Coll. Antropol. **25** (2001) 1: 127–140
UDC 572:519.23-055.5/.7
Original scientific paper

Genealogical Structuring of a Population

M. Kujundžič Tiljak, J. Kern, D. Ivanković, H. Tiljak and S. Vuletić

»Andrija Štampar« School of Public Health, School of Medicine , University of Zagreb, Zagreb, Croatia

A B S T R A C T

The study observed population of 484 generation restrictive genealogies collected in four Croatian regions during 1970/71 within »The study of fat and carbohydrate metabolism indicators correlated with the occurrence of diabetes in animal fat and oil consuming population groups«. The genealogies were collected on two islands (Brač and Hvar) and in two continental regions (Sinjska Krajina and Srijem). Genealogy Structure Index (GSI) was defined as quantitative indicator of genealogy structure. GSI was continuos variable which, by its extreme, describes vertical and horizontal type of genealogy structure, independently of the way genealogies were collected. Genealogy structure of surveyed population was described on the basis of participation of different genealogy types. Populations of the island Hvar and Brač showed to have horizontal genealogy type, continental population of Sinjska Krajina showed to have mixed type and continental population of Srijem showed to have vertical type of genealogy structure.

Introduction

The *genealogies* or pedigrees could be considered as the basic source of information for any inherited characteristic in human population[1]. In medicine genealogies are mostly used for investigation of heredity, etiology discovering or risk assessment. Collecting and analysis of genealogies are especially used in genetic epidemiological researches. *Genetic epidemiology*, by its definition, studies etiology, distribution and control of diseases in a group of relatives, as well as it studies biological and cultural heredity in populations[2–5]. Siblings or a pair of twins can be considered as the minimal group of relatives suitable for genetic epidemiology research[6–9]. At the other side, there are some researches that include very large genealogies[10–12]. However, the aims of genetic epidemiology studies are often to explain the family resemblance[13–17].

One of the common methods used in genetic epidemiology is the analysis of familial aggregation. In these framework studies of population based genealogies are often performed[3,18–21]. These studies

Received for publication March 30, 2001.

are oriented to identification of genetic links between all, or at least the most, living individuals in one defined community. The study of population based genealogies include the ability to:

(1) identify the population of interest and all genealogical links between individuals in that population;

(2) identify health outcomes by direct observation or by record linkage with health registries;

(3) collect information on individuals regarding vital events and pertinent risk factors for the disease or trait to be studied[3,22–25].

Structure of genealogies in a population is very complex. Such a complexity has been implied by the nature of relationships between individuals and the size of genealogy itself[26,27]. Genealogical relationships between individuals can be both blood or non-blood types. Blood-type relationships may be vertical and horizontal. Parent-child relationship is vertical blood-type relationship versus siblings' relationship, which is horizontal blood-type relationship. Horizontal non-blood-type relationship is represented by marriage, while vertical non-blood-type relationship can be realized only through adoption. Higher or lower frequency of the same type relationships in certain genealogy is related to its homogeneity or heterogeneity according to characteristics of its members. Heterogeneity or homogeneity of genealogy is affected by the number of different blood-related groups of individuals, the number of different blood-related founders in the first generation of genealogy and the appearance of new blood-related groups of individuals coming to other genealogy generations. Attributes of any genealogy can be the total number of individuals in genealogy, the number of generations in genealogy (named the genealogy depth), and the maximum number of individuals in one generation (named the genealogy width).

Genealogy data can be derived from census and parish registers, but most frequently genealogical information has been obtained from people, members of the genealogy. Thus the genealogy data collection is very complicated and the results are genealogies that are highly distinctive regarding its structure and its magnitude.

As a result of genealogy structure analysis performed on the population of genealogies of maximum possible range (named non-restrictive genealogies), certain types of genealogies have been recognized. Mainly, they vary according to the participation of different types of relationships, i.e. horizontal and vertical relationships[27].

The aim of this study was to construct the model of analysis of the population according to genealogy structures. The model should define Genealogy Structure Index (GSI) which could specify types of genealogies and consequently describe the genealogical structure of a population.

