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TrueAllele Workshop: Unlocking DNA profile interpretation for Africa

6# African Forensic Forum
University of Cape Town
South Africa
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Cybergenetics, Pittsburgh, PA USA

Cybergenetics

JUSTICE THROUGH SCIENCE

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Kinship IPG

Paternity
Maternity
Missing Person
Mixture with Kinship

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Paternity

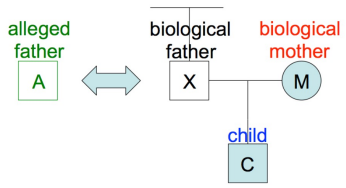

Figure 1. *Paternity example.* The 'X' indicates the inferred biological father genotype.

family	*relation*	*client*	*request*
familyname	child1	KIN	C
familyname	spouse	KIN	M

Table 1. *Example kinship table for a paternity case.*

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Maternity

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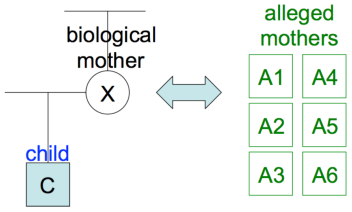

Figure 2. *Maternity example.* The 'X' indicates the inferred biological mother genotype.

family	*relation*	*client*	*request*
familyname	child1	KIN	C

Table 2. *Example kinship table for a maternity case.*

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Missing Person

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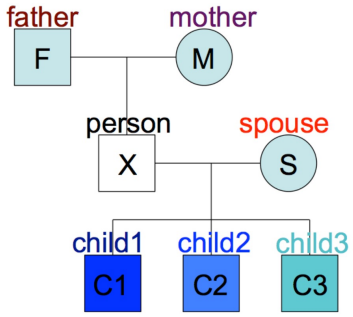

Figure 3. *Missing person example.* The 'X' indicates the inferred missing person genotype.

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Mixture with Kinship

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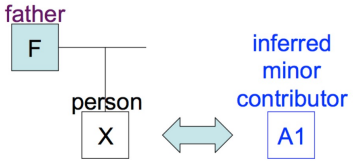

Figure 4. *Mixture and kinship example.* The 'X' indicates the inferred biological son genotype. A1 indicates the inferred minor contributor genotype from an evidence item.

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Kinship Cases

Four half-siblings (UCT)
New York daughter rape
Pennsylvania two-parent murder

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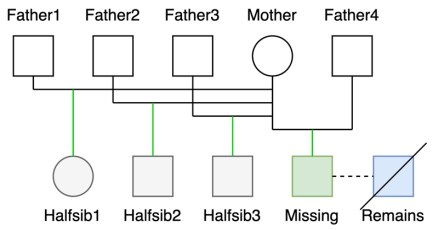
Four half-siblings (UCT)



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4 Half-sib remains identification

		SA mixed		SA total alleles		SA weighted sum	
genotype	KL	logLR	logER	logLR	logER	logLR	logER
kingeno4_child1	1.70	1.56	-2.42	1.52	-2.37	1.42	-2.25
kingeno4_child2	1.47	2.21	-3.36	2.18	-3.38	2.09	-3.22
kingeno4_child3	1.87	1.42	-2.28	1.43	-2.27	1.45	-2.29
kingeno4_child12	2.79	3.29	-4.45	3.28	-4.51	3.14	-4.32
kingeno4_child13	3.44	1.75	-2.71	1.80	-2.74	1.78	-2.72
kingeno4_child23	3.87	3.45	-4.43	3.54	-4.52	3.52	-4.51
kingeno4_child123	**5.71**	**3.60**	**-4.82**	**3.90**	**-5.02**	**3.72**	**-4.85**

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New York daughter rape

Item	Description	4.1 Daughter	5.1 Father
2.2	Stain 9	4.02	17.73
	Stain 21	16.87	14.22
	Stain 26	16.89	14.48
	Stain 30	9.32	17.74
	Stain 31	16.83	16.52

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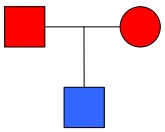
"No conclusions?"

