EDNAP Minutes - 20 April 2014 - Tbilisi Doc: Minutes-EDNAP-Tbilisi-4041.docx

EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

Tbilisi, Georgia

22 April 2014

Host: Ucha Margvelashvili. Chairman: Niels Morling.

A list of participants is attached.

Welcome

Head of The Forensic Biology (DNA) Department Nino Kochiashvili welcomed members to Tbilisi.

Update on exercises

mRNA exercise no 6 and 7 Cordula Haas Cordula Haas has received data from the laboratories participating in exercise 6 and will prepare a manuscript. Cordula Haas offered to organise a collaborative exercise concerning quantification of mRNA. Cordula Haas will discuss the possibility of collaboration with the group of Manfred Kayser and will at the next meeting present a plan for a collaborative EDNAP exercise during the winter 2014/2015 (presentation attached).

The IrisPlex exercise on genetic prediction of eye colour The manuscript is in press in FSI Genetics.

EDNAP ancestry informative marker exercise

Cordula Haas and Walther Parson reported on the status of the Ancestry Informative Marker (AIM) exercise that was very well organized by Christopher Philips and his team at the University of Santiago de Compostela (USC). Twenty-one laboratories received 6 samples (5 controls from different continents and a DNA mixture of two donors with European and East Asian ancestries). Laboratories were also supplied with primer sets for 46 Indel polymorphisms (46-plex) and 34 SNP loci (34-plex). The preliminary results were based on data submissions of 15 laboratories for the 46-plex and 11 laboratories for the 34-plex. Concordance rates were very high, 99.5% for the 46-plex and 96.85% for the 34-plex. USC is providing a web based resource (Snipper) to interpret AIM results. About 50% of the participants used Snipper to infer the ancestry of the individuals investigated resulting in correct ancestry inferences in all cases. The members felt that the exercise should be published and encouraged Chris Phillips to write a draft that can be circulated to the members. (presentation attached).

Updates from other groups

Interpol

Richard Scheithaur

Richard Scheithaur gave a short summary of the DNA activities of Interpol.

Cordula Haas Walther Parson

Niels Morling

Euroforgen: Challenges of interpreting the relevance of trace-DNA

Peter Gill introduced some thoughts and examples from a book in press 'P. Gill: Misleading DNA evidence (reasons for miscarriages of justice' that received funding from the European Union under the grant agreement concerning EUROFORGEN-NoE (presentation attached).

ENFSI

Roman Rhadil gave a short update on the activities of the DNA Working Group of ENFSI. The DNA Working Group was awarded the ENFSI prize for the best working group.

Euroforgen-NoE

Peter Schneider gave an update concerning the project (presentation attached).

EMPOP

Walther Parson reported on the developments of the mtDNA database EMPOP. The Innsbruck team is in the process of re-programming the database and the website. There will be a couple of new features including a more user-friendly tabular summary of query matches, The implementation of a new haplogroup estimation software (EMMA, Röck et al 2013), graphical maps to highlight query matches and haplogroup distribution and Haplogroup browser and a software to better understand and visualize the mitochondrial phylogenetic tree. The website is currently tested externally. Feedback will be evaluated and changes incorporated within the next months (presentation attached).

ENFSI - Evaluative reports in forensic science

Tascha Hicks introduced the draft of the Monopoly project 'ENFSI standard for the formulation of evaluative reports in forensic science'. Members can comment on http://tinyurl.com/ENFSIM1 (presentation, letter and draft attached).

AFSN and HAS (Sout East Asia)

Christopher Syn gave an overview of forensic science/genetics in Southeast Asia and Singapore (presentation attached).

LRmix update and searching

DNA databases with complex mixtures Peter Gill Peter Gill gave an update concerning the project (presentation attached).

ISFG Software Commission

Peter Schneider informed the members that the ISFG has established a commission validation of forensic genetic software with Mike Coble as chairman. June Guiness informed members that the Forensic Science Regulator has started work on the same issue. The possibility of collaboration will be explored.

EDNAP web site update (www.isfg.org/EDNAP)

Members are encouraged to visit the website. Suggestions are welcome.

Future activities

Please see the mRNA and AIMs exercise above.

Next meeting Niels Morling

The next EDNAP meeting will be held on 19 November 2014 at the Institute of Legal Medicine in Zürich and will be organised by Cordula Haas and colleagues. On 18 November 2014, the Steering Group of the DNA Working Group of ENFSI will meet.

Peter Schneider

Peter Schneider

Niels Morling

Christopher Syn

Tascha Hicks

Peter Gill

Roman Hradil

Peter Schneider

Walther Parson

Any other business

Cordula Haas mentioned that the EDNAP exercises are very important and encouraged members to suggest and organise new exercises.

Closing of the meeting

The meeting closed with sincere thanks to Ucha Margvelashvili, Nino Kochiashvili and their colleagues at the laboratory in Tbilisi.

Amendment

Report from EDNAP to ENFSI Niels Morling At the meeting of the DNA Working Group of ENFSI, Niels Morling reported from the EDNAP meeting (presentation attached).

Attachments are found at the EDNAP website http://www.isfg.org/EDNAP/Meetings:

- List of participants
- Presentations
 - o Cordula Haas: mRNA exercise
 - $\circ~$ Chris Phillips, Cordula Haas and Walther Parson: Ancestry marker exercise
 - Peter Gill: Challenges of interpreting the relevance of trace-DNA
 - Peter Schneider: EUROFORGEN-NoE report
 - Walther Parson: EMPOP report
 - \circ Hicks: ENFSI Evaluative reports
 - ENFSI draft of evaluation recommendations
 - \circ ENFSI board member letter
 - o Chris Syn: Forensic science/genetics in Southeast Asia
 - Peter Gill: LRmix update.
 - Niels Morling: Report from EDNAP to ENFSI.

Niels Morling

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EDNAP mRNA profiling exercise 7

Cordula Haas / Erin Hanson / Jack Ballantyne

22. April 2014, Tiflis





EDNAP mRNA profiling exercise 6

 \rightarrow Manuscript in preparation...







human mRNA quant assay

mRNA profiling workflow:

- RNA extraction
- DNase treatment (TURBO DNA-free kit)
- Optional: Quant-iT RiboGreen RNA kit / fluorescence microplate reader or Quant-iT RNA assay kit / Qubit
- Reverse transcription (RT)
- body fluid specific PCR-multiplex
- CE

 \rightarrow a major issue is cross contamination





human mRNA quant assay

- developed by Jack Ballantynes group
- Housekeeping gene
- qPCR assay
- TaqMan MGB probe
- qPCR standard





human mRNA quant assay: human specificity





Agarose gel electrophoresis to test specificity for human RNA

(A) PCR products of primate blood RNA samples, with human RNA as positive control. PM1-Rhesus monkey; PM2-Spider monkey; PM3-Black howler monkey; PM4-Brown lemur; PM5-African green monkey; PM6-Baboon; PM7-Cynomolgous monkey; PM8-Spot nosed guenon; PM9-Pig tailed macaque; PM10-Chimpanzee;

(B)) PCR products of non-primate blood RNA samples, with human RNA as positive control. Mouse-AM1; Duck-AM2; Turtle-AM3; Opossum-AM4; Gopher Tortoise-AM5; Rabbit-AM6; Guinea pig-AM7; Alligator-AM8; Rooster-AM9; Frog-AM10; Calf-AM11; Cow-AM12; Dog-AM13; Cat-AM14; Horse-AM15; Deer-AM16; Pig-AM17; Goat-AM18; Sheep-AM19; Patagonian Cavy-AM20.

(-), RT negative control; (+), RT positive control; EB, extraction blank; RTB, reverse transcription blank; AmpB, amplification blank.

78 bp





human mRNA quant assay: abundance in body fluids



Abundance in human body fluid samples

Total RNA of 25 ng was reverse transcribed and 1/10 of the RT product was used for qPCR. $dCt = Ct^{RT(-)}-Ct^{RT(+)}$





human mRNA quant assay: sensitivity

Sensitivity of qPCR assays in body fluid samples

	Ct	Ct	Ct	Ct	Ct			
qPCR (total RNA/well)	(50ng)	(10ng)	(2.5ng)	(0.5ng)	(0.1ng)	А	R ²	Equation of trendline
Blood-1	NA	19.16	20.76	23.04	25.41	-3.145	0.997	y = -3.1449x + 25.314
Blood-2	NA	18.89	20.35	22.58	25.31	-3.218	0.990	y = -3.2182x + 25.079
Blood-3	NA	19.2	20.11	22.56	25.05	-3.002	0.976	y = -3.0021x + 24.805
Blood-4	NA	18.83	20.2	22.55	25.04	-3.142	0.992	y = -3.1418x + 24.873
Saliva-1	NA	21.3	23.39	26.41	28.03	-3.460	0.987	y = -3.4603x + 28.327
Saliva-2	17.66	19.57	21.26	24.16	26.09	-3.204	0.994	y = -3.2036x + 26.102
Semen-1	NA	25.22	26.55	28.77	31.15	-2.996	0.992	y = -2.9962x + 30.991
Semen-2	NA	24.46	25.81	28.11	30.26	-2.948	0.995	y = -2.9481x + 30.179
Semen-3	NA	22.04	23.39	25.69	28.12	-3.076	0.992	y = -3.0757x + 27.96
Vaginal Secretion-1	NA	25.86	27.47	30.05	31.92	-3.102	0.996	y = -3.1015x + 32.002
Vaginal Secretion-2	21.31	24.22	25.73	27.61	29.82	-3.049	0.992	y = -3.0486x + 29.882
Vaginal Secretion-3	16.88	19	20.52	22.65	25.03	-2.981	0.998	y = -2.9812x + 24.868
Mean	18.62	21.48	22.96	25.35	27.60	-3.110	0.992	
SD	2.36	2.76	2.79	2.78	2.62	0.141	0.006	

NA, not available for the original low concentration of RNA extracts. Ct, Cycle threshold; A, slope; R, coefficient of correlation; SD, standard deviation.





human mRNA quant assay

3 approaches:

- 1. 25 ng RNA into RT
- 2. RiboGreen dependent quant: 25 ng RNA into RT, qPCR \rightarrow copy numbers
- 3. RiboGreen independent quant: 2 ul RNA into RT, qPCR \rightarrow copy numbers
- \rightarrow which is the best approach?
- \rightarrow define the optimal copy number for each body fluid





human mRNA quant assay

mRNA profiling workflow:

- RNA extraction
- DNase treatment (TURBO DNA-free kit)
- Quant-iT RiboGreen RNA kit / fluorescence microplate reader or Quant-iT RNA assay kit / Qubit
- Reverse transcription (RT)
- human mRNA quant assay \rightarrow copy numbers
- optimal copy number input into RT
- body fluid specific PCR-multiplex
- CE





human mRNA quant assay

- \rightarrow other mRNA quant assay (Zubakov, Kayser)?
- → Suggestion for a collaborative exercise on mRNA quantification (EDNAP mRNA exercise 7) at next EDNAP meeting

presumably November 2014 in Zürich









Thank you for your attention!

Cordula Haas / Erin Hanson / Jack Ballantyne

22. April 2014, Tiflis

	10	Chr	Position	Gene	Functional Class
1	s2307666	11	64729920	C11orf85	
2	rs1610863	16	6551830	RSFOX1	
3	rs16635	6	99789775	FAXC	
X	rs1610965	5	79746093	ZFYVE16	
5	rs35451359	18	45110983	-	
-	rs140837	6	3708907	-	
7	rs1160893	2	2.25E+08	WDFY1	
8	+\$2308203	2	1.09E+08	RANBP2	
9	rs33974167	8	87813725	3	
10	rs1160852	6	1.37E+08	IL20RA	
11	rs1610884	5	56122323	MAP3K1	
12	rs2067280	5	89818959	LYSMD3	
13	rs2308067	7	1.27E+08	SND1	
14	rs4183	3	3192524	CRBN	
13	+\$3054057	15	86010538	AKAP13	
18	rs2307840	1	36099090	PSMB2	
17	+\$60612424	6	84017510	ME1	
18	133033053	14	42554496		
19	rs16384	22	42045009	XRCC6	
20	rs34611875	18	67623917	CD226	
21	rs1610859	5	1.28E+08	SLC27A6	2
22	rs3045215	1	2.35E+08	IRF28P2	
23	rs25621	6	1.45+08	-	
24	+\$2307832	1	55590788	USP24	
25	rs16343	4	17635560	FAM184B	
26	rs3031979	8	73501951	KCNB2	
27	+\$34122827	13	63778778	4	
28	rs133052	22	41042364		
29	rs6490	12	1.08E+08	PRDM4	
30	rs4181	2	42577803	COX7A2L	
31	+\$3030826	6	67176774	7	
32	rs140708	6	1.71E+08		
33	rs1611026	5	82545545	XRCC4	
34	rs16438	20	25278470	PYGB	
35	rs2308161	10	69800909	HERC4	
36	rs16687	7	B3887878	-	
37	rs2307998	5	7814345	ADCY2	
38	+\$2307803	3	1.09E+08	-	
39	rs2307930	6	84476378	4	
40	rs25630	6	14734341	-	
Δt	rs2307582	1	2.48E+08	OR2G3	
62	ts2307922	1	39896964	MACFI	
43	rs11267926	15	45526069	1	
44	ts25584	12	1.12E+08	ACAD10	
45	rs2307799	5	70828419	BDP1	
Δį,	1334541393	20	30701405	TM95F4	
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- 3	1 1 46	114	17.64°		Sugar and Press				
	10	Chr	Position	Gene	Functional Class				
r	rs10141763	14	36170607	RALGAPA1	Intronic				
2	rs1024116	18	75432386	-					
3	rs10843344	12	29369871	F.4					
4	rs12913832	15	28365618	HERC2	gramotice for OCA2				
5	rs1321333	20	38849642		and the second				
ŝ,	rs1335873	13	20901724	-					
7	rs1426654	15	48426484	SLC24AS	coding THR 111 ALA.				
ŝ.	151498444	. 1	1.69E+08	-					
ġ.	rs1573020	1	36768200	-					
ġ.	rs16891982	5	33951693	SLC45A2	coding PHE 374 LEU.				
1	rs182549	2	1.37E+08	MCM6	promotor for LCT				
2	rs1886510	13	22374700						
3	rs1978806	10	34755348	PARD3	intronic				
4	132026721	4	1.59E+08	-					
5	rs2040411	22	47836412	+ -					
£	1\$2065160	1	2.05E+08	-					
7	rs2065982	13	34864240						
R	rs2303798	19	42410331	ARHGEF1	intronic				
9	rs2304925	17	75551667	10000000000000000000000000000000000000	1 MARKAN P				
Q.	rs239031	21	17710424	-					
1	rs2572307	21	25672460	-					
ź	132814778	1	1.59E+08	DARC	S' UTR (creates null)				
3	rs3785181	16	90105333	GAS8	Intronic				
à.	rs3827760	2	1.15+08	EDAR	coding VAL 370 ALA				
5	rs4540055	4	38803255	TLR1	intronic				
ŝ	rs5030240	11	32424389	WT1	intronic				
7	rs5997008	22	26350103	MYO18B	intronic				
0	rs722098	21	16685598	-					
5	rs730570	14	1.01E+08	-					
Ö.	rs773658	12	56603834	RNF41	intronic				
1	rs7897550	10	17064992	CUBN	intronic				
ż	rs881929	16	31079371	ZNF668	intronic				
3	rs896788	2	7149155	RNF144A	intronic				
a.	rs917118	7	4457003	-					

32 of 46 Indels are in coding regions but their function or effect on these gene's behaviour is not known yet USC have retained this EUR-informative SNP but it defines the lactase persistence trait - which is *not* an externally visible characteristic

Ricky Ansell asked in Bratislava about the coding status of the 46 Indels and 34 SNPs we proposed to use for this exercise



Opted for the simplest format possible

- Five cell-line DNA preps each from a different continental region
- Used the above as controls for the fullest range of alleles and compared to European 9947a DNA (where a sizable proportion of alleles are absent)
- Additional mixed DNA of E ASN and EUR Kings College donors at 3:1
- Tasks: type 9947a and six DNAs, assign ancestry or identify as mixture



Opted for the simplest format possible

U SC UNIVERSIDADE DE SANTIAGO DE COMPOSTELA

- Five cell-line DNA preps each from a different continental region
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• Chance occurrences were: presence of rare third allele in Indel rs25584 of DNA-C that all labs successfully identified; three alleles in the mixed DNA for tri-allelic SNP rs5030240 that four labs successfully recorded

• On the whole, most labs found the SNP genotyping with SNaPshot a challenge, and Indel genotyping relatively straightforward - for most labs this was their first experience with Indels

• All labs (so far) have correctly assigned ancestries and identified the mixture

20 EDNAP labs took part - Kayser lab at Erasmus also invited



Catherine McGovern / SallyAnn Harbison, NZ Katherine Gettings / Kevin Kiesler / Pete Vallone, US Erin Hanson / Jack Ballantine, US Mayra Eduardoff / Walter Parson, OS Fabrice Noël, BE VLC Subramanyam / Manfred Kayser, NE Francesca Brisighelli / Vince Pascali, IT Regine Banemann / Ingo Bastisch, DE David Ballard / Denise Syndercombe Court, UK Andreas Tillmar / Gunilla Holmlund, SWE Cordula Haas / Walther Bär, SWI . Ana Bento / Maria Joao Porto, PT Helle Smidt / Neils Morling, DK Mike Burrington / Geraldine O'Donnell, IE Vlastimil Stenzl, CZ Karin Resto / Per Hof-Olsen, NO Joyce Harteveld / Titia Sijen, NE Theresa Gross / Peter Schneider, DE Runa Daniel / Roland van Oorschot, AUS Jennifer Templeton / Adrian Linacre, AUS Martina Turanska, SK

• Have we identified the contributors correctly ?

• Fabrice, Valstimil, Martina do you have a 2nd person to nominate ?

3/21 labs have long-standing problems, another 3 sent data late



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Florida has a problem with controls A-D despite two dispatches and DNA being purified cell-line preps. May have lower than expected quantitations but all worked fine elsewhere.

Linköping lab using 3500/POP-7 so need more primers (sent 1/4/14) to optimize their CE.

Prague lab still waiting for a long bureaucratic process to approve purchase of consumables.

These labs sent data after the deadline so it has not been analyzed in depth yet.

More importantly, four labs elected to genotype Indels only, so the 34plex AIM-SNP data analysed so far is from 11/21 labs.



