NEW YORK STATE | DNA Subcommittee

NIST FORENSIC SCIENCES

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Addressing Questions on DNA Interpretation

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Some Key Principles

- Everything in **science involves** mapping observed data to **models** (“hypotheses”)
  - Hardy Weinberg Equilibrium (HWE) models expected genotype frequencies (p^2 or 2pq) assuming unrelated individuals
  - Theta corrections (θ=0.01 or θ=0.03) model potential variation from assumptions of unrelated individuals

- All **models require assumptions,** some of which are more reliable than others depending on data obtained

- **Validation studies generate data that inform the model being used** or enable a model to be constructed
  - For example, a test for HWE is comparing population (validation) data to a model to see goodness-of-fit

- **Genotypes—not alleles—matter** in deciphering mixtures

- **Probabilistic genotyping involves modeling observed data** against potential genotype combinations

Purpose of MIX13 Cases

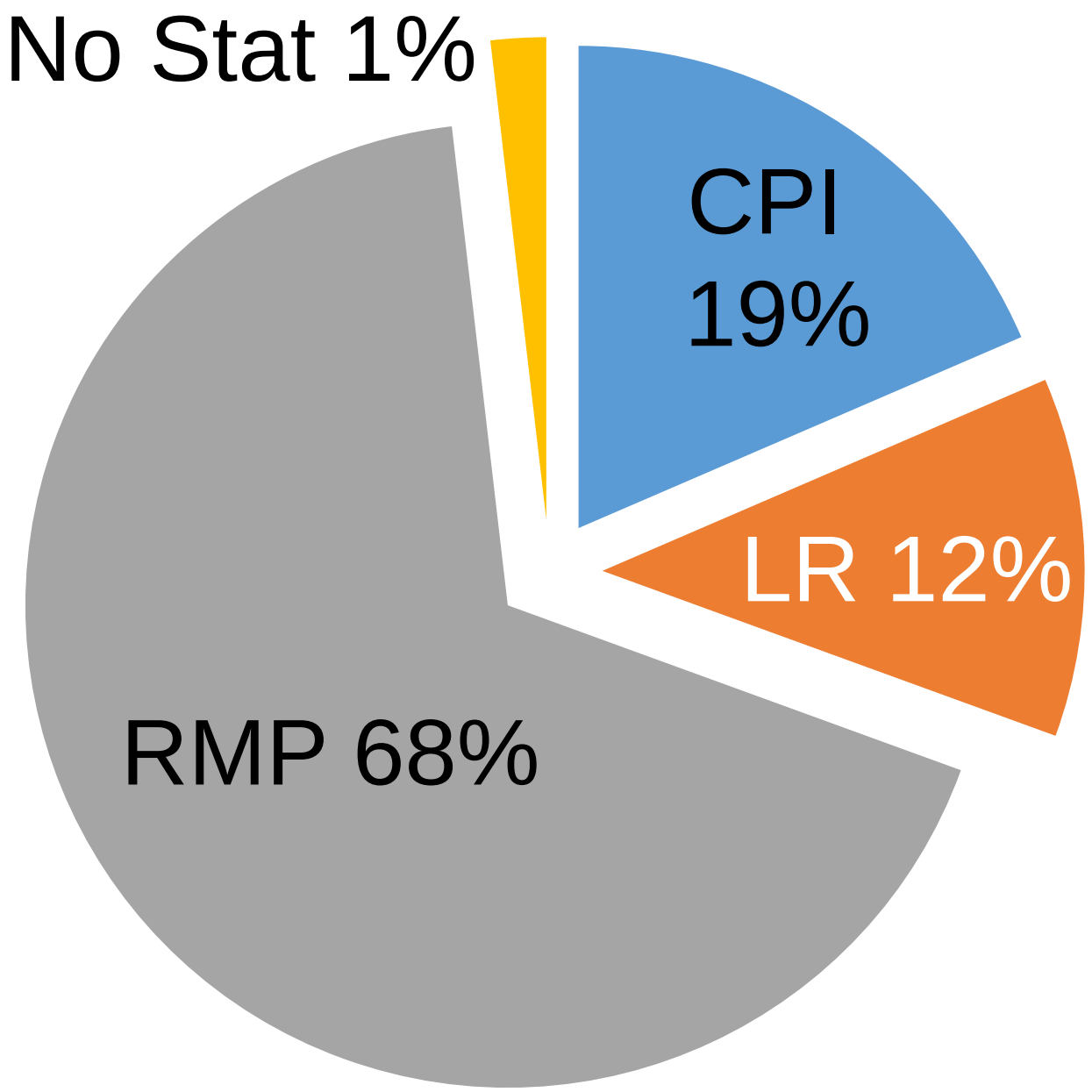
	Challenge provided to study responses
Case 1	~1:1 mixture (2-person)
Case 2	**Low template** profile with potential dropout (3-person)
Case 3	Potential **relative** involved (3-person)
Case 4	Minor component (2-person)
Case 5	Complex mixture (>3-person) with # **of contributors**; inclusion/exclusion issues