Materials and Methods

Genealogies collected within the project »The Study of Fat and Carbohydrate Metabolism Indicators Correlated with the Occurrence of Diabetes in Animal Fat and Oil Consuming Population Groups« in 1970/1971 were used as a basis in the study[28]. There were 484 genealogies collected in four Croatian regions: two islands (99 genealogies in the Brač island and 105 genealogies in the Hvar island), and two continental regions (96 genealogies in the Sinjska Krajina region and 184 genealogies in the Srijem region). All collected genealogies were generation restrictive: they include only examinees aged 25 to 77 years.

The variables measured were as follows:

- number of genealogy members (individuals)
- genealogy depth, i.e. number of generations in the genealogy;
- maximum genealogy width, i.e. maximum number of individuals in any generation;
- number of different entries to the first genealogy generation, i.e. number of different families (blood-related groups) including first generation genealogy members;
- number of entries from the second genealogy generation onwards, i.e. number of different families (blood-related groups) including second and subsequent generation genealogy members;
- number of genealogy members with children;
- number of genealogy members with brothers and/or sisters;
- number of marriages in genealogy.
- A genealogy model with accompanying variable values is shown in Figure 1.

Data analysis consists of:

- factor analysis: confirmatory factor analysis with Procrustean rotation (target matrix is a result of factor analysis of maximum range genealogies collected in the Belec genealogy study[27];
- calculation of Genealogy Structure Index (GSI) for each genealogy;
- analysis of population genealogy structure in four Croatian regions according to the share of different genealogy types based on analysis of frequency distributions of Genealogy Structure Index (GSI). Chi –square test was used to test the differences in genealogy structure between studied regions.

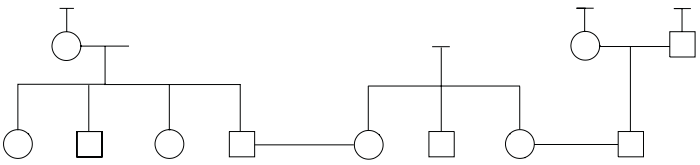
»The SAS System for Windows Release 6,12« was used for statistical analysis.

Results

Comparison of generation restrictive and non-restrictive genealogies

Confirmatory factor analysis with oblique Procrustean rotation was performed on the total population of 484 generation restrictive genealogies. Target matrix was the result of previously performed factor analysis on 132 non-restrictive genealogies from the Belec genealogy study[27].

Seven variables measuring characteristic genealogy dimensions were included in the factor analysis as described previ-



VARIABLE:	Value:	VARIABLE:	Value:
Number of individuals	11	Entries from 2^{-} generation	1
Depth	2	Individuals with children	3
Maximum width	8	Individuals with brothers/sisters	7
Entries in 1^{-} generation	3	Marriages	3

Fig. 1. Genealogy 'Srijem 4' – measured variables.

ously, with exclusion of number of genealogy members (individuals).

Genealogy size is measured by its depth and maximum width. The number of entries in the first generation and the number of entries from the second generation onwards measure heterogeneity of genealogy onwards. The number of individuals with children and the number of individuals with brothers and/or sisters measure blood-type relationships in genealogy. The number of marriages measures non-blood-type relationships in genealogy.

The number of derived latent dimensions was reduced to three of them. The system retained 99.20% of information. Communalities of all the seven variables were pretty high (> 0.9). Factor analysis results are presented in Tables 1–3.

TABLE 1
TARGET MATRIX

	Factor 1	Factor 2	Factor 3
Depth	0.02994	–0.01370	*0.97701*
Maximum width	*1.06263*	–0.03132	–0.12307
Entries in 1st generation	0.00208	*0.99861*	–0.00949
Entries from 2nd generation	*0.93704*	–0.06081	0.08127
Individuals with children	*0.90838*	0.04268	0.10669
Individuals with brothers/sisters	*1.00307*	0.01864	–0.03190
Marriages	*0.91576*	0.06263	0.09236

TABLE 2
PATTERN MATRIX

	Factor 1	Factor 2	Factor 3
Depth	0.02285	–0.01370	*0.99170*
Maximum width	*0.90912*	0.12606	–0.01291
Entries in 1st generation	0.08983	*0.94576*	–0.02868
Entries from 2nd generation	*1.14492*	–0.27576	0.04529
Individuals with children	*0.88784*	0.06554	0.11067
Individuals with brothers/sisters	*0.95030*	0.08204	–0.03174
Marriages	*0.91836*	0.09668	0.02746

