

Helpful Resources on Forensic DNA Testing

Textbooks:

Butler, J.M. (2005) *Forensic DNA Typing: Biology, Technology, and Genetics of STR Markers*, 2nd edition. New York: Elsevier

Michaelis, R.C., et al. (2008) *A Litigator's Guide to DNA: From the Laboratory to the Courtroom*. San Diego: Elsevier Academic Press.

Goodwin, W., et al. (2007) *An Introduction to Forensic Genetics*. Hoboken, New Jersey: Wiley.

Kobilinsky, L. et al. (2005) *DNA: Forensic and Legal Applications*. Hoboken, New Jersey: Wiley.

Websites:

<http://www.ies.krakow.pl/multilingua/welcome/> (see <http://www.enfsi.eu/>)

Multilingua Lexicon established by the European Network of Forensic Science Institutes (ENFSI) with specific words used in forensic science (DNA, fingerprints, statistics, etc.) translated into 16 languages (English, Spanish, Czech, Dutch, French, German, Greek, Hungarian, Italian, Latvian, Norwegian, Polish, Portuguese, Slovak, Swedish, and Turkish)

http://www.ornl.gov/sci/techresources/Human_Genome/elsi/forensics.shtml

some general information on forensic DNA testing

<http://www.dna.gov>

the U.S. Department of Justice website started by the President's DNA Initiative

DNA: A Prosecutor's Practice Notebook: <http://dna.gov/training/prosecutors-notebook/>

Principles of Forensic DNA for Officers of the Court: <http://www.dna.gov/training/otc/>

What Every Law Enforcement Officer Should Know About DNA Evidence: First Responding Officers and Investigators and Evidence Technicians: <http://dna.gov/training/letraining/>

Crime Scene and DNA Basics: <http://dna.gov/training/evidence/>

Laboratory Orientation and Testing of Body Fluids and Tissues: <http://dna.gov/training/forensicbiology/>

Laboratory Safety Programs: <http://www.dna.gov/training/labsafety/>

DNA Extraction and Quantitation: <http://dna.gov/training/extraction/>

DNA Amplification: <http://dna.gov/training/amplification/>

Amplified DNA Product Separation: <http://dna.gov/training/separation/>

STR Data Analysis and Interpretation: <http://dna.gov/training/strdata/>

Population Genetics and Statistics: <http://www.dna.gov/training/populations>

Communication Skills, Report Writing, and Courtroom Testimony: <http://www.dna.gov/training/communicating>

Non-STR DNA Markers: SNPs, Y-STRs, LCN and mtDNA: <http://www.dna.gov/training/markers/>

Advanced and Emerging DNA Techniques and Technologies: <http://www.dna.gov/training/Technology>

STRBase: <http://www.cstl.nist.gov/biotech/strbase/>

resource maintained by NIST on forensic short tandem repeat DNA testing markers and methods

<http://www.cstl.nist.gov/biotech/strbase/weblink.htm>.

DNA Resource.com: <http://www.dnaresource.com>

DNA Policy Community Site: <http://www.dnapolicy.net/>

DNA Litigation Legal Support Page: http://www.denverda.org/DNA/DNA_INDEX.htm

FBI's Combined DNA Index System (CODIS): <http://www.fbi.gov/hq/lab/codis/index1.htm>

National Clearinghouse for Science, Technology, and the Law (NCSTL): <http://www.ncstl.org/home>

National Forensic Science Technology Center (NFSTC): <http://www.nfstc.org>

National District Attorneys Association (NDA): <http://www.ndaa-apri.org/index.html>

National Institute of Justice (NIJ): <http://www.ojp.usdoj.gov/nij/>

National Legal Aid and Defender Association (NLADA): [http://www.nlada.org/Defender/forensics/for lib/Index/DNA](http://www.nlada.org/Defender/forensics/for_lib/Index/DNA)

Basic Glossary of Forensic DNA Terms

Sources of definitions: <http://www.dna.gov/glossary>, <http://www.cstl.nist.gov/biotech/strbase/glossary.htm>, and John Butler FDT3e

