, the theoretical implications of both factors are quite different.

Tab. 34. Some parameters relevant to the evaluation of population structure for American aborigenes, African tribes and white communities studied by different unvestigators (see text)

Community	Country	N *	m	Nm	$\pm \sigma_q$ (p−q)**	F_m **	F_{oq}	α	½ N (%)	½ N x 2000	Reference
American Aborigenes:											
Camayura Indians (lower limit)	Brazil	23	0.12	3	±0.145	0.06944	—	—	2.174	42	Lasker, 1954
Paracho Indians (upper limit)	Mexico	967	0.202	195	±0.019	0.00091	—	—	0.052	1	Lasker, 1954
African Tribes											
Tir - Dinka village	Sudan	109	0.50	55	±0.039	0.00153	—	—	0.459	9	Roberts, 1956
Abuya - Dinka section	Sudan	380	0.35	133	±0.024	0.00098	—	—	0.132	3	Roberts, 1956
Ageir - Dinka tribe	Sudan	3913	0.11	430	±0.012	0.00049	—	—	0.013	0	Roberts, 1956
a) *White populations (beginning of the XIX century):*											
Parish M-a	Sweden	362	0.26	94	±0.028	0.00167	—	—	0.138	3	Alström, 1958
Parish K-g	Sweden	178	0.41	73	±0.033	0.00150	—	—	0.281	6	Alström, 1958
Parish A-y	Sweden	267	0.53	142	±0.025	0.00053	—	—	0.187	4	Alström, 1958
Parish R-a	Sweden	144	0.59	85	±0.032	0.00070	—	—	0.347	7	Alström, 1958
b) *White populations (present time):*											
Religious Isolate (« Dunkers »)	USA	90	ca. 0.15	14	±0.070	0.01417	0.0254*	—	0.555	11	Glass et al., 1952
Susak Island	Yugoslavia	413	0.044	18	±0.059	0.01269	0.02004**	—	0.121	2	Dolinar, 1960
Capivari (rural community)	São Paulo, Brazil	3427	0.315	1080	±0.008	0.00013 (0.00023)***	0.00028	0.00044	0.015	0	Present paper

Key: N - effective population size; m - immigration rate; σ_q - standard deviation of gene frequency estimated by means of formula VII.5.b and assuming p=q (= ca. 68% probability of drift in gene frequency); F_m - inbreeding coefficient estimated by means of formula VII.5.a F_{oq} - inbreeding coefficient estimated by means of formula VII.4.b; α - inbreeding coefficient estimated from the frequencier of consanguineous marriage; 1/2N - probability of heterozygosity decrease assuming 2000 " not fixed " loci;

* These figures were not recalculated because demographic data were not available in the original papers.

** Calculated in the present work or recalculated by using more appropriate formulae (see above).

*** Calculated by means of formula VII.6.a.

https://doi.org/10.1017/S1120962300020321 Published online by Cambridge University Press

It can be seen even in the particular circumstances shown in table 34 that random fluctuations in gene frequency seems to deserve secondary importance as an evolutionary factor among human populations. Since " inter-isolate " migration is very active even among primitive and rural communities, the most extreme effect of random genetic drift should be counterbalanced by the effect of relatively high migration rates. Therefore, in relation to a relatively short period of time, intermigration and admixture should probably be the most important evolutionary factor which should presently be pushing back the inter-group differences made up by selection during a very long time.

It is interesting to observe that among the communities of the Sudan (Roberts, 1956) the " potential " effect of genetic drift is directly proportional to the migration rates precisely because of the " artificial " bounding of " natural " isolates. If the villages are considered as the breeding units, the effective population size is very small but the observed migration rates are too large to be expected a quite significant effect from random fluctuations in gene frequency. Otherwise, if the whole tribe is considered as the breeding unit, the migration rate from outside of the population is relatively low but the effective population size is appreciably larger, so that practically the potential chance for genetic drift cannot account for any genetic change.

In the present investigation, perhaps the most relevant point is that the parish of Capivari represents a common situation to be frequently found at present in different places of the world, mainly in America. So, in this phase of human biological history, genetic changes are determined mostly by migration and admixture of populations with different genetic make up, provided that a short period of time is considered. The present pattern of human evolution is such that cultural factors are relaxing the intensity of natural selection but they are appreciably increasing the dispersion of human beings. Since the selective coefficient of many human genes has approached the neutrality level, the mean effective population size has increased and mutation in the human species is a rare event (probably the average value is not above 10^{-5}), evolutionary changes should be determined proportionately more by migration and admixture, in the future. However, before the " ideal " genetic equilibrium is reached (if ever) it will be " necessary " to have considerable changes in ethical values and/or prejudices of civilized people inhabiting very extensive geographical areas.

