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DNA 101 & Evaluating Post-Conviction DNA Cases

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In partnership with: PC-DNA TTA Program - Quattrone Center for the Fair Administration of Justice

> Hosted by: Forensic TTA Program - RTI International

FORENSICS TTA PROGRAM





Forensics TTA

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Quattrone Center

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DNA 101 & EVALUATING POST-CONVICTION DNA CASES

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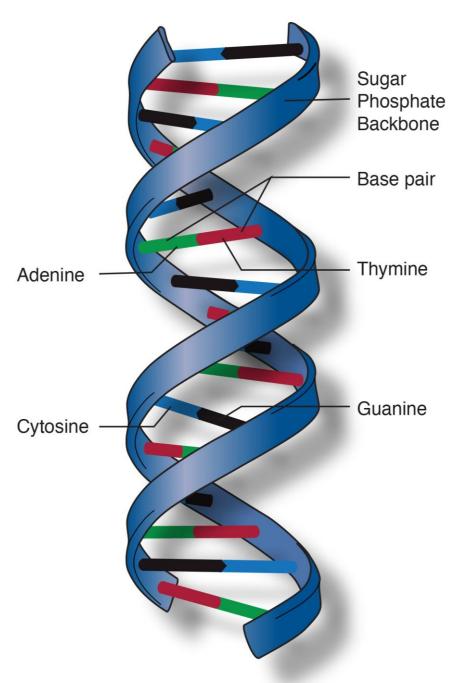


• DNA 101 for criminal justice practitioners

• Reviewing prior testing

• Determine whether new DNA testing will identify the perpetrator

DNA 101 FOR CRIMINAL JUSTICE PRACTITIONERS



https://storymd.com/journal/j69d6q30zj-genetics-glossary-letters-d-ef/page/a8lz3h3y2br-what-is-double-helix

DNA IN CRIMINAL CASES d

- Test on specific areas of the human DNA strand that are more unique to an individual
- Alleles aka your DNA signature: a specific number that symbolizes the genetic makeup at a specific location in your DNA

COMMON

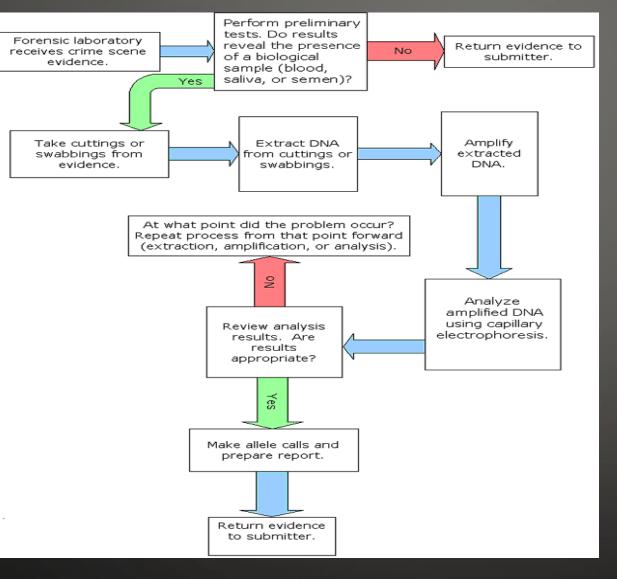
- Blood
- Semen
- Hair*
- Saliva
- Bone
- Skin Cells

*The tests vary depending on what part of the hair is available

EVIDENCE THAT SOURCES OF DNA MAY CONTAIN DNA

MASKS! Sexual Assault Kits Fingernails and Scrapings Weapons: Knives, bats, hammers, guns, blunt objects, etc. Contact Items: Clothing, ropes, telephone cords, key chains, steering wheels, eyeglasses, tape, bed clothes, etc. Saliva: Bottles, cigarette butts, toothbrushes, gum, stamps, envelopes, facial tissue, etc. Jewelry: Rings, bracelets, necklaces, earrings and other piercings, etc. Hair: Brushes, hair ties, combs, hats, bandanas, etc. **Evidence Envelopes that contained items** before

PROCESS



https://oig.justice.gov/sites/default/files/archive/special/0405/chapter2.htm

1. Item goes to the crime lab

2. Prelim Tests/Serology: What type of biological material? (Saliva, osemen/sperm, blood, etc.)

3. Item is swabbed (if not already a swab), scraped or vacuumed to pull the DNA off.

4. Extraction: Turns the item into liquid form and removes potential DNA

5. Quantification: How much DNA is in the sample? How much male?

6. Testing \rightarrow PCR/STR, Y-STR, Mito,

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NGS (next-generation sequencing)

