

50. Probabilities on Inheritance in Consanguineous Families. VI

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VI. Mother-descendant combinations through several consanguineous marriages

1. Special combination with several consanguineous marriages

We have discussed in the preceding chapter¹⁾ the mother-descendant combinations through a single consanguineous marriage. We now attempt to generalize the problem to the case where several consanguineous marriages intervene between a mother and her descendant. Our present purpose is to determine the probability of a combination which consists of the original individual $A_{\alpha\beta}$ and of its descendant $A_{\xi\eta}$, consanguineous marriages interjacent between them occurring t times, and which is designated, with an easily comprehensible routine notation, by

$$\pi_{\mu_1\nu_1; n_1 | \dots | \mu_t\nu_t; n_t}(\alpha\beta; \xi\eta) \equiv \bar{A}_{\alpha\beta} \kappa_{\mu_1\nu_1; n_1 | \dots | \mu_t\nu_t; n_t}(\alpha\beta; \xi\eta)$$

or briefly, provided no confusion can arise, by

$$\pi_{(\mu\nu; n)_t}(\alpha\beta; \xi\eta) \equiv \bar{A}_{\alpha\beta} \kappa_{(\mu\nu; n)_t}(\alpha\beta; \xi\eta).$$

By definition, the reduced probability $\kappa_{(\mu\nu; n)_t}$ is given by

$$\kappa_{(\mu\nu; n)_t}(\alpha\beta; \xi\eta) = \sum \prod_{r=1}^t \kappa_{\mu_r\nu_r; n_r}(a_{r-1}b_{r-1}; a_r b_r)$$

with $a_0b_0 \equiv \alpha\beta$ and $a_t b_t \equiv \xi\eta$, where the summation extends over all the possible sets of $t-1$ genotypes $A_{a_r b_r}$ ($r=1, \dots, t-1$).

We can really permit here also the degenerate cases where some of the n_r 's with $1 \leq r < t$ vanish. But, these exceptional cases will be postponed to a later chapter. In the present section we shall deal with a special case where the n 's are all equal to unity. Its defining equation then becomes

$$\kappa_{(\mu\nu; 1)_t}(\alpha\beta; \xi\eta) = \sum \kappa_{(\mu\nu; 1)_{t-1}}(\alpha\beta; ab) \kappa_{\mu_t\nu_t; 1}(ab; \xi\eta),$$

whence follows, as verified by induction, the formula

$$\begin{aligned} \kappa_{(\mu\nu; 1)_t}(\alpha\beta; \xi\eta) &= \bar{A}_{\xi\eta} + 2^{-t+1} A_t Q(\alpha\beta; \xi\eta) + 4u_t R(\xi\eta) \\ &\quad + 2v_t S(\alpha\beta; \xi\eta) + 2w_t T(\alpha\beta; \xi\eta); \\ A_t &= \prod_{r=1}^t L_r \equiv \prod_{r=1}^t (2^{-\mu_r} + 2^{-\nu_r}), & u_t &= \sum_{r=1}^{t-1} \prod_{s=r+1}^t 2^{-\lambda_s - 2}, \\ v_t &= \sum_{r=1}^{t-1} 2^{-r+1} A_r \prod_{s=r+1}^t 2^{-\lambda_s - 2}, & w_t &= \prod_{s=1}^t 2^{-\lambda_s - 2}; \end{aligned}$$

1) Cf. Proc. Japan Acad. **30** (1954), 152-155. There the value of $S(ij; ik)$ (p. 154, l. 6) should be read $-\frac{1}{2}k(1-4i)$ instead of $-\frac{1}{2}k(1-4k)$.

the last formula remains true even for $t=1$ provided an empty sum is understood, as usual, to be equal to zero.

2. Generic combinations with several consanguineous marriages

We now consider the generic case, i. e. the probability $\kappa_{(\mu\nu; n)_t}$ with $n_r > 1$ ($r=1, \dots, t$). Its defining equation becomes

$$\kappa_{(\mu\nu; n)_t}(\alpha\beta; \xi\eta) = \sum \kappa_{(\mu\nu; n)_{t-1}}(\alpha\beta; ab)\kappa_{\mu_t\nu_t; n_t}(ab; \xi\eta)$$

and leads to the desired formula

$$\kappa_{(\mu\nu; n)_t}(\alpha\beta; \xi\eta) = \bar{A}_{\xi\eta} + 2^{-N_t+1}A_tQ(\alpha\beta; \xi\eta), \quad N_t = \sum_{r=1}^t n_r,$$

which remains valid even when $t=1$. We remark here especially that the present formula remains valid regardless of the values of the n_r 's with $1 \leq r < t$ provided merely $n_t > 1$.

