INTERPRETATION OF DNA EVIDENCE: A PARADIGM FOR FORENSIC VOICE COMPARISON?

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People vs Collins, California 1968

Bystanders to a robbery testified that the perpetrators had been a black male, with a beard and moustache, and a Caucasian female with blonde hair in a ponytail. They had escaped in a yellow car.

An "instructor in mathematics" explained the multiplication rule for probability, and the prosecutor invited the jury to consider the probability that the accused pair, who fitted the above description, were not the robbers. The prosecutor suggested that the jury would be safe in estimating:

Black man with beard	1 in 10
Man with moustache	1 in 4
White woman with ponytail	1 in 10
White woman with blonde hair	1 in 3
Yellow car	1 in 10
Interracial couple in car	1 in 1000

The jury returned a verdict of guilty.

Forensic Identification Using DNA profiles

1. Sample left at scene of crime provides a DNA profile.
2. Defendant's profile matches.
3. Based on a convenience sample of DNA profiles, together with population genetics theory, it is estimated that about 1 person in 1 million will have a matching profile.

How convinced should a juror be that the defendant is the source of the crime sample?

Several answers given in the literature for the probability that the defendant is guilty in an idealised "Island Problem":

1. $P(G) = 1/(1+E(X))$ where X is the number of innocent individuals with the profile on the island.
2. $P(G) = E[1/Z|Z > 0]$ where Z is the number of individuals with the profile on the island before crime sample observed.
3. $P(G) = 1/(1+NP)$, standard application of Bayes Theorem.

The weight-of-evidence formula

Eventually a consensus emerged in favour of Bayes Theorem, which in a general form can be written:

$$\mathrm{P(C{=}S|evidence)} = \frac{\mathrm{P(evidence\ \&\ C{=}S)}}{\sum_X \mathrm{P(evidence\ \&\ C{=}X)}}$$

where

- C is the source of the crime sample;
- S is the defendant;
- the summation $\sum$ is over all individuals X who might be C, including S. (Could be over everyone on earth: the probability vanishes for most.)

Assume that DNA evidence is assessed last. Then can rewrite formula as

$$\text{P(C=S|all evidence)} = \frac{1}{1 + \sum_X w_X R_X}$$

where

$$w_X = \frac{\text{P(C=X|other evidence)}}{\text{P(C=S|other evidence)}} = \text{prior odds ratio}$$

$$R_X = \frac{\text{P(DNA evidence|C=X)}}{\text{P(DNA evidence|C=S)}} = \text{likelihood ratio (LR)}$$

Assumption: DNA evidence is independent of the other evidence, given C.

Some lessons learned from the weight-of-evidence formula ...

A. R_X alone does not determine P(C=S).

Suppose that:

1. each of N possible culprits is, if not for the DNA evidence, in much the same position as the defendant, so that $w_X = 1$ for all these individuals, and $w_X = 0$ for everyone else;
2. R_X is equal to a constant r.

Then

$$\mathrm{P}(\mathrm{C{=}S}|\text{evidence}) = \frac{1}{1 + Nr}$$

So $r = 1/1$ million may not be convincing if N is large, plausible e.g. in a big city.

B. Relatives may be important.

Assume $N + B$ possible culprits (other than S):

- B brothers of S, and $R = 1/100$ for each of them;
- N unrelated men, $R = 10^{-6}$ for each.

If $w = 1$ for each possible culprit, then

$$\mathrm{P(C{=}S|evidence)} = \frac{1}{1 + B/100 + N/10^6}$$

B may dominate even if $N \gg B$ and there is no evidence against any of the brothers.

C. Ethnicity of possible culprits may be important.

- N_1 possible culprits have genetic background similar to that of S, and N_2 have a distinct genetic background.
- Positive correlations due to shared ancestry, and lack of frequency estimates on fine scales $\Rightarrow$ P(X has profile|S has it) is larger for the former group than for the latter.
- So the N_1 "similar genetic background" individuals can dominate, even if $N_1 < N_2$. Argument essentially the same as for brothers; here, difference in R not so great, but N_1 usually much larger than B.

D. Lab/handling error or tampering may be important.

Matching DNA profiles can arise because:

1. S=C and no error occurred;
2. S≠C, but S and C have matching "true" profile;
3. S≠C, different "true" profiles, but one or both were incorrectly recorded, resulting in a match.

- A reasonable juror may assign 1 $\gg$ P(3.) $\gg$ P(2.).
- P(DNA evidence|C=X) involves a sum of P(2.) and P(3.), so 2. may be irrelevant if P(3.) $\gg$ P(2.)
- It's not the probability of *any* error that matters, but an error that results in a match (c.f. Price, lotteries & printing errors).
- The probability of evidence tampering leading to a match is almost always greater than R_X, so arguably R_X is always irrelevant.

E. Effect of database search.

- After observing the crime scene profile, you search a database of the DNA profiles of known individuals and find exactly one match. The matching individual becomes the defendant S.
- The database search is like a hypothesis "trawl", and so the resulting evidence is weakened in proportion to the size of the search.
- For example, if the match probability is 1/1 million, and there are 1 million profiles in the database, then a match is expected even if C isn't in the database. So these data would provide no evidence against S, right?

Wrong!

Probability analysis shows that evidence is slightly stronger after a database search. Reasons:

1. non-matching individuals searched are (in effect) excluded as possible culprits (so "N" becomes smaller);
2. observation of non-matches strengthens the evidence that the profile is rare.
3. The **hypothesis trawl** analogy isn't valid because we know in advance that one of the hypotheses is true. Suppose the database includes every possible culprit. Is the evidence against the unique matching individual weak or strong?
4. The **probability of a match in the database** is irrelevant. Although analogous with a common mode of statistical reasoning, it is simply addressing the wrong problem and in this setting is very misleading. Example: 1 million + 1 possible culprits, of whom 1 million have a profile in the database.

Many difficulties remain

It's almost impossible in practice to keep separate:

1. **likelihood ratios**: the domain of the expert witness
2. **probabilities of hypotheses**: the domain of the finder of fact.

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  - The forensic scientist often has to make subjective judgements about which alternative hypotheses are so implausible that they need not be considered (e.g. the voice is that of a spirit capable of perfect mimicry or aliens from another planet).
  - The number of contributors to a DNA sample can never be bounded above based on the DNA profile alone.
  - The profile evidence is often consistent with a moderately-high rate of contamination, and the decision to assume contamination rate ≈ 0 is based on (subjective) prior knowledge of the profiling process and previous casework.