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A complicating factor was some use of POP-7/3500 detectors - here USC is unable to help predict SNP mobility at the lower size range



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Erin Hanson / Jack Ballantine, US

Mayra Eduardoff / Walter Parson, OS Fabrice Noël, BE VLC Subramanyam / Manfred Kayser, NE Francesca Brisighelli / Vince Pascali, IT Regine Banemann / Ingo Bastisch, DE David Ballard / Denise Syndercombe Court, UK Andreas Tillmar / Gunilla Holmlund, SWE

Cordula Haas / Walther Bär, SWI

Ana Bento / Maria Joao Porto, PT

Helle Smidt / Neils Morling, DK

Mike Burrington / Geraldine O'Donnell, IE

Vlastimil Stenzl, CZ

Karin Resto / Per Hof-Olsen, NO

Joyce Harteveld / Titia Sijen, NE

Theresa Gross / Peter Schneider, DE

Runa Daniel / Roland van Oorschot, AUS

Jennifer Templeton / Adrian Linacre, AUS

Martina Turanska, SK

ELSEVIER





Forensic Science International: Genetics 1 (2007) 273-280

Inferring ancestral origin using a single multiplex assay of ancestry-informative marker SNPs

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POP-6

POP-4



Revision of the SNPforID 34-plex forensic ancestry test: Assay enhancements, standard reference sample genotypes and extended population studies

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"POP-4 is now in increasing use and several SBE primer rearrangements we report here anticipate the discontinuation of AB POP-6 CE polymer used in the original 34-plex assay development"



SNP genotyping with SNaPshot

SNPs gave these no calls / wrong calls for control DNAs A-E:



Lab :	5	6	9	11	12	13	14	16	18	19	20	NO CALL	WRONG	TOTAL	AVERAGE	Code
rs10141763	0/0	0/0	0/0	0/3	0/0	0/1	0/0	0/0	0/0	0/0	0/0	0	4	4	0.8	P11
rs1024116	0/0	0/0	0/0	0/2	0/0	0/0	0/0	1/0	0/0	0/0	0/0	1	2	3	0.6	A29
rs10843344	0 / 1	0/0	0/0	0/1	0/0	0/1	0/0	0/0	0/0	0/0	4/0	4	3	7	1.4	P06
rs12913832	0 / 1	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	1/0	1	1	2	0.4	P08
rs1321333	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	1/0	1	0	1	0.2	P03
rs1335873	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	A52
rs1426654	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P22
rs1498444	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/1	0/0	2/0	2	1	3	0.6	P21
rs1573020	0 / 1	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	1	1	0.2	P13
rs16891982	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P25
rs182549	0/0	0/0	0/0	0/4	0/0	1/0	0/0	0/0	4/0	0/0	0/0	5	4	9	1.8	P12
rs1886510	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	A13
rs1978806	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/1	0/0	0/0	1/0	1	1	2	0.4	P09
rs2026721	0/0	0/0	0/0	0/0	0/0	1/0	0/0	0/0	0/0	0/0	1/0	2	0	2	0.4	P23
rs2040411	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	A40
rs2065160	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P15
rs2065982	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P18
rs2303798	0/0	0/0	0/0	0/0	0/0	1/1	0/0	0/0	0/0	0/0	0/0	1	1	2	0.4	P17
rs2304925	0/0	0/1	0/1	0/0	0/0	0/2	0/0	1/0	0/1	0/0	1/1	2	6	8	1.6	P01
rs239031	1/2	0/1	0/0	0/2	0/1	5/0	0/0	0/0	0/0	0/2	2/2	8	10	18	3.6	P07
rs2572307	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P16
rs2814778	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P04
rs3785181	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P19
rs3827760	0/0	1/0	0/0	0/0	2/0	5/0	0/0	0/0	0/0	0/0	1/0	9	0	9	1.8	P28
rs4540055	0 / 1	0/0	0/0	0/0	0/0	2/1	0/0	0/0	0/0	0/0	0/0	2	2	4	0.8	P24
rs5030240	0/0	0/2	0/0	0/1	0/0	2/0	0/0	1/0	0/1	1/0	1/0	5	4	9	1.8	P27
rs5997008	0/0	1/1	0/0	0/0	0/1	1/0	0/0	0/0	0/0	0/0	1/0	3	2	5	1	P02
rs722098	1/1	0/0	0/0	0/0	0/0	0/2	0/0	0/0	0/0	0/0	1/0	2	3	5	1	A21
rs730570	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P26
rs773658	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P10
rs7897550	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P05
rs881929	0/0	1/0	0/0	0/0	0/0	1/1	0/0	0/0	2/0	0/0	5/0	9	1	10	2	P20
rs896788	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P14
rs917118	0/1	0/0	0/0	0/3	0/0	0/1	0/0	0/0	0/0	0/0	1/0	1	5	6	1.2	A07
NO CALL	2	3	0	0	2	19	0	3	6	1	23					
WRONG CALL	8	5	1	16	2	10	0	1	3	2	3					
TOTAL	10	8	1	16	4	29	0	4	9	3	26					

The overall no call / wrong call rate was 3.15% for control DNAs A-E = 96.85% concordance



Lab :	5	6	9	11	12	13	14	16	18	19	20	NO CALL	WRONG	TOTAL	AVERAGE	Code			
rs10141763	0/0	0/0	0/0	0/3	0/0	0/1	0/0	0/0	0/0	0/0	0/0	0	4	4	0.8	P11			
rs1024116	0/0	0/0	0/0	0/2	0/0	0/0	0/0	1/0	0/0	0/0	0/0	1	2	3	0.6	A29			
rs10843344	0 / 1	0/0	0/0	0/1	0/0	0/1	0/0	0/0	0/0	0/0	4/0	4	3	7	1.4	P06			
rs12913832	0 / 1	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	1/0	1	1	2	0.4	P08			
rs1321333	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	1/0	1	0	1	0.2	P03			
rs1335873	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	A52			
rs1426654	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P22			
rs1498444	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/1	0/0	2/0	2	1	3	0.6	P21			
rs1573020	0 / 1	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	1	1	0.2	P13			
rs16891982	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P25			
rs182549	0/0	0/0	0/0	0/4	0/0	1/0	0/0	0/0	4/0	0/0	0/0	5	4	9	1.8	P12			
rs1886510	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	A13			
rs1978806	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/1	0/0	0/0	1/0	1	1	2	0.4	P09			
rs2026721	0/0	0/0	0/0	0/0	0/0	1/0	0/0	0/0	0/0	0/0	1/0	2	0	2	0.4	P23			
rs2040411	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	A40			
rs2065160	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P15			
rs2065982	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P18			
rs2303798	0/0	0/0	0/0	0/0	0/0	1/1	0/0	0/0	0/0	0/0	0/0	1	1	2	0.4	P17			
rs2304925	0/0	0/1	0/1	0/0	0/0	0/2	0/0	1/0	0/1	0/0	1/1	2	6	8	1.6	P01			
rs239031	1/2	0 / 1	0/0	0/2	0/1	5/0	0/0	0/0	0/0	0/2	2/2	8	10	18	3.6	P07			
rs2572307	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P16			
rs2814778	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P04			
rs3785181	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P19			
rs3827760	0/0	1/0	0/0	0/0	2/0	5/0	0/0	0/0	0/0	0/0	1/0	9	0	9	1.8	P28			
rs4540055	0/1	0/0	0/0	0/0	0/0	2/1	0/0	0/0	0/0	0/0	0/0	2	2	4	0.8	P24			
rs5030240	0/0	0/2	0/0	0/1	0/0	2/0	0/0	1/0	0/1	1/0	1/0	5	4	9	1.8	P27			
rs5997008	0/0	1/1	0/0	0/0	0/1	1/0	0/0	0/0	0/0	0/0	1/0	3	2	5	1	P02			
rs722098	1/1	0/0	0/0	0/0	0/0	0/2	0/0	0/0	0/0	0/0	1/0	2	3	5	1	A21			
rs730570	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P26			
rs773658	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P10			
rs7897550	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P05			
rs881929	0 / 0	1/0	0/0	0/0	0/0	1/1	0/0	0/0	2/0	0/0	5/0	9	1	10	2	P20			
rs896788	0 / 0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P14			
rs917118	0/1	0/0	0/0	0/3	0/0	0/1	0/0	0/0	0/0	0/0	1/0	1	5	6	1.2	A07			
NO CALL	2	3	0	0	2	19	0	3	6	1	23	overall no call rate of 3.15%							
WRONG CALL	8	5	1	16	2	10	0	1	3	2	3 🗖	overall genotype discordancy of 3,15%							
TOTAL	10	8	1	16	4	29	0	4	9	3	26		901007						

Table records the discrepancies amongst 1,870 SNP genotypes

Some 34plex SNP components have known issues, notably P01, P06-7 and P28



Lab :	5	6	9	11	12	13	14	16	18	19	20	NO CALL	WRONG	TOTAL	AVERAGE	Code	
rs10141763	0/0	0/0	0/0	0/3	0/0	0/1	0/0	0/0	0/0	0/0	0/0	0	4	4	0.8	P11	
rs1024116	0/0	0/0	0/0	0/2	0/0	0/0	0/0	1/0	0/0	0/0	0/0	1	2	3	0.6	A29	
rs10843344	0/1	0/0	0/0	0/1	0/0	0/1	0/0	0/0	0/0	0/0	4 / 0	4	3	7	1.4	P06	P06-P07 peak
rs12913832	0/1	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	1/0	1	1	2	0.4	P08	pair very close
rs1321333	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	1/0	1	0	1	0.2	P03	
rs1335873	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	A52	
rs1426654	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P22	
rs1498444	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/1	0/0	2/0	2	1	3	0.6	P21	
rs1573020	0 / 1	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	1	1	0.2	P13	
rs16891982	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P25	
rs182549	0/0	0/0	0/0	0/4	0/0	1/0	0/0	0/0	4 / 0	0/0	0/0	5	4	9	1.8	P12	Iow peaks
rs1886510	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	A13	
rs1978806	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/1	0/0	0/0	1/0	1	1	2	0.4	P09	
rs2026721	0/0	0/0	0/0	0/0	0/0	1/0	0/0	0/0	0/0	0/0	1/0	2	0	2	0.4	P23	
rs2040411	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	A40	
rs2065160	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P15	
rs2065982	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P18	G-like artifactual
rs2303798	0/0	0/0	0/0	0/0	0/0	1/1	0/0	0/0	0/0	0/0	0/0	1	1	2	0.4	P17	neak in NTC
rs2304925	0/0	0/1	0/1	0/0	0/0	0/2	0/0	1/0	0/1	0/0	1/1	2	6	8	1.6	P01	
rs239031	1/2	0/1	0/0	0/2	0/1	5/0	0/0	0/0	0/0	0/2	2/2	8	10	18	3.6	P07	P06-P07 peak
rs2572307	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P16	
rs2814778	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P04	
rs3785181	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P19	
rs3827760	0/0	1/0	0/0	0/0	2/0	5/0	0/0	0/0	0/0	0/0	1/0	9	0	9	1.8	P28	A allele very low
rs4540055	0/1	0/0	0/0	0/0	0/0	2/1	0/0	0/0	0/0	0/0	0/0	2	2	4	0.8	P24	
rs5030240	0/0	0/2	0/0	0/1	0/0	2/0	0/0	1/0	0/1	1/0	1/0	5	4	9	1.8	P27	tri-allelic SNP
rs5997008	0/0	1/1	0/0	0/0	0/1	1/0	0/0	0/0	0/0	0/0	1/0	3	2	5	1	P02	
rs722098	1/1	0/0	0/0	0/0	0/0	0/2	0/0	0/0	0/0	0/0	1/0	2	3	5	1	A21	
rs730570	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P26	
rs773658	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P10	
rs7897550	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P05	
rs881929	0/0	1/0	0/0	0/0	0/0	1/1	0/0	0/0	2/0	0/0	5/0	9	1	10	2	P20	low peaks
rs896788	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	0	P14	
rs917118	0/1	0/0	0/0	0/3	0/0	0/1	0/0	0/0	0/0	0/0	1/0	1	5	6	1.2	A07	
NO CALL	2	3	0	0	2	19	0	3	6	1	23						
WRONG CALL	8	5	1	16	2	10	0	1	3	2	3						
TOTAL	10	8	1	16	4	29	0	4	9	3	26						

Some 34plex SNP components have known issues, notably P01, P06-P07 and P28





Some 34plex SNP components have known issues, notably P01, P06-7 and P28







Indel genotyping with a direct PCR-to-CE system

Indels: 3 no calls and 3 wrong calls (each in same lab) = >99.5% Concordance



Lab	5	6	9	10	11	12	13	14	15	16	17	18	21	19	21	NO CALL	WRONG	TOTAL	DE COMPOS
rs2307666	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1470
rs1610863	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-777
rs16635	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-196
rs1610965	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-881
rs35451359	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-3122
rs140837	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-548
rs1160893	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-659
rs2308203	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2011
rs33974167	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2929
rs1160852	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-593
rs1610884	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-798
rs2067280	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1193
rs2308067	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0 / 0	0/0	0/0	0/0	0/0	0	0	0	MID-1871
rs4183	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-17
rs3054057	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2538
rs2307840	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1644
rs60612424	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-3854
rs3033053	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2275
rs16384	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-94
rs34611875	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-3072
rs1610859	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-772
rs3045215	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2313
rs25621	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-397
rs2307832	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1636
rs16343	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-51
rs3031979	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2431
rs34122827	0/0	0/0	0/0	0/0	0/0	0/2	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	2	2	MID-2264
rs133052	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2256
rs6490	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-128
rs4181	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-15
rs3030826	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2241
rs140708	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-419
rs1611026	0/0	0/0	0/0	0/0	0/0	0/1	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	1	1	MID-943
rs16438	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-159
rs2308161	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2005
rs10087	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-250
rs2307998	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1802
rs2307003	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1607
15200/900	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID 406
1520000	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	2/0	0	0	0 2	
re2207002	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	ۍ ۲	0	<u>ی</u>	MID 1706
152001922 re11067006	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID 2606
1311201920 re95594	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0 0	MID 260
re2207700	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID 1602
re3/5/1202	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID 2710



Ancestry inferences using Snipper



-10

-15

PC1 23.45%

-5

0

5

Bold values here and following indicate completely concordant genotyping for all 80 markers with each sample

1-2,456,273 times more likely E ASIA than AMERICA 2-27,212,309.9343 times more likely E ASIA than AMERICA **4-9,969,649.6410 times more likely E ASIA than AMERICA** 6-1,662,862.3788 times more likely E ASIA than AMERICA **7-9,969,649.6410 times more likely E ASIA than AMERICA** 8-1,915,104,447.7545 times more likely E ASIA than AMERICA **9-9,969,649.6410 times more likely E ASIA than AMERICA** 11-11,368,023.3185 times more likely E ASIA than AMERICA 13-24,639,071.1726 times more likely E ASIA than AMERICA **18-9,969,649.6410 times more likely E ASIA than AMERICA**

DNA-A



1-10,155,216,244,176,471,131,240,267,776.0000 times more likely EUROPE than E ASIA 2-10,155,216,244,176,471,131,240,267,776.0000 times more likely EUROPE than E ASIA 4-10,155,216,244,176,471,131,240,267,776.0000 times more likely EUROPE than E ASIA 6-10,155,216,244,176,471,131,240,267,776.0000 times more likely EUROPE than AMERICA 7-1,527,975,244,180,861,408,820,330,496.0000 times more likely EUROPE than AMERICA 8-16,420,864,767,598,781,007,921,152.0000 times more likely EUROPE than E ASIA 9-10,155,216,244,176,471,131,240,267,776.0000 times more likely EUROPE than E ASIA 11-10,155,216,244,176,471,131,240,267,776.0000 times more likely EUROPE than E ASIA 13-10,155,216,244,176,471,131,240,267,776.0000 times more likely EUROPE than E ASIA 20-299,423,442,787,246,318,685,257,728.0000 times more likely EUROPE than E ASIA

DNA-B



EUROPE



-10

-5

PC1 23.45%

E ASIA

0

5

1-217,912,560,580,145.3125 times more likely OCEANIA than E ASIA 2-378,212,377,874,323.0000 times more likely OCEANIA than E ASIA 4-378,212,377,874,323.0000 times more likely OCEANIA than E ASIA 6-99,975,472,816,742.1094 times more likely OCEANIA than E ASIA 7-11,166,496,196,613.1855 times more likely OCEANIA than E ASIA 8-17,508,652,075.2790 times more likely OCEANIA than E ASIA 9-378,212,377,874,323.0000 times more likely OCEANIA than E ASIA 11-59,143,806,943,454.8984 times more likely OCEANIA than E ASIA* 13-27,723,983,920,520.1211 times more likely OCEANIA than E ASIA 18-59,143,806,943,454.8984 times more likely OCEANIA than E ASIA* 20-43,806,880,061,280.2109 times more likely OCEANIA than E ASIA

DNA-C

* 34plex rs5030240 tri-allelic SNP = NN in both


-10

1-1,064,869,103,473.9652 times more likely AMERICA than E ASIA 2-**86,184,546,277,253.4375 times more likely AMERICA than E ASIA** 4-19,941,398,232,604.1523 times more likely AMERICA than E ASIA 6-182,558,772,642.2585 times more likely AMERICA than E ASIA 7-**86,184,546,277,253.4375 times more likely AMERICA than E ASIA** 8-575,144,413,429.2894 times more likely AMERICA than E ASIA 9-**86,184,546,277,253.4375 times more likely AMERICA than E ASIA** 11-10,653,650,518,249,948.0000 times more likely AMERICA than E ASIA 13-19,642,211,218,158.8125 times more likely AMERICA than E ASIA 18-20,290,522,073,158.6445 times more likely AMERICA than E ASIA

DNA-D



-5

E ASIA

0

5





E ASIA

0

5

1-7,231,484,739,385,767,674,048,692,591,992,730,812,416 times more likely AFRICA than OCEANIA 2-12,039,445,370,327,450,953,488,334,222,996,090,650,624 times more likely AFRICA than OCEANIA **4-261,782,596,050,744,346,488,932,985,848,702,133,338,112 times more likely AFRICA than OCEANIA** 6-466,008,526,513,015,244,257,093,658,959,937,536 times more likely AFRICA than OCEANIA 7-3,731,671,935,222,560,132,104,471,444,636,148,170,752 times more likely AFRICA than OCEANIA 8-6,809,261,373,339,948,161,372,782,888,352,743,424 times more likely AFRICA than OCEANIA **9-261,782,596,050,744,346,488,932,985,848,702,133,338,112 times more likely AFRICA than OCEANIA** 11-261,782,596,050,744,346,488,932,985,848,702,133,338,112 times more likely AFRICA than OCEANIA 13-24,420,193,029,865,759,751,127,191,414,805,507,342,336 times more likely AFRICA than OCEANIA 18-12,039,445,370,327,450,953,488,334,222,996,090,650,624 times more likely AFRICA than OCEANIA 20-534,890,820,487,177,742,550,759,925,126,994,591,744 times more likely AFRICA than OCEANIA DNAs A, C and D are much more closely positioned but can be better differentiated using just a three population PCA





