DNA Identification: Mixture Interpretation

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Cybergenetics

Fingernail DNA Evidence

An autopsy photograph presented in the talk showed a deep defensive knife wound to the back of the victim's right hand. The DNA evidence was extracted from under the indicated fingernails.

Feb 2008: 7% DNA component matches suspect

DNA Evidence

- DNA from under victim's fingernails (Q83)
- two contributors to DNA mixture
- 93.3% victim & 6.7% unknown
- 1,000 pg DNA in 25 ul
- STR analysis with ProfilerPlus®, Cofiler®
- know victim contributor genotype (K53)
- TrueAllele® computer interpretation
  (using genotype addition method)
   infer unknown contributor genotype
- only after having inferred unknown,
   compare with suspect genotype (K2)

Three DNA Match Statistics

Score	Method
13 thousand	inclusion
23 million	subtraction
189 billion	addition

- Why are there different match results?
- How do mixture interpretation methods differ?
- What results should be presented in court?

DNA Mixture Data
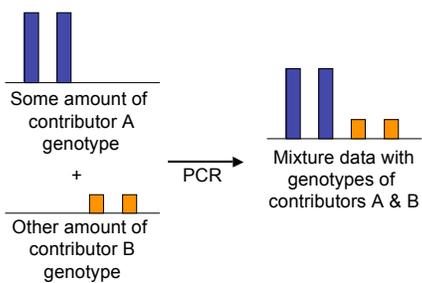

Some amount of contributor A genotype

+

Other amount of contributor B genotype

PCR →

Mixture data with genotypes of contributors A & B
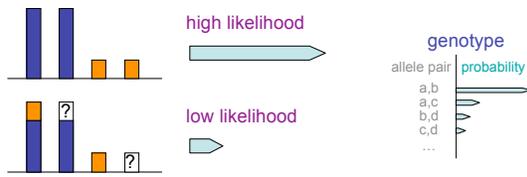

Cybergenetics

D7S820

200   Update

WF070403012Q83-1
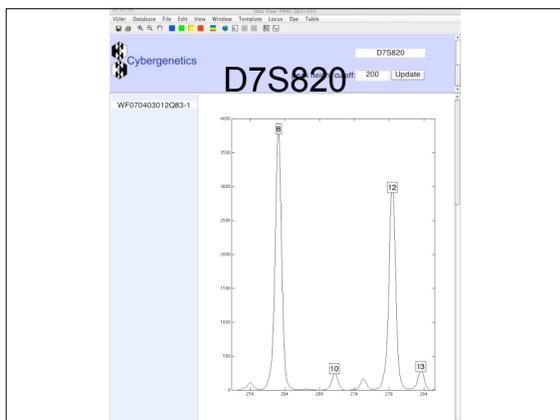
Quantitative Mixture Interpretation

Step 1: infer genotype

- consider every possible allele pair
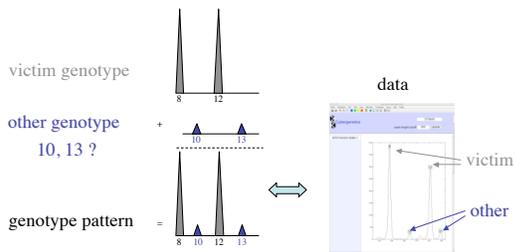- compare pattern with DNA data
- Rule: *better fit's more likely it*

high likelihood

low likelihood

genotype

allele pair	probability
a,b	
a,c	
b,d	
c,d	
…	

Quantitative Genotype

victim genotype

8   12

other genotype
10, 13 ?

+

10   13

data

victim

other

genotype pattern

=

8   10   12   13

Quantitative Information

At the suspect's genotype,
identification vs. coincidence?

after
(evidence)

↑ data

before
(population)

$$\frac{\text{Prob(suspect matches evidence)}}{\text{Prob(suspect matches population)}} = \frac{100\%}{1.72\%}$$

$$= 58$$

Step 2: match genotype

high probability retains LR information

Qualitative Manual Review

Step 1: infer genotype

Rule: *every pair gets equal share*

listed allele pairs
are all assigned
the same likelihood

genotype

allele pair	likelihood
a,a	⊳
a,b	⊳
a,c	⊳
a,d	⊳
…	

Qualitative Genotype

cutoff

8 10 12 13

8 10 12 13

8   10

• apply threshold
• discard peak data
• make all the same

• 10 possible pairs
• equal likelihood
• diffuse probability
• lose match strength
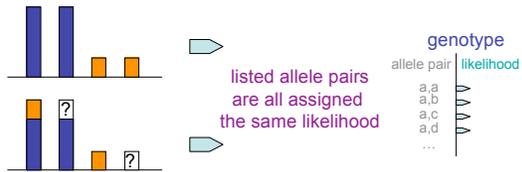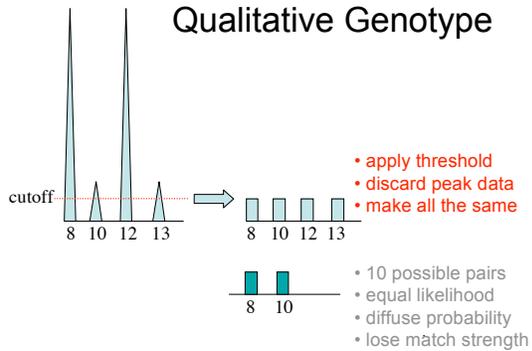
Qualitative Information

At the suspect's genotype,
identification vs. coincidence?

after
(evidence)

↑ data

before
(population)

$$\frac{\text{Prob}(\text{suspect matches evidence})}{\text{Prob}(\text{suspect matches population})} = \frac{4.42\%}{1.72\%}$$

= 2.57

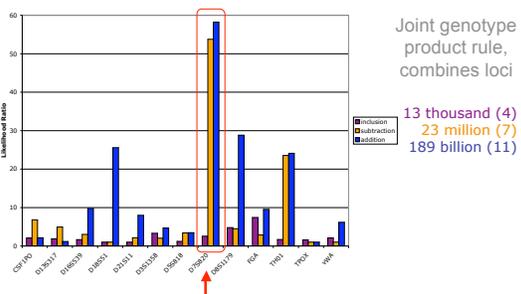Step 2: match genotype

lower probability loses LR information

DNA Match Comparison

Joint genotype product rule, combines loci

13 thousand (4)
23 million (7)
189 billion (11)

Computer Preserves Information

13.26
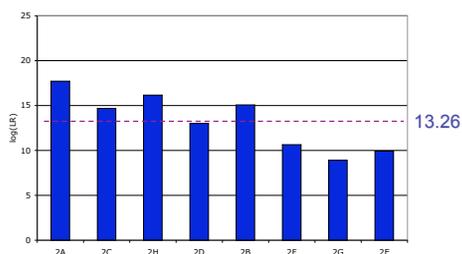
Human Review Loses Information

13.26
6.24
7.03