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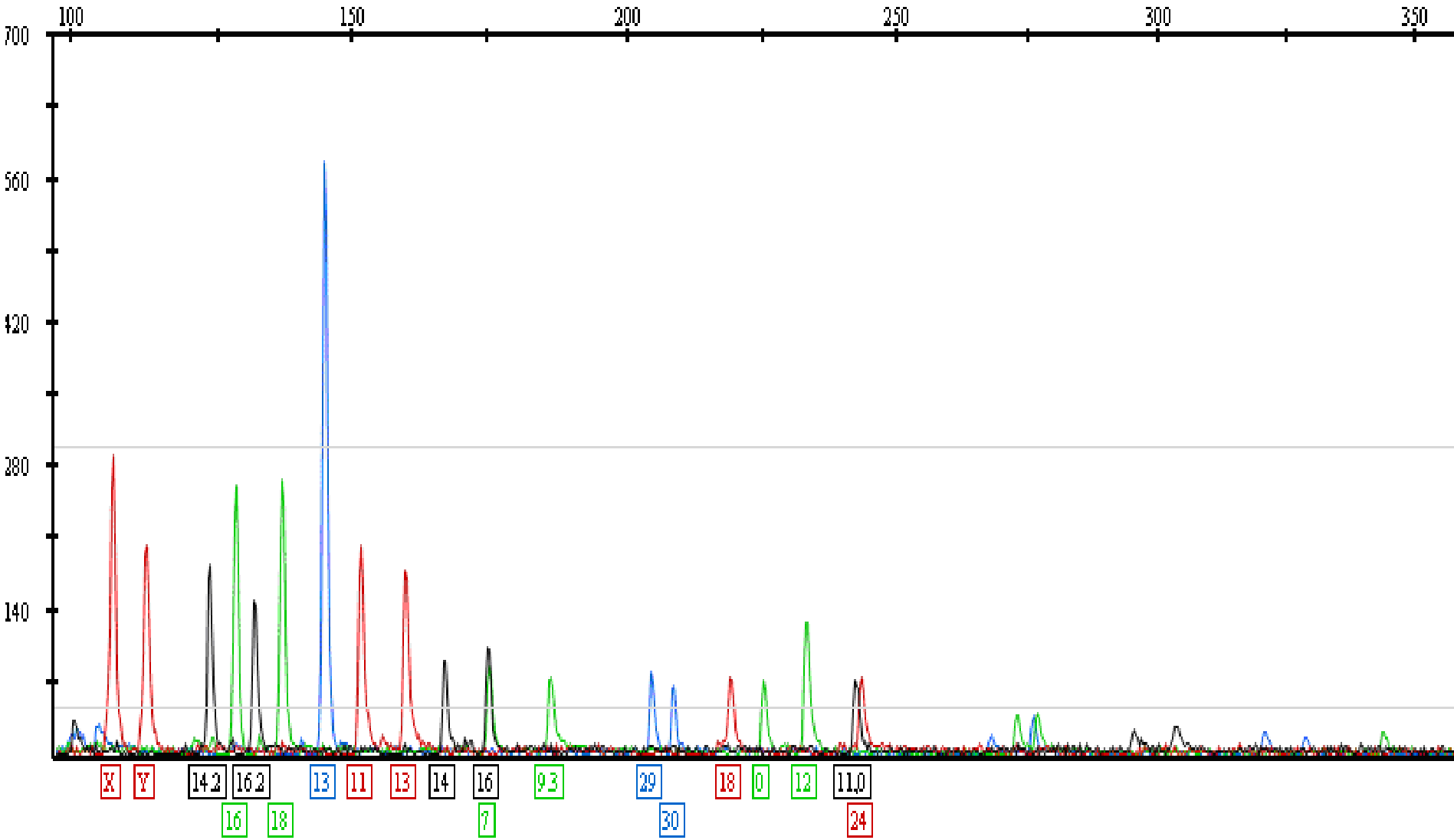
- Although sin is ultimately unavoidable, we should maintain the goal of righteous behaviour and be aware of deviations from it.

Brief introduction to short tandem repeat (STR) profiling

- Short tandem repeat (or "microsatellite") : DNA motif, usually 4 bp, repeated say 5 to 20 times
- High mutation rate: $\sim$ 1 per 500 generations
  - repeat number is highly variable in the population, typically 4–8 common alleles
- Typing is not sequence based
  - measure time to traverse a distance under an electric charge
  - time $\propto$ molecular weight $\Rightarrow$ repeat number can be inferred
  - can analyse multiple loci in one run using both length and colour separation.
- Resulting single-locus genotype: a pair of integers such as 7,9
  - 7 copies of motif on one chromosome, 9 on the other
  - partial repeats sometimes occur, e.g. 9.3
  - whole profile typically 10 – 20 loci.

Electropherogram: mutiplexing via colour separation



Where do the probabilities underlying LRs come from?

- In my subjective opinion frequency-based notions of probabilities have little direct applicability to the central issue in a criminal trial, which focusses on a specific event.
- For DNA evidence, probabilities emerge from population genetics theory;
  - but ultimately the choice of theory is subjective, and all theories include subjective elements.
- The probability that two individuals X and S share a DNA allele depends on their relatedness. Three levels:
  1. Known relatedness: eg. uncle of X = grandfather of S.
  2. Relatedness due to unknown shared ancestors, measured by F_{ST} (or θ).
  3. Completely unrelated.

Where do the probabilities underlying LRs come from?

- Much ink has been wasted on concerns about independence across genetic loci
  - but "independence" of two events is not an absolute yes/no, it depends on the conditioning
  - conditional on relatedness, Mendel's laws imply independence.
- Independence of an individual's two alleles within a population (Hardy-Weinberg Equilibrium) is an empirical approximation, much argued over but actually of little importance:
  - At a single locus

$$R_X = \mathrm{P}(G(X){=}AB | G(S){=}AB),$$

where $G(X)$ denotes "genotype of X". So independence of alleles across S and G matters, but independence of the two alleles of X or of S is peripheral.

Plug-in estimates vs integration

- LRs for DNA profile evidence at a single locus depend on
  1. estimates of allele frequencies in a relevant population, obtained from a database
  2. values for the population genetics parameter F_{ST}; by definition, directly-relevant estimates are not available, but we do have estimates in many different populations.
- Whole-profile LR depends involves products/powers.
- Common to use plug-in estimates
  - this keeps "training" or "background" data logically separate from evidence data.

  Ideally we should integrate over a distribution for unknowns
  - but expectation of a high power can be $\gg$ power of the expectation
- I have worked with an awkward compromise of using plug-in values but at high end of plausible range.

The "Random Man" Fallacy

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- In an attempt to avoid the unavoidable subjectiveness, or through laziness of thought, many writers have (implicitly) assumed that the probabilities underlying the LR are generated by random sampling of suspects in a population.
  - Objective (in a sense), but clearly false.
- This approach leads to numerous problems:
  1. Much ink spilled arguing over "in which population?", but it is like arguing over the number of wings on a tooth fairy. The more narrowly the population is defined, the better for the defendant, but this process leads to a "population" that includes only the defendant.
  2. "Random man" generated all the confusion over the "database search" problem (above).
  3. Leaves no way to think about
     - role of relatives
     - genetic background
     - laboratory error/fraud
     - incorporation of DNA with other evidence.

US National Research Council report

"The evaluation of forensic DNA evidence" (Natl Acad. Press, 1996) is centred on a "random man" view of weight of evidence, based on a null hypothesis of:

I_{NRC} : The defendant was chosen randomly in a population of innocent possible culprits

The report is riddled with errors and confusion, but the prestige of the NRC is such that it still holds sway in the US, and seems in effect unchallengeable.

Misunderstandings in the NRC report

1. NRC: "If the calculated profile probability is small ... even a large relative error will not change the conclusion";

   WRONG (see A above).

2. NRC: "Because one or a few relatives in a large population will have only a very slight effect on [profile frequency], we believe that the importance of unknown relatives has been exaggerated";

   WRONG see B.

3. NRC: "Most of the time, we believe, the subgroup of the suspect is irrelevant";

   WRONG see C.

Misunderstandings in the NRC report

4. NRC: "We believe that a calculation that combines error rates with match probabilities is inappropriate";

   WRONG see D. But it is not for forensic scientist to combine the two; this should be done by jurors themselves, with forensic scientists giving evidence to guide them e.g. describing quality assurance mechanisms and results of blind trials.

5. NRC: "The initial identification of a suspect through a search of a DNA profile database is analogous to performing a coin toss experiment many times";

   WRONG see E. If you toss 20 coins many times, it becomes more and more likely that you will eventually get 20 heads, but this is irrelevant to weight of evidence against the individual who matches; this person has not been "tossed" repeatedly.

Population genetics in the NRC report

- The committee which advised the NRC included prominent population geneticists, and it discussed population genetic issues at length.
- However, it only considered population genetic effects on the profile frequency P(X has profile), rather than on the LR (or match probability) P(X has profile|S has it),
- The report does briefly refer to the match probability, but it assumed independence of X and S without comment, thus missing the point of how population genetics affects DNA profile evidence.
- The NRC report therefore has almost nothing useful to say on the role of population genetic effects on DNA evidence, despite devoting to it many pages of misguided discussion.

