

# Defense Forensic Science Center

## DNA Mixture Interpretation Study: *Inter- and Intra-laboratory Variation*



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# Briefing Overview

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- DFSC mixture study structure
  - Goals and composition
- Review **Preliminary** Results
  - Visualizing variation: In-house Metrics
  - Trends
- Next Steps
  - Participant Reports



# DFSC Mixture Study



- Purpose:
  - To assess the inter- and intra-laboratory variation in DNA examiners' generated genotype interpretations
  - To better understand the current state and potential **limitations** of mixture interpretation in the forensic community
- Participation
  - Initiated Summer 2014
  - n=185 returned datasets



# Study Datasets:

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- Examiners asked to deconvolute 6 identical mixtures:
  - Use their laboratory's SOP
  - Stochastic and analytical thresholds set by DFSC
  - Genotype interpretations recorded on Excel-based worksheet provided (**DEAT: DNA Examiner Assessment Tool**)
  - User assessment form collected from each participant



# What is the Ideal Deconvolution?

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- Correctly determine the number of contributors (NOC) in the sample
- Generate the correct genotypes for each contributor in the sample
- Analyze via metrics:
  - NOC matching
  - Genotype Interpretation Metric (GIM)
  - Allelic Match scoring (AM)



# Determining the NOC



Mixture	Contributor Ratio	Known NOC	Derived NOC (%)				
			2*	At least 2	3*	At least 3	No Answer
1	3.5:1	2	63%	35%	0%	0.5%	1%
2	2:1 W/ Ref	2	64%	29%	1%	2%	5%
3	2:1	2	37%	48%	2%	5%	8%
4	3:1	2	54%	37%	1%	5%	3%
5	4:1:1 W/ Ref	3	0%	1%	18%	75%	5%
6	1:1:1	3	0%	1%	14%	76%	9%

n=185 for each mixture

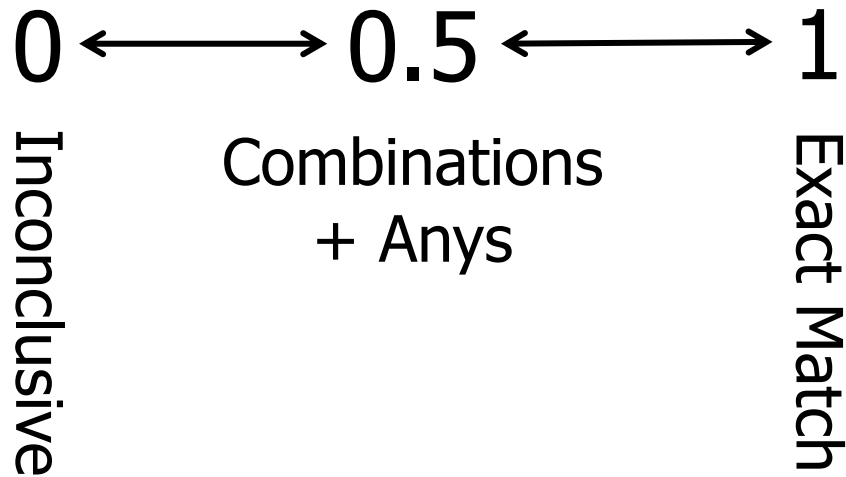
\*Includes "Consistent With"



# Metrics: GIM + AM



GIM:



*How many answers did I provide at each locus?*

AM:

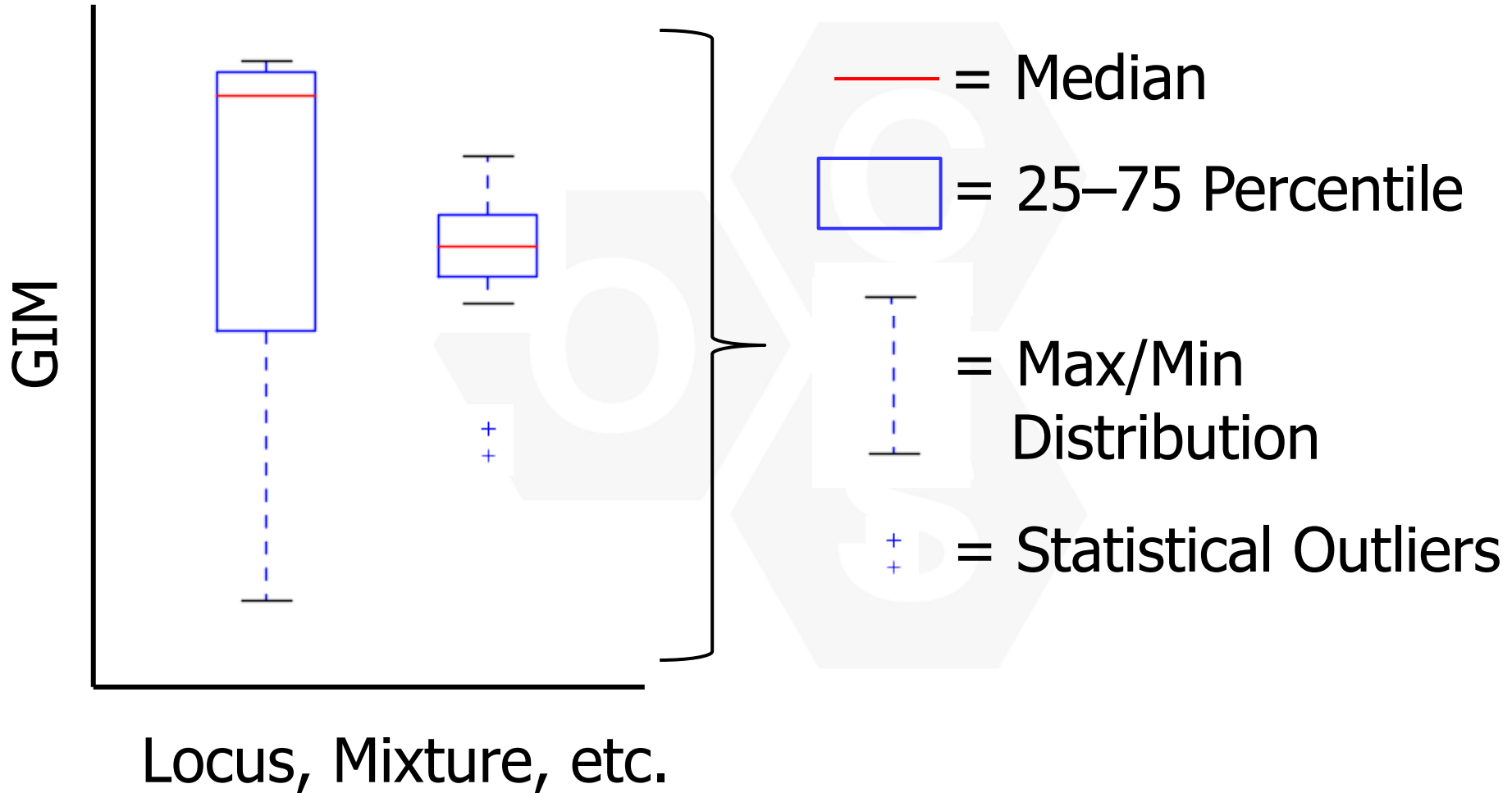
Known	Generated	AT	AF	Inc
11, 12	11, 12	2	0	0
	11, Any	1	0	0
	11, 13	1	1	0
	10, 13	0	2	0
	Inc.	0	0	2

