Forensic DNA Testing: Issues and Problems

Ronald T. Acton, Ph.D., HCLD(ABB), CC(ABB), D(ABMLI), MB(ASCP)^{CM}

Professor Emeritus, School of Medicine

UAB

Chief Consultant

South Arising Consultants

Identity Testing

- Identity testing is an attempt to determine if two biological specimens or two individuals are identical or related.
- Forensic DNA testing is one form of identity testing.
- Paternity testing is another form of identity testing.
- DNA testing is also used to ascertain whether a donor bone graft is successful.
- DNA testing is used to ascertain twins.

Contrasting Forensic DNA Testing With Other Forms of Testing

- In paternity and medical DNA testing the sample is almost always pristine and the amount of the material is adequate.
- Mixtures of DNA profiles rarely a problem.
- More rigid requirements govern paternity and medical testing.
- Statistical treatment of the test results are well established and accepted in the scientific community.
- Forensic evidence samples are often in limited amount, may be degraded, may be contaminated and may contain a mixture of DNA profiles.
- In forensic testing the methodology is often pushed to the limits.

Initial Information the DNA Expert Needs

The laboratory report to determine:

The type of test(s) performed.

Inclusion or exclusion.

Is there a mixture of profiles.

The statistical probability.

The Type of Test(s) Performed

- Variable number of tandem repeats (VNTR).
- Dot blots.
- Short tandem repeats (STR).
- Y short tandem repeats (Y-STR).
- Mitochondrial sequencing.

Additional Information Needed

- Chain of custody documents.
- Curriculum vitae.
- Proficiency testing program results.
- License and/or accreditation.
- Error rate.
- Procedures manual.
- Laboratory bench notes.
- Alleles identified for subjects and evidence items.
- STR or Y-STR electropherograms.
- Mitochondrial sequence data.
- Data base of allele frequencies used in statistical calculations.

Issues/Problems Affecting Data Analyses and Conclusions Drawn

- Laboratory accreditation
- Training of director and analyst
- What constitutes an expert
- Laboratory independence
- Interpretation of results
- Bias
- A mixture of DNA profiles on an evidence item.
- Alleles possessed by the suspect or victim that are not present on an evidence item
- Touch DNA
- Transfer DNA
- Forensic Genealogy
- Statistics

Examples of Test Results

STR Profiles From Single Source Suspect Excluded

Locus	Alleles Detected	
	Evidence	Suspect
D3S1358	14, 17	14, 17
D8S1179	10, 13	9, 13
D21S11	27, 28	29, 32
D18S51	13, 16	13, 16

STR Testing that Includes a Suspect

Locus	Alleles Detected	
	Evidence	Suspect
D3S1358	14, 17	14, 17
D8S1179	10, 13	10, 13
D21S11	27, 28	27, 28
D18S51	13, 16	13, 16

Frequency of DNA profile in white population =

2(0.07 x 0.24) x 2(0.03 x 0.19) x 2(0.07 x 0.26) x 2(0.04 x 0.15) = 0.0000001 or 1 in 10,000,000

Mixture of DNA Profiles in Evidence

Locus	Alleles Detected			Other DNA Profiles
	Victim	Suspect	Nails	
D3S1358	15	14,17	14, 15, 17	14, 15 + 15, 17; 14, 14 + 15, 17
D8S1179	10, 13	14	10, 13, 14	10, 14 + 10, 13; 10, 14 + 13, 14
D21S11	27, 28	26, 29	27, 28, 26, 29	27, 26 + 28, 29; 28, 26 + 27, 29
D18S51	13, 16	15, 17	13, 16, 15, 17	13, 15 + 16, 17; 13, 17 + 16, 15

Profiles consistent with victim and suspect.

Mixture of DNA Profiles in Evidence

Locus	Alleles Detected			Other DNA Profiles
	Victim	Suspect	Vaginal Swab	
D3S1358	15	14,17	14, 15, 16, 17	14, 15 + 15, 17; 14, 14 + 15, 17; 14, 15 + 16, 17
D8S1179	10, 13	14	10, 12, 13, 14	10, 14 + 10, 13; 10, 14 + 13, 14; 10, 12 + 13, 14
D21S11	27, 28	26, 29	27, 28, 26, 29	27, 26 + 28, 29; 28, 26 + 27, 29
D18S51	13, 16	15, 17	13, 16, 15, 17	13, 15 + 16, 17; 13, 17 + 16, 15

Alleles in profiles that are not consistent with victim and suspect.