Low Copy Number (or Low Template) STR profiling

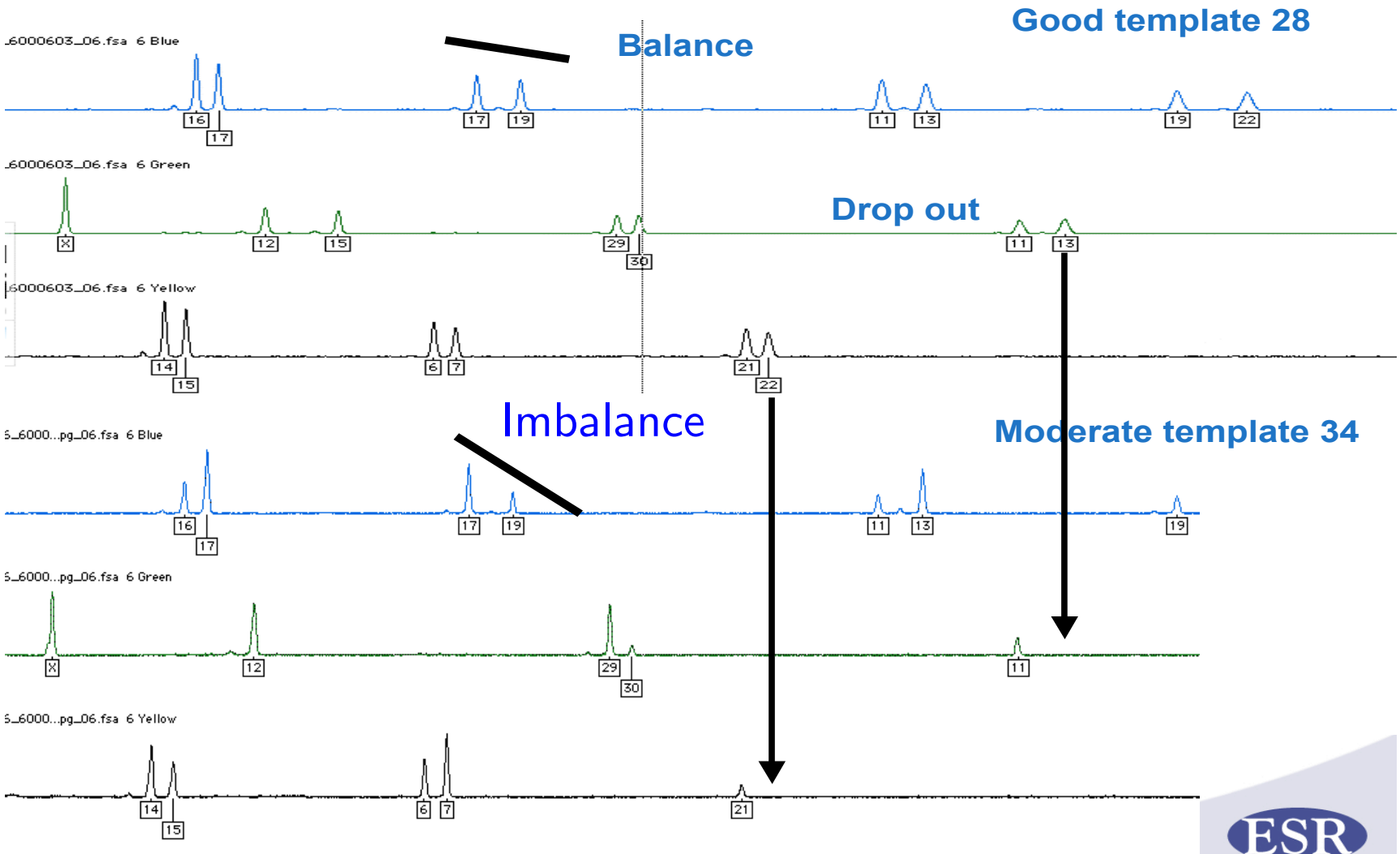
- STR profiling has traditionally used at least 1 ng of DNA
- Recently various enhancements of the technique, such as extra PCR cycles for additional replication, have allowed template levels as low as ∼20 pg to generate DNA profiles
  - this is approximately the DNA content of three cells
- Useful for miniscule traces of DNA e.g. extracted from touch marks or even residue of breath
- Problems:

  dropout e.g. PCR amplification fails entirely

  dropin due to isolated contaminant DNA, e.g. from laboratory plasticware

  peak imbalance can confuse interpretation of mixtures (multi-source profiles)

