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Computer Analysis of Parentage in Relation to Maternity and Paternity

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Summary. General formulas for statistical calculations of parentage by means of blood group analysis are presented in relation to those of maternity and paternity. Based on these formulas, a computer program has been devised to calculate plausibilities, exclusion probabilities, and distributions of log *(Y/X)* of parentage for any blood groups. The program also gives numerical values of these quantities of maternity and paternity. The values of plausibility and exclusion probability are highest for parentage, and decrease in the order of paternity and maternity. Concerning the distribution of log *(Y/X)* for true families, a simple relation holds for the ratio of the mean value of log *(Y/X)* in parentage a_p, to that in paternity a_f, and to that in maternity a_m as,

$$a_p : a_f : a_m = 1 : 0.6 : (0.6)^2$$

This relation holds for all 14 blood groups examined.

Key words: Parentage, computer program - Blood groups, statistical calculation of parentage

Zusammenfassung. Es werden allgemeine Formeln zur statistischen Berechnung der Elternschaft auf Grund der Blutgruppenanalyse beschrieben, die in Beziehung zu jenen für Abstammungsprobleme (Mutter und Vater) gesetzt werden. Auf diesen Formeln basierend wurde ein Computerprogramm entwickelt, mit dem die Wahrscheinlichkeit, Ausschlußmöglichkeit und die Verteilungen von log *(Y/X)* der Elternschaft für jede Blutgruppe berechnet werden kann. Das Programm enthält auch numerische Werte zur Quantifizierung der Maternität und Paternität. Die Werte für die Wahrscheinlichkeit und Ausschlußmöglichkeit sind am höchsten für die Elternschaft und nehmen in der Reihenfolge Vater- und Mutterschaft ab. Was die Werte für den log *(Y/X)* für echte Familien angeht, so ist eine einfache Gleichung für das

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Verhältnis des Mittelwertes von log *(Y/X)* für die Elternschaft a_p zu dem Mittelwert für die Vaterschaft a_f und zu dem für die Mutterschaft a_m zutreffend:

$$a_p : a_f : a_m = 1 : 0{,}6 : (0{,}6)^2$$

Diese Gleichung trifft für alle 14 untersuchten Blutgruppen zu.

Schlüsselwörter: Elternschaft, Computerprogramm zur Berechnung – Blutgruppen, Wahrscheinlichkeit und Ausschlußmöglichkeit für Elternschaft

Introduction

In the field of legal medicine, blood group tests are applied most frequently to the problems of disputed paternity with respect to a putative father, a mother, and a child. There are plenty of works on the paternity on the basis of population genetics for various types of blood groups [1–3]. On the other hand, the blood group tests have been less commonly applied to the problems of disputed maternity with respect to a putative mother and a child [4] as well as disputed parentage with respect to a putative father, a putative mother, and a child [5–7], because these two cases are not so frequent as, e.g., kidnapping or accidental interchange of newborn babies. However, these three cases belong to the problems of the same category in population genetics and are mutually related to one another.

Although there are a few works on the exclusion probability of parentage in a simple blood group, such as MN [5–7], there are no works on the plausibility or the exclusion probability of parentage in a complicated blood group, such as Rh. It can be expected that the plausibility and the exclusion probability increase in the order of maternity, paternity, and parentage, corresponding to the increase in the number of exclusive persons included in each case. Quantitative analysis of this tendency, however, has not yet been performed, because there were no general algebraic expressions applicable to any blood groups used commonly.

In this study, formulas for the calculation of parentage as well as maternity are derived in relation to those of paternity. Based on these formulas, a computer program has been devised to calculate plausibilities, exclusion probabilities, and the distributions of log *(Y/X)* for paternity, maternity, and parentage. Then the numerical evaluation of plausibility and exclusion probability for the three cases was carried out by using 14 blood groups, resulting in a simple relation for the ratio of the mean values of log *(Y/X)*. The results of calculation for parentage are applied to two pairs of parents and two babies when the interchange of infants is disputed.

Case Report

Formulas

The following notations are used in this paper [8]. Phenotypes of a father, a mother, and a child are expressed as F, M, and C, respectively. A phenotypic frequency and a frequency of (father)-mother-child combination are given in square brackets. *NP* indicates the number of phenotypes of an arbitrary blood group.

