AGENDA FOR THE EDNAP MEETING

ROME – 17 APRIL 2018

Expected duration: 09.00 - 17.00

Coffee: 10.30-10.50 – Lunch: 12.30-13.30 – Coffee: 15.30-15.50

Host: Alessandra La Rosa
Chairman: Niels Morling

Welcome	Alessandra La Rosa
Update on activities	
Methylated DNA and age exercise	David Ballard
Exercise no. 2 on mRNA typing with MPS	Cordula Haas
mtDNA quantification exercise - update	Arnoud Kal
Updates from other groups	
Plans in Germany to introduce forensic DNA phenotyping	Peter Schneider
ENFSI	Sander Kneppers
ISFG DNA Commission – Hierarchy of propositions	Peter Gill
Longest Uninterrupted Stretch (LUS) of STRs based on NGS typing	Peter Gill/Jodi Irwin
ISFG	Walther Parson
High quality DNA sequence database	Walther Parson
EMPOP	Walther Parson
STRidER	Walther Parson
The VISAGE project	Walther Parson
Other activities	Eirik Hanssen/
Body fluid recognition from microbial patterns using MPS	Peter Gill
MicroFLOQ Direct	Christopher Syn
Future activities	
Collaborative exercise on mtDNA MPS (long C-tracts)	Walther Parson
EDNAP meeting in the autumn of 2018	Walther Parson
Any other business	Niels Morling

EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

Rome, Italy

17 April 2018

Host: Alessandra La Rosa
Chairman: Niels Morling

A list of participants is attached.

Welcome
Alessandra La Rosa welcomed members to Rome.

Update on exercises

Second exercise on methylated DNA and age David Ballard
David Ballard presented the results of the second collaborative EDNAP exercise on age estimation by means of measurements of methylation of selected DNA positions. A draft of a manuscript is expected to be circulated within the next month (presentation attached).

Exercise no. 2 on mRNA typing with NGS Cordula Haas
Cordula Haas presented the results of the second collaborative EDNAP exercise on discrimination between various tissues and body fluids with mRNA determined with MPS. A draft of a manuscript is expected to be circulated before the next EDNAP meeting in October 2018 (presentation attached).

mtDNA quantification exercise Arnoud Kal
Arnoud Kal presented the plan for a collaborative exercise concerning quantification of mtDNA. Please contact Arnoud Kal if you want to participate (presentation attached).

Updates from other groups
ENFSI. Sander Kneppers
Sander Kneppers reported from the ENFSI DNA Working Group (presentation attached).

Plans in Germany to introduce forensic DNA phenotyping Peter Schneider
Peter Schneider gave an overview of the status of the considerations concerning introduction of forensic DNA phenotyping in Germany (presentation attached).

ISFG DNA Commission – Hierarchy of propositions Peter Gill
Peter Gill presented the considerations of the DNA commission of the ISFG concerning the hierarchy of propositions and evaluation of reporting (presentation attached).

Longest Uninterrupted Stretch (LUS) of STRs based on NGS typing Peter Gill
Peter Gill presented a suggestion for nomenclature of LUS-STRs with respect to the utility for probabilistic genotyping (presentation attached).

EMPOP update Walther Parson
Walther Parson gave a short update of the activities of EMPOP (presentation attached).

High quality DNA sequence database - STRidER Walther Parson
Walther Parson gave a short update on STRidER (presentation attached).

ISFG report Walther Parson
Walther Parson gave an update of the activities of the ISFG (presentation attached).

The EU supported project 'VISAGE' Walther Parson
Walther Parson gave an update on the VISAGE project (presentation attached).

Other activities
Body fluid recognition from microbial patterns using MPS Eirik Hanssen/Peter Gill
Erik Hanssen presented research about identification of tissue and body fluids by means of investigation of microbial DNA (presentation attached).

MicroFLOQ Direct Chris Syn
Chris Syn presented results of STR typing with microFLOQ kit (presentation attached).

Future activities
Collaborative exercise on mtDNA MPS (long C-tracts) Walther Parson
Walther Parson suggested to perform a collaborative exercise on mtDNA heteroplasmy. The Innsbruck laboratory plans to send out DNA extracted from forensically relevant samples (e.g. hair, blood, and saliva) from individuals with point and/or length heteroplasmy. The participating laboratories are invited to contribute to this exercise by providing sequence data of these samples using their preferred mtDNA sequencing method(s). This includes conventional Sanger and Massive Parallel Sequencing technologies. Walther Parson will propose an experimental plan and send it out for review and comments in May 2018. The plan is to begin the exercise in the autumn 2018.

