DNA Mixture Interpretation:
Principles and Practice in Component Deconvolution and Statistical Analysis

Deconvolution of Mixed DNA Profiles Using a Proportionately Shared Allele Approach

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DNA_DataAnalysis

- Developed by Tom Overson at USACIL with input from USACIL DNA Examiners
- Excel VBA Application
- Total Casework Data Management Tool
- (Post GeneMapper ID)

Major Features of DNA_DataAnalysis

- Matching
  - Within cases, between cases, to Staff Database
  - Highlight Foreign Alleles
- Statistical Analysis
  - Single Source
  - Mixed Profile (more than one possibility at a locus)
  - CPI/CPE
  - LR
  - No manual data entry (just mouse clicks)

Major Features of DNA_DataAnalysis

- QA/QC Checks
- Examiner/Reviewer Concordance
- Identifiler, Profiler Plus/COfiler, Yfiler, PowerPlex 16
- CODIS Features
- Mixture Deconvolution

Benefits of Using DNA_DataAnalysis

- Significant time savings
  - Simple case can go from GMID table to write up in 5 minutes (record of alleles, matches, QA checks, stats, profile for CODIS entry)
  - Instantaneous check for Examiner/Reviewer concordance
- Less errors
  - No typos (especially for stats)
  - No transcription (don't need pen and paper)

Benefits of Using DNA_DataAnalysis

- Record of mixture interpretation
- Can document the support of the interpretation
  - Summary of proportions and peak height ratios
  - Graph of contributor proportions
- Consistent math used
  - No fat fingers on the calculator
  - Same equations for every sample
  - Easy to understand and explain

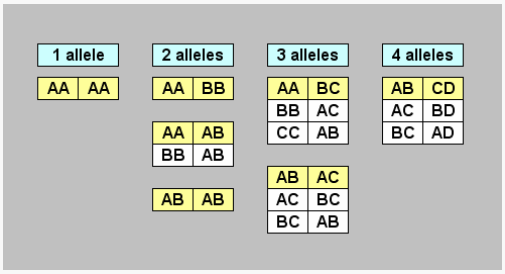
Mixture Deconvolution

- For two- and three- person mixtures
- Considers all possible combinations of alleles at all loci
- Allows for conditioning on required profiles – (can set up to 6 references)
- Creates record of interpretation and rationale of examiner's decision

Mixture Deconvolution

- NOT an Expert System
- A comprehensive tool that assists an expert
- Does everything you would do using pen and paper
- Based on proportionate allele sharing

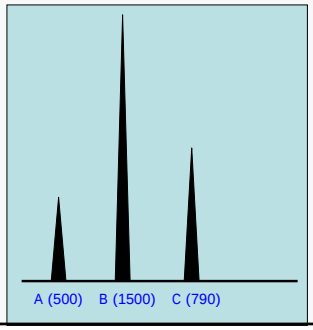
DNA_DataAnalysis calculates for all possible 2-Contributor combinations …



DNA_DataAnalysis calculates for all possible 2-Contributor combinations …

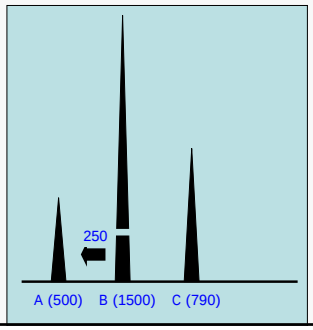
- Only 7 “families” of possibilities
- Only 14 total possibilities
- Largest “family” n=3

Evaluate alleles ABC for genotypes AB & BC



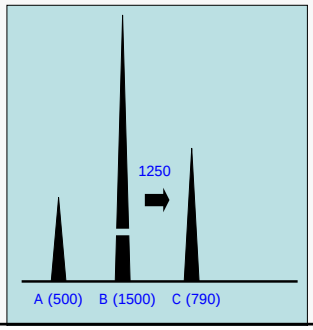
- Scenario: Victim is AB, and this is from vaginal swab
- Is it appropriate to assume a 50% PHr for the AB?