ABI: Applied Biosystems Inc.; an instrument and reagent manufacturing company based in Foster City, California

ABI 310 Genetic Analyzer: a capillary electrophoresis instrument sold by Applied Biosystems and widely used throughout the forensic DNA community since its introduction in 1995; an ABI 310 uses a single capillary filled with a viscous polymer solution to separate DNA molecules based on their relative size; many labs have now adopted a multi-capillary instrument such as the ABI 3100 or ABI 3130xl Genetic Analyzer

ABI 3100 Genetic Analyzer: a multi-capillary electrophoresis instrument sold by Applied Biosystems since 2001; 4-capillary and 16-capillary versions are available; newer versions include a mechanical polymer pump instead of a syringe and are named ABI 3130 or ABI 3130xl

Accreditation: the formal process by which a laboratory is evaluated, with respect to established criteria, for its competence to perform a specified kind of measurement; the evaluation takes place through an audit by an outside, qualified agency and helps provides confidence that the laboratory meets minimum professional standards for general operations

Accuracy: the degree of agreement or conformity of a measured value with its actual (true) value

Allele: An alternative form of a gene or a section of DNA at a particular genetic location (locus); typically multiple alleles are possible with STR markers

Allele Frequency: The proportion of a particular allele among the chromosomes carried by individuals in a population.

Amelogenin: a commonly used sex-typing genetic marker because the gene, which codes for tooth enamel, occurs on both the X and Y chromosomes

Amplification: An increase in the number of copies of a specific DNA fragment; can be in vivo or in vitro.

Amplicon: the product of polymerase chain reaction DNA amplification

Analyst: an individual trained and qualified to report DNA results

Artifact: any non-allelic product of the amplification process (stutter or minus A) or anomaly of the detection process (pull-up or spike)

Autosome: A chromosome not involved in sex determination. The diploid human genome consists of 46 chromosomes, 22 pairs of autosomes, and one pair of sex chromosomes (the X and Y chromosomes).

Basepair: Two complementary nucleotides joined by hydrogen bonds; basepairing occurs between A and T and between G and C.

Base sequence: The order of nucleotide bases in a DNA molecule.

Buccal swab: a relatively non-invasive technique of scrapping the inside of a mouth to collect cells from the inner cheek lining

Capillary electrophoresis (CE): an electrophoretic technique for separating DNA molecules by their size based on migration through a narrow glass capillary tube filled with a liquid polymer

Chromosome: The structure by which hereditary information is physically transmitted from one generation to the next.

CODIS: Combined DNA Index System, which under the direction of the FBI Laboratory, is the software architecture that runs the U.S. national DNA database

CODIS loci: a set of 13 STRs required for inclusion of a DNA profile at the national level in CODIS

Complementary sequences: Nucleic acid base sequences that form a double-stranded structure by matching base pairs; the complementary sequence to G-T-A-C is C-A-T-G.

Complete profile: a full DNA result with values being obtained from all attempted loci

Concordance: obtaining the same value when testing multiple times

Conservative: an assignment of the weight of evidence that is believed to favor the defense

Degradation: the fragmenting, or breakdown, of DNA by chemical or physical means

Deoxyribonucleic acid (DNA): The genetic material of organisms, usually double-stranded; a class of nucleic acids identified by the presence of deoxyribose, a sugar, and the four nucleobases.

Detection limit: the smallest amount of some component of interest that can be measured by a single measurement with a stated level of confidence

Differential extraction: a DNA extraction procedure where the sperm cells are physically separated from the DNA of other cells before the sperm DNA is isolated; generally results in a sperm and non-sperm (epithelial) fraction

DNA databank: a repository of stored bloodstain cards or DNA samples used to generate DNA profiles

DNA database: a computer repository of DNA profiles

DNA profiling or typing: a string of values (numbers or letters) compiled from the results of DNA testing at one or more genetic markers

DNA sequence: The relative order of base pairs, whether in a fragment of DNA, a gene, a chromosome, or an entire genome.