7. Interpret Mixtures with PGS

National Forensic Science Technology Center: DNA Analysis https://www.youtube.com/watch?v=JUoBTk1NDZ8

3 COMMON TYPES OF DNA TESTING

- 1. PCR/STR:
 - MI state lab is currently testing on 23 locations (Loci) and 1 sex chromosome (X,Y)
 - Can be interpreted in STRMix/TrueAllele and uploaded into CODIS
 - Different kits are available at different labs some test up to 27

2. Y-STR:

- Tests on the Y (Male) chromosome that is passed down in a direct line from grandfather to father to son, etc.
- Bio father and son will have the same profile
- Two brothers (same father) will have the same profile

3. Mitochondrial:

- Tests for DNA that is passed down in a direct line from mother to children
- Test used on hair shafts (*Note: STR/Y-STR early testing on shafts now*)

PROBABILISTIC GENOTYPING SOFTWARE

STRMix and TrueAllele are two different software programs a.k.a. competitors

• Interprets complex DNA mixtures using statistics

- It is NOT a test itself; but rather a program used to interpret the results of STR/PCR testing
 - The analysts input various information from the test results the results are NOT automatically inputted from the DNA test results

PCR/STR EXAMPLE

CLocus/Loci (pl) "Location" of the gene on a chromosome

> Different locations in the DNA strand (23 and Amelogenin)

> > Note: Amelogenin XX = Female XY = Male

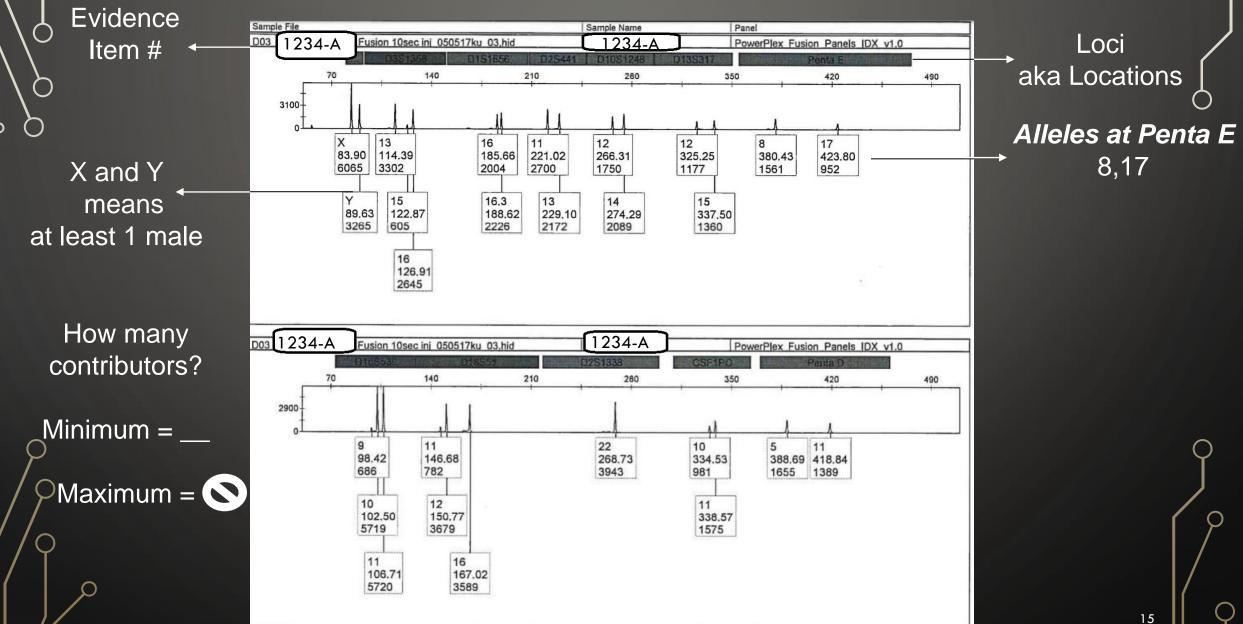
Locus	Known
D3S1358	15, 15
D1S1656	13, 16.3
D2S441	11, 14
D10S1248	13, 14
D13S317	12, 12
Penta E	11, 18
D16S539	10, 12
D18S51	16, 16
D2S1338	19, 23
CSF1PO	11, 11
Penta D	9, 11
TH01	9, 9.3
vWA	15, 16
D21S11	30, 30.2
D7S820	8, 8
D5S818	12, 12
ТРОХ	9, 11
DYS391	10, 10
D8S1179	14, 15
D12S391	16, 17
D19S433	12.2, 14
FGA	22, 22
D22S1045	11, 15