The mixed-source DNA

DNA-F was a 3:1 mixture of a King's Chinese donor + David Ballard





1726 gave very low peaks for lab #20 = accounting for all three no calls

PCA also provides a simple way to infer the ancestry of the mixture contributors (using all recorded alleles to make the mid-cluster point)





Mixture 1:3 (EAS:EUR)



Summary points



• Results so far analyzed indicate SNP genotyping at this scale of multiplexing is difficult to get familiar with - but those labs already acquainted with the 34plex SNaPshot test produced concordant results for most samples

• Indels were readily adopted by all participants and gave near-complete concordance as well as clear signals of imbalance in the mixed DNA

All labs have reported correct ancestries for control DNAs A-E

• Since the USC Indel population pages are now online, is it appropriate to suggest we can generate population data in those EDNAP labs willing to contribute data from their lab collections? (USC can supply all primers needed)

forInDel		forInDel				forInDel USE Nome - SEARCH (keys) about (mport & bug (contact) group multiple populations into a population set to get information withins the set (additive results) distribute populations into different sets to get information between them all (comparative results) multiple of population sets 123±3 IPATIMUP-USC 48-plax (N=949)				
The second	INDEL	internal INDEL code	frequencies MIDDLE EAST (N=163)	CENTRAL-SOL	TH ASLA	IPATIMUP-USC 46-piex	AFRICA C. African Republic - Bake Prgmy D. R. of Congo - Mada Prgmy Renya - Bantu N. E.	C EUROPE C France - Masque C France - Prynch C Bally - Sercinsen	CENTRAL-SOUTH ASIA	Cambodia - Cambodian Cambodia - Cambodian Cambodia - Cambodian Cambodia - Cambodian Cambodia - Cambodian
	152307506 15166053	MID-1470 MID-777 MID-196	A: 0.546 a C: 0.454 c A: 0.322 a C: 0.678 c A: 0.494 a	A: 0.411 C: 0.589 A: 0.500 C: 0.500 A: 0.411	20		Avernade - Sav Naperia - Yandae Senegal - Mandenka South African Benta Amenica Brazi - Kermena Brazi - Sani	Hany - Fuecae Davy - from Bergame Ontway Blands - Occasian Russia - Russian Sussia Caccasa - Adyge HIDDLE EAST Aborn (Nam) - Nosabbe	Pacatan - Baruthy Pacatan - Hazara Pacatan - Kalana Pacatan - Kalana Pacatan - Pathan Pacatan - Pathan Pacatan - Sandhi	China - Han China - Hanhai China - Laha China - Miae China - Miae China - Nael
America Africa Europe Hiddle East Central-South Asia East Asia Oceania SPOMATE (2.1.1.) SECON VERSION 2012.133	rt1610965 rs3545135	MID-681 MID-3122	A: 0.506 A: 0.506 A: 0.196 C: 0.196 C: 0.052 A: 0.163 A: 0.163	A: 0.760 C: 0.240 A: 0.993 C: 0.007 A: 0.134	20		Colombia - Colombias Henco - Haya Henco - Pana	Streef (Carrel) - Drute Streef (Carrel) - Pelastinian Streef (Carrel) - Pelastinian Streef (Regev) - Decourt	C Norpenydie - Hefanestan New Ource - Ropuen	C China - Tai C China - Th C Soleria - Yakyi



Interpretation of complex DNA profiles: a review of recent progress and remaining challenges





1

Lrmix Exploratory approach

- probability of dropout is modelled across the entire range.
- Emphasis on the exploratory approach
- PrD often flat-lines (ie LR is relatively insensitive to PrD)





What about peak height balance?

- It is not true that we ignore this.
- We evaluate this if needed using Forensim Hbsimu() model
- The key difference here is that we do not incorporate the output into the LR model.
- This reinforces the 'exploratory principle' to interpret DNA profiles

Convergence between models

"Continuous models are superior to other models because they make use of all available data"

- Different models generate different numbers
- Ideally, all models should converge
- Modelling assumptions must be reasonable (and this is a crucial point)

Comparative study to illustrate the convergence principle (no drop out in this eg)



The exploratory approach



Interpretation process is an interaction of the expert with a statistical model



"these complex mixture profiles should be subjected to interpretation approaches to see if a true contributor is *appropriately associated* with the mixture and if noncontributors are *appropriately excluded*." John Butler

Why exploratory?

- The purpose is not to give a 'black-box' answer because there is no definitive answer
- All of the answers are conditional hence the function of the 'expert' is to explore the various possibilities, on behalf of the prosecution and defence.
- Some generalisations are possible
- The 'process' used to interpret complex DNA profiles is provided in this talk
- Consider a minor/minor(s) contributors in the following epg. We could regard this as a typical LTDNA profile

Step 1: examine the epg

- And Consider the case circumstances
- Is it a mixture?

EPG

Case circumstances:

≻Epithelial swab from female victim (V)

Sexual assault with two suspects under Hp (S1, S2)



Step 2: Explore the profile

- What kind of mixture is it?
- Choose from following:
 - Major/minor?
 - Even?
- Do we expect drop-out?
 - (compare with logistic regression)

Step 2: Explore the profile

Profile overview for case Case1_data

Alleles that appear in the replicates but not in the profiles

Select	Name	Replicate	Suspect1	Suspect2	Distinct Alleles
V	Epithelial				
V	D3S1358	14 16 17	16 17	15 17	4
1	VWA	16 <u>17</u> 18 19	16 18	18 19	4
1	D16S539	11 12 13 15	12 13	12 12	4
1	D2S1338	17 19 20	19 20	17 18	4
V	D8S1179	9 10 1314	9 13	13 13	4
~	D21S11	29 31 32	28 32	30 30	5
1	D18S51	12 <u>16</u>	12 15	12 20	4
1	D19S433	12 <u>14 15.2</u> 16	12 16	12 15	5
V	TH01	6 9.3	6 9.3	<mark>6 9.3</mark>	2
V	FGA	19 24 26	19 21	20 21	5

A typical low template profile showing PrD range relative to thresholds

Check the peak heights against logistic regression to work out if drop-out is expected



Change in philosophy

- With the old methods we had to 'filter' alleles and there were many restrictions about the kind of analysis that could be undertaken
- The new method can evaluate profiles without filtering alleles and are not restricted by numbers of contributors etc.
- Consequently, we are able to devise simple rules that can be followed to produce an LR.
- The questions shift towards "what are the propositions that should be considered"
- The role of the RO now becomes a *facilitator* of the court going discussion by following a logical process

Exploratory approach (case evaluation)

Profile overview for case Case1_data

Matching alleles in the replicate and Suspect1

Select	Name Replicate		Suspect1	Suspect2	Victim	Distinct Alleles
-	Epithelial					
रा	D3S1358	14 <u>16 17</u>	<u>16 17</u>	15 17	14 16	4
<u>-</u>	VWA	<u>16</u> 17 <u>18</u> 19	<u>16 18</u>	18 19	17 19	4
<u>-</u>	D16S539	11 <u>12 13</u> 15	<u>12 13</u>	12 12	11 15	4
<u>-</u>	D2S1338	17 <u>19 20</u>	<u>19 20</u>	17 18	17 24	<u>5</u>
<u>-</u>	D8S1179	<u>9</u> 10 <u>13</u> 14	<u>9 13</u>	13 13	10 14	4
<u>-</u>	D21S11	29 31 <u>32</u>	28 <u>32</u>	30 30	29 31	<u>5</u>
<u>-</u>	D18S51	<u>12</u> 16	<u>12</u> 15	12 20	16 16	4
1	D19S433	<u>12</u> 14 15.2 <u>16</u>	<u>12 16</u>	12 15	14 15.2	<u>5</u>
<u>-</u>	TH01	<u>69.3</u>	<u>6 9.3</u>	6 9.3	6 9.3	2
T	FGA	<u>19</u> 2426	<u>19</u> 21	20 21	24 26	<u>5</u>

Profile overview for case Case1_data

Matching alleles in the replicate and Suspect2

Select	Name	Replicate	Suspect1	Suspect2	Victim	Distinct Alleles
7	Epithelial					
12	D3S1358	14 16 12	15(13)	15 12	14.66	4
V	VWA	16 17 16 15	10.15	18.19	(UP)	4
V	D165539	11 1213 15	12 (3)	1212	11/20	4
R	D2S1338	1219 20	100	1718	0020	5
P	08S1179	9 10 7 14	9.43	1313	1 941 9-91	4
17	D21S11	29 31 32	29,14	30 30	1444 (March 1997)	5
R.	D18551	1216	37.39	1720	110.72	4
V	D195433	12 14 15.2 16	11114	1215	1 ACREATE	5
2	TH01	69.2	6.63	6.2.7	59.4	2
V	FGA	19 24 26	11.21	20 21	210	1

S1+S2+victim / victim + 2 unknowns



Exploratory approach (Evaluate the LR)

- Why are we doing this?
- The process is exploratory
- So what will happen if we replace a suspect with a random man?
- We would expect the LR to be very low (an exclusion!!)
- Therefore, the performance test is a measure of *robustness* and we consider this to be an important part of model *validation*

Performance test drop-out=0.45

We replace suspect 1



Performance Test Results

Performance test drop-out=0.45

We replace suspect 2



Performance Test Results

Exploratory approach (case re-evaluation)

S1,V,U vs V,U,U

S2,V,U vs V,U,U



'Dissection of the propositions' Simplify the model before you report 20

International efforts



Euroforgen-NoE collaborative exercise on LRmix to demonstrate standardization of the interpretation of complex DNA profiles

L. Prieto^a, H. Haned^b, A. Mosquera^c, M. Crespillo^d, M. Alemañ^e, M. Aler^f, F. Álvarez^a, C. Baeza-Richer^g, A. Dominguez^h, C. Doutremepuich¹, M.J. Farfán^{j,k}, M. Fenger-Grøn¹, J.M. García-Ganivet^m, E. González-Moya^k, L. Hombreiroⁿ, M.V. Lareu^c, B. Martínez-Jarreta^o, S. Merigioli^p, P. Milans del Bosch^q, N. Morling¹, M. Muñoz-Nieto^d, E. Ortega-González^r, S. Pedrosa^s, R. Pérez^d, C. Solís^a, I. Yurrebaso^t, P. Gill^{u,v,*}

ENFSI collaborative exercise on mixtures interpretation: 75 labs

CrossMark

Conclusions

- The LRmix model is based on principles that are described by the DNA commission documents
- The model is exploratory.
- Comparative studies are under way supported by Euroforgen NOE
- A need for transparency in software



EUROFORGEN-NoE update: EDNAP Meeting Tbilisi 2014

Peter M. Schneider

Institute of Legal Medicine University of Cologne (Germany)





EUROFORGEN-NoE is funded by the European Commission within the 7th Framework Programme

22/04/2014 Slide no 1

Project Data



- Funding period:
 - five years (01.01.2012 31.12.2016)
- Total costs / grant requested:
 - €8.1 Mill. / €6.6 Mill.
- Consortium:
 - 12 partners from 8 countries
- Website:
 - www.euroforgen.eu





• WP1

- Project Management: Coordination and communication office
- WP2
 - Integrating research and networking: towards the creation of an European Virtual Center of Research in Forensic Genetics
- WP3
 - Three exemplar reserach projects
- WP4
 - Ethical and legal aspects, and the societal dimension of forensic genetics
- WP5
 - Education, Training and Career Development



WP2 – Integrating research and networking





Geography of European Forensic Genetics







EUROFORGEN-NoE is funded by the European Commission within the 7th Framework Programme

22/04/2014 Slide no 5



• First Call 2013

- 14 fellowships awarded to 13 colleagues from 9 countries
- Details on website

• Second Call 2014-2015

- 20 new fellowships open
 - Laboratory visits for 3-5 days
 - Active participation in workshops related to EFG aims
 - Other research/training activities related to scope of WPs 2-5
- Application details on the website
- Travel support up to EUR 500



The Newsletters



ISSUE 1/2013



WELCOME

Dear colleagues - we want to keep you up-to-date with the activities of the EUROFORGEN Network of Excellence: Our consortium aims to improve the exchange of information about new research developments, funding opportunities, training resources and educational activities, and to explore the impact of modern forensic genetics on the society.

The EUROFORGEN Network of Excellence has now almost completed the second year of the five year funding period 2012-2016. During the first year, numerous activities have been initiated to establish the ground for better networking structures in the field of forensic genetics. So in this first newsletter we will provide you with information about

- first results compiled from European-wide surveys among forensic laboratories to inquire about the situation both in routine work and research.
- and an inquiry among selected National Contact Persons (NCPs) on education and the needs for training, which has helped to compile our "White Book on Education"

These data have beloed us to establish

- a short term fellowship program and

- to initiate a "Train the Trainers" workshop series. We sincerely hope that you and many other colleagues from the field of forensic genetics will benefit from our Network of Excellence and might even join the activities with their own contribution.

Peter M Sohnelder

institute of Legal Medicine, University of Cologne Coordinator of the EUROFORGEN-NoE Consortium



bers at the 2013 General As

THE EUROPEAN RESEARCH DIRECTORY

The Network initiative to distribute a detailed questionnaire to all forensic genetic laboratories across Europe has resulted in the publication of the Directory of Forensic Genetic Research Laboratories in Europe. The questionnaire was designed to explore the activities and needs of the European forensic genetic laboratories. For the report, 146 questionnaires from 31 different countries were analysed.

ization, the educational needs, the research activities and the challenges in the field. 200

Furthermore, a total of 179 laboratories across Europe

were identified as active in the field, and have

accepted to contribute to the activities of

EUROFORGEN-NoE. Significant information was

derived concerning the number and type of labs in the

different European countries, the level of standard-

EUROFORGEN

The European landscape of forensic genetic laboratorie

Official labs generally have a large number of cases per year (more than 1000), while university labs have a reduced volume of cases (less than 200), and private labs an intermediate position with the majority having between 200 and 1000 cases. Most of the labs are carrying out practical casework (mainly criminal casework and paternity testing). There is a good level of standardization in Europe concerning the type of markers; however, statistics is still a challenge and most of the labs have a need for training in statistics and especially in the interpretation of complex profiles.

There is widespread concern for an increased need for education and training in forensic genetics. It is of great importance to establish a global framework across Europe in order to achieve the highest educational standards. Concerning research, a high percentage of labs is interested in research activities 70% of the labs are performing research, of these about 80% carry out practical applied research. In addition, a number of advanced research topics are addressed in a wide range of laboratories. This is perhaps the reason behind the European leadership in this field. However, funding for this type of research is difficult to obtain in most countries, and increased efforts to improve the funding situation are needed so that the European leadership can be maintained.

You find the full report as PDF document and the contact information of all participating laboratories on our website

NoE is funded by the E members: institute of Legal Medicine, University of Cologne (DE) - institute of Legal Medicine, University of Santiag ie (ES) - Norwegian Institute of Public Health, Osio (NO) - King's College, Lo alty of Copenhagen (DK) - Netherlands Forensic Institute, Den Haag (NL) - Institute of Legal Medicine, Innsbruck Medic uity (AT) - Norwegian University of Life Sciences, Osio (NO) - Dept. of Genetics and Evolution, Jagielionian University Krakov raity Centre for Forensic Science, Newcastle (UK) - Exiontis GmbH, Berlin (DE) - GAS

- 2 newsletters published in 2013 ۲
- 1 newsletter published in 2014
- 2 more newsletters planned for 2014 ۲

News Section

Within the News Section you can find recent information on

- Project + Dissemination activities
- Recent * project meetings
- The application rules for the * Short Term Fellowships

We sincerely hope that you and many other colleagues from the field of forensic genetics will benefit from our Network of Excellence and might even join the activities with their own contribution.

We look forward to your feedback - if you wish to support the EUROFORGEN-Network of Excellence. + Contact us!

Peter M. Schneider Institute of Legal Medicine, University of Cologne (Coordinator of the EUROFORGEN-NoE Consortium)

Newsletter

The EUROFORGEN Network of Excellence has completed two years of a five year funding period from 2012-2016. During the first 2 project years, numerous activities have been initiated to establish the ground for better networking structures in the field of forensic genetics.

Please have a look into the recent Newsletters:

- Newsletter 2/2013
- Newsletter 1/2013



EUROFORGEN-NoE is funded by the European Commission within the 7th Framework Programme

22/04/2014 Slide no 7


EP1: Crime scene investigation and human DNA discovery

- mRNA profiling of human body fluids/tissues
 Van der Berge et al.: A collaborative European exercise on mRNA-based body fluid/skin typing and interpretation of DNA and RNA results. FSI Genet. 2014
- EP2: Guiding investigations by genetic analysis of physical traits and tailored multiplex development
 - SNPs as ancestry markers
 Phillips et al.: Building a forensic ancestry panel from the ground up: The EUROFORGEN Global AIM-SNP set. FSI Genet. 2014

EP3: Bioinformatics, in silico modelling and statistics

 Development of software tools for forensic applications
 Prieto et al.: EUROFORGEN-NoE collaborative exercise on LRmix to demonstrate standardization of the interpretation of complex DNA profiles. FSI Genet. 2014





- A <u>competitive call</u> will be published to invite research <u>proposals</u> for 3 new projects & partners
- Projects will be selected based on <u>peer-review evaluation</u>
- <u>3 new projects</u> will be funded for 24 months
- Projects will be <u>fully integrated</u> into the Consortium
- Expected timeline for submission and project selection:

– End of funding:	31 December 2016 .
 Starting date of project: 	1 January 2015
– Amendment of contract:	until 15 December 2014
 Decision announced: 	30 September 2014
 Submission deadline: 	31 July 2014
– Publication date of call:	1 May 2014



White Book on Education & Training



	6	
	EUROFORGEN Network of Excellence	Courses and training in forensic genetics in Europe
		Here we provide you with a detailed overview on
	White Book	 • <u>upcoming courses</u> - courses that are performed only once or only once in a while • <u>regular courses</u> - courses that are performed regularly each year Within each section you have the easy option to sort the courses by country!
	on the current status of education and training in forensic genetics	
	2782 Solda	Please 🗐 contact us if you have new information on courses or training offers!
	in Europe	· · · · · · · · · · · · · · · · · · ·
	March 2013	White Book on the current status of education and training in forensic genetics 2013 ف <u>Download pdf.</u>
	European Forensic Genetics Network of Excellence	
	www.euroforgen.eu	
7	EUROFORGEN-NoE - FP7/2007-2013 under grant agreement no. 285487	22/04/2014 Slide no 10
SEVENTA (BAMERICAR)	Deliverable D5.2	22/04/2014 Slide no 10

White Book on Education & Training



		NCPs		
Content		No.	%	
Common basic courses	General introduction to forensic genetics	2	12	
	Population genetics	1	6	
	Biostatistics and/or software	8	47	
Crime case work	Presumptive tests, DNA extraction, etc.	3	18	
	STR typing with CE	2	12	
	Interpretation of results and weight of evidence	14	82	
Relationship testing	Interpretation of results in complex relationship cases	10	59	
Disaster victim identification	General introduction, management, etc.	6	35	
Advanced technical	mtDNA sequencing	2	12	
education	General course	3	18	
Ethical/legal issues	General introduction, management etc.	4	24	
Other courses	Econometrics	1	6	
	Reporting officers courses	2	12	
	Court presentation	2	12	



* Six countries did not list urgently needed courses.