TABLE 3
STRUCTURE MATRIX

	Factor 1	Factor 2	Factor 3
Depth	0.45123	0.32968	*0.99960*
Maximum width	*0.98929*	0.74070	0.42455
Entries in 1st generation	0.72107	*0.99764*	0.31621
Entries from 2nd generation	*0.97698*	0.51819	0.45577
Individuals with children	*0.98075*	0.70564	0.51928
Individuals with brothers/sisters	*0.99229*	0.71863	0.40946
Marriages	*0.99615*	0.73066	0.45945

Factor analysis results:

1st factor is predominantly defined by the maximum genealogy width, number of entries from the second genealogy generation onwards, number of individuals with children, number of individuals with brothers or sisters and number of marriages. These variables are good descriptors of genealogy magnitude, homogeneity and heterogeneity, i.e. genealogy type. So it can be named as *the genealogy type factor*.

2nd factor merely defined by the number of entries in the first genealogy generation can be named as *the initial genealogy heterogeneity factor*.

3rd factor merely defined by genealogy depth, i.e. number of genealogy generations, can be named as *the genealogy generations' magnitude factor*.

Variance explained by the 1st factor is 5.596 (49.56% of total variance), the variance explained by the 2nd factor is 3.469 (30.72% of total variance) and the variance explained by the 3rd factor is 2.136 (18.92% of total variance).

The genealogy structure index (GSI)

The Genealogy Structure Index (GSI) is quantitative variable, calculated as factor score of the genealogy type factor. Since confirmatory factor analysis has shown that non-restrictive genealogies and generation restrictive genealogies can be described by the same factors, the factor score coefficients obtained in factor analysis of non-restrictive genealogies were used for the calculation of the Genealogy Structure Index (GSI).

Factor score coefficients demand a standardized original variables. Therefore the standardization of original variables measured on 484 studied genealogies was achieved by using the mean and the standard deviation calculated from population of 132 non-restrictive genealogies.

Standardized variables are calculated as follows:

Standardized depth
= (Depth – 4.56818 / 1.24295)

Standardized maximum width
= (Maximum width 13.90909) / 12.96057

Standardized entries in 1st generation
= (Entries in 1st generation
2.21212) / 1.31944

Standardized entries form 2nd generation
= (Entries from 2nd generation
10.21970) / 11.48269

Standardized individuals with children
= (Individuals with children
20,78030) / 20,83901

Standardized individuals with brothers /sisters
= (Individuals with brothers/sisters
23.19697) / 25.08472

Standardized number of marriages
= (Number of marriages
11.53788) / 11.56555.

Figure 2 shows the distribution of Genealogy Structure Index (GSI) calculated for total population of 484 studied genealogies. This GSI distribution is bimodal. For 94 two-member, two-generation genealogies (19.42% of genealogies) GSI value is –4.4324 (= value of Q_1), and they have identical structure with parent (mother or father) and child (son or daughter). For 80 two-member, one-generation genealogies (16.53% of genealogies) GSI values is –3.2757 (= value of Q_3), and they have identical structure consisting of two parentless sibs (brother and sister, two brothers or two sisters).

Figure 3 shows genealogy structure for genealogies with modal values (equal to quartiles) of Genealogy Structure Index (GSI).

Genealogy Structure Index (GSI) calculation formula was as follows:

Genealogy Structure Index (GSI)

= (–1,33914 'Standardized depth') +
(0,67138 'Standardized maximum width') +
(–0,47563 'Standardized entries in 1st generation') +
(0,34421 'Standardized entries form 2nd generation') +
(0,24500 'Standardized individuals with children' +
(0,49284 'Standardized individuals with brothers/sisters') +
(0,25853 'Standardized number of marriages').