Homozygous Same allele from mom and dad

Heterozygous Different allele from mom and dad

Half and Half
 9 from mom; 11 from dad
 or
 11 from dad; 9 from mom

ELECTROPHEROGRAMS



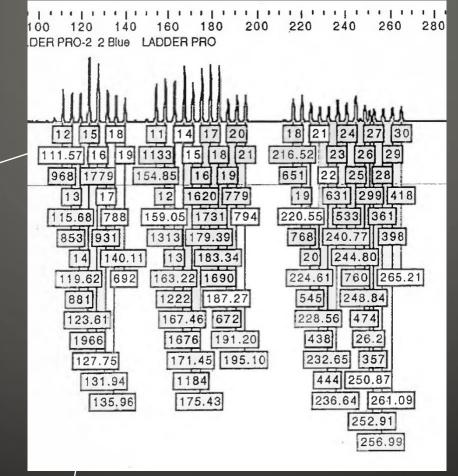
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LADDERS

The most common alleles found at a given location

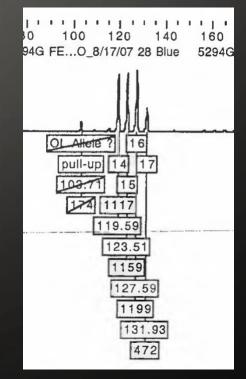
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Ex: At that first location you could have the following alleles: 12,13,14,15, 16,17,18,19



Off Ladder Allele: An allele that is not in those numbers accepted on the ladder.

Could either be an artifact or a contributor with a rare allele



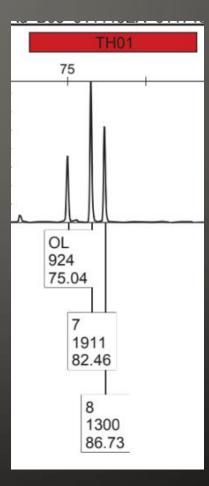
OFF LADDER: ANOTHER EXAMPLE

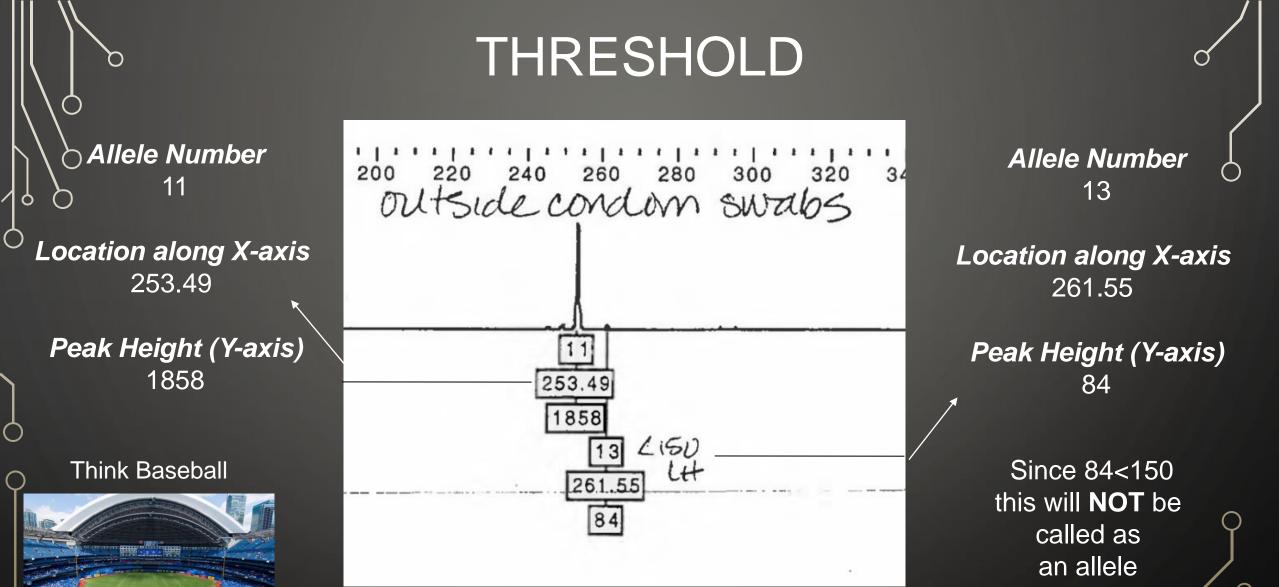
7, 8 = Accepted alleles for THO1 (based on ladder)