Can you assume the B contribution to the AB is 250?



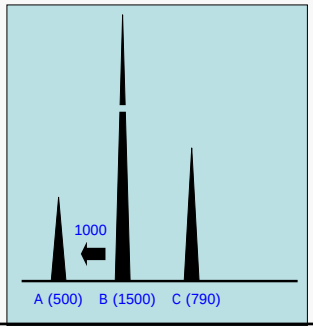
- If so, then AB PHr = 250 ÷ 500 = 0.5
- AB portion = (250 + 500) ÷ t = 0.27
- t = 500+1500+790

Can you assume the B contribution to the AB is 250?



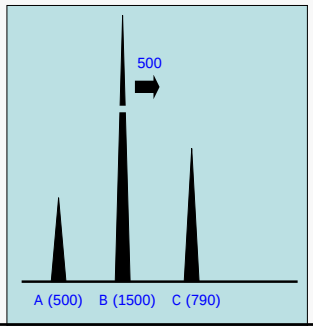
- Then BC PHr = 790 ÷ 1250 = 0.63
- BC portion = (1250 + 790) ÷ t = 0.73

Or do you assume the B contribution to the AB is 1000?



- If so, then AB PHr = 500 ÷ 1000 = 0.5 *(same as Test 1)*
- AB portion = 500 + 1000 ÷ t = 0.54 *(double Test 1)*

Or do you assume the B contribution to the AB is 1000?



- Then BC PHr = 500 ÷ 790 = 0.63

(same as Test 1)

- BC portion of total RFUs = (500 + 790) ÷ t = 0.46

(less than Test 1)

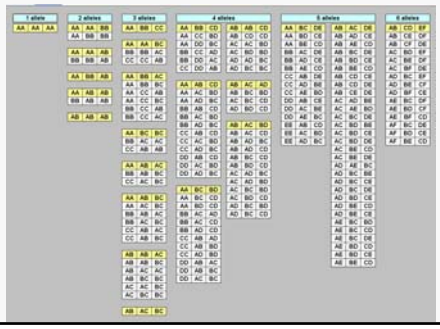
What is the rationale for choosing Test 1 … or Test 2 ... or any other scenario?

Test 1	Test 2
AB PHr = 0.5	AB PHr = 0.5
AB p = **0.27**	AB p = **0.54**
BC PHr = 0.63	BC PHr = 0.63
BC p = **0.73**	BC p = **0.46**

The approach developed for DNA_DataAnalysis

- Calculates PHr's and p's without any arbitrary conditioning based upon a known reference or major contributor
- Uses the same set of calculation rules for every instance
- Calculations based on 3 simple rules
- Proportionate Allele sharing
- PHr and p both important

… and all possible 3-Contributor combinations



… and all possible 3-Contributor combinations

- 23 “families” of possibilities
  - 3 allele pattern has 8 “families” alone
- 150 total possibilities
  - 5 alleles, 3 heterozygotes has 30 possibilities

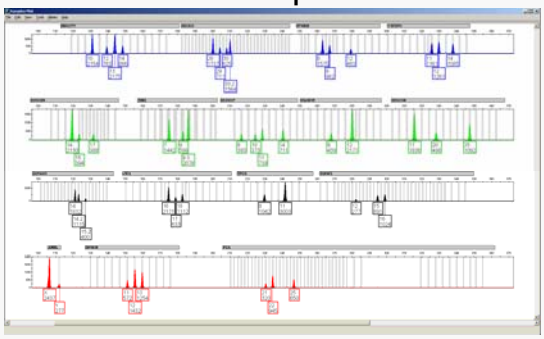
3-contributor combinations

- The same rules apply
- Again, calculates for each and every combination
- Most 3-contributor patterns break down into simple 2-person patterns
  - (plus a somewhat independent 3rd person)

