118. Probability-theoretic Investigations on Inheritance. XV_4. Detection of Interchange of Infants.

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7. Illustrative examples, recessive genes being existent.

Problems and results discussed in the preceding sections have exclusively concerned genotypes. In case of existence of recessive genes, the procedure has only to be modified according to the corresponding dominance relations. We give here, as illustrative examples, the results on ABO, Q as well as $Qq_{\pm}$ blood types.

First, for *ABO blood type*, applying the process explained in § 3 to a corresponding table, we get

$$
\begin{aligned}
&G_0(O, O) && &&= r^4(1-r^2), \\
&G_0(O, A) &&= G_0(A, O) &&= pqr^2(p+2r)(2-q), \\
&G_0(O, B) &&= G_0(B, O) &&= pqr^2(q+2r)(2-p), \\
&G_0(O, AB) &&= G_0(AB, O) &&= 2pqr^2(r^2+2pq), \\
(7.1)\qquad &G_0(A, A) && &&= p^2q(p+2r)^2(2-q), \\
&G_0(A, B) &&= G_0(B, A) &&= 0, \\
&G_0(A, AB) &&= G_0(AB, A) &&= 2p^2qr^2(p+2r), \\
&G_0(B, B) && &&= pq^2(q+2r)^2(2-p), \\
&G_0(B, AB) &&= G_0(AB, B) &&= 2pq^2r^2(q+2r), \\
&G_0(AB, AB) && &&= 4p^2q^2r^2.
\end{aligned}
$$

The total sum of these sixteen quantities represents the probability G_{0ABO} of detecting the interchange of infants within the first triple. The expression for this probability being evidently symmetric with respect to p and q, it can be expressed in a unique manner as a function of two independent variables r and pq. In fact, by remembering a recurrence formula

$$p^\nu+q^\nu=(1-r)(p^{\nu-1}+q^{\nu-1})-pq(p^{\nu-2}+q^{\nu-2}),$$

we obtain an expression

$$
\begin{aligned}
(7.2)\qquad G_{0ABO}=r^4(1-r^2)+2pq(1+r+3r^2+3r^3+2r^4) \\
-p^2q^2(7+12r+5r^2)+2p^3q^3.
\end{aligned}
$$

Next, we obtain in turn

$$
\begin{aligned}
&\Phi(O, O) &&= 0,\\
&\Phi(O, A) = \Phi(A, O) &&= pr^3\{pq(2(p+r)^2+q(p+2r))\\
& && \qquad + r(p+r)(q+r)^2\},\\
&\Phi(O, B) = \Phi(B, O) &&= qr^3\{pq(2(q+r)^2+p(q+2r))\\
& && \qquad + r(q+r)(p+r)^2\},\\
&\Phi(O, AB) = \Phi(AB, O) &&= pqr^2\{p(p+r)^2(p+2r)\\
& && \qquad + q(q+r)^2(q+2r)\},\\
&\Phi(A, A) &&= p^2r^2\{pq(2(p+r)^2+q(p+2r))\\
& && \qquad + (p+r)(p+3r)(q+r)^2\},\\
(7.3)\quad &\Phi(A, B) = \Phi(B, A) &&= pq\{4pqr^2(1-pq)+r(p+r)(q+r)((p+r)^3\\
& && \quad +(q+r)^3)+r^2(2-r^2)(p+r)(q+r)\\
& && \quad +(p+r)(q+r)(p^2(p+2r)+q^2(q+2r))\},\\
&\Phi(A, AB) = \Phi(AB, A) &&= p^2q\{p(p+r)^2(p+2r)^2\\
& && \quad + q(2p+3r)(q+r)^2(q+2r)+4pqr^2(p+r)\},\\
&\Phi(B, B) &&= q^2r^2\{pq(2(q+r)^2+p(q+2r))\\
& && \qquad + (q+r)(q+3r)(p+r)^2\},\\
&\Phi(B, AB) = \Phi(AB, B) &&= pq^2\{q(q+r)^2(q+2r)^2\\
& && \quad + p(2q+3r)(p+r)^2(p+2r)+4pqr^2(q+r)\},\\
&\Phi(AB, AB) &&= p^2q^2\{3p(p+r)^2(p+2r)\\
& && \qquad + 3q(q+r)^2(q+2r)+8pqr^2\}.
\end{aligned}
$$

The total sum of these sixteen quantities represents the probability Φ_{ABO} of detecting the interchange only with reference to the second triple for which we get, similarly as above, an expression

$$
\begin{aligned}
(7.4)\quad \Phi_{ABO} = {} & r^4(1-r^2)+2pq(1+r+r^2+r^3-2r^4)\\
& -p^2q^2(7+12r+5r^2+20r^3-2r^4)+2p^3q^3(1-4r-8r^2)-2p^4q^4.
\end{aligned}
$$

Addition of (7.2) and (7.4) yields the *whole probability* of detecting the interchange of infants in the form

$$
\begin{aligned}
(7.5)\quad G_{ABO} = {} & 2r^4(1-r^2)+4pq(1+r+2r^2+2r^3)\\
& -2p^2q^2(7+12r+5r^2+10r^3-r^4)+4p^3q^3(1-2r-4r^2)-2p^4q^4.
\end{aligned}
$$