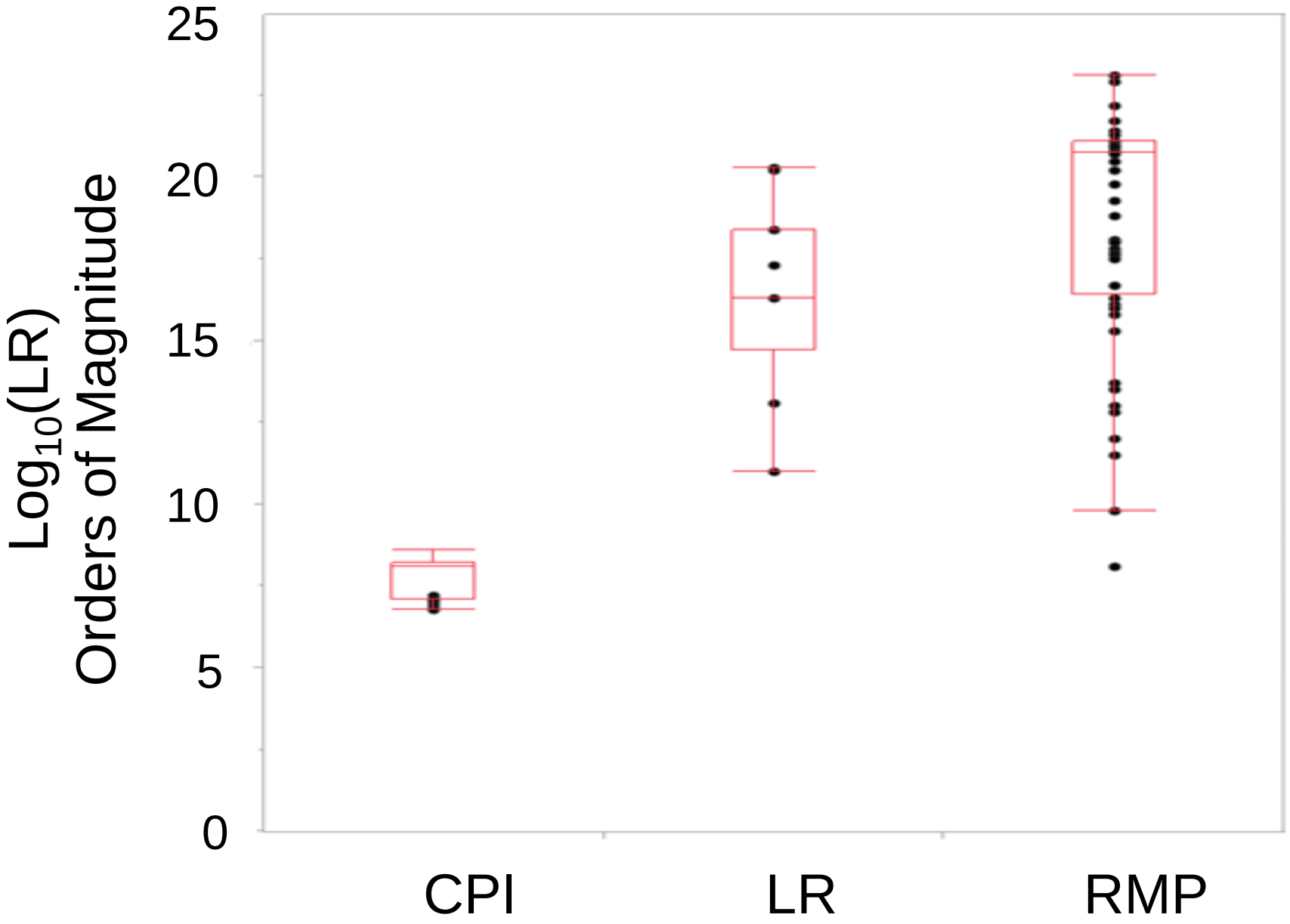
According to German Stain Commission (2009) mixture types: 1 = A, 2 = C, 3 = ?, 4 = B, 5 = ?

MIX13 Study (Case 01)

- Summary – Mock sexual assault, 2 person 50:50 mixture, all alleles above a ST of 150 RFU.

- Purpose – How many labs would consider the victim's profile and determine genotypes (deconvolution) for a mRMP statistic?


