Using probabilistic genotyping to distinguish family members

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Morgantown, WV

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Cybergenetics

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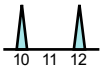
DNA evidence interpretation

Evidence item → Lab → Evidence data → Infer → Evidence genotype

10, 12

DNA from one person



Compare

Known genotype
10, 12

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DNA interpretation challenges

- Low-level DNA
- Degradation
- **Mixtures**

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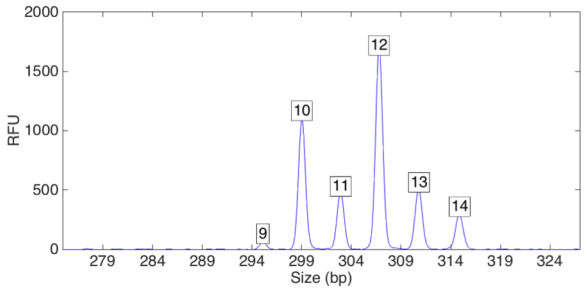
Mixtures

How many contributors?



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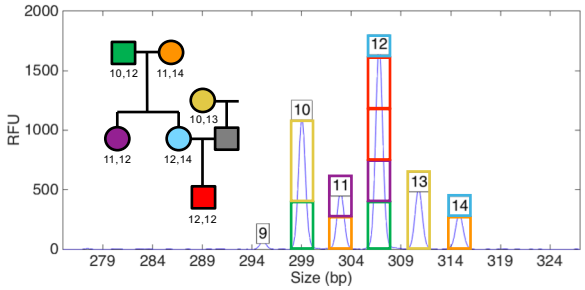
Relatives share alleles



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Science in the name of justice

- Use tools
- Gain knowledge
- Get answers
- Impact society

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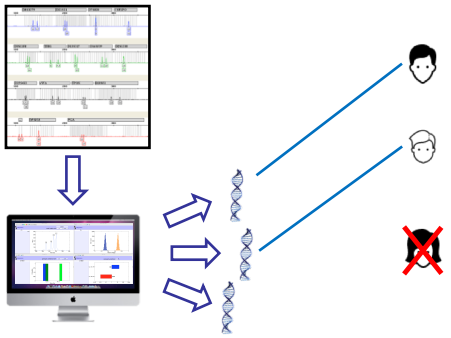
Computer DNA interpretation



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State of West Virginia v. Defendant

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Case context

- April 2016
- House party
- Drugs and alcohol
- Alleged assault

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Case context

- Taken to hospital
- Aware of assault
- Sexual assault kit collected

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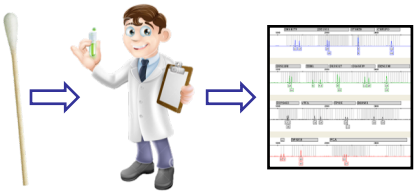
The evidence



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Crime lab findings

The results identified from the "anal swabs x2" (combined sperm and ecell fractions) are consistent with a mixture of DNA. The primary results identified from the ecell fraction are consistent with the DNA profile of [redacted]. The results identified from both amplifications of the sperm fraction are consistent with a mixture of DNA from three or more individuals. Due to the nature of the sample and the large number of possible contributors, no conclusions were made regarding the inclusion or exclusion of [redacted]

The results identified from the "vaginal swabs x2" (combined sperm and ecell fractions) are consistent with a mixture of DNA. The results identified from the ecell fraction are consistent with the DNA profile of [redacted]. The results identified from both amplifications of the sperm fraction are consistent with a mixture of DNA from three or more individuals. Due to the nature of the sample and the large number of possible contributors, no conclusions were made regarding the inclusion or exclusion of [redacted]

Probabilistic genotyping may prove beneficial on these samples. Currently the WV State Police Forensic Laboratory does not perform this type of analysis.

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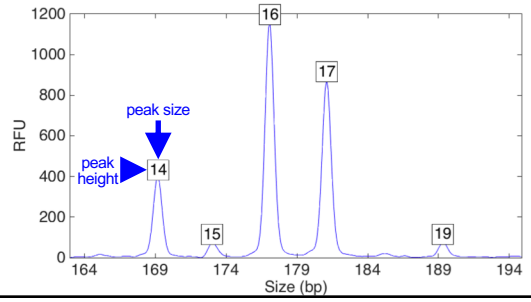
Computers can use all the data

Quantitative peak heights at locus vWA



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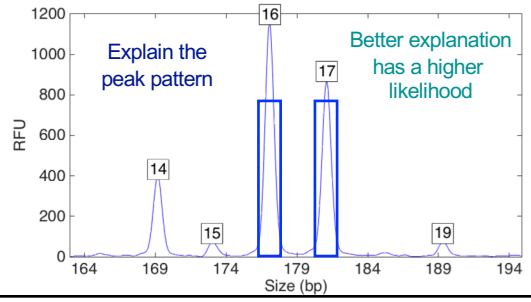
How the computer thinks

