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Research Preview

Jeremy Travis, Director
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Automated DNA Typing: Method of the Future?

A Summary of a Research Study Conducted by Holly A. Hammond and C. Thomas Caskey at the Baylor University College of Medicine

Although highly reliable in clinical or research applications, the standard technology used for DNA typing—known as RFLP-VNTR[1] analysis—has been less satisfactory in the forensic setting. RFLP-VNTR requires abundant and clean specimens; samples typically found at crime scenes, however, are both quantitatively and qualitatively inadequate—very small and often environmentally degraded from exposure to heat, light, and humidity. Further, police investigations call for a quicker turnaround time than is possible with the standard method, which can take months to complete.

An NIJ-sponsored project at Baylor University's College of Medicine sought a DNA typing system that overcomes the limitations of samples found at crime scenes. The project replicated the DNA sample (i.e., synthesized new DNA from existing DNA) to obtain sufficient quantities for analysis and then identified genetic markers[2] for DNA typing. That procedure, known as PCR-STR,[3] can produce reliable results with degraded specimens, is quick, and can be automated to permit the creation of a vastly improved data base of DNA profiles of convicted offenders. PCR-STR promises to extend the application of DNA typing as a powerful criminal justice tool that helps to establish, with a high degree of certitude, the guilt or innocence of suspects.

Basic facts about DNA

Understanding the importance of DNA typing to criminal investigations requires knowledge of some fundamental facts. Each molecule of DNA, the primary carrier of genetic information in living organisms, consists of a very long spiral structure that has been likened to a "twisted ladder." The handrails of the ladder string together the ladder's "rungs," which are called bases. Bases, composed of four varieties of nucleic acid, combine in pairs called "nucleotides." The sequence of these base pairs constitutes the genetic coding of DNA.

DNA in humans is found in all cells that contain a nucleus (i.e., in all except red blood cells). Each nucleated cell (with the exception of sperm and egg cells) usually contains the full complement of an individual's DNA, called the "genome," that is unvarying from cell to cell. The genome consists of approximately 3 billion base pairs, of which about 3 million actually differ from person to person. However, the base pairs that vary represent a virtually incalculable number of possible combinations. Person-to-person differences within a particular segment of DNA sequence are referred to as "alleles."

DNA typing focuses on identifying and isolating discrete fragments of these alleles in a sample and comparing one sample with another. For example, a forensic scientist might compare a semen sample retrieved from a rape victim to a DNA sample taken from a suspect. If identical fragments appear in both samples, a match is declared. To determine the likelihood of a match being mere coincidence, a particular combination of alleles is compared to the frequency with which the combination appears in the statistical population.

PCR-STR procedure

The replication process. Many of the frequently encountered and frustrating shortcomings of forensic specimens—i.e., contamination, environmental degradation, and the generally small quantity of testable material—are surmounted by PCR analysis. This technique involves extracting DNA from a small evidence sample and then replicating it through a complex operation of repeated heating and cooling cycles and exposure to an enzyme. Because each cycle doubles the quantity of DNA, the original extraction can be replicated several million times within a short period of time. By examining several locations (loci) where variation occurs, a typing profile can be produced.

Baylor researchers focused on isolating 13 STR loci, each of which contains a short region where a sequence of three, four, or five nucleotides is repeated a different number of times in different people. Samples identified by a radioactively labeled, allele-specific probe are blotted onto a membrane, according to standard PCR protocol. Each dark spot that appears can be read as "yes" or "no" to the question of whether a particular individual possesses a given allele.

When comparing DNA samples from known individuals with evidence samples, a difference of a single allele can exclude someone as the donor of that evidence sample. The more locations that show the same allele pattern, the stronger the evidence that the two samples came from the same individual. PCR amplification techniques allow DNA typing results to be obtained from even badly degraded samples, and STR analysis permits exact allele designations. Thus, PCR-STR allows samples analyzed at different times to be easily compared.

Automation. STR analysis was initially developed as a manual process, but automated methods are becoming available. Key to this technology is the use of fluorescent chemicals during the PCR process. A laser-generated fluorescent signal from the STR alleles passes information to a computer, where the collected data are analyzed to produce DNA profile information. In addition, robotic workstations are available to process DNA samples and assist with other procedures. With automation, the entire process—from DNA extraction to data interpretation—could be accomplished with little human involvement or manipulation, thereby reducing the possibility of error.

Realization and acceptance of full automation will take some time. All the pieces, however, are currently available and can be integrated. Laboratories, companies, and individuals associated with the Human Genome Project, molecular biology, and forensic science could work together to bring the newest technology in molecular analysis to the crime laboratory. Every criminal case with relevant biological evidence could be analyzed—without the time, cost, and technical limitations that thwart the full potential of forensic DNA typing. Most importantly, accurate and rapid DNA typing capability advances the foremost criminal justice objective: to shield the innocent and convict the guilty.

Notes

1. This acronym stands for restriction fragment length polymorphism-variable number of tandem repeats.

2. A marker is a gene with a known location on a chromosome and a clear-cut phenotype (physical appearance or functional expression of a trait) that is used as a point of reference in the mapping of other locations.

3. PCR stands for polymerase chain reaction and is the technique used to replicate DNA. STR (short tandem repeat) refers to the region on a DNA strand where a sequence of three, four, or five nucleotides is repeated a different number of times in different people—the so-called STR loci.

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