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# General Formulas of the Estimated Likelihood Ratio Y/X in the Diagnosis of Paternity of a Deceased Putative Father

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**Summary.** When the putative father is dead his probable genotype, essential for estimate of likelihood ratio in the diagnosis of paternity, should be deduced from his relatives. In the present paper are described the general method for such deduction of probable genotype and the derivation of the formula of likelihood ratio. $10^4$ examples of the diagnosis of paternity of deceased father are examined using a Monte Carlo method and the distributions of relative frequencies of log(Y/X) are calculated for the true father and non-father. These results indicate that the present method of estimation of Y/X from relatives is quite useful for the diagnosis of paternity of a deceased putative father.

**Key words:** Estimation of paternity likelihood ratio, deduction of probable genotype – Paternity, deceased putative father

**Zusammenfassung.** Wenn der vermutliche Vater verstorben ist, wird sein wahrscheinlicher Genotyp, der für eine Vaterschaftsbeurteilung notwendig ist, aus den Blutgruppenbefunden seiner Verwandten hergeleitet. In der vorgelegten Arbeit wird eine allgemeine Methode für eine solche Ableitung sowie die Herleitung einer Berechnungsformel des Likelihood-Quotienten Y/X dargelegt. $10^4$ mit der Monte-Carlo-Methode hergestellte Fälle mit verstorbenem vermutlichem Vater wurden in dieser Weise ausgewertet und die Verteilung der relativen Häufigkeit von log(Y/X) für Väter und Nichtväter berechnet. Die Ergebnisse zeigen, daß die hier vorgestellte Methode zur Berechnung von Y/X aus Blutgruppenbefunden von Verwandten für die Vaterschaftsbeurteilung eines verstorbenen vermutlichen Vaters brauchbar ist.

**Schlüsselwörter:** Vaterschaftswahrscheinlichkeit, wahrscheinlicher Genotyp bei Verstorbenen – Monte-Carlo-Methode – Putativvater, Vaterschaftswahrscheinlichkeit bei Tod des

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In the diagnosis of paternity there are some cases where the putative father has died. To calculate the probability of paternity the genotype of the deceased putative father should be estimated from his relatives.

Hummel et al. [1, 2] investigated the probability of paternity of the deceased putative father using the blood group results of his parents, those of his mother and three siblings [3], and those of his wife and children [4]. Nakajima et al. [5, 6] also reported on cases where the phenotypes of the widow and children and those of the two siblings of the deceased father were known. Mayr [7, 8] demonstrated the non-paternity of the deceased father by deducing his HL-A genotype from his relatives.

A general method which is applicable to any cases has been derived by Ihm and Hummel [9, 10]. The method is so designed as to derive "frequency X" and "frequency Y" of the different family trees belonging to the two hypotheses. The results are called plausibility of grandparenthood, of siblingshood and so on, according to the kinds of the relatives.

In our present method which is also applicable to all cases, the blood types of the relatives are used for deducing probable genotype of the deceased father and the deduced genotype is used for the calculation of "probability (Px)" of the deceased putative father himself, where "probability" expresses the proportion that children of a certain blood type are born from parents of given blood types. With our method, the combination frequencies of the putative father and any of the relatives are expressed by simple algebraic formulas for the following two kinds of basic frequencies: (1) frequency of genotype (or phenotype) in the general population, and (2) frequency of father-mother-child combination of genotypes (and/or phenotypes).

We have also investigated the statistical tendency of the estimated likelihood ratio using the distribution curves [11] of log(Y/X) and examined the validity of the estimated likelihood ratio in the diagnosis of paternity.

## Methods of Calculation

The methods of calculation are described for the following situations:
(A) frequency of blood type combination of the putative father and any of his relatives,
(B) deduction of probable genotype of the deceased putative father, and
(C) calculation of the estimated likelihood ratio Y/X.

### (A) *Frequency of Blood Type Combination of the Putative Father and any of his Relatives*

Here, the term blood type is used as for both phenotype and genotype. Following formulas of the combination can be applied to genotypic combinations, phenotypic combinations, and partially genotypic combinations.

(1) *Combination Frequency of Father-Mother-Child, Trio (F, M, C).* Let us denote the blood types of father, mother, and child to be F, M, and C, respectively. If blood types in a polymorphic system are expressed as Ty-1, Ty-2, ..., Ty-n, then each F, M, or C is expressed as one of these blood types. Here, $n$ denotes the number of blood types in the system, and parentheses ( ) represent the frequency of corresponding blood type or the frequency of blood type combination. For example, (F) represents the blood type frequency of father in the general

population, and (F, M, C), the blood type frequency of father-mother-child combination, trio. To calculate any kinds of combination frequency of the putative father and any of the relatives, all the blood type frequencies of trios should be provided.

