

## GENETIC STRUCTURE OF THE DOMAHÁZA POPULATION 2. EFFECTIVE POPULATION, MIGRATION, DRIFT

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**Abstract:** A number of variables were examined in order to determine the level of isolation of a Hungarian community living in partial geographic isolation, and thereby to evaluate the random genetic drift potential in the population. The relatively small proportion of migrants (0.3165), the limited individual mobility, as given by marital distance (28.98 km), the small effective size ( $N_e = 142$ ; Kimura and Crow 1963), the effective migration rate ( $m_e = 0.1267$ ; Malécot 1948), and the value of the product  $N_e m_e$  (18) agree with the possibility of genetic drift in this population.

**Key words:** Migration; Individual mobility; Marital distance; Domaháza population (N-Hungary).

### Introduction

Small isolated communities are of great interest from both the theoretical and the practical standpoints. They represent an opportunity to investigate the potential factors of evolution and, particularly, to evaluate the genetic effects of random genetic drift.

The random drift in gene frequencies due to small population number and other causes has played a central role in the evolutionary theory advanced by Sewall Wright (1931, 1969), Kimura and Crow (1963).

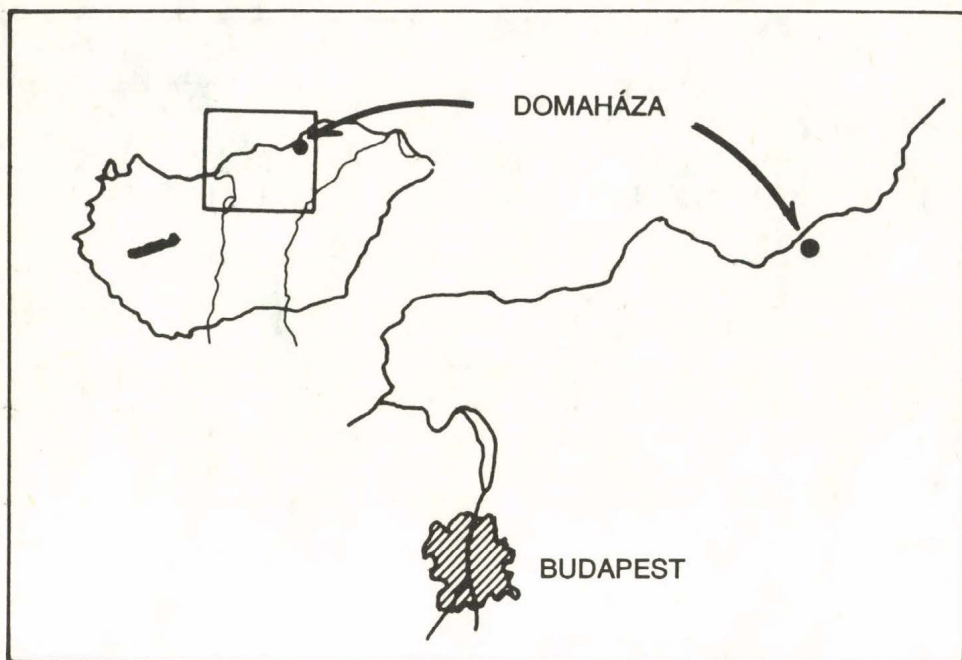
In order to appraise drift effects, many models have been suggested. The first of these was proposed by Wright in 1943, and named the *island model*. It assumes that a population is subdivided into small groups, between which genetic exchanges are low. Inasmuch as a human community fulfils this condition, the island model can be applied particularly in investigations of genetic drift (Magalhães and Arce Gomez (1987).

In the present study investigations were carried out in a partially isolated population in North Hungary. The characteristic features of marriages and the inbreeding tendencies have previously been reported (Pap and Holló 1988).

### The Population

The settlement under study (Domaháza) is situated in Borsod County in the north-western part of the hilly region north of the Bükk Mountains, near to the national frontier (Fig. 1). The settlement lies in the narrow valley of a stream and is connected with the other settlements only by a road running east, this is the only passable road for vehicles. On the other three sides the village is isolated from the other settlements by extensive woods.

The name of the village first appears in written documents in the 14th century. In 1332 it is mentioned in the list of papal tithes as a parish. According to the 1784 census the number of inhabitants was 540. The characteristic feature of the settlement is that the population is made up mainly by the descendants of three clans (Elek, Holló, Kisbenedek).



*Fig. 1: Localization of the population*

The clans, which in the past were assimilated into particular localities, can be traced back as far as 1500 in the documents. From data from the registers and censuses it was established that the growth of the population up till 1965 was 1.77-fold. At the time of our investigation (Holló 1965) the population numbered was 957 individuals, of whom 62% were members of the 3 clans.