  exaggerated stutter can confuse interpretation of mixtures — may mask an allele of a low-level contributor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6000603_06.fsa 6 Green
Drop out
6000603_06.fsa 6 Yellow
Imbalance
Moderate template 34
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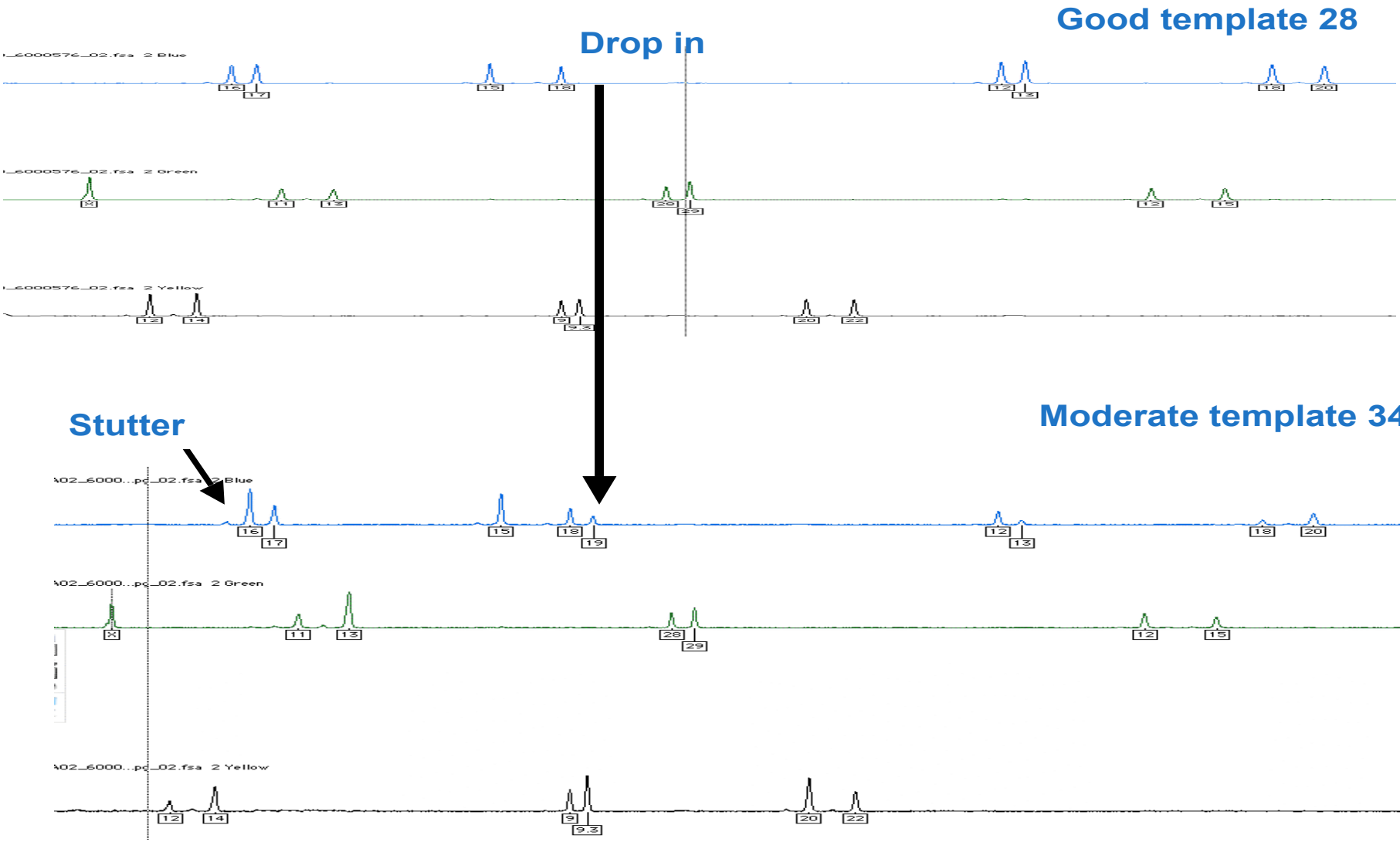
S_6000...pg_06.fsa 6 Green
S_6000...pg_06.fsa 6 Yellow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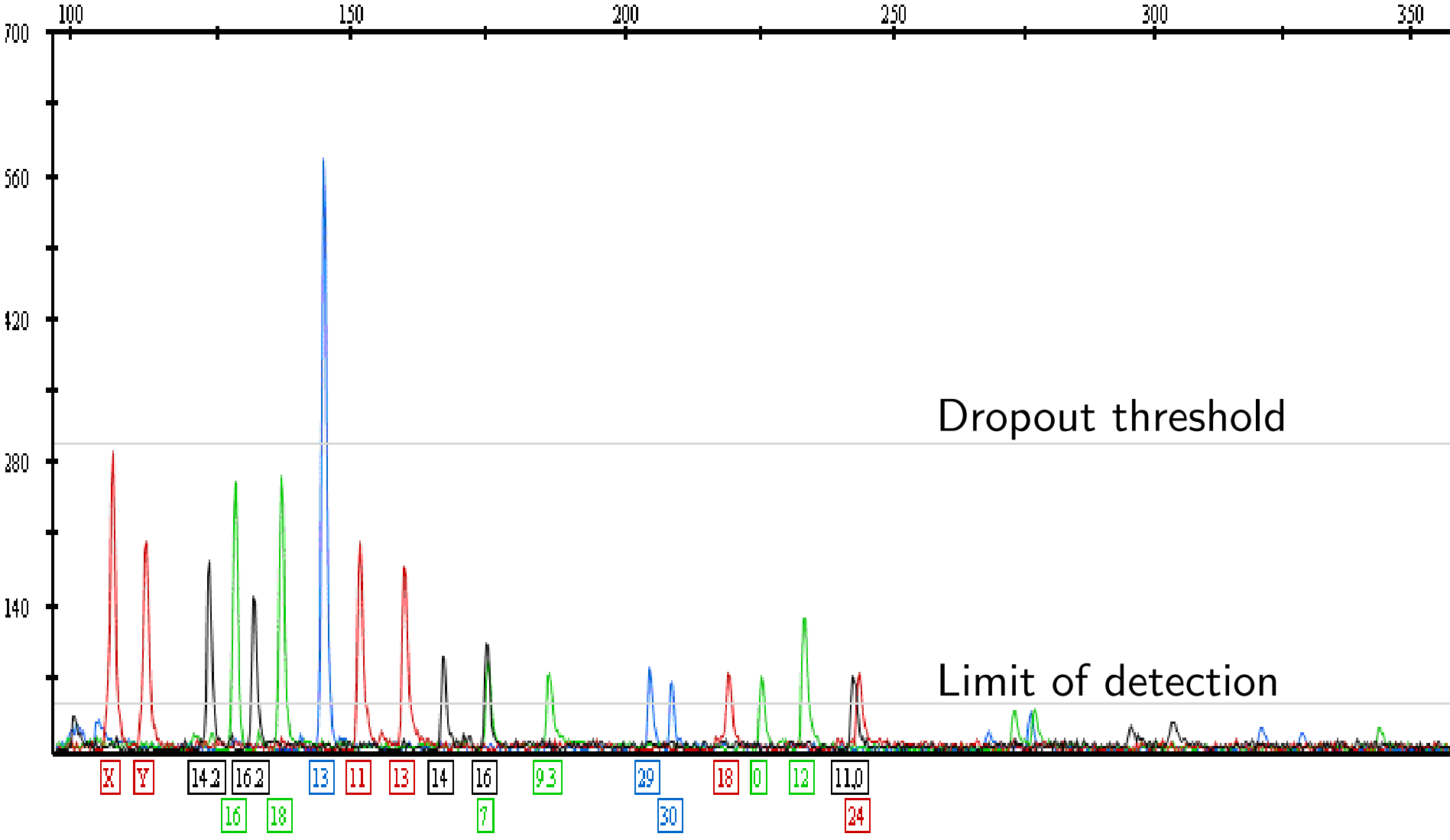

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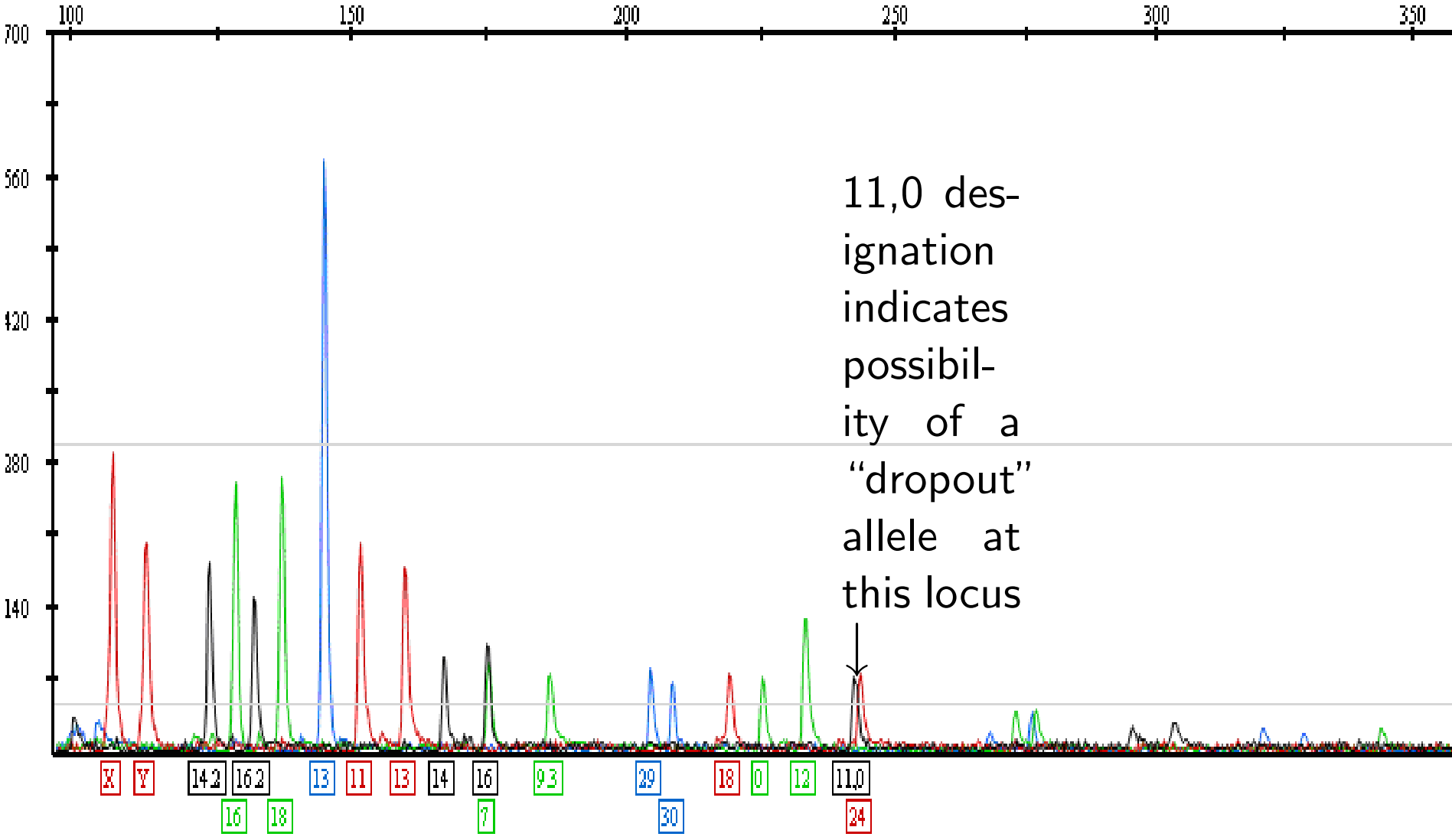

