STR Data Goes to Court: A Laboratory Perspective

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The last ten years have seen the introduction of human DNA identification tests on various biological samples in crime laboratories in the United States and around the world. Laboratories in the United States have acquired vast experience in performing DNA tests and taking the results and conclusions drawn from those tests into the courtroom for presentation to judges and juries. Restriction fragment length polymorphism (RFLP) data have been presented in numerous courts, and there are a significant number of state appellate rulings accepting RFLP data. Similarly, the results from polymerase chain reaction (PCR)-testing have also been presented widely in courts in the United States, resulting in many appellate rulings accepting this technology. Currently, most of the appellate rulings regarding PCR-testing reference data were obtained using the AmpliType® DQα PCR Forensic DNA Amplification and Typing Kit (DQA1; Perkin-Elmer Corporation). However, there have been several more recent rulings accepting AmpliType® PM PCR Amplification and Typing Kit and D1S80 data. Extensive experience has been accumulated regarding the issues that affect the admissibility and presentation of "new and novel" test results in court. The newest form of DNA testing to become commercially available for forensic DNA analysis is STR (short tandem repeat) analysis. STR testing has been performed on DNA isolated from forensic case samples for a variety of reasons. These include: 1) increasing the chance of excluding a falsely-accused individual, 2) determining whether a sample contains a mixture of DNA from more than one individual, 3) assisting in the interpretation of data from samples containing a mixture of DNA and 4) limiting the number of individuals included as possible donors of the DNA obtained from a sample by providing increased statistical frequencies. As scientists, we can rely on our past experience when testifying to scientific data produced using the newer, commercially available STR systems.

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Our role as scientific expert witnesses is to educate the jury and/or judge regarding the type of testing that has been done, the results and conclusions of these tests, and their limitations. It may be helpful to explain the genetic basis for each type of test and the advantages and disadvantages of the systems used. For example, one advantage of PCR-based systems is that they can be used to obtain results from very small samples that do not contain sufficient material for RFLP analysis. There are two major types of variations in nuclear DNA that are used for human identity testing. One type of variation is a single-base change that occurs at a specific location in the DNA (e.g., one person has an "A" and another person has a "G" at the same position). These variations are commonly analyzed using oligonucleotide probes specific for the sequence in amplified PCR products; for example, the dot blot analysis used in the AmpliType® DQα, PM and PM + DQA1 test kits. The second type of variation arises as a result of differences in the number of blocks of tandemly repeated sequences found at a specific location in the DNA. Variations in the number of these repeats between individuals leads to length differences at specific regions. These are analyzed by electrophoresis of the DNA through a gel matrix, followed by observation of differences in the migration rates of the differently sized DNA fragments.

Blocks of large repeated DNA sequences are referred to as variable number tandem repeats (VNTRs) and are analyzed by RFLP. Variable numbers of shorter repeated DNA sequence blocks that are amplified by PCR are commonly referred to as amplified fragment length polymorphisms (AmpFLPs; e.g., D1S80). Short tandem repeats (STRs) refer to tandemly repeated blocks of very short sequences (generally two, three or four bases), and these, like AmpFLPs, are analyzed after amplification of the DNA using PCR. As STR and VNTR sequences are genetically similar, STR and D1S80 testing use a similar technology to that used for VNTR analysis. STR and D1S80 testing combine the analysis of DNA fragment length variations by gel electrophoresis with the advantage of using PCR amplification to generate multiple copies of the target DNA.

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Cellmark analysts have been to court in over 30 cases where the GenePrint™ *STR Multiplex System -CSF1PO, TPOX, TH01 (CTT Multiplex) and/or CTT in conjunction with the* GenePrint™ *Sex Identification System – Amelogenin (CTT-A) data have been presented to the trier of fact.*

These technologies and the use of STR sequences are not "new or novel" to scientists and are widely used in many areas of research and diagnostics outside of the field of forensic human identity testing.

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- Information regarding the widespread use of PCR and STR testing in other fields.
- The genetic basis for the polymorphisms observed.
- A description of the technology used and types of results obtained.
- Validation studies, including relevant publications.
- Training and experience of the scientist and the laboratory.
- Proficiency testing, controls performed, and safeguards in evidence handling and testing to ensure accurate and reliable results.

Presentations at trial have ranged from a brief description of the technology and a summary of the data to more extensive testimony that includes areas routinely covered in admissibility hearings and test results discussed in detail.

The issues raised in cross-examination have generally been similar to those raised previously for other types of PCR testing and for RFLP testing. These include whether STR testing is "new and novel," whether multiplexing compromises the assay, whether the sensitivity of PCR testing means that contamination may invalidate the results, and whether small databases are representative of larger populations. These issues can be addressed by the expert witness by citing publications detailing validation studies and databases, by the use of appropriate laboratory standard operating procedures for evidence handling and testing and for performing controls, by proper training of laboratory staff and the use of proficiency tests, and through the application of relevant guidelines such as those from TWGDAM (1) and the DNA Advisory Board.

The first appellate ruling in which STR testing was reviewed and accepted occurred in 1997 [*Commonwealth of Massachusetts v. Adam Rosier*; 425 Mass. 807, 685 N.E. 2d 739 (1997)].

CONCLUSIONS

STR test results may provide useful information in many cases where DNA testing is possible. Since the genetic analysis of STR sequences has been used widely and has been accepted by molecular biologists in many areas of study for over eight years, the use of STRs is not considered to be a new technique to the scientific community. We as scientists can be confident taking the results of STR testing to court as long as the data are supported by good laboratory practices. Our role as scientific expert witnesses is as follows:

- To be prepared with the appropriate validation studies, training and standard operating procedures, including the use of appropriate controls, proficiency testing, etc., to support the STR data.
- To work closely with attorneys and other relevant individuals to determine which cases can benefit from the use of STR testing.
- To fairly and accurately present STR data to the trier of fact.

REFERENCE

1. The Technical Working Group of DNA Analysis Methods (1995) Guidelines for a quality assurance program for DNA analysis. *Crime Laboratory Digest* **22**, 21.