*Did my genotypes include the “correct answer”?*

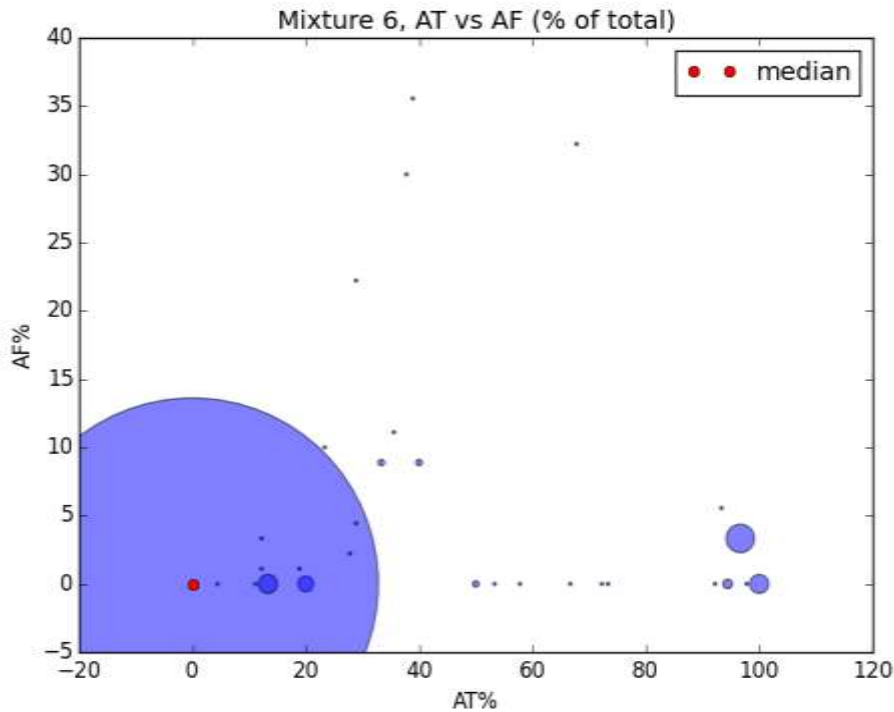




# Visualize Variation: GIM Box Plots



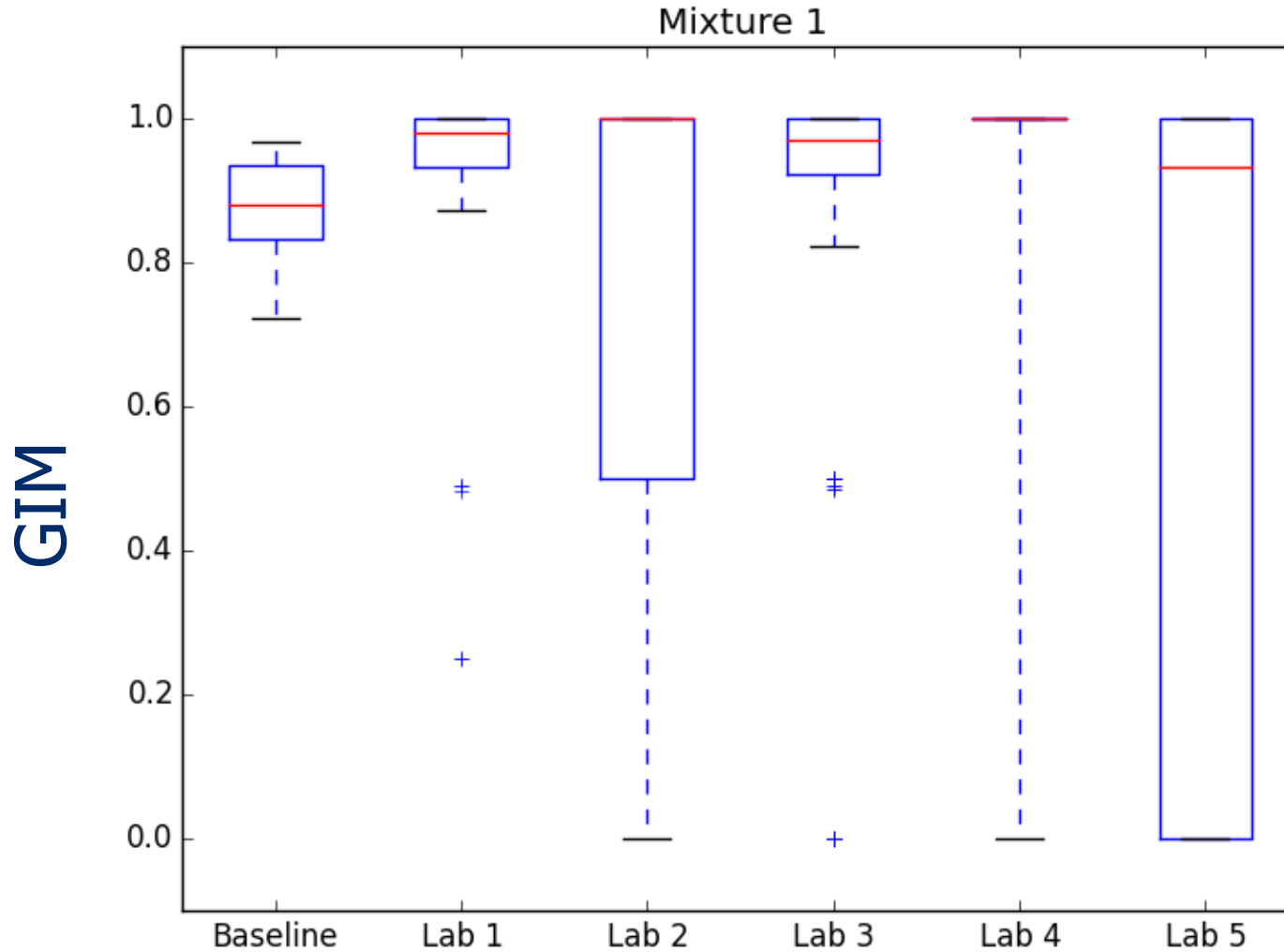
# Visualize AM Variation: Truth vs. Mismatch



- AT vs. AF
- AF vs. GIM
- AF vs. Inc.
- Each dot represents a single examiner
- Larger radius, increased number of examiners with same score