Next meetings Niels Morling
The next EDNAP meeting will take place on 31 October 2018 in Innsbruck together with the meeting of the ENFSI Steering Group. It will be the 30 years anniversary of EDNAP, and there will be a modest celebration.

Any other business Niels Morling
Ingo Bastisch raised the question if the EDNAP group should be updated. The members were in favour of continuing to follow a permissive policy and continue to invite colleagues to participate in the meetings and the collaborative exercises.

Closing of the meeting Niels Morling
The meeting closed with sincere thanks to Alessandra La Rosa and all colleagues, who helped to organise the meeting.

The minutes and attachments are found at the EDNAP website:
http://www.isfg.org/EDNAP/Meetings:

- Agenda
- List of participants
- Presentations
  - David Ballard: Report on methylated DNA and age determination

- Cordula Haas: Report on the second collaborative exercise on mRNA NGS
- Arnoud Kal: Information on the planned mtDNA quantification exercise
- Sander Kneppers: Report from the ENFSI DNA Working Group
- Peter Schneider: Plans in Germany to introduce forensic DNA phenotyping
- Peter Gill: ISFG DNA DNA Working Group - Activity level propositions
- Peter Gill: LUS of STRs based on NGS typing
- Walther Parson: ISFG report
- Walther Parson: EMPOP report
- Walther Parson: STRidER report
- Walther parsons: The VISAGE project
- Eirik Hanssen: Body fluid recognition from microbial patterns using MPS
- Chris Syn: MicroFLOQ Direct
- Walther Parson: Information on the meeting 30 October 2018 in Innsbruck.

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Methylated DNA & Age Exercise

David Ballard

EDNAP, Rome 2018

KING'S College LONDON

Manuscript

- Introduction
- Methods
- Results Phase 1
  - Methylation control reproducibility per lab and marker

Methylation Standards 0-100% run on the MiSeq & PGM

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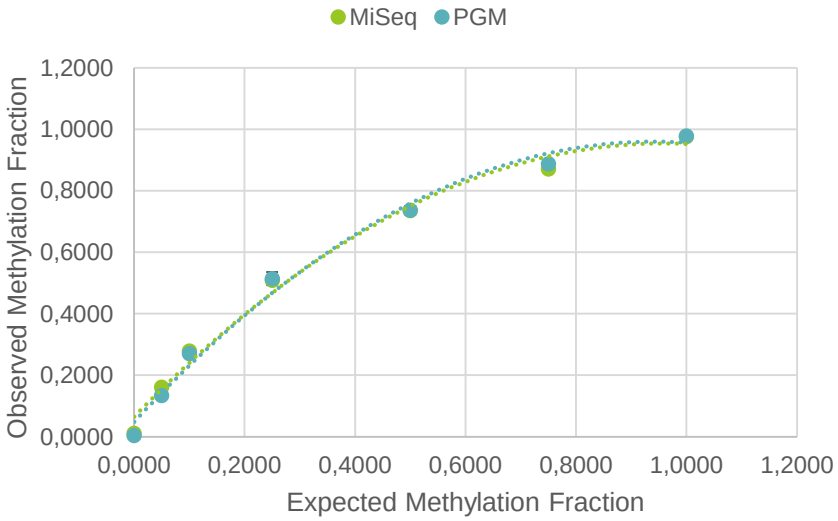

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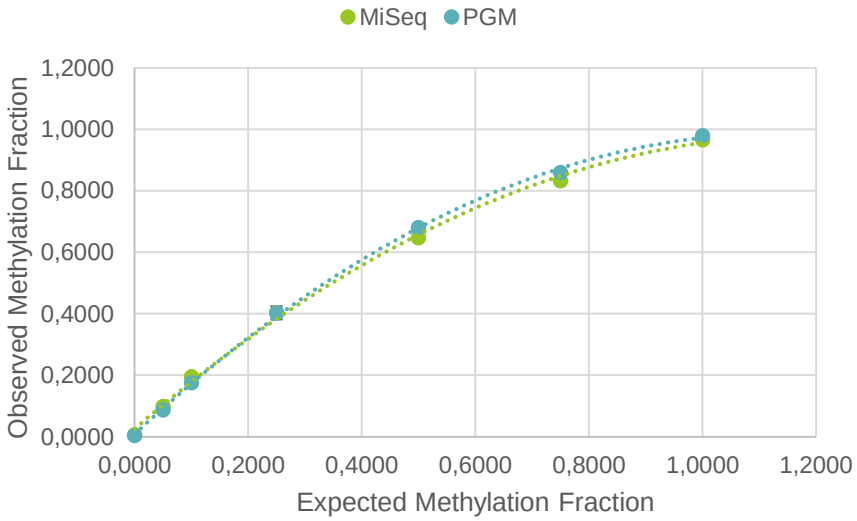