22/04/2014 Slide no 11



KICK-OFF MEETING: 'TRAIN THE TRAINERS' WORKSHOP SERIES

7 – 10 OCTOBER 2013 IN COPENHAGEN

"STATISTICAL METHODS IN FORENSIC GENETICS"

PRACTICAL ORGANISER: NIELS MORLING

FACULTY: THORE EGELAND, OSLO (ORGANISER OF TEACHING)

DANIEL KLING, OSLO

OSKAR HANSSON, OSLO

GURO DØRUM, OSLO

http://arken.umb.no/~theg/Copenhagen2013/





'TRAIN THE TRAINERS' WORKSHOP SERIES

FOLLOW-UP WORKSHOP: 20 - 23 MAY 2014 IN COPENHAGEN

- Participants are expected to organize local workshops on training topics
- Local workshops are planned / considered in:
 - Italy
 - Spain
 - Poland
 - Croatia





- CEPOL course Avila (Spain), June 4-7, 2013
 - "Mixtures, complex DNA profiles, and familial testing: interpretation workshop schedule"
 - Open for members of police laboratories
 - www.cepol.europa.eu



• CEPOL course Madrid (Spain), June 2-6, 2014

- Target Group: Specialist police and forensic experts involved in research for the resolution of crimes by scientific methods of DNA analysis or related to EUROFORGEN project.
- Faculty: Peter Gill, Hinda Haned, Corina Benschop, Thore Egeland, Guro Dørum and Ana Mosquera





• Ethical and Legal Aspects

- <u>Legal Audit</u>: Forensic DNA Profiling and Databasing The Legal Landscape of Europe
- <u>Survey:</u> Public Perceptions of Forensic Genetics
- Funding application to contribute to web-based project
 "Sense about Science" http://www.senseaboutscience.org/
- Public relations conference in Brussels (2014)
 - To raise awareness about forensic DNA achievements
 - To address funding needs
 - To approach decision makers



f EUROFORGEN - European Forensic Genetics Network of Excellence Peter Home Find Friends a - HE -1 Events EUROFORGEN **I** Find Friends GROUPS **Vetwork of Excellence** EUROFORGEN - Euro... 🔄 RuckteKat Friends ... 4 🖌 John McLaughlin 🛛 20+ EUROFORGEN - European ... Photos Files Notifications + Create Group a Members Events 샾 I Forensic DNA ÷Α. ICMP - International C ... Create Group Write Post Add Photo / Video **Ask Question** Add File 96 members ABOUT Open Group APPS Write something. The EUROFORGEN-NoE proposal aims to Games develop a network of excellence for the Photos creation of a E. See More Peter Schneider Pokes 96 members (3 new) Invite by Email March 20 at 11 16pm · Cologne · Edited Notes. + Add People to Group Please take notice of this interesting workshop on databases and DNA T Gifts Taus: data exchange in Brussels! Music Forensic DNA - Science - Genetics LAR PURE METERIZATION, VARIATION O STRENGTIENING OF () On This Day Forensic DNA Data Exchange SUGGESTED GROUPS See All mit Your Neighborhood Games Feed 20+ Romanov News + Join Mike Coble joined FRIENDS Cologne, Germany ... 5 Worldwide Association of + Join Hockey 20+ Women For... Anna Barbaro and 9 DNA 20+ other friends joined 😽 Close Friends Jobs in Sweden -A Family English + Join A Workshop in Brussels 9,010 members INTERESTS. PIES 2014 Workshop on Forensic DNA Data Exchange Rages and Public Fig.. PEOPLE YOU MAY KNOW See All

EMPOP

Dr. Walther Parson, MSc Assoc. Prof. Institute of Legal Medicine, Innsbruck, Austria Adj. Assoc. Prof. Penn State Eberly College of Sciences, PA, USA walther.parson@i-med.ac.at

EMPOP timeline



Гуре 🛞	haplotype as differe	ences to rCRS
Sample Info 😡 Query 🙆	Range Pr	ofile
Options 🥹	Match type pattern literal Disregard InDels in I 16193 309 309 455	Number of differences displayed
Source 😡	🗹 Forensic (25328) 🗹	Literature (9289)



Continuous review of "literature data" with improved QC software FSIG 2007; Parson FSI 2004, FSIG 2006, 2007 2014 Zimmermann FSIG 2010, Brandstätter IJLM 2004,

Em



Only high-quality data are loaded onto EMPOP Do not use forensic/literature categories any more All data undergo the same quality control using EMPOP QC tools







Röck FSIG 2010

SAM - alignment-free search software guarantees that matches are found regardless of alignment and notation of haplotypes







Geographic/metapopulation categories to sort matches according to forensic relevant criteria Include linguistic categories and use map display



Query	Result	Details	Neighbors			
Entire Datab	ase				Frequency	Confidence Interval
				11 / 26127	4.2102e-4	[2.1019e-4, 7.5320e-4]
By Origin					Frequency	Confidence Interval
+ Africa				0 / 1900	0.0000e+0	[0.0000e+0, 1.9396e-3]
+ America				6 / 13829	4.3387e-4	[1.5924e-4, 9.4411e-4]
+ Asia				3/6024	4.9801e-4	[1.0271e-4, 1.4547e-3]
+ Europe				2 / 4374	4.5725e-4	[5.5380e-5, 1.6507e-3]
By Metapopu	ulation				Frequency	Confidence Interval
African				0 / 103	0.0000e+0	[0.0000e+0, 3.5181e-2]
+ Native Am	nerican			0 / 1930	0.0000e+0	[0.0000e+0, 1.9095e-3]
+ East Asian	ı			0 / 3681	0.0000e+0	[0.0000e+0, 1.0016e-3]
+ Eurasian				6/9503	6.3138e-4	[2.3174e-4, 1.3737e-3]
+ Sub-Sahar	ran			0 / 886	0.0000e+0	[0.0000e+0, 4.1549e-3]
+ Afro-Asia	tic			0 / 2049	0.0000e+0	[0.0000e+0, 1.7987e-3]
Afro-Americ	an			2/2823	7.0847e-4	[8.5810e-5, 2.5569e-3]

Tabular summary of matches (and neighbours) providing details relevant to a forensic search by offering sortable columns

E OP





Matches displayed on a geographic map provides a better overview on matches and populations included in a search



Query	Result	Detalls N	eighbors										
11 of 11 haplotypes shown													
	Origin					Metopopula	tion			Haplogroup	9))		
filter Em	fitter arigins					Filter metop	opulations			filter hoplo	group		
EmpAcc#	Continent	Region	Country	Province	City	ы	12	в	Ignored Mutotions	Rank 1	Rank 2	Publications	
EMP00057	Asia	Central Asia	Uzbekiston	Tashkent province	Chirchik	Eurasian	et.	7.	12	H2c2c1	RO 😗	Irwin 2010	
EMP00017	Europe	Western Europe	Germany	Southwest Germony		Eurasian	Indo- European			H2o2o1 👧	RO	Lutz-Bonengel 2009	
EMP00063	Asia	Central Asia	Uzbekistan	Fergana	Ohunbabaev	Eurosion				H202010	RO	Irwin 2010	
EMP00009	America	South America	Argentina	Misiones		Admixed				H2c2a1	RO 🚯	Bobillo 2010	
EMP00231	America	South America	Argentina	Rio Negro		Admixed				H2o2a1	RO	Bobillo 2010	
EMP00057	Asia	Central Asia	Uzbekiston	Tashkent province	Chirchik	Eurosion				H2c2c1	RO 🚯	Irwin 2010	
EMP00466	Americo	Northern America	United States of America	Florido		Eurosian	Indo- European			H2a2a1 🚯	RO	AFDIL 2011	
EMP00473	America	Northern America	United States of America	Idaho		Admixed				H2c2c10	RD 😗	AFDIL 2012	
EMP00514	Europe	Western Europe	Germony	Baden- Württemberg	Freiburg	Eurosion	Indo- European			H2o2o1	RO	Lutz-Bonengel 2012	
EMP00530	America	Northern America	United States of America	Washington		Afro- American				H2c2c1	RO 🚯	AFDIL 2012	

Matches associated with haplogroup status provides phylogenetic background information that may be relevant in the forensic context





Global distribution of haplogroups displayed on a geographic map based on high quality mtDNA data included in EMPOP



Query Result Details Nei

ls Neighbors

92 of 92	2 of 92 haplotypes shown														
	Origin					Metapopula	tion						Hoplogroup		1
filter Em	filter origins	*				filter metap	opulations			filter			filter hoplag	moup	
EmpAcc#	Continent	Region	Country	Province	City	u.	12	13	Cost	Count	Mutotions	ignored Mutations	Rank 1	Rank 2	Publications
EMP00454	Americo	Northern America	United States of Am <mark>erica</mark>	Arizona	Phoenix	Eurasian	indo- European		0.47	1	C16362T (0.47)		н O	RD 😗	AFDIL 2011
EMP00055	Americo	Northern America	United States of America	Kentucky	Louisville	Eurosion	Indo- European		0.77	1	C315.1- (0.77)		H20201	H2o2a1 0	AFDIL 2006
EMP00507	Americo	Northern America	United States of America	Vermont		Eurosian	Indo- European		0.77	1	C315.1- (0.77)		H2o2o1	H2o2o1	AFDIL 2011
EMP00447	America	Northern America	United States of America	Alabama		Admixed			0.77	1	C315.1- (0.77)		H2a2a1 🚯	H2a2a1 🕄	AFDIL 2011
EMP00409	Americo	Northern America	United States of America	Colorado		Eurosion	indo- European		0.77	1	C315.1- (0.77)		H2o2o1	H2o2o1 🕄	AFDIL 2011
EMP00539	Americo	Northern America	United States of America	Arizona	Mesa	Eurosian	Indo- European		0.77	1	C315.1- (0.77)		H20201	H2o2o1 😗	AFDIL 2012
EMP00482	Europe	Western Europe	Germany	Bavaria	Munich	Eurasian	Indo- European		0.77	1	C315.1- (0.77)		H2o2o1 😗	H2a2o1 😗	Eduardoff 2013
EMP00470	America	Northern America	United States of America	Hawaii		Admixed			0.77	1	C315.1- (0.77)		H2c2c1 0	H2a2a1 😗	AFDIL 2012
EMP00497	America	Northern America	United States of America	Ohio		Eurasian	Indo- European		0.77	1	C315.1- (0.77)		H2o2o1	H2o2o1 🕄	AFDIL 2012
EMP00529	Americo	Northern America	United States of America	Texas		Notive American			0.77	1	C315.1- (0.77)		H2a2a1	H2o2o1	AFDIL 2012

Neighbours displayed by distance and costs Costs are estimated based on fluctuation rates neighbours can be sorted in various ways



16024-576 16025C

Entire Database		Frequency			
	0 / 26127	0.0000e+0			
By Origin	Frequency				
+ Africa	0/1900	0.0000e+0			
+ America	0 / 13829	0.0000e+0			
+ Asia	0/6024	0.0000e+0			
+ Europe	0/4374	0.0000e+0			

Query Res	ult Details Neig	hbors
Rank 1: MRCA:	120201	
Costs	Name of Profile	Haplogroup
2.00	H2a2a1	H2a2a1
2.00	H2a2a1a	H2o2o1a
2.00	H2a2a1b	H2a2a1b
2.00	>EU330412.1	H2a2a1b
2.00	>HM589044.1	H2a2a1
2.00	>JQ702287.1	H2a2a1b
2.00	>JQ703864.1	H2a2a1b
2.00	>NC_012920.1	H2a2a1
Rank 2: MRCA:	RO - only 30 most fa	vorable entries out of 28
Costs	Name of Profile	Haplogroup
2.54	H2o2o1d	H2o2o1d
2.65	>JQ704346.1	H2o2o1b
2.67	H2o2o1c	H2a2a1c
2.75	RO	RO
2.75	HV	HV

Non-matching haplotypes can be subjected to automated haplogroup estimation using a maximum likelihood approach





Haplogroup browser

is a dynamic tool to surf the phylogenetic tree and search in mtGenome sequences for (combinations of) mutations





Haplogroup Browser

Find haplogroup in mtGenoms by SNP

16318T×	8137T 🔀	
117		
1175		
U7-1		
0741		
Uraz		
U/a3		
U7a4		
U7a5		
U7b		
U7b1		

Haplogroup browser

is a dynamic tool to surf the phylogenetic tree and search in mtGenome sequences for (combinations of) mutations





Haplogrouping! EMMA?



Phylotree





Haplogroup M Eastern Eurasia Native America



Manual haplogrouping using Phylotree

EMP00539 (Arizona)

16024-576 **16235G 16291T 16293G 263G 315.1C 16235G 16291T 16293G 263G** n.a. - hg H2a2b1







Manual haplogrouping using Phylotree

ABS133 (Argentina) 16024-576 10646 16189C 16292T 16519C 71A 153G 204C 207A 263G 315.1C 373G 10646A

mtGenome - hg H55 **16189C**16519C153G204C263G315.1C750G 1438G 4769G 8860G 10646A 15326G 263G n.a. 750G 1438G 4769G 8860G 10646A 15326G



modified from Phylotree B.16





EMMA

Forensic Science International: Genetics 7 (2013) 601–609 Concept for estimating mitochondrial DNA haplogroups using a maximum likelihood approach (EMMA)⁴

Alexander W. Röck^a, Arne Dür^b, Mannis van Oven^c, Walther Parson^{a,d,*}

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^b Institute of Mathematics, University of Innsbruck, Innsbruck, Austria

^c Department of Forensic Molecular Biology, Erasmus MC, University Medical Center Rotterdam, The Netherlands

^d Penn State Eberly College of Science, University Park, PA, USA

EMMA uses

phylotree haplogroup nomenclature

virtual phylotree haplotypes (4,806; phylotree build 16) curated database of full mtGenomes (19,299; build 16)

improved haplogroup estimates







	worldwide	hg X1'3	hg K	hg T
T16519C	in 18,362 of 40,246	in 8 of 8	in 893 of 931	in 1,124 of 1,171
%	45.6	100	95.5	95.9

Apparently the mutation rates differ between haplogroups need to determine mutation rate within haplogroups 606 discernible CR-HGs (relevant to forensics)

Manual haplogrouping of 19,171 EMPOP CR haplotypes according to phylotree build 12-15 (Nov 2011 - Sep 2012) Requirements:

high quality sequences (EMPOP QC process) consistent phylogenetic alignment (Bandelt and Parson, 2008) 409 CR-HGs >4 haplotypes in EMPOP (R9)



The fluctuation rate (r) is a measure for the stability of a "mutation" within a given haplogroup



T16217C is a stable marker in hgs B4 and HV2 and therefore a strong signature for hg-estimation T152C is strongly fluctuating in all 4 hgs and therefore of little relevance for hg-estimation



The fluctuation rate (r) is a measure for the stability of a "mutation" within a given haplogroup

$$r_{\alpha\beta} = \frac{\sum_{\gamma} \min(n(\alpha, \gamma), n(\beta, \gamma))}{\sum_{\gamma} n(\gamma)}$$

 α , β ... A, C, G or T; α unequal β γ ... runs over all CR-hgs $n(x, \gamma)$... denotes number of samples in CR-hg γ with symbol x $n(\gamma)$... denotes total number of samples in CR-hg γ



Control region fluctuation rates

CR-HG	Ν	T16217C	Difference to majority	T152C	Difference to majority
A2	30	0	0	16	14
B2	40	39	1	19	19
C1	50	0	0	27	23
D1	40	2	2	12	12

 $r_{(T16217C)} = (0+1+0+2) / (30+40+50+40) = 3 / 160 = 0.01875$

 $r_{(T152C)} = (16+19+27+12) / (30+40+50+40) = 74 / 160 = 0.4625$

 $0 \le r \le 0.5$

EN

NPOP



Concept

Compare test haplotype to all database haplotypes by striving for maximum likelihood

$$L_{t}(b) = \prod_{i} r(b_{i} \rightarrow t_{i})$$

b ... database haplotype, t ... test haplotype, i ... positions

Calculation of the product is computationally intensive, therefore minimal costs are computed instead





 $C_t(b) = \lg(\prod_i r(t_i \rightarrow t_i)/L_t(b)) \quad \text{where} \quad \lg(x) = \log 10(x)/3$

For short motif lists, such as differences to rCRS between database and test haplotypes, the cost function can be efficiently evaluated by

$$C_t(b) = \sum_i c(b_i, t_i)$$

and

$$c(b_i, t_i) = \lg(r(t_i \to t_i)/r(b_i \to t_i))$$

are real numbers termed positional costs for the change from the base profile symbol to the test profile symbol

Average "mutations" yield value of approx. 1.0, unobserved transitions 2.0 and unobserved transversions 3.0

Ranking of haplotypes by total costs equals ranking by maximum likelihood



Upcoming meetings





EUROPEAN NETWORK OF FORENSIC SCIENCE INSTITUTES

Monopoly 2010 project M1: Development and implementation of an ENFSI standard for reporting evaluative forensic science

ENFSI DNA working group - Tbilisi, April 23rd 2014 Dr Sheila Willis (Project Coordinator) and Dr Tacha Hicks
Purpose of this afternoon session

- Present the aims of the project.
- Provide information on the feedback mechanism.
- Explain how the document relates to DNA examination and how it can help us.
- Engage a discussion of specific points of the standard.
- See how the approach can be applied in the laboratory

Why is it important for ENFSI ?



ENFSI Mission Statement

The purpose of ENFSI is to share knowledge, exchange experiences and to come to mutual agreements in the field of forensic science. ENFSI is recognized as an expert group in the field of forensic sciences.

- It is a cross cutting area whatever the discipline.
- As research for assessing the value of findings is performed in ENFSI working groups, we need to build context to support mutual understanding and to ease communication.
- We need to rise the standards in the field of interpretation.

What will this initiative bring

- Provides an ENFSI response to the concerns raised in the NAS report.
- A way to harmonize the provision of evaluative reports in Europe, hence promoting the mutualisation and exchange.
- A mechanism to identify the gap in data to support evaluative reporting (e.g., trace DNA).

