8. Probabilities on Inheritance in Consanguineous Families

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In a series of successive papers¹⁾ a unified theory of inheritance has been developed from a probabilistic view-point. In the present Note we consider again a single inherited character which consists of m multiple alleles at one diploid locus denoted by

A_i $(i=1,\ldots,m)$

and of which the inheritance is subject to Mendelian law. Our main purpose is to study the distributions of genotypes in several definite combinations, lineal or collateral, consisting of individuals chosen at random from a consanguineous family; a population in consideration is supposed to be in an equilibrium state unless the contrary is stated. We further suppose that the panmixie takes place at any generation except when the consanguineous marriages are appointed. The problem may be regarded as generalizations of those discussed in the previous papers²).

In the present preliminary Note we confine ourselves to a compendious announcement of the results. The details on the formulas as well as their consequences will be fully discussed in our subsequent paper⁵ which will nearly be published.

The definitions as well as the notations concerning several concepts contained in the previous papers will be retained. However, for the sake of brevity, the *frequencies of the genes* A_i (i=1,...,m) will be denoted merely by

i instead of p_i

provided no confusion can arise. Further, the different Latin letters except those designating the running types in summation are supposed, in principle, to indicate the different genes.

I. Simple lineal combination

1. Mother-nth descendant combination

The probability of mother-child combination $\pi \equiv \pi_1$ is now generalized to that of a combination consisting of an individual $A_{\alpha\beta}$ and

¹⁾ Y. Komatu, Probability-theoretic investigations on inheritance. I-XVI. Proc. Japan Acad. **27-29** (1951–1953).

²⁾ Cf. especially, IV. Mother-child combinations. **27** (1951), 587-620 and **29** (1953), 68-77; V. Brethren-combinations. **27** (1951), 689-699 and **29** (1953), 78-82.

³⁾ Y. Komatu and H. Nishimiya, Probabilistic investigations on inheritance in consanguineous families. Bull. Tokyo Inst. Tech. (1954).

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its *n*th descendant $A_{\xi\eta}$, which will be designated by

$$\pi_n(lphaeta; \xi\eta) \equiv \overline{A}_{lphaeta}\kappa_n(lphaeta; \xi\eta).$$

The reduced probability κ_n satisfies a recurrence equation

$$_{n}(lphaeta; \ \xi\eta) = \sum \kappa_{n-1}(lphaeta; \ ab)\kappa(ab; \ \xi\eta)$$

where the summation extends over all the possible types A_{ab} . The running suffices will be designated, in principle, by letters at the head of Latin alphabet. Now, it is shown that the probability is expressed by the formula

$$\kappa_n(\alpha\beta; \xi\eta) = \overline{A}_{\xi\eta} + 2^{-n+1}Q(\alpha\beta; \xi\eta)$$

where the quantity Q is independent of the generation-number n and its values are given as follows:

Q(ii; ii) = i(1-i),	Q(ii; ik) = k(1-2i),
$Q(ii; kk) = -k^2$,	Q(ii; hk) = -2hk;
$Q(ij; ii) = \frac{1}{2}i(1-2i),$	$Q(ij; ij) = \frac{1}{2}(i+j-4ij),$
$Q(ij; ik) = \frac{1}{2}k(1-4i),$	$Q(ij; kk) = -k^2$,
Q(ij; hk) = -2hk.	

The proof of the formula can be performed by induction⁴⁾ by actually verifying an identity

 $\sum Q(\alpha\beta; ab)\kappa(ab; \xi\eta) = \sum Q(\alpha\beta; ab)Q(ab; \xi\eta) = \frac{1}{2}Q(\alpha\beta; \xi\eta).$

It is noted that the quantity Q satisfies a quasi-symmetry relation

 $\overline{A}_{lphaeta}Q(lphaeta;\ \xi\eta) \!=\! \overline{A}_{\xi\eta}Q(\xi\eta;\ lphaeta)$

and further the identities

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 $\sum Q(a\beta; ab)=0, \qquad \sum \overline{A}_{ab}Q(ab; \xi_{\eta})=0.$

An asymptotic behavior of κ_n as $n \to \infty$ can be readily deduced from its expression derived above. In fact, there holds a limit equation

$$\lim \kappa_n(\alpha\beta; \xi\eta) = \overline{A}_{\xi\eta}$$

stating that a consanguineous correlation between the types of an individual and of its n th descendant becomes negligible for a sufficiently large n. The approach to the limit is for any pair monotone.