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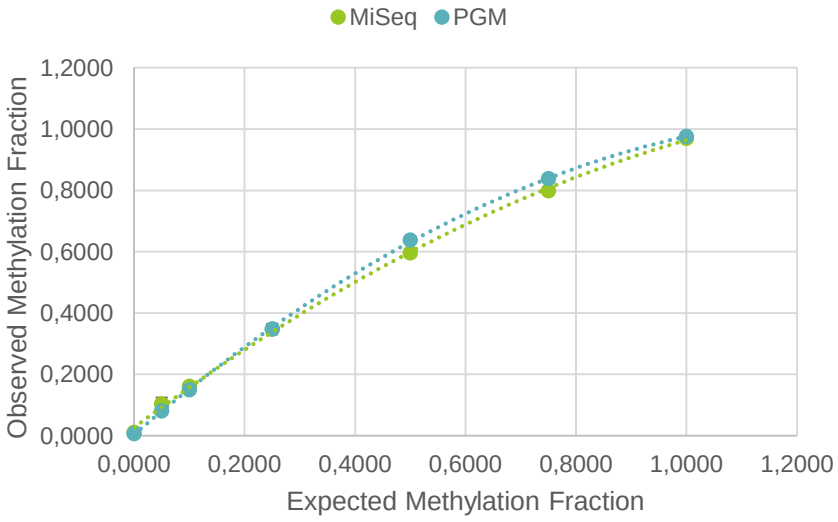

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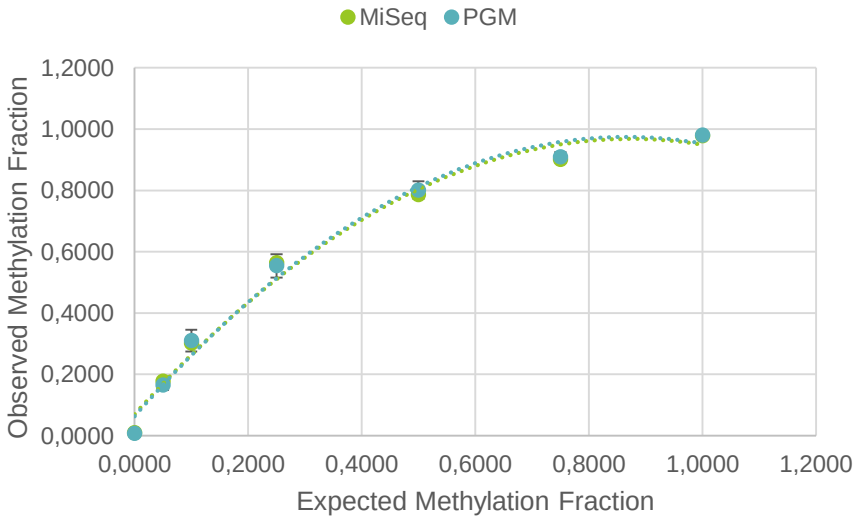