Overall project aims and objectives

- Elaboration of a standard (->guidelines)
- Identification of implementation challenges
 - a roadmap for the future implementation of the approach and an audit template
- Provide training
- Support
 - M1 Project collaborative website
 - Product development process
 - SEFE e-learning programme (Lausanne)
 - FORSTAT meetings (Edinburgh, Cracow)

Project Core Group

- 8 representatives of ENFSI laboratories, currently:
 - Forensic Science Laboratory (EFÉ), Ireland (coordinator of the project)
 - Instytut Ekspertyz Sądowych (IES), Institute of Forensic Research, Krakow, Poland
 - Institut National de Criminalistique et Criminologie (INCC), Belgium
 - Institut de police scientifique (IPS), Université de Lausanne, Switzerland
 - LGC Forensics, UK
 - Netherlands Forensic Institute (NFI), The Netherlands
 - Swedish National Laboratory of Forensic Science (SKL), Sweden
 - Servicio de Criminalistica de la Guardia Civil, Spain

Initial basis for the ENFSI Draft Standard

Science and Justice 49 (2009) 161-164



Standards for the formulation of evaluative forensic science expert opinion



Consultation Process



Product Development Process



B.1.2.1 Each "Draft for Comment" will include the following instructions for submitting comments: "The Core Group encourages stakeholder participation in the preparation of documents. Suggestions for modifications are welcome and may be forwarded to the Secretary in writing. The following information is required as a part of the response:

- (a) Submitter's name
- (b) Affiliation (agency/organization)
- (c) Address
- (d) Telephone number and/or email address
- (e) Document title and version number
- (f) Change from (note document section number)
- (g) Change to
- (h) Basis for change

B.1.4 The decision of the Core Group, with justification, shall be communicated in writing to the submitter by the Chair or his or her designee within 30 days of that decision. In addition, the submitter will be notified of the appeals process as outlined in section B.3.

Dealing with feedback

A A
 Define the formulation of evaluative reports in forensic science
 PDP - Feedback on evaluative standard from Issue 2.5, now Issue 2.6
 Sign in
 PDP - Feedback on evaluative standard from Issue 2.5, now Issue 2.5, now Issue 2.6
 Sign in
 File Edit View Insert Format Data Tools Help Last edit was made 13 days ago by christop
 Working..

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4			Contracting they			Design of the second second		Data da al d
2	Form or Content	Critical or not Critical	standard concerned	Description of the issue	Possible solution	Group	the Standard	Core grout
3	Content	Critical	3.3	Not clear that finding nothing might be a potential finding, if interest for activity level assessment	Add after findings in (): that can include the absence of recovered material	Agreed	Sentence "No result is also a finding" was added in the glossary (Section "Findings").	11/23/2012
4	Form	Not critical	3.7 to 3.9	Should the text say something about laboratory error?	??	It is felt difficult to address that point in the standard	No modification retained.	11/23/2012
5	Content	Critical	3.11	The sentence is very categorical "will not" whereas section 3.2 accepts some exceptions	add "exception made of the conditions under 3.2"	See modification proposed	Section 3.11 was taken out. Further development added in the guidance note.	11/23/2012
8	Form	Not critical	3.10 & 3.12	Is the term "assigned" well chosen for a likelihood ratio or a probability?	33	Group agrees that word is fine.	No change.	11/23/2012
7	Form	Critical	3.15 & Guidance note 4	When the term support for a proposition is used, specify against an alternative systematic	Add "against the other" after proposition	Agreed.	relative to the alternative has been added in 3.15 Guide note 4 was updated as well.	11/23/2012
8	Form	Not critical	5 and References	For the word "proposition", there is a reference to Evett et al. (2005), which is not in the list of references	Should this be Evett et al. (2000)? Or add the missing reference to the list of references	Agreed.	Corrected to Evett & al. 2000a	11/23/2012
9	Form	Not critical	5 and References	There are two "Cook et al. (1998)" and two "Evett et al. (2000)"	add "a" and "b" to the citations	Agreed.	Corrected to 1998a and 1998b	11/23/2012
10	Content	Not critical	4. Guidance note 1	may not be possible to always include "mandate and questions asked"	add if possible?	Agree	Update made (also in 3.13)	11/23/2012
U.	Content	Not critical	Guidance note 2 p6	does "fired ammunition elements" include GSR?	confusing, needs clarification	Agree	Sentence changed to "as such as bullets and cartridge case comparisons"	11/23/2012
12	Content	Critical	guidance note 2 p6/7	Is it correct that the absence of data is not justification to adopt source level propositions?	why not source for example for road traffic accidents where scientist has not seen the damage and has no idea of probability of transfer of paint?	add something in guidance note 2	Update made in Guidance note.	11/23/2012
13	Content	Critical	3.3	PA allows to avoid 'post hoc rationalisation' Here it is unclear if one can give a global LR without assigning the factors that could be influenced by our findings and have therefore to be assigned before examination	Add as a bullet point 'assess factors such as relevance, presence of artfacts, transfer probabilies'. Or consider introducing 'post hoc rationalisation' in the glossary	Not agreed, because the document does not prescribe how to obtain a particular LR in pre- assessment.	No change.	11/23/2012
14	Content	Critical	3.12	Case information should be in the case file	Add case information	Agreed.	Case information added as a first bullet point	11/23/2012
52	Content	Critical	Guidance note 2	Is it on purpose that sub-level is not mentioned	Add (sub-) before source	add something in guidance	Done.	11/23/2012

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Document currently under review with the ENFSI working groups.

ENFSI standard for the formulation

- of evaluative reports in forensic
- science

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Issue	Date	Name
1	08.09.08	S.M.Willis
2.3	15.05.12	Project M1 Core Group (Dublin May 2012 meeting)
2.4	29.06.12	Project M1 Core Group (Dublin May 2012 meeting), CC/AB
2.5	22.08.12	Project M1 Core Group (EAFS Aug 2012 meeting)
2.6	26.11.12	Project M1 Core Group (Dublin Nov 2012 meeting)
2.7	26.04.13	Project M1 Core Group (The Hague April 2013 meeting)

Document 2.8 will be circulated through Roman.

Use the form available on the link below to report to the Core group structured feedback that will be considered in the preparation of the next iteration of the standard:

https://docs.google.com/forms/d/1I5g4wqOG8UZkSMhPGjB8W8dCWru7CZ2V4dSn-5LL600/viewform?pli=1

Key elements for the ENFSI document

Clarity on nature of REPORTS Evaluative reporting based on a likelihood ratio

Proposition/alternative

(sub) Source / activity level propositions

	11000					
neral bics	Source Pre-as	e/activity level	the document			
	Avoidi	ng findings-led eval	uation			
		1. SCOPE 2. EVALUATIVE REPORTING				
ha		3. STANDARD				
d			4.1 Reporting requirements			
	of the		4.2 Propositions			
ndard	orthe	4. GUIDANCE NOTES	4.3 Data used to assess the strength of the findings			
			4.4 Meaning of the LR in an evaluative report			
		5. GLOSSARY				
		REFERENCES				

1. Scope of the standard

This standard is only for <u>evaluative</u>
 <u>reporting</u>, not for investigative, nor for intelligence reports, nor for analytical reports.

An evaluative report is to help the Court with the issue at hand.

It does not mean that we cannot use the framework for investigation, but it is not the scope of the document and the project M1.

Example of an investigative case:



Helping formulate propositions in forensic DNA analysis

John Buckleton^{a,} , Jo-Anne Bright^{a, b}, Duncan Taylor^e, Ian Evett^e, Tacha Hicks^a, Graham Jackson^{f,} ^g, James M. Curran^b

Show more

http://bib-ezproxy.epfi.ch:2120/10.1016/j.scijus.2014.02.007

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Investigative example

- Murder of 4 related people.
- A bloodstain was recovered associated with the accused that could be explained as a mixture of all 4 deceased.
- However 27 people from the pedigree were sampled by the authorities and given as reference samples (trawl).
- Taken individually, 7 of these cannot be excluded from the mixture.

Why investigative?

- If there are *M* persons of interest, to a *N* contributor profile, then there are many pairs of hypotheses that could be considered.
- Our LR depends on the hypotheses.
- With no case information (in particular what is the issue for the Court), we cannot help formulate propositions.
- Thus, the weight of our findings cannot be given.

2. What is evaluative reporting?

- When one is asked to assess and report the value of the findings.
- Evaluation of forensic science findings for use in court uses probability as a measure of uncertainty. This is based upon the findings, associated data (including expert knowledge), case specific propositions and conditioning information.

3. Standard itself

- Likelihood ratio
- Hierarchy of propositions
 - Sub-Source level (analytical characteristics)
 - The DNA came from Mr A versus from an unknown person
 - Source level proposition
 - The blood came from Mr A versus from an unknown person
 - Activity level (both intrinsic and extrinsic characteristics)
 - Mr A had intercourse with Ms B versus they only had social contact as defined in the case information

Activity versus (sub)-source

- The further down the hierarchy the scientist operates the more the responsibility for interpreting the evidence is transferred to the court or to other experts.
- So, if the case goes to Court and that the issue regards the activities, then, we have a duty to report given activity level propositions.
- If not, this could be misleading.

Activity level propositions: when?

- When transfer mechanisms, persistence and background levels of the material has a significant impact on the understanding of the alleged activities and requires expert knowledge.
- In order to avoid bias (i.e., findings led) preassessment should be conducted.
- There is uncertainty, and the Court should be made aware of this.
- This will take time (research, education)
- Working groups are the ideal forum for this.

Sub(source) level propositions: when?

- Helping to address source level propositions is adequate in cases where there is no need for expert knowledge to take the results in relation to source level propositions and consider them in the context of the alleged activities in the case.
- Here is an example..

Sub(source) level propositions: when?

- Example:
 - A large bloodstain is recovered at the point of entry on a burglary scene and delivered at the laboratory for a DNA analysis. Combination of presumptive test and appearance allows the scientist to establish the nature of the body fluid (here blood).
 - Further, a party says that he has never been in the premises. The set of propositions will be (1) the bloodstain came from the defendant and (2) the bloodstain came from another unknown individual.

4. Guidance notes

- These notes explain the standard in further detail.
 - Reporting requirements
 - Propositions
 - Data used
 - Meaning of the likelihood ratio (i.e., the expression of the value of the evidence)

5. Glossary

All terms underlined in the standard are defined in the glossary.

Examples

Examples of statements will be provided.



Roadmap towards the implementation of the ENFSI standard for the formulation of evaluative reports in forensic science

Step 1 Managing the change

- Identifying **key personnel responsible** for the implementation
- Deciding on a **strategy** to approach each forensic discipline covered by the laboratory (focus groups, leaders in each discipline, etc.)
- Adopting a project plan with defined objectives and timeline

Step 2 Training

- Providing training and workshops on the standard (i.e. framework of circumstance, propositions, likelihood ratio, workshops per discipline)
- Identifying what is covered by evaluative reports (compared to factual or investigative reports)
- Training should include **competency testing**.
- Providing information and training to the stakeholders (e.g. police officers, judiciary, mandating authority) in relation to the changes associated with the standard in particular the exchange of information at the outset of the case and the reporting practice.

Step 3 Identifying the issues

- Implementing the mechanisms to establish the **key issues** in the submitted cases by adapting the exchange of information between the forensic laboratory and the mandating authority
- •Setting an appropriate framework of propositions (including dealing with "no comment" interviews)
- Identifying the levels of propositions (source or activity level) that best help address the key issues
 If appropriate, carrying out a pre-assessment of cases
- and communicating with the mandating authority
- •Identifying the *data* requirements (*data* as defined in the standard) to help address the issues. If needed, undertake structured data acquisition.
- •Optional: Developing a uniform verbal scale to support consistent reporting within the laboratory

Step 4

Reporting according to the standard

- Reporting on the **probability of the findings given the propositions** and relevant background information which leads to a **likelihood ratio**
- •Avoiding in reports statements that are transposing the conditional (i.e. not reporting on the probability of the propositions given the observations)
- •Auditing the casework using the audit template associated with the standard

Discussion

- The document is a draft and we welcome this discussion as it is the opportunity to improve it.
- We cannot amend it today: it has to follow a defined process (PDP).
- But, we can **discuss** the any point raised.
- If questions/propositions remain, then it will be your responsibility to raise them to the Core group through the structured feedback mechanism.
- http://tinyurl.com/ENFSIM1
- Welcome comments from the floor

Take home message

- This document is a framework to help us avoid misleading our readers (e.g., the Court).
- It helps identify where there are gaps in our knowledge.
- These gaps can be filled through research carried out in the working groups.
- Trace DNA brings on new challenges that we ought to tackle.
- We should give the value of our findings based on logic.

Thank you very much for your kind attention

ENFSI standard for the formulation of evaluative reports in forensic

3 science

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7 ENFSI standard for the formulation 8 of evaluative reports in forensic 9 science

10 **1. SCOPE**

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1.1 This document provides forensic scientists with a standard for formulating
 evaluative reports and related requirements for the case file.¹ It does not cover the
 requirements for intelligence², investigative or technical reporting.

1.2 Forensic scientists working with various types of known items and questioned
 or recovered items (e.g., traces), and different legal systems ultimately have a duty to
 assist the judicial system. This can be achieved by the production of intelligence,
 investigative, technical or evaluative reports.

1.3 Experts will not report on matters outside their own area of expertise. Experts
will not usually give <u>conclusions</u> on issues that do not require specialist knowledge.
However, if asked, they may do so provided it is made clear that this is not part of an
expert evaluation. They should conform to the ENFSI code of conduct (BRD-GEN003).

1.4 The document requires formulating evaluative reports within a <u>hierarchy of</u>
 propositions and defines the conditions to operate within that hierarchy.

30 2. EVALUATIVE REPORTING

31

32 2.1 Evaluative reports for use in court should be produced when two conditions
 33 are met:
 34

- 35 36 37
- The forensic scientist has been asked by a <u>mandating authority</u> or party to examine, quantify and/or compare material (typically recovered trace material with reference material from known potential sources);
- 38 sources);
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43 2.2 Evaluative reports should be labelled (where practicable) or identified as such
44 by the agency in order not to be confused with the other types of reports (intelligence,
45 investigative or technical).

¹ The elaboration of this document is based on previous works published by the Association of Forensic Science Providers (AFSP, 'Standards for the formulation of evaluative forensic science expert opinion', Science & Justice, 2009, 49, 161-164).

² All terms underlined in the document find a definition in the glossary at the end of the document.

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47 2.3 Evaluation of forensic science findings in court uses probability as a measure
48 of uncertainty. This is based upon the <u>findings</u>, associated <u>data</u> (including expert
49 knowledge), case specific propositions and <u>conditioning information</u>.

51 2.4 Evaluation will follow the principles outlined in Guidance note 1. It is based on
52 the assignment of a <u>likelihood ratio</u>. Reporting practice should conform to these
53 logical principles.

54 **3.0 STANDARD**

55 3.1 The key issue(s) in the case will be established by: 56 57 Considering all available, relevant information and, where necessary, 58 requesting additional information Agreeing by discussing - when possible or necessary - with the 59 • 60 relevant mandating authority or party (e.g., magistrate, prosecution or 61 defence team) 62 63 3.2 On the basis of the case circumstances and the agreed key issue(s), 64 competing propositions at a given level in the hierarchy are set [guidance note 2]. 65 Propositions set should ideally not be changed at any stage unless: 66 67 Key issues in the case change and/or • The conditioning information changes 68 • 69 Forensic findings lead to new investigative avenues 70 71 3.3 Pre-assessment helps achieve balance and assures that scientists formulate 72 potential findings explicitly before the examination. Case pre-assessment may not 73 always be necessary for source level propositions, but should be conducted in cases 74 when activity level propositions are set. Given the chosen propositions, and the 75 circumstances of the case, pre-assessment aims to: 76 77 specify main potential findings of scientific examinations of the items • 78 submitted; 79 assign probabilities (i.e., their order of magnitude) for potential • 80 findings regarding each proposition. This leads to an assignment of 81 likelihood ratios for potential findings at this stage. 82 83 When results are already known (e.g., results of a DNA-database search), and initial 84 pre-assessment was not conducted, every effort should be made to avoid to be led by the findings. This may involve having another scientist carry out the assessment 85 without the results. 86 87 88 3.4 If, as a result of the pre-assessment, scientific examinations are unlikely to 89 assist in differentiating between the propositions, the mandating authority or party will 90 be advised accordingly. Such advice and the result of it must be documented in the 91 case file. 92 93 If a mandating authority or party dictates an examination strategy that, in the 3.5 94 opinion of the forensic scientist, is inappropriate then this authority or party must be

advised accordingly and the advice and conversations must be made explicit on the

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96 case file. Any resulting limitations on the interpretation[s] must be described in the
97 report.
98

99 **3.6** If access to relevant items, as identified through the assessment, is denied or
100 unavailable then the mandating authority or party will be advised as to the limits of
101 any resulting interpretation. This advice should be made clear through the report.
102

103 3.7 Examination of specimens and / or items is carried out on the assumption that 104 such specimens or items have been recovered, packaged, preserved and 105 transported in accordance with accepted protocols or best practice unless there is 106 good reason to believe otherwise - e.g. from the submission form, the container or 107 packaging. In such cases further enquiries will be made to confirm or otherwise such 108 suggestions and discussions will take place with the mandating authority or party to 109 agree a way forward. This may result in the items not being examined or, if they are, 110 the results and conclusions may be subject to limitations the extent of which should 111 be expressed.

3.8 Pre-assessment, examinations, observations, analyses and evaluation
 carried out should be valid and in accordance with an established and controlled
 methodology.

3.9 Pre-assessment, examinations, observations, analyses and evaluation should
be made by competent and trained personnel.

3.10 Based on the findings of the examination and their probabilities assigned during pre-assessment, a likelihood ratio is assigned. The assigned probabilities (at the pre-assessment stage) may be refined in the light of the findings e.g., a rare glass or fibre type. Justification for changes will be documented.

- 125 **3.11** The case file should include (not exhaustive list):
- Case information (verbatim, or as otherwise received)
- Mandate and questions asked, if available
- 128 Materials and items received
- The key issue(s) and propositions of interest
- All discussions with mandating authorities and parties in the case
- Examination strategy

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- Potential outcomes and assigned probabilities when pre-assessment was
 carried out
- Relevant <u>data</u> used in probability assignments [guidance note 3]
- Observations made and analytical results
- Discussion and evaluation of the strength of support that the findings provide to help to resolve the issues (and related propositions) dictated by the purpose and the circumstances of the case
- Conclusions and report given to the mandating authority or party.
- 141 **3.12** Reports should include (not exhaustive list):
- Conditioning information used
- Mandate and questions asked, if required
- The propositions addressed
- Relevant items collected/received
- Items examined
- Significant findings
- 148 Discussion and evaluation
- Conclusion(s)
150

3.13 The conclusion(s) in the report must be related to the propositions under consideration and the assigned likelihood ratio [**guidance note 4**].

153 154 **3.14** The conclusion must be expressed either by a value of the likelihood ratio 155 and/or using a verbal scale related to the value of the likelihood ratio. The verbal 156 equivalents must express a degree of support for one of the propositions relative to 157 the alternative, and be defined from ranges of likelihood ratios. The choice of the 158 reported verbal equivalent is based on the likelihood ratio and not the reverse. The 159 full verbal scale used will be provided in the report for reference [guidance note 4].