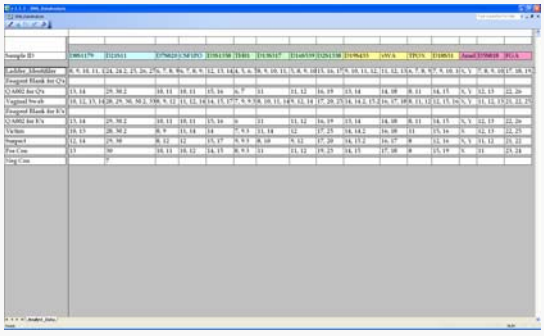
3-contributor combinations

- Where 2-contributors share a type, the p is a combined proportion
- Four situations with “double sharing” are treated differently
  - Cannot calculate PHr or p
  - Valid combinations described as “not excluded” based on required PHr and mPH

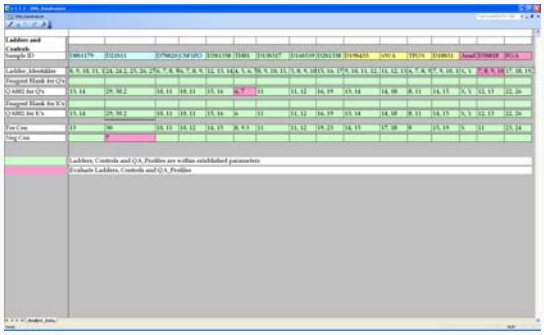
Example



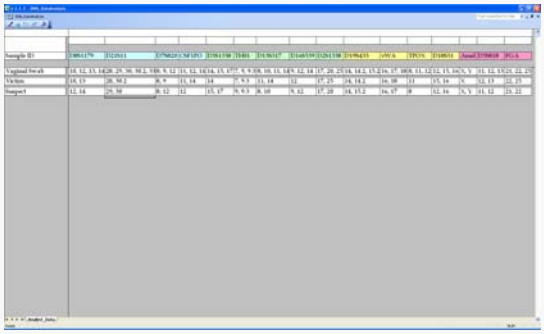
Main Screen View



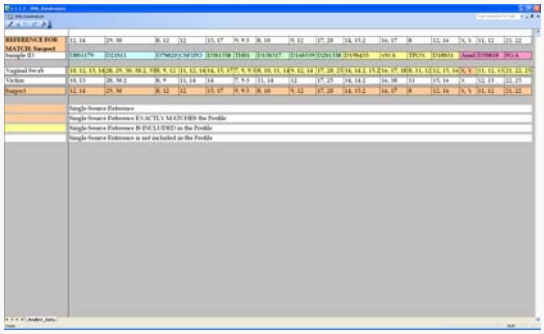
QA Check



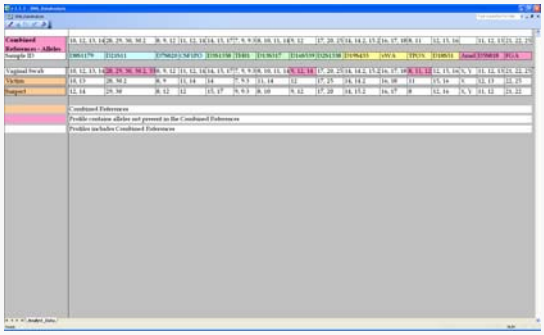
Samples View



Matching View

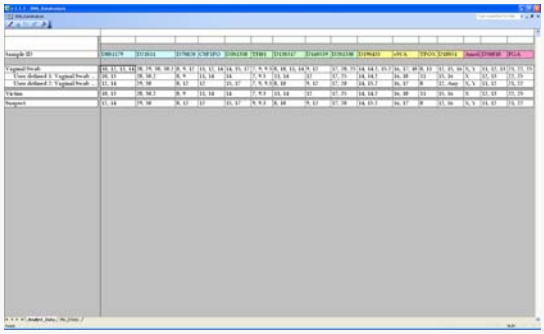


Foreign Allele View

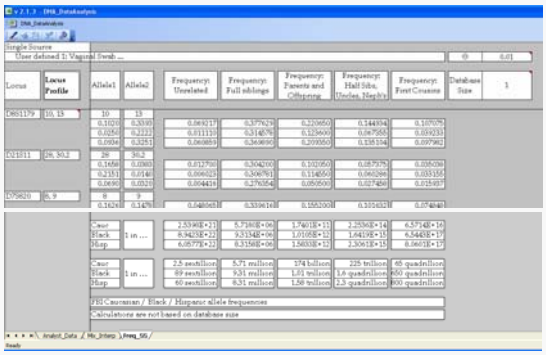


Mixture Interpretation Tool

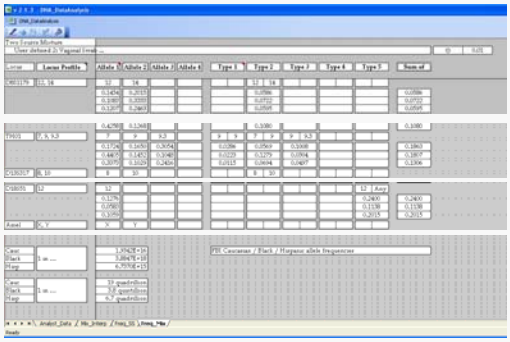
User Deconvoluted Profiles Added



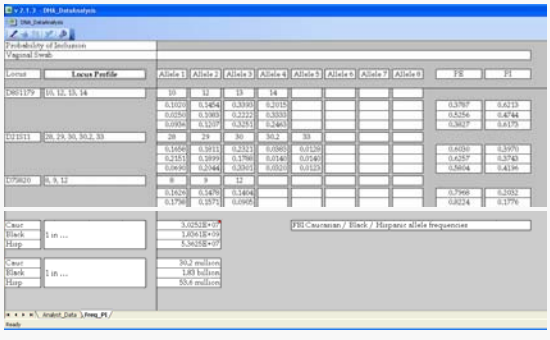
Single Source Stats



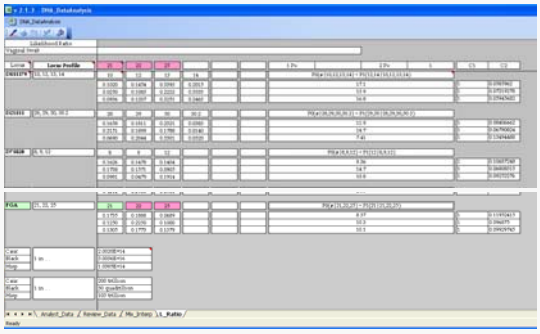
Mixture Stats



CPI Stats



