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Short communication

The evolution of DNA databases—Recommendations for new European STR loci

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Abstract

Following a recent meeting by the ENFSI and EDNAP groups on the 4–5 April, 2005, in Glasgow, UK, it was unanimously agreed that the process of standardization within Europe should take account of recent work that unequivocally demonstrated that chance of obtaining a result from a degraded sample was increased when small amplicons (mini-STRs) were analysed. Consequently, it was recommended that existing multiplexes are re-engineered to enable small amplicon detection, and that three new mini-STR loci with alleles <130 bp (D10S1248, D14S1434 and D22S1045) are adopted as universal. This will increase the number of European standard Interpol loci from 7 to 10.

Keywords: National DNA database; STR; Evolution; European standards

1. Introduction

The European Network of Forensic Science Institutes (ENFSI) and the European DNA Profiling Group (EDNAP) has worked collaboratively to achieve standardisation of DNA profiling throughout Europe. The purpose is to facilitate comparisons of DNA profiles between laboratories. Currently, there are seven Interpol short tandem repeat (STR) loci that are in common to all European laboratories [1,2], but this is insufficient to accommodate the potential numbers of comparisons that may be made because chance matches will be commonplace.

2. Standardisation of STR loci

Several multiplexes are in common use throughout Europe (http://www.enfsi.org/ewg/activities), although the 'official' number of Interpol loci is just 7, many laboratories show a much higher degree of compatibility because they use the same multiplex systems. In particular, more than half of the laboratories questioned utilise AmpFlSTR SGM Plus or an equivalent multiplex that incorporates the same loci. Nevertheless, the conclusions of the ENFSI/EDNAP groups after recent collaborative experimentation; [3] (manuscript in preparation) are that there is a need to alter existing multiplexes to improve success rates when degraded DNA is analysed. A very high level of standardization has already been achieved at the scientific and technical level to allow rapid implementation of new database loci.

Because differences exist between countries in their choice of loci for the purpose of constructing National DNA databases, it is accepted that any technical changes

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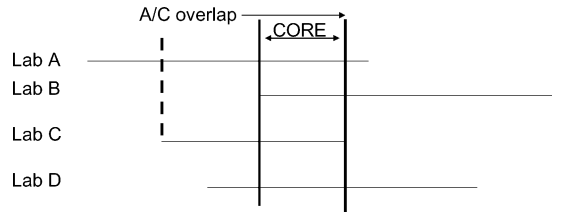

Fig. 1. Four laboratories run four different multiplexes represented by the horizontal lines. There is some overlap between all bounded by the vertical lines—but some labs overlap more than others, e.g. labs A and C have greater compatibility than A and B.

to existing multiplexes will have to accommodate national requirements. In particular, it would not be feasible for laboratories to abandon loci; rather the requirement is to design multiplexes that incorporate new loci in addition to those currently used.

We also accept that there are legislative and political issues to solve—in some jurisdictions it may be a requirement to alter the law in order to allow exchange of information. This paper is solely concerned with the scientific issues.

Rather than using a centralised database, where profiles are physically held, it is proposed that the most cost effective and efficient method would be to link database laboratories together using virtual networks. Search engines would then be used to interrogate databases. This way, each country would retain control over their respective databases, and the scope for searches would be easily controlled according to local legislative requirements. Also searches will not be limited by the Interpol core loci, as many labs will be compatible at many more than the core group (Fig. 1; http://www.enfsi.org/ewg/activities).

However, we recognise that the databases should be dynamically evolving to keep pace with developments in the area. We assume that over time the database will change, but we need to carefully address the reasons for change. Briefly, they can be summarised as follows:

(1) To improve the discriminating power.
(2) To improve the sensitivity of testing so that smaller amounts of DNA may be detected.
(3) To improve robustness or the quality of the result.

Adding loci will automatically improve the discriminating power—but should these comprise existing STR systems

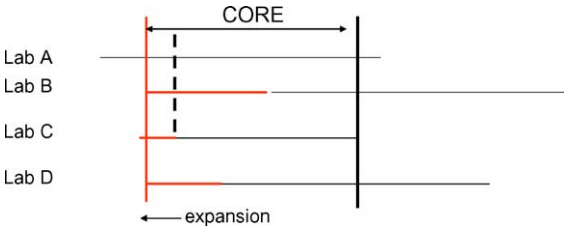

Fig. 2. Labs B, C and D introduce more loci into their multiplex systems, whilst retaining capability to analyse older loci.