1. Plausibility

1.1 Paternity. The plausibility of paternity, W_f was defined by Essen-Möller [1,2] as

$$W_f = X_f / (X_f + Y_f) \tag{1}$$

where X_f is the frequency that a certain phenotype of father F is found among true fathers for a given mother M and a child C.

$$X_f = [F, M, C] / [M, C] \tag{2}$$

where [F, M, C] and [M, C] are a combination frequency of father-mother-child and that of mother-child, respectively. Y_f is the frequency that a certain phenotype F is found in non-fathers, i.e., in normal male population.

$$Y_f = [F] \tag{3}$$

Therefore the likelihood ratio in paternity is

$$(Y/X)_f = [F] \cdot [M, C] / [F, M, C] \tag{4}$$

1.2 Maternity. The frequencies X_m and Y_m in maternity are defined as in paternity [4]. X_m is the frequency that a certain phenotype of mother M is found in true mothers for a given child C.

$$X_m = [M, C] / [C] \tag{5}$$

Y_m is the frequency that a certain phenotype M is found in non-mothers, i.e., in normal female population.

$$Y_m = [M] \tag{6}$$

Therefore, the likelihood ratio in maternity is

$$(Y/X)_m = [M] \cdot [C] / [M, C] \tag{7}$$

1.3 Parentage. Based on the above definitions (1.1 and 1.2), X_p, Y_p and $(Y/X)_p$ are defined as follows,

$$X_p \quad = [F, M, C] / [C] \tag{8}$$

$$Y_p \quad = [F] \cdot [M] \tag{9}$$

$$(Y/X)_p = [F] \cdot [M] \cdot [C] / [F, M, C] \tag{10}$$

where X_p is the frequency that true parents having their phenotypes F and M are found in true father-mother-child combination within the group where the phenotype of child is fixed as C. Y_p is the frequency of a man with his phenotype F and a woman with her phenotype M to cross.

The above Eqs. 8–10 can also be derived by using the following alternative definition. X_p' is the frequency that true child is found in true father-mother-child combination in the condition that the phenotypes of a father and a mother are F and M, respectively. Y_p' is the frequency that a certain child C is found in normal children.

$$X_p' \quad = [F, M, C] / [F] \cdot [M] \tag{11}$$

$$Y_p' \quad = [C] \tag{12}$$

$$(Y/X)_p = [F] \cdot [M] \cdot [C] / [F, M, C] \tag{13}$$

The derived equation, Eq. 13 is identical with Eq. 10.

2. Exclusion Probability

2.1 Paternity. a) For a certain combination of a mother M and a child C, the exclusion probability $E_f(\mathrm{M},\mathrm{C})$ is calculated by summing up the frequencies of phenotypes of non-fathers who are excluded among all phenotypes, *NP*, in a blood group.

$$E_f(\mathrm{M},\mathrm{C}) = \sum_{F=1}^{NP}{}' \,[\mathrm{F}] \quad \text{with } [\mathrm{F},\mathrm{M},\mathrm{C}] = 0 \tag{14}$$

where Σ' means that the summations are made under the condition, $[\mathrm{F},\mathrm{M},\mathrm{C}] = 0$.

b) The mean exclusion probability of the blood group is calculated as the sum of the products of a combination frequency of mother-child [M, C], and the exclusion probability of the combination, $E_f(\mathrm{M},\mathrm{C})$ for all combinations of mother and child.

$$E_f = \sum_{M=1}^{NP} \sum_{C=1}^{NP} [\mathrm{M},\mathrm{C}] \cdot E_f(\mathrm{M},\mathrm{C}) \tag{15}$$

2.2 Maternity. a) The exclusion probability for a certain child C, $E_m(\mathrm{C})$, is calculated by summing up the phenotypic frequencies of non-mothers

$$E_m(\mathrm{C}) = \sum_{M=1}^{NP}{}' \,[\mathrm{M}] \quad \text{with } [\mathrm{M},\mathrm{C}] = 0 \tag{16}$$

where Σ' is defined in the same way as (14).

b) For a certain blood group, the exclusion probability E_m is defined in the same way as (15).

$$E_m = \sum_{C=1}^{NP} [\mathrm{C}] \cdot E_m(\mathrm{C}) \tag{17}$$

2.3 Parentage. a) The exclusion probability of parentage for a certain child C, $E_p(\mathrm{C})$, is calculated by summing up the combination frequencies of excluded father-mother combinations among all possible combinations.

