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ISOMORPHISMS OF MENDELIAN POPULATIONS

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INTRODUCTION

Genotypes can be characterized either by the probability laws of their inheriting from parents to offspring, or by the way in which they influence the phenotype.

The probability laws of heredity can be formally described with the help of the notion of population. For further details of basic mathematical concepts of population genetics see [17], from where, with a small change, the notion of population has been taken.

Formal description of the nature of the genotype's influence on the phenotype has not yet been achieved in a greater detail. The most elaborate conception we know about is [2]. This paper introduces the notion of phenotype system, i.e., a function from a set of genotypes into a set of phenotypes, and the notion of phenogram, i.e., a class of permutationally equivalent phenotype systems.

We have tried to define the terms phenotype system and phenogram with regard to the fact that there is a structure of a population on the set of the genotypes. From this point of view it is useful to investigate the characteristics of the group of isomorphisms of this population. Our paper aims at proving the proposition appearing in [2], namely that the isomorphisms of the mendelian population correspond in a one – one manner to the permutations of the set of loci and the sets of the alleles of every locus. This can help in finding the cyclic index of the group of the isomorphisms of this population.

Standard set-theoretical notation is used throughout the paper. Index of symbols can be found in Appendix.

1. Definition. *A population is a couple* $\mathbf{Z} = \langle Z, P \rangle$ *such that* Z *is a finite non-empty set and* P *is a function from* Z^3 *into the closed interval of real numbers* $\langle 0, 1 \rangle$, *such that for every* $\iota, \varkappa \in Z$ *holds* $\sum_{\lambda \in Z} P(\iota, \varkappa, \lambda) = 1$. *The elements of* Z *are called the genotypes of* $\mathbf{Z}$.

2. Definition. *Let $\boldsymbol{Z}_i = \langle Z_i, P_i \rangle$, $i = 1, 2$ be two populations. A one–one function φ from Z_1 onto Z_2, such that*

$$\iota, \varkappa, \lambda \in Z \rightarrow P_1(\iota, \varkappa, \lambda) = P_2(\varphi(\iota), \varphi(\varkappa), \varphi(\lambda))$$

is called an isomorphism between $\boldsymbol{Z}_1$ and $\boldsymbol{Z}_2$. The set of all isomorphisms between $\boldsymbol{Z}_1$ and $\boldsymbol{Z}_2$ is denoted by $Is(\boldsymbol{Z}_1, \boldsymbol{Z}_2)$.

Let us note, that for every population $\boldsymbol{Z}$, $Is(\boldsymbol{Z}, \boldsymbol{Z})$ with the operation composition of functions is a group.

3. Definition. *A phenotype system is a triple $H = \langle \boldsymbol{Z}, f, F \rangle$ such that $\boldsymbol{Z} = \langle Z, P \rangle$ is a population, f is a function from Z into a finite non-empty set F. The elements of F are called phenotypes.*

4. Definition. *We say that two phenotype systems $H_i = \langle Z_i, f_i, F_i \rangle$, $i = 1, 2$ represent the same phenogram, if there exist φ from $Is(\boldsymbol{Z}_1, \boldsymbol{Z}_2)$ and a one–one mapping τ from F_1 onto F_2 such that the following diagram is commutative:*

$$\begin{array}{ccc} Z_1 & \xrightarrow{\varphi} & Z_2 \\ {\scriptstyle f_1}\downarrow & & \downarrow{\scriptstyle f_2} \\ F_1 & \xrightarrow{\tau} & F_2 \end{array}$$

The relation "to represent the same phenogram" is an equivalence relation on the class of all phenotype systems. The equivalence classes defined by this relation will be called the phenograms. We say, that a phenotype system H represents a phenogram $\boldsymbol{H}$ if $H \in \boldsymbol{H}$.

The following problem is considered in [2]. Given a population $\boldsymbol{Z}$ and a set F of phenotypes, which are the phenograms of the form $\langle \boldsymbol{Z}, f, F \rangle$? This is the reason why we deal with the characteristics of the group $Is(\boldsymbol{Z}, \boldsymbol{Z})$.