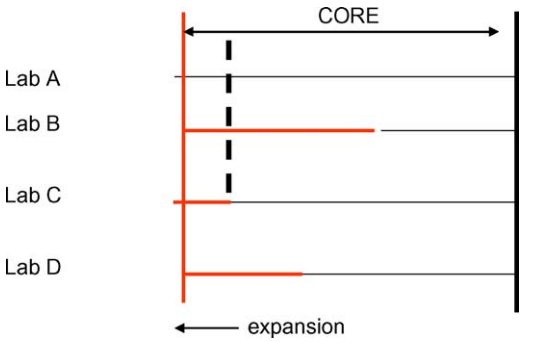

Fig. 3. Over time it may be possible to abandon original loci—at this stage, DNA loci will be standardized around the core group.

or should they comprise new low molecular weight markers such as SNPs or mini-STRs, which could be more suitable for typing of heavily degraded DNA?

Under the assumption that it is agreed to increase the number of loci in order to achieve commonality throughout Europe, the question is how can this be achieved?

Given that substantial national DNA databases have already been constructed using divergent multiplexes, it is unrealistic to suggest that laboratories can change by abandoning loci in favour of new ones. Rather, it is proposed that new core loci are decided and then laboratories expand their systems whilst retaining their existing set of STRs (Fig. 2). The new core loci will be decided after collaborative experimentation by the EDNAP group, leading to recommendations and subsequent ratification by the ENFSI group.

A natural consequence of the proposed locus expansion strategy will be the requirement for labs to use a different multiplex to that previously used. Furthermore, different labs will require custom multiplexes. New biochemical methods may be required to facilitate a flexible amplification strategy of currently used and newly selected database systems.

After a period of time, it may be possible to abandon some loci (although they would remain on the database). Further expansion into new loci may occur by agreement, and thus the database becomes dynamic where loci may be added or subtracted from a multiplex over long periods of time. The process will be driven by joint recommendations and considerations of the ENFSI and EDNAP groups (Fig. 3).

3. Decisions on new European loci and method of implementation

At concurrent meetings held on 4–5 April, 2005, in Glasgow, UK by the EDNAP and ENFSI groups, as a result of collaborative exercises and a review of the literature, the following recommendations were made:

(1) Mini-STRs to be adopted as the way forward to increase both the robustness and sensitivity of analysis.

(2) The role of SNPs to be considered further in relation to the analysis of discrete samples such as bone—we consider that the prime utilisation of SNPs will be in relation to mass disasters, where analyses have to deal with highly compromised samples, but can be carried out independent of a national DNA database [4].
(3) Existing core-loci used in national DNA databases will be retained and converted into mini-STRs by re-engineering, so that the primers are built close to the repeat region [5,6].
(4) Furthermore, we recommend that European laboratories adopt three new mini-STR loci, namely: D10S1248, D14S1434 and D22S1045 [7,8].
(5) We have compiled a secondary list of loci, which may be converted into useful mini-STRs: D12S391, D1S1656, TPOX. These loci are not 'core' but we recommend that these are used if a manufacturer wishes to consider additional loci to those listed in (4) [6,9,10].
(6) New multiplexes should be equivalent in sensitivity to existing multiplexes, which means that full profiles are achieved down to ca. 250 ng undegraded genomic DNA using standard PCR amplification methods. It is important to recognize that the primary purpose is not to generate systems that are 'super-sensitive' or low-copy-number equivalent. The primary purpose is to facilitate detection of partly degraded DNA. This strategy also concurrently increases the discriminating power, which improves the effectiveness of comparisons between National DNA databases. Conversely, the larger the multiplex, the less efficient the amplification may become. This means that success rates could be compromised. Clearly there is a balance to be struck between the size of the multiplex and its efficiency.
(7) It is proposed that the ENFSI/EDNAP groups evaluate new multiplexes according to the above criteria. Manufacturers will be actively encouraged to provide suitable candidate multiplexes.

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