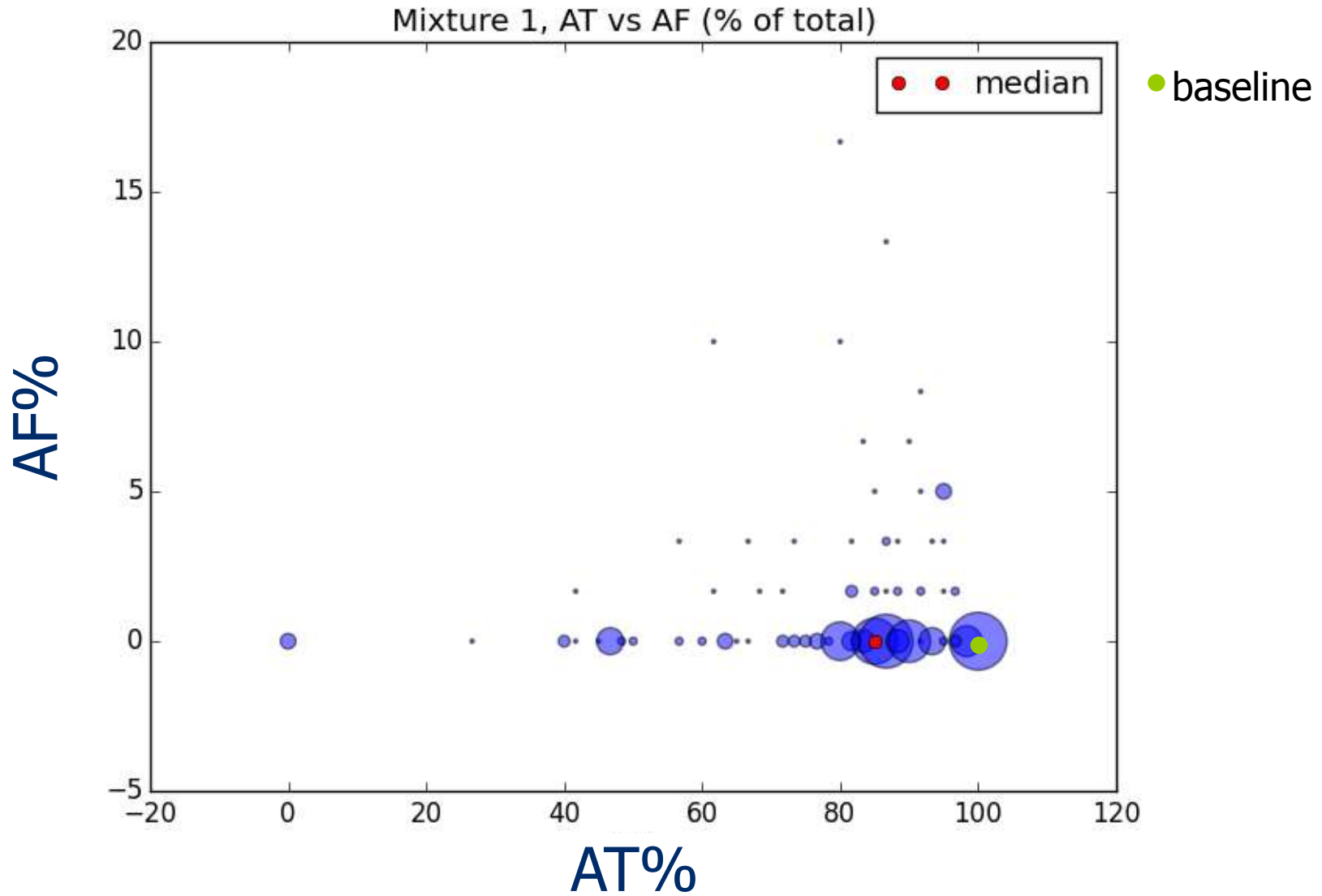


# Mixture 1: 3.5:1, 2-person





# Mixture 1: 3.5:1, AF vs. AT

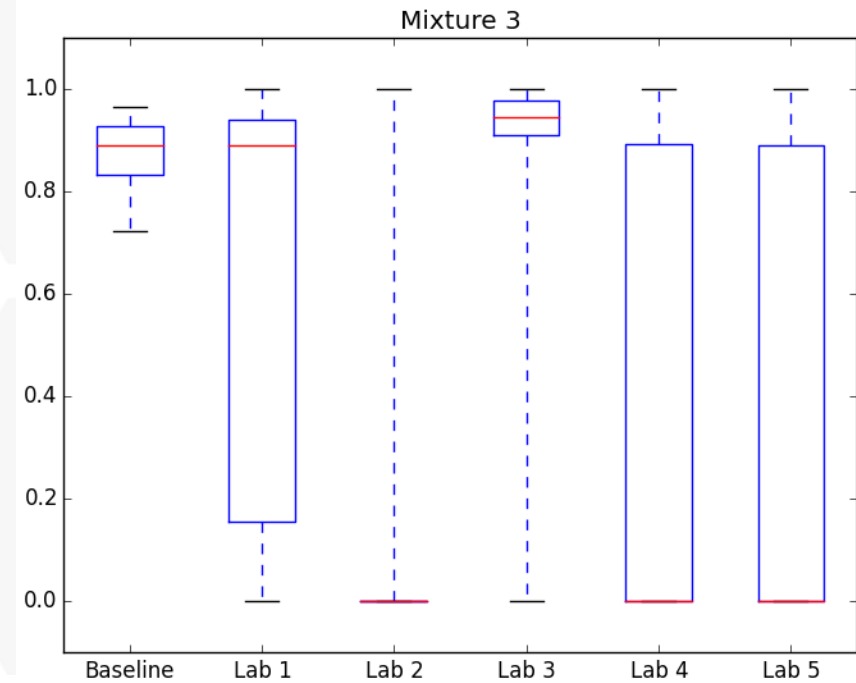
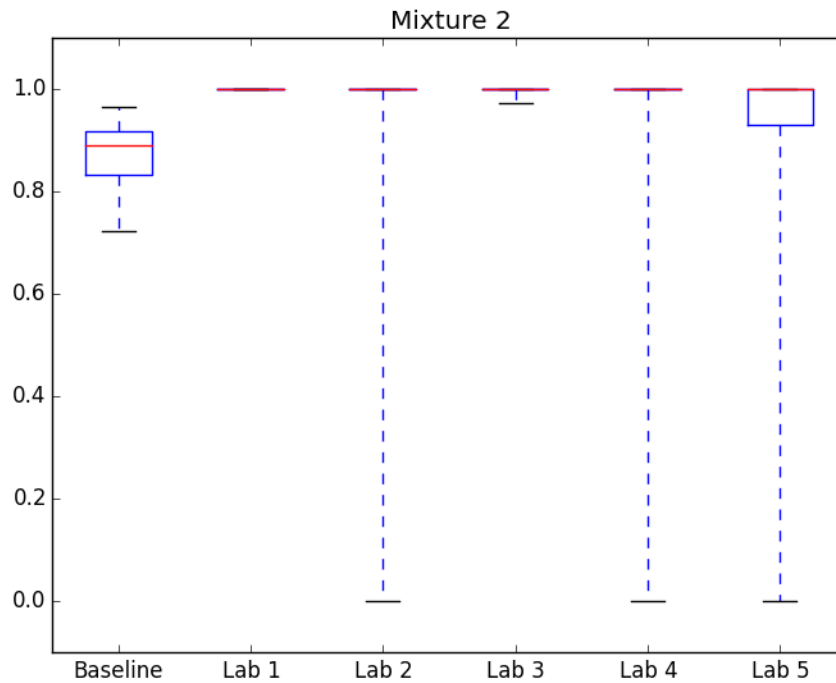