Consider every possible genotype solution



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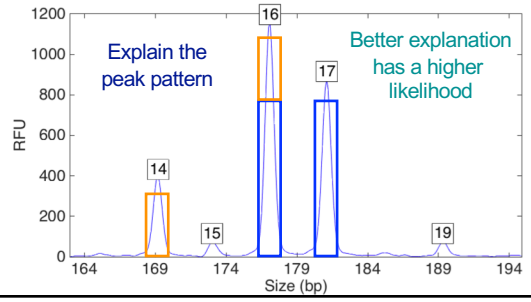
How the computer thinks

Consider every possible genotype solution



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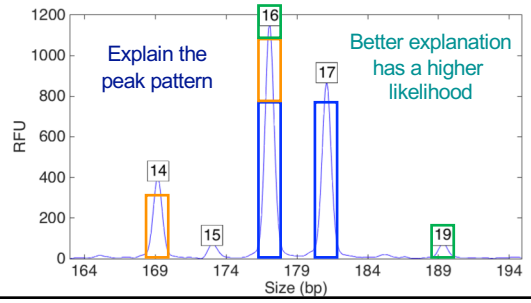
How the computer thinks

Consider every possible genotype solution



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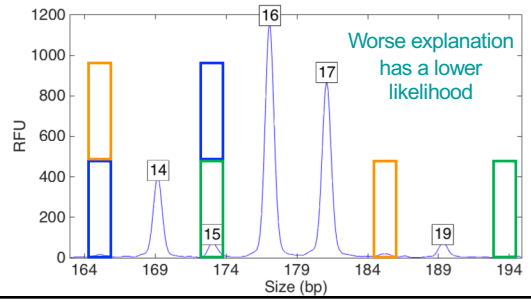
How the computer thinks

Consider every possible genotype solution



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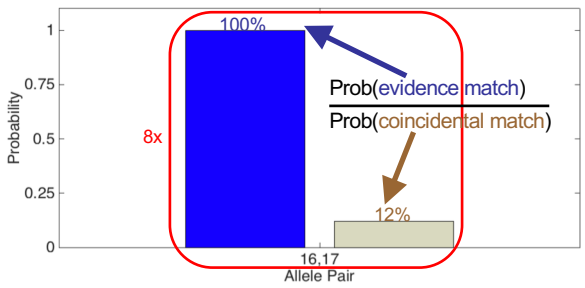
Evidence genotype

Objective genotype determined solely from the DNA data.

Never sees a comparison reference.



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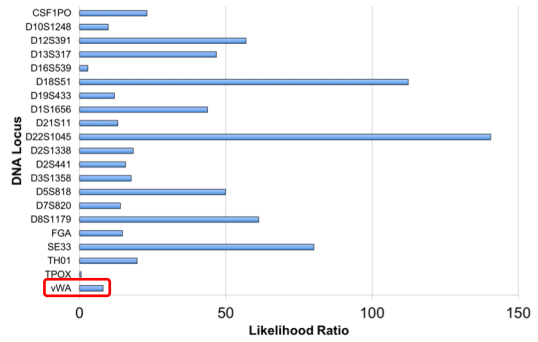
Match information at 21 loci



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Is the victim in the evidence?

A match between the vaginal swab and victim is:

55.2 nonillion times more probable than a coincidental match to an unrelated African-American person

4.07 octillion times more probable than a coincidental match to an unrelated Caucasian person

3.99 octillion times more probable than a coincidental match to an unrelated Southeast Hispanic person

11.5 octillion times more probable than a coincidental match to an unrelated Southwest Hispanic person

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Match statistics

Item	Victim	Suspect	Brother	Defendant	Suspect
Vaginal swabs	27.60				

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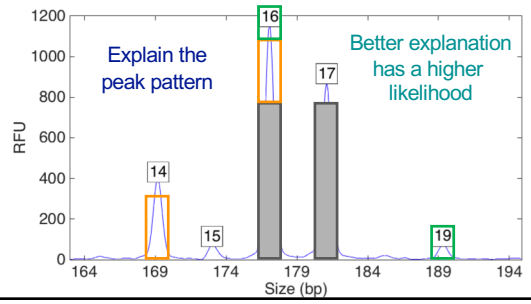
How the computer thinks

Peeling process assuming victim as known

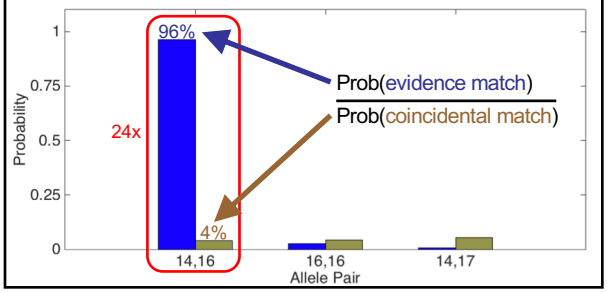


23

Evidence genotype

Objective genotype determined solely from the DNA data.

Never sees a comparison reference.