VIII. Summary and conclusions

A demographic and genetic investigation was carried out in a rural community (Parish of Capivari) of the State of São Paulo. This parish was chosen because it is located in an area which received very large numbers of northeastern Italian immigrants. The most important points and conclusions of the study were:

1. Because the present investigation is concerned with the " breakdown " of an isolate, some methodological considerations are discussed, mostly those of Wahlund, Dahlberg, Wright, and Haldane. It is concluded that the " break-down of isolates ",

that is, the increase of human dispersion should determine: (*a*) decrease of random inbreeding rates expected as a consequence of increase of population size; (*b*) decrease of variance in gene frequency (Wahlund's effect); (*c*) increase of heterozygote frequency for both rare and common genes, permitting new genetic combinations to be " tested "; (*d*) chance decrease of random genetic drift as a consequence of the increase in population size; and (*e*) disturbance of equilibrium between mutation and selective elimination rates for recessive genes because of the decrease of homozygote frequency caused by (*a*). As a consequences of (*e*) the gene frequency of recessive genes is increasing.

2. The demographic characteristics of the community were presented and the origin of the population was discussed. It was shown by means of examination of the Roman Catholic marriage records, that, from 1890 to 1920, the parish of Capivari received very large numbers of immigrants. The parish registers revealed that most of the immigrants came from three Italian regions: Veneto, Lombardia, and Emilia-Romagna. It is also shown, on the basis of the parental origin of 3253 males whose files are recorded in the Office of Recruitment for Obligatory Military Service, that more than 50% of the individuals of the present generation have, at least, one parent of Italian derivation. Some data concerned with female fertility and number of children per family in the present population were considered.

3. The " break-down of the isolate " was analysed by means of immigration rates and frequencies of consanguineous marriages for different periods of time. The analysis was based on information obtained from Roman Catholic marriage records. The study of dispersion was based on the birth place of the individuals marrying into the community during the following periods: 1830-35, 1860-65, 1890-95, 1920-25, and 1950-55. The trend of immigration rates has shown, in agreement with more indirect demographic data, that " the break-down of the isolate " commenced about two generations ago. The migratory movement, because of its extent, should have caused deep changes in the genetic structure of the community.

The neighbourhood concept was applied to the migration data. The mean and variance of distance between birth and marriage places were presented for different periods, including and excluding non-Brazilian individuals. The distance analysis has confirmed the conclusions obtained by other procedures. However, it was concluded that the neighbourhood concept as applied to civilized populations, particularly those under complex demographic, cultural and social conditions, should not produce sound results because human dispersion, as a populational movement, is not a random process.

The frequencies of different types of consanguineous marriage were obtained from Roman Catholic marriage records for the following periods: 1830-35, 1860-65, and 1890-1955. From these data the mean coefficient of inbreeding and isolate size (based on the frequency of first cousin marriage) were estimated. A progressive and uniform decrease of cousin marriage rates was demonstrated following 1890, when immigration into the community increased. From 1920 the inbreeding rates were shown to be stable, on a very low level. During this period the mean coefficient

of inbreeding decreased to about one tenth. Since inbreeding rate reflects isolate size, the consanguinity data suggest a marked " break-down of the isolate ".

An attempt was made to consider critically the available consanguinity data. The frequencies of first cousin marriage based on the catholic registers were compared with those obtained from civil registers for three different periods. The differences found were discussed on the assumption of some biases concerned with the information sources of consanguinity data. Non-randomness in frequency of first cousin marriage was checked by comparing the observed rate in the present generation with that expected from the actual number of parents between 15 and 39 (25 years per generation) and the average number of children per family observed in the parish of Capivari. The expected frequency on the basis of Dahlberg's formula was very close to the figure obtained from catholic registers. Assuming that first cousin marriages were occurring at random, a tentative evaluation was made of the extent that observed frequencies of other types of consanguinity depart from the values expected according to the isolate theory. It was concluded that the isolate concept, from a formal point of view, can only represent a situation prevailing among stable and isolated small populations. However, the inter-regional comparisons of isolate sizes estimated from the frequencies of the same type of cousin marriage was believed to be a good indication of the genetic isolation of the individuals in different places.

