2018 Rapid DNA Maturity Assessment

Erica Romsos & Peter Vallone

Applied Genetics Group, NIST

November 7, 2018

NIST APPLIED GENETICS

Steps in Forensic DNA Analysis

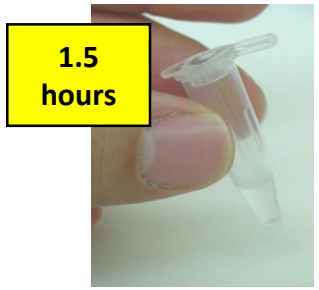
Steps Involved

- Collection
- **Extraction**
- **Quantitation**
- **Multiplex PCR**
- **STR Typing**
- **Interpretation of Results**

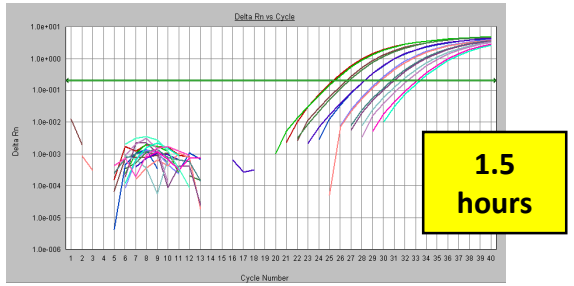
Blood Stain

Buccal swab

Sample Collection

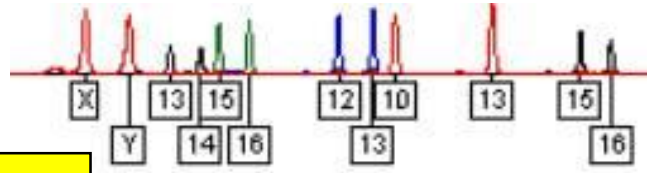


DNA Extraction



DNA Quantitation

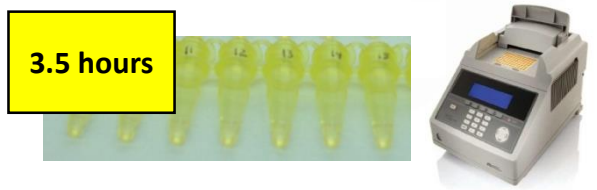
DNA separation and sizing



1.5 hours

STR Typing

Interpretation of Results



Multiplex PCR Amplification

1-2 day process (a minimum of ~8 hours) with current laboratory procedures and technology

Commercial RDNA Instruments

IntegenX

- RapidHIT 200
  - PowerPlex 16HS
  - Globalfiler
- RapidHIT ID
  - Globalfiler

3-7 Samples
2 hour run time

1 Sample
90 minute run time

ANDE

- ANDE/DNAScan
  - PowerPlex 16
- ANDE
  - FlexPlex (27 loci)

5 Samples
86 minute run time

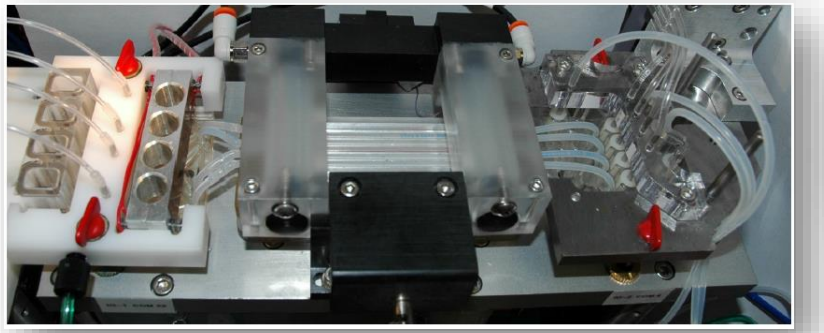
Rapid DNA Instrument Testing

2012-2015: Ongoing communication with Developers

2008 2010 2012 2014 2016 2018

08/2017: Rapid DNA Act Passes

2010: Apollo 200 Testing (**136 samples**)



09/2012: First prototypes delivered to NIST (DHS Instruments)

2015: NIST Participates in IXI PP16 Dev. Validation (**100 samples**)

2016: NIST Participates in ANDE PP16 Dev. Validation (**150 samples**)

2016: NIST Participates in testing ParaDNA

2013: NIST Interlab Study (**350 samples**)

2014: NIST Maturity Assessment (**280 samples**)

2017: NIST Participates in ANDE FlexPlex Dev Validation (**250 samples**)

2018: NIST Rapid DNA Maturity Assessment (**240 samples**)

Forensic Science International: Genetics Supplement Series 5 (2015) e1–e2

Contents lists available at ScienceDirect

Forensic Science International: Genetics Supplement Series

journal homepage: www.elsevier.com/locate/FSIGSS

ELSEVIER

Rapid DNA maturity assessment

Erica L. Romsos[a,*], Sanae Lembirick[b], Peter M. Vallone[a]

[a] U.S. National Institute of Standards and Technology, NIST, 100 Bureau Drive, Gaithersburg, MD 20899-8314, USA
[b] Montgomery College, Rockville, MD 20850, USA

NIST: Rapid DNA Maturity Assessments

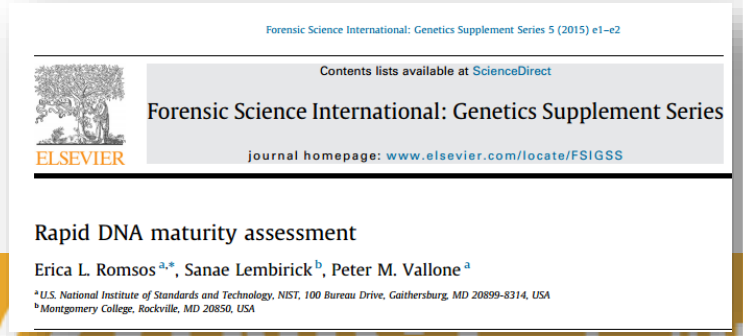
- Collection and distribution of samples to all participating laboratories
  - 2013: 3 labs, 150 samples
  - 2014: 7 labs, 280 samples
  - 2018: 9 participants, 240 samples
- Coordination of all testing sites to include return of all data to NIST for analysis and review
  - Includes IRB approval for swab collection and MTA for distribution
- Analysis and compilation of all data
- Summary of results presented across multiple meetings within the forensic and biometric communities

2013 Rapid DNA Maturity Assessment

- Summer 2013 we felt confident that we could carry out an interlab assessment of the R-DNA prototypes
- Data was collected and analyzed in August 2013
  - All instruments had the same version of software and scripts for testing
- Three federal testing sites (7 individual instruments)
  - 50 single source reference buccal swabs were provided by NIST for each instrument tested
- 350 single source reference buccal swabs tested
  - Success defined as the automated calling of the 13 CODIS core STR loci
- **Overall success = 88.3%**

2014 Rapid DNA Maturity Assessment

- Fall 2014 assessment of the current status of rapid DNA typing technology for the CODIS Core Loci
  - In support of lab and future booking station Rapid DNA implementation
- Many modifications to both hardware and software were made between 2013 and 2014
- 7 participating laboratories (11 independent instruments)
  - 20 single source reference buccal swabs tested
- 280 single source reference buccal swabs tested
  - Success defined as automated (lights out) calling of the 13 CODIS core loci
- **Overall Success = 76.1%**