TABLE 4
GENEALOGY STRUCTURE INDEX (GSI) DISTRIBUTION – IN FOUR RESEARCH AREAS

	Brač	Hvar	Sinj	Srijem	Ukupno
N	99	105	96	184	484
$\bar{x}$	–3.3834	–3.5494	–3.6780	–3.9116	–3.6786
s	2.9841	3.6395	2.0322	1.8299	2.6042
Q_1	–4.3797	–4.4324	–4.4320	–4.4324	–4.4324
Median	–4.1401	–4.1360	–4.2342	–4.3413	–4.2699
Q_3	–3.2757	–3.2757	–3.2757	–3.8314	–3.2757
$Q_3 - Q_1$	1.1040	1.1567	1.1563	0.6011	1.1567
Mod	–3.2757	–3.2757	–4.4324	–4.4324	–4.4324
Min	–4.7146	–4.9834	–4.5083	–4.6074	–4.9834
Max	19.0322	32.9442	13.4420	18.5219	32.9442
Range	23.7468	37.9276	17.9503	23.1293	37.9276

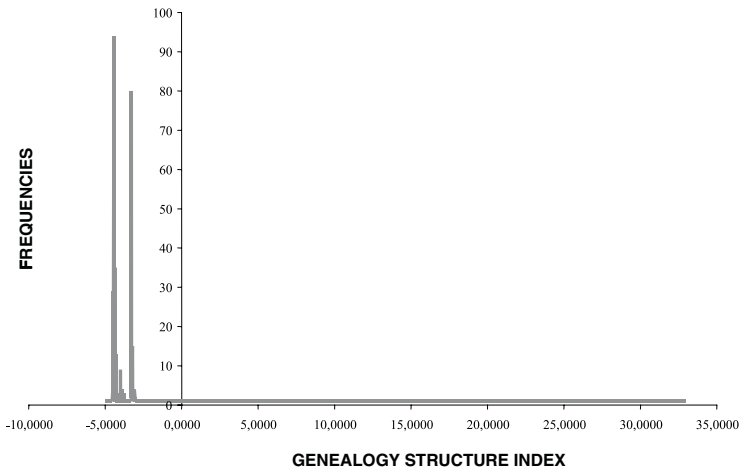


Fig. 2. Distribution of Genealogy Structure Index (GSI) total population of 484 genealogies.

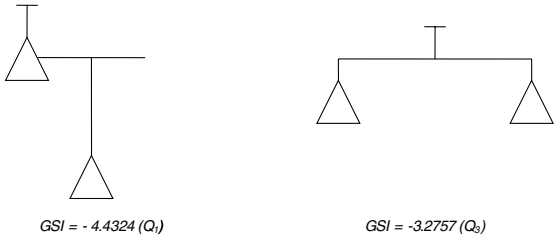


Fig. 3. Genealogy structure – modal values of Genealogy Structure Index (GSI).

Table 4 and Figures 4–7 show Genealogy Structure Index (GSI) distributions for each studied region – Brač, Hvar, Sinj and Srijem. These four distributions are also bimodal, as well as in total population of 484 studied genealogies.

Genealogical structuring of a population

Population's genealogy structure is based on the participation of different genealogy types.

Analysis of frequency distribution of Genealogy Structure Index (GSI) has shown that genealogies with prevalent vertical blood-type relationships, i.e. parent-child relationship, have low values of GSI. At the other side, the genealogies with prevalent horizontal blood-type relationships, i.e. sibs' relationships, have high values of GSI.

The Genealogy Structure Index (GSI) Q_1 value (25th centile value) for the studied 484 genealogies was –4.4324. This was also the first modal value of the Genealogy Structure Index (GSI). It reflects structure of two-member, two-generation genealogies consisting of parent (mother or father) and child (son or daughter), i.e. genealogies with only one vertical blood-type relationship. The genealogies with the Genealogy Structure Index (GSI) values lower than Q_1 value had more shares of vertical blood-type relationships. Thus the share of vertical blood-type relation-

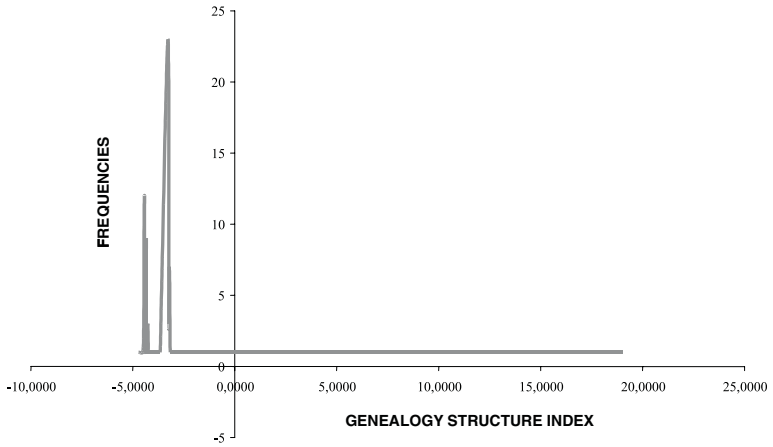


Fig. 4. Distribution of Genealogy Structure Index (GSI) 99 genealogies from Brač.