The first peak marked "OL" for "Off Ladder" as it was not a common allele at TH01

Notice how tall the peak is = 924

Either an artifact (not an allele) OR an allele from a contributor





Threshold at Labs may vary **MSP's threshold was 150 when this was conducted** (250 at one point)

https://i.pinimg.com/736x/71/97/53/7

197537cc3a88e1d95bb7005b86ee1

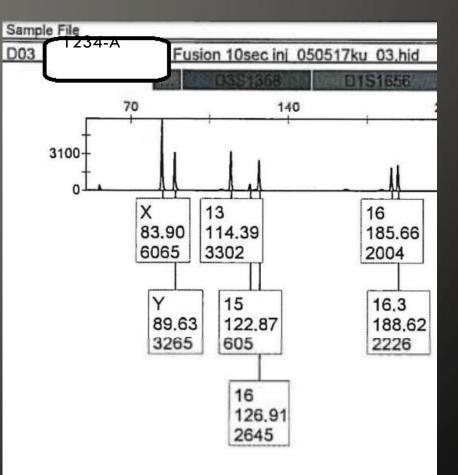
be.jpg

ARTIFACTS

A "glitch" in the process ... not an allele:

- Most Common = Stutter:
 - A "fake" allele that is either before or after an allele
- Pull-up
- Dye Blobs

If you see an allele marked artifact, stutter, pull-up, or a blob, ASK QUESTIONS!



Specific number assigned to case by the lab

Evidence is assigned a specific set of numbers/letters (usually the first few numbers are the same)

> Type of DNA test conducted (Note: STRMix was used to interpret)

	1234	EPORT
Laboratory No. Investigating Ofcr. Agency Agency No.		Record No. Date Received Time Received Date Completed
Nature of Offense:		
1100-1 - Sexual	Assault CSC 1st	
Victim(s):		
Victim		
Suspect(s):		
John Doe	\supset	
Evidence:		
1234-A 1234-B	Vaginal swabs enve Known buccal from	
Poculte of Examina	ation:	

Results of Examination:

Deoxyribonucleic acid (DNA) recovered from the above submitted samples was processed using the polymerase chain reaction (PCR) and the PowerPlex® Fusion System. These profiles were evaluated using STRMix[™], a probabilistic genotyping software application.

Conclusions:

234-B 1. A DNA profile was obtained from item

1234-A A partial DNA profile of two donors was obtained from item (Vaginal swabs envelope) cuttings - Fraction 1). Due to the limited data obtained, no conclusions can be made.

Report number (starting at

Make sure you have them all

Fraction 1 v. Fraction 2 Epithelial v. Sperm* F/M Mix v. Male (attempt to separate the sperm from the sample)

> *Sperm Fraction DOES NOT MEAN SPERM/MALÉ DNA WAS THERE

STRMix Probabilistic Genotyping Software Not a DNA test! Program that assists with interpretation and statistical weighting of DNA results 3. Interpretation of item 1234-A (Vaginal swabs envelope cuttings - Fraction 2) was performed assuming that the DNA profile originated from one individual.

Hypotheses:	H ₁ : 1234-B (Known buccal from John Doe)
	H ₂ : An unrelated, unknown contributor
Conclusion:	Based on the DNA typing results obtained from the vaginal swabs envelope
	cuttings - fraction 2, it is at least 1.1 million times more likely if it originated from
	John Doe than if it originated from an unrelated, unknown contributor.
Verbal Scale:	This analysis provides very strong support that is a contributor to
	the DNA profile developed from the vaginal swabs envelope cuttings - fraction 2.

Remarks:

Other members of the Forensic Science Division may have processed evidence associated with this report, in addition to the reporting analyst.

The propositions were formed from the information available to the undersigned at the time of analysis. If this information changes or if other propositions should be considered, the analyst is able to undertake them if instructed with sufficient time.

