## 61. Probability-theoretic Investigations on Inheritance. X<sub>3</sub>. Non-Paternity Concerning Mother-Child-Child Combinations.

## By Yûsaku Komatu.

Department of Mathematics, Tokyo Institute of Technology and Department of Legal Medicine, Tokyo Medical and Dental University. (Comm. by T. FURUHATA, M.J.A., May 13, 1952.)

## 5. Illustrative examples, recessive genes being existent.

General discussions developed in the preceding sections have exclusively concerned genotypes. If, however, phenotypes containing recessive genes are taken as basic unit, the circumstances will become somewhat different. As frequently mentioned, if recessive genes are existent, genotype of an individual cannot necessarily be determined from its phenotype in a unique manner. Corresponding to the fact that a directly observable character is a phenotype, probabilities a posteriori of possible types of father against every given mother-child combination are also to be determined based upon phenotypes, what will be illustrated by various human blood types.

We first consider ABO blood type. Now, given a mother-child combination (O; O), and types except O alone may be possible as type of father, of which the probabilities a priori for O, A, B have to be taken as

$$\bar{O}=r^2$$
,  $\bar{A}=p(p+2r)$ ,  $\bar{B}=q(q+2r)$ ,

respectively. On the other hand, the mating  $O \times O$ ,  $A \times O$ ,  $B \times O$ , orders being taken into account, produce a child O with probabilities

$$1, \qquad \frac{r}{p+2r}, \qquad \frac{r}{q+2r},$$

respectively. Hence, due to Bayes' theorem, probabilities a posteriori of types of father being O, A, B are respectively given by

$$Z(O, O; O) = 1 \cdot \overline{O} / \left( 1 \cdot \overline{O} + \frac{r}{p+2r} \cdot \overline{A} + \frac{r}{q+2r} \cdot \overline{B} \right) = r,$$
  

$$Z(A, O; O) = \frac{r}{p+2r} \cdot \overline{A} / \left( 1 \cdot \overline{O} + \frac{r}{p+2r} \cdot \overline{A} + \frac{r}{q+2r} \cdot \overline{B} \right) = p,$$
  

$$Z(B, O; O) = \frac{r}{q+2r} \cdot \overline{B} / \left( 1 \cdot \overline{O} + \frac{r}{p+2r} \cdot \overline{A} + \frac{r}{q+2r} \cdot \overline{B} \right) = q.$$

In similar ways, the remaining probabilities a posteriori will be determined. The results, together with those similarly obtained on Q and  $Q_{q\pm}$  blood types, are tabulated as follows. It is to be noticed that the results on  $Qq_{\pm}$  blood type reduce essentially to those on Q blood type by putting  $(v_1, v_2) = (v, 0)$  or  $(v_1, v_2) = (0, v)$  and unifying  $q_-$  and  $q_+$  into q.

Mother	Father Child	0	A	В	AB
	0	r	p	q	0
0	A	0	$p\!+\!r$	0	q
	В	0	0	$q\!+\!r$	p
	0	r	p	q	0
A	A	$rac{r^2(p\!+\!r)}{p^2\!+\!3pr\!+\!r^2}$	$rac{p(p\!+\!r)(p\!+\!3r)}{p^2\!+\!3pr\!+\!r^2}$	$rac{qr(p\!+\!r)}{p^2\!+\!3pr\!+\!r^2}$	$rac{pq(p+2r)}{p^2+3pr+r^2}$
	В	0	0	$q\!+\!r$	p
	AB	0	0	$q\!+\!r$	p
	0	r	p	q	0
	A	0	p+r	0	q
В	В	$rac{r^2(q+r)}{q^2+3qr+r^2}$	$rac{pr(q+r)}{q^2+3qr+r^2}$	$rac{q(q+r)(q+3r)}{q^2+3qr+r^2}$	$rac{pq(q+2r)}{q^2+3qr+r^2}$
	AB	Ō	p+r	0	q
	A	$\frac{r^2}{p+r}$	$rac{p(p+2r)}{p+r}$	$\frac{qr}{p+r}$	$rac{pq}{p+r}$
AB	В	$rac{p+r}{rac{r^2}{q+r}}$	$\frac{pr}{q+r}$	$rac{q(q+2r)}{q+r}$	$rac{pq}{q+r}$
	AB	0	$\frac{p(p+r)}{p+q}$	$rac{q(q+r)}{p+q}$	$rac{2pq}{p+q}$
			Fa-		

				Mother	Fa- ther Child	Q	<i>q</i> -	<i>q</i> +
Mother		Q	q	Q	Q	$\frac{u(1+2v)}{1+uv}$	$\frac{v_1(v+v_2)}{1+uv}\\v^2+v_1v_2$	$\frac{{v_2}^2}{1+uv}\\ {v_2}^2$
Q	Child Q	$\frac{u(1+2v)}{1+uv}$	$\left  \frac{v^2}{1+uv} \right $	-	q q+	u	$\overline{\begin{array}{c}v+v_2\\v_1\end{array}}$	$\overline{v+v_2} \ v_2$
8	q	u	v		Q	1	0	0
q	Q	1	0	$q_{-}$	<i>q</i> _	u	$rac{vv_1(v+2v_2)}{v^2+v_1v_2}$	$rac{vv_2^2}{v^2\!+\!v_1v_2}$
А	q	u	v		$q_+$	u	$v_1$	$v_2$
					Q	1	0	0
				$q_+$	<i>q</i> _	u	v	0
					<i>q</i> +	u	$v_1$	$v_2$

Making use of the thus determined probabilities a posteriori, we can deduce the results in these concrete cases, constructing the following tables; probabilities corresponding to (4.2) as well as to (4.21) are listed.

