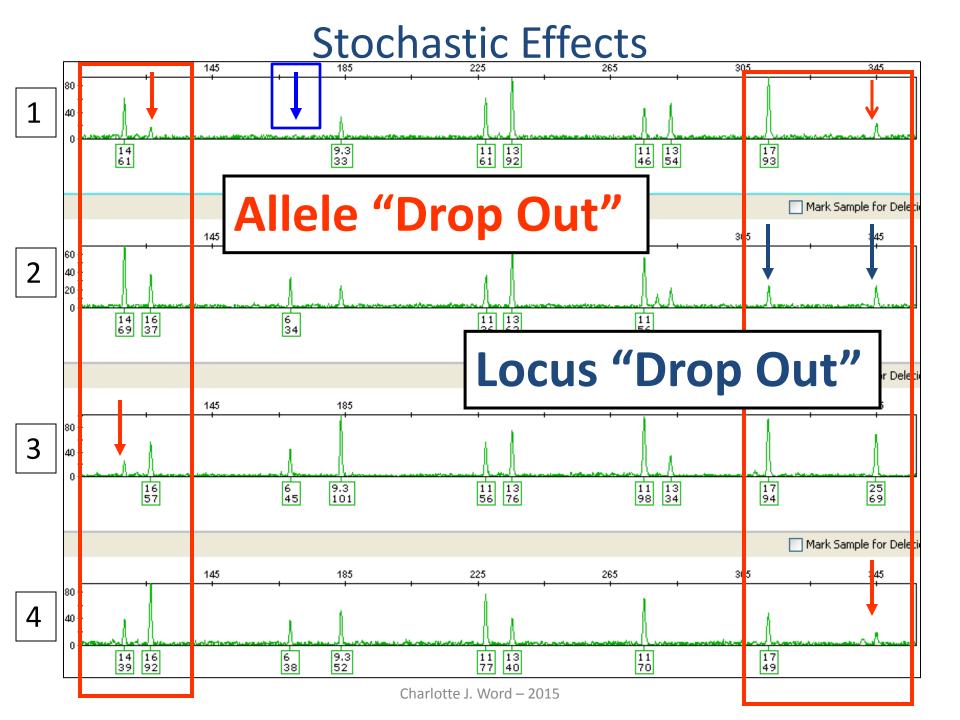


## Errors in Interpretation of DNA Profile Data

Charlotte J. Word, Ph.D. July 22, 2015

Low Template DNA & Stochastic Effects **Elevated Stutter/** 9.3 33 11 13 61 92 Allele Drop In **Peak Imbalance** 14 16 69 37 34 11 13 36 62 56 Mark Sample for Deletion 9.3 101 56 76 98 94 Mark Sample for Deletion 9.3 52 77 40 70 49 Charlotte J. Word – 2015



#### DNA NOT the Gold Standard?

#### **Bulk of Samples Tested Today:**

- Low Template (LT) DNA
  - Technical "errors" from stochastic effects
    - Drop in, elevated stutter
  - Lack of reproducibility of alleles, peak heights, peak height ratios
  - Uncertainty in data
- Complex Mixtures
  - High sensitivity amplification kits → 1 or more contributors are LT DNA

#### DNA NOT the Gold Standard?

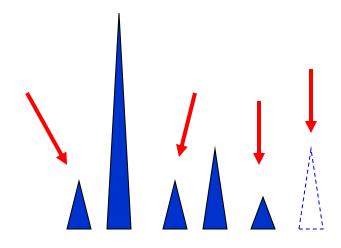
- DNA profile interpretation & comparison difficult
  - False inclusions, False exclusions
  - Incorrect use of CPI calculation
  - Lack of consistency
- Is Probabilistic Software the solution?
  - -Maybe...
  - -But there are still issues....

#### **Complex Mixtures**

#### Uncertainty

- > Peak vs. Artifacts
  - ➤ Stutter?
  - ➤ Pull-up?
  - ➤ Real Allelle?

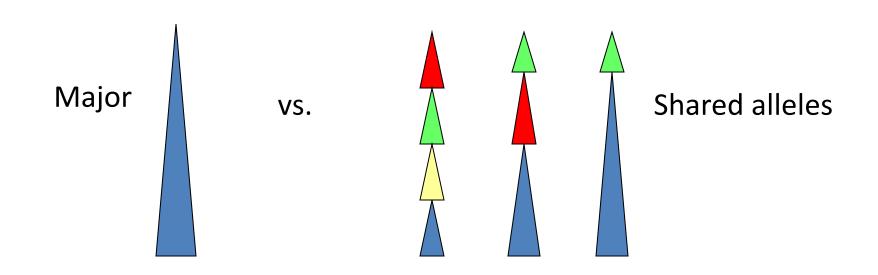
➤ Missing Alleles?



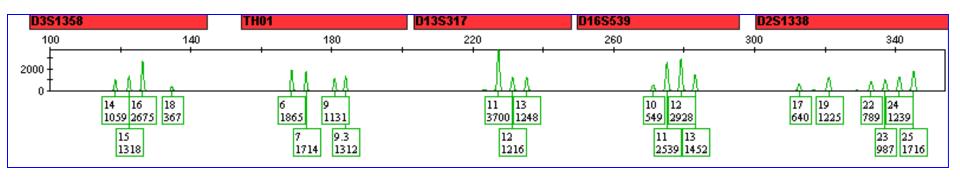
#### **Complex Mixtures**

#### **More Uncertainty**

- Major contributor?
- Shared Alleles?
- Related contributors?