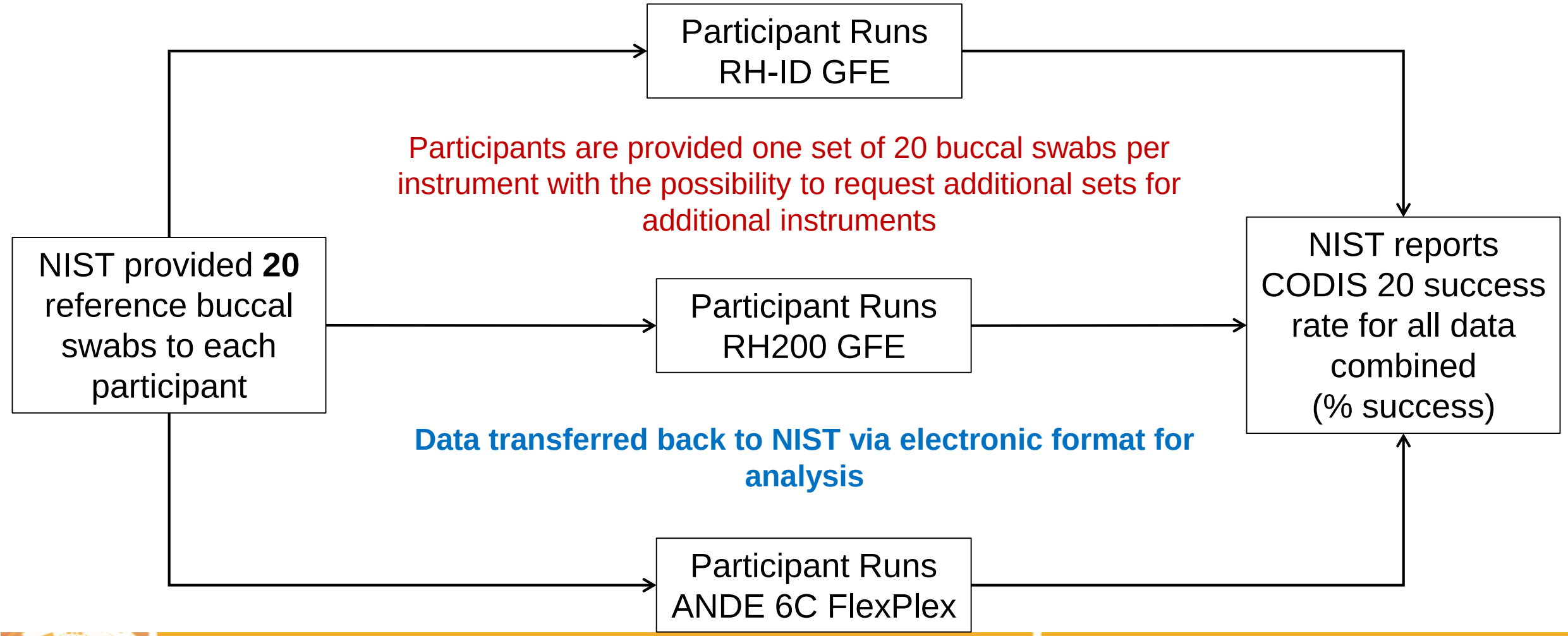
2018 Rapid DNA Maturity Assessment

- **Goal**: To measure the status of rapid DNA typing technology for the 20 CODIS core loci in support of booking station Rapid DNA implementation

- Rapid DNA instruments capable of genotyping the 20 CODIS core loci were eligible for participation

- 20 single source reference buccal swabs were distributed to participating laboratories

2018 Rapid DNA Maturity Assessment Study Outline



Timeline of 2018 Maturity Assessment

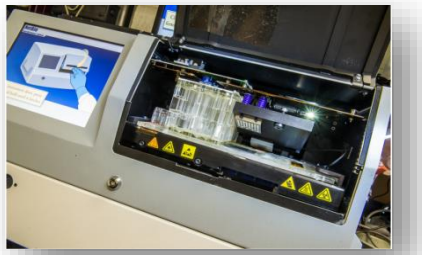
February 2018-Present: Buccal samples collected at NIST and stored at RT (>900 swabs have been collected from >50 unique individuals)

May 2018: Official call for Participants in the 2018 Rapid DNA Maturity Assessment for the CODIS core 20 loci

July 2018: Samples shipped to participants

August 2018: Data returned to NIST for analysis

20 Swabs provided

Participants	Instrument Platforms	Chemistry	Independent Instruments	Total Samples Tested	Analysis Method
Federal	ANDE 6C 	FlexPlex	5	100	Rapid DNA Analysis
State Police	IntegenX RapidHIT 200 	GlobalFiler Express	3	60	Modified Rapid DNA Analysis
Vendor	IntegenX RapidHIT ID 	GlobalFiler Express	4	80	Modified Rapid DNA Analysis
9 Participants	3 Platforms		12 Instruments	240 Samples	

Success Metrics

- Success was measured by **complete and concordant genotypes** produced for the 20 CODIS core loci

- Allele calls by the integrated rapid DNA devices were compared to lab generated profiles for concordance
  - Fusion 6C, PP21, GFE on a 3500xL

- Two interpretation parameters were implemented
  - Rapid DNA Analysis: Without human intervention
  - Modified Rapid DNA Analysis: Expert interpretation and analysis of electropherogram https://www.fbi.gov/file-repository/addendum-to-qas-for-rapid-dna.pdf/view

Rapid DNA Analysis-ANDE

Automated (lights-out) analysis without human intervention

Effective June 1, 2018, the following Rapid DNA system is approved for use at NDIS by an accredited forensic DNA laboratory:

Rapid DNA Analysis System for Accredited Laboratory Use

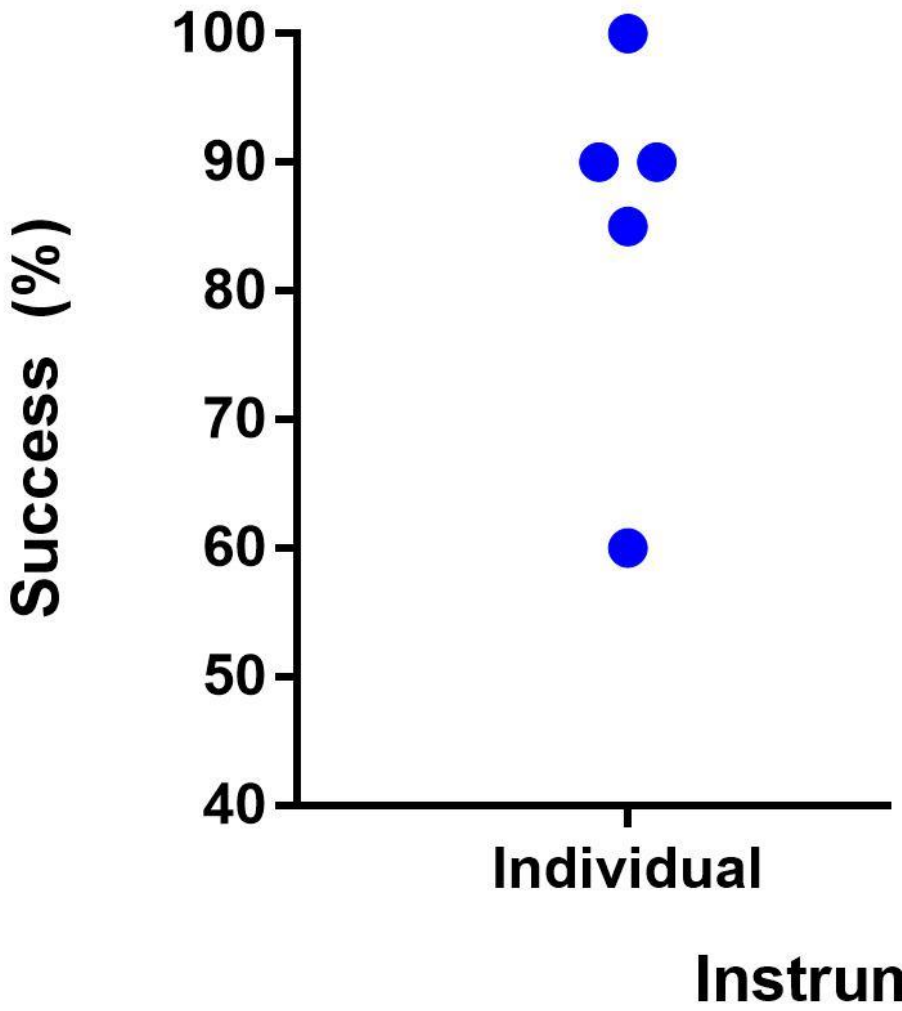
Component	Name	Part/Version Number
Rapid DNA Instrument	ANDE 6C Instrument	A0120001003
Typing Kit	FlexPlex27	FlexPlex27
Cartridge	ANDE A-Chip (FlexPlex)	A0210001057
System Software	ANDE System Software	2.0.6
Expert System Software	ANDE Expert System	2.0.5