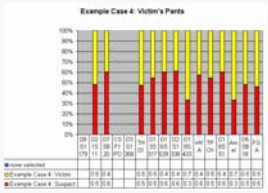
LR Stats



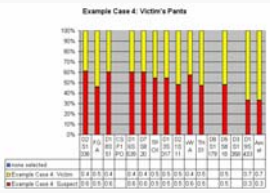
Male + Female: 1:1, 1:2, 1:3, 1:10

4 stains from one Case: Suspect + Victim

Plotting by locus size can help with degraded DNA

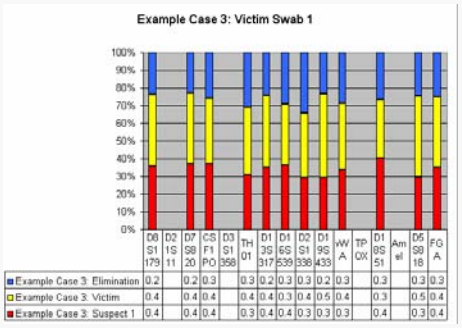


Identifiler Order

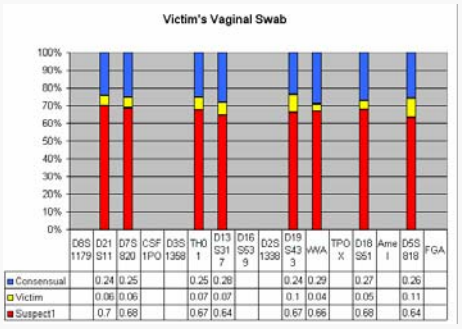


Large to small order

3-Contributors



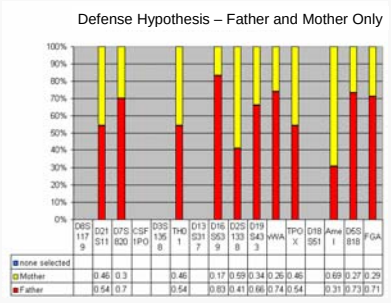
3-Contributors



Hypothetical case

- 14 year old girl accuses Dad of raping her
- Semen stain from parent's bed
- All alleles from profiles of Dad, Mom, and Victim found in stain
- Prosecution hypothesis: the evidence is from the Father + Mother + Daughter
- Defense hypothesis: the evidence is from the Father + Mother
  - Of course the daughter's alleles – or any child's - are included
  - There is no reason to believe the daughter contributed to the stain

Hypothetical case

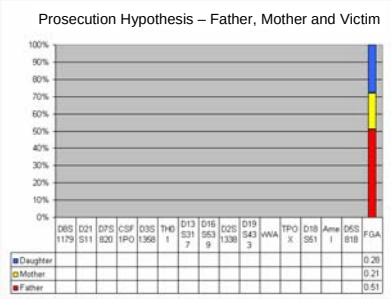


