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Diagnosis of Paternity for Cases Without Mother and Without Both Mother and Putative Father Based on Blood Group Findings from the Relatives

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Summary. Formulas of the estimated likelihood ratio Y/X are derived for cases without mother as well as those without both mother and putative father, by using blood group findings of their relatives.

The distribution curves of the relative frequencies of log(Y/X) for these cases are calculated with respect to 10^4 families which are created by a Monte Carlo simulation. The extent of success in the paternity diagnosis is clarified by the statistical analysis based on these distribution curves.

According to the above analysis, fairly high chance of success can be obtained in the diagnosis of such ambiguous cases without the plaintive mother and/or the putative father, if their relatives are alive. It is also concluded that the genetic information as to the parents of the deceased person increases the exclusion probability, whereas that as to the spouse and children increases the fraction of $\log(Y/X) > 1$ for non-father, corresponding to the fraction where the Essen-Möller value is less than 9%.

Key words: Estimation of paternity likelihood ratio, probable genotype of deceased person – Paternity, blood group findings of relatives

Zusammenfassung. Für Fälle, in denen die Mutter oder in denen die Mutter und der Putativ-Vater verstorben sind, werden Formeln für die Berechnung des Likelihood-Quotienten Y/X aus den Blutgruppenbefunden der Verwandten abgeleitet.

Die Verteilungskurven der relativen Häufigkeiten der Funktion log(Y/X) werden anhand von 10^4 Fällen mit der Monte-Carlo-Methode errechnet. Durch statistische Analyse der Verteilungskurven läßt sich die Erfolgschance bei der Vaterschaftsbestimmung abschätzen. Nach dieser Analyse gibt es eine recht hohe Erfolgschance in Fällen, in denen Mutter und Putativ-Vater verstorben sind, wenn die Verwandten noch leben. Es kann weiterhin daraus entnommen werden, daß genetische Informationen über die Eltern der verstorbenen Person die Ausschlußwahrscheinlichkeit erhöhen, während Daten

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über Mutter oder Vater und Kinder den Teil der Funktion $\log(Y/X) > 1$ für Nicht-Väter vergrößern, der einem Essen-Möller-Wert kleiner als 9% entspricht.

Schlüsselwörter: Vaterschaft, bei Tod der Mutter und des Putativ-Vaters – Monte-Carlo-Methode, Likelihood-Quotient aus Befunden der Verwandten

In the diagnosis of paternity for cases without the plaintive mother, the quantities of X and Y are used conventionally for the calculation of Essen-Möller values of paternity probability, where X is the frequency of father-child combination for the certain plaintive child, and Y for the frequency of the corresponding blood type of the putative father [1, 2, 3]. The deceased plaintive mother is considered as one of general person in the population.

In such cases, however, if the genotype of the deceased mother can be deduced from her relatives, then the diagnosis of paternity can be performed with more certainty than that by the conventional method described above. Therefore, in the present paper the formulas of the likelihood ratio in these cases are derived by deducing the genotype of the decreased plaintive mother. This method is also applied to more ambiguous cases where both the putative father and the plaintive mother are deceased, but their relatives are alive.

Deduction of the Probable Genotype of the Deceased Person

If someone has deceased, his genotype cannot be singled out from his relatives in most cases. His probable genotype may contain all genotypes as its components. The probability of occurrence of each genotypes Ge-i is expressed by π_i ($i = 1, \ldots, n$), where n is assumed to be the number of genotypes in the system. The method of deducing the probable genotypes of the decreased person, same as explained in the previous report [4], is briefly described here.

Let us denote the blood type (i.e., phenotype or genotype) of father, mother, and k children in a pedigree (Fig. 1) to be F, M, and $C_1, \ldots, C_k$, respectively, and parentheses ( ) to be the frequency of a blood type or the frequency of combination: (F) expresses the frequency of father in the general population and (F, M, C), the frequency of father-mother-child combination [5]. Relations between the deceased person and his relatives are expressed by using various combination frequencies. For example, if the deceased person is the father who is supposed to have a genotype Ge-i, and his relatives are his wife and children in a pedigree, this relation is expressed by the combination frequency (Ge-i, M, $C_1, \ldots, C_k$), where Ge-i is one of the genotypes in the system ($1 \leq i \leq n$). The probability π_i that the deceased person may possess Ge-i is calculated by using the following equation,

$$\pi_i = \frac{\text{combination frequency of Ge-i and the phenotype of the relatives}}{\sum_{i=1}^{n} \text{combination frequency of Ge-i and the phenotypes of the relatives}} \quad \ldots \text{Eq. (1)}$$

where the following relations must hold

$$0 \leq \pi_i \leq 1 \quad \text{and} \quad \sum_{i=1}^{n} \pi_i = 1.$$

If independent information of two kinds are available from the relatives, the probability π_i that the deceased person may possess Ge-i is proportional to the product of the two independent probabilities [4].