Electrophoresis: a technique in which molecules are separated by their velocity in an electric field

Epithelial cells: skin cells, vaginal cells, or other cells that are normally found on an inner or outer body surface

Eukaryote: an organism with cells containing a nucleus

Evidence sample: biological sample collected from a crime scene or people or objects associated with a crime scene; sometimes referred to as the "Q" or question sample

Exclusion: a DNA test result indicating that an individual is excluded (does not match at the tested DNA locations) as the source of the DNA evidence; in a criminal case, "exclusion" does not necessarily equate to "innocence"

Fluorescence: the emission of light from a molecule following its excitation by light energy; in the context of DNA analysis, different fluorescent dyes permit simultaneous detection of similar size PCR products through fluorescence emission in different colors

Forensic science: the application of scientific knowledge to questions of civil and criminal law, typically through presentation of results from evidence in court

Gene: the basic unit of heredity; a sequence of DNA nucleotides on a chromosome

Genetics: the study of the patterns of inheritance of specific traits

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

Genotype: the genetic makeup of an organism, as characterized by its physical appearance or phenotype

Guidelines: a set of general principles used to provide direction and parameters for decision making

Hereditry: the transmission of characteristics from one generation to the next

Heterozygosity: The presence of different alleles at one or more loci on homologous chromosomes.

Homologies: Similarities in DNA or protein sequences between individuals of the same linear sequences, each derived from one parent.

Homologous chromosomes: A pair of chromosomes containing the same linear gene sequences, each derived from one parent.

Identifier: a multiplex STR typing kit from Applied Biosystems that co-amplifies 15 STRs and the sex-typing marker amelogenin; the STR loci amplified are D8S1179, D21S11, D7S820, CSF1PO, D3S1358, TH01, D13S317, D16S539, D2S1338, D19S433, VWA, TPOX, D18S51, D5S818, and FGA

Inconclusive: A situation in which no conclusion can be reached regarding testing done due to one of many possible reasons (e.g., no results obtained, uninterpretable results obtained, no reference sample available for testing)

Internal size standard (ISS): specific DNA fragments of known length that are used to size other DNA fragments in a sample being measured

Intimate sample: a biological sample obtained directly from a victim's body such as a vaginal swab

In vitro: Outside a living organism

Kinship analysis: DNA evaluations using biological relatives to predict expected genotypes in missing individuals; serves as an indirect form of human identification when no direct reference samples are available

Linkage: The proximity of two or more markers (genes, etc.) on a chromosome; the closer together the markers are, the lower the probability that they will be separated during DNA repair or replication process, and hence the greater the probability that they will be inherited together.

Locus (pl. loci): The specific physical location of a gene on a chromosome.

Marker: a gene or specific DNA sequence of known location on a chromosome; used as a point of reference in the mapping of other loci

Match: when genetic profiles show the same types at all loci tested and no unexplainable differences exist

Mitochondrial DNA (mtDNA): a small, circular DNA molecule located in the mitochondria that contains approximately 16,500 nucleotides; the abundance of hundreds of copies of mtDNA in each cell make it useful with samples originating from limited or damaged biological material

Multiplex PCR: co-amplification of multiple regions of a genome with more than one set of primers; enables information from the different target sequences to be collected simultaneously

Mutation: any inheritable change in DNA sequence; an alteration or change of an allele at a genetic locus resulting in genetic inconsistency between a biological parent and offspring

Non-sperm cell fraction: the portion of a sample produced during differential extraction containing DNA from non-sperm cells; generally epithelial cells from the female victim in the case of sexual assault evidence

Nucleotide: A unit of nucleic acid composed of phosphate, ribose or deoxyribose, and a purine or pyrimidine base.

Nucleus: The cellular organelle in eukaryotes that contains the genetic material.

Polymerase chain reaction (PCR): An in vitro process that yields millions of copies of desired DNA through repeated cycling of a reaction involving the DNA polymerase enzyme.