No Stat 1%
CPI 19%
LR 12%
RMP 68%


25
20
15
10
5
0
Log10(LR)
Orders of Magnitude
CPI
LR
RMP

MIX13 Study (Case 02)

- Summary – Mock handgun (touch DNA), 3 person **6**:**1.5**:**1** mixture, total DNA amplified was 300 pg, potential for drop-out with the 2 low-level contributors. An additional contributor profile (suspect D) was provided, but is not in the mixture.

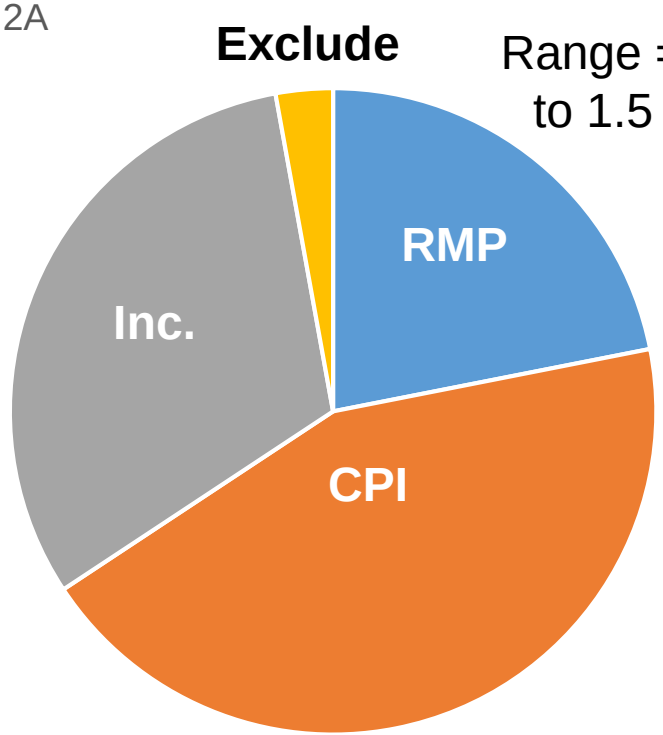
- Purpose – How many labs would consider this mixture as too complex to interpret?

Primary Goals

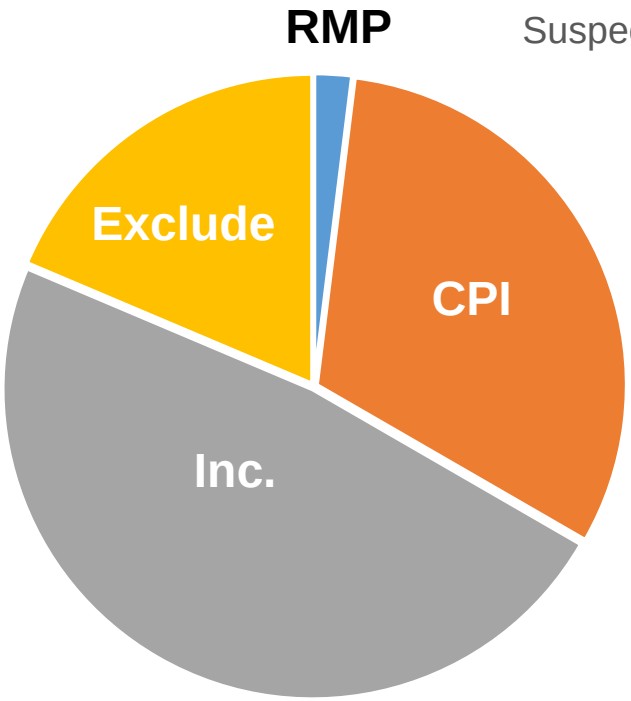
- Most labs – CPI for some combination of Suspects A, B and C using a limited number of loci.

- One lab included Suspect D (Not in the mixture).