$$E_p(\mathrm{C}) = \sum_{F=1}^{NP}{}' \sum_{M=1}^{NP}{}' \,[\mathrm{F}] \cdot [\mathrm{M}] \quad \text{with } [\mathrm{F},\mathrm{M},\mathrm{C}] = 0 \tag{18}$$

b) For a certain blood group, the exclusion probability is as follows

$$E_p = \sum_{C=1}^{NP} [\mathrm{C}] \cdot E_p(\mathrm{C}) \tag{19}$$

Corresponding to the alternative definition of parentage, the exclusion probabilities are also defined as

a') For a certain father-mother combination, the exclusion probability of non-child $E_p(\mathrm{F},\mathrm{M})$ is calculated by summing up the phenotypic frequencies of excluded children among all phenotypes.

$$E_p(\mathrm{F},\mathrm{M}) = \sum_{C=1}^{NP}{}' \,[\mathrm{C}] \quad \text{with } [\mathrm{F},\mathrm{M},\mathrm{C}] = 0 \tag{20}$$

b')For a certain blood group, the exclusion probability E_p is

$$E_p = \sum_{F=1}^{NP} \sum_{M=1}^{NP} [\mathrm{F}] \cdot [\mathrm{M}] \cdot E_p(\mathrm{F},\mathrm{M}) \tag{21}$$

This formula (21) gives the same values as formula (19).

Program Specification

The input data necessary for the calculation are as follows: (1) name of the blood group, (2) number of phenotypes of the blood group, (3) number of genes of the blood group, (4) the gene frequencies, and (5) translational matrix defined in ref. [8]. Details are explained in our previous work [8]. The same input data can be used for the computer programs written by us [8, 9].

The output data are as follows.

a) Mean exclusion probabilities of the blood group for maternity, paternity and parentage (E_m, E_f, E_p).

b) A table for indices of maternity for all possible combinations of mother-child (M, C, X_m, $(Y/X)_m$, E_m(C)).

c) A table for indices of paternity as well as parentage for all possible combinations of father-mother-child (F, M, C, $X_f(=X_p)$, $(Y/X)_f$, E_f(M, C), $(Y/X)_p$, E_p(F, M)).

d) Distributions of the relative frequencies of log (Y/X) for maternity, paternity and parentage with their means and variances.

The list of the program may be obtained from authors free of charge.

Results and Discussion

Table 1 indicates the results of calculation for parentage, such as mean values of the distributions of log (Y/X), variances, and exclusion probabilities for 14 blood groups in Japanese population. The gene frequencies used have been taken from our previous work [10]. a indicates the mean value of the distribution of log (Y/X) for true father-mother-child combinations and b, that for non father-mother-child combinations. These values indicate the efficiencies of the diagnosis of each blood group. Furthermore, these values are used in the following way: When a blood group (i) is independent of each other, following formulas hold for

Table 1. Mean values and variances of the distributions of log (Y/X) and exclusion probabilities for parentage in various blood groups

	a	σ_a^2	b	σ_b^2	E
1. Inv	−0.1046	0.0338	−0.0046	0.0542	0.1260
2. P	−0.1197	0.0367	−0.0108	0.0582	0.1477
3. Duffy	−0.1263	0.0609	−0.0181	0.0419	0.1468
4. AcP	−0.1640	0.0526	−0.0577	0.0452	0.2230
5. Hp	−0.1730	0.0498	−0.0725	0.0412	0.2435
6. EsD	−0.1826	0.0463	−0.0917	0.0340	0.2669
7. GPT	−0.1859	0.0449	−0.0993	0.0308	0.2753
8. Kidd	−0.1880	0.0440	−0.1044	0.0286	0.2809
9. AB0	−0.2408	0.0548	−0.1029	0.0624	0.3291
10. Rh	−0.2789	0.1140	0.0329	0.4031	0.3076
11. MNSs	−0.2847	0.1060	−0.1044	0.0641	0.3498
12. PGM	−0.3846	0.1394	−0.1490	0.0797	0.4450
13. Gc	−0.4238	0.1028	−0.2578	0.0518	0.5337
14. Gm	−0.4656	0.0738	−0.3132	0.0590	0.5895

