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SYMPOSIUM

Population genetic research in Hungary

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ABSTRACT In this short retrospective study, the author gives account of some aspects and the main results of human population genetical studies in Hungary. In the foreground of his interest is research taking as its basis the population as a genetic unit, which demonstrates the genetic structures of the populations, the geographic (and other) mechanisms of isolation, migration, the variability of genetic markers (blood group-, serum- and enzyme polymorphisms, deutan/protan anomaly), *i.e.,* which are related to the problems of human microevolutional processes.
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KEY WORDS

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The human biological research into the structure, genetic variability and functional relationship with the environment of the human population has provided a great deal of information for the estimation of the factors of microevolution and the causes of genetic variability, for the knowledge of the biological status of human populations. By doing so population-oriented human biological (biological anthropological) research has opened up new roads towards the revelation of the inheritance conditions of man and the continuously evolving interactions in the man versus environment relationship. Basic research has set out as its main objectives the exploration of the gene pool of human populations, the properties of multifactorial character variations, estimation of the advantageous or disadvantageous effects prevalent in the populations, the interpretation of microevolutional processes (Pap 1998).

Within the frame of this short review our aim is to present some genetic and biodemographic aspects of population structure. We wish to outline here the population-oriented approach in Hungarian population research, the theoretical and practical sources and methodological orientation forming the conceptual basis of the projects.

Population genetic approach, Mendelian populations

One of the essential results of the theory of evolution, and within this frame, of human evolution, following the work of Darwin and Mendel, was the elaboration of the concept of the gene-pool. The gene-pool approach, rather than placing the individual in the focus of evolution, concentrates on the population, and regards as the basic material of evolution the totality of the genes or alleles of the individuals concomitantly alive in the breeding population. The concept of gene pool is, anyhow, of human genetic, of anthropological origin, and an important role in its propagation was played by Dobzhansky (1955, 1985). The criterion of up-to-date human biology is the methodology approaching the structure of the biological, that is, breeding populations from a population-oriented aspect. Humanity as a biological species is, in a wide general sense, a Mendelian population. Evolving in it was a geographically or socially isolated hierarchy of the subordinated Mendelian (sub)populations. These subpopulations are not only of socio-economic, but also of biological character. This may be disturbing for some scholars of the social sciences, nevertheless it is totally independent of the scholars' endeavours to delimit their disciplines from the science of biology.

It is a fact that the delimitation of the Mendelian populations is mostly uncertain, since their gene pools, their total morphological patterns are not wholly differentiated. (Wright 1967-1977; Harrison and Boyce 1972). Therefore, it is important to emphasize that in the course of our human population studies (Pap 1978) the point of orientation was the smallest Mendelian population, the nuclear family. The parents and their children are genetically the closest relatives. Population structure can be delineated from these smallest genetic units (Fig. 1). Another source, valid only for human population research is the database of the registers. This system allows genealogical studies, family reconstruction, retrospectively as far back as several hundred years (in Hungary back to the 1750s). This is an important opportunity for the determination of the biological structure and status of the populations.

We think that the factors that determine the structure and dynamics of the given population with the greatest probability can be revealed at the population level. The spatial and temporal differences in the population are, essentially, results of the microevolutional events, *i.e.*, the genetic processes.

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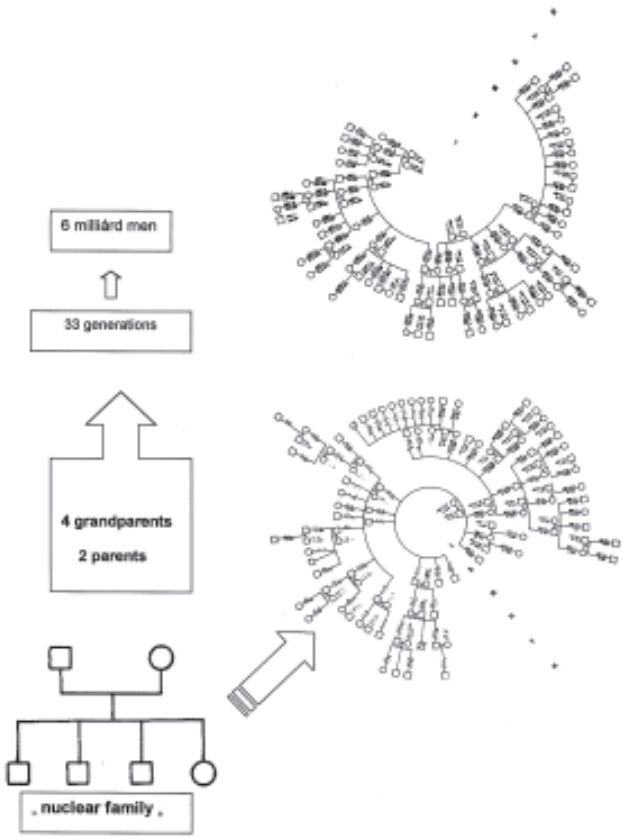

Figure 1. A "nuclear family", as the smallest Mendelian population.