In this way, the probable genotype of the deceased person (deceased putative father, deceased plaintive mother) can be expressed as a linear combination of all the genotypes in the system and used for the calculation of likelihood ratio in the following section.

Derivation of the Likelihood Ratio

In this section F, M, and C represent the blood type of the putative father, plaintive mother and child, respectively, when they are alive; π_i and π_j represent the probabilities that the genotype of the putative father is Ge-i and the genotype of the plaintive mother is Ge-j, respectively, when they are deceased.

Let us define Py and Px as follows:

Px is the probability that children of the blood type C are born from parents of the blood types F and M;

Py is the probability that children of the blood type C are born from mothers of the blood type M and general men in the normal population.

$$\text{Px} = \frac{(\text{F, M, C})}{(\text{F})(\text{M})} \qquad \text{Py} = \frac{(\text{M, C})}{(\text{M})} \qquad \ldots \text{Eq. (2)}$$

Group 1. Estimated Likelihood Ratio in Cases Without Mother on the Basis of Blood Group Findings from her Relatives

When the probable genotype of the deceased mother (i.e., π_j) is deduced from the phenotypes of her relatives, the Px and Py are derived as follows

$$\text{Px} = \sum_{j=1}^{n} \pi_j \frac{(\text{F, Ge-j, C})}{(\text{F})(\text{Ge-j})} \qquad \text{Py} = \sum_{j=1}^{n} \pi_j \frac{(\text{Ge-j, C})}{(\text{Ge-j})} \qquad \ldots \text{Eq. (3)}$$

As a special case, if no information is available on the relatives of the deceased plaintive mother, the mother is considered as one of persons in the general population. Therefore, the probability that the mother has the genotype Ge-j is equal to the frequency of the genotype Ge-j in the system ($\pi_j = (\text{Ge-j}), 0 \leq j \leq n$). In this case Py/Px becomes as follows

$$\text{Py/Px} = \sum_{j=1}^{n} (\text{Ge-j})(\text{Ge-j, C})/(\text{Ge-j}) \Big/ \sum_{j=1}^{n} (\text{Ge-j})(\text{F, Ge-j, C})/\{(\text{F})(\text{Ge-j})\}$$

$$= \frac{(\text{C})}{(\text{F, C})/(\text{F})} = \frac{(\text{F})}{(\text{F, C})/(\text{C})} = \text{Y/X} \qquad \ldots \text{Eq. (4)}$$

where X is the frequency of father-child combination for the certain plaintive child and Y is the frequency of the corresponding phenotype of the putative

father defined by Essen-Möller. According to Eq. (4), the ratio Py/Px is the same as the ratio Y/X.

Group 2. Estimated Likelihood Ratio in Cases Without Both Putative Father and Plaintive Mother on the Basis of Blood Group Findings from Respective Relatives

When both the putative father and the plaintive mother have deceased, their probable genotypes (i.e., π_i and π_j) are deduced from respective relatives, and Px and Py are derived as follows,

$$\mathrm{Px} = \sum_{i,j=1}^{n} \pi_i \pi_j \frac{(\mathrm{Ge\text{-}i},\ \mathrm{Ge\text{-}j},\ \mathrm{C})}{(\mathrm{Ge\text{-}i})(\mathrm{Ge\text{-}j})} \qquad \mathrm{Py} = \sum_{j=1}^{n} \pi_j \frac{(\mathrm{Ge\text{-}j},\ \mathrm{C})}{(\mathrm{Ge\text{-}j})} \qquad \ldots \mathrm{Eq.}\,(5)$$

Computer Simulation

In the pedigree shown in Fig. 1, the putative father and his relatives are shown in the left side; the plaintive mother and her relatives as well as the plaintive child are shown in the right side. To clarify the statistical tendency of the estimated likelihood ratio, 10^4 pedigrees are created by a Monte Carlo simulation by using Japanese gene frequency [4]. To calculate the distribution of log(Y/X) for *true father,* families containing the putative father, his relatives, the plaintive mother, her relatives, and the plaintive child are created altogether according to the law of inheritance [6]. To calculate the distribution of log(Y/X) for *non-father*, families containing the putative father and his relatives are chosen at random with respect to families containing the plaintive mother, her relatives, and the child.