161 **4.0 GUIDANCE NOTES**

162 **Guidance Note 1: Reporting requirements**

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The reporting of the value of scientific findings must conform to four requirements:
 Balance, Logic, Robustness and *Transparency*. These requirements are met by
 following the principles of forensic evaluation.

167

168 The standards set out in this document describe the mechanism by which these 169 requirements are met in formulating such reports.

170

Balance - The findings should be evaluated given at least one pair of propositions: usually one based upon one party's account of the events and one based upon an <u>alternative</u> (opposing party's account of the events). In the absence of a reasonable alternative the value of the findings cannot be assessed, it can only be shown that it is or is not in contradiction with the only proposition that has been put forward. In that case, scientists should state clearly that they are not reporting upon the value of the findings.

178

Logic - Reporting scientific findings should address the probability of *the findings* given the propositions and relevant background information and not the probability of
 the propositions given the observations and background information. The report
 should not contain statements that are transposing the conditional.

183

184 *Robustness* - The reporting should be capable of sustaining scrutiny by other experts 185 and cross-examination. It should be based upon sound knowledge and experience of 186 the trace type(s) and the use, wherever possible, of pertinent databases, published 187 data or ad hoc case based experimentation. The scientist will be satisfied that the 188 results of the observations and analyses upon which inferences and conclusions are 189 drawn are robust. Robustness is understood here as the scientist's ability to explain 190 the grounds for his opinion together with his degree of understanding of the particular 191 trace type.

192

Transparency - The reported conclusions should be derived from a demonstrable process in both the case file and the report (see also 3.11 and 3.12). The report should be written in way that is suitable for a wide audience of readers (i.e. participants in the justice system). It may include expert supplements explaining the technical background.

198 **Guidance Note 2: Propositions**

200 Level in the hierarchy

An evaluative statement will generally relate to propositions either at (sub-) source or
activity level.

It is the duty of the expert to help the court by explaining the significance of their findings within the context of the case. To do this, the expert either considers the findings given the propositions proposed by the parties, or in their absence, the expert proposes the most reasonable propositions from the case circumstances.

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Addressing source level propositions is adequate in cases where there is no need for expert knowledge to take the results in relation to source level propositions and consider them in the context of the alleged activities in the case. The following example will illustrate the above.

Example: A bloodstain is recovered at the point of entry on a burglary scene and
delivered at the laboratory for a DNA analysis. Combination of presumptive test and
appearance allows the scientist to establish the nature of the body fluid (here blood).
Further, a party says that he has never been in the premises. The set of propositions
will be (1) the bloodstain came from the defendant and (2) the bloodstain came from
another unknown individual.

221

Evaluating analytical results at source level is adequate here because expert knowledge is not necessary to evaluate the findings at activity level. Indeed, the evaluation here does not require knowledge from the DNA scientist with regards to transfer, persistence and recovery of bloodstain.

226

The above applies to many other types of physical traces (e.g. footwear marks, toolmarks, fingermarks) - typically marks and materials left at crime scenes. It also applies to trace types such as hairs/fibres and paint when the material can reasonably be assumed by the expert to be the result of the alleged activity (e.g., tuft of fibres at point of entry, semen on the crotch of panties).

232

233 However, activity level propositions should ideally be used when the consideration of 234 transfer mechanisms, persistence and background levels of the material has a 235 significant impact on the understanding of the alleged activities and requires expert 236 knowledge. Indeed, phenomena such as secondary (or tertiary) transfer, 237 contamination or fortuitous presence of such material in the environment affect the 238 case evaluation of the findings, in particular when small quantities of material are 239 recovered. This is typically the case for trace types such as microtraces (fibres, 240 glass, gunshot residues, other particles), small quantities of DNA, drugs or 241 explosives.

242

In areas such as bullets and cartridge case comparisons, handwriting, speaker
 recognition, physical fits, there is, in general, no distinction between source level and
 activity level propositions.

246

247 Absence of specified propositions248

When a proposition cannot be specified, the expert should provide an intelligence, an investigative or a technical report as deemed appropriate in the context of the case.

251

Experts will establish the alternative propositions from the investigator, mandating authorities and parties, or use their own judgement to highlight reasonable alternatives.

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In cases where it proved impossible (e.g. one party makes "No comment"), in order to evaluate the findings to offer a *balanced* conclusion, the expert requires to consider an alternative proposition. There are three options available in such circumstances:

- Adopt alternative propositions that most likely reflect the party's position and prepare an evaluative report. Only this option can lead to the production of an evaluative report meeting the requirements of this standard.
 - 2. Explore a range of <u>explanations</u> for the findings and prepare an investigative report.
 - 3. State the findings in a technical report and state whether they are in agreement or in conflict with the only proposition that has been put forward. The report should stress that in the absence of an alternative, it is impossible to evaluate the findings logically.
- 272 Changing propositions

273 274 In principle, propositions are not changed unless the key issues in the case and/or 275 the conditioning have changed. For example, when the issues at hand are at activity 276 level, the absence of data on transfer, persistence or background level of the trace type under consideration is not a justification to change the set of activity level 277 278 propositions to a set of source level propositions. Indeed the requirement for 279 considering activity level propositions does not derive from the availability of data in 280 relation to the findings and the type of trace, but solely from the consideration that 281 phenomena such as transfer, persistence and background levels crucially affect the 282 strength of the information that can be reported.

283

284 Example: In a case in which a considerable quantity of DNA is recovered from the 285 hands of a suspect, and it is alleged that the suspect digitally penetrated a victim, it is 286 relevant to consider factors such as background and persistence of such trace material; in particular if it is alleged by the suspect that recovered DNA here on his 287 288 fingers is the consequence of a legitimate social contact. If, in such a case, the 289 examiner fell short of structured data to guide as to the factors relevant for evaluation 290 given activity level propositions, it would be inappropriate to retreat to source level 291 propositions (stating the victim versus an unrelated person as the source of the 292 recovered DNA). The reason for this is that, firstly, it is not contested that the victim is 293 the source of the recovered DNA (hence the propositions are irrelevant). Secondly, 294 and more importantly, the potentially large likelihood ratio for source level 295 propositions bears a risk to be misinterpreted as a conclusion with respect to activity 296 level propositions (i.e., the actual issue in the case).

Nevertheless, if the examiner chooses in this case to report the findings at source level (arguing, for example, that the suspect is not saying anything about any alternative activity), the examiner must mention explicitly with appropriate caveats the factors that have a bearing on the assessment of the findings at activity level. Alternatively, the expert may explain the possible activities that may have led to the findings in an investigative report.

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The next example illustrates the fact that propositions should not be adapted in the light of the forensic results obtained but should remain anchored on the framework of circumstances.

308

Example: Consider a case where it is alleged that an offender broke a double glazed 309 310 window (made of two differentiable sheets of glass respectively A and B). From the 311 alleged circumstances, the following propositions were set to pre-assess the case at 312 activity level: (1) the individual broke the double-glazed window as alleged, versus (2) the individual has nothing to do with the breaking, nor was he near the scene. For 313 314 illustration, assume that during pre-assessment the examiner expected under 315 proposition (1) to recover from the garment worn by the offender a large amount of 316 glass fragments from both windows. However, the examination led to the recovery of only two glass fragments of one group indistinguishable of sheet A. In such a case, 317 318 the forensic findings still require to be assessed in the context of the above 319 propositions (including the consideration of the small number of fragments 320 associated with sheet A and the absence of any glass fragments associated with the 321 sheet B). It would be misleading to adapt the propositions at activity level to a new pair of propositions at source level, i.e.: (1) the two recovered fragments come from 322 323 sheet A, vs. (2) the two recovered fragments come from an unknown source of glass.

324

325 It is recognized that there are cases where propositions are set following forensic 326 examinations. Typical examples occur in early stages of investigations.

327

Example: Assume a case involving a dead body, and an unknown cause of death. The medical examiner may find a bullet, considered lethal, and this may lead to the formulation of a particular set of propositions for a firearms examiner who may conduct comparative examinations with bullets fired under controlled conditions using the weapon of a suspect.

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334 Guidance Note 3: Data used to assess the strength of the 335 findings

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337 Likelihood ratios are based on the assignments of the probability of the findings given each of the competing propositions. These assignments must be based on data and 338 documented on the case file. This standard invites disclosure of the data that were 339 used to base conclusions on. Published data will be used wherever possible as a 340 341 basis for these assessments, provided they are deemed relevant by the scientist and 342 fit for purpose. If published data are not available then data from unpublished 343 sources may be used as long as they are documented on file. Regardless of the 344 existence of sources (published or not) of structured data, personal data such as 345 experience in similar cases and peer consultations may be used provided that the 346 forensic scientist can justify the use of such data. It is expected that the expert will 347 document the grounds on which an assessment is based. For example, if that 348 assessment is based on experience, the expert will be able to demonstrate relevant 349 and documented previous professional activity.

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In particular, in cases where the material or trace type is rarely encountered then the
 probabilities will be informed by either specialist knowledge and / or case tailored
 simulations or surveys.

355 Guidance Note 4: Meaning of the likelihood ratio in an 356 evaluative report

The conclusion should express the degree of support provided by the forensic findings for one proposition or the specified alternative(s) depending upon the magnitude of the likelihood ratio (LR).

For a LR assigned as one the conclusion should be to the effect that the findings provide no assistance in addressing the issue covered by the propositions.

For values of LR greater than one the conclusion should be that the findings are more probable if the first proposition (in the numerator) is true rather than the alternative (in the denominator). For values of LR less than one then the conclusion should be that the findings are more probable if the alternative is true, than if the first proposition is true.

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This, in effect, is indicating a degree of support of the forensic findings for one
 proposition relative to the other.
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The degree of support will relate to the magnitude of the likelihood ratio. A likelihood ratio may be expressed by a verbal equivalent according to a scale of conclusions (see also Nordgaard & al., 2012). An example is provided below:

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Values ³ of likelihood ratio	Verbal equivalent (two options of phrasing are suggested)			
1	The forensic findings <i>do not support</i> one proposition over the other.			
	The forensic findings <i>provide no assistance</i> in addressing the issue.			
2 - 10	<i>Weak</i> support ⁴ of the forensic findings for the first proposition compared to the alternative.			
	The forensic findings are <i>slightly more probable</i> given one proposition rather than the other.			
10 - 100	Moderate support			
	are <i>more probable</i> given			
100 - 1000	Moderately strong support			
	are appreciably more probable given			
1000 - 10,000	Strong support			
	are <i>much more</i> probable given			
10,000 - 1,000,000	Very strong			
	are <i>far more</i> probable given			
1,000,000 and	Extremely strong			
above	are exceedingly more probable given			

381

Although the choice of terms, number of steps and intervals may vary between
laboratories, the scale and its principles will apply across all forensic disciplines
covered within laboratories.

385 When source level propositions are considered, and when the likelihood ratio 386 amounts to the reverse of a conditional match probability $(CMP)^5$ – typically in a DNA

 $^{^3}$ Likelihood ratios corresponding to the inverse (1/X) of these values (X) will express the degree of support for the specified alternative compared to the first proposition.

⁴ Scientists or their reports should avoid conveying the impression that a statement of the kind: "the forensic findings provide *weak support* for the first proposition compared to the alternative" is meaning that the findings provide (strong) support for the stated alternative. It just means that the findings are up to 10 times more probable if the first proposition is true than when the stated alternative is true. This is also the reason why the alternative should be explicitly stated. In cases where the reader could be expected to misread as described above, scientists must add additional comments.

case involving a large unmixed stain – the expert may choose to report the
 conditional match probability instead of the likelihood ratio.

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Another special instance of source level conclusions occurs when the likelihood ratio
(i.e., its numerator) is equal to zero. In this case, the term 'exclusion' is commonly
used as a conclusion.

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⁵ The term conditional match probability (CMP) expresses the probability of an adventitious correspondence conditional on a case-tailored alternative proposition. This term is more general than the more widely known but restrictive term 'random match probability (RMP)'.

395 **5.0 Glossary**

397 Preliminary note

Many of the distinctions between the terms described in this section are not rigid and exclusive. The reader should allow for a flexible view and accept that, in some situations, one term may appear more suitable in one situation than in another.

403 Case file

404 All laboratory notes and correspondence associated with the case and which may, 405 under certain circumstances, be disclosed.

407 Classification

The assignment of a person or object to a particular category is called classification (see also examples given in the paragraph <u>technical report</u>).

411 Conclusion

In evaluative reports, the conclusion is a statement that answers particular questions
and is reached on the basis of a reasoning process that conforms to the <u>principles of</u>
forensic evaluation. It is formulated as a <u>likelihood ratio</u>.

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416 **Conditioning information**

Conditioning information is the relevant information that helps the expert recognise the pertinent issues, select the appropriate propositions and carry out the case preassessment. It must always be regarded as provisional and the expert must be ready to re-evaluate findings if the conditioning information changes. Examples of relevant information that could change include the nature of the alleged activities, time interval between incident and the collection of traces (and reference items) and the suspect's/victim's account of their activities.

424 More formally, conditioning information is an essential ingredient of the assignment of 425 probabilities, since all probabilities are conditional. In forensic evaluation, it is 426 important not to focus on all possible information, but only on the information that is 427 relevant to an allegation of interest. Forensic reporting requires scientists to make 428 clear their perception of the conditioning information at the time they conduct their 429 examination (see also principles of forensic evaluation). Conditioning information is 430 sometimes known as the framework of circumstances (or, background information). 431 Much of the non-scientific information will not have a bearing on the scientific 432 findings, but it is essential to recognise those aspects of the non-scientific information 433 that do. Further examples of relevant information include the origin of the perpetrator 434 $(\neq$ the suspect) and the nature of garments and surfaces.

436 **Data (associated with the evaluation of a given trace type)**

Throughout this document, the term data is not used to describe results of 437 438 examinations associated with the items in the case at hand. Data will refer to the 439 technical and empirical knowledge associated with a given trace type. It refers to 440 general (empirical) observations, such as the occurrence of DNA profiles among 441 members of a relevant population or the expected number of glass fragments 442 transferred on garments as a result of breaking glass. Such data can take, for 443 example, the structured form of scientific publications, databases or internal reports 444 or be part of the expert knowledge built upon experiments conducted under 445 controlled conditions, training and experience.

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449 **Evaluative report**

An evaluative report is a report that evaluates the forensic findings in the light of at least one pair of propositions. Therefore it is based on a <u>likelihood ratio</u> and conforms to the <u>principles of evaluation</u>. Most of the time, evaluative reports will follow from comparative examinations between material of unknown source and reference material from one or several potential source(s).

456 Evidence

The term 'evidence' is a generic term. From a strict scientific point of view, evidence refers to outcomes of forensic examinations (<u>findings</u>) that, at a later point, may be used by legal decision-makers in a court of law to reach a reasoned belief about a proposition. Evidence should be a term kept for lawyers.

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462 **Examinations (tests and analyses)**

In their general meaning, examinations, tests and analyses refer to all technical operations conducted - in controlled conditions and/or according to a predefined protocol - by forensic scientists for the purpose of making observations (that will constitute the findings) deemed to be relevant to help address the <u>key issue(s)</u> in a case.

468

469 Explanation

470 In the context of a forensic science evaluation, explanations have been recognised 471 as intermediate considerations when exploring less formal alternatives. While they 472 have the potential to account for particular observations, they do not qualify as formal 473 propositions essentially because - often - they may be a statement of the obvious, 474 speculative or fanciful. Moreover, explanations can be offered provided that no 475 exclusive alternatives have been presented by parties. A further characterising 476 feature of explanations is that their use as a conditional leads to a probability of one 477 for any given outcome. Consequently, no probative value can be assigned to the 478 respective outcome. See also Evett & al. (2000a).

479 480 **Findings**

481 Findings are the result of observations and measurements that are made on items of 482 interest. They can be qualitative (nominal or ordinal) or quantitative (discrete or 483 continuous). No result is also a finding. Examples for qualitative results (typically, descriptors for categories) are fibre types and blood groups. These are nominal 484 485 because they have no natural ordering. Qualitative results are said to be ordinal if 486 they have an underlying order even though it is generally not quantifiable (e.g., the 487 damage of car involved in an accident, described as none, slight, moderate, severe, 488 very severe). Examples for discrete quantitative results are counts of glass fragments 489 or gunshot residues (in terms of integer values). Examples for continuous results are 490 measurements of physical quantities such as length, weight or refractive index (in terms of any value on a continuous interval). 491

Generally, all results (i.e., material differentiated from the specimen and material that was not differentiated) should be included in the evaluation, as it is not balanced to assess only findings that correspond to a potential source. Observations are made in a case, not as part of a series of experiments where an outlier can be eliminated.

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497 Intelligence report

In intelligence proceedings, scientists provide indicators (based on physical remnants of events) to link cases, events, and situations in the form of strategic intelligence (threat evaluation, measuring impact of ongoing crime phenomena) in order to help design strategies. This may lead to operational and investigative measures by determining trends and helping to design coordinated action. Operational measures

- 503 may be crime disruption, prevention, etc, whereas investigative strategies lead to 504 operational crime/case analysis.
- Intelligence reports address questions relating to phenomena and may be in the form
 of analytical products (such as crime pattern) or intelligence products (such as
 specific crime series to inform decisions on the prioritization of problems and targets).

509 Investigative report

510 Investigative reports are case specific or focus on a series of cases. They may 511 describe a modus operandi, type of traces observed in related cases to enhance the 512 detection and the relevance of collected traces. They are asked to help produce 513 explanations in order to account for observations (the outcome of analytical tests or 514 visual examinations as provided for example in a technical report).

- 515 Often, such explanations may refer to criminal phenomena that span on several 516 criminal events without one or several particular suspect(s) being available at the 517 time when the explanation is produced. The provision of an explanation is to be 518 distinguished from evaluative reporting that addresses more formally defined 519 propositions related to a criminal case in particular.
- They may be given orally, but should be confirmed in a brief statement (type of trace
 to look for, map, MO) or log book.

523 Item

524 An item is, in a very general sense, an object on which examinations are conducted. 525 An item can originate from a known source (in which case it could be a reference), 526 but can also be an object of unknown source seized for example at a crime scene (in 527 which case it would be a questioned item).

528

529 Likelihood ratio

A likelihood ratio is a measure of the relative strength of support that particular findings give to one <u>proposition</u> against a stated alternative (Aitken, Roberts & Jackson, 2011; Aitken & Taroni, 2004). It is defined in terms of the ratio of two <u>conditional probabilities</u>: (i) the probability of the <u>findings</u> given that one proposition is true and given the <u>conditioning information</u>; and (ii) the probability of the <u>findings</u> given that the other proposition is true and given the conditioning information.