4. The frequency of some congenital abnormalities in the present population of the parish of Capivari was analysed. Information was obtained from the examination of 7746 children in the Public Health post of the community. The individuals were classified, according to parental origin, in four groups: descendants of Brazilians, descendants of Italians, descendants of Brazilian and Italian and a heterogeneous remaining group. The following defects were considered: hare lip and cleft palate, malformations of upper and lower extremities, congenital heart diseases, mongolism, epilepsy, inguinal hernia and gross mental retardation. A brief consideration of the hereditary basis of each abnormality was presented. The overall frequency of the congenital defects among the groups classified by parental origin was significantly different. Only one abnormal child (a mongolian idiot) was found in the " hybrid " group. The results were discussed on theoretical grounds and some explanations were suggested.

5. The distribution of stature and six other " normal " hereditary traits were also studied in the present population of the parish of Capivari. The investigation of stature and eye colour was based on the files of 2882 males examined for military service. The investigation of the remaining traits, namely, taste sentitivity to phenylthiourea, middle phalangial hair, ear lobe attachment, and colour-blindness was based on the examination of 290 individuals, 93 males and 97 females, in the public local high school Individuals were classified, for each trait, in four categories: descendants of Brazilians, descendants of Italians, descendants of Brazilians and Italians and a heterogeneous remaining group.

The genetic basis of stature was reviewed and the present trend of average stature among human populations was discussed. Some relevant hypotheses, mainly those

proposed by Dahlberg and Penrose were discussed under ... Consideration of recent publications. Mean stature among the four groups, classified by parental... origin, was shown to be significantly different. While the average stature of the " hybrid " group was quite similar to that of the tallest, its variance was significantly less than those of " pure " groups. Since the three relevant groups were apparently living under the same ecological and social conditions, it was suggested that the differences should be caused by genetic factors. It was considered that the data support the hypothesis of dominant genes determining human stature. The alternative explanation based on overdominant genes could not be discarded. However, it was emphasized that differences in variance were too large to be explained by hereditary factors alone.

Distribution of eye colour among the four groups was significantly heterogeneous. No correlation between stature and eye colour was found. The mean heights of dark-eyed and light-eyed groups were not significantly different.

The distribution of the remaining five common traits was analysed and the genetic basis of each was briefly discussed. Only the distribution of taste sensitivity to phenylthiourea was significantly different among the four groups classified by parental origin. No colour-blind was found in the " hybrid " group and the observed frequency of colour blind females departed significantly from the expected one on the basis of sex-linkage theory. Correlation tests between taste sensitivity to phenylthiourea, middle phalangial hair and ear lobe attachment, produced no significant result at the 5% level of probability in every case.

6. The relative effectiveness of evolutionary factors causing genetic changes in the present population was considered. Some theoretical concepts and formulae, mainly those developed by Wright were briefly presented. On the basis of demographic and genetic data obtained for the parish of Capivari, the effective population size was estimated. Variance of gene frequency and the probability of random fluctuations in gene frequency were too low to be of evolutionary significance.

Mean inbreeding coefficients were calculated indirectly from the migration rates and from the gene frequencies observed in the present population. These figures were compared with the values obtained directly from the frequencies of different types of cousin marriages. By considering the probable basic populations from which the population of the parish of Capivari was derived, the relative amount of Italian and non-Italian admixture was estimated. These figures were compared with those obtained by a more direct procedure.

The parameters related to population structure of the parish of Capivari were compared with those obtained from other communities under different ecological and social conditions. The relative importance of random genetic drift and migration among recent human populations was discussed. It was emphasized that the conditions prevailing in the parish of Capivari represent a relatively common situation in America, so that migration and admixture should be an important evolutionary factor in the present period of human biological history.

IX. References

ALSTRÖM, C. H.: First cousin marriages in Sweden 1750-1844 and a study of the population novement in some Swedish subpopulations from the genetic-statistical view - point. Acta genet. (Basel), *8:* 295-369 (1958).

BERNSTEIN, M. M.: The middigital hair genes. Their inheritance and distribution among white races. J. Hered., *40:* 127-131 (1949).