# Mixture 2 and 3: W/ and W/O Reference

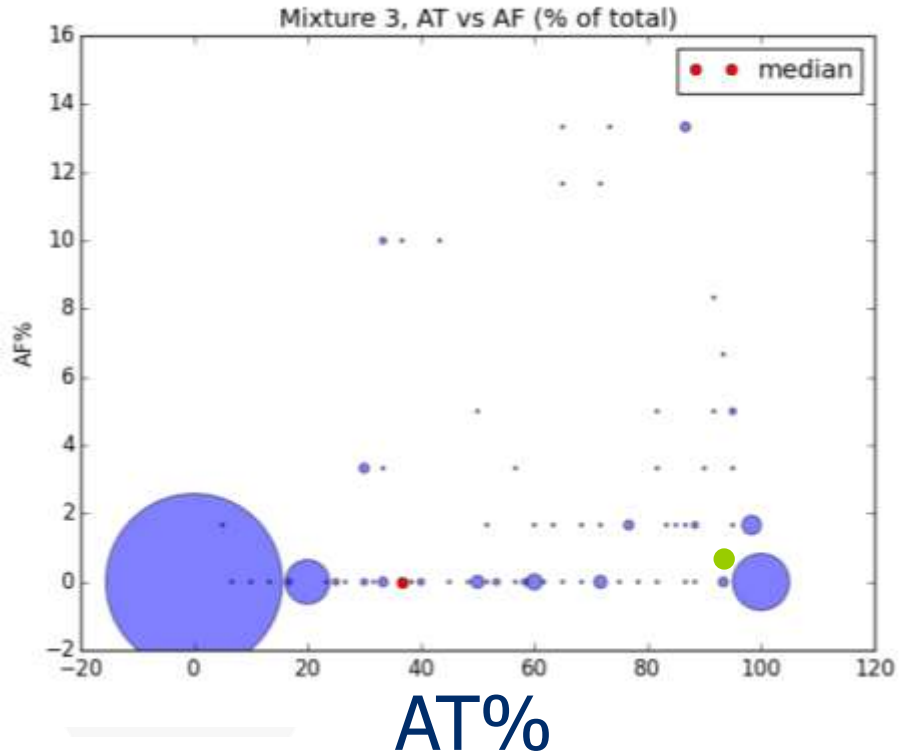
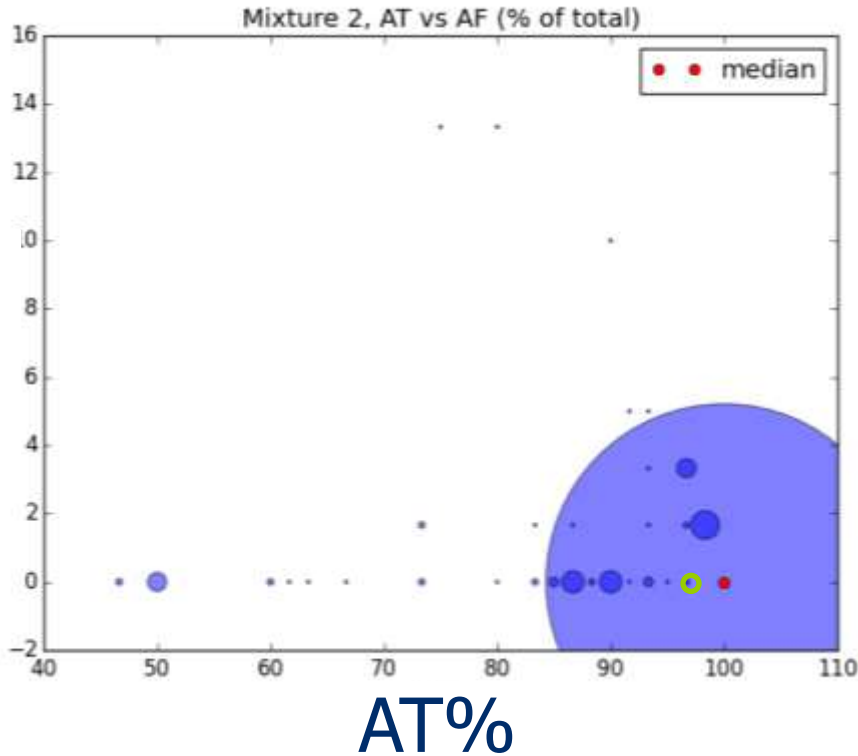