We next consider more generally a probability of the form

$$\kappa_{(\mu\nu; n)_t | (\mu'\nu'; 1)_t}(\alpha\beta; \xi\eta) \equiv \kappa_{\mu_1\nu_1; n_1 | \dots | \mu_t\nu_t; n_t | \mu'_1\nu'_1; 1 | \dots | \mu'_t\nu'_t; 1}(\alpha\beta; \xi\eta)$$

with $n_r \geq 1$ for $1 \leq r < t$ and $n_t > 1$. It is defined by an equation

$$\kappa_{(\mu\nu; n)_t | (\mu'\nu'; 1)_t}(\alpha\beta; \xi\eta) = \sum \kappa_{(\mu\nu; n)_t}(\alpha\beta; ab)\kappa_{(\mu'\nu'; 1)_t}(ab; \xi\eta).$$

We obtain the formula

$$\begin{aligned} \kappa_{(\mu\nu; n)_t | (\mu'\nu'; 1)_t}(\alpha\beta; \xi\eta) &= \bar{A}_{\xi\eta} + 2^{-N_t-t'+1}A_tA'_tQ(\alpha\beta; \xi\eta) \\ &\quad + 4(u'_t + w'_t)R(\xi\eta) + 2^{-N_t+1}A_t(v'_t + 2w'_t)S(\alpha\beta; \xi\eta), \end{aligned}$$

where we put

$$\begin{aligned} A'_t &= \prod_{r=1}^{t'} (2^{-\mu'_r} + 2^{-\nu'_r}), & u'_t &= \sum_{r=1}^{t'-1} \prod_{s=r+1}^{t'} 2^{-\lambda'_s-2}, \\ v'_t &= \sum_{r=1}^{t'-1} 2^{-r+1}A'_r \prod_{s=r+1}^{t'} 2^{-\lambda'_s-2}, & w'_t &= \prod_{s=1}^{t'} 2^{-\lambda'_s-2}. \end{aligned}$$

It should be noted that the last formula for $\kappa_{(\mu\nu; n)_t | (\mu'\nu'; 1)_t}$ loses its effect for $t=0$ as well as for $t'=0$.

3. General mother-descendant combinations through several consanguineous marriages

In the present section we shall deal with the probability $\kappa_{l | (\mu\nu; n)_t}$. It is given by a recurrence equation

$$\kappa_{l | (\mu\nu; n)_t}(\alpha\beta; \xi\eta) = \sum \kappa_l(\alpha\beta; ab)\kappa_{(\mu\nu; n)_t}(ab; \xi\eta).$$

By retaining the notations introduced in the preceding sections, we first get

$$\begin{aligned} \kappa_{l | (\mu\nu; 1)_t}(\alpha\beta; \xi\eta) &= \bar{A}_{\xi\eta} + 2^{-l-t+1}A_tQ(\alpha\beta; \xi\eta) \\ &\quad + 4(u_t + w_t)R(\xi\eta) + 2^{-l+1}(v_t + 2w_t)S(\alpha\beta; \xi\eta). \end{aligned}$$

In any case with $n_t > 1$, we get briefly

$$\kappa_{l | (\mu\nu; n)_t}(\alpha\beta; \xi\eta) = \bar{A}_{\xi\eta} + 2^{-l-N_t+1}A_tQ(\alpha\beta; \xi\eta).$$

Finally, in case corresponding to the last one of the preceding section, i. e. in case $n_t > 1$, we get

$$\begin{aligned} \kappa_{l | (\mu\nu; n)_t | (\mu'\nu'; 1)_t}(\alpha\beta; \xi\eta) &= \bar{A}_{\xi\eta} + 2^{-l-N_t-t'+1}A_tA'_tQ(\alpha\beta; \xi\eta) \\ &\quad + 4(u'_t + w'_t)R(\xi\eta) + 2^{-l-N_t+1}A_t(v'_t + 2w'_t)S(\alpha\beta; \xi\eta). \end{aligned}$$

4. Distribution of genotypes after consanguineous marriages

The present section is devoted to illustrate a very notable

phenomenon concerning the distribution of genotypes in a generation after consanguineous marriages. Up to now we have dealt with several mother-descendant combinations $(A_{\alpha\beta}; A_{\xi\eta}) \dots$, of which the probabilities are designated by $\pi \dots (\alpha\beta; \xi\eta) \equiv \bar{A}_{\alpha\beta\kappa} \dots (\alpha\beta; \xi\eta)$, respectively. For each combination, if we eliminate mother's type by summing up over whole possible range, then we obtain the probability of descendant $A_{\xi\eta}$ alone, i. e. the relative frequency of genotype $A_{\xi\eta}$ in the generation of descendant, which will be designated by

$$\bar{A} \dots (\xi\eta) = \sum \pi \dots (ab; \xi\eta) \equiv \sum \bar{A}_{ab\kappa} \dots (ab; \xi\eta).$$

In case of a simple mother-descendant combination, with probability κ_n , without intervention of any consanguineous marriage, it is given by

$$\bar{A}_n(\xi\eta) = \sum \pi_n(ab; \xi\eta) = \bar{A}_{\xi\eta}.$$

The last relation shows that the distribution of $A_{\xi\eta}$ in the generation of descendant then coincides just with one in the original generation.