By using the formulas derived in the previous section, the distribution curves of logarithm of the likelihood ratio [4, 7] are calculated for the cases without mother and/or without putative father based on blood types of their relatives.

The Three Following Cases are Considered in Group 1:

Case (a) the genotype of the plaintive mother is unknown.
Case (b) that of the plaintive mother is deduced from her husband and two children.
Case (c) that of the plaintive mother is deduced from both her parents.

The Six Following Cases are Considered in Group 2:

Case (a) the genotype of putative father is deduced from both his parents, and that of plaintive mother is unknown.
Case (b) that of putative father is deduced from both his parents, and that of plaintive mother is deduced from her husband and two children.
Case (c) that of putative father is deduced from both his parents, and that of plaintive mother is deduced from both her parents.

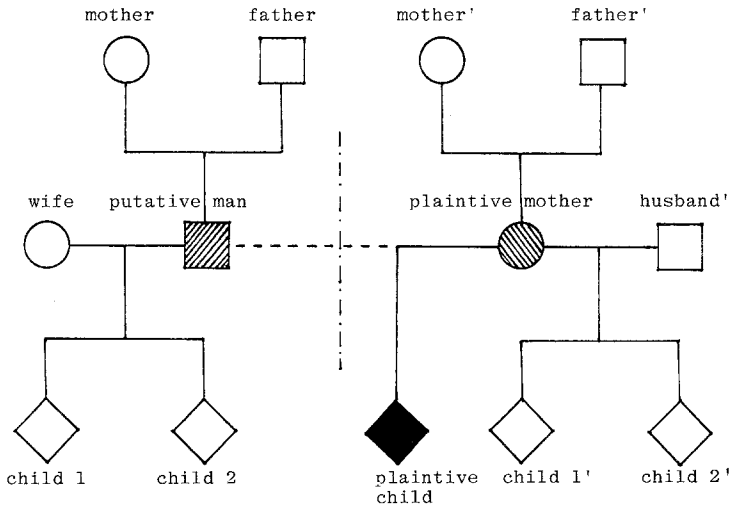

Fig. 1. The pedigree shows the putative father with his relatives in the left side and the plaintive mother with her relatives and the plaintive child in the right side

Case (a)′ that of putative father is deduced from his wife and two children, and that of plaintive mother is unknown.

Case (b)′ that of putative father is deduced from his wife and two children, and that of plaintive mother is deduced from her husband and two children.

Case (c)′ that of putative father is deduced from his wife and two children, and that of plaintive mother is deduced from both her parents.

Fourteen blood group systems are used as in the previous report [4]. Results are shown in Figs. 2—4. The relative frequency of log(Y/X) for *true father* and *non-father* are designated in white circles and black circles, respectively. Those for *non-father* excluded are expressed in black sticks. The mean values, standard deviations of the distributions, the relative area under the curve for log(Y/X) <-2, (or <-1, >1, >2, respectively) and the exclusion probabilities are summarized in Table 1. The corresponding Essen-Möller values of log(Y/X) = -2, -1, 1 and 2 are 99.0, 90.9, 9.1 and 1.0%., respectively. The values -1 and 1 of log(Y/X) are found near the lower limits of such regions of verbal predicates as "likely true father" and "likely non-father", respectively [3, 7].

For convenience's sake let us define verbal predicates "at least likely true father" and "at least likely non-father". That is, the former represents a group of *true father* whose value of log(Y/X) is less than -1, and the latter represents a group of *non-father* whose value of log(Y/X) is more than 1.

In these 14 systems, 85% of *true father* belongs to the categories of "at least likely true father" and the exclusion probability of *non-father* is 93% in usual cases where all the putative father, the plaintive mother and the child are alive [4].