GIM





# AF vs. AT: W/ and W/O Reference



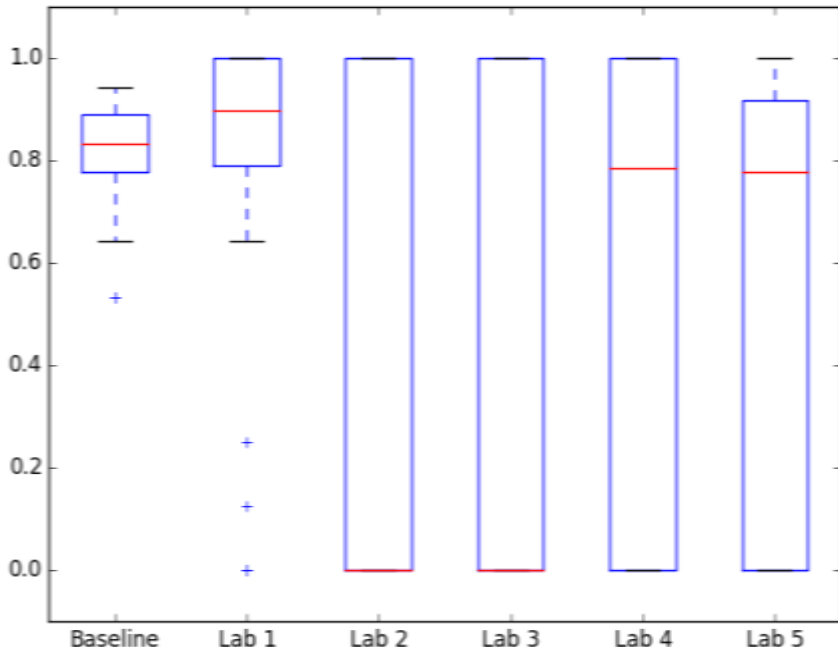
● baseline



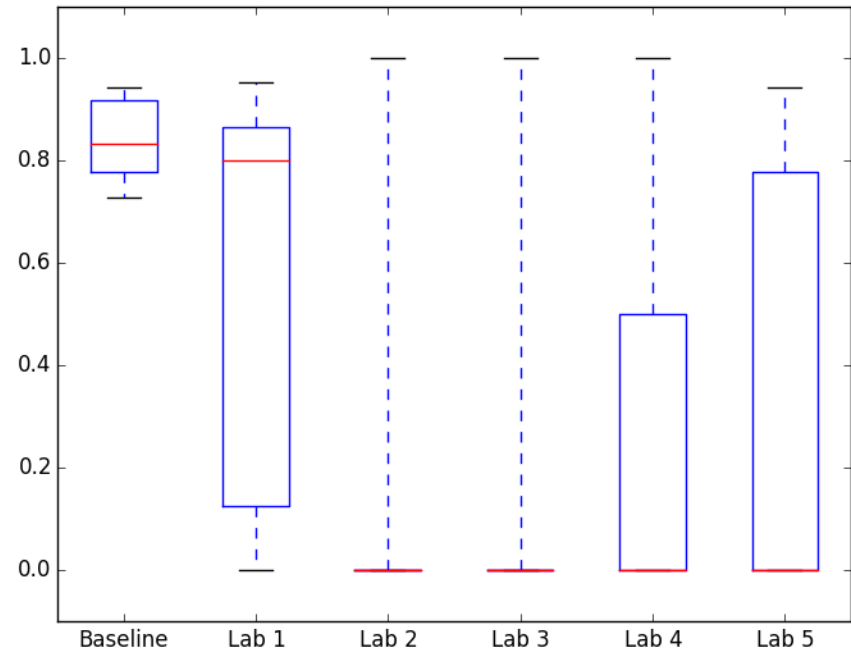