We next consider the mother-descendant combination denoted by $(A_{\alpha\beta}; A_{\xi\eta})_{(\mu\nu; 1)_t}$. The distribution in the generation of descendant is then given by

$$\bar{A}_{(\mu\nu; 1)_t}(\xi\eta) = \bar{A}_{\xi\eta} + 4(u_t + w_t)R(\xi\eta),$$

where

$$u_t + w_t = \sum_{r=0}^{t-1} \prod_{s=r+1}^t 2^{-\lambda_s - 2}, \quad \lambda_s = \mu_s + \nu_s - 1;$$

$$R(ii) = \frac{1}{2} i(1-i), \quad R(ij) = -ij \quad (i \neq j).$$

In case of mother-descendant combination $(A_{\alpha\beta}; A_{\xi\eta})_{(\mu\nu; n)_t}$ with $n_t > 1$, we get

$$\bar{A}_{(\mu\nu; n)_t}(\xi\eta) = \bar{A}_{\xi\eta}.$$

In quite a similar manner, we obtain

$$\bar{A}_{(\mu\nu; n)_t | (\mu'\nu'; 1)_{t'}}(\xi\eta) = \bar{A}_{\xi\eta} + 4(u'_{t'} + w'_{t'})R(\xi\eta),$$

and furthermore

$$\bar{A}_{t | (\mu\nu; 1)_t}(\xi\eta) = \bar{A}_{\xi\eta} + 4(u_t + w_t)R(\xi\eta),$$

$$\bar{A}_{t | (\mu\nu; n)_t}(\xi\eta) = \bar{A}_{\xi\eta} \quad \text{for } n_t > 1,$$

$$\bar{A}_{t | (\mu\nu; n)_t | (\mu'\nu'; 1)_{t'}}(\xi\eta) = \bar{A}_{\xi\eta} + 4(u'_{t'} + w'_{t'})R(\xi\eta),$$

where

$$u'_{t'} + w'_{t'} = \sum_{r=0}^{t'-1} \prod_{s=r+1}^{t'} 2^{-\lambda'_s - 2}, \quad \lambda'_s = \mu'_s + \nu'_s - 1.$$

We can thus assert the following proposition: *The distribution of genotypes deviates in a generation immediate after any consanguineous marriage, while the deviation disappears in a generation*

distant after the last consanguineous marriage. The deviation of genotype $A_{\xi\eta}$ in the former case amounts to $R(\xi\eta)$ multiplied by a definite positive factor depending only on the generation-numbers which antecede the last marriage and refer to last successive collateral separations occurring immediately after consanguineous marriages. Consequently, the frequency of $A_{\xi\eta}$ in the generation of descendant increases or decreases according to whether it is homozygous or heterozygous.

Thus, we are led to a conjecture concerning practical observations. Suppose, in fact, there exists a population which is isolated hereditarily from its neighboring populations and in which consanguineous marriages take place to a considerable extent. For instance, one may imagine a population living on an isolated island which lacks facilities of communication. If one will observe frequencies of an inherited character in the population, it will then be found out that the frequencies of homozygous and heterozygous types are more than and less than those expected from the supposition of an equilibrium state, respectively.

We have seen above that the deviation of distribution disappears in the second generation after a consanguineous marriage. This phenomenon resembles the stability of an equilibrium state of distribution based on the *buffer-effect* by random matings. However, these two phenomena are really distinct in nature. In fact, the disappearance of deviation is caused, in our present case, by random matings *between* a deviated population and an undeviated one. On the contrary, for disappearance of deviation in case of buffer-effect, the random matings are supposed to take place *within* the deviated population. It is however shown that the result of buffer-effect coincides also quite with the ordinary one. Consequently, although our discussions have been exclusively based on the random matings between a deviated population and an undeviated one, the results remain valid even when the random matings take place within the deviated population.