The magnitude of the likelihood ratio relates to the degree of support provided by the evidence under the tested hypotheses and assumptions. The qualitative statement for a comparison of the evidentiary typing results to a person of interest is based on the following table:

Likelihood Ratio	Qualitative Equivalent	
0	Exclusion	
>0 to 99	Uninformative	
100 to 999	Moderate Support	
1,000 to 9,999	Strong Support	
10,000 and greater	Very Strong Support	

Disposition of Evidence:

The Biology Unit is no longer retaining known or evidentiary samples in long term storage. All items of evidence will be returned to the submitting agency. Samples, or portions thereof, that have been retained for DNA have been consumed in analysis. All DNA evidence and extracts will be returned to the submitting agency.

Where the evidentiary samples were sent post-testing

COMMON QUESTIONS

What is masking?

Imagine a jar full of 100 black jellybeans (victim) and 1 pink jellybean (John Doe). If you reach inside the jar, it would be very difficult for you to pick out the 1 pink jellybean over the 100 black jellybeans with your eyes closed. Same goes for DNA.

- Common masking: Intimate samples (vaginal/anal swabs) or victim's blood mixed on a knife the perpetrator touched
 - In both of these examples, the victim's DNA will mask the perpetrator's
 - This is where creative/complex DNA testing plans are imperative

Can a partial profile be used for comparison purposes? Your report reads: "A partial DNA profile was obtained from item 1234-B (John Doe known) that is sufficient for comparison purposes." This means that there is not a full 23-loci profile for John Doe. A partial profile can be used for comparison purposes. Still scrutinize the results.

SCIENTIFIC DEFINITIONS AND SOURCES

Allelic Drop-out

Failure to detect an allele within a sample, or allele doesn't amplify during PCR.

Amelogenin

- The sex determination locus.

Analytical threshold

Minimum height requirement to call an allele and not noise. Generally either a true allele or artifact.

Artifact

Any non-allelic products of the amplification process (ex. Stutter), anomalies of the detection process (ex. pull-up), or by-products of primer synthesis (ex. Dye Blob).

Sources:

National Institute of Justice, DNA for the Defense Bar (June 2012); SWGDAM Interpretation Guidelines for Autosomal STR Typing (2010).

SCIENTIFIC DEFINITIONS AND SOURCES

The graphic representation of the separation of molecules by electrophoresis or other means of separation.

Locus (Loci)

The specific physical location(s) of gene(s) on a chromosome.

Masked allele

- An allele of a minor contributor that may not be readily distinguishable from the alleles of the major contributor or an artifact.

Touch DNA

DNA that is left behind, typically from skin (epithelial) cells, when a person touches or otherwise comes into contact with an item or person.

Stutter

Well researched phenomenon which causes a byproduct during the PCR process. Stutter is typically 1 repeat unit smaller, and reproducibly shorter than the true DNA allele.

Sources:

National Institute of Justice, DNA for the Defense Bar (June 2012); SWGDAM Interpretation Guidelines for Autosomal STR Typing (2010).

REVIEWING PRIOR DNA TESTING

REQUEST THE DATA

1. For all DNA testing that was conducted in relation to this matter, please provide all documents and supporting evidence used in the formulation of any expert opinion, *including, but not limited to the following:*

i. All electropherograms;ii. All laboratory submission reports;iii. All bench notes;iv. All graphs and tray information;

v. All laboratory log-in records;
vi. All communications between the laboratory and third-parties;
vii. All photographic or written inventories; and
viii. All other supporting or underlying evidence or data.

2. Legible copies of all documents used by the People's experts to render their opinions regarding the biological evidence in this matter, including the evidence collection (crime scene notes), serology, and DNA analysis.

3. All digital media (photographs, video, audio) and raw data **(.fsa or .hid files)** created as a part of the evidence collection, serological analysis, and DNA analysis of evidentiary items in this matter.

