Summer Institute in Statistical Genetics
University of Washington, Seattle
Forensic Genetics Module

**An introduction to the evaluation
of evidential weight**

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The notes for my lectures today and tomorrow are in the form of a 51-page document extracted from the book.

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Weight-of-Evidence for Forensic DNA Profiles

Second Edition

STATISTICS IN PRACTICE

WILEY

First we'll do some warm-up exercises.

1. The famous Collins case.
2. Rare disease testing.
3. Coloured taxis.

You have seen this sort of example before but the "base rate fallacy" seems such a natural error for human brains that frequent refreshers are useful.

- For 2 and 3 above, the problem is set up so that there is a right answer.
- That's not so for 1, we won't propose a solution here and indeed there is no full solution, but I hope by the end of this course you will have ideas about how to approach evidence evaluation in such cases.

One mantra that I would suggest you recite for an hour every evening is

Always focus on the right question.

Much confusion is caused by straying from this path.

Yesterday's news headline

Melania Trump: Astrophysicist calculates there was one in 87 billion chance speech was not plagiarised

- What's wrong with the headline?
- What's needed to make it right?

In the UK case of Sally Clark, the medical expert Roy Meadows testified something like

> *The probability for two babies in a family to die of SIDS is 1 in 73 million.*

The number is wrong, but let's ignore that. What is the right question?

For DNA profile evidence the question is usually

> *Did the DNA in the crime sample come from the alleged contributor(s)?*

We'll approach answering this question slowly, by way of a remote island where crime is rare, but did once happen.

- The "island problem" represents a simplification of a forensic identification problem.
- Don't be tricked into thinking it isn't important in practical problems – many of the key ideas are present.
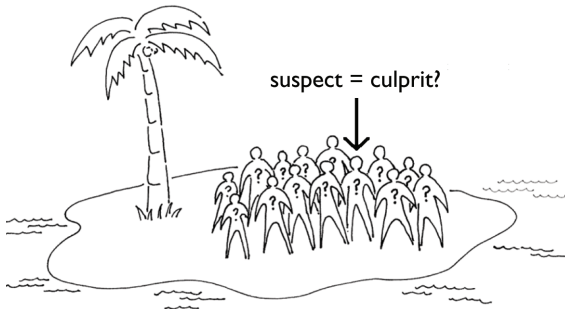
suspect = culprit?

- All 101 islanders are initially equally under suspicion;
- The culprit has Υ;
- The suspect has Υ;
- The Υ-states of the other islanders are unknown;
- We expect on average about 1 person in 100 to have Υ.

What is the probability that the suspect is guilty?

Lessons from the island problem

1. The fact that Υ is rare (i.e. p is small) does not, taken alone, imply that Q is likely to be guilty.

2. Uncertainty about p does not "cancel out". Ignoring uncertainty is unfavourable to defendants.

3. The overall weight of evidence against Q involves adding together the probability of a "chance match" and the probability of a match due to a typing error.

4. In the case of a search of possible culprits to find a "match" with crime scene evidence, the longer the search (i.e. the more individuals found not to match) the stronger the case against the one who is found to match.

Section 2.1.3: Application of the formula

There are a lot of ideas in this section so a summary may be useful:

- The order in which different items of evidence are analysed ultimately doesn't matter, but we need to be clear about order to avoid misunderstandings:
  - If the DNA evidence is considered first there may be a large set of alternative possible contributors, whereas the other evidence may, if accepted by the finder of fact, narrow it down to a small number of individuals.

- The weight-of-evidence formula doesn't solve all the problems, but it can guide thinking in the right direction.

- It can help delineate the roles of juror and expert witness.
  - the expert can advise on values for the LR for one or more hypothesis pairs (e.g. Q vs unrelated, Q vs sibling).
  - it's generally not the job of the expert to assess the other evidence (base rate or prior information) but illustrative calculations can be helpful (see next slides)

Illustrative probability calculation.