The number of these phenograms can be obtained with the help of the Bruijn's formula

$$N = P_D\left(\frac{\partial}{\partial x_i}, \quad i = 1, 2, \ldots, |\boldsymbol{D}|\right) P_R[\exp(j \sum_{i=1} x_{ji}), \quad j = 1, 2, \ldots, |\boldsymbol{R}|]/x_k = 0$$
$$\text{for all } k$$

(see [2] and [3]), expressing the total number N of patterns of mappings of a set $\boldsymbol{D}$ into a set $\boldsymbol{R}$, provided that D, R are groups if permutations of $\boldsymbol{D}$, $\boldsymbol{R}$, with cyclic indices PD and PR respectively. We use the special case $\boldsymbol{D} = Z$, $\boldsymbol{R} = F$, $D = Is(\boldsymbol{Z}, \boldsymbol{Z})$ and R is the group of all permutations of F.

5. Example. Diallelic mendelian population.

Here $Z = \{aa, ab, bb\}$ and P is defined by the table where the value in the row ι and column $\varkappa$ represents $\sum_{\lambda \in Z} P(\iota, \varkappa, \lambda)\, \lambda$. For diallelic mendelian population and the set $F = \{0, 1, 2\}$ of phenotypes, there are just four phenograms. These phenograms can be represented (for instance) by the following four functions:

aa	ab	bb	
0	0	0	1. the trivial case
0	0	1	2. dominance-recesivity
0	1	0	3.
0	1	2	4. intermediare heredity

$\iota \backslash \varkappa$	aa	ab	bb
aa	aa	$\frac{1}{2}aa + \frac{1}{2}ab$	ab
ab	$\frac{1}{2}aa + \frac{1}{2}ab$	$\frac{1}{4}aa + \frac{1}{2}ab + \frac{1}{4}bb$	$\frac{1}{2}ab + \frac{1}{2}bb$
bb	ab	$\frac{1}{2}ab + \frac{1}{2}bb$	bb

6. Example. Let A be a finite non-empty set. The population $\langle [A]^2, q \rangle$, where q is defined for any $\iota, \varkappa, \lambda \in [A]^2$, and $\lambda = \{a, b\}$ by the formula

$$q(\iota, \varkappa, \lambda) = \frac{Card(\lambda) \,.\, [Card(\iota \cap \{a\})\, Card(\varkappa \cap \{b\}) + Card(\iota \cap \{b\})\, \mathrm{Card}(\varkappa \cap \{a\})]}{2Card(\varkappa)\, Card(\lambda)}$$

is called mendelian population of one locus with the set A of alleles.

Let us note that for $Card(A) = 2$ this population is isomorphic to the diallelic mendelian population. If ϱ is a permutation of A, then $q(\iota, \varkappa, \lambda) = q(\varrho''\iota, \varrho''\varkappa, \varrho''\lambda)$ holds for every $\iota, \varkappa, \lambda$ from $[A]^2$. It is easy to see that the properties of mendelian population of one locus with the set A of alleles depend on the cardinality of A only.

7. Definition. *Any function from a positive integer to the set of all positive integers is called dimension. We say, that $\langle Z, P \rangle$ is a mendelian population of dimension d, if the following condition is satisfied:*

$$Z = \{\iota;\ Fuc(\iota),\ D(\iota) = D(d),\ i \in D(\iota) \rightarrow \iota(i) \in [d(i)]^2\}$$

and P is defined for any $\iota, \varkappa, \lambda \in Z$ by the formula

$$P(\iota, \varkappa, \lambda) = \prod_{i \in D} q(\iota(i), \varkappa(i), \lambda(i))\,,$$

where q is defined in the same way as above. The elements of $D(d)$ are called the loci and the elements of $d(i)$ are called the alleles of the i-th locus. The genotypes of this population are functions over the set of the loci associating every locus with a couple of its alleles.

A genotype ι from Z is said to be homozygote or heterozygote in the i-th locus, if $Card(\iota(i)) = 1$ or $Card(\iota(i)) = 2$, respectively. We shall denote $Hom(\iota) = \{i \in D(d); Card(\iota(i)) = 1\}$ and $Het(\iota) = \{i \in D(d); Card(\iota(i)) = 2\}$.