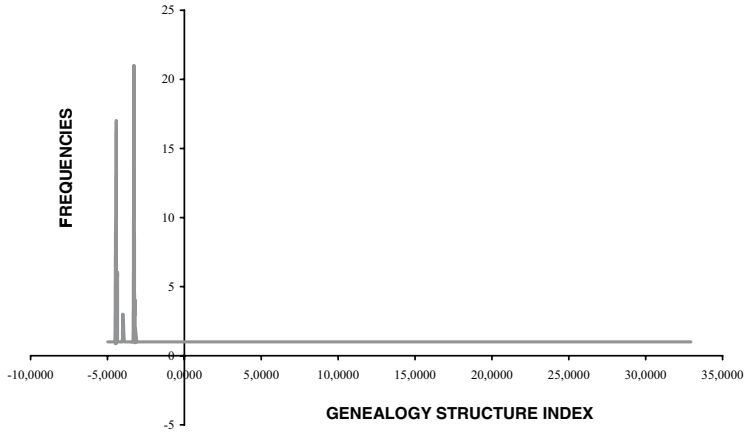


Fig. 5. Distribution of Genealogy Structure Index (GSI) 105 genealogies from Hvar.

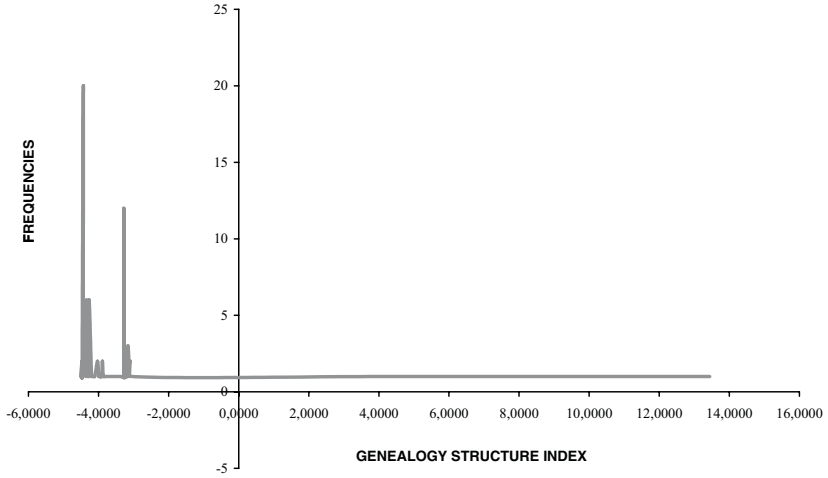


Fig. 6. Distribution of Genealogy Structure Index (GSI) 6 genealogies from Sinjska Krajina.

ships in a genealogy is as higher, as the Genealogy Structure Index (GSI) value is lower.

The Genealogy Structure Index (GSI) Q_3 value (75^{th} centile value) for the studied 484 genealogies was –3.2757. This was also the second modal value of the Genealogy Structure Index (GSI). It reflects structure of two-member, one-generation genealogies consisting of two parentless sibs (brother and sister, two brothers or two sisters), i.e. genealogies with only one horizontal blood-type relationship. The genealogies with the Genealogy Structure Index (GSI) values higher than Q_3 value had more shares of horizontal blood-type relationships. Thus the share of horizontal blood-type relationships in a genealogy is as higher, as the Genealogy Structure Index (GSI) value is higher.