# 3-Person Mixtures:

GIM

Mixture 5

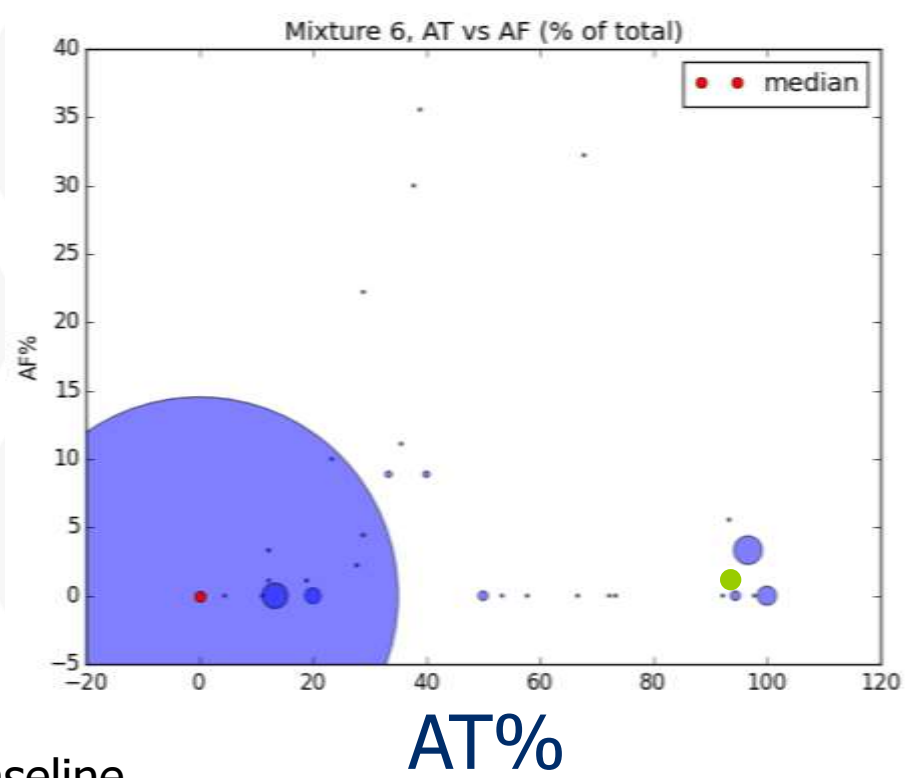
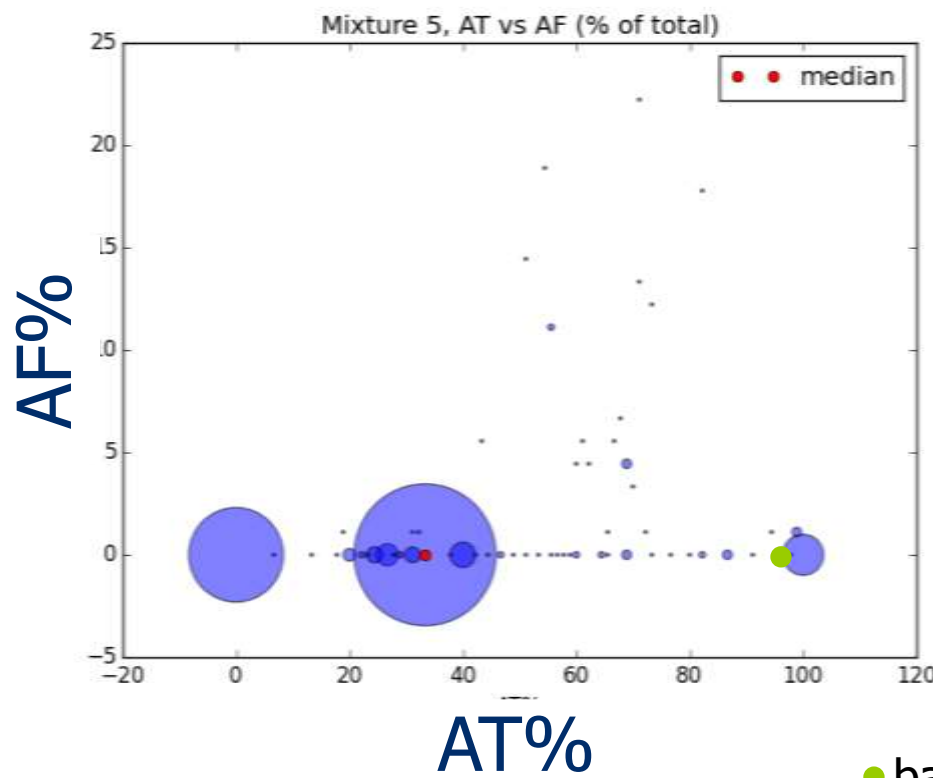