The proposed representation of genotypes is based on the idea of numbering the loci and the alleles of every locus, and the expressing P by q. It may seem a little obscure. The more usual and more graphic way is to represent the genotypes by the words of the form $a_1 b_1 * \ldots * a_k b_k$, where $1, 2, \ldots, k$ are the loci, a_i, b_i are alleles of the i-th locus, the words $a_1 b_1 * \ldots * a_k b_k$, $a'_1 b'_1 * \ldots * a'_k b'_k$ such that $\{a_i, b_i\} = \{a'_i, b'_i\}$ being identified.

8. Lemma. *Let d be a dimension and π a permutation of $D(d)$. Mendelian population of dimension d is isomorphic to mendelian population of dimension $d \circ \pi$.*

9. Lemma. *Let d_1 and d_2 be two dimensions, $d_1 \subseteq d_2$ and $W(d_2 \setminus d_1) = 1$. Then m.p. of dimension d_1 is isomorphic to m.p. of dimension d_2.*

It is obvious that in studying the isomorphisms we can confine ourselves to dimensions d such that $d(i) \geqq 2$ for any $i \in D(d)$.

10. Proposition. *Let $\boldsymbol{Z}$ be mendelian population of dimension d such that $d(i) \geqq 2$ for any $i \in D(d)$. Then there exists a one-one correspondence between $Is(\boldsymbol{Z}, \boldsymbol{Z})$ and the set S of all ordered pairs $\langle \pi, \varrho \rangle$ satisfying the following conditions*

π is a permutation of $D(d)$, $d \circ \pi = d$,

ϱ is a function such that the value $\varrho(i)$ is a permutation of $d(i)$ for each $i \in D(\varrho) = D(d)$

Before proving Proposition 10 we state and prove the following lemma.

11. Lemma. *Let $\boldsymbol{Z} = \langle Z, P \rangle$ be mendelian population of a dimension d, $\iota, \varkappa \in Z$, $\varphi \in Is(\boldsymbol{Z}, \boldsymbol{Z})$. Then*

1. *$Hom(\iota) \cap Het(\iota) = 0$ and $Hom(\iota) \cup Het(\iota) = D(d)$.*
2. *$Card(Hom(\iota)) = Card(Hom(\varphi(\iota)))$.*
3. *For every $\lambda \in Z$ it holds $P(\iota, \varkappa, \lambda) \leqq (\frac{1}{2})\, Card(Het(\iota) \cup Het(\varkappa))$.*
4. *If $\iota/Het(\iota) \subseteq \varkappa$ then there exists a genotype $\lambda \in Z$ such that $P(\iota, \varkappa, \lambda) = (\frac{1}{2})\, Card(Het(\varkappa))$.*
5. *If $\iota/Hom(\iota) \subseteq \varkappa$ then $\varphi(\iota)/Hom(\varphi(\iota)) \subseteq \varphi(\varkappa)$ and $Het(\varphi(\varkappa)) \subseteq Het(\varphi(\iota))$.*
6. *If $Het(\iota) \subseteq Het(\varkappa)$ then $Het(\varphi(\iota)) \subseteq Het(\varphi(\varkappa))$.*
7. *If $Hom(\iota) = Hom(\varkappa)$ then $Hom(\varphi(\iota)) = Hom(\varphi(\varkappa))$.*
8. *If $\iota/Hom(\iota) = \varkappa/Hom(\varkappa)$ then $\varphi(\iota)/Hom(\varphi(\iota)) = \varphi(\varkappa)/Hom(\varphi(\varkappa))$.*

Proof of Lemma 11.

1. follows immediately from the definition.

2. is a consequence of 1. and the equality

$$(\tfrac{1}{2})\, Card(Het(\iota)) = P(\iota, \iota, \iota) = P(\varphi(\iota), \varphi(\iota), \varphi(\iota)) = (\tfrac{1}{2})\, Card(Het(\varphi(\iota))).$$

3. follows from the definition of P.