In many traditional reports, the calculation of blood type combination of trios is not always easy [12]. Recently, however, a very simple method has been proposed by Asano et al. to calculate blood type frequency of trio using computer method, even if the modes of inheritance are as complicated as HL-A and Rh [13].

(2) *Combination Frequency of Father, Mother, and k Children* (F, M, $C_l, \ldots, C_k$). In the case of $k$ children, the blood type of each child is denoted as $C_l, \ldots, C_j, \ldots, C_k$. The combination frequency of father, mother and $k$ children (F, M, $C_l, \ldots, C_j, \ldots, C_k$) is defined by the combination frequency of each trio (F, M, $C_j$) and the blood type frequencies of father (F) and mother (M).

$$(\mathrm{F}, \mathrm{M}, C_l, \ldots, C_k) = (\mathrm{F}) \times (\mathrm{M}) \times \prod_{j=1}^{k} \frac{(\mathrm{F}, \mathrm{M}, C_j)}{(\mathrm{F}) \times (\mathrm{M})} \quad \ldots\ldots(1)$$

The product extends to all children.

(3) *Combination Frequency of Mother (or Father) and k Children* (M, $C_l, \ldots, C_k$) [or (F, $C_l, \ldots, C_k$)]. The combination frequency of mother (or father) and $k$ children is given as the sum of the combination frequencies of father-mother-$k$ children over all blood types of fathers (or mothers).

$$(\mathrm{M}, C_l, \ldots, C_k) = \sum_{F=1}^{n} (\mathrm{F}, \mathrm{M}, C_l, \ldots, C_k) \quad \ldots\ldots(2)$$

$$(\mathrm{F}, C_l, \ldots, C_k) = \sum_{M=1}^{n} (\mathrm{F}, \mathrm{M}, C_l, \ldots, C_k) \quad \ldots\ldots(2)'$$

(4) *Combination Frequency of k Children* ($C_l, \ldots, C_k$). The combination frequency of $k$ children ($C_l, \ldots, C_k$) is given as the sum of the combinations of mother and $k$ children over all blood types of mothers.

$$(C_l, \ldots, C_k) = \sum_{M=1}^{n} (\mathrm{M}, C_l, \ldots, C_k) \quad \ldots\ldots(3)$$

In this way can be calculated various kinds of combination frequencies of the putative father and the relatives in a pedigree, using the basic combination of the trio (F, M, C).

(B) *Deduction of Probable Genotype of the Deceased Putative Father*

Although the blood type of the putative father is confined to one blood type in a system when he is alive, the blood type of the deceased putative father cannot be confined to one in most cases. Therefore, in such cases the probability of each genotype in the system should be estimated from the phenotypes of the relatives. Let $\pi_l, \ldots, \pi_i, \ldots, \pi_n$ represent probabilities that the genotype of the putative father are Ge-1, ..., Ge-i, ..., and Ge-n, respectively ($\sum_{i=1}^{n} \pi_i = 1$, $0 \leq \pi_i \leq 1$). Then the probable genotype of the putative father can be defined as a linear combination of all kinds of genotypes in the system. The probability $\pi_i$ is calculated from the phenotypes of the relatives according to the following equation:

$$\pi_i = \frac{\text{combination frequency of Ge-i and the phenotypes of the relatives}}{\sum_{i=1}^{n} \text{combination frequency of Ge-i and the phenotypes of the relatives}} \quad \ldots\ldots(4)$$

Detailed descriptions are given in the following cases. Let us assume the pedigree written in Fig. 1.

*Case (a).* To illustrate the method for deducing the genotype of the putative father from his wife M and two children $C_1$, $C_2$, let us denote one of the possible genotype of the putative father as Ge-i. The Ge-i is any one of the genotypes in the system. The combination frequency of the putative father and his wife and two children is written as (Ge-i, M, $C_1$, $C_2$). In this pedigree, Ge-i corresponds to the father. The probability that the putative father possesses Ge-i, is written as follows:

$$\pi_i = \frac{(\text{Ge-i, M, } C_1, C_2)}{\sum_{i=1}^{n} (\text{Ge-i, M, } C_1, C_2)} = \frac{(\text{Ge-i, M, } C_1, C_2)}{(\text{M, } C_1, C_2)} \quad \ldots\ldots(5)$$

where the sum extends over all genotypes of father, Ge-i and is equal to the combination frequency of mother and the two children (M, $C_1$, $C_2$).

