Presidential Task Force on
Missing and Murdered American Indians and Alaska Natives
Operation Lady Justice

Understanding DNA Testing and Reporting

Utilizing technological advances in DNA testing and leveraging database searches in the Combined DNA Index System (CODIS) have, in part, driven testing of biological evidence in violent crime cases, including cold cases. If there is documentation—such as a police report—that a crime occurred, then any foreign DNA obtained from evidence may be valuable to help identify a perpetrator and link that individual to other crimes through a database search.

Evidence from cold cases still may be suitable for DNA testing—even if it was previously tested using biological fluid screening only or early DNA tests, such as restriction fragment length polymorphism (RFLP). These examination results may even help determine whether there is viable evidence to proceed with additional testing using newer DNA technology. Even if evidence was partially or mostly consumed during prior testing, extracts and other byproducts of previous processes may be used.

Challenge: Obtain CODIS-eligible DNA from serology-negative cases

- DNA quantification and amplification kits are more sensitive in detecting DNA than typical biological screening processes.
- Enzymes used in serological detection break down over time, but viable DNA may still be possible.
- DNA extracts can provide viable DNA to test using the following:
    - Expanded short tandem repeat (STR) loci kits,
    - Y-STR loci kits, and
    - advanced DNA testing applications.

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STR Technology: Prominent DNA Test

DNA is an initialism for deoxyribonucleic acid. Found within a cell's nucleus in the human body, DNA is known as the biological blueprint of life. Nuclear DNA is passed down from generation to generation, with half a person's DNA coming from each parent. Humans are very similar, but—with the exclusion of monozygotic multiple births (e.g., identical twins)—there are small, though unique, differences in our DNA that identify us. Forensic DNA testing focuses on the parts of the DNA that are different among humans.

Common forensic nuclear DNA tests look at STRs in our DNA, in which:

- **short** refers to a small segment of DNA,
- **tandem** refers to being right next to each other, and
- **repeats** means replication.

A forensic DNA test examines the number of times an STR is replicated; each replication is called an allele (see Example 1). Because half a person's DNA comes from each parent, each person will have two alleles at each location tested (see Example 2). A DNA profile is created when many STRs (typically more than 20 locations) are examined. The DNA profile also indicates whether the individual is female (X,X) or male (X,Y).

Example 1. STR Profile at Three Marker Locations

STR Location	Allele 1	Allele 2
D8S1179	10	12
FGA	24	24
Amelogenin	X	Y

Note: In Example 1, D8S1179 is the name of an STR location. The results at this location are 10,12 (see Example 2). FGA is a second STR location, and the results are 24,24. Amelogenin is a sex-determining marker; the results X,Y indicate the DNA is from a male.

Example 2. STR Structure, D8S1179

TCTA-TCTA-TCTA-TCTA-TCTA-TCTA-TCTA-TCTA-TCTA-TCTA = 10 repeats
TCTA-TCTA-TCTA-TCTA-TCTA-TCTA-TCTA-TCTA-TCTA-TCTA-TCTA-TCTA = 12 repeats

STR technology is utilized widely in forensics. With STRs, data can be obtained from a small amount of sample (i.e., a pin drop), the regions are stable but also highly variable among humans, and the regions can be targeted simultaneously for efficiency. These features also make this technology popular for paternity testing, ancestry testing, and human identification in mass disasters and missing person cases.

Expanded DNA Testing: Y Chromosome and Mitochondrial DNA

Other types of DNA tests can be encountered in a forensic setting. Y-chromosome testing looks at STR regions on the Y chromosome, which is passed down through the paternal lineage (i.e., from father to son only). This testing can be useful when very little male DNA is detected in the presence of high amounts of female DNA. By focusing on the male DNA, forensic examiners can develop a Y-STR profile—essentially ignoring the impact of the female DNA. This makes Y-chromosome testing a viable option for detecting low levels of male DNA in forensic evidence, such as sexual assault kits. Y-STR analysis is an effective tool for resolving inconclusive STR results and determining the number of male contributors in a mixture.

