What Your DNA Report Means To You

Criminalists in all sections of the laboratory must deal with scientific concepts on a daily basis in their routine casework. Moreover, with science also comes highly specific terminology—those words and phrases that require a dictionary and a college professor in order to comprehend. Criminalists, however, cannot escape the fact that technical wording is necessary in reports to convey accurate conclusions. Often times, the DNA section is in the spotlight concerning the particular wording in a report. Therefore, the DNA section has compiled definitions of the more commonly used expressions, terms, and concepts used in reports to assist our submitting agencies in understanding what their lab results are telling them.

**DNA**: refers to deoxyribonucleic acid; DNA is the genetic blueprint in most body cells that makes people the same (two arms, two eyes, etc.), yet different (blue eyes, red hair, etc.). Studies indicate that 99.9% of human DNA is identical from one person to the next. The 0.1% that is genetically different among individuals is where the crime lab focuses. Ultimately, these variances allow us to include or eliminate participants associated with crimes.

**Gene**: code area for a particular trait (such as hair color).

**Genome**: a person’s entire genetic code, from beginning to end.

**Locus**: describes the location of a particular gene on the DNA chain; loci is the plural form of locus.

**Allele**: one of several alternative forms of a gene at a locus, such as the A, B, AB, and O alleles for blood typing. In forensic analysis, primarily one to two alleles are detected at each locus for one person. The distinct combination of alleles over the entire genome makes each individual unique.

**Profile**: a visual representation of a person’s unique pattern of alleles. Software assigns numbers to the alleles present at each of the 13 core loci we examine (locus names are available upon request). Generating a profile allows us to compare one profile to another to look for similarities or differences. Sometimes with degraded or small amounts of DNA, we can only generate a partial profile, where less than the 13 loci are present. However, this may be useful information for eliminations, etc.

**Polymerase chain reaction (PCR)**: a step in generating a profile where a small amount of DNA is copied many times to obtain the amount needed for analysis.

**Applied Biosystems**: a vendor who manufactures chemicals used to analyze DNA. Vendors put these chemicals into specially prepared kits for our convenience. Currently, we use kits named Profiler Plus and COFiler. Profiler Plus provides 9 of the 13 loci we use, and COFiler provides the other 4. Accreditation guidelines require us to report the method we use to analyze the DNA. Vendors and kit names may change from time to time as technology progresses.

**CODIS**: refers to the FBI's Combined DNA Index System. This is the DNA database which stores DNA profiles from unknown crime samples, missing persons, and convicted offenders. CODIS compares these profiles against one another for possible matches (hits).

**DNA Typing**: also known as profiling; refers to the process of generating a profile.

**Standard**: a DNA sample (usually blood or buccal cells) collected directly from a person associated with a crime: a suspect, victim, or even a person who may need to be eliminated as a participant in the crime. All standards must be individually labeled as to who gave the sample. If a standard is not available, contact the lab to explore other options.

**Blood Standard**: a blood sample drawn into a blood tube. The DNA section prefers purple capped blood tubes since they contain the appropriate preservative for blood to prevent degradation. The Toxicology section requires gray capped blood tubes for analysis.

**Buccal Standard**: a swabbing of the inside of the mouth on the cheek. Actual skin cells from the inside cheek are required for DNA, not the saliva. Saliva, like sweat, is a fluid and contains no cells and, therefore, no DNA. Saliva, however, moistens the swab.
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and aids in cell collection. These swabs, just like any swabs collected from a crime scene, must be dried to prevent mold formation. Place the swabs in an envelope or paper bag—nothing plastic or airtight.

screening: a technique used to locate blood or semen stains or other potential sources of DNA. These tests do not confirm actual blood or semen, but assist in focusing on what could possibly be blood or semen. Screening is also referred to as presumptive testing.

confirmation: final testing to identify the presence of semen or human blood.

Semen confirmation consists of one test specific for a protein produced by the prostate gland in a male.

Human blood confirmation consists of two tests. One test determines the presence of blood. The other test determines the presence of human protein, which indicates that the stain is of human origin.

If there is a very small amount of sample, confirmation is not conducted, and the stain proceeds directly to the DNA step. During confirmation, a portion of the sample is consumed and is gone forever. Therefore, generating a DNA profile is often more important than consuming a stain for confirmation.

comparison: visually placing an unknown profile next to a known profile to determine how many alleles, if any, are in common between the profiles.

consistent: a profile that matches another profile exactly at the 13 loci we examine. If the match is not perfect between the profiles, then the profiles are inconsistent with each other.

mixture: alleles existing from more than one person in a profile, such as two people bleeding on a piece of evidence near the same vicinity. Sometimes, analysts are able to determine if one person’s DNA is present in a higher concentration than the other person’s DNA. In that case, the report may state that there is a major contributor (the person who contributed the most DNA) and a minor contributor (the person whose DNA is present in a lesser concentration in a mixture).

exclusion (elimination): when a known profile from a standard is compared to an unknown profile and the profiles are not the same. The person could not have left the DNA, so the person is excluded as a contributor.

sperm detected: semen is present, but may not necessarily contain intact sperm cells.