*Case (b).* To deduce the genotype of the putative father from his parents, F, M, and his brother, $C_1$, the combination frequency of the putative father and his father-mother-brother is written as (F, M, $C_1$, Ge-i). In this pedigree, Ge-i corresponds to $C_2$. The probability that the putative father possesses Ge-i, is written as follows:

$$\pi_i = \frac{(\text{F, M, } C_1, \text{Ge-i})}{\sum_{i=1}^{n} (\text{F, M, } C_1, \text{Ge-i})} = \frac{(\text{F, M, } C_1, \text{Ge-i})}{(\text{F, M, } C_1)} \quad \ldots\ldots(6)$$

where the sum of (F, M, $C_1$, Ge-i) extends over all genotypes of the child, Ge-i, and is equal to the combination frequency of the trio (F, M, $C_1$).

*Case (c).* Estimation from both of his parents, his brother, his wife and his two children [combination of case (a) and case (b)]. This is the case where two kinds of information about the relatives are independent of each other. Let us denote $\pi_{ia}$ the probability that the putative father possesses Ge-i in case (a), and $\pi_{ib}$ the probability that he possesses Ge-i in case (b). The probability $\pi_i$ that the putative father possesses Ge-i is proportional to the product of the two probabilities, $\pi_{ia}$ and $\pi_{ib}$, as a consequence and is written as follows according to Bayes's theorem.

$$\pi_i = \frac{\pi_{ia} \times \pi_{ib}}{\sum_{i=1}^{n} \pi_{ia} \times \pi_{ib}} \quad \ldots\ldots(7)$$

In this way the genotype of the putative father can be estimated according to the principle of equation (4).

(C) *Calculation of the Estimated Likelihood Ratio Y/X*

Let us define the probabilities Px and Py as follows: Px is the probability that children of the phenotype C are born from mothers of the phenotype M and fathers of the genotype Ge-F. Py is the probability that children of the phenotype C are born from mothers of the phenotype M and the general men in the normal population.

$$\text{Px} = \frac{(\text{Ge-F, M, C})}{(\text{Ge-F})(\text{M})} \qquad \text{Py} = \frac{(\text{M, C})}{(\text{M})} \quad \ldots\ldots(8)$$

When the putative father has been dead, his probable genotype should be deduced as described in the preceding section (B) and is expressed as a linear combination of all genotypes in the system. $\pi_i$, the coefficient of each Ge-i, corresponds to the probability that the putative father possesses Ge-i. In such cases, the estimated probabilities Px and Py are derived as follows;

$$\text{Px} = \sum_{i=1}^{n} \pi_i \frac{(\text{Ge-i, M, C})}{(\text{Ge-i})(\text{M})} \qquad \text{Py} = \frac{(\text{M, C})}{(\text{M})} \quad \ldots\ldots(9)$$

Therefore, the estimated likelihood ratio Y/X (i.e., Py/Px) of the deceased putative father is as follows;

$$Y/X = 1 \Big/ \sum_{i=1}^{n} \pi_i \frac{(\text{Ge-i, M, C})}{(\text{Ge-i})(\text{M, C})} = 1 \Big/ \sum_{i=1}^{n} \frac{\pi_i}{[Y/X]_{\text{Ge-i}}} \quad \ldots\ldots(10)$$

When several systems are used, the products of Y/X of each system should be considered.

## Computer Simulation and Discussion

In the pedigree shown in Fig. 1, the relatives of the putative father are written in the left side and the plaintive mother-child couple is written in the right side. To examine the validity of the derived formulas, the estimated values of Y/X are calculated for three cases (a), (b) and (c) described in the previous section and compared with the values of Y/X calculated directly, using the phenotype of the putative father. (If the paternity of a man is denied on the basis of phenotypes of paintive mother-child couple, the value Y/X of the man is designated as $\infty$ and distinguished from others.)

In addition, such calculations are performed for a large number of examples to look into the statistical tendency of the estimated values of Y/X. For instance, $10^4$ pedigree with Japanese gene frequencies (Table 1) are created by a Monte Carlo simulation according to the slightly modified method reported previously [9, 13]. In cases of *true father*, families which contain the putative father and his relatives as well as the plaintive mother-child couple are created and used for the calculation of Y/X. In cases of *non-father*, families including putative father and his relatives are chosen at random with respect to random plaintive mother-child couples. To calculate the values Y/X, frequencies of father-mother-child combinations are stored in the memory of computer. Figure 2 shows the distributions of relative frequency of log(Y/X), which are obtained for *true father*