Another type of DNA test uses mitochondrial DNA (mtDNA). Instead of looking at nuclear DNA, pieces of mtDNA—found within the cell but outside the nucleus—are sequenced to create an mtDNA profile. This type of DNA is passed down maternally (i.e., from mother to her children) and is useful for testing hair shafts (where nuclear DNA is limited) or skeletal remains, which often have compromised nuclear DNA. Due to inheritance patterns, Y-STR and mtDNA profiles cannot uniquely identify an individual, but they can provide very important information to help with an investigation.

DNA: The Laboratory Process

A person has the same DNA in all of their cells. DNA usually can be obtained from biological fluids (e.g., blood, saliva, semen); hair roots; skin cells; tissues; and skeletal elements. Consequently, evidence submitted for possible DNA testing can vary greatly (see Example 3). A forensic examiner may attempt to identify a possible biological fluid or submit items directly for DNA testing. Using C0020hemicals, the examiner will remove possible DNA from the submitted substrate (e.g., swab) and purify it. Next, the amount of human DNA retained from the item is determined; at this stage, the amount of male DNA present also can be established. If the amount of recovered DNA is below established detection limits, laboratories may choose to not proceed with further testing. However, by processing further, DNA is copied by targeting the STR regions that are different among humans. The STR regions are separated, and the forensic examiner uses computer software to record each location's test result and summarize the results and any comparisons, conclusions, and statistics in a forensic case report.

Example 3. Items Commonly Submitted for DNA Testing

- Bedding
- Bones
- Bottles, straws, cups
- Cigarette butts
- Clothing (e.g., hats, shirts, pants, sneakers, underwear, gloves)
- Condoms and wrappers
- Fingernail scrapings
- Hair
- Ligatures
- Sexual assault evidence collection kits (e.g., vaginal swabs, anal swabs, oral swabs)
- Swabs from firearms
- Swabs from surfaces (e.g., window, steering wheel, door handle)
- Swabs of possible stains (e.g., blood, saliva, semen)
- Weapon handles

DNA Reports and Conclusions

Forensic DNA reports have common, standardized elements that include report date, case identifier, description of the technology, DNA locations tested or chemistry utilized, description of the evidence examined, results, disposition of evidence, and the signature and title of the person authorizing the report. When applicable, conclusions and a quantitative or qualitative interpretation statement are included. If the case was screened for biological fluids (e.g., semen, blood, saliva), a section or separate report will explain the screening, results, and conclusions—as well as if the item proceeded to, or is recommended for, DNA testing. Depending on the amount and quality of DNA, the result may have data at every location tested (i.e., a full profile) or data at some of the locations tested (i.e., a partial profile). If no DNA is detected—for example, no DNA is deposited on a tested item, or the DNA is degraded completely due to varying conditions over time, temperature, and humidity—there will be no detectable results. Profiles that contain data from more than one individual are referred to as DNA mixtures. Because one person has two alleles at each STR location, three or more different alleles detected at a single location could indicate multiple contributors. The totality of the profile is used to determine the results and make conclusions.

The DNA profile from an evidence item can be compared to known profiles obtained from the victim, suspect, or elimination samples. Results are commonly referred to as inconclusive, excluded, or included. When a result is inconclusive, there is typically not enough information, or the information is too complex to make a definitive conclusion; the DNA from that item is not reliable for making comparisons. An exclusion supports that a known profile cannot be contained within the profile generated from the evidence item, whereas an inclusion supports that a known profile cannot be omitted from the DNA profile generated from the evidence item. Sometimes an inclusion also will be called a match when there is a single DNA profile from one individual. An inclusion or match always should be supported with a quantitative statistical calculation that helps to explain the rarity of the inclusion (e.g., a random match probability or likelihood ratio).

Lack (or presence) of DNA should always be examined within the totality of all evidence in an investigation. When there is an inclusion, a quantitative statistic represents the rarity of the DNA profile and cannot convey the chance that an individual committed or did not commit the crime. Thus, DNA cannot convey guilt or innocence. Currently, DNA evidence cannot determine the DNA's or the donor's age, but research is being conducted in these areas.

Author

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