To date, ANDE 6C is the only rapid DNA system to be NDIS approved for automated rapid DNA analysis

https://www.fbi.gov/services/laboratory/biometric-analysis/codis/rapid-dna

n=85

CODIS 20 Success: Automated Analysis



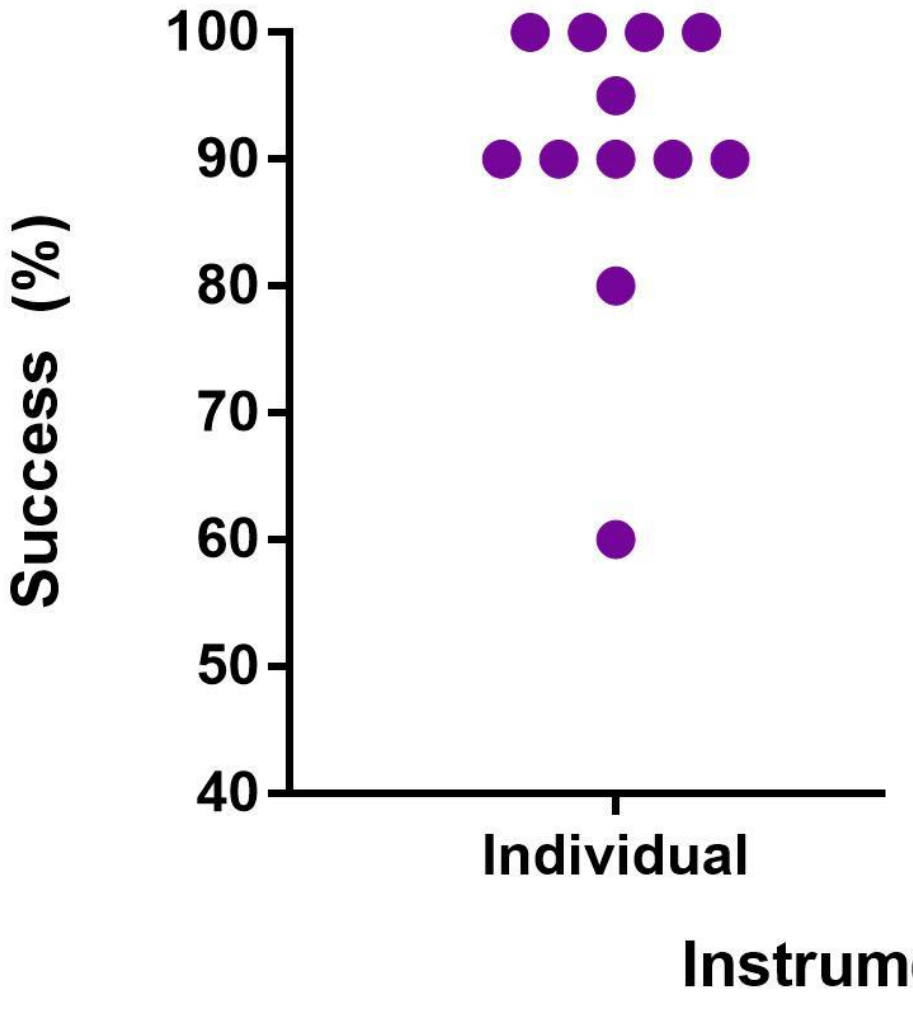
Average Success: 85%

Modified Analysis Parameters

- Samples were manually reviewed (n=240)
  - ANDE profiles were analyzed in GeneMapper IDX v1.5
  - RapidHIT profiles were reviewed in GeneMarker HID v2.8.2
  - Heterozygote balance filter set at 0.25
- After manually interpreting a profile (PHR >0.25, low AT, recovery of "lost" data), concordance was checked against the laboratory generated reference profile
- Success was determined by **complete and concordant** profiles for the 20 CODIS core loci