4. Let us define λ by the formula $\lambda(i) = \iota(i) \cup \varkappa(i)$ for each $i \in D(d)$.

5. If $\iota/Hom(\iota) \subseteqq \varkappa$ then for every λ such that $P(\iota, \varkappa, \lambda) > 0$ it holds $Card(Het(\lambda)) \leqq \leqq Card(Het(\iota))$. If the condition $\varphi(\iota)/Hom(\varphi(\iota)) \subseteqq \varphi(\varkappa)$ is not satisfied, than there exists $\mu \in Z$ such that $Het(\varphi(\iota)) \subsetneqq \subsetneqq Het(\mu)$ and $P(\varphi\iota(\iota), \varphi(\varkappa), \mu) > 0$. Using 2. for $\lambda = \varphi^{-1}(\mu)$ we obtain $P(\iota, \varkappa, \lambda) > 0$ and $Card(Het(\lambda)) = Card(Het(\mu))$. Hence $\varphi(\iota)/Hom(\varphi(\iota)) \subseteqq \varphi(\varkappa)$ and consequently $Het(\varphi(\varkappa)) \subseteqq Het(\varphi(\iota))$.

6. a) We first prove 6. for ι, $\varkappa$ such that $\iota/Het(\iota) \subseteqq \varkappa$. 4. asserts that there exists λ with the property $P(\iota, \varkappa, \lambda) = (\frac{1}{2})\, Card(Het(\varkappa))$. By 3. it is $P(\iota, \varkappa, \lambda) = P(\varphi(\iota), \varphi(\varkappa), \varphi(\lambda)) \leqq (\frac{1}{2})\, Card(Het(\varphi(\iota)) \cup Het(\varphi(\varkappa)))$. Hence $Card(Het(\varphi(\iota)) \cup Het(\varphi(\varkappa))) \leqq \leqq Card(Het(\varkappa)) = Card(Het(\varphi(\varkappa)))$, that is, $Het(\varphi(\iota)) \subseteqq Het(\varphi(\varkappa))$.

b) Now we prove 6. for every $\iota, \varkappa \in Z$. Let us denote $\lambda = \iota/Het(\iota) \cup \varkappa/Hom(\iota)$. By a) $Het(\varphi(\iota)) \subseteqq Het(\varphi(\lambda))$ and by 5. $Het(\varphi(\lambda)) \subseteqq Het(\varphi(\varkappa))$. 6. is proved.

7. and 8. are consequences of 5. and 6.

Proof of Proposition 10. We shall prove that there exist a one-one function g from S into $Is(\boldsymbol{Z}, \boldsymbol{Z})$ and a function h from $Is(\boldsymbol{Z}, \boldsymbol{Z})$ into S such that $g \circ h$ is the identity on $Is(\boldsymbol{Z}, \boldsymbol{Z})$.

Let us denote $H_i = \{\iota \in Z;\ Hom(\iota) = \{i\}\}$ and $H_{i,a} = \{\iota \in H_i;\ \iota(i) = \{a\}\}$ for any $i \in D(d)$, $a \in d(i)$.

Let us define the function g by the equation $g(\pi, \varrho)(\iota)(i) = \varrho(\pi^{-1}(i))''\, \iota(\pi^{-1}(i))$ for $\langle \pi, \varrho \rangle \in S$, $\iota \in Z$, $i \in D(d)$. It follows from the properties of the function q that $g(\pi, \varrho) \in Is(\boldsymbol{Z}, \boldsymbol{Z})$. It holds $\iota \in H_{i,a}$ if and only if $g(\pi, \varrho)(\iota) \in H_{\pi(i),\varrho(i)(a)}$. Hence if $\langle \pi, \varrho \rangle \neq \langle \pi', \varrho' \rangle$ then there exist $i \in D(d)$ and $a \in d(i)$ such that $\langle \pi(i), \varrho(i)(a) \rangle \neq \neq \langle \pi'(i), \varrho'(i)(a) \rangle$. If $\iota \in H_{i,a}$ then $g(\pi, \varrho)(\iota) \in H_{\pi(i),\varrho(i)(a)}$ and $g(\pi', \varrho')(\iota) \in H_{\pi'(i),\varrho'(i)(a)}$ Hence g is one-one.