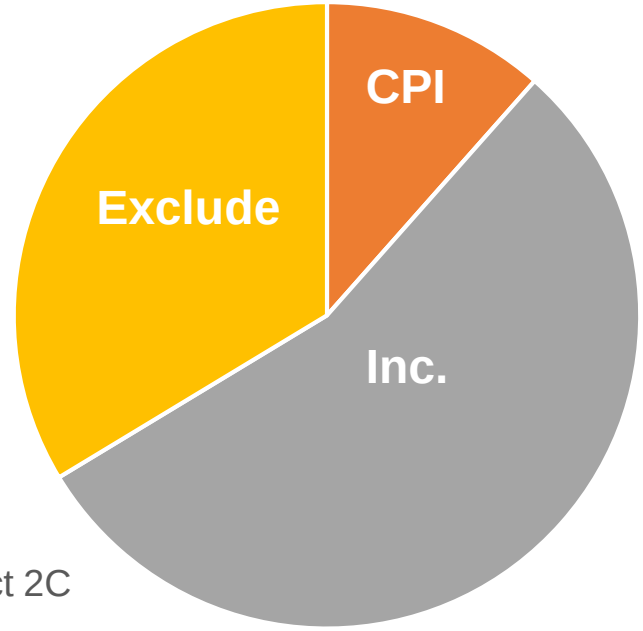
Suspect 2A



Range = 100M to 1.5 Quad

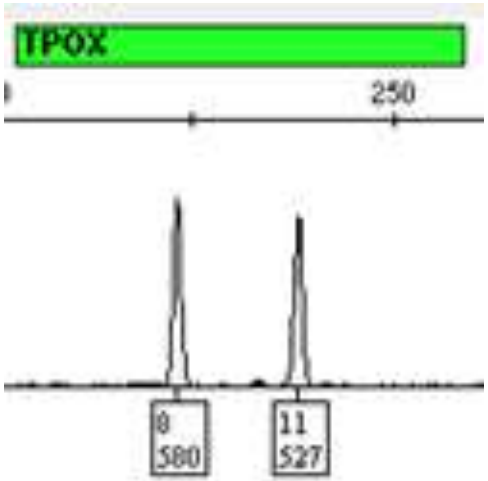


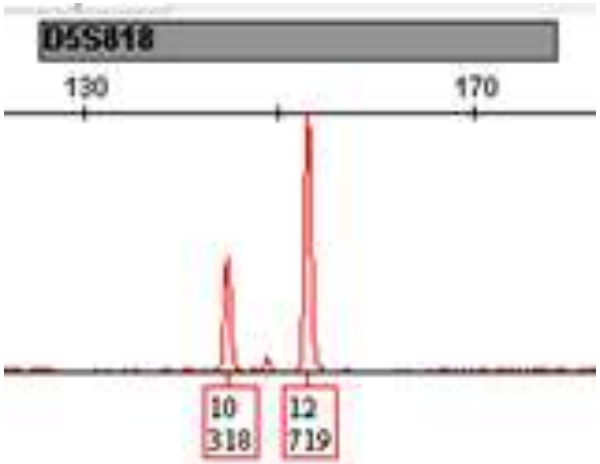
Suspect 2B

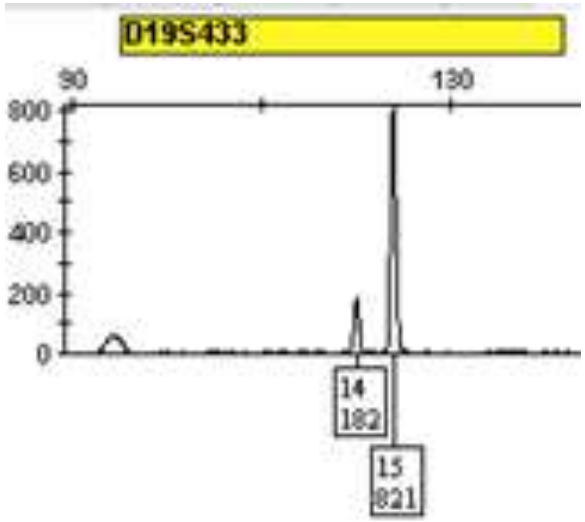


Range = 2.8 to 15K

Suspect 2C


TPOX
250
8
580
11
527


D5S818
130
170
10
318
12
719


D19S433
90
130
800
600
400
200
0
14
182
15
821

~ 1 in 35

Intra-Laboratory Results (n = 8)

Analyst	Suspect A	Suspect B	Suspect C	Suspect D
1	Inconclusive - A, B, C			Excluded
2	6.74 Quad	**23.6**	Excluded	Excluded
3	Inconclusive - A, B, C			Excluded
4	**9.4** for A, B, C			Excluded
5	4.1 Quint	**37**	Excluded	Excluded
6	**230** for A, B		Inconclusive	Excluded
7	**9.4** for A, B		Excluded	Excluded
8	**37.3** for A, B		Excluded	Excluded

MIX13 Study (Case 03)

- Summary – Mock sexual assault, 3 person **7**:**2**:**1** mixture, The two minor contributors are brothers, An additional contributor profile (suspect 3B) was provided, but is not in the mixture.

- Most of the suspected brother's alleles are masked in the mixture

- Purpose – Given the relatedness of the individuals in the mixture, is this too complex for interpretation?