n=217

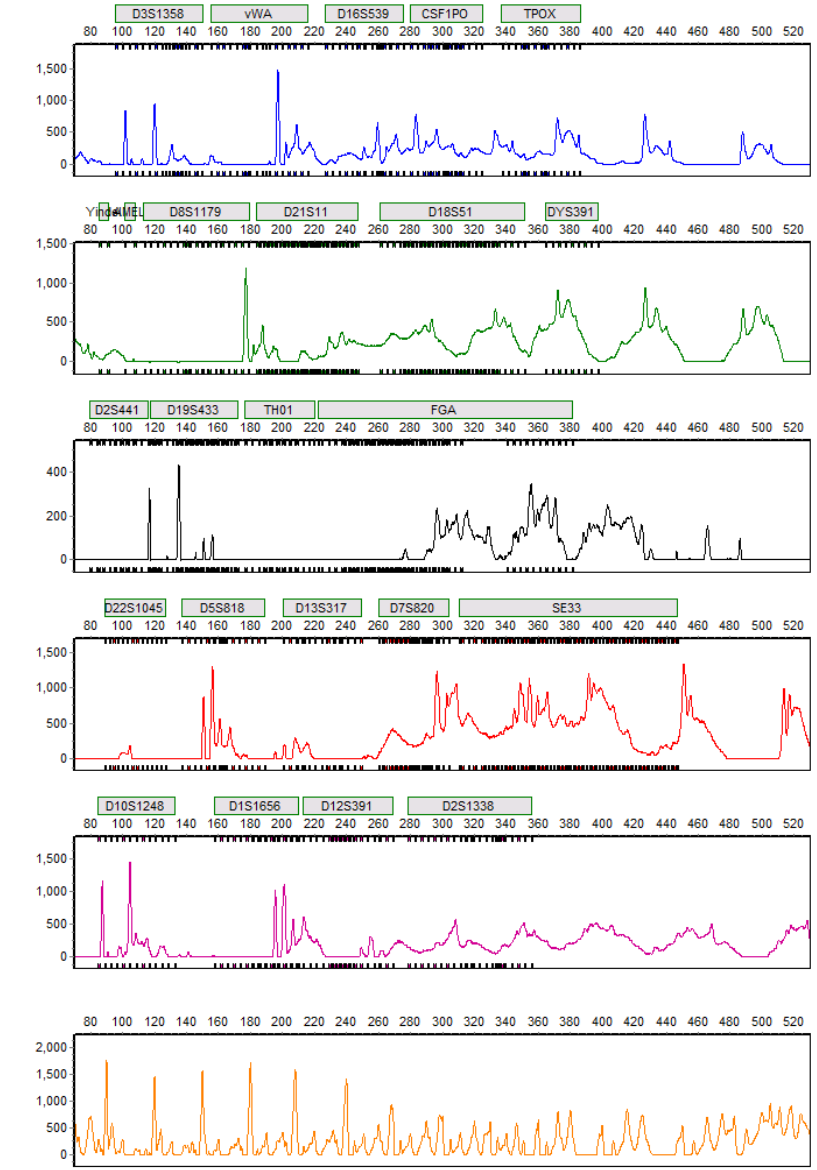
CODIS 20 Success: Modified Analysis



Average Success: 90 %

Unrecovered Samples

n=23

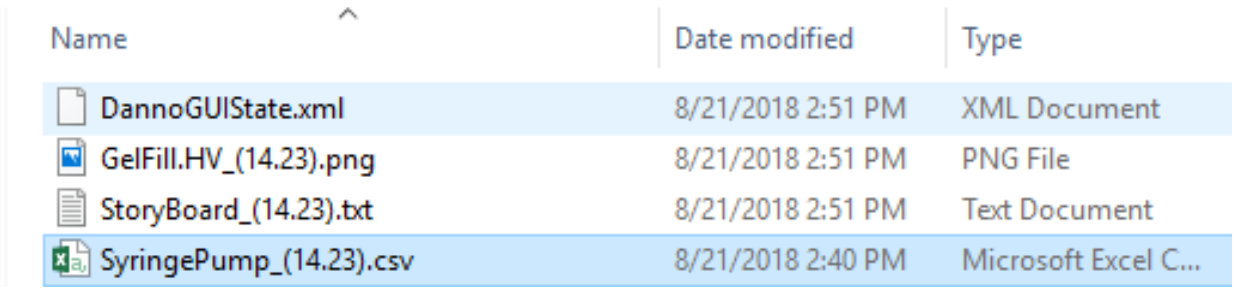


Unrecovered Samples n=23

Instrument Related

Unknown n=9

Data Transfer Failure n=2



Name	Date modified	Type
DannoGUIState.xml	8/21/2018 2:51 PM	XML Document
GelFill.HV_(14.23).png	8/21/2018 2:51 PM	PNG File
StoryBoard_(14.23).txt	8/21/2018 2:51 PM	Text Document
SyringePump_(14.23).csv	8/21/2018 2:40 PM	Microsoft Excel C...