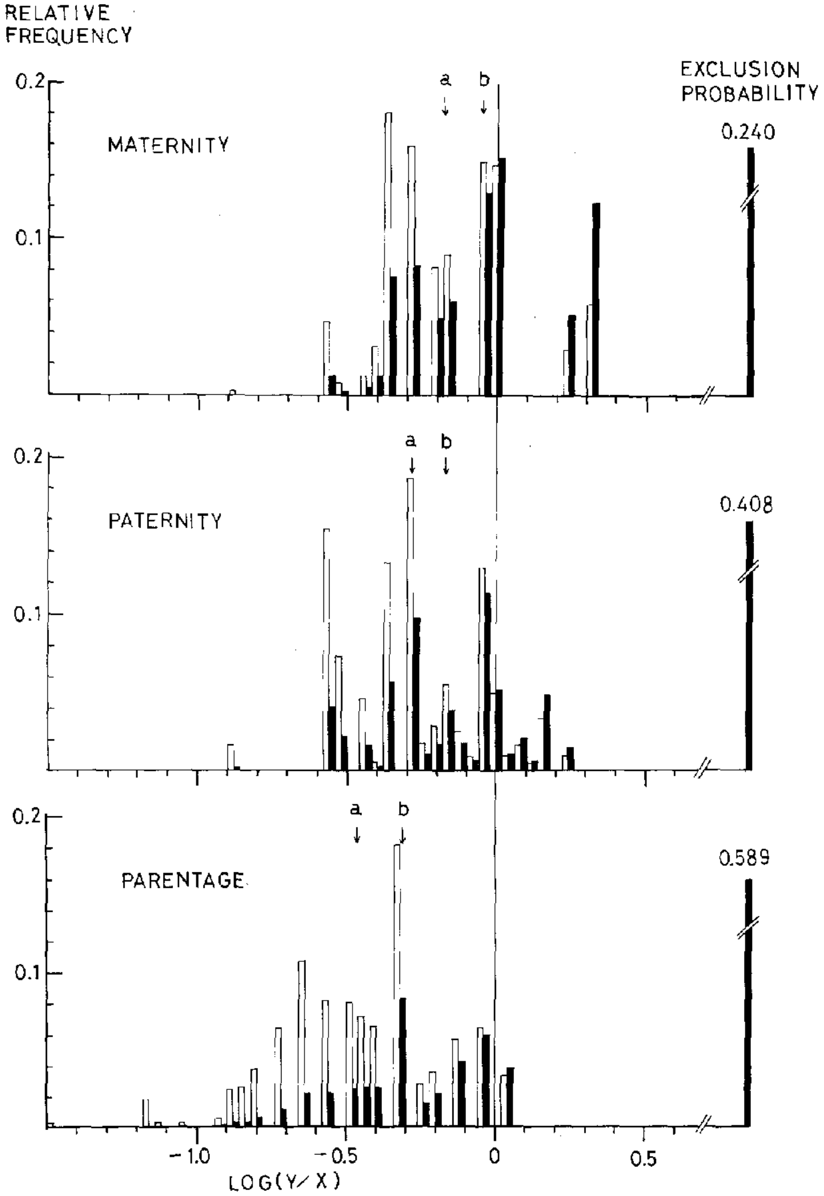

Fig. 1. Distributions of log *(Y/X)* for Gm blood group in Japanese population. The relative frequencies of log *(Y/X)* for true father-mother-child combinations are expressed by *white sticks* and their mean value is indicated by *a*. The relative frequencies of log *(Y/X)* for non father-mother-child combinations are expressed by *black sticks* and their mean value is indicated by *b*. The class interval is 0.04 in the logarithmic scale