— and BURKS, B. S.: The incidence and mendelian transmission of mid-digital hair in man. J. Hered., *33:* 45-53 (1942).

BIRDSELL, J. B.: Some implications of the genetical concept of race in terms of spatial analysis. Cold Spr. Harb. Symp. quant. Biol., *15:* 259-314 (1950).

BLAKESLEE, A. F.: Corn and men. J. Hered., *5:* 511-518 (1914).

BOAS, F.: Changes in bodily form of descendants of immigrants (Columbia Univ. Press, New York 1912).

BÖÖK, J. A.: The incidence of congenital diseases and defects in a South Swedish population. Acta genet. (Basel), *2:* 289-311 (1951); Genetical investigations in a North-Swedish population. Population structure, spatic oligophrenia, deaf mutism. Ann. hum. Genet., *20:* 239-250 (1956).

BOWLES, G. T.: New types of old Americans at Harvard (Harvard University Press, Cambridge, Mass. 1932).

BOYD, W. C.: Genetics and races of man. (Little Brown, Boston 1950).

BROMAN, B.; DAHLBERG, G. and LICHTENSTEIN, A.: Height and weight during growth. Acta paediat., (Uppsala) *30:* 1-66 (1942).

BRUES, A. M.: A genetic analysis of human eye colour. Amer. J. phys. Anthrop., n. s. 4: 1-36 (1946); Linkage of body build with sex, eye color, and freckling. Amer. J. Hum. Genet., *2:* 215-239 (1950).

CAMPOS, V.: Fundações municipais paulistas nos séculos XVIII e XIX. Tomo I. Capivari (Instituto Histórico e Geográfico de São Paulo, São Paulo, Brasil 1952).

CARNEIRO, J. F.: Imigração e colonização no Brasil (Publ. avulsa no. 2, Cadeira de Geografia do Brasil, Faculdade Nacional de Filosofia, Universidade do Brasil, Rio de J. 1950).

CARTER, C. and MCCARTHY, D.: Incidence of mongolism and its diagnosis in the newborn. Brit. J. prev. soc. Med., *5:* 83-90 (1951).

CAVALLI-SFORZA, L. L.: Some data on the genetic structure of human populations. Proc. Inter. Congr. Genet., *1:* 389-407, Montreal 1958; Demographic attacks on genetic problems. Some possibilities and results (Seminar on the use of vital statistics and health for genetic and radiation studies, UN/WHO 1960).

CLARK, P. C.: The heritability of certain anthropometric characters as ascertained from measurements of twins. Amer. J. Hum. Genet., *8:* 49-54 (1956).

CONRAD, C.: Die erbliche Fallsucht; in Guetts' Handbuch der Erbkrankheiten, vol. 3, part 2 (Thieme, Leipzig 1940).

CONTERIO, F. and CAVALLI-SFORZA, L. L.: Evolution of the human constitutional phenotype: An analysis of mortality effects. Ric. sci., suppl. ano *29:* 1-14 (1959).

COON, C. S.: The races of Europe (Macmillan, New York 1939).

CROW, J. F. and MORTON, N. E.: Measurement of gene frequency drift in small populations. Evolution, *9:* 202-214 (1955).

CUNHA, A. X. and ABREU, M. D. A.: A sensibilidade gustativa da feniltiocarbamida em Portugueses. Contr. Anthrop. Portug., *5:* 85-96 (1956).

DAHLBERG, G.: Twin births and twins from a hereditary point of view (Bekförlages, Stockholm 1926); Inbreeding in man. Genetics *14:* 421-454 (1929); On rare defects in human populations, with particular regard to inbreeding and isolate effects. Proc. Roy. Soc. Edinb., *58:* 213-232 (1938); Rare psychological defects from the point of view of the population. Proc. 7th. Inter. Genetical Congr., p. 94, Edinb. (1939); Mathematical methods for population genetics. Karger, Basle/New York (1947); Genetics of human populations. Advanc. genet., *2:* 67-98 (1948).

DANFORTH, C. D.: Distribution of hair on the digits of man. Amer. J. Phys. Anthrop., *4:* 189-204 (1921).

DAS, S. R.: Inheritance of the P. T. C. taste character in man: An analysis of 126 Rárhi Bráhmin families of West Bengal. Ann. hum. Genet., *22:* 200-212 (1958).

DAVENPORT, C. B.: Heredity of human eye color. Bibl. genet., *3:* 443-463 (1927).