Mixture 6





# 3-person Mixtures: AF vs. AT







# AM: Summaries

## Mixture 1: Major and Minor\*

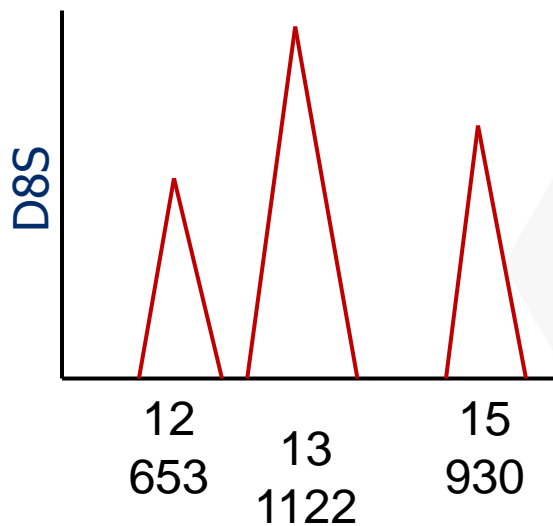
Locus	AT%	AF%	INC%		Locus	AT%	AF%	INC%
D8S1179	65.8%	0.8%	31.6%		D2S1338	89.0%	0.3%	10.3%
D221S11	89.4	0.3	10.3		D19S433	77.3	1.0	12.6
D7S820	88.2	0.5	11.3		vWA	76.6	0.7	11.9
CSF1PO	59.0	0.3	37.4		TPOX	63.2	0.8	32.9
D3S1358	87.1	1.3	8.1		D18S51	86.1	1.0	11.6
THO1	83.7	2.4	8.4		D5S818	81.0	0.5	11.9
D13S317	70.7	0.8	16.5		FGA	88.5	0.8	10.7
D16S539	76.9	0.5	12.9					
<b>MIXTURE AVG</b>	<b>78.8</b>	<b>0.8</b>	<b>15.9</b>					

\*ID+, n=155

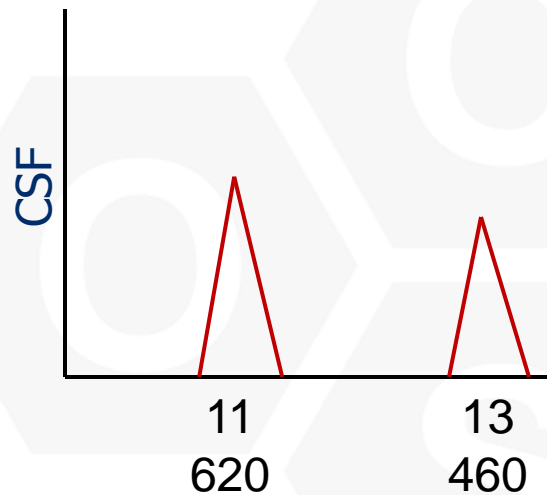


# Inc's: Mixture 1

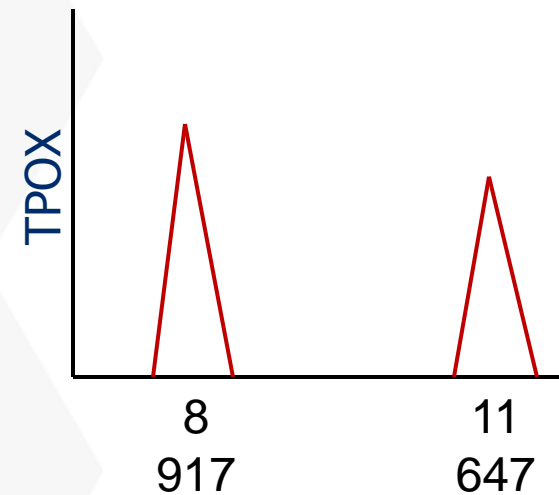
## 3.5:1, 2-person mixture



Known Major: 13,15  
Known Minor: 12,12  
Inc= 31.6%



Known Major: 11,13  
Known Minor: 11,11  
Inc= 37.4%



Known Major: 11,13  
Known Minor: 11,11  
Inc= 32.9%

AVG Inc<sub>LocI</sub> across mixture= 15.9%



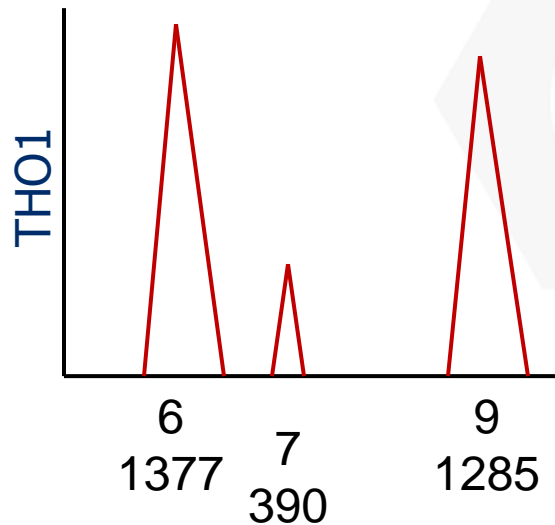
# AM: THO1, Mixture 1



3.5:1, 2-person mixture

Known Major: 6,9

Known Minor: 6,7



Locus	AT%	AF%	INC%
THO1	83.7	2.4	8.4

↓

Locus	Minor	%
THO1	7,7	100

↓

Lab	Total Lab n=	AF n=	% of Entire Lab
A	11	6	55%
B	14	2	14
C	4	2	50
D	10	2	20
E	17	1	6
F	2	1	50
G	5	1	20