Pennsylvania v James Yeckel



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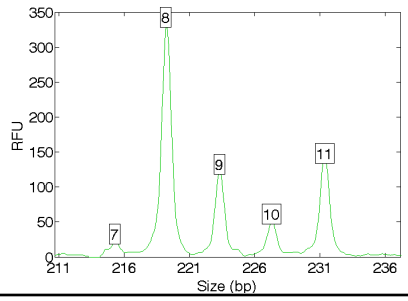
STR data

Quantitative peak heights at locus D7S820



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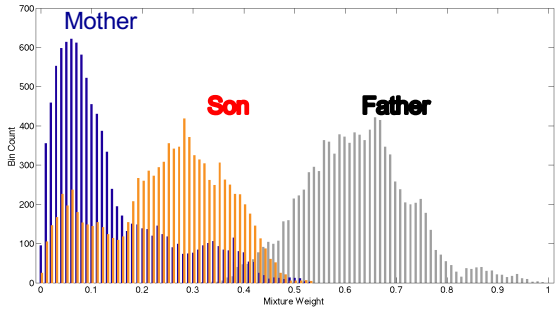
Mixture separation



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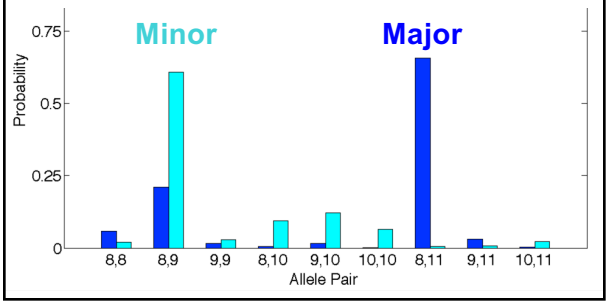
Genotype separation

Separate the mixture data into contributor genotypes



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Reported match statistic

The computer's LR information

A match between the shotgun shell
and James Yeckel is
6.13 trillion times more probable than
a coincidental match to an unrelated Caucasian person

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Why DNA investigation fails

Rape kits are processed, but:

Data isn't used
Mixtures aren't separated
Genotypes aren't uploaded
Database isn't searched

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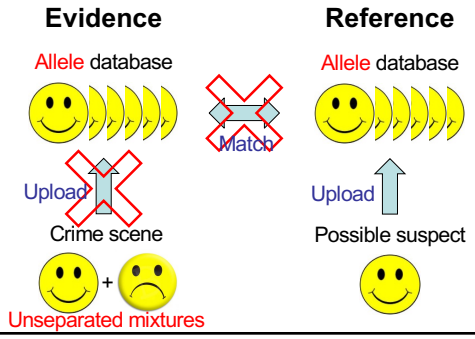
DDG CODIS database we have



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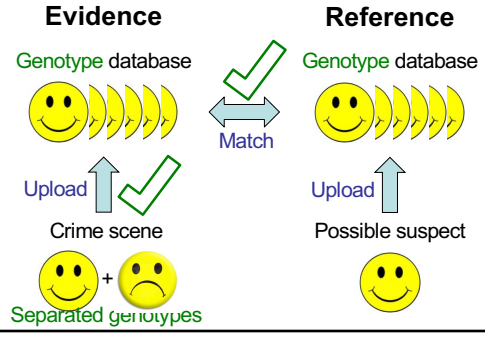
TrueAllele IPG database we need



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Identifying Victim Remains Using Kinship Genotype Inference

American Academy of Forensic Sciences
Criminalistics Section

February 2023
Orlando, FL

William Allan, MS
Mark Perlin, PhD, MD, PhD

Cybergenetics

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African Bus Crash

- 2008 bus crash in Komatipoort, South Africa
- Police recover burned victim remains
- Relatives submit DNA to help identify remains
- Lab couldn't identify remains. Sent DNA data to Cybergenetics.

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Victim Remains

Victim Remains
AO0553E
AO0554E
AO0555E
AO0556E
AO0557E
AO0558E
AO0559E
AO0560E
AO0561E
AO0562E
AO0563E
AO0564E
AO0565E
AO0566E
AO0567E

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Relatives of Missing People

Sample	Relation
AP2438C	Grandfather
AP2439C	Daughter
AP2440C	Son
AP2441C	Mother
AP2442C	Son
AP2443C	Sister
AP2444C	Father
AP2445C	Son
AP2446C	Unknown
AP2447C	Brother
AP2448C	Brother
AP2449C	Sister
AP2450C	Mother
AP2451C	Son
AP2452C	Father
AP2455C	Son

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Task: Find DNA Matches

Victim Remains
AO0553E
AO0554E
AO0555E
AO0556E
AO0557E
AO0558E
AO0559E
AO0560E
AO0561E
AO0562E
AO0563E
AO0564E
AO0565E
AO0566E
AO0567E

?