Mother	2nd child 1st child	0	V	B	AB	Part. prob. w. r. t. mother and 1st child
	0	0	$pr^{3}(q+r)$	$qn^3(p+r)$	l	$r^{3}(pr+qr+2pq)$
0	A	$pqr^3$	0	$pqr^2(p+r)$	I	$pqr^2(p+2r)$
	В	$pqn^3$	$pqr^2(q+r)$	0	1	$p \bar{q} r^2(q+2r)$
	0	0	0	$rac{pqr^3(p+r)}{n+2r}$	$\frac{pqr^2(p+r)^2}{p+2r}$	$pqr^2(p+r)$
	Ą	$p^2qr^2$	0	$\frac{pqr(p+r)(p^2+3pr+r^2)}{n+2r}$	$rac{pq(p+r)^2(p^2+3pr+r^2)}{n+2r}$	$iggl\{ pq(p+r)(p^2+3pr+r^2)\ +p^2qr^2$
${V}$	В	$\frac{p^2qr^3}{p+2r}$	0	0	0	$rac{p^2qr^3}{p+2r}$
	AB	$\frac{p^2qr^2(p+r)}{p+2r}$	0	0	0	$\frac{p^2qr^2(p+r)}{p+2r}$
	0	0	$rac{pqr^{3}(q+r)}{q+2r}$	0	$\frac{pqr^2(q+r)^2}{q+2r}$	$p \bar{q} r^2 (\bar{q} + r)$
	V	$\frac{pq^2r^3}{\alpha+9m}$	0	0	0	$\frac{pq^2r^3}{q+2r}$
B	В	$pq^{2r^2}$	$\frac{pqr(q+r)(q^2+3qr+r^2)}{q+2r}$	0	$\frac{pq(q+r)^{2}(q^{2}+3qr+r^{2})}{q+2r}$	$\left\{ p  {\displaystyle {{{\bar q}}}  {\left( {{q + r}}  ight)} \left( {{{\bar q}}^2 + {3{{\bar q}}r + {r^2}}}  ight)} + {p  {{\bar q}}^{2{r^2}}}  ight.$
	AB	$\frac{pq^2r^2(q+r)}{q+2r}$	0	0	0	$\frac{pg^2r^2(q+r)}{q+2r}$
	A	1	0	0	$rac{1}{2}pqr^2(p+q)$	$\frac{1}{2}pqr^2(p+q)$
AB	В	l	0	0	$\frac{1}{2}pqr^{2}(p+q)$	$rac{1}{2}p q r^2(p+q)$
	AB	1	0	0	0	0
		_	_	-	$\frac{1}{L_{AB0} = p(1 - 1)}$	$L_{AB0} = p(1-p)^{\frac{1}{4}} + q(1-q)^{\frac{1}{4}} + pqn^{2}(3-r)$

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_	Mother	2nd child 1st child	Q	ą	l n	Part. p nother	rob. w. r. t. and 1st child
	0	Q	0	0			0
	Q	q	0	0			0
q		Q	0	0			0
		q	$uv^4$	0	$uv^4$		$uv^4$
		1	1		1	$L_Q$ =	$=uv^4$
lother	2nd c 1st child	hild	Q		<i>q</i> _	<i>q</i> +	Part. prob. w. r. t. mother and 1st child
	Q		0		0	0	0
Q	<i>q</i> _		0		0	0	0
	$q_+$		0		0	0	0
	Q		0		0	0	0
$q_{-}$	<i>q</i> _	$uvv_1$	$(v^2 + v_1 v_2)$		0	0	$u \pi v_1 (v^2 + v_1 v_2)$
	<i>a</i> +	11	$nn_1n_2^2$		0	0	$uvv_1v_2^2$

Mother	Ist child	Q	<i>q</i> _	$q_+$	Part. prob. w. r. t. mother and 1st child
	Q	0	0	0	0
Q	$q_{-}$	0	0	0	0
	$q_+$	0	0	0	0
	Q	0	0	0	0
q	<i>q</i> -	$uvv_1(v^2+v_1v_2)$	0	0	$u \pi v_1 (v^2 + v_1 v_2)$
	$q_+$	$uvv_1v_2^2$	0	0	$uvv_1v_2^2$
	Q	0	0	0	0
$q_+$	$q_{-}$	$uvv_1v_2^2$	0	0	$uvv_1v_2^2$
	$q_+$	$uvv_2{}^3$	$v_1 v_2^4$	0	$(uv+v_1v_2)v_2^3$
	, ,		,	1	$L_{Qq\pm} = uv^4 + v_1v_2^4$

It will be noticed, as really seen from the tables, that, corresponding to (4.33), every total probability coincides with the one for one-child case also when recessive genes are existent; that is,

 $L_{AB0} = P_{AB0}, \quad L_{Qq\pm} = P_{Qq\pm}, \quad L_{Q} = P_{Q}.$ 

## 6. Maximizing distributions for $J_0$ .

The distribution maximizing the probability  $J_0$  obtained in §2 will be determined by a similar procedure as in the preceding chapters.