Group 1

Results of the calculation in cases of group 1 are shown in Fig. 2. In case (a) the phenotypes of the relatives of the deceased mother are not available. About 50%

Table 1. Parameters of the distribution curves of log(Y/X)

Group	Case	Mean value in logalithm	Standard deviation	Relative area under the curve				Exclusion probability
				log(Y/X) <−2	<−1	>1	>2	
				%	%	%	%	%
1	(a) True father	−1.08	0.69	9.24	54.10	0.18		
	Non-father	0.02	0.68	0.04	2.00	2.19	0.05	71.39
	(b) True father	−1.37	0.81	21.14	68.46	0.21		
	Non-father	0.36	0.96	0.09	1.97	6.74	1.27	72.92
	(c) True father	−1.46	0.73	22.22	72.94	0.01		
	Non-father	0.16	1.00	0.07	2.12	1.07	0.48	86.38
2	(a) True father	−0.51	0.55	0.37	18.21	0.30		
	Non-father	0.32	0.84		1.16	9.04	2.35	26.73
	(b) True father	−0.68	0.63	1.86	30.69	0.59	0.01	
	Non-father	0.52	0.99	0.01	1.80	16.75	4.25	29.18
	(c) True father	−0.74	0.58	2.20	31.97	0.13		
	Non-father	0.22	0.96	0.01	1.69	5.43	2.54	51.49
	(a)′ True father	−0.55	0.65	1.13	24.08	1.12		
	Non-father	0.53	0.72		1.26	24.33	2.48	4.48
	(b)′ True father	−0.71	0.75	4.26	34.37	1.32	0.05	
	Non-father	0.67	0.80	0.02	1.54	31.49	4.88	5.40
	(c)′ True father	−0.73	0.75	4.31	35.48	1.44	0.01	
	Non-father	0.67	0.82	0.01	1.47	31.51	5.36	8.21

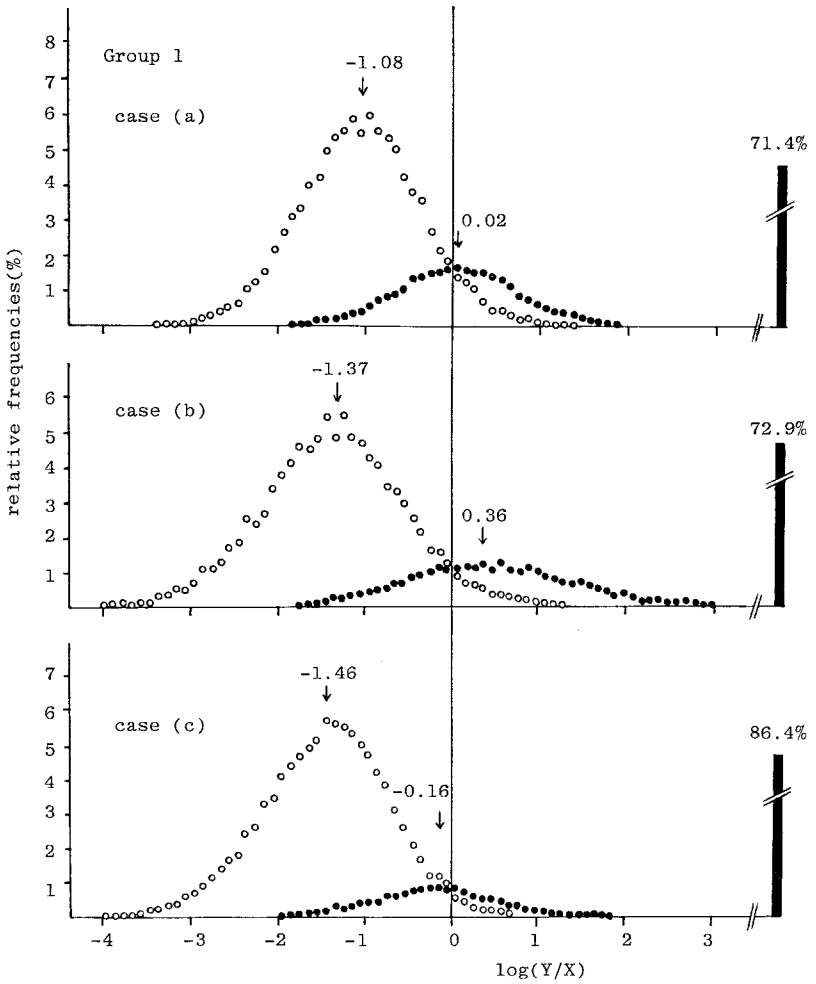

Fig. 2. The distribution curves of log(Y/X) for *true father* and *non-father* in cases without mother. Cases *(a), (b),* and *(c)* in group 1 express situations where the genotype of the mother is *(a):* unknown, *(b):* deduced from her husband and two children, *(c):* deduced from both her parents, respectively. The relative frequencies of log(Y/X) for *true father* and *non-father* are designated in white circles (○) and black circles (●), respectively. Those for *non-father* excluded are expressed separately in black sticks. The arrows indicate the mean positions of the distributions for *true father* and for *non-father* who is not excluded. The class-interval of log(Y/X) is 0.1. The relative frequencies which are greater than 0.001 are drawn in every distributions