— and Love, A. G.: Army anthropology (Med. Dept. U. S. Army in the World War, vol. 15, part 1, Government print. off., Wash. 1921).
Davis, J. S.: The incidence of congenital clefts of and palate. Ann. Surg., *80:* 363-374 (1924).
Dolinar, Z.: Micro-evolution among the Susak Islanders. Inbreeding, sterility, blood groups and red hair. Ann. hum. Genet., *24:* 15-22 (1960).
Dunn, L. C.: The effects of isolates on the frequency of a rare human gene. Proc. nat. Acad. Sci., Wash., *33:* 359-363 (1947).
Ehrat, R.: Die Missbildungen der Neugeborenen an der Universitatsfrauen Klinik Zürich 1921-1944. Villiger, Zürich, 1948).
Finney, D. J.: Tests for sex-linkage in a quantitative character. Ann. Eugen. (Lond.), *9:* 203-207 (1939).
Fisher, R. A.: The correlation between relatives on the supposition of Mendelian inheritance. Trans. Roy. Soc. Edinb., *52:* 399-433 (1918).
— and Gray, H.: Inheritance in man: Boas's data studied by the method of analysis of variance. Ann. Eugen. (Lond.), *8:* 74-93 (1937).
Fogh-Andersen, P.: Inheritance of hare lip and cleft palate. Opera. ex Domo Biol. Hered. Hum. Univ. Hafn. (Copenhagen), *4:* 1-257 (1943).
Fraccaro, M.: Breeding structure of human populations - An analysis of distance. Eugen. Quart., 6: 32-35 (1959).
Freire-Maia, N.: Frequencies of consanguineous marriages in Brazilian populations. Amer. J. Hum. Genet., *4:* 194-203 (1952); Inbreeding in Brazil. Amer. J. Hum. Genet., *9:* 284-298 (1957).
Frota-Pessoa, O.: The estimation of size of isolates based on census data. Amer. J. Hum. Genet., *9:* 9-16 (1957).
Gates, R. R.: Étude sur le croissement de races. III Nouvelles observations concernant les oreilles, en particulier les lobes. J. Génét. Hum., *3:* 95-112 (1954).
Gini, C.: Alcuni resultati preliminari dell'indagine antropometrica sui soldati Italiani (Comitato Italiano per lo Studio dei Problemi della Popolazione, ser. 1, vol. 5, p. 589-607, Roma 1935); The distribution of stature is hypernormal. Acta Genet. (Basel), *1:* 361-376 (1949-1950).
Glass, B.: Genetic changes in human populations, especially those due to gene flow and genetic drift. Advanc. Genet., *6:* 95-139 (1954); On the unlikelihood of significant admixture of genes from the North American Indians in the present composition of the Negroes of the United States. Amer. J. Hum. Genet., *7:* 368-385 (1955); On the evidence of random genetic drift in human populations. Amer. J. Phys. Anthrop. n. s., *14:* 541-555 (1956).
Glass, B. and Li, C. C.: The dynamics of racial intermixture — an analysis based on the American Negro. Amer. J. Hum. Genet., *5:* 1-20 (1953).
— Sachs, M. S.; Jahn, E. F. and Hess, C.: Genetic drift in a religious isolate: an analysis of the causes of variation of blood group and other gene frequencies in a small population. Amer. Natur., *86:* 145-159 (1952).
Goldstein, M. S.: Demographic and bodily changes in descendants of Mexican immigrants (Inst. Latin-Amer. Stud., Austin, Texas 1943).
Haldane, J. B. S.: The spread of harmful autosomal recessive genes in human populations. Ann. Eugen. (Lond.), *9:* 232-237 (1939).
— and Moshinsky, P.: Inbreeding in Mendelian populations with special reference to human cousin marriage. Ann. Eugen. (Lond.), *9:* 321-340 (1939).
Hogben, L.: Nature and Nurture (London, 2nd ed. 1939, Allen and Unwin).
Holmgren, I.: The increase in the height of Swedish men and women from the middle of the 19th century up to 1930, and the changes in the height of the individuals from ages of 26 to 70. Based on measurements of 46,000 persons. Acta med. scand., *142*: 267-390 (1952).
Hooton, E. A.: Up from the ape (Macmillan rev. ed., New York 1946).
Howells, W. W.: Birth order and body size. Amer. J. Phys. Anthrop., n. s. *6:* 449-460 (1948).
Hulse, F. S.: Exogamie et hétérosis. Arch. suiss. Anthrop. Gén., *22:* 103-125 (1957).
Ito, P. K.: Anthropometric study of new-born infants of Japanese parents in America. Amer. J. Dis. Child., *52:* 321-330 (1936).
Kalmus, H.: Defective colour vision, P. T. C. tasting and drepanocytosis in samples from fifteen Brazilian populations. Ann. Hum. Genet., *21:* 313-317 (1957).
Kemp, T.: The frequency of diseases affected by heredity in Denmark. Cold Spr. Harb. Symp. quant. Biol., *15:* 129-140 (1950); Genetics and disease (Munksgaard, Copenhagen 1951).