Primary Goals

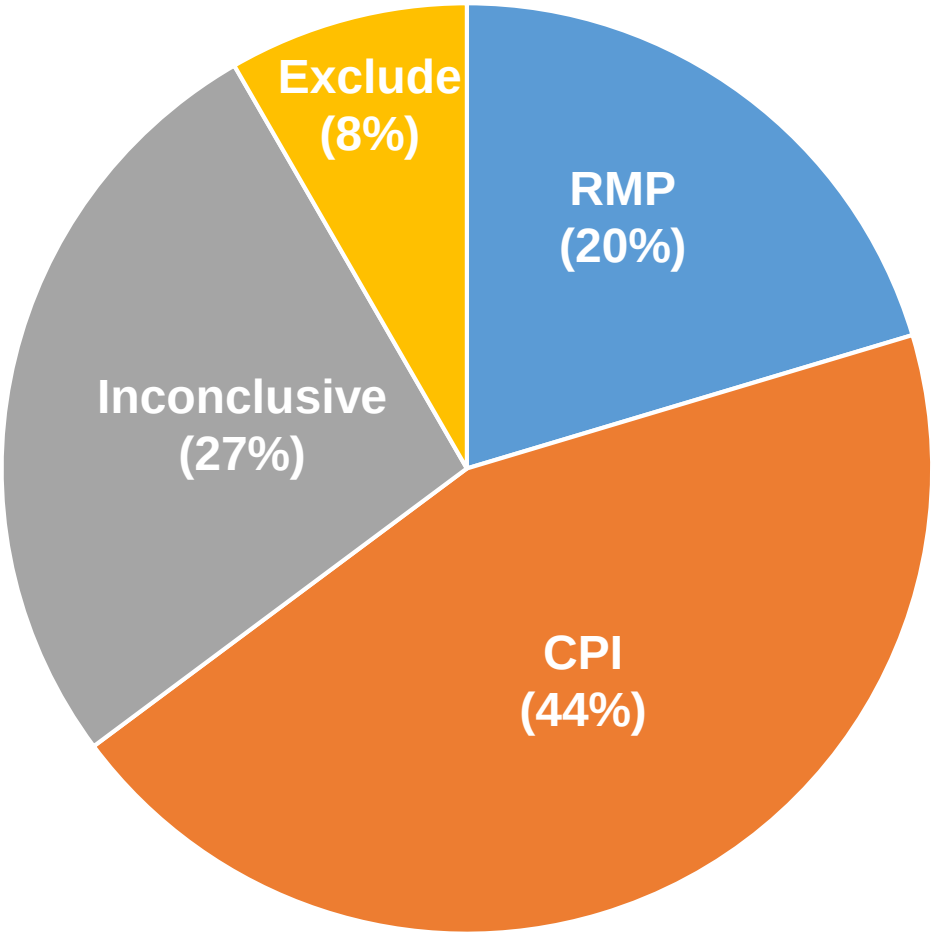
- Only one lab included Suspect B (Not in the mixture)
- Most labs are using CPI stats for this case…

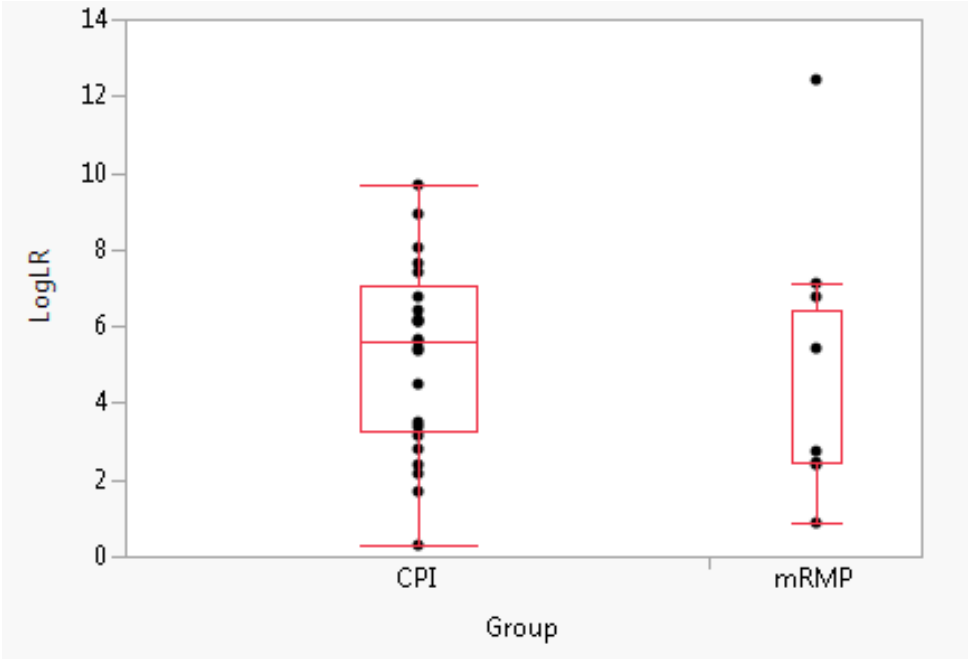
RMNE

- Random Man Not Excluded (CPE/CPI) – The probability that a ***random person*** (unrelated individual) would be excluded as a contributor to the observed DNA mixture.

