Int J Leg Med (1994) 107:159–160

TECHNICAL NOTE

DNA recommendations – 1994 report concerning further recommendations of the DNA Commission of the ISFH regarding PCR-based polymorphisms in STR (short tandem repeat) systems

Introduction

The DNA Commission of the International Society of Forensic Haemogenetics (ISFH) convened during the 15th International Congress in Venice in October 1993.

Major topics of discussion were the construction and nomenclature of allelic ladders, exchange of data and establishment of international data banks.

The recommendations are summarized as follows:

General

The previous recommendations provided by the DNA Commission are still valid (Int J Leg Med 1992, 105: 63–64; Forensic Sci Int 1992; 55:1–3) and the forensic use of the STR loci should conform to these standards.

Allelic ladders and nomenclature

Allelic ladders should be used for all STR systems detected by manual electrophoretic systems and should be based on the predominant simple repeat motif of the system in question. The commonly occurring alleles should be present in the ladder.

More complex STR loci, such as human beta-actin related pseudo gene H-beta-Ac-psi-2 (ACTBP2, also known as SE33), may contain such hypervariable sequences that generating a ladder based on the consensus repeat sequence will be impractical. However, regularly spaced ladders can and should be constructed. Interlaboratory exchange of data in such systems is difficult as the sequence variation in the alleles can affect the relative mobility under different electrophoretic conditions. Therefore for this purpose, detailed information on electrophoretic conditions must be available. All alleles in an allelic ladder should be sequenced to establish the sequence of the repeat unit(s), the number of repeats present and the actual size of the allelic fragment.

Although sequencing of alleles in the allelic ladder is recommended, alleles in a population database or in an identity testing case do not need to be sequenced. Firstly, such an endeavour is not practical, and secondly alleles are operationally defined by comparison with a sequenced ladder making additional sequencing superfluous.

Published sequenced allelic ladders should be made available to a reasonable number of scientists on request. Once commercially available they can be obtained from the commercial source.

Consistent with the construction of an allelic ladder alleles should, when possible, be designated by the number of repeats they contain even if the sequence of the repeats is different (e.g. VWA). We recognize that alleles with the same classification may actually vary in their sequences and in the actual number of repeats due to insertions or deletions of repeats or the flanking regions, but this should not present a problem either for data base generation or identity testing.

When an allele does not conform to the standard repeat motif of the system in question it should be designated by the number of complete repeat units and the number of base pairs of the partial repeat. These 2 values should be separated by a decimal point. For example, HumTHO1 alleles differ in size by 1 repeat unit of 4 bp except for one relatively common allele in Caucausians. This allele is 1bp shorter than 10 repeat units and should therefore be designated 9.3.

Some analytical systems do not require an allelic ladder as a reference for allele typing. For example, semi-automated methods which employ fluorescent tags to identify alleles contain internal standards within the same electrophoretic lane as the sample being tested. The alleles are characterized by their fragment size in base pairs but should be converted to the aforementioned allele designation protocol. Additionally, it should be stated which primer has been labelled and which label has been used. If an allelic ladder is labelled it should be consistent with the labelled primer used to amplify the STR alleles.

Both native and denaturing gels are acceptable for the electrophoretic separation of STR alleles as long as detailed information is provided.

Population data

When publishing population data on PCR-based DNA systems it should be stated whether the published ISFH guidelines have been adhered to.

The DNA-Commission consisted of the Executive Committee of the ISFH (W. Bär, B. Brinkmann, P. Lincoln, W. R. Mayr, U. Rossi) and coopted external experts (B. Budowle, C. Bell, A. Carracedo, A. Eisenberg, R. Fourney, P. Gill, A. Kloosterman, K. Monson, O. Pascal, S. Rand, J. Robertson, A. van Daal).