The probability in case of MN blood type, given in (2.17), i.e.,

$$(6.1) J_{0MN} = s^2 t^2 (2 - 3st)$$

is regarded as a function of the product st alone, ranging over  $0 \leq st \leq 1/4$ . The derivative

(6.2) 
$$dJ_{0MN}/d(st) = st(4-9st)$$

remains positive for 0 < st < 1/4, the maximizing distribution is attained if and only if st=1/4 and hence

(6.3) 
$$s=t=1/2, \quad \overline{M}=\overline{N}=1/4, \quad \overline{MN}=1/2;$$

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the maximum being

$$(6.4) \qquad (J_{0MN})^{\max} = 5/64 = 0.0781.$$

In case of general result (2.16), the stationary value

(6.5) 
$$(J_0)^{\text{stat}} = \left(1 - \frac{1}{m}\right) \left(1 - \frac{3}{m} - \frac{1}{m^2} + \frac{15}{m^3} - \frac{31}{2m^4}\right),$$

attained by symmetric distribution

(6.6)  $p_i = 1/m$  (i = 1, ..., m),

will probably be the actual maximum.

In case of ABO blood type, the probability given in (2.18), i.e.,

(6.7) 
$$J_{0ABO} = p^2 (1-p)^4 + q^2 (1-q)^4 + \frac{1}{2} p q r^2 (1+7r^2),$$

is regarded as a function of p and q  $(r \equiv 1-p-q)$ . The system of equations  $\partial J_{0ABO}/\partial p = \partial J_{0ABO}/\partial q = 0$  yields the maximizing distribution

(6.8) 
$$p=q=0.2481, r=0.5038;$$
  
 $\overline{O}=0.2538, \overline{A}=\overline{B}=0.3116, \overline{AB}=0.1230;$ 

the common value of p and q is determined as a root quartic equation

$$(6.9) 174x^4 - 300x^3 + 196x^2 - 57x + 6 = 0.$$

The maximum of (6.7) corresponding to (6.8) is equal to

$$(6.10) (J_{0ABO})^{\max} = 0.0610.$$

In case of Q blood type, we get similarly for (2.19), i.e.,

$$(6.11) J_{0Q} = u^2 v^4;$$

the maximizing distribution and the maximum:

(6.12) 
$$u=1/3, v=2/3; \overline{Q}=5/9, \overline{q}=4/9;$$

$$(6.13) (J_{0Q})^{\max} = 16/729 = 0.0219.$$

In case of  $Qq_{\pm}$  blood type, the probability given in (2.20), i.e.,

$$(6.14) J_{00\sigma_{\pm}} = u^2 v^4 + (2u + v_1) v_1 v_2^4$$

can be regarded as a function of v and  $v_2$  (u=1-v,  $v_1=v-v_2$ ). The system of equations determining maximizing distribution becomes

(6.15) 
$$0 = \partial J_{0Qq\pm} / \partial v = 2(1-v)(v^3(2-3v)+v_2^4), \\ 0 = \partial J_{0Qq\pm} / \partial v_2 = 2v_2^3(2v(2-v)-v_2(5-3v_2));$$

the roots satisfying  $0 < v_2 < v < 1$  being desired. The system (6.15) is written in an equivalent form

(6.16) 
$$v_2 = \frac{v(100 - 98v - 6v^2 + 69v^3)}{5(25 - 24v + 12v^2)}, \quad 2v(2 - v) = v_2(5 - 3v_2).$$

By eliminating  $v_2$ , an equation of degree seven with respect to v will be obtained. But, the detailed discussion will be omitted. We notice here, remembering (6.12), only an estimation stating that

$$(6.17) \qquad (J_{0Qq\pm})^{\max} \ge (J_{0Q})^{\max} + \underset{0 \le v_2 \le 2^{1/3}}{\max} \left(2 \cdot \frac{1}{3} + \frac{2}{3} - v_2\right) \left(\frac{2}{3} - v_2\right) v_2^4 \\ = \frac{16}{729} + \left(\frac{4}{3} - \frac{15 - \sqrt{33}}{18}\right) \left(\frac{2}{3} - \frac{15 - \sqrt{33}}{18}\right) \left(\frac{15 - \sqrt{33}}{18}\right)^4 \\ = 0.0315.$$