- Only a few labs have stated this – “Due to the relatedness of the exemplars submitted for comparison, a statistical analysis cannot be provided at this time.”

Case 03


Exclude
(8%)
RMP
(20%)
Inconclusive
(27%)
CPI
(44%)


14
12
10
8
6
4
2
0
LogLR
CPI
mRMP
Group

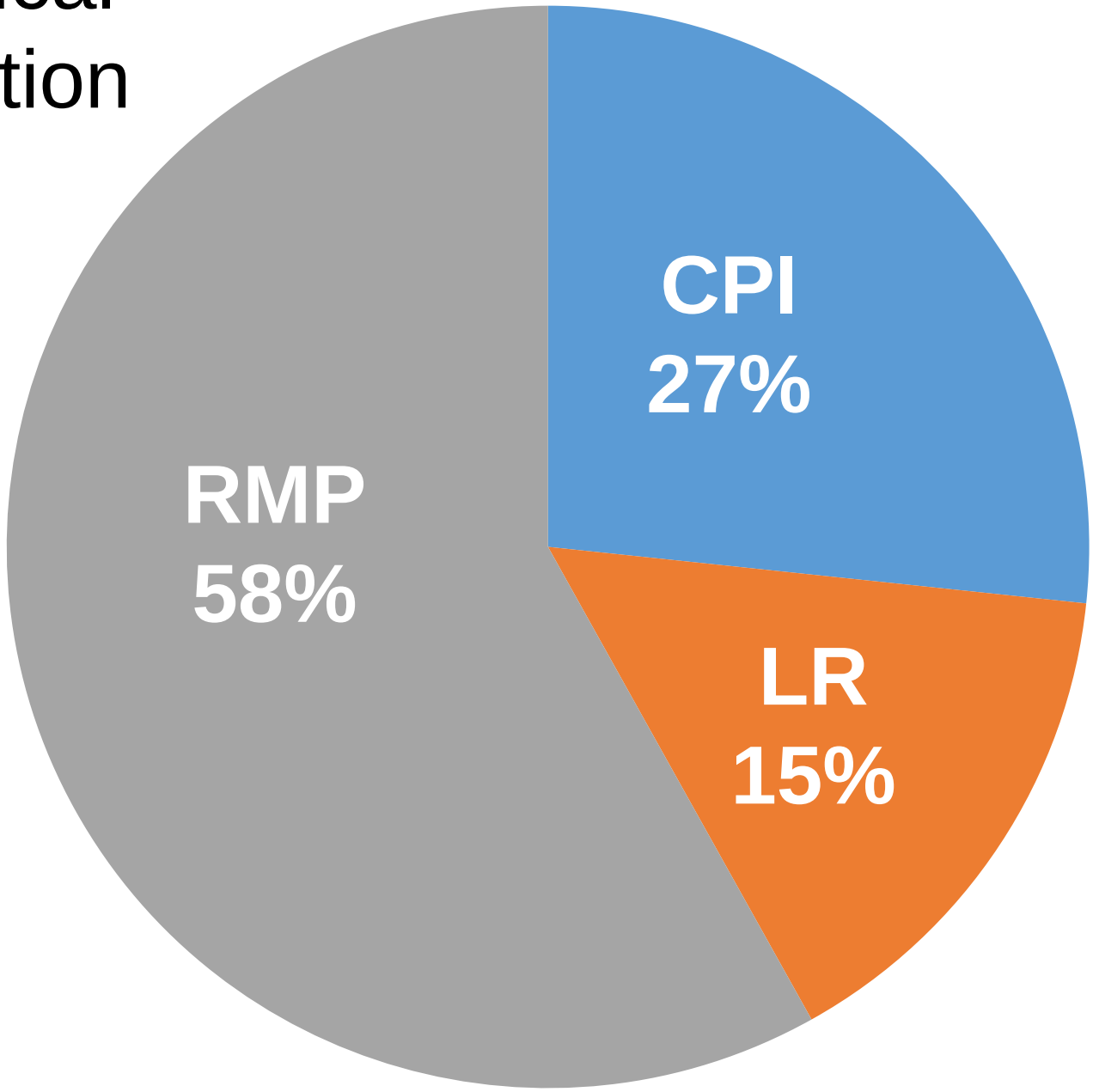
MIX13 Study (Case 04)