Sample	Relation
AP2438C	Grandfather
AP2439C	Daughter
AP2440C	Son
AP2441C	Mother
AP2442C	Son
AP2443C	Sister
AP2444C	Father
AP2445C	Son
AP2446C	Unknown
AP2447C	Brother
AP2448C	Brother
AP2449C	Sister
AP2450C	Mother
AP2451C	Son
AP2452C	Father
AP2455C	Son

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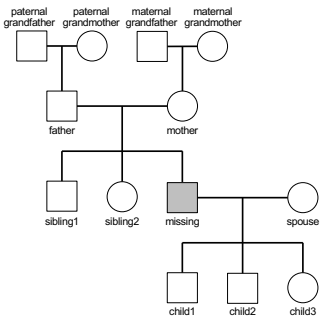
Kinship Relations



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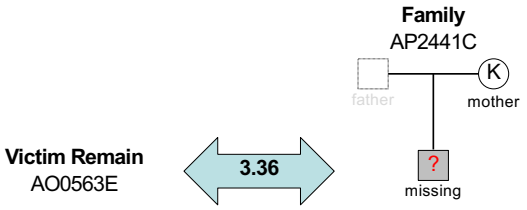
Matching Genotypes



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DNA Match Results

Relations of missing persons

Victim remains	AP2439C	AP2440C	AP2441C	AP2442C	AP2444C	AP2445C	AP2446C_3	AP2451C	AP2455C
AO0553E									
AO0554E							4.45		
AO0555E								1.88	
AO0556E				5.03					
AO0557E						3.57			
AO0558E									
AO0559E									2.95
AO0560E									
AO0561E					2.34				
AO0562E									
AO0563E			3.36						
AO0564E									
AO0565E									
AO0566E	4.03								
AO0567E		2.25							

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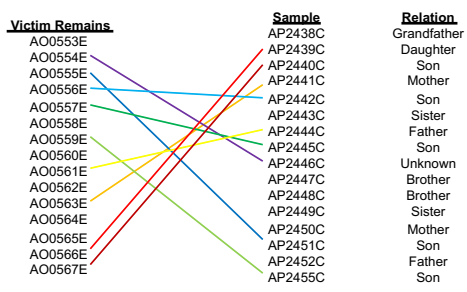
Matches TrueAllele Found



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Student Exercise

- 1 TrueAllele database server
- 12 interpretation processors
- 20 students (3rd training day)

Task: have every student use TrueAllele to identify the victim remains

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Student Outcome

15 victim items	15 victim genotypes
16 family items	16 family genotypes
16 family references	16 kinship genotypes

15 victim genotypes x 16 kinship genotypes = 240 genotype comparisons

240 comparisons x 20 students = 4,800 total comparisons

before lunch:	students upload genotypes
during lunch:	TrueAllele infers and matches
after lunch:	students review identification results

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Conclusions

TrueAllele identifies victim remains using kinship genotype inference

The TrueAllele approach is:
- Automated
- Informative
- Easy to use

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Database Cases

World Trade Center disaster
England v. Stuart Burton rape
Texas v. Lydell Grant exoneration

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World Trade Center attack

September 11, 2001 in New York City

18,500 remains

2,700 victims

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Identify victim remains

Victim Remains

Match

Missing People

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Southampton rapist

Stranger rape on New Year's Eve

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DNA rape kit report

A trace amount of semen detected on vaginal swabs was submitted for DNA profiling

The DNA profile contained limited information. A speculative search was done of the National DNA Database. The search found 13 profiles, 1 in the Hampshire area.

Due to the low level and incomplete nature of the DNA profile, it is not suitable for routine statistical evaluation.

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Is the suspect in the evidence?

A match between the vaginal swabs and Stuart Ashley Burton is 17 thousand times more probable than coincidence.

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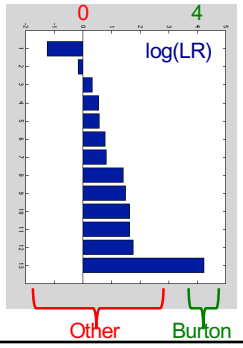
Separating database hits



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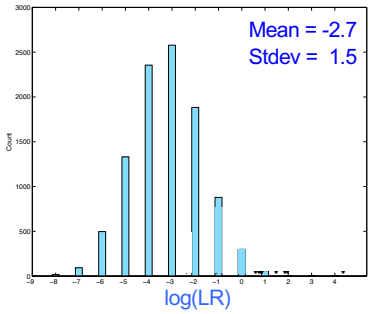
Non-contributor distribution



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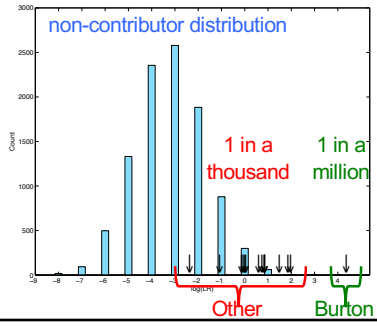
Separating hits statistically



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DNA leads to conviction

Mr. Burton pleaded guilty to the
rape committed on January 1, 2014.
He was sentenced to 12 years in prison.