No data was transferred from the instrument to analyze

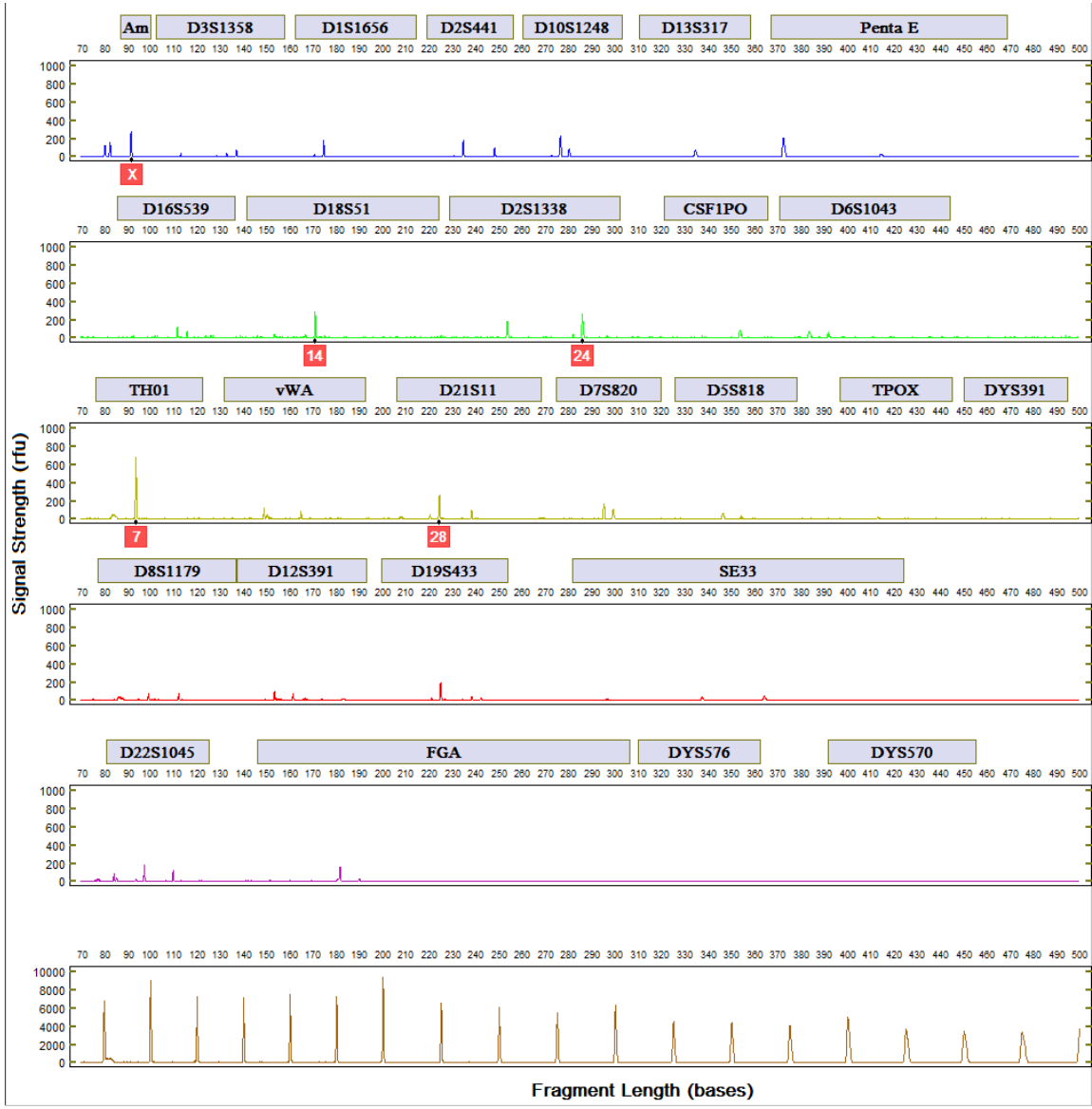
Unrecovered Samples n=23

Instrument Related

Partial Profile n=10

Unknown n=9

Data Transfer Failure n=2



Unrecovered Samples

n=23

Instrument Related

Partial Profile

n=10

Unknown

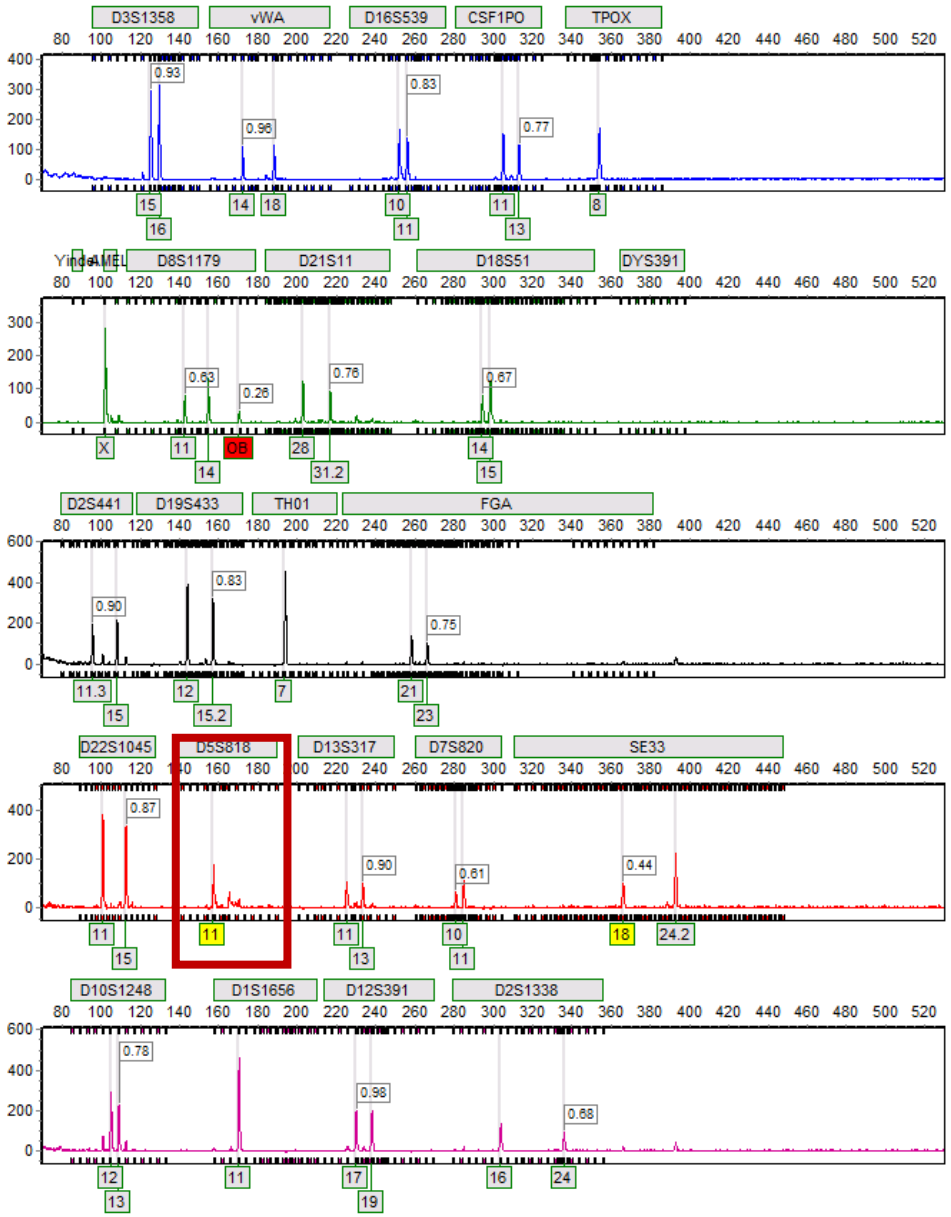
n=9

Data Transfer Failure

