Appendix C
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Appendix D
Acronymns and Glossary

Acronyms

A —adenine
AABB —American Association of Blood Banks
AAFS —American Academy of Forensic Sciences
AFIS —Automated Fingerprint Identification Systems
APB —Advisory Policy Board (NCIC)
ASCLD —American Society of Crime Laboratory Directors
ASHG —American Society of Human Genetics
ASHI —American Society of Histocompatibility and Immunogenetics
BJS —Bureau of Justice Statistics (DOJ)
C —cytosine
CAC —California Association of Criminalists
CACLD —California Association of Crime Laboratory Directors
CLIA —Clinical Laboratory Improvement Amendments of 1988
CTS —Collaborative Testing Services
DHHS —U.S. Department of Health and Human Services
DNA —deoxyribonucleic acid
Doc —U.S. Department of Commerce
DOJ —U.S. Department of Justice
FBI —Federal Bureau of Investigation (DOJ)
FSA —Forensic Science Associates
FSF —Forensic Science Foundation
FSRTC —Forensic Science Research and Training Center (FBI/DOJ)
G —guanine
HCFA —HealthCareFinance Administration (DHHS)
HGML —Human Gene Mapping Library
HLA —human leukocyte antigen
ISFH —International Society for Forensic Haemogenetics
mtDNA —mitochondrial DNA
NCIC —National Crime Information Center
NIH —National Institutes of Health
NIJ —National Institute of Justice
NIST —National Institute of Standards and Technology (DOC)
NLETS —National Law Enforcement Telecommunications System
OMIM —On-Line Mendelian Inheritance in Man
OTA —Office of Technology Assessment
PCR —polymerase chain reaction
PDB —Protein Data Bank
PIR —Protein Identification Resource
RFLP —restriction fragment length polymorphism
SSN —social security number
T —thymine
Triple I —Interstate Identification Index (NCIC)
TWGDAM-Technical Working Group on DNA Analysis Methods (FBI/DOJ)
VNTR —variable number of tandem repeats

Glossary of Terms

Allele: Alternative form of a genetic locus (e.g., at a locus for eye color there might be alleles resulting in blue or brown eyes); alleles are inherited separately from each parent.

Autoradiogram: **An x-ray** film image showing the position of radioactive substances. Sometimes called ''Autocad. ''

Autoradiograph: See *autoradiogram.*

Autoradiography: A technique for identifying radioactively labelled molecules or fragments of molecules.

Autosome: Chromosome not involved in sex determination. In a complete set of human chromosomes, there are 44 autosomes (22 pairs).

Band shift: The phenomenon of DNA fragments in one lane of a gel migrating slower or faster than identical fragments in another lane. As visualized on an autoradiogram, the overall patterns would be the same, but out of register. Factors responsible for band shift include contaminants, salt concentration, and DNA concentration.

Base pair: Two complementary nucleotides (adenosine and thymidine or guanosine and cytidine) held together by weak bonds. Two strands of DNA are held together in the shape of a double helix by the bonds between base pairs.

Blot: See *Southern blot.*

Cell: The smallest component of life capable of independent reproduction and from which DNA is isolated for forensic analysis.

Chromosome: A threadlike structure that carries genetic information arranged in a linear sequence. In humans, it consists of a complex of nucleic acids and proteins.

Controls: Tests designed to demonstrate that a procedure worked correctly and performed in parallel with experimental samples. Controls yield certain expected results; when the observed results for the controls deviate from what is expected, then the results for the case samples cannot be considered reliable.

Deoxyribonucleic acid (DNA): The molecule that encodes genetic information. DNA is a double-stranded helix held together by weak bonds between base pairs of nucleotides.

DNA: See *deoxyribonucleic acid.*

DNA band: Referring to the visual image, e.g., on a autoradiograrn or an ethidium bromide stained gel, that represents a particular DNA fragment.

DNA probe: Short segment of DNA that is labeled with a radioactive or other chemical tag and then used to detect the presence of a particular DNA sequence through hybridization to its complementary sequence.

DNA sequence: Order of nucleotide bases in DNA.

Electrophoresis: Technique used to separate molecules such as DNA fragments or proteins. In forensic uses of DNA tests, electric current is passed through a gel, usually composed of a substance called agarose, and the fragments of DNA are separated by size. Smaller fragments will migrate farther than larger pieces.

Enzyme: A protein that acts as a catalyst, speeding the fate at which a biochemical reaction proceeds, without being permanently altered or consumedly the reaction so that it can be used repeatedly.

Gel: The semi-solid matrix used in electrophoresis to separate molecules. In forensic DNA analysis, the substance usually used is agarose, although acrylamide can also be used

Gene: The fundamental unit of heredity; an ordered sequence of nucleotide base pairs to which a specific product or function can be assigned.

Genome: All the genetic material in the chromosomes of a particular organism; its size is generally given as its total number of base pairs.

Genotype: The genetic constitution of an organism, as distinguished from its physical appearance, or phenotype.

Hardy-Weinberg equilibrium: In a large, random, intrabreeding population, not subjected to excessive selection, migration, or mutation, the gene and genotype frequencies will remain constant overtime, so that for most single-locus probe analyses, the likelihood of being a homozygote (one band) with genotype $a_i a_i$ will be (pl)2, where p_i is the frequency in the population of allele a_i. For heterozygotes with two bands, the chance that a person will have genotype a_i/a_2~ will be $2p_i/2p_2$, where p_1 and p_2 are the respective frequencies of how often bands a_1 and a_2 occur.