of *true father* belongs to the categories "at least likely true father", whereas the exclusion probability of *non-father* is 71% and the fraction of "at least likely non-father" is 2%. The theoretical exclusion probability of this case calculated in terms of algebraic formulas of gene frequencies in these 14 systems is 71%. In cases (b) and (c), the phenotypes of the relatives of the deceased mother are available. About 70% of *true father* belongs to the categories "at least likely true father" and this fraction is appreciably larger than that in case (a). Although the increase in exclusion probability is quite small in case (b) where relatives of deceased mother are her husband and two children, fairly large increase in the

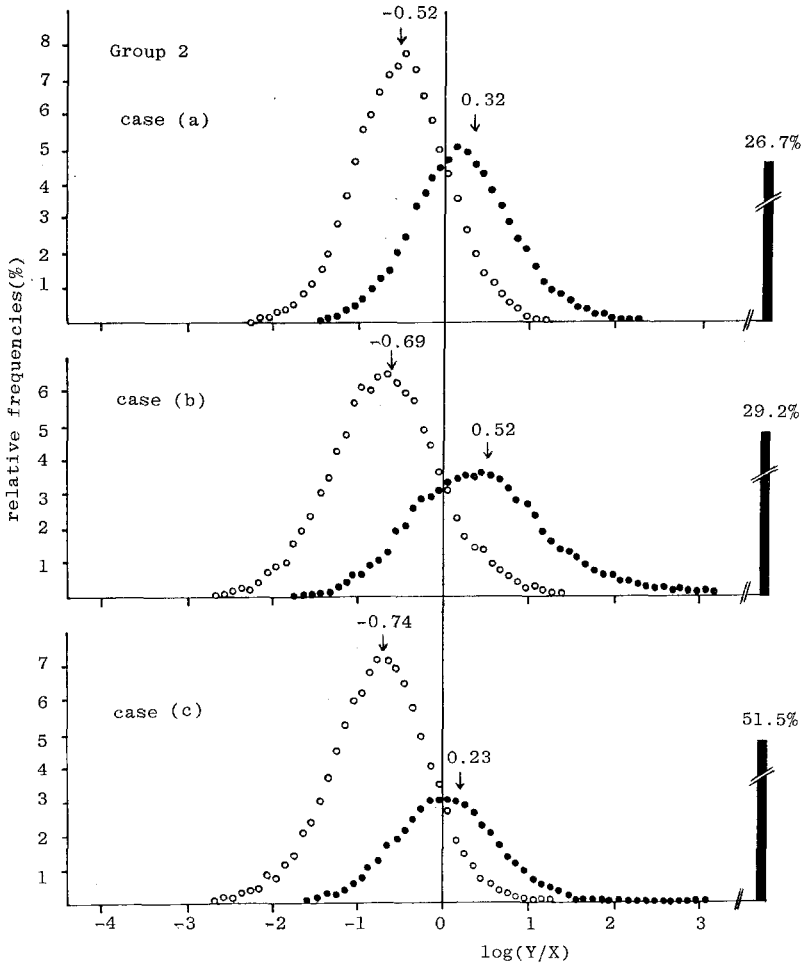

Fig. 3. The distribution curves of log(Y/X) for *true father* and *non-father* in cases without both putative father and plaintive mother. Cases *(a), (b),* and *(c)* in group 2 express situations where the genotype of the putative father is deduced from both his parents, and the genotype of the plaintive mother is *(a):* unknown, *(b):* deduced from her husband and two children, *(c):* deduced from both her parents, respectively

fraction of "at least likely non-father" is observed. Therefore, the sum of exclusion probability and the fraction of "at least likely non-father" amounts up to 80%. In case (c) where the relatives of the deceased mother are both her parents, large increase in exclusion probability is obtained. The sum of the exclusion probability and the fraction of "at least likely non-father" amounts up to 87%.