Finally, it is to be remarked that the values of $\kappa_n(\alpha\beta; \xi\eta)$, and hence also of $Q(\alpha\beta; \xi\eta)$, are evidently independent of $\alpha\beta$ provided the genotype $A_{\xi\eta}$ possesses no gene in common with $A_{\alpha\beta}$. Consequently, the values of $\kappa_n(ij; kk)$ and $\kappa_n(ij; hk)$ coincide with $\kappa_n(ii; kk)$ and $\kappa_n(ii; hk)$, respectively. Similar remarks will be availed passim.

2. Parents-n th descendant combination

Let a pair of parents with fixed types $(A_{\alpha\beta}, A_{\gamma\delta})$ be given, the order of members being taken into account. We then denote by

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⁴⁾ Another deductive derivation of the formula is found in Y. Komatu and H. Nishimiya, Lineal combinations on a Mendelian inherited character. Rep. Stat. Appl. Res., JUSE **3** (1953).

 $\varepsilon_n(\alpha\beta, \gamma\delta; \xi\eta)$

the probability that the pair is accompanied by an *n*th descendant $A_{\varepsilon_{\eta}}$. The values of $\varepsilon \equiv \varepsilon_1$ have already been availed in a previous paper⁵). The probability in question is given by a recurrence formula

 $\varepsilon_n(\alpha\beta, \gamma\delta; \xi\eta) = \sum \varepsilon(\alpha\beta, \gamma\delta; ab)\kappa_{n-1}(ab; \xi\eta).$

It is shown that the probability is expressed by the formula

$$\epsilon_n(\alpha\beta, \gamma\delta; \xi\eta) = \overline{A}_{\xi\eta} + 2^{-n+1}E(\alpha\beta, \gamma\delta; \xi\eta)$$

valid for $n \ge 2$. The factor E in the residual term is then defined by

$$E(\alpha\beta, \gamma\delta; \xi_{\eta}) = 2 \sum \epsilon(\alpha\beta, \gamma\delta; ab)Q(ab; \xi_{\eta})$$

and is symmetric with respect to $\alpha\beta$ and $\gamma\delta$. Its values are listed as follows:

E(ii, ii; ii) = 2i(1-i),E(ii, ii; ig) = 2g(1-2i), $E(ii, ii; gg) = -2g^2$, E(ii, ii; fg) = -4fg; $E(ii, ik; ii) = \frac{1}{2}i(3-4i),$ $E(ii, ik; ik) = \frac{1}{2}(i+3k-8ik),$ $E(ii, ik; kk) = \frac{1}{2}k(1-4k),$ $E(ii, ik; ig) = \frac{1}{2}g(3-8i),$ $E(ii, ik; kg) = \frac{1}{2}g(1-8k),$ $E(ii, ik; gg) = -2g^2,$ E(ii, ik; fg) = -4fg;E(ii, kk; ii) = i(1-2i),E(ii, kk; ik) = i + k - 4ikE(ii, kk; ig) = g(1-4i), $E(ii, kk; gg) = -2g^2$, E(ii, kk; fg) = -4fg;E(ii, hk; ii) = i(1-2i), $E(ii, hk; ik) = \frac{1}{2}(i+2k-8ik),$ $E(ii, hk; kk) = \frac{1}{2}k(1-4k),$ $E(ii, hk; hk) = \frac{1}{2}(h+k-8hk),$ E(ii, hk; ig) = g(1-4i), $E(ii, hk; kg) = \frac{1}{2}g(1-8k),$ $E(ii, hk; gg) = -2g^2$, E(ii, hk; fg) = -4fg;E(ij, ij; ii) = i(1-2i),E(ij, ij; ij) = i + j - 4ij, $E(ij, ij; gg) = -2g^2,$ E(ij, ij; ig) = g(1-4i),E(ij, ij; fg) = -4fg; $E(ij, ik; ij) = \frac{1}{2}(i+2j-8ij),$ E(ij, ik; ii) = i(1-2i), $E(ij, ik; jj) = \frac{1}{2}j(1-4j),$ $E(ij, ik; jk) = \frac{1}{2}(j+k-8jk),$ E(ij, ik; ig) = g(1-4i), $E(ij, ik; jg) = \frac{1}{2}g(1-8j),$ $E(ij, ik; gg) = -2g^2,$ E(ij, ik; fg) = -4fg; $E(ij, hk; ii) = \frac{1}{2}i(1-4i),$ $E(ij, hk; ij) = \frac{1}{2}(i+j-8ij),$ $E(ij, hk; gg) = -2g^2$, $E(ij, hk; ig) = \frac{1}{2}g(1-8i),$ E(ij, hk; fg) = -4fg.It is noted that the quantity E satisfies the identities

 $\sum E(\alpha\beta, \ \gamma\delta; \ ab) = 0, \qquad \sum \overline{A}_{ab} E(\alpha\beta, \ ab; \ \xi\eta) = Q(\alpha\beta; \ \xi\eta).$ An asymptotic behavior of ε_n as $n \to \infty$ is obvious; namely, we get

$$\lim_{n\to\infty} \varepsilon_n(\alpha\beta, \ \gamma\delta; \ \xi\eta) = \overline{A}_{\xi\eta}.$$

5) Cf. I. Distribution of genes. 27 (1951), 371-377.

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II. Simple collateral combinations

1. Collateral combination originated from same couple

By generalizing the probability of brethren combination $\sigma \equiv \sigma_{11}$, we introduce now that of a combination consisting of μ th and ν th collateral descendants $A_{\xi_1 \gamma_1}$ and $A_{\xi_2 \gamma_2}$, respectively, originated from the same couple, which will be designated by

$$\begin{split} \sigma_{\mu\nu}(\xi_1\eta_1, \ \xi_2\eta_2) &\equiv \overline{A}_{\xi_1\eta_1}\tau_{\mu\nu}(\xi_1\eta_1, \ \xi_2\eta_2). \\ \text{It is proved that there holds an identical relation} \\ \tau_{\mu\nu}(\xi_1\eta_1, \ \xi_2\eta_2) &\equiv \kappa_{\mu+\nu-1}(\xi_1\eta_1; \ \xi_2\eta_2) \end{split}$$

provided $\mu + \nu > 2$, while it should be remembered that τ_{11} is not identical with κ_1 . The last relation shows, in particular, that the dependence of $\tau_{\mu\nu}$ with $\mu + \nu > 2$ on the generation-numbers is subject merely to their sum $\mu + \nu$.

2. Collateral combination originated from different couples We next designate by

At designate by

 $\sigma^0_{\mu
u}(\xi_1\eta_1, \xi_2\eta_2) \equiv \overline{A}_{\xi_1\eta_1} \tau^0_{\mu
u}(\xi_1\eta_1, \xi_2\eta_2)$

the probability of a combination consisting of μ th and ν th descendants $A_{\xi_1\eta_1}$ and $A_{\xi_2\eta_2}$, respectively, originated from two different couples with a female alone in common.

It is proved that there holds an identity

for any pair of μ and ν with $\mu, \nu \geq 1$, and hence $\tau^0_{\mu\nu} = \tau_{\mu+1,\nu} = \tau_{\mu,\nu+1}$.