KIMURA, M.: Solution of a process of random genetic drift with a continuous model. Proc. Nat. Acad. Sci., Wash. *41:* 144-150 (1955).

KLOEPFER, H. W.: An investigation of 171 possible linkage relationships in man. Ann. Eugen. (Lond.), *13:* 35-71 (1946).

KRAUS, B. S. and WHITE, C. B.: Micro-evolution in a human population: a study of social endogamy and blood type distributions among the Western Apache. Amer. Anthrop., *58:* 1017-1043 (1956).

LASKER, G. W.: Migration and physical differentiation. Amer. J. Phys. Anthrop., n. s. *4:* 273-300 (1946); Human evolution in contemporary communities. Southwestern J. Anthrop., *10:* 353-365 (1954); Migration, isolation, and ongoing human evolution. Human Biol., *32:* 80-88 (1960).

LAUGHLIN, W. S.: Blood groups, morphology, and population size of the Eskimos. Cold Spr. Harb. Symp. quant. Biol., *15:* 165-173 (1950).

LEJEUNE, J.; TURPIN, R. and GAUTHIER, M.: Le mongolisme, le premier exemple d'aberration autossomique humaine. Ann. Génét. (Paris), *1:* 41-49 (1959).

LENZ, W.: Über den Einfluss der Homogamie auf die Verteilungskurven der menschlichen. Acta Genet. (Basel), *3:* 97-100 (1952).

LERNER, I. M.: Genetic homeostasis (Wiley, New York 1954).

LEVIT, S. G.: The problem of dominance in man. J. Genet., *33:* 410-434 (1936).

LI, C. C.: Population genetics (Univ. Chicago Press 1955).

LUNDMAN, B. J.: Über die Körperhöhensteigerung in den nordischen Ländern nach dem Weltkriege. Z. Rassenk., *11:* 1-5 (1940).

MACMAHON, B. and MCKEOWN, T.: The incidence of hare lip and cleft palate related to birth rank and maternal age. Amer. J. Hum. Genet., *5:* 176-183 (1953).

— MCKEOWN, T. and RECORD, R. G.: The incidence and life expectation of children with congenital heart disease. Brit. Heart J., *15:* 121-129 (1953).

MAXWELL J.: Social implications of the 1947 mental survey (London 1953).

MCKEOWN, T.; MACMAHON, B. and PARSONS, C. G.: The familial incidence of congenital malformation of the heart. Brit. Heart J., *15:* 273-277 (1953).

— and RECORD, R. G.: Malformations in a population observed for five years after birth; In Ciba Found. Symp. on congenital malformations, pp. 2-16 (Little Brown, Boston 1960).

MERTON, B.: Taste sensitivity to PTC in 60 Norwegian families with 176 children. Confirmation of the hypothesis of single gene inheritance. Acta Genet. (Basel), *8:* 114-128 (1958).

MOHR, O.: Heredity and disease (Norton, New York 1934).

MORANT, G. M.: Secular changes in the heights of British people. Proc. Roy. Soc. B., *137:* 443-452 (1950).

MORTARA, G.: Essai de calcul du taux de reproduction pour la population de l'etat de São Paulo (Inter. Statist. Inst., 29th session, Rio de J. (1955).

MORTON, N. E.: Non-randomness in consanguineous marriages. Ann. Hum. Genet., *20:* 116-124 (1955).

NEEL, J. V.: A study of major congenital defects in Japanese infants. Amer. J. Hum. Genet., *10:* 398-445 (1958).

NEWMAN, H. H.; FREEMAN, F. N. and HOLZINGER, K. J.: Twins: A study of heredity and enviroment (Univ. Chicago Press 1937).

