DNA PROFILING IN CRIME INVESTIGATIONS: AN EUROPEAN OVERVIEW

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History

Towards the end of 1988, when single locus probing (SLP) analysis was becoming a reality for routine casework in forensic science, there was an announcement that there was an intention to relax border restrictions within the European Community. As this was designed to allow greater freedom of unchecked movement between the various member countries there was an implication that there would be the potential for an increase in cross-border crime. It was therefore realised by forensic scientists that there could be a requirement for laboratories to compare and exchange results obtained in crime case investigations in the event that a suspected perpetrator resided in another country. In order to facilitate an interchange of DNA profiling data it was imperative that all laboratories would be using the same systems and methods. Therefore a meeting was arranged for representatives from laboratories in Europe who had started to use the technology in order to decide on a set of SLP probes and, in particular, a restriction enzyme, which would be common to all participants. It took two meetings to arrive at a point of agreement in early 1989. The restriction enzyme Hinf1 together with the probes YNH24 and MS43A were selected to form the basis of an inter-laboratory study to evaluate the ability of the group to harmonize on common systems. As a result of these meetings the European DNA Profiling (EDNAP) group was born and a programme for European integration was initiated.

Originally, EDNAP had only one objective which was to ensure that common systems would be used throughout the European Community in order that DNA results from crime cases could be exchanged. There was, additionally an agreement that the results from all inter-laboratory exercises would be published in the scientific literature starting with the SLP study [1]. Following a successful period with the use of SLP analysis, attention was focused on the need to harmonize on systems which employed amplification methodology. There was unanimous agreement that the way forward lay with the use of short tandem repeat (STR) technology and future effort would be concentrated on finding a set of loci which could form a core system. A number of different STRs were the subject of inter-laboratory exercises but, in the event, HUMTH01 and vWA were chosen to be the common loci [2]. By this stage of the development of integration within Europe, although only two loci were designated as common, there was a far larger group of loci which were being used by most forensic science laboratories.

With the prospect of the formation of a national DNA database, the Forensic Science Service (FSS) in the UK had been developing a multiplex which could be used for routine casework and form the basis of a national index [3,4]. Following a couple of false starts the hexaplex of STR loci together with amelogenin (Second Generation Multiplex (SGM)) was validated and used to develop a national DNA database which came into operation in 1995. This has proved to be a very successful venture and has attracted much interest from other laboratories in Europe; many of which tested the multiplex and now use it operationally.

Five years ago the European Network of Forensic Institutes (ENFSI) became involved with a Europeanwide programme to set standards for scientific work carried out in crime investigations within state-run organizations. The DNA group within ENFSI has made great strides in raising the standard of operational work by conducting inter-laboratory exercises to determine the reliability of a variety of STR loci. Recently they carried out an extensive study to evaluate the performance of commercially available STR multiplexes in the hands of the various member laboratories. There is little doubt that the availability of commercial systems is attractive to many laboratories who do not have the time or the facilities to produce and maintain their own STR primer sets. Therefore the results of this testing could determine the common system for the majority of laboratories within Europe.

In addition to the testing and evaluation of the genomic STR loci both EDNAP and ENFSI have been active in evaluating the reliability of the use of mitochondrial DNA (mtDNA).

Current technology

There has been a steady evolution in the use of STR loci in forensic science throughout Europe. Two years ago a questionnaire indicated that there were more than fifty different STR loci in regular use with a number of systems employed for their detection. A point has now been reached where the vast majority of

laboratories have settled on the use of a similar set of STR loci which are separated by vertical gel electrophoresis or capillary electrophoresis with laser detection of fluorescent dye-labelled amplified products.

