139. On Probabilities of Non-Paternity with Reference to Consanguinity. I

By Yûsaku KOMATU and Han NISHIMIYA

Department of Mathematics, Tokyo Institute of Technology and
Department of Mathematics, Shibaura Institute of Technology

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In a series of papers we have dealt with an inherited character subject to Mendelian law from probabilistic view point. Considering a single inherited character which consists of m multiple alleles at one diploid locus denoted by

$$A_i \qquad (i=1,\cdots,m),$$

we have determined, among others, the probability of non-paternity, i.e. the probability that a putative man has a genotype inconsistent with those of a mother-child combination, both the man and the combination being chosen at random from a population; cf. esp. [1]. It has been supposed that there exists no consanguineous relationship among the putative man and the parents of the child. Accordingly, the distribution of genotypes in the population has been supposed to be an ordinary one in an equilibrium state.

On the other hand, in another series of papers we have discussed the distributions of genotypes in several definite combinations which consist of individuals chosen at random under various imposed consanguineous relationships. In particular, we have shown that the probabilities of sib-combinations are to be modified according to the presence of consanguinity; cf. esp. [2].

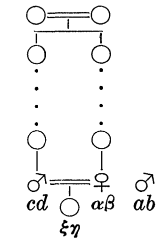
Now the question arises to determine how the probabilities of non-paternity are to be modified correspondingly. In the present paper this problem will be studied for a few particular types of consanguinity existent among a triple of relevant individuals. Namely, we restrict ourselves here to consider triples for which (μ,ν)th sibship exists between parents of a child or between a putative man and one of the parents. A remarkable monotoneity caused by the presence of consanguinity will become clear while it seems previously plausible to some extent. In fact, it will be verified that in every case the probability of non-paternity always decreases by virtue of the presence of consanguinity. According to circumstances, each kind of consanguineous relationships will be considered separately in the sequel.

Various definitions and notations concerning several concepts contained in the previous papers will be retained here also. In particular, the frequencies of the genes A_i $(i=1,\cdots,m)$ will be denoted often merely by i instead of p_i respectively, provided no confusion can arise. Further, we suppose as before that different Latin letters except those designating the running suffices in summation indicate, in principle, different genes.

1. **Non-paternity of a putative man not related with the child.** We begin with the case where the parents of a child under consideration have μth and νth antecedants in common while a putative man is in no consanguineous relation with them. The probability of this mother-child combination $(A_{\alpha\beta}; A_{\xi\eta})$ is given by

$$\pi(\alpha\beta; \xi\eta \mid \sigma_{\mu\nu;1}) = \sum \sigma_{\mu\nu}(\alpha\beta; cd)\varepsilon(\alpha\beta, cd; \xi\eta)$$

where the summation extends over all the possible types A_{cd} (of the true father). Inserting the values of $\sigma_{\mu\nu}$ and ε already determined in the previous papers, we obtain the following result, the integer λ being defined, as before, by



$$\lambda = \mu + \nu - 1:$$

$$\pi(ii; ii \mid \sigma_{\mu\nu;1}) = i^3 + 2^{-\lambda} i^2 \ (1-i),$$
$$\pi(ii; ik \mid \sigma_{\mu\nu;1}) = i^2 k - 2^{-\lambda} i^2 k;$$
$$\pi(ij; ii \mid \sigma_{\mu\nu;1}) = i^2 j + 2^{-\lambda-1} ij \ (1-2i),$$
$$\pi(ij; ij \mid \sigma_{\mu\nu;1}) = ij\,(i+j) + 2^{-\lambda} ij\,(1-i-j),$$
$$\pi(ij; ik \mid \sigma_{\mu\nu;1}) = ijk - 2^{-\lambda} ijk.$$

Though the formula for $\sigma \equiv \sigma_{11;1}$ is exceptional against that for $\sigma_{\mu\nu;1}$ with $\lambda > 1$, the result just derived for $\pi(\alpha\beta; \xi\eta \mid \sigma_{\mu\nu;1})$ remains valid for any $\lambda \geqq 1$ without exception.