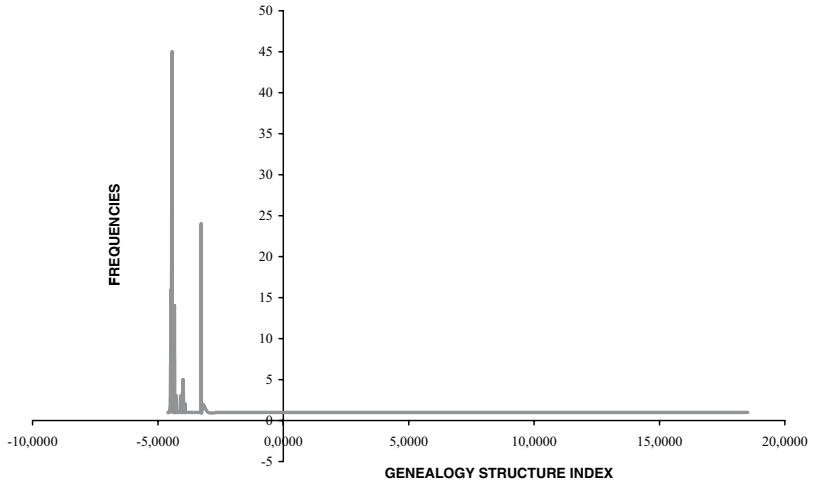


Fig. 7. Distribution of Genealogy Structure Index (GSI) 184 genealogies from Srijem.

The Genealogy Structure Index (GSI) median value for the 484 studied genealogies was –4.2699 and reflects mixed structure with absence of prevailing specific type of relationships, i.e. mostly the same share of vertical and horizontal blood-type relationships in a genealogy. As the Genealogy Structure Index (GSI) value is lower than median, the share of vertical blood-type relationships is as higher and the share of horizontal blood-type relationships is as lower. At the other side, as the Genealogy Structure Index (GSI) value is higher than median, the share of vertical blood-type relationships is as lower and the share of horizontal blood-type relationships is as higher.

To achieve the better description of the genealogy structure of the population (the population's genealogy structure) the genealogies collected in four studied areas were categorized in four groups according to the calculated values of the Genealogy Structure Index (GSI):

1st group: the Genealogy Structure Index (GSI) value below or equal to the Q_1 value;

2nd group: the Genealogy Structure Index (GSI) value above to the Q_1 value and below or equal to the median.

3rd group: the Genealogy Structure Index (GSI) value above to the median and below or equal to the Q_3 value.

4th group: the Genealogy Structure Index (GSI) value above to the Q_3 value.

The genealogies in the 1st group have the highest share of vertical blood-type relationships, while the genealogies in the 4th group have the highest share of horizontal blood-type relationships. The genealogies in the 2nd and the 3rd group have mixed structure, but with more share of vertical blood-type relationships in the 2nd group, and with more share of horizontal blood-type relationships in the 3rd group of genealogies. So the described groups of the studied genealogies can be named as follows:

1st group = vertical genealogies (V-genealogies), i.e. genealogies prevailing vertical component;

2nd group = vertical-horizontal genealogies (VH-genealogies), i.e. mixed geneal-

ogies with some prevailing vertical component;

3rd group = horizontal-vertical genealogies (HV-genealogies), i.e. mixed genealogies with some prevailing horizontal component;

4th group = horizontal genealogies (H-genealogies), i.e. genealogies prevailing horizontal component.

Among the genealogies collected in Brač, Hvar and Sinjska Krajina the highest share has the group of HV-genealogies (Table 5). In the genealogies collected in Srijem the same share have the group of V-genealogies and the group of HV-genealogies. Among the genealogies from Hvar, Sinjska Krajina and Srijem the lowest share has the group of H-genealogies, while the lowest share in the genealogies from Brač has the group of V-genealogies. Figure 8 shows the shares of different groups of genealogies among the genealogies from the different areas of study.

In the region of Brač the share of the V-genealogies was higher than the share of H-genealogies, while in other research areas (Hvar, Sinjska Krajina and Srijem) the share of H-genealogies was much higher than the share of V-genealogies. In all four-research areas the share of both mixed genealogies (VH-genealogies and HV-genealogies) was higher of share of both V-genealogies and H-genealogies. In all research area there were more HV-genealogies then VH-genealogies.

The total share of genealogies with prevalent horizontal component (H-genealogies and HV-genealogies) in the studied populations of Brač (53.5%) and Hvar (55.2%) was higher in comparison to the total share of genealogies with prevalent vertical component (V-genealogies and VH-genealogies). In Srijem there were more genealogies with prevalent vertical component (58.7%), while in Sinjska Krajina the share of genealogies with prevalent vertical component was equal to the share of genealogies with prevalent horizontal component (both 50.0%).