- 536 The two conditional probabilities forming the likelihood ratio may be assigned either 537 on the basis of published data or the general knowledge (base) of the expert (see 538 also 'Probability, conditional').
- 539 The use of a likelihood ratio does not generally imply that one of the two propositions 540 considered must be true. Though the considered propositions are deemed most 541 relevant, they do not need to be exhaustive, so both propositions could be false. The 542 likelihood ratio says nothing about propositions other than the two that were 543 considered.

544 545 **Key Issue(s)**

The key issue(s) represent those aspects of a case on which a Court, under the law of the case, seeks to reach a judgement. The key issue(s) provide the general framework within which <u>requests</u> to scientists and <u>propositions</u> (for evaluative reporting) are formally defined.

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551 Mandating authority or submitting parties

552 Mandating authorities or submitting parties are the persons or institutions that submit 553 items to forensic scientists (i.e., to the institutions to which they are affiliated).

554

555 **Pre-assessment**

556 Case pre-assessment seeks to specify potential findings prior to performing any 557 analyses or prior to knowing the results, to assess the potential value associated with

each of these findings, as well as the probability with which these results may be
obtained under each of the competing propositions. The purpose is to (i) avoid
evaluations biased by the findings, and (ii) devise an examination strategy on which a
mandating authority or party can - in terms of expected results and associated
evidential value - agree (Cook & al., 1998a).

563 To ensure a balanced approach scientists should - prior to any examinations -564 formulate potential outcomes (along with probabilities for these outcomes) given that 565 each of the competing propositions is true. Otherwise an evaluation may be biased. 566 For example, a statement of the kind 'These observations correspond well to my expectations⁶ if the prosecution's proposition is true' is more trustworthy if the 567 568 scientist can demonstrate that the respective expectations (including assignments for 569 factors such as transfer and persistence) have been formulated prior to conducting 570 any examinations.

571

572 Principles of forensic science evaluation

573 The choice of <u>probability</u> as a measure for uncertainty suggests three precepts for 574 evaluation in forensic science (here adapted from Evett & al., 2000b, p. 235):

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 1. Interpretation of scientific findings is carried out within a framework of circumstances. The interpretation depends on the structure and content of the framework.
 - 2. Interpretation is only meaningful when two or more competing propositions are addressed.
 - 3. The role of the forensic scientist is to consider the probability of the findings given the propositions that are addressed, and not the probability of the propositions.
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585 **Probability, conditional**

Probability is a concept by which one can express uncertainties (about an event or, more generally, an unknown state of affairs). The laws of probability define the values that probability can take and how probabilities combine (Aitken & Taroni, 2004). Among forensic scientists and other members of the judicial area at large, it is standard to view probabilities (i) as conditional on the <u>information</u> available to the individual who makes a probability assignment (i.e., all probabilities are conditional) and, thus, (ii) as personal degrees of belief (Taroni, Aitken & Garbolino, 2001).

594 **Propositions**

Propositions are statements that are either true or false, and that can be affirmed or denied (Anderson, Schum & Twining, 2005). Propositions should be formulated in pairs (e.g., views put forward by the parties to the cases) and against a background of information and assumptions. Moreover, they should be amenable to a reasoned assignment of credibility by a judicial body and be useable for rational inference. Propositions should be distinguished from <u>explanations</u> that do not have the aforementioned properties. See also Evett & al. (2000a).

602

603 **Proposition, alternative**

An alternative proposition is mutually exclusive with respect to another competing proposition with which it forms a pair. Typically, the proposition put forward by the opponent party is referred to as an alternative proposition. Evaluative reporting requires the consideration of at least one pair of mutually exclusive propositions. It may involve the consideration of multiple propositions.

⁶ Notice that this use of the term 'expectation' is a generic one should be distinguished from its more restricted meaning and use in statistical literature.

610 **Propositions, hierarchy of**

In the context of criminal proceedings, propositions can be classified into broad 611 612 categories (or, hierarchical levels), such as 'crime level' (propositions that refer to the 613 commission of a criminal offence), 'activity level' (propositions about a human activity or a happening, 'source level' (propositions about the source of physical matter). See 614 615 also Cook & al. (1998b). 'Sub-source' represents a further propositional level. It may 616 be appropriate when it is not possible to attribute analytical findings to specific source 617 material. In DNA profiling, for example, it may be that a profile cannot be attributed to 618 a particular crime stain, item of tissue or other particularised source material. See 619 also Evett & al. (2002).

620

621 Request(s)

The request(s) is (are) the question(s) that <u>mandating authorities or parties</u> submit to forensic scientists.

624

625 Sample

The notion of 'sample' as it is considered in this section refers to a representative selection of items from a population of items (or, more generally speaking, the extraction of a representative part of a whole). Such a choice is made in a way that should allow reasoning about the properties of the source population. This is typically the case with seizures of items thought to contain something illegal. The notion of sample is appropriate when referring to the collection of representative material from a known source.

633

634 Specimen

Like a <u>sample</u>, a specimen is also part of a whole, yet it is fundamentally different from a sample. In a great majority of forensic contexts, a specimen represents a single (possibly degraded or even contaminated) <u>item</u>, such as a stain, a fingermark, a shoemark, etc. found on a crime scene. A characteristic feature of a specimen of this kind is that it does not offer the same qualities as a sample because there is a fundamental uncertainty arising from its nature as trace material. For example, it may not be representative and/or replicable.

642

643 Strength of support of the findings

This is the expression of the extent to which the observations (i.e, findings) support one of the two competing propositions. The extent of the support is expressed to the mandating authority or party in terms of the magnitude of the <u>likelihood ratio</u>. It can also be expressed using a verbal scale related to the magnitude of the likelihood ratio.

649

650 **Technical (factual) report**

651 A technical report is one that does *not* involve a formal evaluation under a pair of 652 competing <u>propositions</u>, expressed in terms of a likelihood ratio.

653 In a strict sense, purely technical or factual reporting is confined to a statement not 654 subjected to any sort of evaluation. It amounts to a descriptive account of findings. In 655 certain situations, the descriptive statement of observations may lead to particular 656 conclusions, such as a statement about the nature of particular physical matter, or -657 more formally - the assignment of an object to a class (i.e., classification). A technical report is often restricted to the results associated with the observations of items 658 without any comparative work against known sources. However, it may also involve 659 660 the reporting of quantitative measure(s) of an attribute (such as weight or 661 concentration) associated with the item. These measure(s) are generally reported alongside with some indications of their associated uncertainties (precision, accuracy 662 663 of the technique). Even though such reports may contain elements of statistical

664 evaluation, they remain descriptive and do not constitute evaluative reports as 665 defined in this document.

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667 Below are a few examples of technical reporting:

- This electropherogram shows at that locus two peaks, one at position a and one at position b. Given the criteria for allelic designation, we can conclude that the genotype of the donor of the stain is ab for that locus.
 - These transparent fragments have the following properties: size inferior to 2mm, have anisotropic optical properties, etc. They are glass fragments.
 - This powder of unknown composition has a strong kerosene smell, has a white and partially yellow colour, and leads to particular GC-MS results (i.e., chromatogram), hence it fulfils all the criteria to consider this substance as cocaine. When quantified, the results showed a concentration of XX% (± YY%).
 - The application of ESDA to the questioned document allowed the detection of the following indented numbers written on the document: 1, 10, 34, 22, 4.
 - The submitted document has been produced by a xerographic device such as a laser printer.

683 In order to place a technical report appropriately into context, experimental and 684 observational conditions need to be mentioned in the report.

686 **Transposing the conditional**

687 In legal contexts, a statement is a transposed conditional if it fallaciously equates (or, 688 confuses) the probability of particular findings given a proposition with the probability 689 of that proposition given these findings.

690

Example: Assume a bloodstain recovered from a crime scene that led to a DNA profile that corresponds to that of a suspect. If the probability of finding this DNA profile in an unknown person is, for example, 1 in 500 million, it would be fallacious to conclude that there is a probability of only 1 in 500 million that the suspect is not the donor of the stain. It is particularly important to remind this in cases in which the potential source has been found as a result of searching a – possibly large – DNA database.

698

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Ewa Klimuk Secretary

July 4th, 2013

Standard for reporting evaluative forensic evidence (Project M1) / EC monopoly work programme 2010 'Strengthening the Evaluation of Forensic Results across Europe (STEOFRAE).

Dear ENFSI Member Representatives,

Dear Expert Working Group Chairs,

A Core Group of representatives of the following 8 ENFSI institutes started the above mentioned monopoly 2010 project M1 in 2012:

- Forensic Science Laboratory (EFÉ), Ireland (coordinator of the project)
- Instytut Ekspertyz Sądowych (IES), Institute of Forensic Research, Krakow, Poland
- Institut National de Criminalistique et Criminologie (INCC), Belgium
- Institut de police scientifique, Université de Lausanne, Switzerland
- LGC Forensics, UK
- Netherlands Forensic Institute (NFI), The Netherlands
- SKL, Sweden
- Servicio de Criminalistica de la Guardia Civil, Spain

Based on the publication "Standards for the formulation of evaluative forensic science expert opinion"¹ the Core Group developed in five meetings and after consulting QCC the enclosed

¹ Science & Justice, Vol. 49, 159 - 228

DRAFT version 2.7 of an "ENFSI standard for the formulation of evaluative reports in forensic science."

As it was announced at the last Joint Meeting in Berlin, the Core Group would like to consult the WG Chairpersons and ENFSI Members on the DRAFT version 2.7 as a basis for discussion in the whole ENFSI community. Please consider the following information:

(1) The document is a DRAFT and should be considered as such. The objective of the consultation is to further improve the document based on the feedback from all stakeholders. The project consultation phase is conducted according to a product development process that guarantees due consideration to be given by the Core Group to all formal feedback received. That process is briefly outlined on the feedback form (see (4) below).

(2) The document is presented without the set of examples covering various forensic disciplines that the project team is currently building up. These examples will help in due time and they will also be shaped as a function of the feedback from the WGs consultation process. In other words, the final document will come with dedicated examples that should illustrate the principles set forth in the document.

(4) The Core Group is available to facilitate the discussion within the working groups. It can take the form of a general presentation to the working group annual meeting, dedicated workshops, etc. If the chairpersons see a need for discussion, they can simply contact Sheila Willis (SMWillis@fsl.gov.ie).

(4) Feedback can be logged formally using the following form:

https://docs.google.com/forms/d/1I5g4wqOG8UZkSMhPGjB8W8dCWru7CZ2V4dSn-5LL600/viewform?pli=1

Feedback can be either submitted through the WG chairperson or ENFSI Member Representative (to avoid duplication of comments), but also on an individual basis. The first option is ideal from an efficiency perspective, but the matter under consideration is so important that any feedback is welcome. In function of the amount of feedback received (either from a group or from a laboratory), the core team may suggest a meeting to be held with that group. Such a process has already been successfully applied during the internal consultation phase at the NFI (leading to the DRAFT 2.7).

On behalf of the Board I encourage you to peruse the enclosed DRAFT version 2.7, discuss it within your WG or Institute and give your feedback. It is up to you to evaluate the specified procedures and their applicability in daily forensic casework.

Yours sincerely,

Promas Anderman

Dr. Thomas Andermann Member of the 17th ENFSI Board

Forensic Science in Southeast Asia – AFSN and HSA, Singapore





Dr Chris Syn Director | Biology Division | Health Sciences Authority, Singapore



The Asian Forensic Sciences Network (AFSN) - A Fruit of International Cooperation

The LEADING INNOVATIVE AUTHORITY protecting and advancing NATIONAL HEALTH and SAFETY

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Before October 2008

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Two important developments that led to the formation of AFSN

HSA, Singapore 1st Regional DNA Symposium on Forensic DNA and Population

Statistics

ISA



Sep 2006 92 participants, 8 countries

Eijkman Institute, Indonesia 1st National DNA Symposium with invited speakers from Malaysia, Singapore and Australia



Feb 2007

Nov 2007 - A Regional Forensic DNA Profiling (REAFD) Workgroup was formed in with the inaugural meeting in Kuala Lumpur, Malaysia.



2nd meeting was held in Bangkok, Thailand, in Nov 2008.

7 Countries

UNDCP organised a regional Consultative Meeting for the Heads of Drug Testing Laboratories in Southeast Asia. Led to the publication of a annual regional drug newsletter

DrugNetAsia.



UNODC Project H44 on precursors and drug analysis – formation of a Forensic Science Network for this region was suggested by Dr Barbara Remberg of UNODC.



1999 Publication of *DrugNetAsia*

Year 2006 - 2007

A total of 3 meetings that brought together Drug Testing Laboratories and Law Enforcement Agencies from 11 Countries in Asia





Forerunner Group Meeting 14-15 Oct 2008

Brunei Darussalam (DSS), Malaysia (DOC), Philippines (NBI), Singapore (HSA), Thailand (CIFS), Vietnam (VFSI) UNODC: Dr Barbara Remberg AICEF: Prof Jose Lorente









Forerunner Group Meeting

Major Decisions

- Region Asia
- Membership Institute
- Voting rights Country
- Membership fees none
- o Constitution
- Code of Conduct
- Name of network
- Interim Board Formed

- 2 yr Presidency
- Secretariat of AFSN organisation from which President comes from
- AFSN website hosted by HSA
- AFSN Newsletter published by HSA

Formation of AFSN in Oct 2008



The scale symbolizes the justice The atomic orbit symbolizes the science The map of Asia symbolizes the region



Purpose of AFSN

To provide a forum for discussion



To enhance the quality of forensic services



To establish links with other networks



To formulate strategies



Growth of AFSN

Inaugural Meeting 2009, Kuala Lumpur, Malaysia



5th Annual Meeting 2013

Singapore





AFSN Meeting Attendance





Growth of AFSN

SCIEN

ETWOR



HSA Health Sciences Authority

Growth of AFSN

1.	National Forensic DNA Profiling Laboratory, Bangladesh	19.	Guangzhou Forensic Science Institute, People's Republic of China	2
2.	Department of Scientific Services, Brunei Darussalam	20.	Institute of Forensic Science, Ministry of Public Security, People's Republic of China	
3.	Centre for DNA Fingerprinting and Diagnostics (CDFD), India	21.	Institute of Forensic Science, Tianjin Public Security Bureau, People's Republic of China	
4.	Department of Police Medicine of the Indonesian National Police, Indonesia	22.	The Institute of Evidence Law and Forensic Science, China University of Political Science and Law, People's Republic of China	36. Korea Coast Guard Research Institute
5,	Eijkman Institute for Molecule Biology, Indonesia	23.	Laboratory Service, Philippines Drug Enforcement Agency, Philippines	37. CyberSecurity,
6.	Forensic Laboratory Centre of Indonesian National Police Headquarters, Indonesia	24.	National Bureau of Investigation, Philippines	Malaysia
7.	Indonesian Association of Forensic Pathologist, Indonesia	25,	Natural Sciences Research Institute, University of the Philippines Diliman Quezon City, Philippines	38. Philippines National Police Crime
8.	Laboratory of National Narcotics Board, Indonesia	26.	Health Sciences Authority, Singapore	Laboratory
9.	National Digital Forensic Centre (NDFC) of Supreme Prosecutor's Office, Korea	27.	Central Institute of Forensic Science, Thailand	
10.	National Forensic Service, Korea	28.	Department of Forensic Medicine, Faculty of Medicine, Chulalongkorn University, Thailand	
11.	Scientific Investigation Laboratory, Korea	29.	Department of Forensic Medicine, Faculty of Medicine, Siriraj Hospital, Thailand	
12.	Food and Drug Quality Control Center, Lao PDR	30.	Department of Medical Sciences, Thailand	
13.	Forensic Science Department of Judiciary Police, Macau SAR	31.	Faculty of Medicine, Chiang Mai University, Thailand	
14.	Department of Chemistry, Malaysia	32.	Human Genetics Unit, Department of Pathology, Faculty of Medicine, Ramathibodi Hospital, Thailand	
15.	Royal Malaysia Police Forensic Laboratory (RMP Forensic Lab), Malaysia	33.	Institute of Forensic Medicine, Police General Hospital, The Royal Thai Police, Thailand	
16.	Mongolian National Institute of Forensic Science, Mongolia	34.	Office of Narcotics Control Board, Thailand	13 Countries
17.	Forensic Science Division, Department of Fujian Provincial Public Security, People's Republic of China	35.	Vietnam Forensic Science Institute, Vietnam	38 Member Institutes
18.	Forensic Science Center of Guangdong Provincial Public Security Department, People's Republic of China			E AUTHORITY ONAL HEALTH and SAFETY

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Collaboration





Collaboration through AFSN Workgroups and Committee

Formation of workgroups/committee

- DNA Workgroup
- Illicit Drugs Workgroup
- Quality Assurance & Standards Committee
- Trace Evidence Workgroup
- Toxicology Workgroup
- Crime Scene Workgroup



DNA Workgroup

Collaboration – Training

- Training at Department of Chemistry, Malaysia
 - o 2 officers from Eijkman Institute, Indonesia, 22 Nov 04 Dec 2010
 - 2 officers from Hue University, Vietnam & 1 officer from Medical Forensics Center, Vietnam, 9 – 14 Jan 2011
- Training by HSA, Singapore in Aug 2009
 - 2.5 day QA workshop based on ASCLD/LAB and FBI DNA QAS QA for CIFS, Thailand
- HSA, Singapore organised a DNA Workshop on "New Frontiers in Forensic DNA" on 17-21 Oct 2011
 - Topics: population statistics, DNA databasing, and emerging DNA technologies for regional labs
 - Speakers: Dr Bruce Budowle and Dr Angela van Daal
 - o 58 participants from 7 countries



DNA Workgroup

Collaboration – Creating Standards

- HSA, Singapore was involved in the special committee to discuss the ISO standard for consumables used in DNA processing
- Countries: Australia, United Kingdom, Germany, US and Singapore
- IFSA Minimum Standards Document for DNA Analysis and Interpretation – for emerging laboratories

Collaboration - with other networks

- Participation in the DNA proficiency test
- Seeking SWGDAM observer status





Other Workgroups

Illicit Drugs Workgroup

- Organise workshops on UNODC International Collaborative Exercises "ICE" Program participation
- Representative in SWGDRUG to represent the Asian drug testing laboratories

Trace Evidence Workgroup

- > Inter-lab comparison of protocols on fire debris and vehicle paint analysis
- Contact with SWGMAT and ENFSI Textile and Hair Working Group

Toxicology Workgroup

- Contact with SMANZFL Toxicology SAG
- HSA provided Toxicology QA training to CIFS, Thailand in 2010
- First draft of AFSN Alcohol Analysis Guideline

Quality Assurance & Standards Workgroup

- Participation in Hair Proficiency Program provided by NFS, Korea
- Planning other PT programs