11,0 designation indicates possibility of a "dropout" allele at this locus

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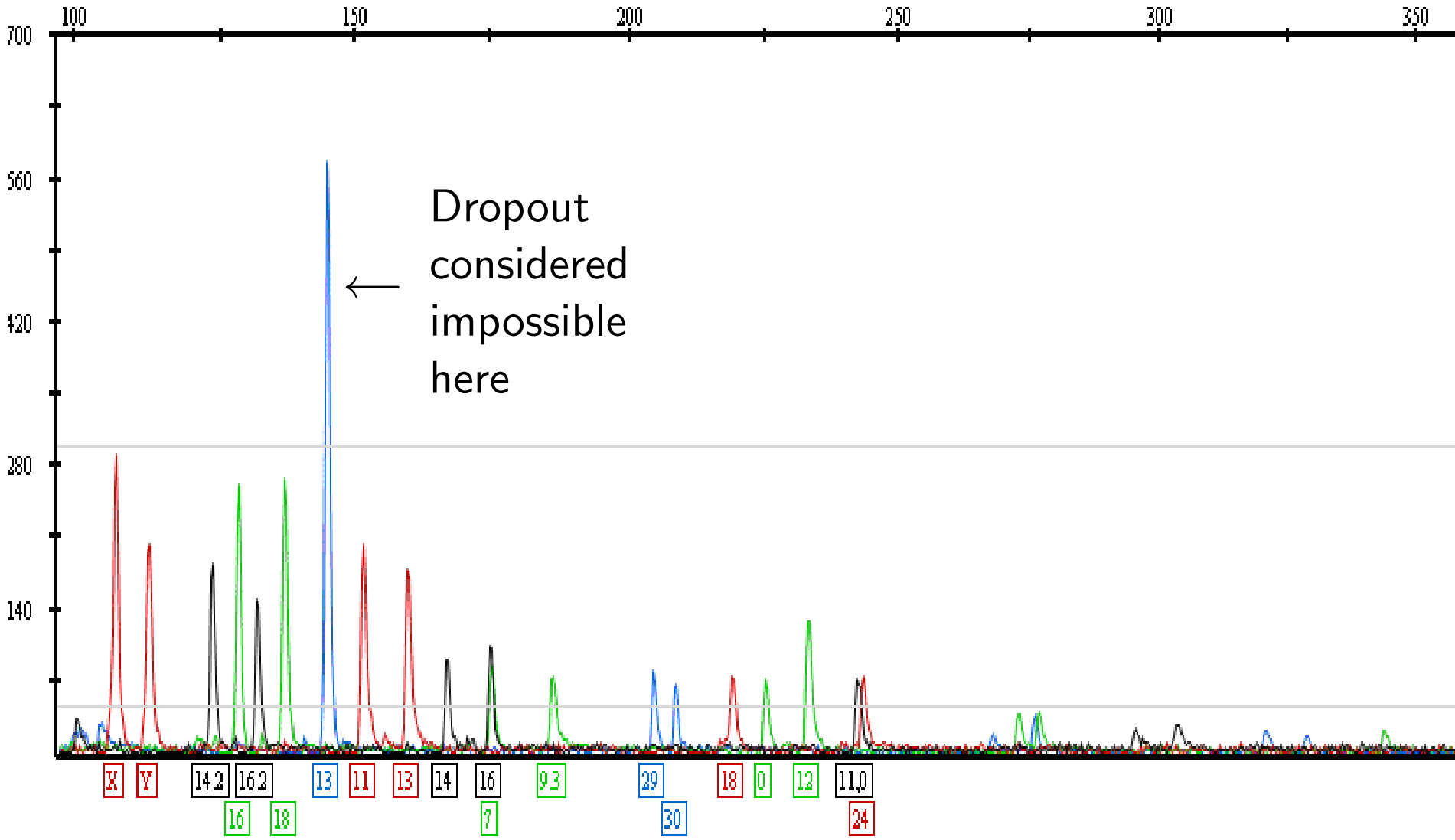
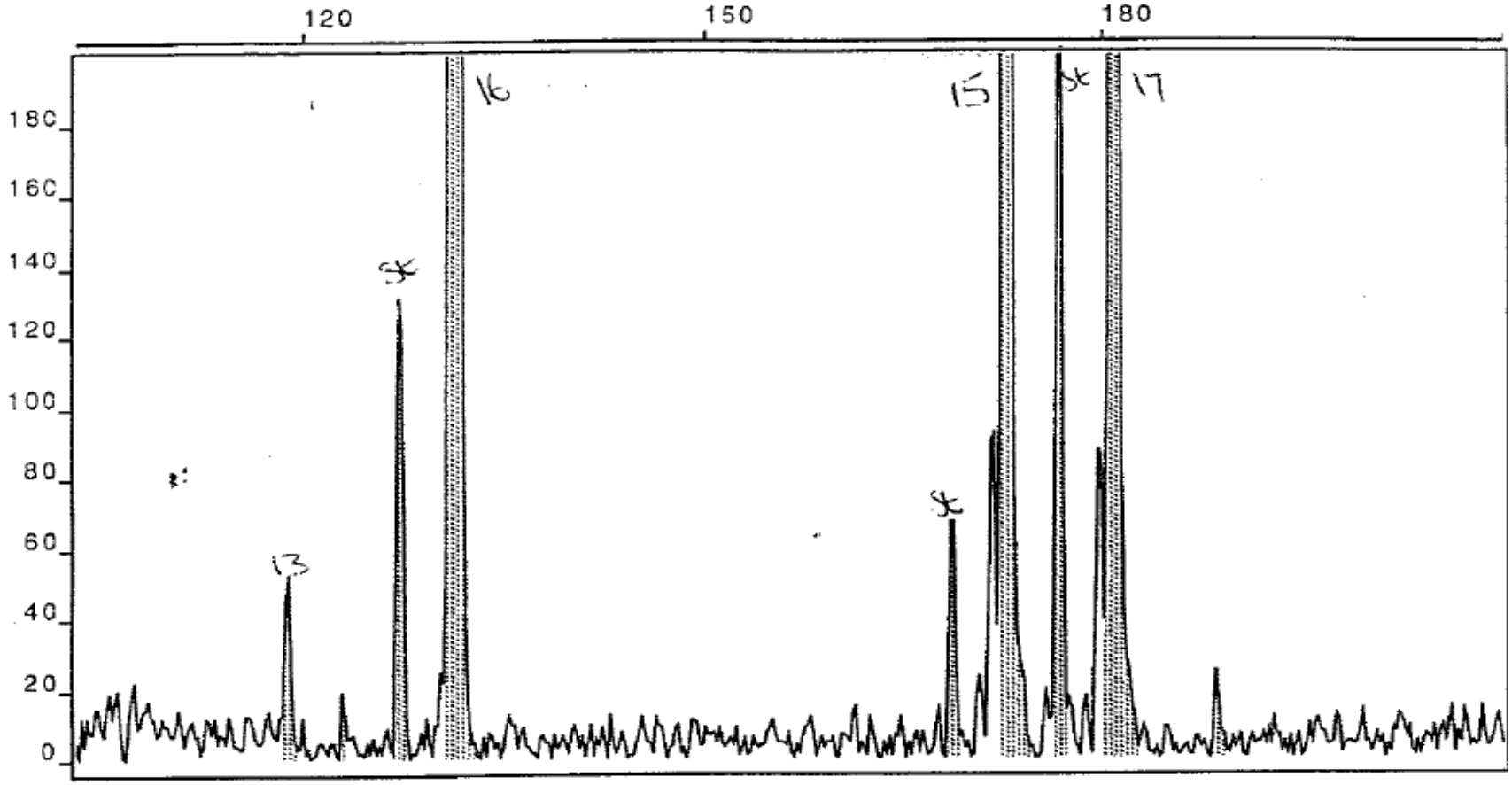


