

EDITORIAL

RECOMMENDATIONS OF THE SOCIETY FOR FORENSIC HAEMOGENETICS CONCERNING DNA POLYMORPHISMS

The new techniques allowing the characterisation of individuals at the DNA level has been the subject of a recent article this journal (*Forensic Science International*, 41 (1989) 197–203). The topicality of the subject and its importance in forensic science is reflected in the literature and the programme content of current scientific meetings in this field. The International Society for Forensic Haemogenetics set up a Committee in 1987 to discuss these exciting developments in relation to their use in the medico-legal context and the following recommendations have been made by the Society:

DNA Recommendation

A. General statement

The history of forensic sciences show that research and science are closely related to the practical work in this field and vice versa. A major aim of the International Society for Forensic Haemogenetics is to advance the quality of forensic evidence. For this purpose, the development of new technology is essential. It must, however, meet the requirements defined by legislation and the quality standards which have been established over the past decades. Based on these considerations, the DNA Committee has agreed on the following statements: (1) Recombinant DNA technology is becoming an essential tool for the improvement of forensic evidence, but is only part of the expertise. (2) In order to achieve high reproducibility and to guarantee the important principle of a second opinion, certain criteria must be fulfilled for the investigation of DNA polymorphisms: (i) DNA probes should be generally available; (ii) DNA probes should only be used, if at least two independent laboratories have the necessary expertise; (iii) Basic reliability and biostatistical standards which have been established in the classical field of forensic haemogenetics must be applied to the forensic use of DNA.

B. Requirements for the application in paternity testing

(1) *Definition of systems and "alleles"*. (a) Generally a DNA system is defined by a DNA probe recognising a genomic sequence and by the specificities (cleavage sites) of restriction enzymes and/or the unique identification of complementary primer sequences. (b) "Alleles" are defined by DNA fragments of variable length, which are detected in a polymorphic system and

agreeing with a formal genetic model. They are detected by means of conventional Southern blot analysis or comparable methods. They are usually represented by one or two restriction fragments of a given size generated by the use of one enzyme (or two enzymes in double digestions) and detected with one probe. (c) The "allele" designation should be preferentially in kilobase size.

(2) *General and developmental requirements.* (a) Collaboration and exchange of data should be encouraged to establish the usefulness of a system. (b) DNA polymorphisms should be defined by family and population studies. At least 500 meioses and an adequate population sample should have been tested and published, before a polymorphism can be introduced into paternity testing. (c) The chromosomal localisation and linkage data to other polymorphisms used in paternity testing should be available. This information should be documented in the publications of the International Human Gene Mapping Workshop. (d) The description must include: information on the type of probe, estimation of the number of alleles, size of constant and variable fragments, proof of Mendelian inheritance, "allele" or haplotype frequencies, frequencies of mutations and/or recombinations and a check of the Hardy-Weinberg equilibrium.

(3) *Requirements for methodology and standardization.* (a) Size markers with discrete fragments of known size should span and flank the entire range of the DNA system being tested. (b) A human control DNA of known allele composition should be included on each gel. (c) Intactness of the individual genomic DNA before restriction enzyme digestion and complete digestion of the DNA should be assured by appropriate control experiments.

(4) *Suggestions and requirements concerning the type of system.* So far, two different types of systems have to be taken into consideration:

(4.1) Single locus systems (sls) — (i) The mutation rate should be 10^{-4} or less, using a single restriction enzyme. (ii) Systems with higher mutation rates should not be used routinely and require special considerations.

(4.2) Multi locus systems (mls) — (i) Mutation rates of up to 10^{-2} and higher have to be expected. (ii) Since calculated probabilities on the statistical basis for paternity as mentioned in Section A(2) cannot be given, only verbal opinions about exclusion or non-exclusion are possible. (iii) The use of mls's without biostatistical evaluation is currently not acceptable in routine paternity testing.

(5) *Conclusions.* Paternity testing with conventional techniques is a well established procedure as evidence in Court cases. DNA polymorphisms need to be further explored. At the present time, DNA techniques should be used only in combination with conventional systems.

C. Specific requirements for the application in criminal investigations

In this section some specific requirements are listed with regard to the analysis of stains; however, it is stressed that most of the requirements in parts A and B are also relevant under this heading. The sections in parts A

and B not applicable to the analysis of stains are as follows: B(2) paragraph 2 references to family studies, the sub-clause of B(2) paragraph 4 referring to mutations and also the references to mutations and paternity under B(4) and B(5).

(1) *Somatic stability.* The application of DNA analysis to criminal investigations is mainly concerned with the comparison of results obtained from a control blood sample with that obtained from a body fluid stain. The stain may be a deposit of blood, semen, vaginal fluid, saliva or even a smear of tissue. Also the analysis of hairs, in particular hair roots, may be undertaken. The system used should therefore be shown to be somatically stable.

(2) *Statistical interpretation and band matching.* In addition to A(2) above it is recognised that the reproducibility of any method may lead to the imprecise alignment of bands which are nevertheless considered to match. Furthermore in stain analysis it may be that interfering substrates or degradation of the DNA has caused minor variations in band position again leading to some distortion. Such positional variations must be shown to be within the expected experimental variation and wherever possible the statistical assessment should be correspondingly adjusted. A record of the analysis, the associated results and the method of statistical evaluation should be readily available for examination by a second independent analyst.

(3) *Analysis of semen-contaminated vaginal swabs.* When differential extraction of vaginal swabs is carried out the supernatant containing the female DNA should be tested. Generally for this analysis the removal of all of the female DNA should be avoided as it may provide a useful internal marker.

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