Reported LR: 3 million (unrelated)

> *... consider the hypothetical scenario in which, on the basis of the non-DNA evidence, a juror considered that there were 1 million men who could be the questioned DNA source: X and 999,999 men unrelated to him. If each is initially considered equally likely to be the source, the effect of the DNA evidence would be to change the probability that X is indeed the correct source from 1 in 1 million (or 0.0001%) up to 75%. If initially there were 10,000 men each equally likely to be the source of the DNA, the effect of the DNA evidence would be to change the probability that X is the true source from 1 in 10,000 (or 0.01%) up to 99.7%. Finally, if the other evidence were such that a juror considered that only 100 equally likely men could be the source of the DNA, the effect of the DNA evidence would be to change the probability that X is the true source from 1 in 100 (or 1%) up to over 99.99%.*

Illustrative probability calculation.

Reported LR: 61 (brother)

> *Again hypothetically, if a juror judged that the DNA must have come from either X or a specified brother of X, both initially equally likely, then the effect of the DNA evidence would be to change the probability that X is the true source from 1 in 2 (or 50%) up to over 98%.*

Section 2.2.4: Laboratory and handling errors

The strength of DNA evidence requires the probabilities of

1. a chance match with an alternative possible contributor, and
2. a false match due to an error.

The probability of any error occurring in the handling and analysis of a DNA profile is typically much higher than 1 above.

- Not relevant: only the probability of *an error causing a false match*.
- Example of newspaper report of winning lottery ticket.

2 above is essentially limited to the suspect's DNA being present in the crime sample for reasons other than committing the crime, such as:

- Cross contamination in the lab of evidence samples from different crime scenes (UK 2012 case of Adam Scott)
- Deliberate planting of DNA to frame a suspect.

Even if considered very unlikely (no evidence to suggest either), the possibility of cross contamination or of planting of evidence may be considered by a reasonable juror to be more likely than a chance match.

- If so, a DNA match probability may be essentially irrelevant.

However that's an assessment to be made by jurors.

- An expert can give jurors some information about laboratory errors but very little about evidence tampering.
- It's still useful to report match probabilities even if a juror later assess this to be unimportant.

Why not declare uniqueness of the profile?

- How to decide the threshold for uniqueness?
    - Low-template and mixed samples often generate modest LRs.
- What to do about other evidence?
    - All evidence in favour of Q is evidence against uniqueness.

What's wrong with RMNE (inclusion probability)?

It doesn't answer the right question!

- How to incorporate other evidence?
- All evidence counts as evidence against the defendant - not always realistic.
- Problematic to compute RMNE for low-template profiles and multiple contributors of interest.
- Weight of evidence doesn't depend on the profile of the alleged contributor.
    - e.g. two co-defendants both alleged to be contributors.

Calculating LRs allowing for coancestry

- The match probability for X given the profile of Q depends on relatedness: both close relatedness and more distant "coancestry".

- There exists some very nice population genetics theory that allows us to compute match probabilities allowing for relatedness or coancestry, the latter measured by the coefficient F_{ST}.

- F_{ST} has several interpretations, one convenient interpretation is:

    The probability that two alleles sampled in a subpopulation are identical through inheritance from an ancestor within the subpopulation.

- Genotype matches at distinct loci are generally not independent,
    - but relatedness is the cause of non-independence and after adjusting for relatives/coancestry it is reasonable to proceed assuming independence (multiply across loci).

Computing match probabilities

The match probabilities depend on:

- population allele fractions (the p).
    - These are unknown but can be estimated from a population database.
    - Sampling variation should be taken into account: several ways to do this; it usually doesn't make much difference.
- The coancestry coefficient F_{ST}.
    - Also unknown, and cannot be directly estimated for a particular crime scenario.
    - Appropriate value can vary over different X for the given Q: simplest to proceed using an upper bound.
    - Fortunately high mutation rate at STR loci seems to keep F_{ST} relatively low: we have found that 3% is generally a safe value to use even if the wrong database is specified for X.

Match probabilities for mixture profiles

How many and which contributors to include in the competing hypotheses can be difficult.

- If the amount of DNA from each contributor is estimated, then overstating the number of contributors presents only a computational problem
    - if not really a contributor, the estimated amount of DNA will be low.
- Low-level contributors that can be distinguished from the contributor(s) of interest can be treated like "dropin" (so the exact number of contributors need not be known).
- For multiple contributors of interest, we need to take care about specifying hypotheses.
    - It is usually not appropriate to compare "both are contributors" with "neither is a contributor".
    - Instead we have to proceed one contributor at a time, with and without the other contributor being assumed present.
    - Thus 4 LRs for two queried contributors.