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Match statistics

Item	Victim	Suspect	Brother	Defendant	Suspect
Vaginal swabs	27.60	29.94			

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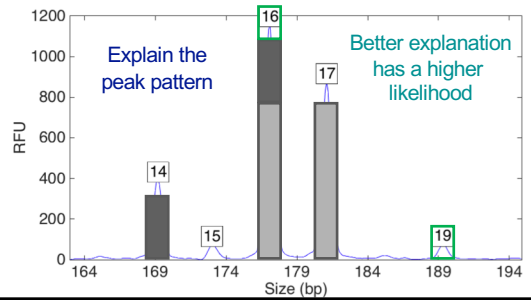
How the computer thinks

Peeling process assuming victim and suspect as known



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Match statistics

Item	Victim	Suspect	Brother	Defendant	Suspect
Vaginal swabs	27.60	29.94	11.84		

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Vaginal swabs vs. defendant

Number of contributors	Assumed references	Average match statistic
3	none	-9.94
3	victim	-11.40
3	victim, suspect	-12.74
3	victim, brother	-41.39
4	victim, suspect, brother	-3.44

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Anal swabs vs. defendant

Number of contributors	Assumed references	Average match statistic
3	none	-23.77
3	suspect	-28.13
3	suspect, victim	-33.56
3	suspect, brother	-39.67
4	suspect, victim, brother	-1.99

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TrueAllele Results

Vaginal swabs

Item 01.001, vaginal swabs

TrueAllele assumed that the evidence sample data (Item 01.001) contained three or four contributors, and objectively inferred evidence genotypes solely from these data. Reference genotypes were assumed as known in some calculations that involved comparisons to other reference genotypes. Single and joint data interpretation was performed. Following genotype inference, the computer then compared separated genotypes from this evidence item to provided reference genotypes (Items 01.011, 05.001, 06.001, and 08.001), relative to ethnic populations, to compute LR DNA match statistics. Based on these results:

A match between the vaginal swabs (Item 01.001) and brother (Item 06.001) is:
4.37 trillion times more probable than a coincidental match to an unrelated African-American person,
693 billion times more probable than a coincidental match to an unrelated Caucasian person,
3.6 trillion times more probable than a coincidental match to an unrelated Southeast Hispanic person, and
8.77 trillion times more probable than a coincidental match to an unrelated Southwest Hispanic person.

A match between the vaginal swabs (Item 01.001) and defendant (Item 05.001) is:
6.31 quadrillion times *less* probable than a coincidental match to an unrelated African-American person,
67.1 quadrillion times *less* probable than a coincidental match to an unrelated Caucasian person,
22.8 quadrillion times *less* probable than a coincidental match to an unrelated Southeast Hispanic person, and
21.4 quadrillion times *less* probable than a coincidental match to an unrelated Southwest person.

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TrueAllele Results

Anal swabs

Item 01.006, anal swabs

TrueAllele assumed that the evidence sample data (Item 01.006) contained three or four contributors, and objectively inferred evidence genotypes solely from these data. Reference genotypes were assumed as known in some calculations that involved comparisons to other reference genotypes. Single and joint data interpretation was performed. Following genotype inference, the computer then compared separated genotypes from this evidence item to provided reference genotypes (Items 01.011, 05.001, 06.001, and 08.001), relative to ethnic populations, to compute LR DNA match statistics. Based on these results:

A match between the anal swabs (Item 01.006) and brother (Item 06.001) is:
4.28 sextillion times more probable than a coincidental match to an unrelated African-American person,
37.5 quintillion times more probable than a coincidental match to an unrelated Caucasian person,
23.1 quintillion times more probable than a coincidental match to an unrelated Southeast Hispanic person, and
97.6 quintillion times more probable than a coincidental match to an unrelated Southwest Hispanic person.

A match between the anal swabs (Item 01.006) and defendant (Item 05.001) is:
26.5 septillion times *less* probable than a coincidental match to an unrelated African-American person,
497 septillion times *less* probable than a coincidental match to an unrelated Caucasian person,
267 septillion times *less* probable than a coincidental match to an unrelated Southeast Hispanic person, and
386 septillion times *less* probable than a coincidental match to an unrelated Southwest Hispanic person.

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DNA evidence in the courtroom

- Trial in April 2018
- Teaching about probabilistic genotyping
- Cross exam
- Outcome:

Not guilty of sexual assault

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Validation studies

36 studies, 7 peer-reviewed publications

Cybergenetics. Australia TrueAllele® Validation Report. 2011.

Caponera, J. New York State Police Crime Laboratory System TrueAllele® Casework Validation Addendum. 2013.

Guest, K., Ludvico, L., Ferrara, L., Perlin, M. Development of Kinship Mixtures and Subsequent Analysis Using TrueAllele® Casework. 2015.

Greenspoon, S.A., Schiermeier-Wood, L., and Jenkins, B.C. Establishing the limits of TrueAllele® Casework: a validation study. Journal of Forensic Sciences, 60(5):1263-1276, 2015.

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Thank you!

For more information

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