We have here to mention that an explicit expression for the whole probability has previously been obtained and expressed by Wiener[1] in the form

$$
\begin{aligned}
& G_{ABO} = 2\{(\overline{A}+\overline{AB})(\overline{B}+\overline{O})^2+(\overline{B}+\overline{AB})(\overline{A}+\overline{O})^2-\overline{O}^2\overline{AB}^2\\
(7.6)\quad & \quad -\overline{AB}(\overline{A}+\overline{O})(\overline{B}+\overline{O})\}+2\overline{O}\,\overline{AB}\{2+\overline{AB}-(1-\overline{AB})(1+\overline{O})\\
& \quad -\overline{A}(q-\tfrac{3}{4}\overline{AB})-\overline{B}(p-\tfrac{3}{4}\overline{AB})\}.
\end{aligned}
$$

Replacing the frequencies of phenotypes in Wiener's expression by those of genes, one can easily verify that (7.6) is essentially identical with (7.5).

An inequality corresponding to (6.1) remains here also valid

1) A. S. Wiener, On the usefulness of blood grouping in medicolegal case involving blood relationship. Journ. Immun. **24** (1933), 443-454,

based on the same reason stated there. But, it can be verified directly in a rather simple way. Namely, the difference of (7.2) and (7.4) becomes

$$G_{0ABO}-\Phi_{ABO}=4pqr^2(1+r+2r^2)+4p^2q^2r^3(5-r)+4p^3q^3r(1+r)+2p^4q^4, \tag{7.7}$$

which is evidently non-negative always and moreover positive unless $pq=0$.

Next, for *Q blood type*, a fact that the mating $q\times q$ is unable to produce Q, implies

$$G_0(q, q)=\bar{q}^2\bar{Q}=uv^4(1+v). \tag{7.8}$$

On the other hand, if at least one of parents is Q, then the child can be Q as well as q and hence

$$G_0(Q, Q)=G_0(Q, q)=G_0(q, Q)=0. \tag{7.9}$$

The second quantity of (5.2) can be determined as follows:

$$\begin{aligned} \Phi(Q, Q) &=u^2(1+2v)v^4, \\ \Phi(Q, q)=\Phi(q, Q) &=uv^6, \\ \Phi(q, q) &=0. \end{aligned} \tag{7.10}$$

Thus, we conclude

$$G_{0Q}=\Phi_Q=uv^4(1+v), \tag{7.11}$$

$$G_Q=2uv^4(1+v). \tag{7.12}$$

An inequality corresponding to (6.1) reduces here to an identity.

Last, for *$Qq_\pm$ blood type*, the following results will be derived in a similar manner as above:

$$\begin{aligned} &G_0(q_-, q_-) =v_1^2(v+v_2)^2u(1+v), \\ &G_0(q_-, q_+)=G_0(q_+, q_-)=v_1v_2(v+v_2)u(1+v), \\ &G_0(q_+, q_+)=v_2^4(u(1+v)+2uv_1(1+v)(v+v_2)), \\ &G_0(Q, Q) =G_0(Q, q_-)=G_0(q_-,Q)=G_0(Q, q_+)=G_0(q_+, Q)=0; \end{aligned} \tag{7.13}$$

$$\begin{aligned} &\Phi(Q, Q) =u^2(1+2u)v^4+u^2v_1(v+v_2)v_2^4, \\ &\Phi(Q, q_-) =\Phi(q_-, Q) =uv_1(v+v_2)v^4+uv_1(v^2+v_1v_2)v_2^4, \\ &\Phi(Q, q_+) =\Phi(q_+, Q) =uv_2^2v^4+uv_1v_2^6, \\ &\Phi(q_-, q_-)= =vv_1^2(v+2v_2)v^4, \\ &\Phi(q_-, q_+)=\Phi(q_+, q_-)=vv_1v_2^6, \\ &\Phi(q_+, q_+) =0; \end{aligned} \tag{7.14}$$

$$G_{0Qq\pm}=\Phi_{Qq\pm}=uv^4(1+v)+v_1v_2^4(v+v_2), \tag{7.15}$$

$$G_{Qq\pm}=2uv^4(1+v)+2v_1v_2^4(v+v_2). \tag{7.16}$$

In conclusion it should be noticed that the *discontinuity* of the sort stated in § 6 of VII appears here again. In fact, we see, from (7.5) and (2.16),

$$[G_{ABO}]^{r=0}-[G_{MN}]^{(s,t)=(p,q)}=-16p^3q^3. \tag{7.17}$$

The deficiency is of course caused by the existence of a recessive gene as explained in detail in § 6 of VII. However, there is no discontinuity between ABO and Q types. In fact, by putting $(p, q, r) = (u, 0, v)$ or $(p, q, r) = (0, u, v)$ in (7.5), one will arrive exactly at (7.12). There is no discontinuity also between $Qq_{\pm}$ and Q types, as seen from (7.12) and (7.16).

8. Maximizing distributions.

We shall now proceed to determine, the distribution of genes which maximizes the respective probability derived in the preceding sections.