Heterozygous: Having two different alleles at a particular locus. For most forensic DNA probes and individuals, if the person is heterozygous at the locus the probe detects, the autoradiogram displays two bands.

HLA: See *human leukocyte antigen.*

Homozygous: Having the same allele at a particular locus. For most forensic DNA probes and individuals, if the person is homozygous at the locus the probe detects, the autoradiogram displays a single band.

Human leukocyte antigen (HLA): Located on the surface of most cells, except blood cells, these protein-sugar structures differ among individuals and are important for acceptance or rejection of tissue or organ grafts and transplants. The locus of one particular class of these antigens, HLA *DQx-1,* is useful for forensic analysis using PCR.

Hybridization: The process of joining two **complementary strands** of DNA, or of DNA and RNA, together to form a double-stranded molecule.

Junk DNA: Sequence of DNA for which no specific coding function has yet been assigned. Also called noncoding DNA.

Linkage disequilibrium: The phenomenon of a specific allele of one locus being associated with an allele of another locus on the same chromosome with a frequency greater than expected by chance.

Locus: A specific, physical position on a chromosome.

Marker: A gene with a known location on a chromosome and a clear-cut phenotype that is used as a point of reference when mapping another locus; or, referring to DNA fragments of known base pair length run on gels from which the size of unknown DNA sample fragments can be determined.

Mitochondria: Structures, or organelles, found within a cell that are responsible for generating the cell's (and hence organism's) energy. Mitochondria contain DNA molecules that are inherited only from an individual's mother. An individual and his or her siblings will share the same mitochondrial DNA pattern-the pattern of their mother (and other maternal relatives, including the maternal grandmother and maternal aunts and uncles).

Multilocus probe: DNA probe that detects genetic variation at multiple sites in the genome. An autoradiogram of a multilocus probe application yields a complex, stripe-like pattern of 30 or more bands per individual. Compare *Single-1ocus probe.*

Nucleotide: The unit of DNA consisting of one of four bases—adenine, guanine, cytosine, or thymine-attached to a phosphate-sugar group. The sugar group is deoxyribose in DNA. (In RNA, the sugar group is ribose and the base uracil substitutes for thymine.)

Phenotype: The appearance of an individual or the observable properties of an organism that result from the interaction of genes and the environment.

Polymerase chain reaction (PCR): An in vitro process, through which repeated cycling of the reaction reproduces a specific region of DNA, yielding millions of copies from the original.

Polymorphism: The existence of more than one form of a genetic trait.

Probe: In forensic applications, a short segment of DNA tagged with a reporter molecule, such as radioactive phosphorus (32P), used to detect the presence of that particular complementary DNA sequence.

Protein: A biological molecule whose structure is determined by the sequence of nucleotides in DNA. Proteins are required for the structure, function, and regulation of cells, tissues, and organs in the body. Some traditional forensic genetic markers are proteins.

Recombinant DNA technology: Processes used to form a DNA molecule through the union of different DNA

molecules, but now commonly used to refer to any techniques that directly examine DNA.

Replication: The synthesis of new DNA from existing DNA. PCR is an in vitro technology based on the principles of replication.

Restriction endonuclease: An enzyme that has the ability to recognize a specific DNA sequence and cut it at that sequence.

Restriction enzyme: See *restriction endonuclease.*

Restriction fragment length polymorphism (RFLP): Variations in the size of DNA fragments produced by a restriction endonuclease at a polymorphic locus.

RFLP analysis: DNA technique using single-locus or multilocus probes to detect variation in the DNA sequence by revealing size differences in DNA fragments produced by the action of a restriction enzyme. See *restriction fragment length polymorphism.*

Serology: Scientific discipline concerned with the study of body fluids.

Single-locus probe: DNA probe that detects genetic variation at only one site in the human genome. An autoradiogram using one single-locus probe usually displays one (homozygote) or two (heterozygote) bands. Compare *multilocus probe.*

Southern blot: The nylon membrane to which DNA has adhered after the process of Southern blotting.

Southern blotting: The technique for transferring DNA fragments separated by electrophoresis from the gel to a nylon membrane, to which DNA probes that detect specific fragments can then be applied.

Standardization: In forensic uses of DNA tests, refers to a national system that uses a single restriction endonuclease with, in whole or part, certain designated DNA probes; critical to databanking considerations. Compare *standards.*

Standards: Criteria established for quality control and quality assurance; or, known test reagents, such as molecular weight standards. Compare *standardization.*

Tandem repeats: Multiple copies of the identical (or nearly identical) DNA sequence arranged in direct succession at a particular site on a chromosome. See *variable number of tandem repeats.*

Tag polymerase: DNA polymerase-the enzyme used to form double-stranded DNA from nucleotides and a single-stranded DNA template-isolated from the bacterium *Thermus aquaticus,* which normally lives in hot springs. *Taq* polymerase can withstand the high temperatures required in the repeating cycles of PCR.

Variable number of tandem repeats (VNTR): Repeating units of a core DNA sequence, for which the core number varies between individuals, thus providing the basis for RFLP analysis. See *tandem repeats.*