### Population Surveys

The study of the whole population involved processing of the state and church registers containing the births, marriages and deaths between 1779 and 1965. Processed, in addition, were the documents in the archives, records of legal proceedings and conscription data. After collecting this information, data were obtained by interview from each family and individual, such as: full name, place and data of birth, place and date of marriage, reproductive history of the couples, names and birth data of parents, brothers and sisters. If the marriage was consanguineous, the pedigrees were constructed. Detailed genealogical analyses were performed concerning the whole population. The data collection was conducted and performed by one of the authors (Holló, L.) who is a member of one of the clans and is personally acquainted with the majority of the population.

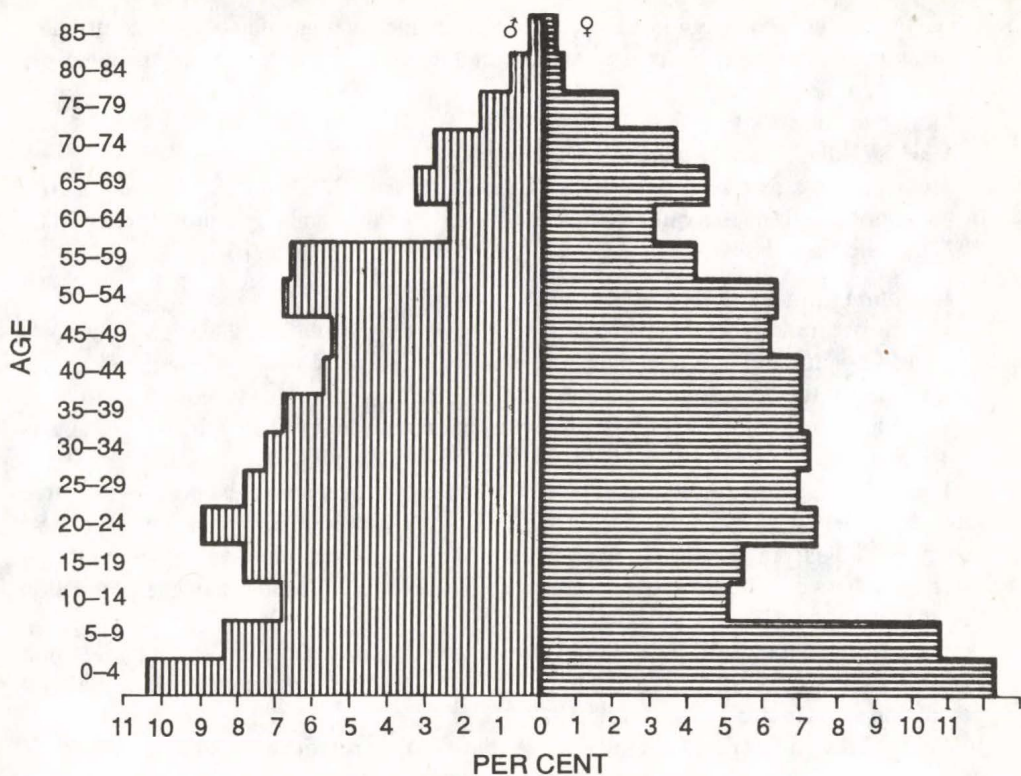


Fig. 2: Age and sex distribution in 1965 (Domaháza)

## Results and Discussion

### *Age and Sex Distribution*

In 1965 the population size of the settlement was 957 individuals. The number of males (469) and females (488) was approximately identical. The age pyramid shows a harmonic distribution (Fig. 2); one third of the population (34%) is younger than 20 years of age.

### *Effective Population*

The breeding population  $N_r$  is defined herein according to Glass et al. (1952) and to Freire-Maia et al. (1978) as the number of individuals with at least one living offspring aged 0-30 years, and was estimated as 297 individuals or 31.03% of the total population ( $N=957$ ). This estimation is not much lower than the value commonly found in human populations: 34% of  $N$  (cf. Freire-Maia 1974; Freire-Maia and Cavalli 1978). In a relatively isolated Hungarian population (Tiszaomgyorós) the value of  $N_r$  was 32.5% (Pap 1979).

The effective population size ( $N_e$ ) was estimated with Kimura and Crow's (1963) formula,

$$N_e = (2N_r - 2) / (xk - 1) + V_k/k$$

where  $N_r$  is the breeding population size,  $\bar{x}_k$  is the average number of contributed gametes per parent and  $V_k$  is the variance of the number of gametes contributed per individual in the present generation.