5. Asymptotic behaviors of the probabilities

The asymptotic behaviors of the probability $\kappa_{l|(\mu\nu; n)_t}$, as each among l , μ_r , ν_r and n_r tends to infinity, can be readily deduced from its expression derived above, though the results which will be obtained are regarded as quite plausible. We obtain, in fact, the following limit equations:

$$\begin{aligned} \lim_{\mu_z \rightarrow \infty} \kappa_{l|(\mu\nu; n)_t}(\alpha\beta; \xi\eta) &= \kappa_{l|(\mu\nu; n)_{z-2} | \mu_{z-1} \nu_{z-1}; n_{z-1} + \nu_z + n_z | (\mu' \nu'; n')_{t-z}}(\alpha\beta; \xi\eta), \\ \lim_{\mu_z \rightarrow \infty} \kappa_{l|(\mu\nu; n)}(\alpha\beta; \xi\eta) &= \bar{A}_{(\mu' \nu'; n')_{t-z}}(\xi\eta), \\ \lim_{l \rightarrow \infty} \kappa_{l|(\mu\nu; n)_t}(\alpha\beta; \xi\eta) &= \bar{A}_{(\mu\nu; n)_t}(\xi\eta), \end{aligned}$$

where we put $\mu'_s = \mu_{z+s}$, $\nu'_s = \nu_{z+s}$, $n'_s = n_{z+s}$ for $1 \leq s \leq t-z$. In these equations, the generation-numbers except the one which tends to infinity may be quite arbitrary; in particular, l may be equal to zero, a case which will be easily comprehensible.

We now observe the asymptotic behaviors of the probability $\kappa_{(\mu\nu; n)_t}$, as $t \rightarrow \infty$. It depends on the choice of a sequence $\{\mu\nu; n\} \equiv \{\mu_r, \nu_r; n_r\}_{r=1}^\infty$.

It will be sufficient to investigate an essential case where $n_r = 1$ ($r = 1, 2, \dots$). We suppose that an arbitrary sequence $\{\mu\nu\}$ consisting of positive integers μ_r, ν_r ($r = 1, 2, \dots$) is given. *If there exists a number τ such that*

$$\lambda_r = \lambda_\infty \text{ (const) } \quad \text{for } r > \tau$$

then we have

$$\lim_{t \rightarrow \infty} \kappa_{(\mu\nu; 1)_t}(\alpha\beta; \xi\eta) = \bar{A}_{\xi\eta} + \frac{4}{2^{\lambda_\infty+2} - 1} R(\xi\eta),$$

and if

$$\lim_{r \rightarrow \infty} \lambda_r = \infty$$

then we have

$$\lim_{t \rightarrow \infty} \kappa_{(\mu\nu; 1)_t}(\alpha\beta; \xi\eta) = \bar{A}_{\xi\eta}.$$

The latter case may be regarded as a particular one included in the former. In fact, λ_r being restricted to an integer, the assumption $\lambda_r = \lambda_\infty$ for $r > \tau$ is equivalent to $\lim_{r \rightarrow \infty} \lambda_r = \lambda_\infty$.

Otherwise, the sequence $\{\kappa_{(\mu\nu; 1)_t}(\alpha\beta; \xi\eta)\}$ oscillates, as t increases, within certain upper and lower bounds.

To state more precisely, we introduce the notations defined by

$$\begin{aligned} \bar{\lambda} &= \overline{\lim}_{r \rightarrow \infty} \lambda_r, & \underline{\lambda} &= \underline{\lim}_{r \rightarrow \infty} \lambda_r, \\ \bar{\nu} &= \frac{4}{2^{\bar{\lambda}+2} - 1}, & \underline{\nu} &= \frac{4}{2^{\underline{\lambda}+2} - 1}, \end{aligned}$$

for which there hold evidently the inequalities

$$1 \leq \underline{\lambda} \leq \bar{\lambda} \leq \infty, \quad 0 \leq \underline{\nu} \leq \bar{\nu} \leq 4/7.$$

We can then assert that, in general, *there hold, for any sequence $\{\mu\nu\}$ the estimations*

$$\begin{aligned} \bar{A}_{ii} + \underline{\nu}R(ii) &\leq \underline{\lim}_{t \rightarrow \infty} \kappa_{(\mu\nu; 1)_t}(\alpha\beta; ii) \leq \overline{\lim}_{t \rightarrow \infty} \kappa_{(\mu\nu; 1)_t}(\alpha\beta; ii) \leq \bar{A}_{ii} + \bar{\nu}R(ii), \\ \bar{A}_{ij} + \bar{\nu}R(ij) &\leq \underline{\lim}_{t \rightarrow \infty} \kappa_{(\mu\nu; 1)_t}(\alpha\beta; ij) \leq \overline{\lim}_{t \rightarrow \infty} \kappa_{(\mu\nu; 1)_t}(\alpha\beta; ij) \leq \bar{A}_{ij} + \underline{\nu}R(ij). \end{aligned}$$