Let us denote $h_1(\varphi)(j) = \bigcup Hom(\varphi(\varkappa))$ for $\varphi \in Is(\boldsymbol{Z}, \boldsymbol{Z})$, $j \in D(d)$, $\varkappa \in H_j$ and $h_2(\varphi)(j)(a) = \bigcup W(\varphi(\varkappa)\, Hom(\varphi(\varkappa)))$ for $\varkappa \in H_{j,a}$. It follows from 11.7 and 11.8 that $h_1(\varphi)$ and $h_2(\varphi)(j)$ are defined correctly, independently of the choice of $\varkappa$. $h_1(\varphi)$ is a permutation of $D(d)$. $h_2(\varphi)(j)$ is a function from $d(j)$ onto $d(h_1(\varphi)(j))$, hence $d(j) = d(h_1(\varphi)(j))$, that is $d \circ h_1(\varphi) = d$. It holds $\iota \in H_{i,a}$ if and only if $\varphi(\iota) \in H_{h_1(\varphi)(i),h_2(\varphi)(i)(a)}$. It can be proved that $(h_1(\varphi))^{-1} = h_1(\varphi^{-1})$ and $(h_2(\varphi)(i))^{-1} = = h_2(\varphi^{-1})(h_1(\varphi)(i))$.

Let us define the function h by the equation $h(\varphi) = \langle h_1(\varphi), h_2(\varphi) \rangle$. It is sufficient to prove $g(h(\varphi)) = \varphi$, that is $(h_2(\varphi)((h_1(\varphi))^{-1}(j)))''\, \iota(h_1(\varphi^{-1})(j)) = \varphi(\iota)(j)$ for every $\iota \in Z$ and $j \in D(d)$. From above, it is sufficient to prove $(h_2(\varphi^{-1})(j))^{-1''} \cdot \iota(h_1(\varphi^{-1})(j)) = \varphi(\iota)(j)$. Let $\varphi(\iota)(j) = \{c, d\}$. Then there exist $\gamma \in H_{j,c}$ and $\delta \in H_{j,d}$ such that $P(\gamma, \delta, \varphi(\iota)) = 0$, since $P(\varphi^{-1}(\gamma), \varphi^{-1}(\delta), \iota) > 0$ and $\varphi^{-1}(\gamma) \in H_{h_1(\varphi^{-1})(j), h_2(\varphi^{-1})(j)(c)}$ and $\varphi^{-1}(\delta) \in H_{h_1(\varphi^{-1})(j), h_2(\varphi^{-1})(j)(d)}$. Hence $\iota(h_1(\varphi^{-1})(j)) = \{h_2(\varphi^{-1})(j)(c), h_2(\varphi^{-1})(j)(d)\}$, that is, $\{c, d\} = (h_2(\varphi^{-1})(j))^{-1''} \iota(h_2(\varphi^{-1})(j))$. Proposition 10 is proved.

APPENDIX

We use the following notions and notation of the set theory. Let x be a set and f a function. Then

$\bigcup x$ denotes the union of x.
$Card(x)$ denotes the number of elements of x.
$[x]^2$ denotes the set $\{y \subseteqq x;\ 1 \leqq Card(y) \leqq 2$.
$Fuc(f)$ means that f is a function.
$D(f)$ denoted the domain of f.
f/x denoted the restriction of f to x.
$W(f)$ denoted the range of f.
$f''x$ denoted the range of the restriction of f to x, that is, the set $W(f/x)$.

We identify, as usual, the positive integer k with the set $\{0, 1, \ldots, k - 1\}$, the zero 0 with the empty set.

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Souhrn

ISOMORFISMY MENDELOVSKÝCH POPULACÍ

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V článku jsou navrženy nebo zobecněny některé pojmy matematické populační genetiky sloužící jednak k popisu způsobu, jakým genotyp ovlivňuje fenotyp, jednak k popisu pravděpodobnostních zákonů dědičnosti. Hlavní výsledek článku je v důkazu tvrzení o vzájemně jednoznačném vztahu mezi isomorfismy mendelovské populace a permutacemi množiny lokusů a množin alel každého lokusu.

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