If a male has had a vasectomy or is incapable of producing sperm, then sperm would be absent from the semen.

For successful DNA typing, intact sperm cells must be present in the semen. Semen stains undergo a special technique to separate the sperm from any other body cell (male and/or female) that may be present. The goal is to isolate the sperm cells. Sometimes, complete sperm isolation cannot be achieved, resulting in mixtures in either separation.

sperm fraction: of the two semen separations, the one containing sperm is called the sperm (or male) fraction; may contain no male DNA if no intact sperm cells are present in the semen.

nonsperm fraction: of the two semen separations, the one containing all of the cellular DNA (female and/or male) other than intact sperm is called the nonsperm (or female) fraction.

insufficient amount of DNA: indicates that a sample has no DNA; the DNA is too degraded; there may be DNA, but not enough to type; or the stain was not human.

EXAMINATIONS NOT PERFORMED...

Several reasons exist to explain why a report states that certain examinations were not conducted:

1. If an analyst makes a link between the victim and suspect, then the remaining blood evidence is usually not examined. For instance, in a rape case, if the suspect’s profile is on the victim’s vaginal swab, the lab probably will not examine the bedding or clothing. The strongest association has been made by showing that actual vaginal penetration and ejaculation occurred. Semen on bedding is a weaker link and merely indicates that the suspect was on the bed, and the victim may or may not have been present. Developing the suspect’s profile on every piece of evidence is overkill and a waste of time and resources that could be spent on another case.

2. Analysts do not examine items on which they normally expect to locate the suspect or victim profiles. For example, a suspect’s clothing is usually not examined for semen because we would expect to find his semen on his own clothes, especially underwear.

3. The specimen may not be acceptable for DNA typing, such as blood collected from a bleeding suspect’s hands to blood on a shirt. If the blood is dried, it may not be suitable for examination.

4. The specimen may be a duplicate. Sometimes, agencies submit both a blood tube and a buccal swab for a standard. The analyst will choose which one to type, and the other may not be examined.

5. Another section obtains better results from the evidence. A suspect’s bloody fingerprint on a knife would be better evidence for fingerprint examination, since DNA testing destroys the print.

6. The analyst determines that DNA would not be beneficial to the case. A prime example is cigarette butts found in a public bathroom at a homicide. One could argue that these butts could be from anyone at any time, and any DNA results could be misleading as to the true suspect.

frequency: refers to the approximate number of times we would expect to find that particular profile in the population if we typed everyone in the world. This is NOT the probability that the suspect committed the crime.

FREQUENCIES VERSUS IDENTITY...

Frequencies tend to produce quite a bit of confusion not only for submitting agencies, but for attorneys and juries, as well. Why can’t the lab just come right out and identify the suspect as the one committing the crime? The DNA section applies statistics to profiles, rather than reporting the DNA belongs to the suspect, simply because analysts do not have the luxury of comparing a person’s entire DNA genome, just the 13 loci. Therefore, we can only apply a frequency relating how rare the profile is in the population; in other words, how many times we would expect to see that profile in the population if we typed everyone, so we have to settle for statistics instead of an identity statement.

The lab does not report frequencies for profiles we would find in normal places, such as the victim’s profile on her own vaginal swab, or for mixtures where major and minor components cannot be differentiated.

SAMPLE REPORT...

Semen was detected on the vaginal swab, indicated as from Victim A. Human blood was detected on the pants, indicated as from Victim B. The lab from Victim A screened positive for the presence of blood; however, human blood confirmation was not conducted due the limited amount of sample.

DNA profiles were developed from the vaginal swab and the pants and compared to the DNA profiles developed from the standards from Victim A, Suspect 1, and Suspect 2.

The nonsperm fraction from the vaginal swab is characteristic of a mixture. The most common mixture is consistent with Victim A. The minor component of the mixture is a partial profile from which Suspect 2 cannot be eliminated.

The sperm fraction from the vaginal swab is consistent with Suspect 2. This profile has an approximate frequency of 1 in 68/78/65/32/82/101/112/111/114/116/117/114/32/89/111/110/104/37/97/116/32/82/101/110/115/111/114/116/117.

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