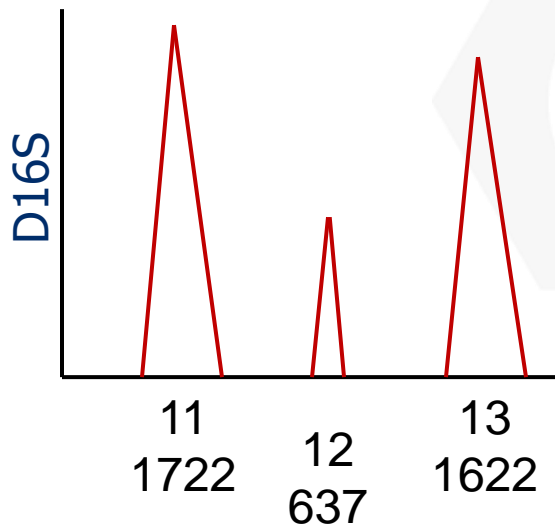
# AM: D16S, Mixture 4



3:1, 2-person mixture

Known Major: 11,13

Known Minor: 11,12



Locus	AT%	AF%	INC%
D16S	83.2	3.7	8.7

↓

Locus	Minor	%
D16S	12,12	90
D16S	11,11;11,13;13,13	10

↓

Lab	Total Lab n=	AF n=	% of Entire Lab
A	11	7	64%
B	14	3	22
D	10	2	20
E	17	2	12
F	2	1	50
G	5	3	60

D16S Minor 12,12



# AM: Within a Laboratory

Lab	Examiner	AF on THO1	AF on D16S
A	1	Yes	Yes
	2	Yes	Yes
	3	Yes	Yes
	4	No	Yes
	5	Yes	No
	6	Yes	Yes
	7	Yes	Yes
	8	No	Yes
	9	No	No
	10	No	No
	11	No	No



	Across Lab	
Lab	% AF Both	% At least 1
A	45%	73%



# Summary

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- Continue to analyze data
- Classification of errors: transcriptional, SOP-related, examiner generated
- Large variation differences exist in different aspects of interpretation
- Potential use of DEAT in training, assessment, and proficiency testing of DNA examiners
  - **23July2015, 2:10 (QA)**



# Acknowledgement

Office of the Chief Scientist, DFSC



Forensic Exploitation Directorate, DFSC

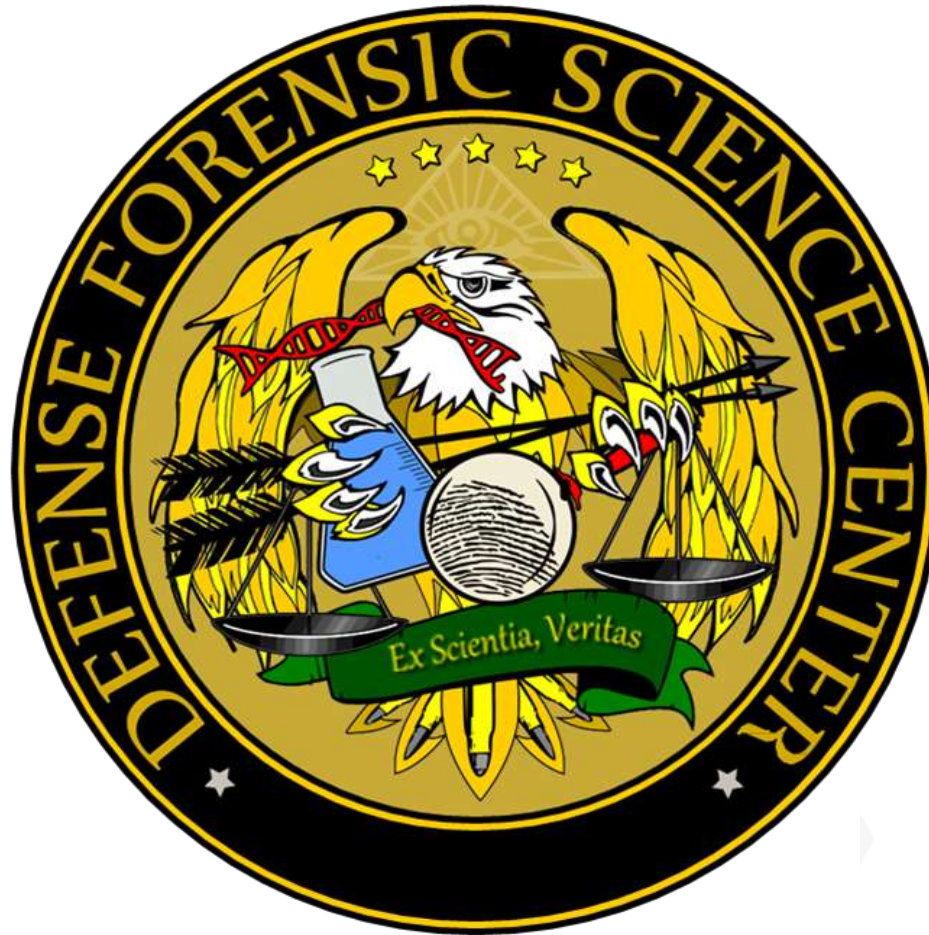


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