R v. Bates and Garside (London, 2003-2006)

- Bates alleged to have murdered Mrs Garside, in collusion with Mr Garside
- multiple DNA samples obtained from crime scene, many profiling runs on a 10-locus STR system
- Most incriminating profile was a mixture:
  - major component from victim: 17 distinct alleles
  - weak minor profile from a male: 8 alleles not masked by major profile
- Bates' profile includes 11 alleles NOT included in Mrs Garside's profile
  - all 11 expected in minor profile if Bates were source



Dye/Sample Peak	Minutes	Size	Peak Height	Peak Area	Data Point
26B,41	46.99	119.05	54	238	1762
26B,42	48.03	123.25	20	42	1801
26B,43	49.07	127.43	131	592	1840
26B,47	50.11	131.58	2167	10448	1879
26B,56	60.03	168.96	68	350	2251
26B,59	61.12	172.95	1433	7711	2292
26B,61	62.21	176.93	235	1182	2333
26B,64	63.31	180.91	1462	8049	2374
26B,67	65.47	188.77	26	105	2455

Locus	Mrs Garside	Bates	Minor component observed	Minor component expected if Bates
D3	16,16	13,16	13	13
VWA	15,17	16,16	16	16
D16	11,12	11,12	—	—
D2	20,20	**19**,22	22	**19**,22
D8	12,13	8,13	8	8
D21	30,32.2	30,31.2	31.2	31.2
D18	14,14	**12,15**	—	**12,15**
D19	12,14	12,15	15	15
THO1	9.3,9.3	7,7	7	7
FGA	23,25	21,21	21	21

Discordant alleles

3 defendant alleles missing under prosecution case

- referred to as “voids” by successive judges
  - misleading term, I prefer “discordant” (with prosecution hypothesis)
- one in stutter position to a major-profile homozygote allele
- two at a high molecular weight locus
  - more susceptible to dropout?
- some signs of an allele in some profiling runs, not to reporting standards
  - threshold of 50 rfu

Inclusion probabilities and the "2p rule"

The **inclusion probability** (or Random Man not Excluded, RMNE) is the probability that a "random man" would not be excluded by the DNA evidence. At a single locus:

$$\mathrm{P}(RMNE) = \begin{cases} \text{if no minor component allele observed} & 1 \\ \text{if allele A only observed} & 2p_A \\ \text{if A and B (single contributor)} & 2p_Ap_B \\ \text{if A and B (multiple contributors)} & (p_A+p_B)^2 \end{cases}$$

For > 2 alleles use square of sum of observed allele proportions.

- if no allele observed: no information; so in effect neutral
- if 1 allele: $\mathrm{P(RMNE)} = 1-(1-p_A)^2 = 2p_A - p_A^2 < 2p_A$
  - called the "2p rule"
  - claimed to be conservative because of "$<$"
- if 2 alleles (single contributor): no dropout
  - usual match probability applies.

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- difficult to combine with other evidence.

Likelihood ratio: one unknown and one known contributor

We seek expressions for single-locus likelihood ratios of the form

$$LR = \frac{\mathrm{P}(\text{DNA evidence} \mid s \text{ and } v \text{ source})}{\mathrm{P}(\text{DNA evidence} \mid x \text{ and } v \text{ source})}$$

where:

$s \equiv$ defendant (or suspect)
$v \equiv$ known contributor (e.g. victim)
$x \equiv$ an unknown individual assumed to be unrelated to s

and the DNA evidence consists of the profiles of s and v and the crime scene profile (CSP).

Data Reduction: observed alleles and dropout

Ideally we would analyse all the DNA evidence – including all the details of every epg. We would then model the background noise, and all the peak heights whether allelic or not.

- Very ambitious task for statistical modelling.

Instead, we assume that the full epg data are reduced to a list of alleles reaching a threshold. We then need to define

- **dropout**: an allele that generates a subthreshold peak, or no peak, due to part or complete failure of the PCR reaction.

The heights of sub-threshold peaks are informative

- but difficult to analyse in a systematic way, so
- ignoring this information may be a pragmatic option.

Our LR is based only on yes/no information for alleles observed and dropped out.

Dropout probabilities for a single allele

$D_a \equiv$ P(allele a drops out)

- depends on DNA template level, allele fragment length and level of degradation of the sample. It can also vary according to locus-specific and lab-specific factors.
- Features of the epg, such as heights of observed peaks, total number of observed alleles, and number of loci showing complete dropout, can be informative about D_a, together with the results of controlled experiments.
- Here we will not focus on estimating D_a but instead explore how the LR varies as a function of D_a.
- Simplifying assumption: $D_a \equiv D$ for all a
  - not realistic
  - may be a pragmatic assumption when data are lacking for modelling D_a.

Dropout probabilities for homozygotes

Assume we know D_a

- what is D_{aa}, the probability of dropout for an *aa* homozygote?

First guess: dropout is all-or-nothing, and occurs independently for the two alleles of a homozygote

- then $D_{aa} = D_a^2$

But often dropout (in our sense) is partial. Two signals may individually fail to reach the reporting threshold but reach it when superposed

- so $D_{aa} < D_a^2$.

On the basis of a small amount of data kindly provided by LGC, I have assumed that $D_{aa} = \alpha D_a^2$ and in practice used $\alpha = 1/2$.

- not realistic when $D \to 1$ (since D_{aa} is never $> \alpha$)

LR calculation: no v; $s \equiv AB$, $CSP \equiv A$; x, s unrelated

We assume D_a and D_{aa} constant over a (write D and D_2).

$$\begin{aligned} LR &= \frac{\mathrm{P}(CSP \equiv A, s \equiv AB | H_s)}{\mathrm{P}(CSP \equiv A, s \equiv AB | H_x)} \\ &= \frac{\mathrm{P}(CSP \equiv A | s \equiv AB, H_s)}{\mathrm{P}(CSP \equiv A | s \equiv AB, H_x)} \\ &= \frac{(1-D)D}{p_A^2(1-D_2) + 2p_A(1-p_A)(1-D)D} \end{aligned}$$

- In numerator, allele B of s dropped out but A did not.
- Denominator involves sum over all possible genotypes for x.
- We assume HWE and known population allele proportion p_A.

Allowing for masking

Masking can be from alleles of v directly, or from artefacts such as stutter from an allele of v.

- Treating stutter as masking means that there may or may not be an allele of x underlying a stutter peak generated by an allele of v.

Let M denote the set of masking alleles, $m = |M|$, and write p_M for the sum of the allele proportions of the elements of M. Here CSP denotes only the non-masking alleles, so that $CSP \cap M = \emptyset$. Then

$$\begin{aligned} LR &= \frac{\mathrm{P}(CSP \equiv A | s \equiv AB, M, H_s)}{\mathrm{P}(CSP \equiv A | s \equiv AB, M, H_x)} \\ &= \frac{D(1-D) \quad \text{if } B \notin M, \quad (1-D) \quad \text{otherwise}}{p_A^2(1-D_2) + 2p_A(1-D)[(1-p_A-p_M)D + p_M]} \end{aligned}$$

Always $<$ non-masking LR.

Allowing for dropin

- Dropin refers to presence of tiny fragments of DNA that can generate a spurious allele, but not a whole genome contribution from an additional unknown contributor.
- Relevant even if not evident under prosecution case.
- Multiple dropin events might be better modelled as an additional contributor with dropout.