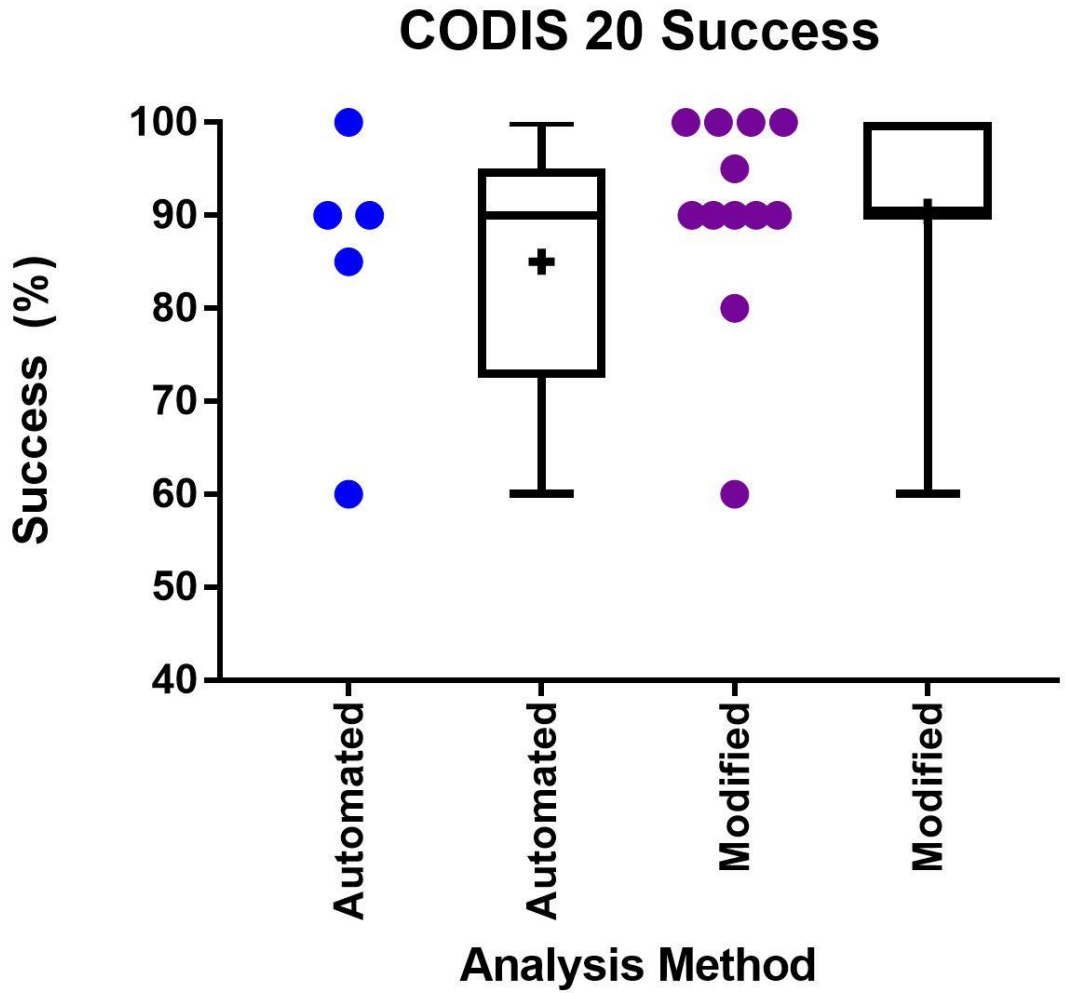
n=2

Single Locus Dropout

n=2



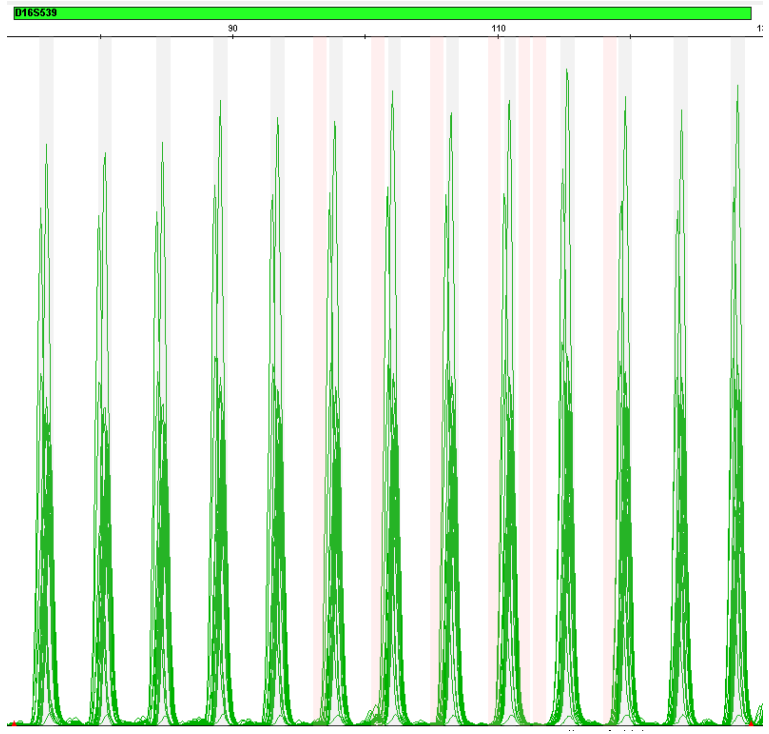
Summary of Success

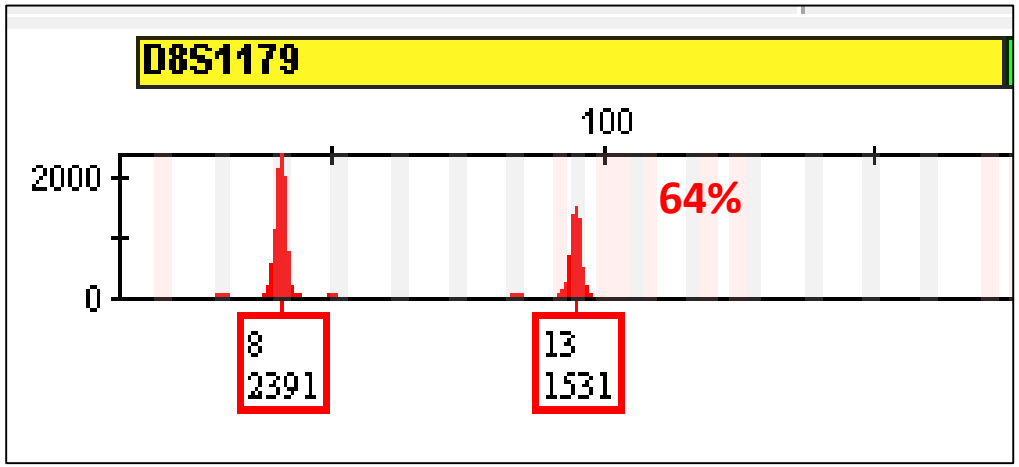


- **Automated success rate of 85% (n=100)**
  - ANDE 6C is the only NDIS approved instrument for automated analysis
  - xml files for the unsuccessful profiles were not generated by the instrument (15%)