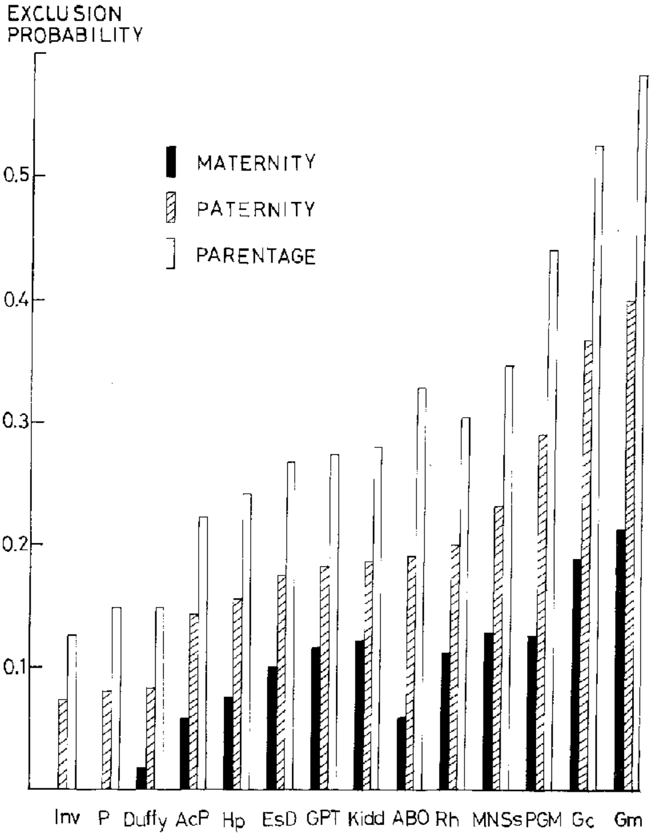

Fig. 2. Comparison of exclusion probabilities in maternity, paternity and parentage. The exclusion probabilities in maternity, paternity and parentage are expressed by three kinds of sticks. The blood groups are arranged in the order that their exclusion probabilities in paternity increase from left to right

the system consisting on n blood groups, as pointed out by Hummel in the distribution of paternity [11, 12] and in the exclusion probability E.

$$a = \sum_{i=1}^{n} a_i \qquad b = \sum_{i=1}^{n} b_i$$

$$\sigma^2 = \sum_{i=1}^{n} \sigma^2_{a_i} \qquad \sigma^2 = \sum_{i=1}^{n} \sigma^2_{b_i} \tag{22}$$

$$1-E = \prod_{i=1}^{n} (1-E_i)$$

Therefore, the distribution of parentage for any combinations of blood groups can be constructed theoretically by using these values in Table 1.

Figure 1 indicates the results of computer calculation in Gm. The exclusion probability increases in the order of maternity, paternity and parentage. The

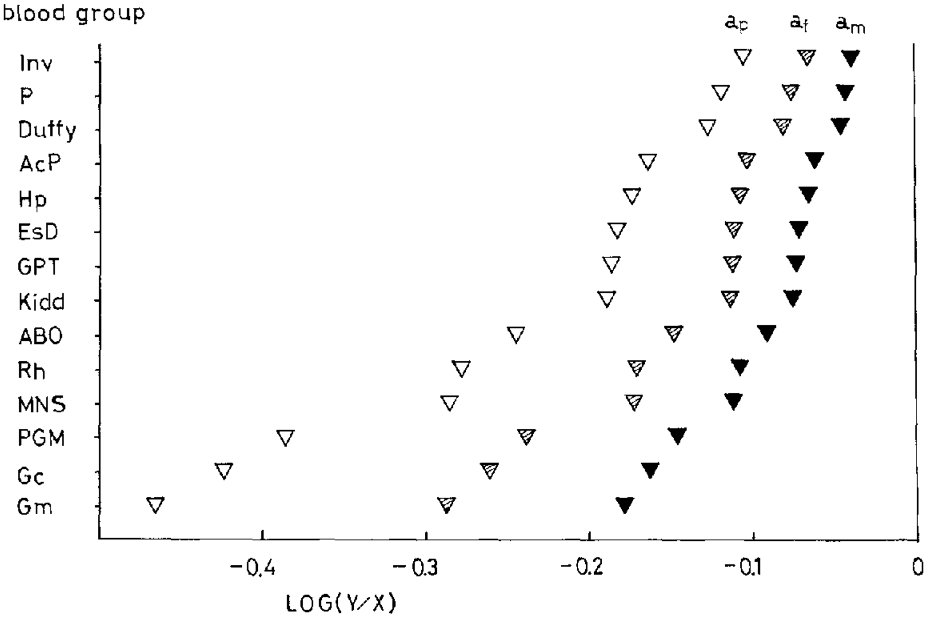

Fig. 3. Mean values of the distributions of log *(Y/X)* in maternity, paternity and parentage for true father-mother-child combinations. Symbols a_m (▼), a_f (▽), and a_p (▽) indicate the positions of mean values in maternity, paternity and parentage, respectively

mean value a decreases in the order of maternity, paternity and parentage, corresponding to the increase in the plausibility in this order. However, it is difficult to elucidate some quantitative relations between these values because there are no general algebraic expressions for the exclusion probability applicable to any blood groups, except to very simple blood group such as MN, nor those for the plausibility applicable to any blood groups. Therefore, we have calculated these values by using computer to find out some quantitative relations among the values in maternity, paternity and parentage.