The value of intelligence databases has long been realised by law enforcement agencies and therefore the need for effective DNA databases became a driving force for the identification of packages of STRs which would be sufficiently reliable and robust for the storage and searching of personal data. It was appreciated that databasing would involve the batch analysis of large numbers of samples and therefore research centered on the need for systems in which a number of primer sets could be simultaneously multiplexed. In 1993, in the UK, the FSS developed a multiplex of 4 STR loci, termed the Quad, which was designed for operational casework but with databasing in mind. The Quad consisted of the four loci TH01, vWA, FES and F13A. In the event this multiplex was deemed to have insufficient discriminating power for use in searching large numbers of samples and subsequent development the SGM was produced which consisted of 6 STR loci: TH01, vWA, FGA, D8S1179, D18S51 and D21S11. This proved to be a robust multiplex and was adopted by a number of forensic science laboratories within Europe. It also continues to form the basis of the UK national DNA database.

An Interpol Working Party on DNA Profiling [5] was set up to produce a set of loci which could be used for a register of offenders and, based on the initial EDNAP exercises and on recommendations by ENFSI, four STRs were identified: THO1, vWA, D21S11 and FGA. These four loci have now been accepted as the European standard and are referred to as European core loci. Three more loci, D3S1358, D8S1179 and D18S51, are currently being considered to extend this list.

The most recent developments have been associated with the introduction of commercially prepared kits and the introduction of the North American (CODIS) DNA database where a set of 13 loci have been chosen for sample analysis. Commercial kits have become available for testing and all 6 of the SGM loci are represented in the multiplexes. These have been tested extensively within Europe through ENFSI interlaboratory exercises [6] to determine which systems performed best within the European environment. Although some of the primers within the kits are different from those in the original SGM multimix, interlaboratory testing will determine compatibility with the results from previously analysed samples. It is expected that commercially produced kits will be popular with laboratories because, aside from the cost, they eliminate the need for the labour intensive manufacture and associated quality control and circumvent the problems associated with licensing issues. Although the advantage might be negligible for the few laboratories involved with the process of large numbers of samples, the gain should be considerable for those laboratories who have a relatively small throughput.

Regardless of the evolution of harmonisation in terms of agreed loci it is likely that STR analysis will continue to form the basis for the analysis of samples within crime cases. However, the platform for their detection could change. The emergence of miniaturised systems which include chip technology, capillary electrophoresis and mass spectroscopy could have a considerable effect on the working practices within Europe. Those laboratories with a need for multi-sample analysis will be looking for the introduction of greater levels of automation to reduce costs and all police forces would welcome a shorter analysis time.

Alongside the examination of the autosomal STRs there have been a number of exercises to develop the use of Y-chromosome STRs. Indications are that these markers can be extremely useful especially in the examination of sexual assault cases. The most informative of these loci appears to be DYS385 [7] and has been the subject of an EDNAP inter-laboratory exercise. However, a pentaplex of Y-linked STRs (DYS19, DYS389I, DYS389 II, DYS390 and DYS393) is currently being assessed, using both singleplex and multiplex conditions, by the group members for its usefulness in casework investigations. So far the results appear encouraging.

Since the introduction of DNA profiling scientists have been able to obtain more results from less, or more degraded, material but there is still a requirement to go further. Consequently there has been a push to use mtDNA analysis in an increasing number of cases; especially those involving hair or bone fragments. Recently EDNAP carried out an inter-laboratory exercise to determine whether uniformity of sequencing results could be achieved across the spectrum of laboratories using a range of different methodologies [8]. Further exercises are underway against the background of the heteroplasmy debate.

Databases

While there has been some progress with the details of the introduction of national DNA databases no major developments have been made in the last year [9].

The first of the national DNA databases to be formed occurred in the UK in 1995 and during its short life it has demonstrated its effectiveness in the detection of the perpetrators of crimes. A change in legislation was required in order to make it a successful venture. The database now has entries of personal profiles totalling nearly half a million and it is therefore not surprising that there are around 700 matches achieved each week. Such an enterprise requires a massive investment in both staff and equipment but it has the potential to be cost effective in the amount of investigative time that can be saved.