Polymorphism: Difference in DNA sequence among individuals. Genetic variations occurring in more than 1% of a population would be considered useful polymorphisms for linkage analysis.

Population: A group of individuals residing in a given area at a given time.

Power of discrimination: the potential power of a genetic marker or set of markers to differentiate between any two people chosen at random; equal to one minus the sum of the square of the genotype frequencies

PowerPlex 16: a multiplex STR typing kit from Promega Corporation that co-amplifies 15 STRs and the sex-typing marker amelogenin; the STR loci amplified are D3S1358, TH01, D21S11, D18S51, Penta E, D5S818, D13S317, D7S820, D16S539, CSF1PO, Penta D, VWA, D8S1179, TPOX, and FGA

Precision: a measure of the closeness of results when experiments are repeated

Presumptive test: an initial examination of evidence to indicate the possible source of the sample (e.g., blood, saliva, semen, etc.); usually followed up by a confirmatory assay or DNA analysis

Primer: a short preexisting polynucleotide chain, usually 18-30 bases long, which targets a specific region of the template DNA and allows a DNA polymerase to initiate synthesis of a complementary strand

Probability of exclusion (PE): the percentage of the population that can be excluded as potential contributors to a DNA mixture result

Product rule: the combination of genotype frequency estimates from multiple loci through multiplying the individual locus genotypes together

Proficiency test: a quality assurance measure used to monitor performance of an analyst and identify areas in which improvement may be needed; can be internal (produced by the agency undergoing the test) or external (produced by an outside test provider); external proficiency tests can be either open or blind

Profiler Plus: a commercial STR typing kit from Applied Biosystems that utilizes multiplex PCR to amplify and provide genetic information for 9 STRs and the sex-typing marker amelogenin; the STR loci examined are D3S1358, VWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, and D7S820

Quality assurance (QA): a system of activities whose purpose is to provide to the producer or user of a product or service the assurance that it meets defined standards of quality

Random Match: a match in the DNA profiles of two sample, where one is drawn at random from the population

Random Match Probability (RMP): the chance of a specific profile occurring in a specific population based on observed allele frequencies for that population

Reference sample: a sample (typically blood or buccal swab) taken from a known person that is used for comparison purposes to an evidentiary sample; sometimes referred to as the "K" or known sample

Reproducibility: the ability to obtain the same result when a test or experiment is repeated

Sex chromosomes (x and y chromosomes): Chromosomes that are different in the two sexes and involved in sex determination.

Short tandem repeats (STR): multiple copies of an identical (or similar) DNA sequence arranged in direct succession where the repeat sequence unit is 2 bp to 6 bp in length; because STRs generally occur in the "junk" DNA outside of the constraints of genes, the number of repeat units can vary between individuals in an accordion-like fashion

Single nucleotide polymorphism (SNP): any polymorphic variation at a single nucleotide; most SNPs are biallelic (e.g., either C or T) with the minor allele being observed at least 1% of the time

Sperm cell fraction: the portion of a sample produced during differential extraction containing DNA from sperm cells; generally from the male perpetrator in the case of sexual assault evidence

Standards: criteria established for quality assurance purposes that place specific requirements on laboratories and analysts; also refers to well-characterized samples that can aid calibration of measurements

SWGDM: Scientific Working Group on DNA Analysis Methods; formerly known as TWGDM, Technical Working Group on DNA Analysis Methods; an FBI sponsored group that develops quality assurance standards and guidelines for laboratories in the United States

Validation: the process by which a sample, measurement method, or a piece of data is deemed useful for a specified purpose; the process of extensive and rigorous evaluation of DNA methods before acceptance for routine use

Verification: confirmation by examination that specified requirements have been met

X-chromosome: one of the sex chromosomes; normal females possess two copies and males one

Y-chromosome: one of the sex chromosomes; normal males possess one copy and females none

Y-STR: short tandem repeat markers found on the Y-chromosome that enable male-specific DNA testing

Zygote: cell formed when the nuclear DNA from a father's sperm cell combines with nuclear DNA in a mother's egg restoring the diploid chromosome count