Frequency distribution of four groups of genealogies categorized according to the Genealogy Structure Index (GSI) is

TABLE 5
FREQUENCY DISTRIBUTION OF DIFFERENT GROUPS OF GENEALOGIES IN FOUR STUDIED AREAS

	Brač		Hvar		Sinj		Srijem		Total	
	f	%	f	%	f	%	f	%	f	%
V-genealogies (IGS Q_1)	16	16.2	30	28.6	24	25.0	67	36.4	137	28.3
VH-genealogies (Q_1 < IGS Q_2)	30	30.3	17	16.2	24	25.0	41	22.3	112	23.1
HV-genealogies (Q_2 < IGS Q_3)	35	35.3	42	40.0	31	32.3	67	36.4	175	36.2
H-genealogies (Q_3 < IGS)	18	18.2	16	15.2	17	17.7	9	4.9	60	12.4
Total	99	100.0	105	100.0	96	100.0	184	100.0	484	100.0

Among 484 studied genealogies the highest share of 36.2% (175 genealogies) have the HV-genealogies. 28.3% of studied genealogies (137 genealogies) were V-genealogies, 23.1% of genealogies (112 genealogies) were VH-genealogies, while only 12.4 % (60 genealogies) were H-genealogies.

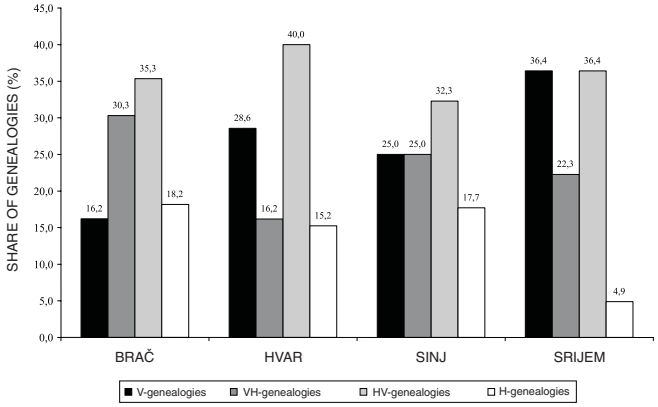


Fig. 8. The share of different types of genealogies in four studied areas.

significantly different between four studied population from Brač, Hvar, Sinjska Krajina and Srijem (^{2} = 29.142; df = 0; p = 0.001). H-genealogies from Srijem have shown the highest contribution (8,361) to the ^{2}, followed by V-genealogies from Brač (5.1582) and from Srijem (4.2726).

Discussion

The genealogy offers the information about its structure, as well as the information about biological and cultural inheritance. It has been recognized as significant analytical problem for a long time.

Since 1927 Weinberg has performed so called Weinberg proband method of segregation analysis for the estimation of proportion of children with certain illness characteristic. Afterwards, development of many statistical methods for the segregation analysis succeeded, i.e.:

- mathematical and statistical methods for the estimation of segregation evolved by Morton[3,4,5,29–31];
- methods for genealogy analysis developed by Thompson[1,10–12,32,33], including intraclass correlation, path-analysis and special procedures for the analysis of huge genealogies;
- methods of correlation and regression in estimation of family resemblance, including intraclass and interclass correlation and statistical methods for the estimation of the twins resemblance evolved by Donner[34–37];
- segregation analysis in sibships and segregation analysis in pedigrees described by Khouri[3,38].

All these methods attempt to analyze specific connections between individuals in genealogy. That means the analyze of specific structure of such o data for a correct risk assessment and the prevalence of pathological states or illness between sibs, especially twins, in nuclear families, in wide genealogies or in total populations.

It could be expected that genealogical relationships, i.e. genealogical structuring of a population, reflecting genetic or cultural similarity, could influence population characteristics like mortality, morbidity, etc. So it was worthy to find variable describing type of genealogies and consequently population where these genealogies are coming from.

