

Adventures in Forensic DNA: Cold Hits, Familial Searches, and Mixtures

Sandy Zabell

Departments of Mathematics and Statistics
Northwestern University

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The approach of this talk

Some simple conceptual issues

... sometimes illustrated by examples from actual cases

... and an occasional reality check

Footnote for the bar: slide language is intentionally kept simple

Forensic DNA 101

PCR: *Polymerase chain reaction*

- ▶ “molecular xeroxing”
- ▶ very sensitive
(potential for contamination and trace DNA)

STR: *Short tandem repeat*

Example: ATCC ATCC ATCC ... ATCC (n times)

DNA Profile: typically 9–15 pairs of repeat numbers

Forensic DNA 101 concluded

Most common scenario: two sources of DNA:

“Known” (victim or suspect)

“Unknown” (evidence)

9–13 locus concordance usually very strong evidence of identity.

(“one in a gazillion”)

“Cold Hits”

“Probable cause” vs. cold hit scenario

Common intuition: Evidence more compelling in first case.

NRC committees, distinguished statisticians have differed on this!

NRC 2: if p is match probability, but searched database of size n ,

use $1 - (1 - p)^n \approx np$ instead of p .

Resolution of the (apparent) paradox

Use Bayes's theorem:

$$\frac{P(H_1 | E)}{P(H_0 | E)} = \frac{P(E | H_1)}{P(E | H_0)} \cdot \frac{P(H_1)}{P(H_0)};$$

- ▶ : E : DNA evidence
- ▶ : H_0 : target not source
- ▶ : H_1 : target is source

The likelihood ratio is (largely) unchanged, but prior odds differ.

For the non-Bayesians ...

Suppose

- ▶ $p = 1/1,000,000$ (match probability)
- ▶ $n = 100,000$ (size of database)
- ▶ $N = 10,000,000$ (size of potential suspect pool)

$np = 1/10$... but expect about 10 profiles in pool.

Explaining these issues to trier of fact can be complicated.

Familial Searches

Search a database for “near misses”

Rationale: relatives are much more likely to have matching profiles

Example: IN v. Flowers

1. Steven Myers *et al.*, *Forensic Science International: Genetics*, 5 (2011), pp. 493–500.
2. David H. Kaye, “The genealogy detectives: a constitutional analysis of “familial searching” , to appear.

Mixtures

Two or more sources of DNA are present

For example, might see n alleles A_1, \dots, A_n

There are $n + \binom{n}{2} = \frac{n(n+1)}{2}$ consistent genotypes

The CPI (*Combined Probability of Inclusion*): uses

$$(p_{A_1} + \dots + p_{A_n})^2$$

The likelihood ratio

Recommended by NRC2, but less commonly used.

If all alleles are present and accounted for, a simple formula exists,
... thanks to

Weir, B.S., et al. (1997). Interpreting DNA mixtures. *Journal of Forensic Sciences* 42, pp. 213–222.

If ...

In fact many complications exist:

- ▶ The number of contributors may be unknown
- ▶ The amounts of DNA may differ
...and most feared of all ...
- ▶ **Allelic dropout**

Technical issues

Alleles are scored using *peaks* on an *electropherogram*

The fine print:

- ▶ “stochastic thresholds”
- ▶ “analytical thresholds”
- ▶ “peak height ratios”

Lab protocols leave the analyst great leeway about scoring alleles.

Enormous activity in this area recently

Resource: NIST, “STRbase”

In particular, see “Information on DNA Mixture Interpretation”
(<http://www.cstl.nist.gov/strbase/mixture.htm>)

John Butler:

[M]any labs are doing or attempting more complex mixtures often without appropriate underlying validation support or consideration of complicating factors.

The single most important consideration: one should:

Make decisions on the evidentiary sample and document them prior to looking at the known(s) for comparison purposes.

[Again Butler, but my emphasis]

Many forensic scientists resist or do not understand this basic scientific principle.

This problem is not restricted to forensic DNA.

Nevertheless . . .

The use of DNA typing (justly) remains the gold standard of current forensic identification.

Questions?

Thank you!