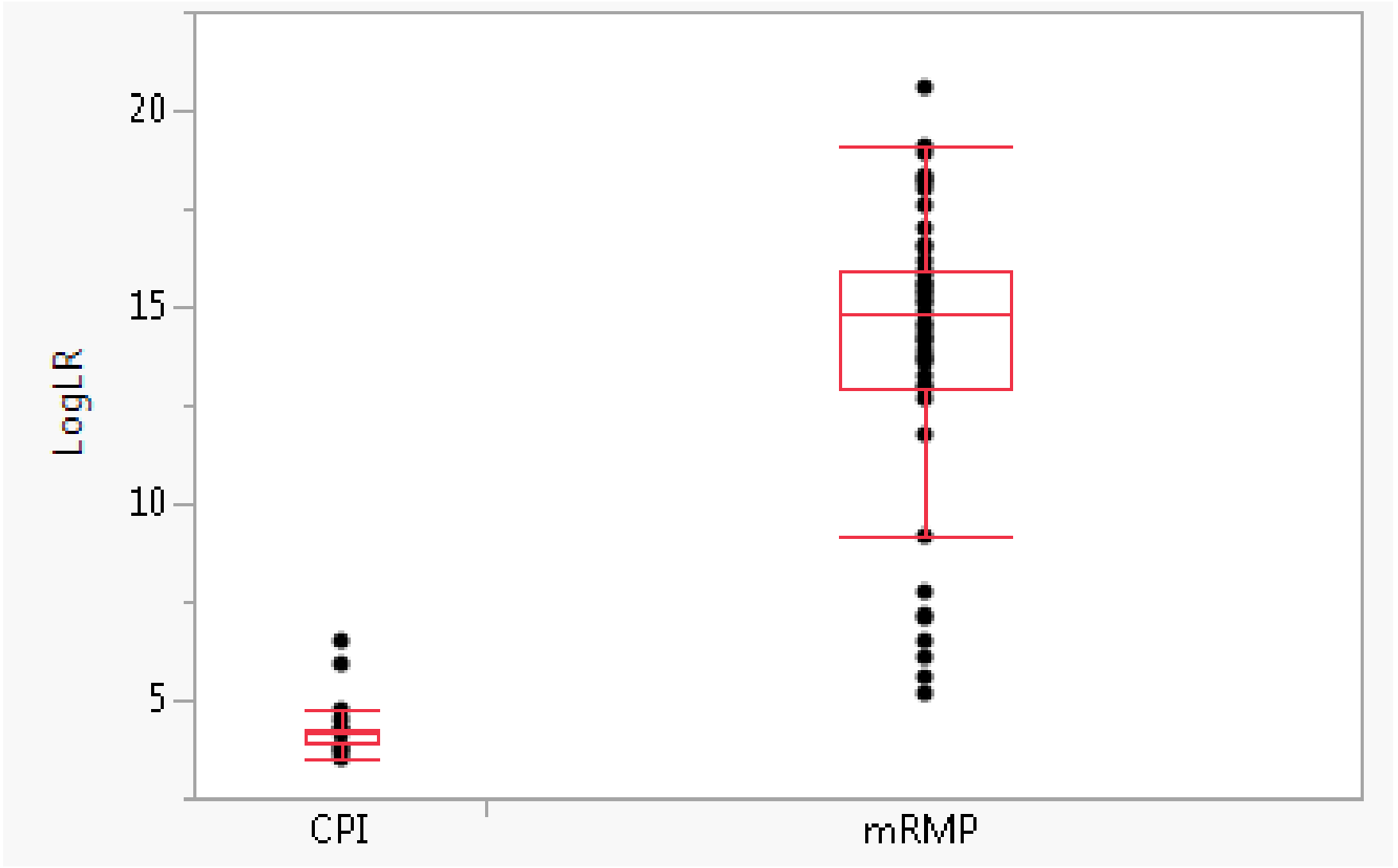
- Summary – Mock sexual assault, 2 person **3.5**:**1** mixture, minor component has alleles below the ST of 150 (required by all labs!)

- Purpose – How many labs would attempt to separate the two components?