- **Modified analysis success rate of 90%**
  - Manual interpretation of all samples (n=240 samples)
  - Increase of success for manually reviewed profiles

n=217 successful profiles





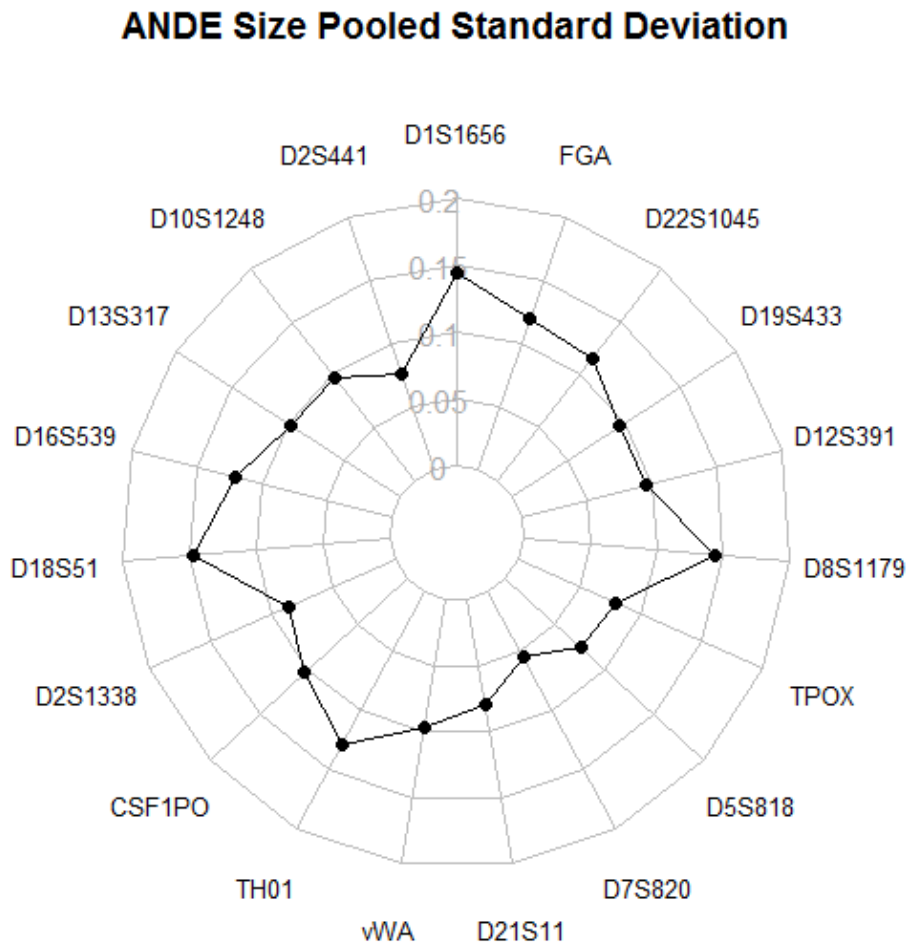
Additional Metrics Analyzed

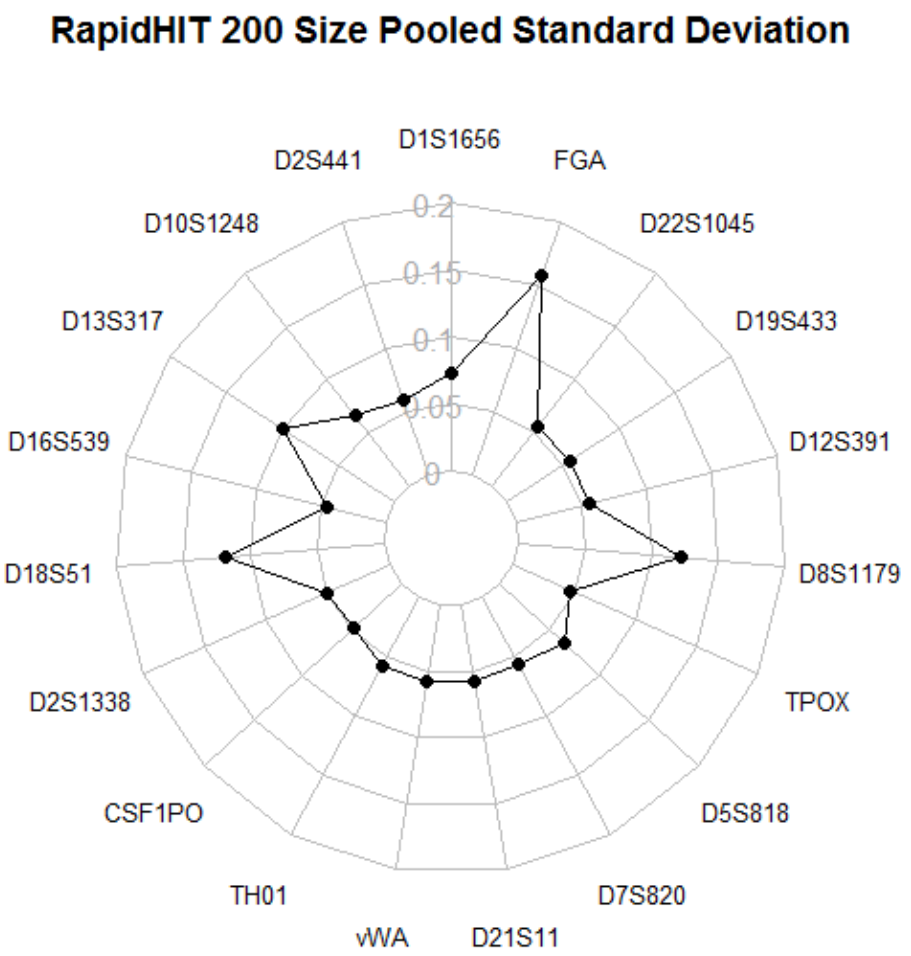
Base pair sizing precision

Heterozygote Balance

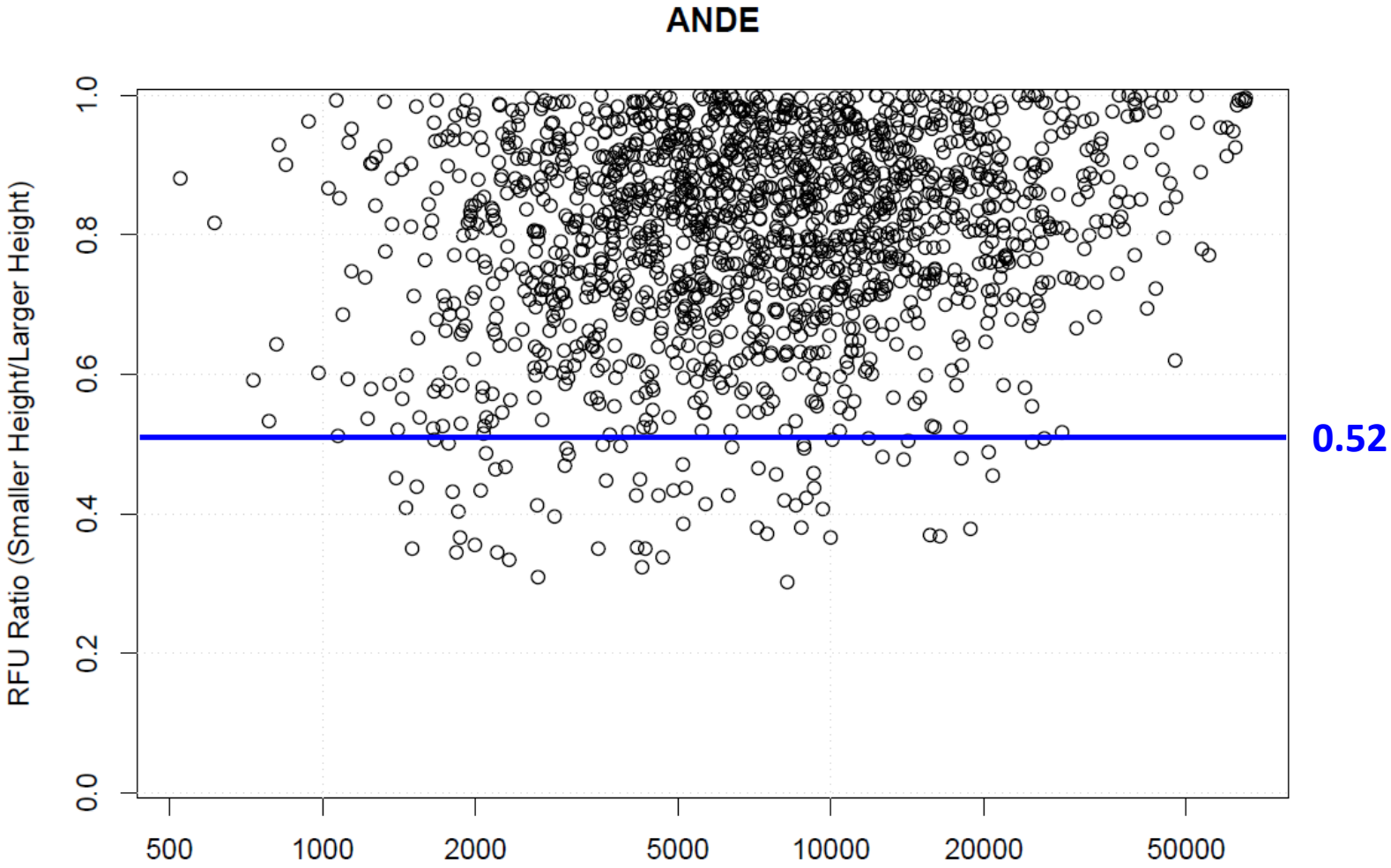
n=19 ladders for ANDE
n=8 ladders for RH 200