ÖSTER, J.: Mongolism. Opera. ex Domo Biol. Hered. Hum. Univ. Hafn. (Copenhagen), *32:* 1-206 (1953).

OTTENSOOSER, F.: Calculo do gráu de mistura racial através dos grupos sangüineos. Rev. Brasil. Biol., *4:* 531-537 (1944).

PENROSE, L. S.: The biology of mental defect (Sidgwick and Jackson 2nd. ed., London 1954); a) Parental age and mutation. Lancet *1:* 312-313 (1955); b) Evidence of heterosis in man. Proc. Roy. Soc. B., *144:* 203-213 (1955).

PRICE, B.: Primary biases in twin studies. A review of prenatal and natal differences producing factors in monozygotic pairs. Amer. J. Hum. Genet., *2:* 293-352 (1950).

POWELL, E. F. and WHITNEY, D. D.: Ear lobe inheritance. J. Hered., *28:* 184-186 (1937).

ROBERTS, D. F.: The dynamics of racial intermixture in the American Negro: some anthropological considerations. Amer. J. Hum. Genet. *7:* 361-367 (1955); A demographic study of a Dinka village. Human Biol., *28:* 323-349 (1956); Some genetic implications of Nilotic demography. Acta Genet (Basel), *6:* 446-452 (1956-1957).

SALDANHA, P. H.: Gene flow from white into Negro populations in Brazil. Amer. J. Hum. Genet., *9:* 299-309 (1957); Taste thresholds for phenylthiourea among Japanese. Ann. Hum. Genet., *22:* 380-384 (1958); Efeito da migração sôbre a estrutura genética de uma comunidade paulista (Doctoral thesis, Univ. São Paulo 1959); *Frequencies of consanguineous marriages in Northeast of São Paulo, Brazil.* Acta Genet. (Basel), *10:* 71-88 (1960).

— CAVALCANTI, M. A. A. and LEMOS, M. L.: Incidence of congenital anomalies in the population of the state of São Paulo, Brazil (Seminar on use of vital statistics and health for genetic and radiation studies, UN/WHO 1960).

— FROTA-PESSOA, O.; EVELETH, P.; OTTENSOOSER, F.; CUNHA, A. B. and CAVALCANTI, M. A. A.: Estudo génético e antropológico de uma colonia de holandeses do Brasil. Rev. Antrop., Sao Paulo, *8:* 1-42 (1960).

— and GUINSBURG, S.: Taste thresholds for phenylthiourea among students in Rio de Janeiro. Rev. Brasil. Biol., *14:* 285-290 (1954).

SHAPIRO, H. L. and HULSE, F.: Migration and environment (Oxford Univ. Press 1939).

SANDERS, J.: Inheritance of hare lip and cleft palate. Genetica, *15:* 433-510 (1934).

SANGHVI, L. D. and KHANOLKAR, V. R.: Data relating to seven genetical characters in six endogamous groups in Bombay. Ann. Eugen. (Lond.) *15:* 52-76 (1949).

SILVESTRONI, E. and BIANCO, I.: Richerche sulla la sensibilita gustativa dei microcitemici sani e dei soggetti normali alla feniltio-carbamide. Ric. Sci. no., *20:* 1856-1860 (1950).

SIRSAT, S. M.: Effect of migration on some genetical characters in six endogamous groups in India. Ann. Hum. Genet., *21:* 145-154 (1956).

SNYDER, L. H.: The inheritance of taste deficiency in man. Ohio J. Sci., *32:* 433-440 (1932); The principles of heredity (Heath 4th ed., Boston 1951).

SPIER, L.: Growth of Japanese children born in America and in Japan (Univ. Wash. Publ. Anthrop. no. 3).

SPUHLER, J. N.: Physical anthropology and demography; in the study of population, eds. Hauser and Ducan, pp. 728-758 (Univ. Chicago Press 1959).

SUTTER, J. and TRAN-NGOC-TOAN: The problem of the structure of isolates and of their evolution among human populations. Cold Spr. Harb. Symp. quant. Biol., *22:* 379-386 (1957).

SUZUKI, A.: Genetics studies on the human lappets. On the inheritance of the lobulus auriculae (in Japanese with an English summary). Jap. J. Genet., *25:* 157-159 (1950).