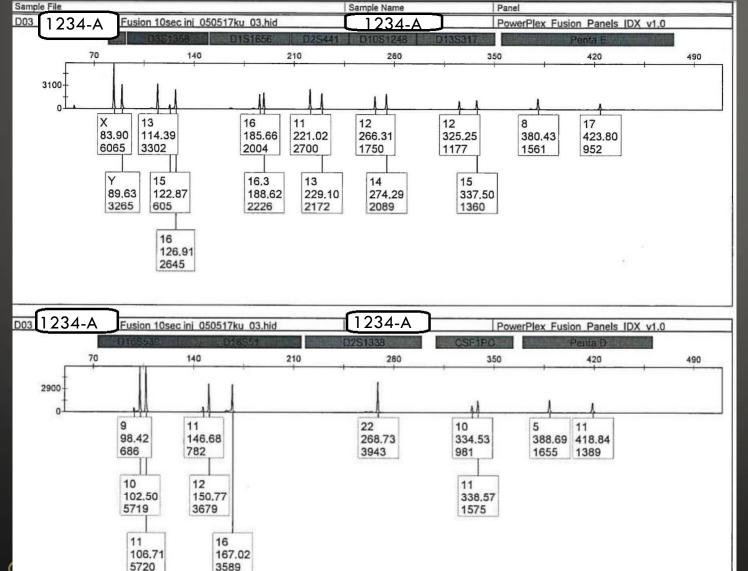
CREATE AN EVIDENCE CHART

 Tracks ALL evidence collected in the case: physical items, fingerprint lifts, ballistics, hair, cars, cell phones, etc.

- Include ALL evidence not just "material to perpetrator" you don't know what may be important
- Not all evidence will get interacted with after it is first collected
- Steps in assigning tag numbers to crime scene evidence:
 - Collected by Law Enforcement who assigns the evidence a tag number
 - Lab receives evidence and assigns their own tag numbers
 - Lab may also assign new numbers for different fractions, extractions, swabs, etc.
 - New post-conviction lab does re-testing and assigns their own tag numbers

				/
ITEM	PD TAG NUMBER	ORIGINAL TESTING (1998)	LAB 1 (2018)	LAB 2 (2022)
Vag swab (unsure how many swabs)	92241	5A - no analysis	Negative for semen and processed STR: Partial profile. Female (no Y) consistent with V YSTR = no results Detected some male	E02 = 1 vaginal swab E02A1 = swab consumed Male DNA not detected at quant NO YSTR PROFILE FROM EITHER EF OR SF There were peaks below threshold. Nothing above threshold
Postal sweb		6A no analysis	6A: Negative for semen. No male found. STR: Partial female – consistent with V, plus one additional DNA type foreign to V (allele or artifact)	E01a : swab and extracts consumed - nothing left.
Rectal swab (3 swabs)	92242	6B no analysis		E01b: quant - zero for male. YSTR - no results. No data at all.
		6C no analysis		E01c: quant - zero for male. YSTR - no results. Some below threshold - none above
D Known		12	STR and YSTR	Full profile: YSTR and STR
Victim known (from ME)		13	Partial victim profile (not suff for comparison)	Full profile: STR
Knife - Just the blade (no handle found)	92278	processed for prints		E04 = Blade E04A1 = swabbed knife blade No male DNA detected at quant Hold off for now: Thoughts of doing STR on this given that female may be involved (suspect)
	92235			
Oral Swab	92244			
Leather shoes (D)	92490	no ignitable liquids		
Two pair of blue jeans and blue shirt (D)	92491	no ignitable liquids		
Pair of blue jeans and gray tshirt (D)		no ignitable liquids		
Hair sample (found near V)	92254			
Distorted plastic piece	92253			
Charred clothing pieces	92222			
				26

CREATE A PROFILE (ALLELIC) CHART



	Vaginal Swab
Locus	1234-A
AMELOGENIN	X,Y
D3S1358	13,15,16
D1S1656	16,16.3
D2S441	11,13
D10S1248	12,14
D13S317	12,15
PENTA E	8,17
D16S539	9,10,11
D18S51	11,12,16 📍

Do this on your own so you know how strong the results are

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It can get complex...

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SO	organization	is
	key!	