Precision-Base Pair Sizing



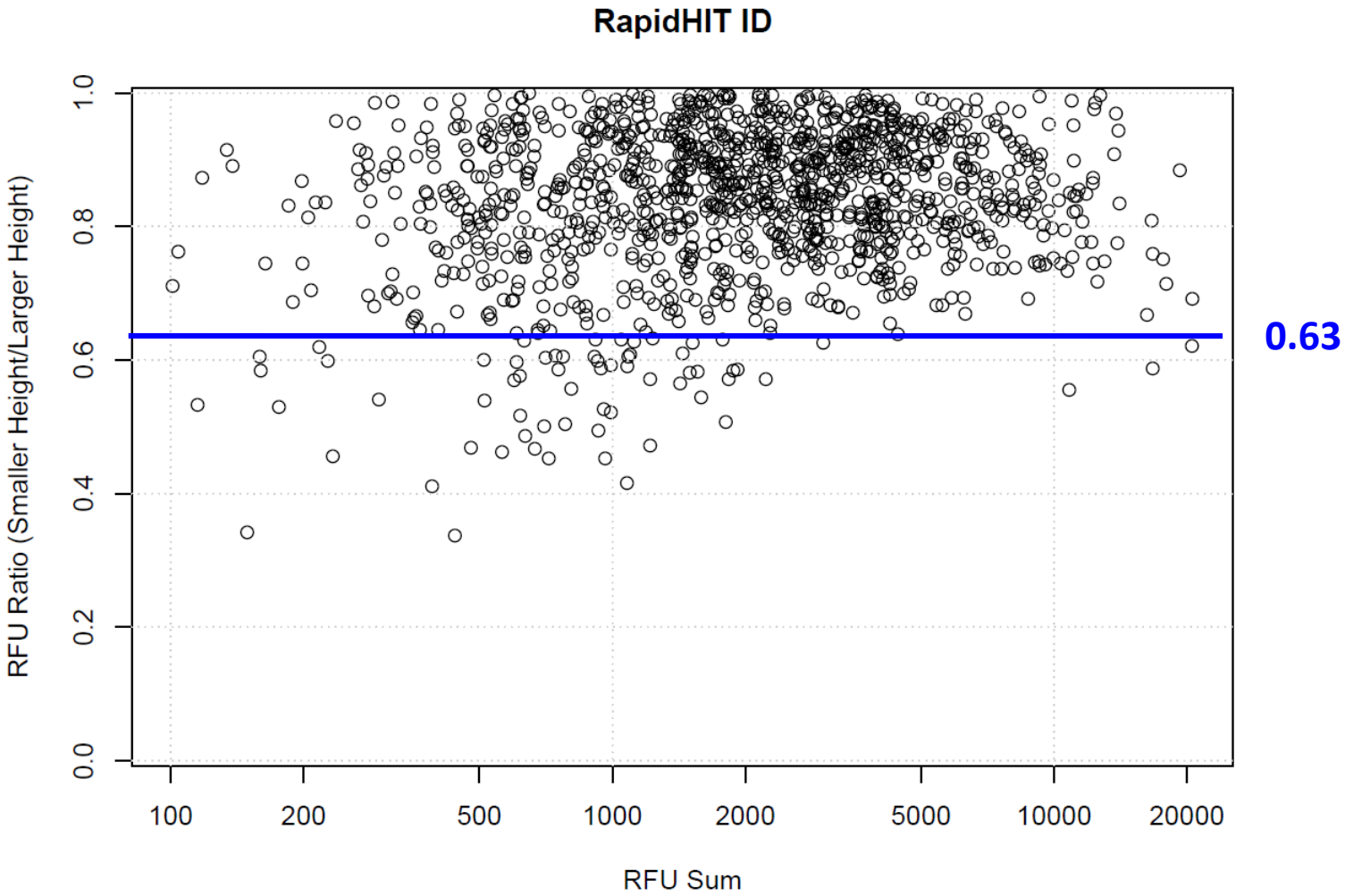


Heterozygote Balance



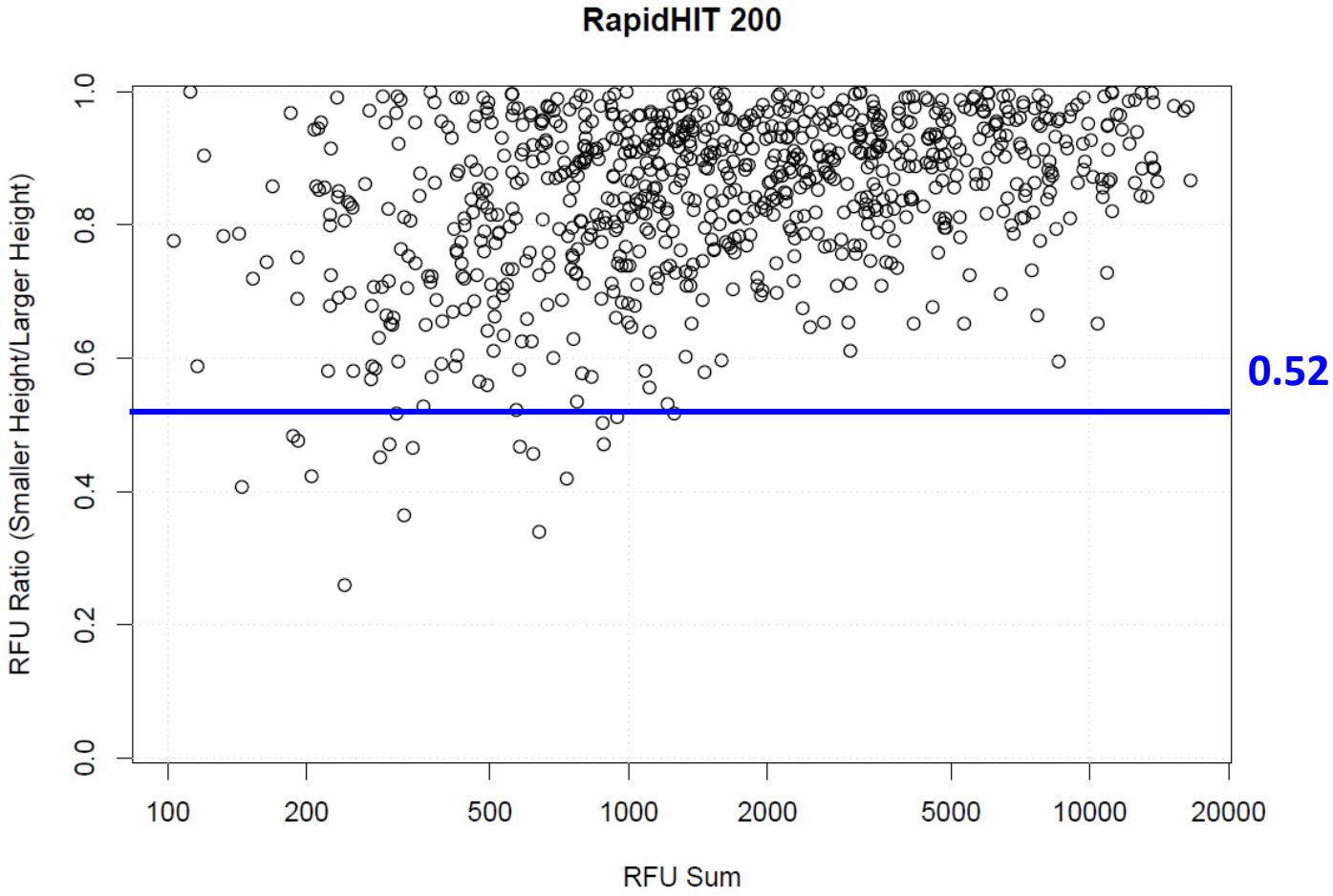
95% of the samples were greater than 52%

Heterozygote Balance



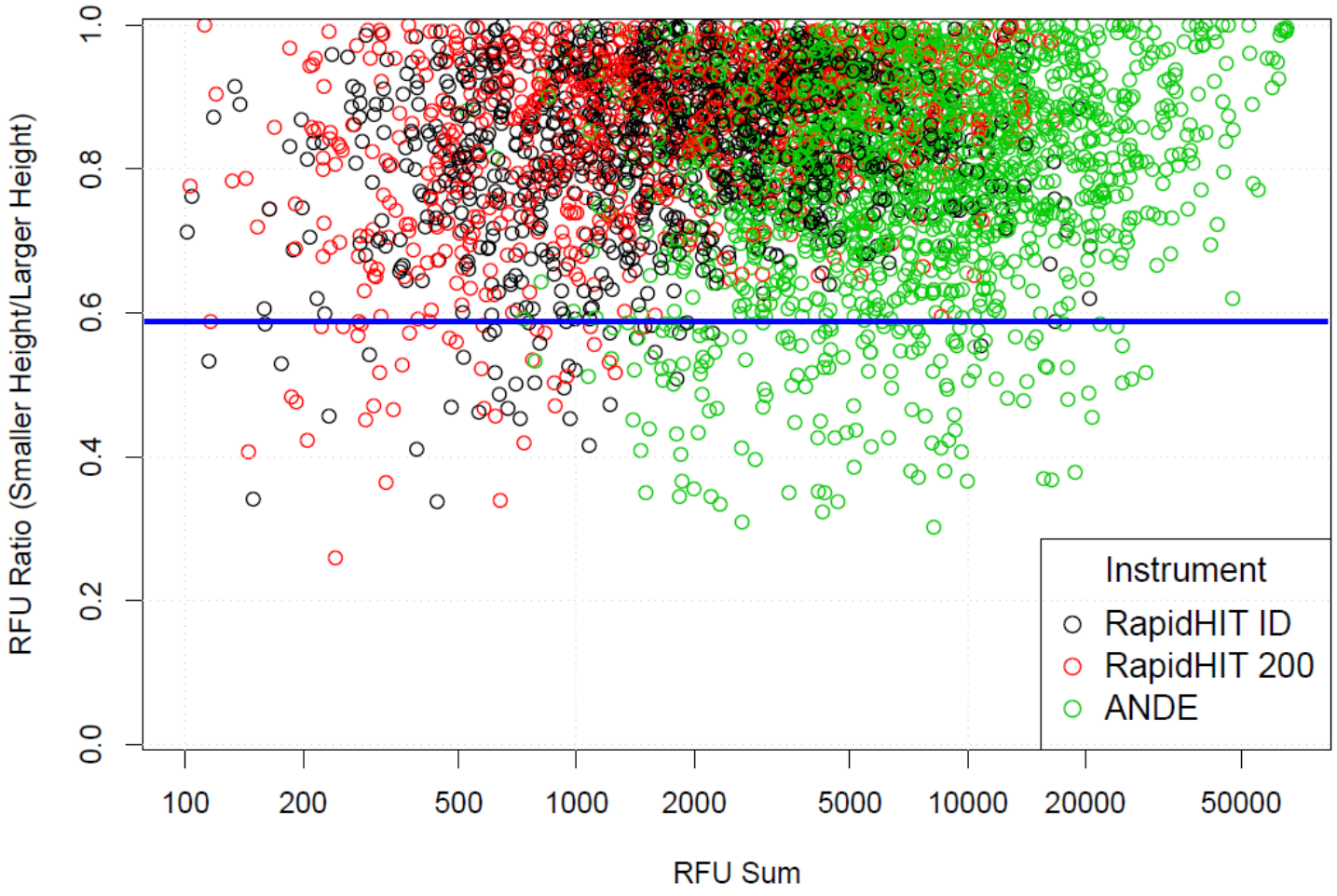
95% of the samples were greater than 63%

Heterozygote Balance



95% of the samples were greater than 52%

Heterozygote Balance



95% of the samples were greater than 59%

Maturity Assessment Summary

- 12 instruments tested across 9 laboratories
- Total of 240 samples examined
  - 85% success rate for the CODIS 20 using **Rapid DNA Analysis**
  - 90% success rate for the CODIS 20 using **Modified Rapid DNA Analysis**
  - Success ranged from 60% to 100%
  - Precision was below 0.17 bp on for both ANDE 6C and RapidHIT 200
  - Combined heterozygote balance (all three instruments) was above 59%

Acknowledgements

Thank you to our participants

- ANDE
- Arizona Department of Public Safety
- Bensalem Police Department
- Federal Bureau of Investigation Laboratory
- Louisiana State Police Crime Laboratory
- Miami Beach Police Department
- Miami Dade Police Department
- NIST (DHS instruments, run at SNA Intl.)
- U.S. Army Criminal Investigation Laboratory

NIST – Applied Genetics Group
Peter Vallone
Steven Lund

Funding
FBI Biometrics Center of Excellence: Forensic DNA Typing as a Biometric tool.

Contact Information
erica.romsos@nist.gov