First, in case of *MN blood type*, by differentiating G_{MN} given in (2.16) with respect to a variable st, we see that the derivative

$$dG_{MN}/d(st) = 4((1-4st)(1-3st) + s^2t^2(3-2st))$$

remains positive throughout the interval $0 \leqq st \leqq 1/4$. Hence, G_{MN} is maximized, as usual, at the distribution

$$(8.1) \qquad s = t = 1/2; \quad \overline{M} = \overline{N} = 1/4, \quad \overline{MN} = 1/2;$$

the maximum value being, as already shown by Wiener[2],

$$(8.2) \qquad (G_{MN})^{\max} = 55/128 = 0.4297.$$

The whole probability (5.41) for *general case* attains a stationary value

$$(8.3) \qquad (G)^{\text{stat}} = \left(1 - \frac{1}{m}\right)\left(1 + \frac{1}{m} - \frac{2}{m^2} - \frac{4}{m^3} - \frac{12}{m^4} + \frac{32}{m^5} - \frac{9}{m^6}\right)$$

at the symmetric distribution

$$(8.4) \qquad p_i = 1/m \qquad (i = 1, \ldots, m),$$

which will perhaps be the actual maximizing one. The value (8.3) increases with m and tends to 1 as $m \to \infty$. In fact, differentiating $(G)^{\text{stat}}$ given in (8.3) with respect to $1/m$, regarded as if a continuous variable, we get

$$\frac{d}{d(1/m)}(G)^{\text{stat}} = -\left(1 - \frac{2}{m}\right)^2\left(\frac{4}{m} + \frac{25}{m^2} + \frac{116}{m^3} + \frac{144}{m^4}\right) - \left(1 - \frac{2}{m}\right)\frac{358}{m^5} - \frac{77}{m^6},$$

which remains negative for $m \geqq 2$.

Next, in case of *ABO blood type*, the probability G_{ABO} given in (7.5) may be regarded, in view of an identity $p+q+r=1$, as a function of two independent variables p and q. Then, the system of equa-

2) A. S. Wiener, Chances of detecting interchange of infants, with special reference to blood groups. Zeitschr. f. ind. Abstam.-u. Vererbungslehre. **59** (1931), 229-235.

tions $\partial G_{ABO}/\partial p=\partial G_{ABO}/\partial q=0$ will imply the maximizing distribution. It is given by

$$(8.5)\qquad \begin{aligned} &p=q=0.2036, \qquad r=0.5928;\\ &\bar{O}=0.3514,\quad \bar{A}=\bar{B}=0.2828,\quad \overline{AB}=0.0830 \end{aligned}$$

the value of p and q in (8.5) is a root of the equation of degree seven:

$$(8.6)\qquad 34x^7-154x^6+285x^5-320x^4+193x^3-63x^2+12x-1=0.$$

The maximum value is equal to

$$(8.7)\qquad (G_{ABO})^{\max}=0.5482,$$

a value which has been announced in a paper of Wiener[3].

In case of *Q blood type*, we get, by differentiating G_Q given in (7.12) with respect to $v=1-u$, the derivative

$$dG_Q/dv=4v^3(2-3v^2)$$

which vanishes for $v=\sqrt{2/3}$. Hence, the maximizing distribution is given by

$$(8.8)\qquad \begin{aligned} &u=1-\sqrt{2/3}=0.1723, \qquad v=\sqrt{2/3}=0.8277;\\ &\bar{Q}=1/3=0.3333, \qquad \bar{q}=2/3=0.6667; \end{aligned}$$

the maximum value being, as also already shown by Wiener[4],

$$(8.9)\qquad (G_Q)^{\max}=8/27=0.2963.$$

Finally, the probability (7.16) for $Qq_\pm$ *blood type* may be regarded as a function of two independent variables v and $v_2(=v-v_1)$:

$$G_{Qq\pm}=2v^4(1-v^2)+2v_2^4(v^2-v_2^2).$$

Differentiation with respect to each of these variables leads to a system of equations for determining the maximing distribution stating

$$0=\partial G_{Qq\pm}/\partial v=4v(2v^2-3v^4+v_2^4),$$

$$0=\partial G_{Qq\pm}/\partial v_2=4v_2^3(2v^2-3v_2^2).$$

Since the case $v=0$ or $v_2=0$ is inadequate for maximization, we get from this system the maximizing distribution:

$$(8.10)\qquad \begin{aligned} &u=1-3\sqrt{2/23}=0.1154, \qquad v=3\sqrt{2/23}=0.8846\\ &v_1=3\sqrt{2/23}-2\sqrt{3/23}=0.0480, \quad v_2=2\sqrt{3/23}=0.8366;\\ &\bar{Q}=5/23=0.2174,\quad \bar{q}_-=6/23=0.2609,\quad \bar{q}_+=12/23=0.5217; \end{aligned}$$

the maximum value being

$$(8.11)\qquad (G_{Qq\pm})^{\max}=6^3/23^2=216/529=0.4083.$$

3) A. S. Wiener, loc. cit. 1)

4) A. S. Wiener, loc. cit. 2)