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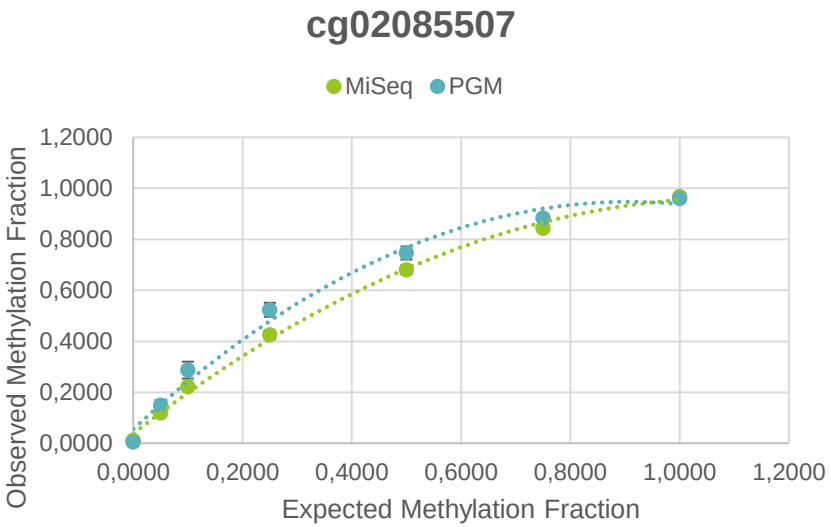
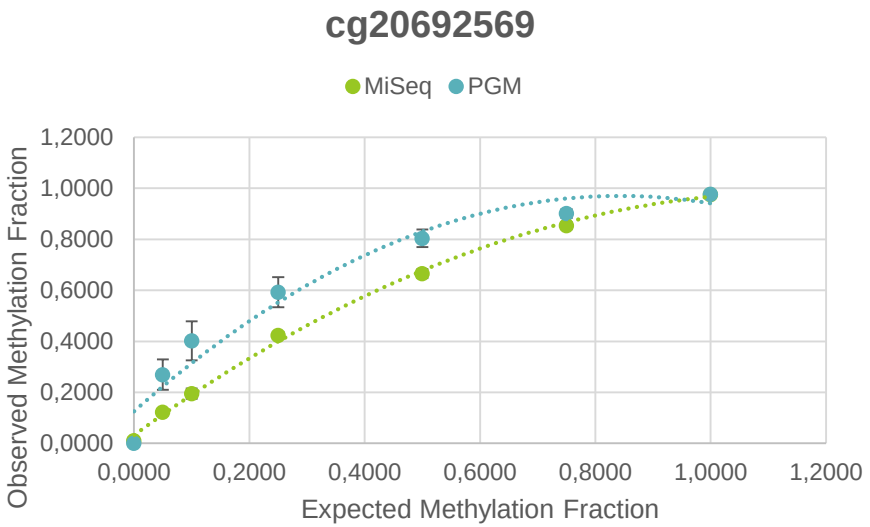
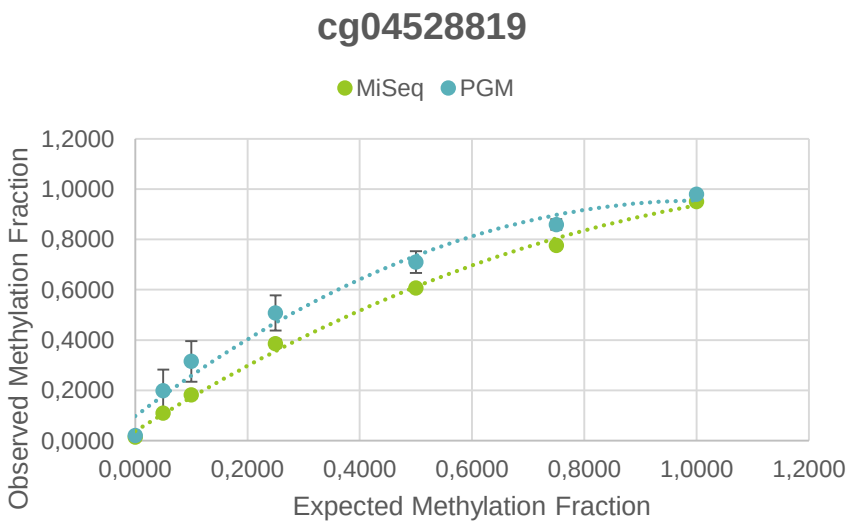
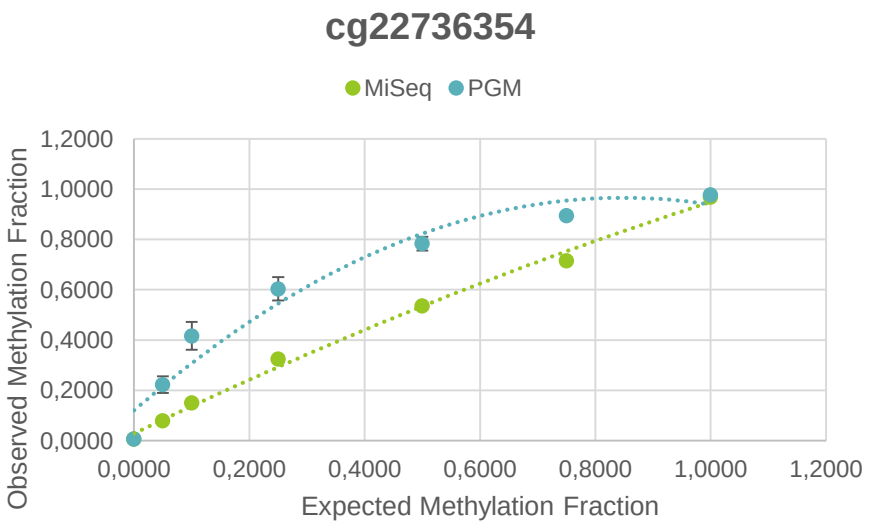
Methylation Standards 0-100% run on the MiSeq & PGM