#### **Apparent Three Person Mixture**

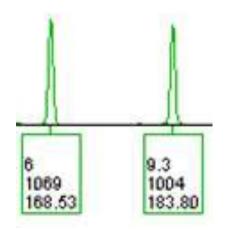


## But is it? How confident can we be in that assumption?

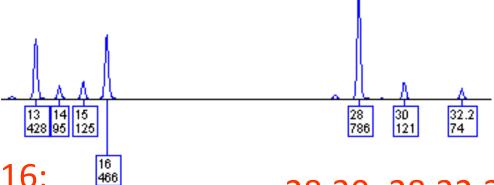
 High risk that 4 person (>70%) or 5 person (40%) or 6 person (14%) mixture would look like a 3 person

What Number of Contributors to assume for calculations?

## Exclusion Criteria Can Exclude:



All but 6,9.3



13,14; 13,15; 14,16;

15,16; 13,13; 16,16

any without

13,14,15,16

28,30; 28,32.2;

30,30; 32.2, 32.2;

any without 28, 30

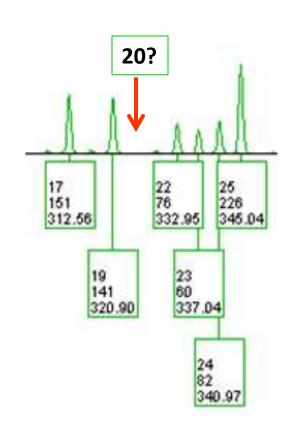
or 33.2

#### How do we RELIABLY Exclude a noncontributor?

#### What are our Exclusion Criteria?

If "missing" an allele in a profile...is it due to:

- Stochastic effects and failure to amplify? OR
- Person is not a contributor to the mixture?



Is it a 3 or 4 or 5 person mixture?

#### What do the DNA data REALLY mean?

"Four person mixture" on a handled item – "cannot be excluded"

- He touched the item
  - Secondary, tertiary transfer?
- "Major" contributor
  - He was the last person to touch the item
  - Sweating the most, high shedder, .....
  - Is there even a major contributor profile?
  - Confirm major DNA profile with a CODIS "hit"
    - Circular argument is it the "right" person?

#### What do the DNA data REALLY mean?

- He is actually a true contributor "he's in there"
  - But really only a possible contributor
- Guilty of the crime (based on possible presence)

What is the investigator telling the suspect? What is the prosecutor telling the suspect? What is the defense attorney telling the suspect? suspect?

What is being said in opening and closing statements to the trier of fact?

Is it right?

#### LR Calculations

- What assumptions to use?
  - -Hp vs Hd?
  - -3, 4, 5 contributors?
  - What if program can only do 3 contributors? 4 contributors?
  - What to report?
- Does the defense have a hypothesis? Do they need to have one?

## Do We Know What the LR Means? Are We Communicating LR Effectively?

- LR = 1 million What if both hypotheses are wrong?!
  - -For single source means 1 in 1 million (RMP)
  - "Very strong support" that "he contributed the DNA" (transposed conditional?)
- What does it mean for a 4 person mixture?
- What does LR of 10,000 mean? 1000? 100?
   10? 3?
- DQ/PM days 1 in 1000 limited meaning for single source sample

#### Are We Answering the Right Question?

#### The questions that **need** answering are:

- 1. Is he a contributor?
- 2. Does the presence of the DNA mean anything in relation to the crime?
- 3. Is he guilty?

What does it mean that this observed profile is X times more likely if he is a contributor along with Y other unknown contributors?

### Questions to Ask? Do we need....?

- Additional validation studies?
- Better proficiency tests? Inter-laboratory studies?
- Additional training?
  - For analysts?
  - For law enforcement?
  - For attorneys?
  - For judges?



Improved standards and recommendations?

#### Questions to Ask?

- Should we be testing these LT DNA and complex mixture samples?
  - Technical and interpretation issues
  - Meaning and relevance to the case
  - High risk of misinterpretation and misrepresentation
- Should we modify case acceptance policies?
- Improve collection techniques? Handling techniques?
- Where do we stop? 3 person? 4 person? 5 person? More?

#### Questions to Ask?

- What does it mean to have DNA from 50 cells?
   20 cells? 4 cells? With no visible "stain"?
  - Mixed with several other people?
- Is the DNA even relevant to the case?
- How will a falsely-accused individual "prove" his innocence?
  - No replicate testing done in US (except NYC)
  - No duplicate samples
  - Consumed samples not option to re-test if "better" test comes out

# WHO is talking about the limitations of these samples and assays?

Who is measuring the error(s) in the processes?

Are we doing the best that we can?

#### Thank You!

- For your attention!
- NIST
- Previous speakers
- John Butler, Mike Coble, Robin Cotton, Catherine Grgicak