Group 2

The paternity diagnosis, in cases where both the putative father and the plaintive mother have deceased, becomes more ambiguous. Results of the calculation in cases (a), (b), and (c) are shown in Fig. 3 and cases (a)′, (b)′, and (c)′ are shown in

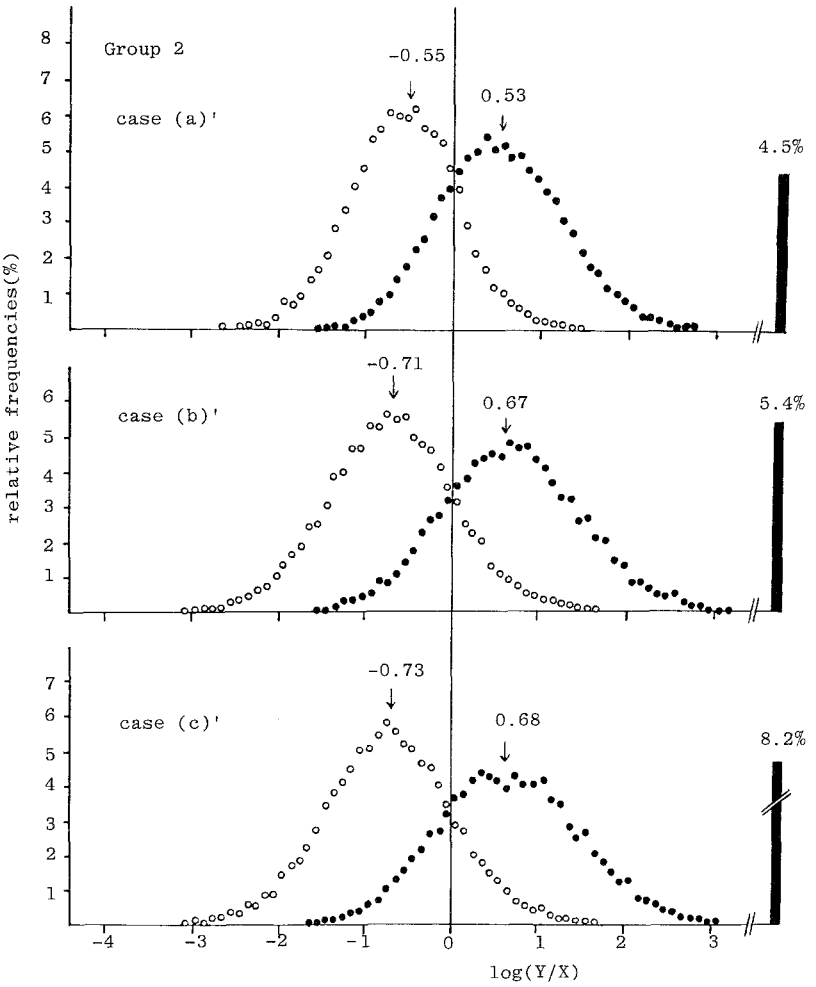

Fig. 4. The distribution curves of log(Y/X) for *true father* and *non-father* in cases without both putative father and plaintive mother. Cases *(a)', (b)',* and *(c)'* in group 2 express situations where the genotype of the putative father is deduced from his wife and two children, and the genotype of the plaintive mother is *(a)':* unknown, *(b)':* deduced from her husband and two children, *(c)':* deduced from both her parents, respectively

Fig. 4, respectively. Here in cases (a) and (a)' the phenotypes of the relatives of the deceased mothers are not available. In cases (a) and (a)' 18% and 24% of *true father* belong to the categories "at least likely true father" and the sums of exclusion probability and the fraction of "at least likely non-father" are 35% and 29%, respectively. In cases (b), (c), (b)' and (c)' 30—35% of *true father* belongs to the categories "at least likely true father" and the sums of the exclusion probability and the fraction of "at least likely non-father" are 37—57%. These results suggest the fairly high possibilities of success of paternity diagnosis in spite of such uncertain cases where both the putative father and the plaintive mother have deceased.

It is concluded that the information from parents increases the exclusion probability, whereas those from spouse and children increases the fraction of "at least likely non-father".

The chance of success is reduced in the paternity diagnosis of cases without the plaintive mother, although the paternity probability can be calculated from the child and the putative father. The diagnosis in such cases becomes more successful by the two following methods. The one is to increase the number of serological tests of the putative father and the child, and the other is to examine the blood types of the relatives of the deceased mother. The serological tests of the relatives can be performed as routine works in addition to those of persons concerned. Therefore, if the blood types of the relatives are available, the method described here serves to promote the paternity diagnosis in cases without plaintive mother and/or putative father.

The computer calculation has been performed on a Facom 230-75 at Nagoya University Computation Center.

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