Missing Alleles

- It is a biological fact that every cell in your body, except red blood cells, contain all your genes.
- If one conducts a DNA test to assess the presence of alleles at a certain number of loci, results should be obtained at all loci.
- Therefore, cells left on an evidence item contains all the genes possessed by the person leaving their cells.
- Failure to detect alleles at all loci indicates that something is wrong with the test.

Alleles Missing in Evidence

Alleles	Alleles Detected			Alleles Missing
	Victim	Suspect	Sock	
D3S1358	15, 16	16, 17	15, 16, 17	
D8S1179	13	12, 16	12, 13, 16	
D18S51	17, 18	12, 17	12	17
D7S820	8, 10	8, 11	8	11
D16S539	10, 12	9, 11	-	9, 11

Allele Drop-Out

Allele drop-out occurs when a sample is analyzed, and one or more alleles are not present. This can be due to a variety of factors none of which can be accurately ascertained:

- The initial input quantity of DNA is too low, resulting in the failure to amplify one or more alleles in the sample.
- A mutation in the primer binding site is present, which causes a failure in the amplification of the allele.
- An allele sizes outside of the normal calling range for a particular locus and goes undetected.

Statistics

“There are three kinds of lies: lies, damn lies, and statistics.”

Benjamin Disraeli , the prime minister of the British Empire from 1874-1880, was reported by Mark Twain to have uttered this statement on statistical analysis. Incorrect.

Statistical Issues to Consider

- Database of allele frequencies
- Method of calculating probabilities

FBI'S Database of STR Allele Frequencies

The FBI's database of STR allele frequencies used by most forensic laboratories in statistical calculations consist of 1100 DNA profiles from African Americans, Caucasians, Southwest Hispanics, Bahamians, Jamaicans, Trinidadians, Filipinos and Chamorros.

Database Issues

- STR allele frequencies vary between racial and ethnic groups and racial and ethnic groups living in different geographic regions.
- Are STR allele frequencies randomly distributed in the population?
- Is there the possibility of population substructure in the area where the crime was committed?
- Are STR alleles frequencies representative of the population where the crime was committed?

Statistical Statement in Report

It can be concluded that to a reasonable degree of scientific certainty subject is the source of the DNA profile from the evidence.

Reasonable degree of scientific certainty means that one is at least 99% certain that this DNA profile would not be seen in a sample of 300 million randomly selected unrelated individuals.

Statistical Statement in Report

Subject cannot be excluded as a potential contributor to this mixed profile. Approximately 99.999998% of the African-American population would be excluded as potential contributors to this mixed profile or approximately 1 in 59 million African-Americans would be included as potential contributors to this mixed profile.

Statistical Statement in Report

Subject cannot be excluded as a contributor to this mixed partial Y-STR profile. In addition, all patrilineal related male relatives and an unknown number of unrelated males cannot be excluded as potential contributors to this mixed partial Y-STR profile.

Approximately 99.97% of the male population can be excluded as a contributor of this mixed Y-STR profile or approximately 1 in 2,900 males can be included as being the source of this Y-STR profile .

Statistical Statement in Report

The genetic traits detected from the evidence are a mixture of at least two individuals, at least one of which is male. The major component of the genetic traits detected from this item and the DNA profile of the suspect match.

This combination of genetic traits occurs in approximately 1 of 1.15 octillion (1 of 1.15E+29) random, unrelated Caucasian individuals, and 1 of 1.05 octillion (1 of 1.05E+27) random, unrelated African-American individuals

How Reasonable is This Number?

As of 2018 the world population is 7.7 billion people.

"Modern" *Homo sapiens* (that is, people who were roughly like we are now) first walked the Earth about 50,000 years ago. Since then, more than 108 billion members of our species have ever been born, according to estimates by Population Reference Bureau (PRB).

1 billion = 109**
1 octillion = 1027**

115 x 1027 octillion/108 x 10** 9 billion = 1.06 x 10 ** 18 more people than have ever been born on earth.**

Probabilistic Genotyping

- TrueAllele® (Cybergenetics)
- STRmix™ (STRMIX LIMITED)

Summary

DNA identity testing is a powerful tool for forensic analysis

As with any complex test that uses the principles and methodology from several scientific and medical disciplines, the test can be improperly performed, the test results improperly interpreted, and errors can occur.