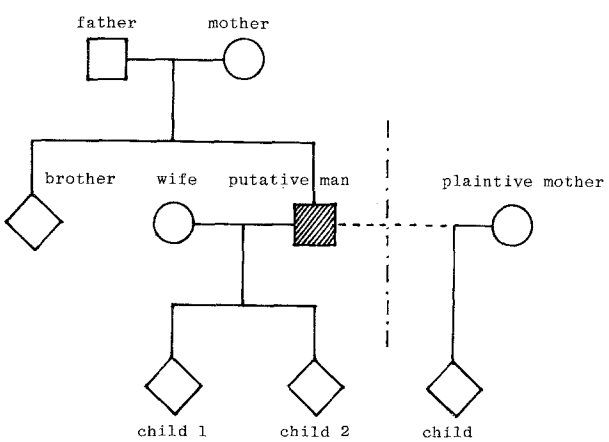


**Fig. 1.** Pedigree. The putative father and his relatives are written in the left side and the plaintive mother-child in the right side. The method of the creation of this pedigree is as follows: using a Monte Carlo method, the genotypes of the genetically independent persons are generated according to the natural occurrence of Japanese population (Table 1) and used for the creation of the genotypes of the genetically decendant persons, and then, the genotypes of living persons are converted into the phenotypes

**Table 1.** Genes and gene frequencies in Japanese population

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| 1 | ABO | O | 0.561 | A | 0.268 | B | 0.171 |
| 2 | MNSs | MS | 0.0321 | Ms | 0.5137 | NS | 0.0165 |
| | | Ns | 0.4377 | | | | |
| 3 | Rh | $R^1$ | 0.6533 | $R^2$ | 0.2556 | r | 0.0375 |
| | | r'' | 0.0332 | $R^o$ | 0.0121 | r' | 0.0062 |
| | | $R^z$ | 0.0014 | $r^y$ | 0.0007 | | |
| 4 | P | P | 0.171 | nonP | 0.829 | | |
| 5 | Duffy | $Fy^a$ | 0.897 | $Fy^b$ | 0.103 | | |
| 6 | Kidd | $JK^a$ | 0.477 | $JK^b$ | 0.523 | | |
| 7 | Gm | $Gm^{1,21}$ | 0.437 | $Gm^{1,16}$ | 0.261 | $Gm^{1,2,21}$ | 0.173 |
| | | $Gm^{1,5}$ | 0.129 | | | | |
| 8 | Km | $Km^1$ | 0.302 | $Km^{non\ 1}$ | 0.698 | | |
| 9 | Gc | $Gc^1$ | 0.752 | $Gc^2$ | 0.248 | | |
| 10 | Hp | $Hp^1$ | 0.263 | $Hp^2$ | 0.737 | | |
| 11 | $PGM_1$ | $PGM_1^1$ | 0.777 | $PGM_1^2$ | 0.223 | | |
| 12 | AcP | $AcP^a$ | 0.213 | $AcP^b$ | 0.787 | | |
| 13 | sGPT | $sGPT^1$ | 0.597 | $sGPT^2$ | 0.403 | | |
| 14 | EsD | $EsD^1$ | 0.650 | $EsD^2$ | 0.350 | | |

and *non-father* [11, 14—17]. Those distributions are calculated using 14 systems written in Table 1. 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 from the phenotypes of mother-child couples are expressed separately in black sticks. The theoretical probability of exclusion is 0.93 in those systems for a random putative father.

Although the relative frequency for *true father* can be calculated theoretically from that for *non-father* (and vice versa), two frequencies are calculated independently to avoid large errors in the region with low relative frequency. The mean values and the standard deviations of the distributions as well as probability of exclusion are summarized in Table 2. The relative area under the distribution curve for $\log(Y/X)<-2$, (or $<-1$, $>1$, $>2$, respectively) is also listed in Table 2. Several features of the distribution observed in Fig. 2 are as follows.

The distribution of *non-father* in the case (a) has appreciably large area under the curve for $\log(Y/X)>1$ and low exclusion probability. On the other hand, the distribution of *non-father* in the case (b) has fairly high exclusion probability. As the information from relatives increase in such a case as (c), the mean of the distribution of the *true father* is shifted toward lower values of log(Y/X). In each case of the distribution of estimated log(Y/X), there are appreciable fractions of putative fathers who can be diagnosed at least as "likely *true father*" [$\log(Y/X)<-1$] or "likely *non-father*" [$\log(Y/X)>1$] [16, 17].

Therefore, the diagnosis of paternity of the deceased putative father can be performed satisfactorily using the estimated likelihood ratio.