From the values  $\bar{x}_k = 3.78$  and

$V_k = 5.23$ , the effective population size  $N_e = 142$ .

The value of effective population is 14.83% of  $N$  (or 47.81% of  $N_r$ ). In the majority of human populations the value of  $N_e$  is about 25% of the total population (Freire-Maia 1977). In the Tiszamogyorós population  $N_e$  is 19.34% of  $N$  (Pap 1976).

#### *Migration Rate and Effective Migration*

The proportion of individuals born and living in the population, their contribution to the breeding and effective populations, then the number of immigrants and their proportion in the breeding population were determined. The fraction of the total population born, outside the village (total migration rate) is formed by the immigrants is 0.3164.

These two values (0.0982 and 0.3164) represent overestimates of the effective migration rates, that take into account the fact that migration tends to occur over short distances and is preferential as regards family composition, thus leading to a strong correlation between the gene frequencies among immigrants and those of the population that receives them (Wright 1931).

The effective migration rate should, therefore, be lower than the overall migration rate in the same way as the effective population size is generally smaller than the total population size (Freire-Maia, Cavalli 1978).

Among the migrants obviously only those that reproduce make any genetic contribution to the population that accepts them. From this fraction the effective migration rate ( $m_e$ ) was estimated, which is the fraction of migration of genetic importance.

Effective migration rate can be estimated through the use of Malécot's (1948) approximation

$$m_e = \sqrt{m(m+2k)},$$

where  $m$  is the long-range and  $k$  the short-range migration rate after  $m$  has been eliminated (Morton et. al. 1976).

Since it is impossible to discriminate 'short-range' and 'long-range' migration with accuracy (Freire-Maia, Cavalli-Sforza 1978).

The following procedure was adopted. Location of the birth places of the migrants on a map showed that they tended to cluster around a radius of 40 km from the village. Those coming from inside this area were designated as short-range migrants; and coming from more distant places as long-range ones those. The calculations were repeated taking 100 km as a conservative radius.

Table 1 presents the data and the corresponding estimates of  $m_e$ . The mean marital distance of the breeding population (i.e. the distance between the birth places of fertile mates) is 28.98 km. This number is compatible with considerable isolation of the Domaháza population.

Table 1. Some parameters related to migration  
(for an analysis of the product  $N_e m_e$ )

	Radius km	Short range	Long range	Total migration	Effectiv migration	$N_e m_e$
Breeding population	40 100	0.2895 0.3030	0.0269 0.0135	0.3165 0.3165	0.1276 0.0914	18 13

The majority of the breeding population is made up of the clans Elek, Holló and Kisbenedek.

The members of the clans were born of marriages within the clan (46.80% of  $N_p$ ) or between individuals with different surnames but of Domaháza birth (16.83% of  $N_p$ ).

The distance between individuals born and living at Domaháza was taken symbolically as 1, and the mean migration distance was calculated with this value.

The average migration distance of the breeding population (i.e., the distance between birth places and Domaháza village; defined by Morton 1969) equals 11.04. These values are in good agreement with the inbreeding tendencies and our findings related to consanguineous marriages (Pap and Holló 1988).

#### *Isolation and Genetic Drift*

The effective size ( $N_e = 142$ ) of the village population is less than half the breeding population ( $N_p = 297$ ). The value of the effective migration rate is about 13% of the breeding population. Therefore the village of Domaháza can be considered fairly isolated. This raises the question of the possibility of the potential effectiveness of drift as a cause of evolutionary change in this population (Magelhães and Arce-Gomez 1987). Wright (1931, 1969) has suggested a way of measuring the effects of genetic drift from the effective size and migration rates.

The product  $N_e m_e$  (known as the coefficient of breeding isolation) is crucial in the determination of the action of random genetic drift on a given population (Wright 1931, 1969).

According to Wright, the action of drift is not negligible in populations with an isolation index between 5 and 50, whereas it is fairly extensive in populations with an isolation index equal to or lower than 5.

The estimate of  $N_e$  obtained through the formula of Crow and Kimura (1970) is 142 and that of  $N_e m_e$  is 18. Thus, the contribution of random genetic drift to evolutionary change is not negligible in this population, as was expected.

The possible consequences of genetic drift were assessed on the basis of investigations on the clinically detectable abnormalities, diseases and genetic polymorphisms.

#### Summary

The village of Domaháza is relatively isolated geographically. The relatively small proportion of migrants (0.3165), the limited individual mobility as given by marital distance (28.98 km), the small effective size ( $N_e = 142$ ), the effective migration rate ( $m_e$

= 0.1276), and the coefficient of breeding isolation ( $N_e m_e = 18$ ) agree with the possibility of genetic drift in this population.

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