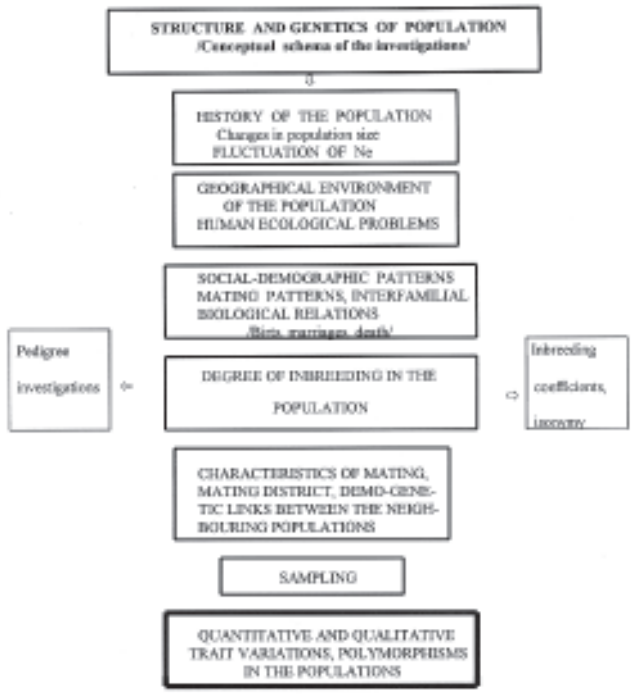

Figure 2. Conceptual scheme of the investigations (Pap 1979).

On concluding this introductory chapter we call attention to the recommendations of the Human Adaptability Section of the International Biological Project (IBP), according to which the human biological research of the populations cannot be considered complete without the presentation of the specific traits of the physical and biological environment affecting the local population (Harrison 1977).

Review of the studies

In Fig. 2 the conceptual outlines of our own research (Pap 1978, 1979) are presented. This concept follows, in general, the procedures, research aspects used in Hungary and abroad. It is important to underline the following: history of the population, fluctuations of the effective population size, geographical environment of the population (human ecological problems), the socio-demographic patterns, the interfamilies biological relations, inbreeding, degree of endogamy in the population, marriage patterns, mating area, vicinity. The investigations are not necessarily carried out in this order, however, in many cases (*e.g.* sampling) they took place along the demo-genetic analysis outlined (*e.g.* examination of blood groups, enzyme and serum groups, multifactorial characters). Thus, this is sampling based on a known population structure.

Demogenetic structure of the populations. Family reconstruction

The knowledge of the genetical-biological status of human populations makes the analysis of past biological events necessary. It is important to reveal the origin, continuity of the families in the population and the genetic relations among the individuals. In the Hungarian literature we can find good examples for the determination of the genetic status of the local populations of a population or a landscape unit. Investigations based on the databases of the registers will be mentioned below. The up-to-date population genetical approach is seen in the investigations in the village Tépe by Csörsz (1927). The study started in 1944 and continued by Nemeskéri and co-workers for decades is well-known in scientific circles in Hungary and abroad (Nemeskéri 1944, 1976).

The complex problems of demo-genetic structure can hardly be revealed without historical-demographic data, information confirming migration. These questions are given detailed answers in reports on investigations in the Bodrog-Tisza Interfluve (Nemeskéri and Walter 1966; Walter and Nemeskéri 1967), in the villages Benk and Tiszamogyorós (Pap 1970, 1979), in Turricse (Nemeskéri et al. 1973), in

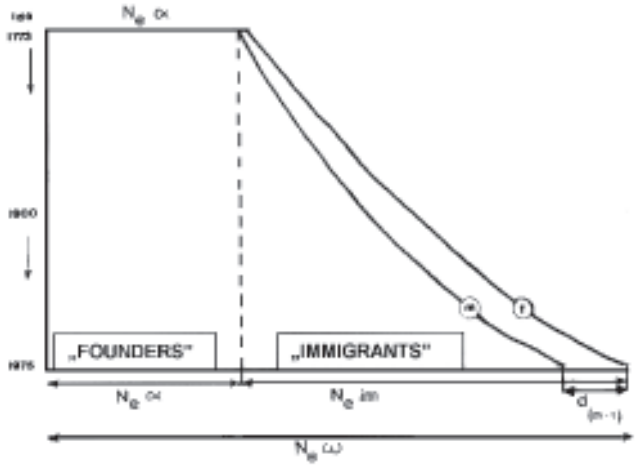

Figure 3. Estimation of the demo-genetic structure of the population (PSE model) (Pap 1978).