So far three other European countries have successfully introduced national DNA databases, Austria, Germany and the Netherlands. Due to the limitations imposed by the various legal systems these databases are not as comprehensive as that in the UK and therefore will not hold the same level of personal profiles (see later). The STR loci used to form these databases are as follows:

UK:	TH01, vWA,	FGA, D8S1179, D18S51, D21S11
The Netherlands:	TH01, vWA,	FGA, D8S1179, D18S51, D21S11
Austria:	TH01, vWA,	FGA, D8S1179, D18S51, D21S11
Germany:	TH01, vWA,	FGA, SE33, D21S11

Even though the German set of loci does not contain the complete SGM, the European core system is present and there is sufficient overlap for the exchange of results. In all cases there is a requirement to obtain another sample for analysis should a match be determined. The second analysis will be confirmed with another set of loci.

Legislation for the formation of databases in other European countries is well advanced especially in the Scandinavian countries. To date preparations to introduce national databases are underway in Belgium, Denmark, Finland, France, Norway, Sweden and Switzerland. The extent of the legislation is not entirely clear and therefore the future effectiveness of the databases cannot be assessed at this stage. Also, decisions have yet to be made on which loci will be used but it is confidently expected that at least the European core system will be used. Now that some databases are a reality and others will soon be in place, if legislation allows there is the possibility to exchange results between the various European countries. In these circumstances quality issues become important. Maintenance and security of the stored information becomes of paramount importance and local legislation must be observed in all details. Systems must be in place to ensure that the data is correct and that a personal profile is removed if the person is acquitted of the crime or if no further action is taken.

It is inevitable that, in the early stages of database operations, there will not be any dramatic successes; these will only come when there is sufficient data for matches to be made. The UK DNA database is now beginning to show how useful these systems can be. In the last two years the database has identified approximately 70 suspects in murder cases, 250 in sexual assaults and more than 200 in robberies. Also identifications have been made in over 20,000 burglaries. This must represent a very considerable saving in the amount of police time spent on the investigations.

Ethical and legal issues

The forensic scientists within Europe have long seen the advantages of DNA databases and through good discussion and a good deal of compromise a level of harmonization has been reached which would allow for the exchange of information. Networked databases would make it much easier to identify criminals who are responsible for crimes in other countries but the different criminal justice systems which currently exist are unlikely to make this a reality in the near future. For a variety of historical and political reasons, civil rights and human dignity issues are viewed differently within the separate countries. In the UK the 1995 Criminal Justice Act allowed police to take non-intimate samples for DNA analysis from anyone suspected of committing a recordable offence (in general terms this means a crime which could attract a custodial sentence). If the person is found guilty the DNA profile can stay on the database forever and, furthermore, should new technology become available the sample can be re-tested using the new system. If the person is acquitted, however, the result must be removed from the database. In other European countries more restrictive legislation is envisaged whereby personal profiles can only be entered onto a database if the individual is convicted of a serious offence. The reasoning behind the UK decision came from statistics which showed that the vast majority of men found guilty of sexual assaults had previous convictions for more minor crimes. Thus a person who sets out to be a serial rapist could be denied his ambition because he would be identified from the database after the first assault.

In Germany the samples for analysis are provided anonymously to the laboratories, whereas the resulting DNA profiles are entered, together with a name, onto the database. While this has the advantage of

preventing misuse of the DNA sample it provides a very cumbersome system for the scientist to operate. In other jurisdictions there are legal decisions which will, unwittingly, make the databases less effective. In Holland a sample for DNA processing can only be taken if it helps to prove the case. Therefore if a suspect admits a sexual offence, no sample is taken and no personal profile is entered onto the database. While it is true that there has been some debate concerning the storage of genetic profiles which might, in the future, provide more information than was previously thought, the whole subject of storing DNA results has become emotive. Legislation for security of the data could be enacted to prevent any unauthorised agencies from obtaining the information. Wide-ranging fingerprint databases have been in use around the world for a long time and fears about their misuse are generally unfounded.

All of these issues were debated at a meeting in Germany [10] where it was generally agreed by scientists that use of comprehensive DNA databases can be extremely effective in linking scenes of crime and identifying the perpetrators of a wide spectrum of cases. Forensic scientists have been successful with the integration of DNA systems throughout Europe and it is now the turn of the legislators to provide a harmonized framework of criminal justice in which to operate.

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