Kinship plausibilities from DNA fingerprints

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In our previous paper on the biostatistical evaluation of multilocus pherograms for normal mother-child-?father cases we dealt with two points we should like to apply to a practical question in this paper: firstly, how to determine both the total number of bands, visible and invisible, recorded by the probe and their mean frequency with the aid of the maximum likelihood technique, and, secondly, how to calculate a probability of paternity using the Essen-Möller formula, assuming that every band belongs to a corresponding di-allele system with a silent antithetical property and further that each is genetically independent. With these two steps we can biometrically evaluate pherograms of multilocus probes in deficiency cases, that is, in cases without findings either for the mother or for the putative father (though findings for close kin may compensate). For an exclusion expectation in deficiency cases one must have findings for both parents of either the child's mother or the putative father. Only in this case can a non-father be excluded without any reservations, in other words, will the W_A value (= probability of paternity for non-excludable men for a given child-mother constellation) be fully valid. In practically all other cases proof of non-kinship through "exclusion" (= genetic incompatibility) is either rare or, as the following cases, impossible:

- questionable paternity without findings for the mother;
- questionable maternity without findings for the father;
- questionable paternity without findings for the putative father, but with findings for his siblings and/or real children;
- questionable grandmaternity without findings for the putative father and the putative grandfather;
- questionable grandpaternity without findings for the putative father and the putative grandmother; and
- disputed full or half-sibship without findings for one or both of the parents.

In all these cases only biostatistics can provide positive or negative proof. To establish parentage a second or third probe may be necessary in some cases. The biostatistical results from the probes are combined with one another and with the results of the blood-group and HLA opinions. This gives an overall W value for the case in question.

As a rule, deficiency cases are 2-hypotheses cases: the allegation is true or not. However, there are also cases with more than two hypotheses, e.g. when neither the one nor the other hypothesis, but a third possibility is correct (= 3-hypotheses case). For every hypothesis there is a pedigree. With a special kinship algorithm, and

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using a mean band frequency, one calculates the frequency of the pedigrees. In the 2-hypotheses case these are f(X) and f(Y). They are inserted in the Essen-Möller formula:

$$W = \frac{1}{1+[f(Y)/f(X)]}$$

which gives the appropriate probability of paternity, maternity, grandmaternity, grandpaternity or sibship. In a case of disputed grandmaternity (= mother of the deceased putative father), to which the multilocus probe 33.15 was applied, the biostatistical result was a plausibility of W = 99,2%. The value speaks very much in favour of real grandmaternity (predicate: 'highly probable'). Moreover, this value is extremely typical of real grandmothers for the given child-mother constellation: their mean value is W = 77%, that is, well below 99,2% (false grandmothers have a mean of W = 29%).

The scatter of the W values for real grandmothers in the present case lies between W = 77% and at least W = 99,2%; in the other direction values of less than W = 50% may be expected. Hence, only high W values clearly indicate kinship; low values speak neither for nor against.

The correctness of this evaluation of W values in deficiency cases was borne out of 190 cases of disputed paternity without findings for the mother. In 95 cases the real father was involved, and in 95 cases a non-father. In 125 analogous cases blood-group and HLA-A,B findings were also available. Practically all non-fathers were excluded from paternity in the blood-group or - more often - the HLA opinion. However, there was not a single exclusion with the multilocus probe, only probability values. Most of these were below 50%; those in the positive range were as high as W = 90% and above. The distribution of the W values for real fathers from blood-group and HLA findings is similar to those for the DNA analysis with the probe 33.15. However, closer examination reveals that the serological expertise provides values in the highest W-values range, and hardly any W values below 50%. For the multilocus probe 33.15, on the other hand, W values for real fathers of below 10% cannot be excluded, and there are fewer results in highest W value classes than for the blood-group and HLA opinions.

The following is an example of a relatively complicated deficiency case - a 2-man case.

A child - now adult - doubts whether his deceased legal father is his real father. Two men are eligible for paternity: his uncle by marriage, previously married to his mother's deceased sister, and an unknown man. Blood-group findings and DNA pherograms are available for 5 persons:

- for the child;
- for his brother (whose parentage is not in dispute);
- for their mother;
- for the daughter of the mother's deceased sister and the uncle by marriage; and
- for the daughter of the mother's deceased sister and another man.

There are three alternatives for the child's paternity (= 3 hypotheses):

1. the child's father is the legal father;
2. the child's father is his uncle by marriage;
3. the child's father is an unknown man.

The blood-group and DNA findings produced the following W values:

	real father is legal father	real father is uncle by marriage	real father is unknown man
blood group opinion	15%	3%	82%
DNA analysis: 2 single-locus probes	16,7%	16,7%	66,6%
DNA analysis: multilocus probe 33.15	0,2%	1,2%	98,6%
combined:	0,008%	0,01%	99,98%

The combined value for the findings of the blood-group opinion and those of the 2 single-locus probes is W = 90%, that is, it is 'probable' that an unknown man is the child's real father. The multilocus probe 33.15 brought the real breakthrough. With the combined W value of 99,98% it is 'practically proven' that an unknown man is the child's real father, and 'practically excluded' that his legal father or his uncle by marriage is.

I should like to very briefly explain the mathematics of the biostatistical evaluation. The blood-group findings as well as the radiograms with the two single-locus probes were evaluated with an extended version of the kinship algorithm Ihm and I developed in 1975 (that is, 14 years ago). This gives the frequencies for the 3 pedigrees. The ratios between them give the probability values for the three hypotheses; the sum of these values is 1 (= 100%).

In principle, the fingerprints obtained with the multilocus probe 33.15 are treated in the same way. However, there is one pecularity: the pherograms gave 12 different band constellations for the five people in the case, with frequencies of occurence between 1 and 8. A 13th constellation was calculated theoretically with n = 33. Each of the 13 band constellations gives a probability value for each of the 3 hypotheses.If a constellation occurs more than once (in the present case: up to 33 times), this affects the probabilities of the hypotheses accordingly. Combining the probabilities obtained from all 13 fingerprints, we get W = 0,2% for the hypothesis 'the legal father is the real father of the child', W = 1,2% for the hypothesis 'the uncle by marriage is the real father of the child', and W = 98,6% for the hypothesis 'an unknown man is the real father of the child'.