	Panties	Breast	Fingernail	Thighs	Thighs	Thighs	Labia (1E)
AMELOGENIN	XY	X, Y	X,Y	X,Y	Х, Ү	X, Y	X, Y O
		14, 15,	9, 13, 14,	13,14,15,1			14, 15, 16,
D3S1358	14,16,17	16, 17	16, 17	6,17	14, 15, 16	15, 16	17
		13, 15,					
		15.3, 16,					\bigcirc
	15,16.3,1	16.3, 17,	15, 16, 16.3,	14,15,15.3,			16.3, 17,
D1S1656	7	17.3	17	16,16.3,17	16.3, 17.3	16.3, 17.3	17.3
		10, 11,	10, 12, 13,	10,11,11.3,	10, 11,	10, 11,	
D2S441	10,12,14	11.3, 14	14	12,13,14	11.3	11.3	11, 11.3, 14
		13, 14,					
D10S1248	13,14	16	13, 14, 16	13,14,15	13, 16	13, 16	13, 14, 16
D13S317	11,12,13	11, 12	11, 12, 13	11,12	12	12	11, 12
PENTA E	5,7,10	5, 8, 18	5, 7, 10, 12	5,7	12, 19	12, 19	5, 12, 19
		9, 10,					
		11, 12,	10, 11, 13,	9,10,11,12,	10, 11, 12,		10, 11, 12,
D16S539	11,13,14	13	14	13,14	13, 14	11,13, 14	13, 14
		11, 12,					
		13, 14,					0
		15, 16,					
		17, 18,	14, 15, 16,				15, 16, 17,
D18S51	15,16,17	19	17	7,18,19	16, 17, 18	17,18	18, 9
		17, 20,					
	18,21,22,		18, 21, 22,	19,20,21,2			20, 22, 23,
D2S1338	24	24	23, 24	2,23,24,27	23, 24	20,24	24 (
							30

ELIMINATE YOUR KNOWNS

	Vaginal Swab	DOE, J.	VICTIM, V.
Locus	1234-A	1234-B	1234-C
AMELOGENIN	X,Y	X,Y	X,X
D3S1358	13,15,16	16, 17	13
D1S1656	16,16.3	13, 16.3	16
D2S441	11,13	10, 14	11,13
D10S1248	12,14	14, 16	12,14
D13S317	12,15	12	12,15
PENTA E	8,17	9, 10	8,17
D16S539	9,10,11	9, 13	9,10
D18S51	11,12,16	16, 19	11,12
D2S1338	22	19, 27	22
CSF1P0	10,11	10	10,11
PENTA D	5,11	5, 8	5,11

THOUGHTS? CSC of female by male perp

1. XY = Male contributor

- 2. 3 alleles at 3 locations = more than 1 contributor
- 3. Eliminate Victim from sample and see what is left over

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ELIMINATE YOUR KNOWNS

	Vaginal Swab	DOE, J.	VICTIM, V.	
Locus	1234-A	1234-B	1234-C	
AMELOGENI				
Ν	XY	X,Y	X,X	
D3S1358		16, 17	13	
D1S1656	13	13, 16.3	16	
D2S441		10, 14	11,13	
D10S1248	() , ()	14, 16	12,14	
D13S317		12	12,15	
PENTA E		9, 10	8,17	
D16S539	Q , Q (11)	9, 13	9,10	
D18S51		16, 19	11,12	
D2S1338	<u>S</u>	19, 27	22	
CSF1P0	N	10	10,11	
PENTA D		5, 8	5,11	

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THOUGHTS? XY = at least 1 Male

- 3 alleles at 3 locations = more than 1 contributor
- Eliminate Victim from sample and see what is left over =
- Eliminate the defendant.
 Anything left over? =

What are these extra alleles?

INTERPRET RESULTS

	Vaginal Swab	DOE, J.	VICTIM, V.
Locus	1234-A	1234-B	1234-C
D3S1358		16, 17	13
D1S1656		13, 16.3	16
D16S539		9, 13	9,10
D18S51		16, 19	11,12

Results:

- Victim is major profile = makes sense as it is her intimate sample
- There are 3 alleles at 4 locations = more than 1 contributor
- There was an X,Y and victim is female = at least 1 male contributor
- 4 extra alleles found at 4 locations that are not consistent with the victim:
 - Out of the 4 extra alleles = John Doe only matches 2
 - Who do the other 2 (... or 4...) belong to?

INTERPRET RESULTS

	Vaginal Swab	DOE, J.	VICTIM, V.
Locus	1234-A	1234-B	1234-C
D3S1358		16, 17	13
D1S1656		13, 16.3	16
D16S539		9, 13	9,10
D18S51		16, 19	11,12

Results:

- FACTS MATTER!
 - Where did the sample come from, who had access/could have touched it/left DNA behind, when was it collected, innocent explanation?
 - For sex crimes: victim have consensual sex? How many alleged perps?
 - Ex: If Victim was assaulted by 1 perp and didn't have consensual sex with anyone within 96 hours:
 - Victim + one perp = Makes sense with data and not John Doe
 - Victim + two perps = Doesn't make sense with data (reminder we only saw a max of 3 alleles at any location) or case facts (see above). So if it all doesn't match John Doe then consult an expert for insight.