Challenges

- A very young network
- Governance
- Great variation in capabilities and QA systems of member laboratories
- Setting up of different workgroups and committees
- Funding
- Language

www.asianforensic.net



World Forensic Festival 2014



AFSN 2015 – hosted by IFS, China



Singapore



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Singapore

- Independent sovereign nation: 1965
- Land mass: 710 sq km
- Population of 5.3m (3.8m citizens & PRs)
 - ~ 40% have tertiary education
 - ~ 74% Chinese, 13% Malays, 9% Indians
- Median gross annual income: USD 35k
- Median annual household income: USD 75k
- GDP per capita PPP: USD 53k
- Home ownership: ~ 90%
- Unemployment rate: 1.8%
- Crime rate per 100,000 population: 549
 - Homicides: < 20 per year
 - Drug abusers arrested: 3,574 (in 2013)





- Ministry of Health → Health Sciences Authority →
 Forensic science testing
- Ministry of Home Affairs → law enforcement agencies
- Ministry of Law → creation and amendment of legislation
- Attorney-General's Chambers → Organ of State → prosecution
- Supreme Court and State Court → Organ of State



A Statutory Board of the Ministry of Health

Vision To be the LEADING INNOVATIVE AUTHORITY protecting and advancing NATIONAL HEALTH and SAFETY

Mission -

- To **Wisely regulate** health products
- To **Serve** the administration of justice
- To Secure the nation's blood supply
- To safeguard public health



Corporate Headquarters • Health Products Regulation Group • Blood Services Group • Applied Sciences Group

A Statutory Board of the Ministry of Health | The Singapore Public Service : Integrity + Service + Excellence



APPLIED SCIENCES GROUP



Chemical Metrology (22 staff)

Biology



 Forensic labs have USD 36m contract with law enforcement Accredited under ASCLD/LAB since 1996 (International program) since 2012)

When ANALYTICAL TOXICOLOGY DIVISION



Drug Abuse Testing Lab

- Analysis for Drugs and Metabolites in Urine Specimens
- Hair analysis unit



 Clinical & Forensic Toxicology Lab

- Ante-mortem specimens from suspected overdose cases, organ transplants
- Post-mortem specimens from accidents, homicides or suspicious deaths



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FORENSIC SCIENCE DIVISION

Forensic Chemistry & Physics Lab

- Trace evidence
 - Fibres / paints & surface coatings / glass / arson
- Firearms / Toolmarks / Impressions
- Explosives & pyrotechnics
- Physical examinations
- Chemical analysis
- Crime scene visits
- Blood Pattern Analysis
- Questioned Document
- Traffic accident reconstruction











ILLICIT DRUGS DIVISION

61 staff

USD 9.4m

Illicit Drugs Lab

- Qualitative and Quantitative Analysis of drug seizures
 - Heroin, morphine, opium
 - Cannabis, cannabis mixture
 - > Amphetamines ice, ecstasy, MDA ...
 - Cocaine
 - Ketamine
 - > LSD
 - Benzodiazepines sleeping pills
 - ➤ Legal highs "Bath salts" …
 - Clandestine Laboratory Investigation







Fig. 2. "Ecstasy' tablets in different shapes

To be the LEAI protecting



BIOLOGY DIVISION



Analysis of biological fluids

DNA profiling for identification

Parentage analysis, DVI

DNA Profiling Lab @ Outram

- Police major crime and volume crime
- Brunei
- Singapore Armed Forces
- Commercial cases



85 staff

USD 14.4m

- DNA Profiling Lab @ Synapse
 - Central Narcotics Bureau drug cases
 - Police volume crime



DNA Database Lab

Owned by SPF, managed by HSA

To be the LEADING INNOVATIVE AUTHORITY protecting and advancing NATIONAL HEALTH and SAFETY **Forensic Samples**



Samples:

~ 9,100 samples per annum

Examination for Biological Material









DNA Extraction

Organic phenol-chloroform





2 x QiaCube





Volume crime reports: 56 days Major crime reports: 210 days Urgent results: 1-3 days

Interpretation & Stats Analyses











PCR (x7) ID+, ESX



'F' Div 1 \bullet FTA* Classic Card igodolIN IN CHILD

- ~ 22,000 samples p.a.
- Processed in duplicate
- TAT of ~ 14 days

DNA Database Laboratory

- Convicted Offenders, Suspects, and Crime Scene profiles
 - Launched 14 Feb 2003
 - ~ 230,000 profiles





ID-Direct



Detection

~ 50 matches per month





What's next – Faster and Better

IntergenX RapidHit 200

FlapidHIT

➢ QiaCube



Illumina miSeq



Promega PowerPlex Y23

RapidHIT



≻ Leica LMD-6500



EADING INNOVATIVE AUTHORITY ting and advancing NATIONAL HEALTH and SAFETY



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Book

Not...

- Not a text book
- Not about DNA profiling as such

Why?

- Complex mixture interpretation is very well advanced now
- But interpretation of the evidence is still at a relatively primitive stage.
- New thinking is urgently required in relation to 'trace-DNA' evidence
- Note that techniques are now ultra-sensitive
- LCN used to be 'controversial' now everyoe does it – but what are the pit-falls?

ls....

- About reporting of evidence
- A list of examples where miscarriages of justice have occurred
- A detailed analysis of scientific reports from publicly available material
- A recognition that:
 - Miscarriages of justice arise from 'accepted practice'
 - The reports are not 'one-off' they represent one of hundreds of similar reports.
 - Only a tiny fraction of miscarriages of justice are captured
 - We have a responsibility to adjust practice accordingly to ensure mistakes are not repeated.

Solutions

- Provided in one chapter
- Not rocket science but it is science:
- Emphasis is made because much reporting is speculation (not science)
- What is science?
 - Scientific method described by Isacc Newton 350years ago

What is science

- The essentials are:
 - The hypothesis is constructed from belief of some principle
 - The hypothesis is tested by experiment and a body of data is generated to support the belief
 - Peer review is carried out to verify the procedure and the belief may be modified accordingly.

Examples of poor reporting

- Adam Scott (UK)
- R. Farah Jama (Melbourne)
- R v. Kerby (UK)
- R. v. Cleobury (UK)
- Case of 'death of Merdedith Kercher (Perugia, Italy)

Miscarriages of justice often have things in common

• But first we have to explain a few principles:

Definition of `trace-DNA'

- The term `trace-DNA' was used in a recent review by van Oorschot et al
- "Trace DNA samples may be defined as any sample which falls below recommended thresholds at any stage of the analysis"

My definition is deliberately vague

 Trace-DNA is defined as any sample where there is uncertainty that it may be associated with the crime event itself - so that it is possible that the transfer may have occurred before the crime event (innocent transfer) or after the crime event (investigator mediated)."

The statement of limitations Published c. 2000 when LCN was

introduced

- 1. Although a DNA profile has been obtained, it is not possible to identify the type of cells from which the DNA originated, neither is it possible to state when the cells were deposited.
- 2. It is not possible to make any conclusion about transfer and persistence of DNA in this case. It is not possible to estimate when the suspect last wore the [watch]2, if it is his DNA. Because the DNA test is very sensitive, it is not unexpected to find mixtures. If the potential origins of DNA profiles cannot be identied,
- 3. it does not necessarily follow that they are relevant to this case, since transfer of cells can occur as a result of casual contact.

Transfer methods – two types

- Aerosol DNA eg saliva from talking/shouting; housedust
- Sticky DNA deposited on surfaces, touched and transferred to other surfaces
- Latex gloves are 'high risk'
- DNA is everywhere in the environment and we currently have limited understanding about the risks, however the work carried out so far demonstrates these are real and serious.

Interpretation of evidence (framework of propositions is useful)

- The *sub-source* level refers to the strength of evidence of the DNA profile itself
- The *source* level refers to an evaluation of strength of evidence if a DNA profile can be associated with a particular body fluid, such as semen, or blood
- The *activity* level associates the DNA profile with the crime itself e.g. sexual assault
- The highest level deals with the ultimate issue of *guilt/innocence*.

The association fallacy

- The association fallacy is more serious than the better known prosecutor's fallacy
- Definition: A probability is transposed from one level of the framework of propositions to higher level. For example, the strength of evidence of a sub-source DNA profile may be directly applied to a source e.g. blood.
- The fallacy is to assume that the likelihood ratio of the sub-source level is the same as the source level. The uncertainty of the association of the DNA profile with its source will reduce the combined strength of evidence.

The naïve investigator effect

- A candidate is found on a DNA database that matches a crime-scene stain.
- The LR is very high, but there is no other evidence in the case. In fact the 'other' non-DNA evidence may point to the innocence of the suspect.
- The 'other evidence' is ignored and the prosecution is carried out solely on the basis of the DNA evidence
- I call this the 'swamping effect' (we are blinded by a very big number)

The 'hidden perpetrator effect'

- A crime-stain is recovered. The expectation is that DNA has been transferred during the course of an offence (such as sexual assault).
- In reality no such transfer of DNA is detected, although body fluid transfer cannot be eliminated. It is likely that DNA from innocent
- individuals will be implicated as potential offenders and the true perpetrator effectively eliminated from the enquiry.
- Phantom of Heilbron is a good example

Confirmation bias

- A well characterised psychological effect
- The forensic scientist ignores inconvenient evidence and 'fits' the evidence to the prosecution argument committing a series of fallacies (the compounded error effect) – especially linked to the association fallacy, where the fact of the DNA profile is explained at the 'activity level' or higher.
- All humans are innately biased
- Humans don't know when they are being biased
- How can we avoid innate human biasness?

An example of the compounded error effect. The miscarriage of justice "Adam Scott"

- Circumstances:
 - Serious sexual assault
 - DNA profile from vulval swab composed of victim, victim's boyfriend and unknown male.
 - Sperm were identified
 - Database search revealed Adam Scott
 - Arrested and incarcerated for several months pending trial.

The statement

- The association fallacy is illustrated:
- `It is estimated that the chance of obtaining matching DNA components if the DNA came from someone else unrelated to Adam Scott is approximately one in one billion (one billion is one thousand million). In my opinion the DNA matching that of Adam Scott has most likely originated from semen'.

Confirmation bias and more association fallacies

 The DNA detected in the sample recovered from (victim's name) vulval swab (GE2b) can be accounted for by a mixture of DNA from (victim's boyfriend) and Adam Scott. In my opinion these findings are what I would expect if Adam Scott had some form of sexual activity with (victim's name).

What do the defence say?

 Adam Scott has never visited Manchester in his life and was hundreds of miles away at the time of the offence.

What does the prosecution say?

 DNA doesn't lie - I have a huge number that proves he was responsible (no-one actually said this but this is what they thought)
Response of the scientist (more confirmation bias)

- In order to assess the overall findings in this case I have therefore considered the following propositions:
- Adam Scott had vaginal intercourse with (victim's name)
- Adam Scott has never been to Manchester and does not know (victim's
- name)

In my opinion, the scientific findings in relation to (victim's name) vulval swab provide strong scientific support for the view that Adam Scott had sexual intercourse with (victim's name) rather than he did not. However, <u>given</u> <u>the position of the semen matching Adam Scott</u> and an absence of semen on (victim's name) internal swabs, the findings do not specifically support vaginal penetration with ejaculation inside the vagina. They may also support vaginalpenile contact with external ejaculation or vaginal intercourse with no internal ejaculation.'

Activity

- Described as 'strong scientific support' without any explanation of the rationale
- i.e. it is completely 'made-up'
- There is a clear impression in the poorly worded report that the 1 in 1 billion statistic is transposed to the activity itself.
- This is perilously close to the ultimate issue of guilt vs innocence.

Muddled propositions are prosecution biased

Note that the alternatives evaluated in Scott were:

- Adam Scott had vaginal intercourse with (victim's name)
- Adam Scott has never been to Manchester and does not know (victim'sname).
 But there is no way to tell from a DNA profile if someone has visited
 Manchester.

Muddled propositions

• The problem with this arrangement is that there are effectively three different sets of hypotheses combined together. In a proper analysis,

these sets of hypotheses would be expressed and evaluated as follows:

– Set A:

Adam Scott had vaginal intercourse with (victim's name)

Adam Scott did not have vaginal intercourse with (victim's name)

– Set B

Adam Scott has been to Manchester

Adam Scott has never been to Manchester

– Set C

Adam Scott does know (victim's name).

Adam Scott does not know (victim's name).

The case illustrates an unfortunate way of thinking that is prosecution biased

- Note the DNA from Adams was from saliva (not sperm)
- The sperm came from the boyfriend (you cannot imply <u>all</u> the male DNA was from sperm)
- Resulted from a laboratory contamination incident
- Although the lab was reprimanded for the contamination incident, the statement itself was not subject to regulatory scrutiny and it appears that this kind of reporting is accepted practice in the UK

Evidence given by a scientist is not the same as scientific evidence

- The report is embellished with details that are clearly unscientific, yet the scientist attempts to justify the report as scientific (simply because he/she purports to being a scientist).
- The report is based on entirely on speculation, not science.





FEED BACK FROM EDNAP TO ENFSI MEMBERS

APRIL 2014 TBILISI

NIELS MORLING SECRETARY OF EDNAP



mRNA – NEW PUBLICATION





Forensic Science International: Genetics 8 (2014) 203-212

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

RNA/DNA co-analysis from human menstrual blood and vaginal secretion stains: Results of a fourth and fifth collaborative EDNAP exercise

C. Haas ^{a,*}, E. Hanson ^b, M.J. Anjos ^k, K.N. Ballantyne ^w, R. Banemann ^r, B. Bhoelai ^e, E. Borges ^j, M. Carvalho ^k, C. Courts ^h, G. De Cock ^d, K. Drobnic ^p, M. Dötsch ^r, R. Fleming ^f, C. Franchi ^x, I. Gomes ⁿ, G. Hadzic ^p, S.A. Harbison ^f, J. Harteveld ^e, B. Hjort ^v, C. Hollard ⁱ, P. Hoff-Olsen ^g, C. Hüls ¹, C. Keyser ⁱ, O. Maroñas ^t, N. McCallum ^m, D. Moore ^q, N. Morling ^v, H. Niederstätter ^o, F. Noël ^d, W. Parson ^o, C. Phillips ^t, C. Popielarz ^j, A.D. Roeder ^s, L. Salvaderi ^y, E. Sauer ^h, P.M. Schneider ⁿ, G. Shanthan ^g, D. Syndercombe Court ^c, M. Turanská ^u, R.A.H. van Oorschot ^w, M. Vennemann ¹, A. Vidaki ^c, L. Zatkalíková ^u, J. Ballantyne ^b





mRNA - MANUSCRIPT IN PREP





University of Zurich[™]

Institute of Legal Medicine

EDNAP mRNA profiling exercise 6

Skin cells

 \rightarrow Manuscript in preparation...







mRNA EXERCISE IN PREP





Institute of Legal Medicine



human mRNA quant assay

- → other mRNA quant assay (Zubakov, Kayser)?
- → Suggestion for a collaborative exercise on mRNA quantification (EDNAP mRNA exercise 7) at next EDNAP meeting

19 November 2014 in Zürich





IRISPLEX - EYE COLOUR



Accepted Manuscript

Title: Collaborative EDNAP exercise on the IrisPlex system for DNA-based prediction of human eye colour

Author: Lakshmi Chaitanya Susan Walsh Jeppe Dyrberg Andersen Ricky Ansell Kaye Ballantyne David Ballard Regine Banemann Christiane Maria Bauer Ana Margarida Bento Francesca Brisighelli Tomas Capal Lindy Clarisse Theresa E. Gross Cordula Haas Per Hoff-Olsen Clémence Hollard Christine Keyser Kevin M. Kiesler Priscila Kohler Tomasz Kupiec Adrian Linacre Anglika Minawi Niels Morling Helena Nilsson Lina Norén Renée Ottens Jukka U. Palo Walther Parson Vincenzo L. Pascali Chris Phillips Maria João Porto Antti Sajantila Peter M. Schneider Titia Sijen Jens Söchtig Denise Syndercombe-Court Andreas Tillmar Martina Turanska Peter M. Vallone Lívia Zatkalíková Anastassiya Zidkova Wojciech Branicki Manfred Kayser







Organisers: Chris Phillips and colleagues, Santiago de Compostela

Opted for the simplest format possible

Five cell-line DNA preps – each from a different continental region

Used the above as controls for the fullest range of alleles and compared to European 9947a DNA (where a sizable proportion of alleles are absent)

Additional mixed DNA of E ASN and EUR – Kings College donors at 3:1

Tasks: type 9947a and six DNAs assign ancestry or identify as mixture



INDEL TYPING OF AIMs



Indels: 3 no calls and 3 wrong calls (each in same lab) = >99.5% Concordance



I also	5	8	0	10	11	10	19	14	15	18	17	18	91	10	21	NOCALL	WRONG	TOTAL	DE COMPOS
Lab	0.0	0.00	0.00	0.(0	0.00	0.0	0.0	0.40	0.00	0 (0	0.00	0.00	21	0.00	21	NUCALL	whoma	TOTAL	1000
rs2307000	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1470
rs1610663	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-777
rs10630	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-196
rs1610965	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-881
rs35451359	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-3122
rs140837	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-648
rs1160893	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-659
rs2308203	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2011
rs33974167	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2929
rs1160852	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-593
rs1610884	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-798
rs2067280	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1193
rs2308067	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1871
rs4183	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-17
rs3054057	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2538
rs2307840	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1644
rs60612424	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-3854
rs3033053	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2275
rs16384	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-94
rs34611875	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-3072
rs1610859	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-772
rs3045215	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2313
rs25621	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-397
rs2307832	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1636
rs16343	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-51
rs3031979	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2431
rs34122827	0/0	0/0	0/0	0/0	0/0	0/2	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	2	2	MID-2264
rs133052	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2256
rs6490	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-128
rs4181	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-15
rs3030826	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2241
rs140708	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-419
rs1611026	0/0	0/0	0/0	0/0	0/0	0/1	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	1	1	MID-943
rs16438	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-159
rs2308161	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2005
rs16687	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-250
rs2307998	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1802
rs2307803	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1607
rs2307930	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1734
rs25630	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-406
rs2307582	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	3/0	3	0	3	MID-1386
rs2307922	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1726
rs11267926	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-3626
rs25584	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-360
rs2307799	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-1603
rs34541393	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0/0	0	0	0	MID-2719



SNP TYPING OF AIMs







ESTIMATION OF ANCESTRY WITH SNIPPER







ESTIMATION OF ANCESTRY WITH SNIPPER - MIXTURE



Mixture 1:3 (EAS:EUR)





SNP AND INDEL TYPING OF AIMs



Results so far analyzed indicate SNP typing at this scale of multiplexing is difficult to get familiar with - but those labs already acquainted with the 34plex SNaPshot test produced concordant results for most samples

Indel typing was readily adopted by all participants and the results were almost completely concordant; clear signals of imbalance in the mixed DNA

All labs reported correct ancestries for 5 individuals