Defendant also convicted of
having sex with 13 year old girl

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Texas v. Lydell Grant

2011, Houston lab fails to interpret DNA mixture
from fingernails of 2010 murder victim;
crime lab testifies DNA is inconclusive

Lydell Grant convicted of murder
and receives a life sentence

2019, Innocence Project of Texas
sends DNA data to Cybergenetics

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Match statistics

		12.1.1	13.1
Item	Description	Aaron Scheerhoorn	Lydell Grant
12.2.1.1	right hand fingernails	18.61	-12.93

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Non-matching evidence genotype

TrueAllele also inferred a **non-matching evidence genotype** from the right-hand fingernails. The probabilistic genotype of this unknown contributor has an expected LR match statistic of 18.2 trillion.

A **CODIS-searchable allele list** was derived from the probabilistic genotype at a 90% credible level.

Should additional reference genotypes become available, **Cybergenetics can compare** them with the probabilistic genotype to calculate DNA match statistics.

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Unprecedented CODIS search

- In 2019, TrueAllele crime lab searches CODIS
- Search finds the unknown fingernail person
- Confronted in Georgia, killer confesses to crime

Government was not pleased by our use of better science to reveal the truth in this case

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Grant released, exonerated

- Grant eventually released from prison
- In 2021, Lydell Grant finally exonerated

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TrueAllele reliability

Over 40 validation studies
- 8 peer-reviewed papers
- ≤10 contributors, low DNA

Over 50 admissibility rulings
- accepted in US, UK, etc.

Over 500 client agencies
- including FBI

Sensitive, specific, accurate
- informative, easy to use

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Peer-reviewed validation studies

Perlin MW, Sinelnikov A. An information gap in DNA evidence interpretation. *PLoS ONE*. 2009;4(12):e8327.

Ballantyne J, Hanson EK, Perlin MW. DNA mixture genotyping by probabilistic computer interpretation of binomially-sampled laser captured cell populations: Combining quantitative data for greater identification information. *Science & Justice*. 2013;53(2):103-114.

Perlin MW, Hornyak J, Sugimoto G, Miller K. TrueAllele® genotype identification on DNA mixtures containing up to five unknown contributors. *Journal of Forensic Sciences*. 2015;60(4):857-868.

Greenspoon SA, Schiermeier-Wood L, Jenkins BC. Establishing the limits of TrueAllele® Casework: a validation study. *Journal of Forensic Sciences*. 2015;60(5):1263-1276.

Bauer DW, Butt N, Hornyak JM, Perlin MW. Validating TrueAllele® interpretation of DNA mixtures containing up to ten unknown contributors. *Journal of Forensic Sciences*. 2020; 65(2):380-398.

Perlin MW, Legler MM, Spencer CE, Smith JL, Allan WP, Belrose JL, Duceman BW. Validating TrueAllele® DNA mixture interpretation. *Journal of Forensic Sciences*. 2011;56(6):1430-1447.

Perlin MW, Belrose JL, Duceman BW. New York State TrueAllele® Casework validation study. *Journal of Forensic Sciences*. 2013;58(6):1458-1466.

Perlin MW, Dormer K, Hornyak J, Schiermeier-Wood L, Greenspoon S. TrueAllele® Casework on Virginia DNA mixture evidence: computer and manual interpretation in 72 reported criminal cases. *PLOS ONE*. 2014;(9)3:e92837.

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TrueAllele education

TrueAllele training courses
- introduction to methods
- advanced problem solving
- self-paced workbooks

Free for Africa
- UCT – 2023
- Zambia – 2024

6#AFF attendees
- follow-up course

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TrueAllele for Africa

DNA information solves crime
- prevents crime, promotes justice

IPG TrueAllele database
- connects crimes and criminals
- DDG CODIS is less effective

Computer automation
- accurate genotyping and match
- scales up for Africa at low cost

Easy to understand and use

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More information

http://www.cybgen.com/information

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- Reliability
- Webinars

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