- Assuming just the Father + Mother, the proportions vary greatly…

Hypothetical case

- … and the Father + Mother hypothesis only supported at 10 loci …
- … inclusion is expected at every locus

Hypothetical case

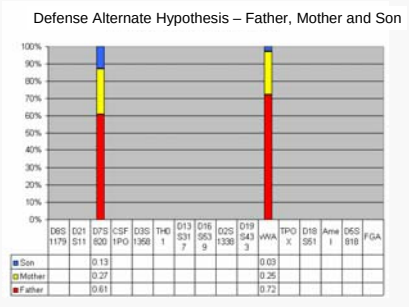


- Although the Father + Mother + Daughter chart may not appear to support the prosecution's hypothesis …

Hypothetical case

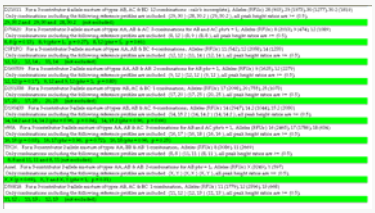
- ... calculations support inclusion of the Father + Mother + Daughter at every locus
- Remember: ratios and proportions can not be calculated for every scenario

Hypothetical case



If evaluating an alternate hypothesis: Father + Mother + Son

Hypothetical case



- … the hypothesis may be supported at 9 loci …
- … but inclusion is expected at every locus

Thanks to…

- Tom Overson
  - USACIL (Ret)
- USACIL DNA Examiners
- John Butler, PhD
  - NIST
- Rhonda Roby, MPH, MS
  - NIJ Consultant