The exclusion probabilities in 14 blood groups are shown in Fig. 2. These results confirm that the exclusion probabilities increase in the order of maternity, paternity, and parentage in any blood groups. We tried to find some relations as to the plausibilities in maternity, paternity and parentage. As we reported in our previous work [13], a and b are functions of exclusion probability E and variance σ^2 as

$$\begin{aligned} a &= \log(1-E) - \sigma^2/\log e \\ b &= \log(1-E) + \sigma^2/\log e \end{aligned} \tag{23}$$

In most blood groups σ^2 increases as E increases. The mean value a decreases as E increases from a blood group to another as shown in Fig. 3. Mean value b, however, does not always decrease as E increases from a blood group to another, because b is the sum of negative value $\log(1-E)$ and positive value $\sigma^2/\log e$ [13]. Therefore, we considered only mutual relation of mean values a_m, a_f and a_p. The differences of mean values $a_f - a_m$ and $a_p - a_f$ increase as the exclusion probability

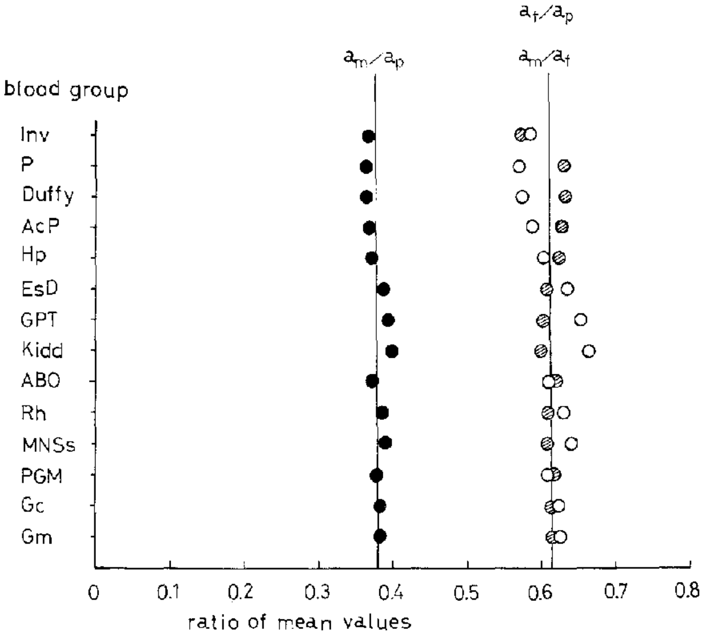

Fig. 4. Comparison of ratios of mean values of log *(Y/X)* for true father-mother-child combinations. Three kinds of ratios of mean values are indicated. Symbols ●, ◍, and ○ indicate a_m/a_p, a_f/a_p, and a_m/a_f, respectively

of a blood group increases, although no quantitative relations are found between both differences. The ratios a_m/a_p and a_f/a_p are calculated and plotted in Fig. 4. The former ratio distributes within the values from 0.36 to 0.40, whereas the latter ratio from 0.57 to 0.64. The ratio a_m/a_f also distributes within the values from 0.57 to 0.64. Therefore, these results are summarized in the following simple relation, and this relation holds not only for simple blood groups, such as AcP and Hp, but also for complicated blood groups, such as Rh and Gm.

$$a_m : a_f : a_p = (0.6)^2 : 0.6 : 1 \qquad (24)$$

This relation is helpful to construct the distribution of plausibility in parentage or that in maternity from that in paternity. For example, if a "typical value" in paternity is 90% in a certain system [2, 11, 12], the "typical value" in parentage and that in maternity are expected to be 97.3% and 79.4%, respectively, by using the relation.

Such a quantitative relation is not found in the exclusion probability. The difference of the exclusion probability in paternity and that in maternity $E_f - E_m$ varies from 0.05 to 0.18, and difference $E_p - E_f$ varies from 0.05 to 0.18. The ratio of the exclusion probability in maternity to that in paternity E_m/E_f varies from 0.0 to 0.44 and the ratio E_f/E_p varies from 0.55 to 0.7. These results are not summarized in a simple relation.

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