If one dropin allele possible:

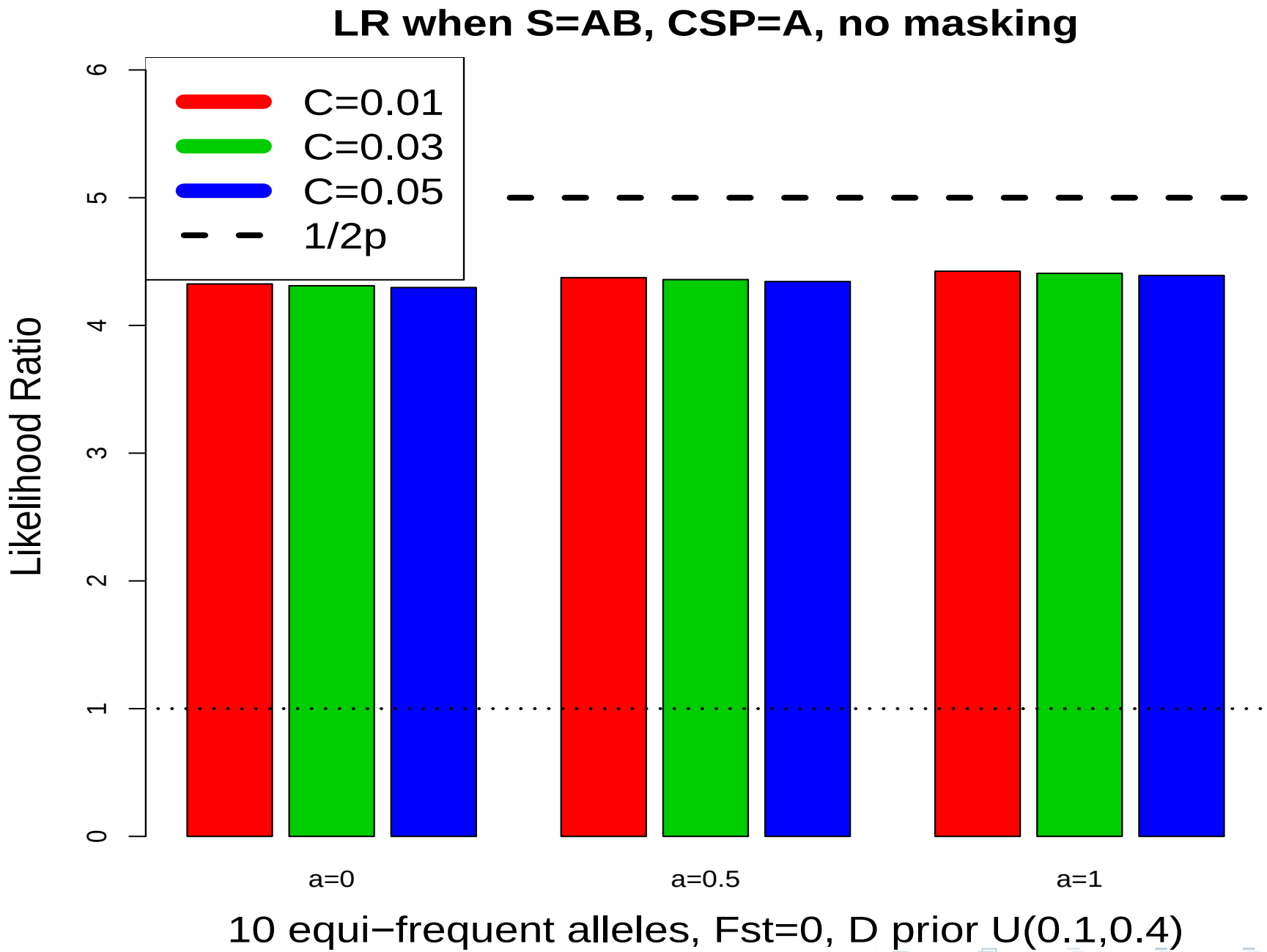
$$LR = \frac{(1-D)D(1-C)}{Q(1-C) + Q'Cp_A} < \text{no dropin } LR$$