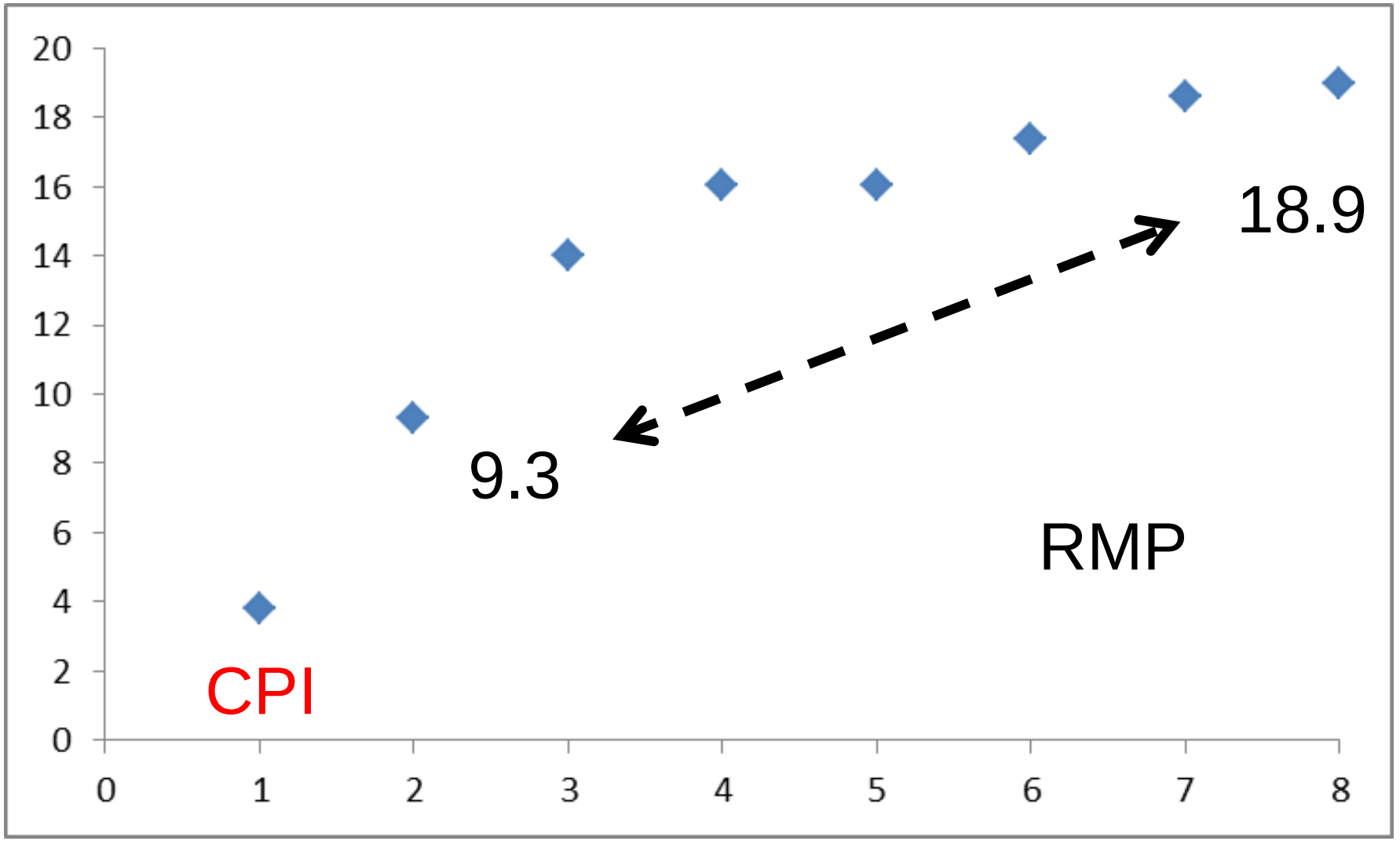
- With all labs using the AT/ST – how much variation is expected?

Statistical Evaluation




LogLR
20
15
10
5
CPI
mRMP

Intra-Laboratory Results (n = 8)



MIX13 Study (Case 05)

- Summary – Mock bank robbery with ski mask evidence (touch DNA), 4 person **1:1:1:1** mixture.

- However – this mixture had no more than 4 alleles at any locus (appears as a 2p mixture). 2 of the 4 contributors were provided along with a non-contributor.

- Purpose – How many labs would consider this mixture as too complex to interpret?

MIX13 Case 5 Outcomes with Suspect C
(whose genotypes were not present in the mixture)

# Labs	Report Conclusions	Reasons given
7	Exclude Suspect C	detailed genotype checks (ID+); TrueAllele negative LR (ID+); assumed major/minor and suspects did not fit (ID+); 4 of 18 labs noted Penta E missing allele 15 (PP16HS)
3	Inconclusive with C only (A & B included)	All these labs used PP16HS
22	Inconclusive for A, B, and C	
76	**Include & provide CPI statistics**	*All over the road…*

Range of CPI stats for Caucasian population:

FBI allele frequencies: **1 in 9** (Labs 12 & 54) to **1 in 344,000** (Lab 107)

National Commission on Forensic Science (NCFS):
www.justice.gov/ncfs

Organization of Scientific Area Committees (OSAC):
www.nist.gov/forensics/osac/index.cfm

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