On the other hand, the probability that, for a fixed mother-child combination $(A_{\alpha\beta}; A_{\xi\eta})$, a putative man chosen at random is inconsistent with this combination is evidently given by

$$V(\alpha\beta; \xi\eta) = \sum_{ab \in \Omega} \bar{A}_{ab}$$

where the summation extends over the set $\Omega = \Omega(\alpha\beta; \xi\eta)$ of types A_{ab} which together with $A_{\alpha\beta}$ can not produce $A_{\xi\eta}$. This probability has been already determined and may be reproduced here as follows:

$$V(ii; ii) = (1-i)^2, \qquad V(ii; ik) = (1-k)^2;$$
$$V(ij; ii) = (1-i)^2, \qquad V(ij; ij) = (1-i-j)^2,$$
$$V(ij; ik) = (1-k)^2.$$

The probability that a mother-child combination is $(A_{\alpha\beta}; A_{\xi\eta})$ and a putative man chosen at random can prove his non-paternity against this combination is then given by the product

$$P(\alpha\beta; \xi\eta \mid \sigma_{\mu\nu;1}) = \pi(\alpha\beta; \xi\eta \mid \sigma_{\mu\nu;1}) V(\alpha\beta; \xi\eta).$$

By summing up this quantity with respect to all combinations $(\alpha\beta; \xi\eta)$, we get the desired probability, i.e. the total probability of non-paternity with respect to the triples under consideration which will be denoted by $P(\sigma_{\mu\nu;1})$. The final result becomes

$$
\begin{aligned}
P(\sigma_{\mu\nu;1}) &= P-2^{-\lambda-1}(2S_2-3S_3-6S_2^2+7S_4+6S_2S_3-6S_5) \\
&= (1-2^{-\lambda})P+2^{-\lambda-1}(2-6S_2+5S_3+2S_2^2-3S_4)
\end{aligned}
$$

where S_k denotes, as before, the power-sum defined by

$$S_k=\sum_{i=1}^{m} p_i^k \qquad (k=1,2,\cdots;\ S_1=1)$$

and P is the probability of ordinary non-paternity without any consanguineous relationship. The latter is equal to

$$P=1-2S_2+S_3-2S_2^2+2S_4+3S_2S_3-3S_5.$$

It may be noted that the value of $P(\sigma_{\mu\nu;1})$ depends on the generation-numbers μ and ν through $\lambda=\mu+\nu-1$ only and hence the interchange of μ and ν has no effect.

As previously noticed, it seems remarkable that the probability of non-paternity always *decreases* by virtue of the presence of consanguinity under consideration, i.e.

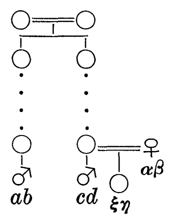
$$P(\sigma_{\mu\nu;1})\leqq P.$$

In fact, the factor of the residual term in the above formula can be written in the form

$$
\begin{aligned}
&2^{\lambda+1}(P-P(\sigma_{\mu\nu;1}))=2S_2-3S_3-6S_2^2+7S_4+6S_2S_3-6S_5 \\
&=\sum{}' ij(2(i+j)-(i^2+j^2)-12ij+6ij(i+j)) \\
&=\sum{}' ij(2(i+j+3ij)(1-i-j)+(i-j)^2),
\end{aligned}
$$

where the prime attached to the summation symbol means that i and j extend over all the possible pairs with $i<j$. It is clear from the last expression that this quantity remains always non-negative; we see further that if every gene is really present, the strict inequality $P(\sigma_{\mu\nu;1})<P$ holds unless $m=2$ and $p_1=p_2=1/2$. More precisely, $\{P(\sigma_{\mu\nu;1})\}$, regarded as a sequence depending on $\lambda=\mu+\nu-1$, increases monotonously and tends to the limit P as $\lambda\to\infty$.