Figure 4. Cluster diagram of the genetic distances between the Tiszamogyorós and other poulations (PGM, AK, EsD, AP1 and AP2 enzymes) after Pap and Barta (1995).

Mezőkövesd (Martos et al. 1982) in Domaháza (Holló and Pap 1989/90) in Szamosangyalos (Csoknyai 1978). More recent are those performed in 17 populations in the Tiszahát microregion, the family reconstruction work based on registry data, the studies presenting the genetic structures (Koertvelyessy et al. 1987; Koertvelyessy et al. 1990, 1992).

Beyond the biological relationship of the local populations basically important are the changes in the intrapopulational sctructure. The genetic structure of a population can be analysed in different ways. Starting with the demographic parameters one can proceed towards the genetic markers, polymorphisms and arrive as far as the gene frequencies, upon which further analyses can be based.

Inbreeding, consanguinity, endogamy

Endogamy and consanguinity can basically influence the frequencies of the genetic markers in the population. The pedigrees including consanguineous marriages contribute important pieces of information to the detection of the genetic structure. Advantageous for such investigations are the small populations found in great numbers in Hungary. Studies on small (in cases endogamic) populations are important parts of population genetic research for they can serve as special genetic models in the measurement of interpopulational genetic similarity and (the complementary) genetic distance.

The intrapopulational inbreeding (F) and the mean inbreeding coefficient values were determined in the Ivád population (a= 0.00300) by Nemeskéri and Thoma (1961). In a detailed examination on a NE Hungarian population (Tiszamogyorós) Pap (1979) found a mean inbreeding coefficient of a = 0.002445, which demonstrates a high degree of endogamy.

On the grounds of the analysis of the genetic structure of the Tiszamogyorós population a "population-structure estimating model" was developed (Pap 1979) (Fig. 3), which can be used to clear up the role of inbreeding in a subpopulation, its effects on the gene pool. (Fig. 4, Table 1). The validity and practical significance of the model is confirmed by investigations at Benk (Pap 1970, 1979), at Domaháza (Holló and Pap 1989/90) and in Mezőkövesd (Martos et al. 1982).

Table 1. Projection of the Tiszamogyorós and other population groups on axes 1 and 2 of the principal component analysis (on the grounds of PGM, AK, EsD, AP1 and AP2 enzymes gene frequencies), after Pap and Barta (1995).

Population	PGM	AK	EsD	AP1	AP2	References
Tiszamogyorós	0.75	0.93	0.89	0.37	0.55	present study
Ivád-Full	0.85	0.97	0.87	0.26	0.69	[10]
Ivád-Half	0.83	0.98	0.88	0.31	0.65	[10]
Mezõkövesd	0.72	0.98	0.89	0.32	0.62	[3]
Nyíregyháza	0.76	0.99	0.90	0.40	0.58	[3]
Mátraderecske	0.71	0.98	0.88	0.41	0.54	[3]
Õriszentpéter	0.84	0.95	0.90	0.23	0.74	[3]
Egyházaskozár	0.76	0.95	0.89	0.37	0.59	[3]
Jászboldogháza	0.68	0.97	0.85	0.31	0.66	[3]
Kisújszállás	0.74	0.98	0.90	0.38	0.54	[3]

Genetic markers

As to research into the role of genetic markers the results of investigations on blood-group, enzyme and serum polymorphisms should be emphasized:

AB0, Rh, MNS, Pp Kk, by Kell, Duffy, Diego (a), Lewis, (a,b) investigations: Backhausz and Nemeskéri (1955, 1960), Joó-Szabados and Rackwitz (1968), Rex-Kiss and Horváth (1971), Balogh (1975), Pap (1979). Tauszik et al. (1980-1986), Tauszik and Simonovits (1980), Walter and Nemeskéri (1967, 1969, 1972), Czeizel et al. (1991), Walter (1997).

Out of the serum groups for the gene frequencies of Hp, Gc, Tf, AMY_2, PLG, Gm and Km markers data were published by Horváth and Simon (1963). Walter (1965), Walter and Nemeskéri (1967), Rex-Kiss and Szabó (1971), Hevér (1976), Pap (1979), Pap et al. (1978), Czeizel et al. (1991), Walter (1997).

Enzyme polymorphisms (SP, PGM_1 AK, ADA, GPT, EsD, GLO, 6-PGD, PGP, HbS, G-6-PD def.: Walter et. al. (1965), Pap (1979), Pap and Béres (1981), Szabó (1980), Czeizel et al. (1991).

Finally, from among the investigations on genetic markers it is important to mention the mapping of protan and deutan disturbances, the frequency of colour vision disorders: Eiben and Bakonyi (1971), Eiben and Kardos (1978), Pap (1979), Pap et al. (1983).

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