TANNER, J. M. and HEALEY, M. J. R.: The genetics of human morphological characters. Advancement of Sci, Lond., *51:* 192-194 (1956).

THOMPSON, G. N.: The trend of Scottish intelligence (London 1949).

UNITED NATIONS: Report of the United Nations Scientific Committee on the Effects of Atomic Radiation (General Assembly, 13th session, suppl. 17 (A/3838), New York 1958).

WAHLUND, S.: Zusammensetzung von Populationen und Korrelationserscheinungen von Standpunkt der Vererbungslehre aus betrachtet. Hereditas (Lund), *11:* 65-106 (1928).

WEIMER, B. R.: Congenital inheritance of inguinal hernia. J. Hered., *40:* 219-220 (1949).

WIENER, A. S.: Complications in ear genetics. J. Hered., *28:* 425-426 (1937).

WRIGHT, S.: Coefficients of inbreeding and relationship. Amer. Natur., *56:* 330-338 (1922); Evolution in Mendelian populations. Genetics., *16:* 97-159 (1931); Size of population and breeding structure in relation to evolution. Science, *87:* 430-431 (1938); Statistical genetics in relation to evolution. (Hermann, Paris 1939); Breeding structure of the population in relation to speciation. Amer. Natur., *74:* 232-248 (1940); Statistical genetics and evolution. Bull. Amer. Math. Soc., *48:* 223-248 (1942); Isolation by distance. Genetics, *28:* 114-138 (1943); Isolation by distance under diverse systems of matings. Genetics, *31:* 39-59 (1946); On the roles of directed and random changes in gene frequency in the genetics of populations. Evolution, *2:* 279-294 (1948); Adaptation and selection; in Genetics, Paleontology and Evolution, eds. Jepsen et al., pp. 365-389 (Princeton Univ. Press 1949); The genetical structure of populations. Ann. Eugen. (Lond.), *15:* 323-354 (1951).

RIASSUNTO *

È stata condotta un'indagine demografica e genetica in un distretto dello Stato di Sao Paulo (Brasile), che ha accolto un gran numero di emigranti italiani. Interessando tale indagine l'« esplosione » degli isolati, sono state prese in considerazione alcune teorie metodologiche ed è stato analizzato il fenomeno per mezzo del tasso di emigrazione e della frequenza dei matrimoni tra consanguinei in differenti periodi di tempo. Dalla consultazione dei documenti di matrimonio cattolico e dei registri tenuti presso il Distretto Militare sono stati ricavati dati sufficienti a facilitare gli studi sulle caratteristiche demografiche della comunità e sull'origine della popolazione. È stata analizzata la frequenza di alcune anormalità congenite nella presente popolazione ed è stata considerata la relativa efficacia dei fattori di evoluzione causante in essa variazioni genetiche.

I parametri relativi alla struttura della popolazione di tale distretto sono stati paragonati con quelli di altre comunità ed è stato messo in rilievo che le condizioni prevalenti del distretto in questione rappresentano una situazione relativamente comune in America, tanto che emigrazione e incrocio potrebbero essere un importante fattore di evoluzione nel presente periodo della storia biologica umana.

RÉSUMÉ

On a conduit une investigation démographique et génétique dans un département de l'Etat de Sao Paulo (Brésil), qui a reçu un gran nombre d'émigrants italiens. Puisque cette investigation concerne l'*éclat* des isolats, on a examiné quelques théories méthodologiques, en analysant le phenomène au moyen du taux d'immigration et de la fréquence des mariages entre consanguins dans les différentes périodes.

De la consultation des registres paroissiaux et de ceux du Département Militaire on a obtenu des données suffisantes à faciliter les études sur les caractères démographiques de la communauté et sur l'origine de la population. On a analysé la fréquence de quelques anomalies congénitales dans la population présente et on a considéré l'efficacité relative des facteurs d'évolution qui y auraient causé des variations génétiques.

Les paramètres concernant la structure de la population de ce département ont été rapportés à ceux d'autres communautés et on a relevé que les conditions trouvées dans ce département représentent une situation assez commune en Amérique, de sorte que migration et croisement pourraient représenter un facteur important d'évolution dans la période présente de l'histoire biologique humaine.

* Questo Riassunto e la corrispondente traduzione in francese sono opera della Redazione.

https://doi.org/10.1017/S1120962300020321 Published online by Cambridge University Press