2. Non-paternity of a putative man related with the father but not with the mother. Suppose now that a putative man and the true father of a child have antecedants of μth and νth generations respectively in common while the mother is in no consanguineous relation with them. Then, the probability of the triple which consists of a putative man A_{ab} and a mother-child combination $(\alpha\beta;\xi\eta)$ under the imposed relationship is given by



$$\bar{A}_{ab}\pi(\alpha\beta;\xi\eta\mid ab\,\sigma_{\mu,\nu+1}^{\male\female})=\bar{A}_{\alpha\beta}\sum\sigma_{\mu\nu}(ab,cd)\,\varepsilon(cd,\alpha\beta;\xi\eta)$$

the summation extending over all the possible types A_{cd} (of the true father).

Now the probability that a mother-child combination is $(\alpha\beta;\xi\eta)$ and a putative man chosen at random can prove his non-paternity against this combination is given by

$$P(\alpha\beta;\xi\eta\mid\sigma_{\mu,\nu+1}^{\male\female})=\sum_{\Omega}\bar{A}_{ab}\pi(\alpha\beta;\xi\eta\mid ab\,\sigma_{\mu,\nu+1}^{\male\female})$$

where the summation extends over the set $\Omega=\Omega(\alpha\beta;\xi\eta)$ of types A_{ab}

which together with $A_{\alpha\beta}$ can not produce $A_{\xi\eta}$. Thus, it becomes necessary to know the value of the summand at least for $ab \in \Omega$. But it can be shown directly that there exists a remarkable relation

$$\pi(\alpha\beta;\xi\eta \mid ab\,\sigma_{\mu,\nu+1}^{\male\female})=(1-2^{-\lambda})\pi(\alpha\beta;\xi\eta) \quad \text{for any } ab\in\Omega.$$

It should be noticed, however, that this relation does not necessarily hold for $ab \notin \Omega$. For instance, we have

$$\pi(ii;ii \mid ii\,\sigma_{\mu,\nu+1}^{\male\female})=(1-2^{-\lambda})\pi(ii;ii)+2^{-\lambda}i^2,$$

$$\pi(ij;ii \mid ik\,\sigma_{\mu,\nu+1}^{\male\female})=(1-2^{-\lambda})\pi(ij;ii)+2^{-\lambda-1}ij, \quad \text{etc.}$$

In view of the relation established just above, the subsequent calculation can be economized and really reduced to the previous one. In fact, we get

$$P(\alpha\beta;\xi\eta \mid \sigma_{\mu,\nu+1}^{\male\female})=\sum_{\Omega}\bar{A}_{ab}(1-2^{-\lambda})\pi(\alpha\beta;\xi\eta)$$

$$=(1-2^{-\lambda})\pi(\alpha\beta;\xi\eta)\sum_{\Omega}\bar{A}_{ab}=(1-2^{-\lambda})\pi(\alpha\beta;\xi\eta)V(\alpha\beta;\xi\eta).$$

Consequently, by summing up this quantity with respect to all combinations $(\alpha\beta;\xi\eta)$, we get the total probability of non-paternity for the present case which is simply expressed by the formula

$$P(\sigma_{\mu,\nu+1}^{\male\female})=(1-2^{-\lambda})P.$$

It is evident that the probability of non-paternity *decreases* by virtue of the presence of consanguinity also in this case, a fact which seems qualitatively quite plausible according to the circumstances. However, the above formula shows that its decrement compared with the ordinary one is due to a multiplicative factor $1-2^{-\lambda}$. In particular, the sequence $\{P(\sigma_{\mu,\nu+1}^{\male\female})\}$ increases monotonously with respect to λ and tends to the limit P as $\lambda\to\infty$.

In the argument performed just above for deriving the partial probability $P(\alpha\beta;\xi\eta \mid \sigma_{\mu,\nu+1}^{\male\female})$, the summation process has been made in two steps, namely first with respect to cd and then to ab. But the order of summation may be inverted. Of course, the same result will then be obtained again.

References

[1] Komatu, Y.,: Probability-theoretic investigations on inheritance. VII. Non-paternity problems; XI. Absolute non-paternity, Proc. Japan Acad., **28**, 102-108; 311-316 (1952).

[2] Komatu, Y., and Nishimiya, H.,: Probabilities on inheritance in consanguineous families, Proc. Japan Acad., **30**, 42-45 (1954).