COMPARE RESULTS

• Using the lab reports, compare your findings to the Lab's conclusions:

- We found that two extra alleles were found that were NOT consistent with John Doe or the Victim
- "This analysis provides very strong support that John Doe is a contributor to the DNA profile from the vaginal swabs ..."
- Scrutinize their findings and what they aren't including
- Scrutinize how many contributors they are claiming and where those assumptions came from (Check bench notes and correspondence between the lab and police departments)
 - Cognitive Bias

CONSULT AN EXPERT!!!

EXPERTS

Consult a DNA expert. *Make sure you have all supportive data including raw⁽⁾ electronic data*

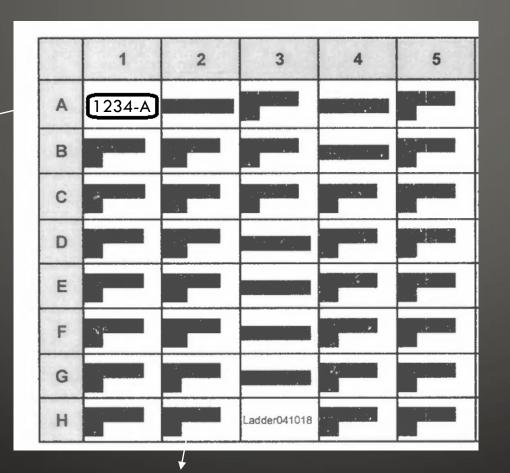
• Is there more testing you can do? Private v. public lab

• For PGS interpretation: can you contact a competitor to see if they will run the same DNA data and obtain different result?

Quattrone Center for the Fair Administration of Justice: If you have a Postconviction Testing of DNA Evidence Grant, they will find an expert and pay for the expert to work with you.

CHECK THE TRAYS

Find all the samples tested in your case using the assigned evidence number



Trays are usually filled with samples from multiple cases ... not just yours!

- Each sample is placed in its own tube in a tray
- Starting at A1, the scientist puts the samples into each tube. Once filled, the tube is closed (to avoid contamination).
- Check to make sure known samples aren't tested directly before the evidence in the case
 - Small particles from your client's known sample can contaminate the evidence

DETERMINE WHETHER NEW DNA TESTING WILL IDENTIFY THE PERPETRATOR

► EVALUATE THE CASE FROM COLLECTION ~ TO REPORTING

- Collection: contamination, fingerprint an item and now we want to DNA test
- Other items/swabs not tested resource issues for state labs
- Example: SAK and multiple swabs of same item
 - Example: murder weapon only testing bloody part
- Extracting the DNA: your results are only as good as your extraction
- Stopping at Quantification:
 - Presumptive test = not 100% accurate
 - Found human DNA but no male = I've had cases where Y is 0, but then obtained YSTR partial profile
- The DNA test: use a different test, Kit used (more locations, more sensitive)
- Reports/Results: mixture interpretation, underlining data that an expert can opine to, PGS, were all suspects/eliminators tested and compared
 - Example: Victim not compared in a CSC mixture interpretation issue

CODIS

- Combined DNA Index System DNA database
 - Different levels have different requirements
- STR profiles stored STR results ONLY
 - Pre-2017 = 13 Loci
 - Post-2017 = 20 Loci
 - Minimum of 8 Loci
- Several evidence requirements and lab requirements
 - Even if suspects in CODIS, doesn't mean tested with updated Kit (more loci)
- 1. Unknown DNA profile obtained from evidence sample
- 2. Analyst inputs profile into CODIS
- 3. Evidence Profile matches offender profile in CODIS aka CODIS Hit
- 4. Lab Confirm the CODIS Hit by obtaining sample of matched offender



Contact Information



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Contact Information





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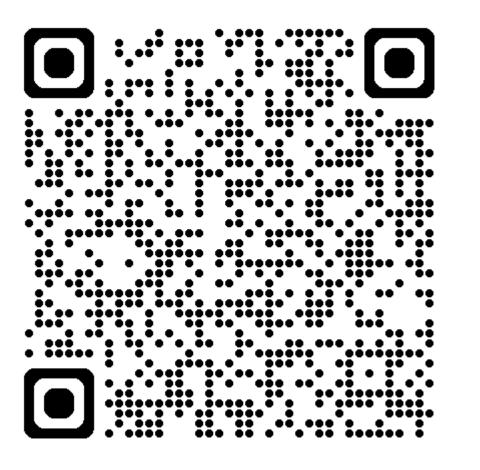
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