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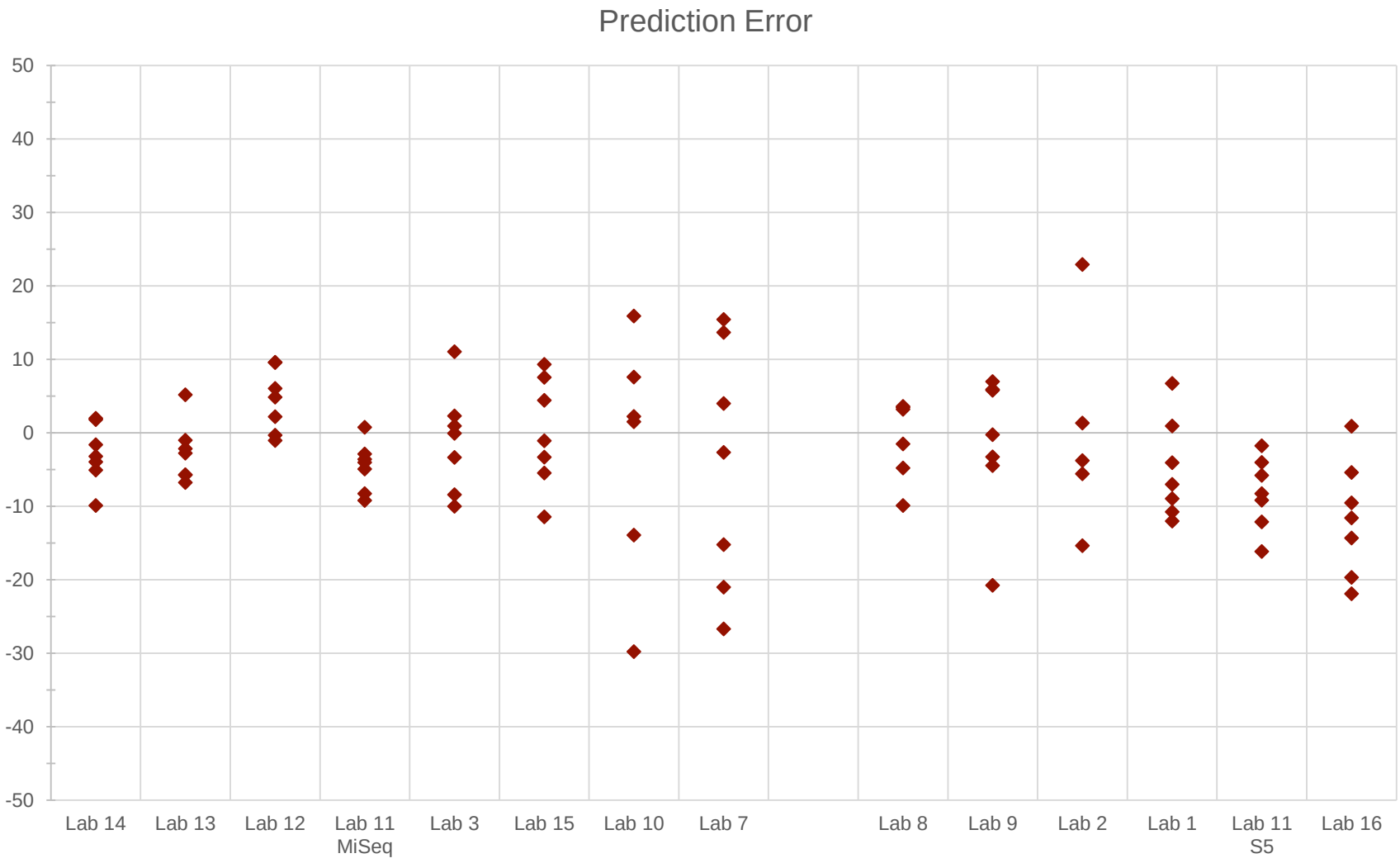
- Results Phase 2
  - Age prediction reproducibility per lab