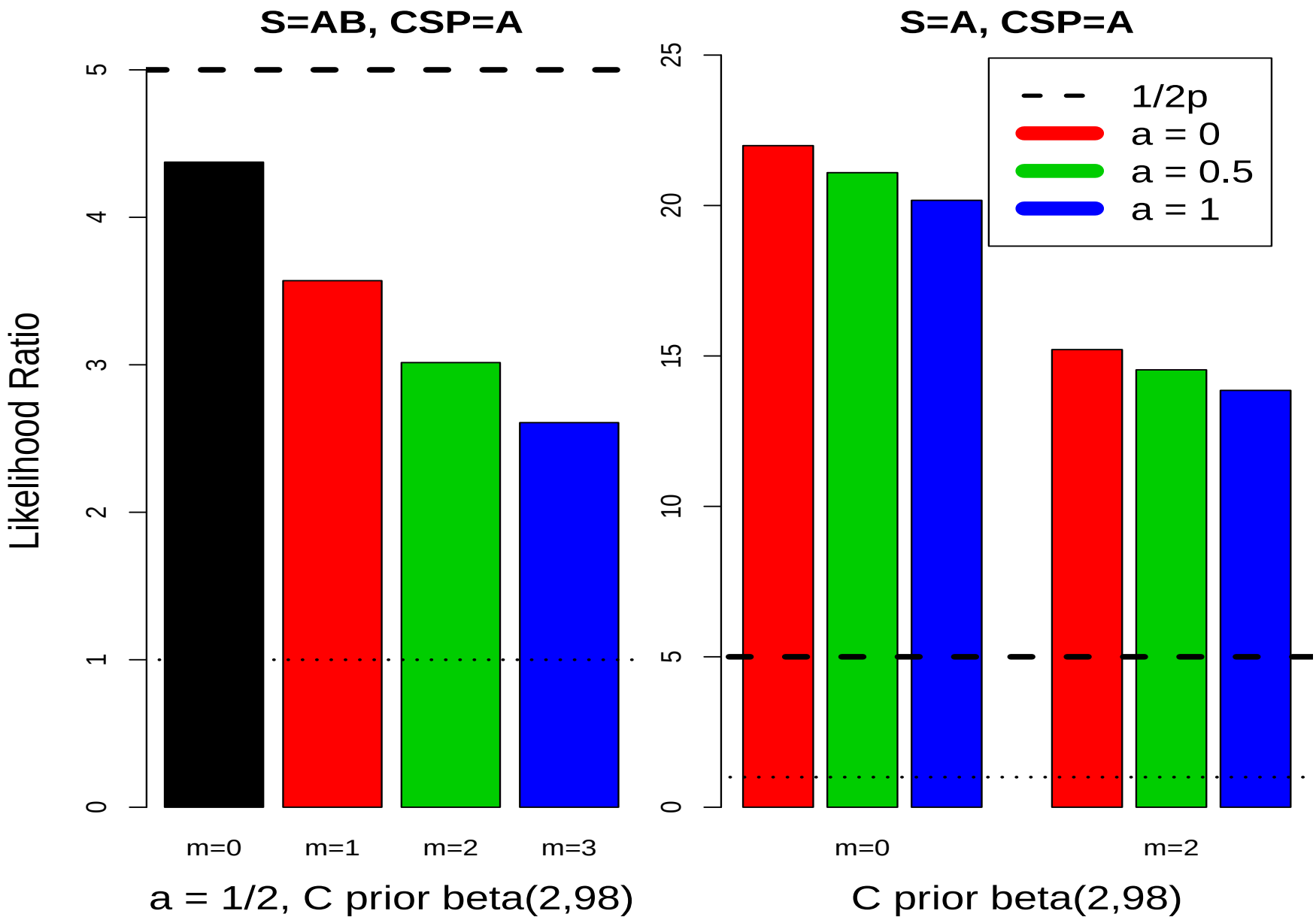
where

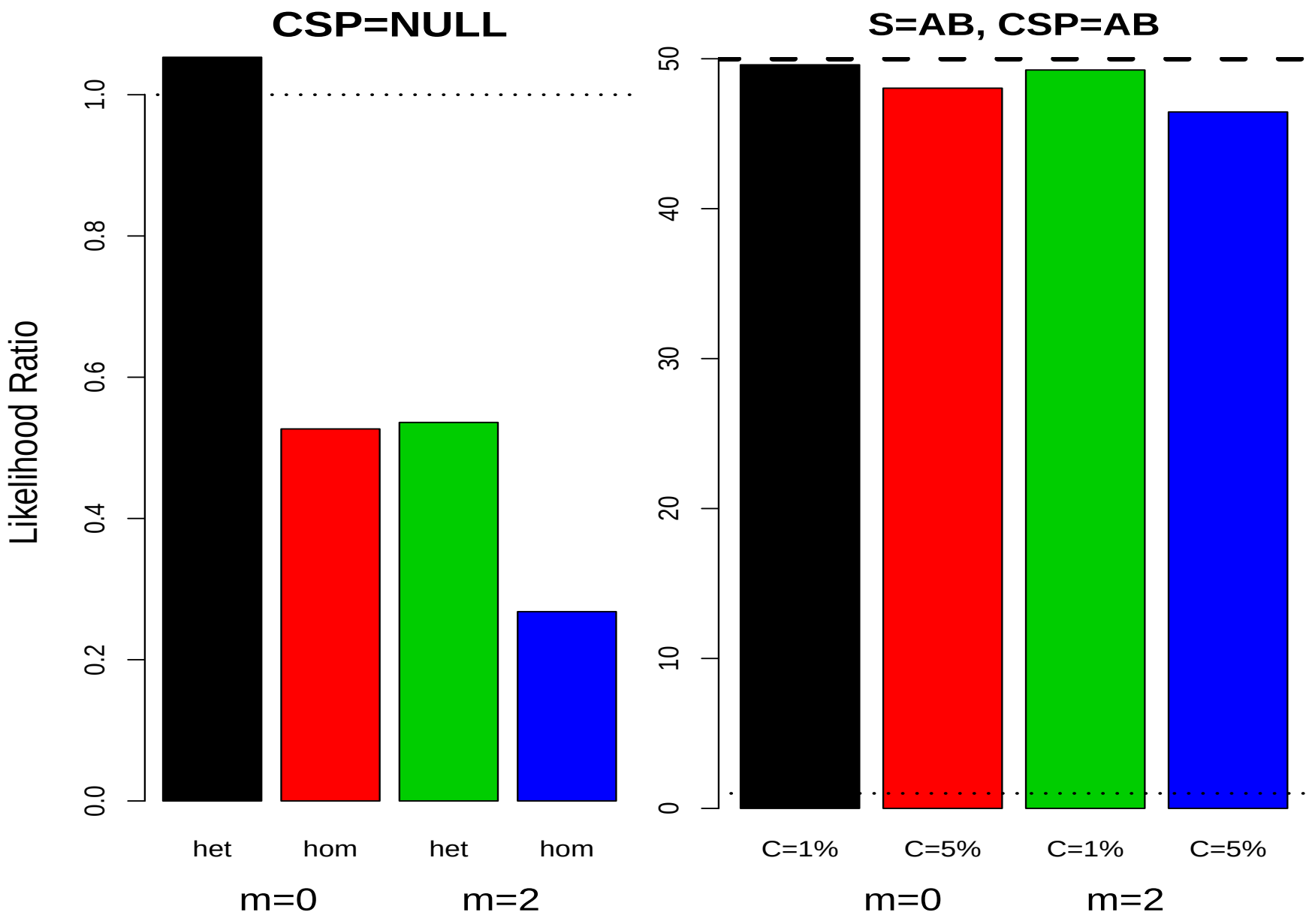
$$\begin{aligned} Q &= \text{denominator from masking } LR \\ Q' &= \text{P(both alleles of } s \text{ drop out)} \end{aligned}$$

In the plots that follow we assume ten alleles at the locus, each with $p = 0.1$;

The 2p rule is not conservative




S=AB, CSP=A
S=A, CSP=A
Likelihood Ratio
0
1
2
3
4
5
0
5
10
15
20
25
1/2p
a = 0
a = 0.5
a = 1
m=0
m=1
m=2
m=3
m=0
m=2
a = 1/2, C prior beta(2,98)
C prior beta(2,98)


CSP=NULL
S=AB, CSP=AB
Likelihood Ratio
0.0
0.2
0.4
0.6
0.8
1.0
0
10
20
30
40
50
het
hom
het
hom
C=1%
C=5%
C=1%
C=5%
m=0
m=2
m=0
m=2

LR values vs RMNE

- The RMNE formulae are almost always unfair to defendants when dropout and dropin are possible.
  - A null observation can be very informative in presence of masking.
  - Even with no masking, a null result it is (slightly) incriminating for a heterozygote and exculpatory for a homozygote.
  - $2p$ rule is always unfair to defendant, especially with masking.
  - Even $2p_A p_B$ (when A and B alleles are observed) is not fair when dropout and dropin are possible, and particularly when there is masking.
- LR can point towards or away from s, possibly depending on D and D_2.
  - RMNE can never favour defendant.

Case of Smith (Sacramento, CA)

- CSP shows mixture of DNA from at least 2 persons
  - one known male contributor k, profiled
  - from X and Y peak heights, roughly even mixture of male and female
  - assume one unknown (female) contributor
  - suspected female contributor s, profiled
- Large amount of dropout from both contributors
  - no alleles observed at 5 out of 15 loci $\Rightarrow D \approx 0.75$
- Prosecution reported a probability of 1 in 96K, obtained by applying a multiple-contributor RMNE only to the seven loci at which an allele of s was observed in the *CSP*.

Locus	Known k	Suspected s	CSP	RMNE
D8	13,16	12,13	12,13,16	4
D21	28,30	29,29	28	1
D7	8,10	9,10	—	1
CSF	8,10	10,11	—	1
D3	14,16	16,17	16	4.3
THO1	7,7	9.3,9.3	7	1
D13	11,13	8,12	—	1
D16	12,13	11,12	12,13	4.0
D2	19,24	17,25	24	1
D19	12,13	13,15	12,13	6.5
VWA	18,20	19,20	18,20	18
TPO	9,9	11,12	9,11	7.0
D18	13,15	12,17	—	1
D5	8,12	11,13	8,11,12	1.7
FGA	21,22	22,24	—	1
Product				96K

Criticisms

- at 4 of 7 loci used for RMNE probability, all CSP alleles are in known profile
  - little or no support for claim of suspect DNA being present
- RMNE takes no account that at two loci for which suspect is homozygous, the allele was not observed in CSP (both homozygotes of known contributor were observed)
- LR calculation
  - assume D and D_2 are the same for both known k and suspected s contributors
  - two assumptions about the 9 alleles shared by k and s (5 of these appear in CSP, 4 do not):
    1. they are assumed "masked" by k with certainty
    2. they are assumed to come from k with probability 0.5, otherwise from unknown contributor.
  - results below are under assumption 1; assumption 2 leads to slightly smaller LR.

	Known	Suspected		LR	RMNE	
Locus	k	s	CSP	D=0.5	court	modified
D8	13,16	12,13	12,13,16	4.5	4	3.4
D21	28,30	29,29	28	0.38	1	1
D7	8,10	9,10	—	1.1	1	1
CSF	8,10	10,11	—	1.1	1	1
D3	14,16	16,17	16	1.4	4.3	1
THO1	7,7	9.3,9.3	7	0.38	1	1
D13	11,13	8,12	—	1.1	1	1
D16	12,13	11,12	12,13	0.93	4.0	1
D2	19,24	17,25	24	0.84	1	1
D19	12,13	13,15	12,13	1.1	6.5	1
VWA	18,20	19,20	18,20	1.4	18	1
TPO	9,9	11,12	9,11	1.3	7.0	2.0
D18	13,15	12,17	—	1.1	1	1
D5	8,12	11,13	8,11,12	0.88	1.7	1.2
FGA	21,22	22,24	—	1.1	1	1
Product				2.0	96K	8.3

Conclusions

- Much progress has been made in the interpretation of DNA profile evidence.
- Likelihood ratios are crucial to this advance
  - must be understood within the context of Bayes Theorem
- Many problems yet to be overcome.
- Some instances of good practice in presenting DNA evidence in court.
- Much remains unsatisfactory.