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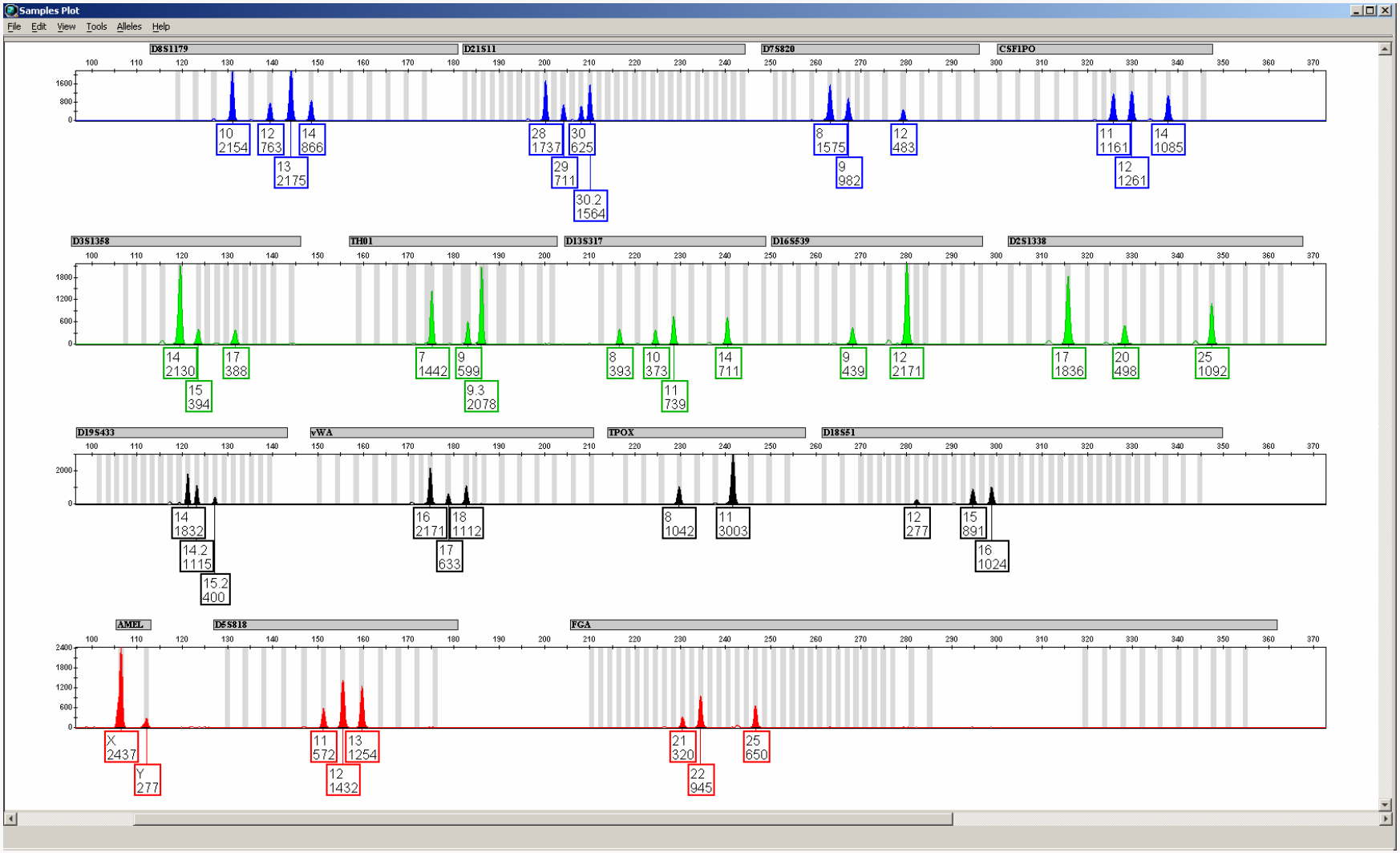
Example


Samples Plot
File Edit View Tools Alleles Help
D8S1179
D21S11
D7S820
CSF1PO
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12 763
13 2175
14 866
28 1737
29 711
30.2 1564
30 625
8 1575
9 982
12 483
11 1161
12 1261
14 1085
D3S1358
TH01
D13S317
D16S539
D2S1338
14 2130
15 394
17 388
7 1442
9 599
9.3 2078
8 393
10 373
11 739
14 711
9 439
12 2171
17 1836
20 498
25 1092
D19S433
vWA
TPOX
D18S51
14 1832
14.2 1115
15.2 400
16 2171
17 633
18 1112
8 1042
11 3003
12 277
15 891
16 1024
AMEL
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FGA
X 2437
Y 277
11 572
12 1432
13 1254
21 320
22 945
25 650