**Table 2.** Parameters of the distribution curves of log(Y/X)

| | Mean value in logalithm | Standard deviation | Relative area under the curve | | | | Exclusion probability |
|---|---|---|---|---|---|---|---|
| | | | log(Y/X) <−2 | <−1 | >1 | >2 | |
| | | | % | % | % | % | % |
| (a) True fathers | −0.84 | 0.86 | 8.52 | 42.65 | 1.84 | 0.07 | |
| Non-fathers | 0.82 | 0.96 | 0.08 | 2.26 | 36.85 | 9.93 | 10.4 |
| (b) True fathers | −0.91 | 0.63 | 4.85 | 43.33 | 0.10 | | |
| Non-fathers | 0.14 | 1.07 | 0.07 | 2.61 | 4.18 | 2.19 | 63.5 |
| (c) True fathers | −1.20 | 0.73 | 13.33 | 60.73 | 0.19 | | |
| Non-fathers | 0.46 | 1.69 | 0.12 | 2.68 | 4.51 | 1.29 | 66.8 |
| (d) True fathers | −1.75 | 0.75 | 35.05 | 84.66 | 0.01 | | |
| Non-fathers | −0.58 | 0.76 | 0.14 | 1.74 | 0.22 | 0.05 | 92.6 |

(a), the values of Y/X are estimated from the wife and two children of the putative father; (b), those estimated from both of his parents and his brother; (c), those estimated from both of his parents, his brother, his wife and his two children; (d), those calculated from putative father himself

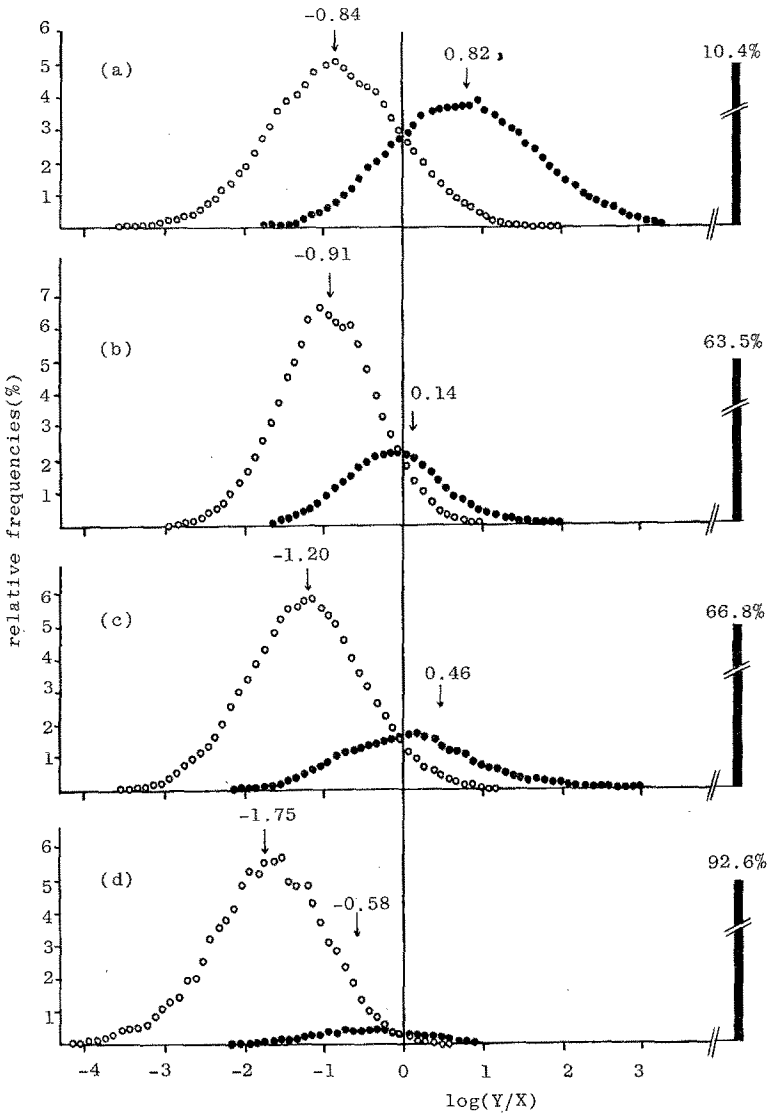


**Fig. 2a–d.** The distribution curves of log(Y/X) for *true father* and *non-father*. **a** The values of Y/X are estimated from the wife and two children of the putative father; **b** those estimated from both of his parents and his brother; **c** those estimated from both of his parents, his brother, his wife and his two children; **d** those calculated from putative father himself. 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 from the phenotypes of mother-child couples 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

Although gene frequencies of Japanese population are used in the calculation, the observed statistical tendency also holds for other populations.

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

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