The Genealogy Structure Index (GSI) has been defined in purpose of better and simpler description of different genealogy

structures. Since there was no difference in analysis of genealogy structure of generation restrictive and non-restrictive genealogies[27], the calculation of Genealogy Structure Index (GSI) has been based on population of non-restrictive genealogies.

The lowest values of Genealogy Structure Index (GSI) have had the genealogies with prevalent vertical component. At the other side, the genealogies with prevalent horizontal component have had the highest values of Genealogy Structure Index (GSI).

Genealogy Structure Index (GSI) centered genealogy analysis was tested on 484 genealogies collected during the four-region population study from 1970/71[28]. According to the calculated value of Genealogy Structure Index (GSI) for each genealogy, the studied 484 genealogies have been divided into four groups: V-genealogies, VH-genealogies, HV-genealogies and H-genealogies.

In four studied regions there were different participation of different genealogy types. The studied islands' populations in Brač and Hvar had horizontal type of genealogy structure: prevailing H-genealogies and HV-genealogies with higher values of GSI. The continental population in Srijem had vertical type of genealogy structure: prevailing V-genealogies and VH-genealogies with lower values of GSI. The continental population in Sinjska Krajina had mixed type of genealogy structure: 50% of genealogies with prevalent vertical component (V-genealogies and VH-genealogies) and 50% of genealogies with prevalent horizontal component (H-genealogies and HV-genealogies).

The islands' population are commonly much more isolated and closed in comparison to the continental populations. The results of previous studies performed on the different Croatian islands, including Brač and Hvar, have shown that the studied populations are quite closed and isolated[39–48]. In more detailed analysis, the genealogies collected in islands of Brač and Hvar, had on average more number of sibs, more number of entries from second generation afterwards and more number of marriages. These characteristics are usually related to the horizontal type of genealogy structure. In some very huge genealogies collected in islands (for example genealogy of 437 individuals from Hvar) the marriages between the same families were found, i.e. brother or sister from one family was married to the brother or sister from another family. Certain isolation of studied islands' populations could be the reason for such phenomenon. However, these huge genealogies were still with prevalent horizontal component with great number of sibs and marriages in four generations. On the other hand, in the continental population from the Srijem region there were traditionally fewer children in families, mostly only one. For this reason the prevalent vertical component has been expected and confirmed in the studied genealogies.

Conclusions

1. One number – the Genealogy Structure Index (GSI), could express genealogy structure.
2. The Genealogy Structure Index is a continuous variable the extremes of which are vertical (lower GSI values) and horizontal type (higher GSI values) of genealogy structure.
3. Irrespectively of the method of genealogy collection, the Genealogy Structure Index (GSI) acts similar in the separation of vertical and horizontal genealogies.
4. The shares of different genealogy types describe the population genealogy structure.

5. The studied continental population had more genealogies with vertical components, while islands' population had more genealogies with horizontal components.

Acknowledgement

We are grateful to Professor Pavao Rudan for great support and encouragement.

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M. Kujundžić Tiljak

»Andrija Štampar« School of Public Health, School of Medicine, University of Zagreb, Rockefellerova 4, 10000 Zagreb, Croatia

GENEALOŠKA STRUKTURIRANOST POPULACIJE

S A Ž E T A K

U radu je ispitivana populacija od 484 generacijski ograničene genealogije prikupljena u četiri područja u Republici Hrvatskoj tijekom rada na projektu »Istraživanja indikatora metabolizma masti i ugljikohidrata vezanih uz pojavu dijabetesa u populacijama koje se hrane mastima životinjskog podrijetla i koja se hrane uljima«, provedenog 1970/71. godine. Genealogije su prikupljene na dva otoka (Brač i Hvar) te u dva kontinentalna područja (Sinjska Krajina i Srijem). Definiran je *indeks genealoške strukture (IGS)* kao kvantitativni pokazatelj genealoške strukture. IGS kontinuirana je varijabla koja u svojim ekstremima opisuje vertikalni i horizontalni tip genealoške strukture, i to neovisno o načinu prikupljanja genealogija. Na temelju udjela skupina genealogija različitih tipova genealoške strukture opisana je genealoška strukturiranost ispitivanih populacija. Otočke populacije Hvara i Brača imale su horizontalnu, kontinentalna sinjska populacija mješovitu, a kontinentalna srijemska populacija vertikalnu genealošku strukturiranost.