Combined prediction results from EDNAP labs for samples A-G



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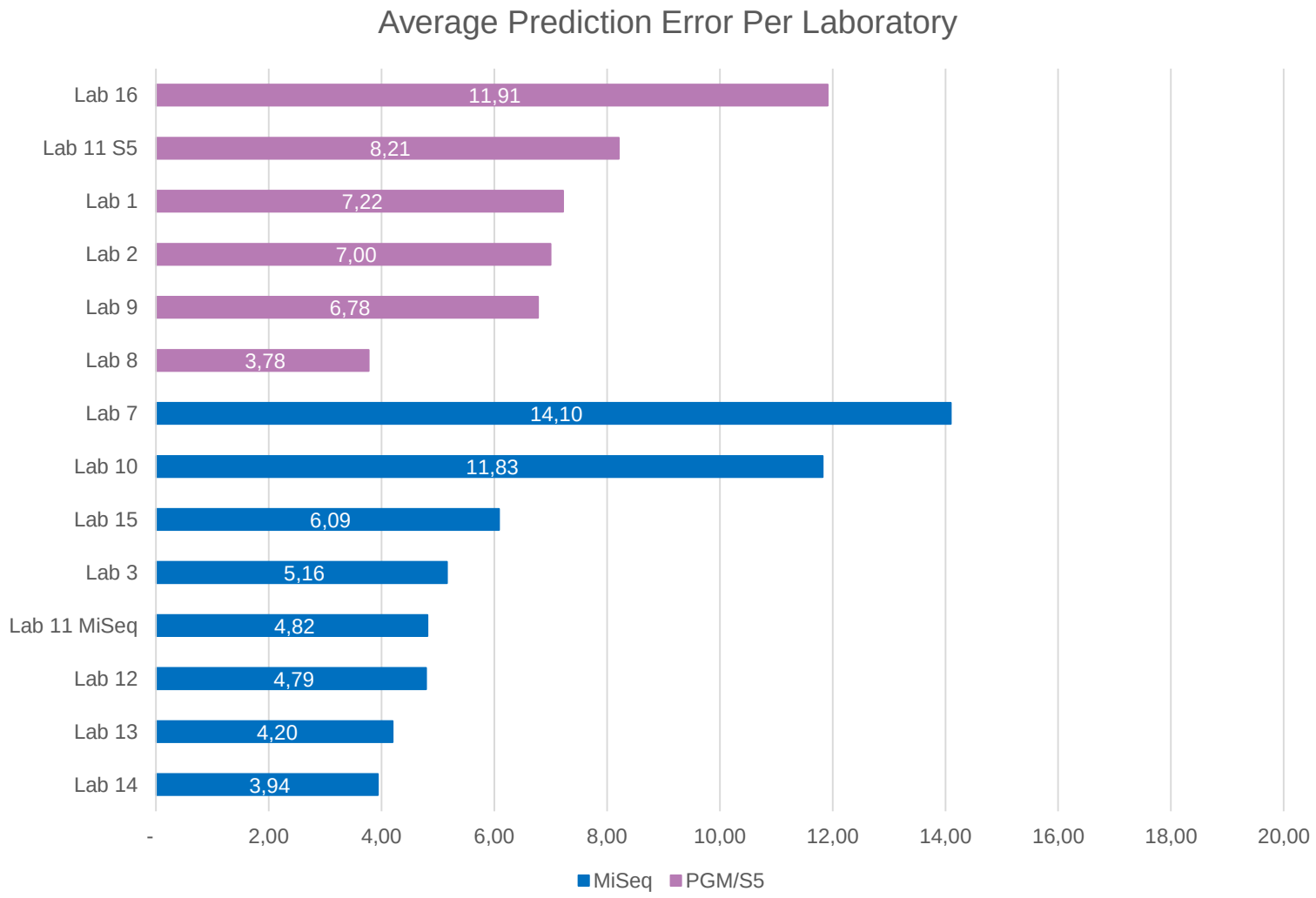

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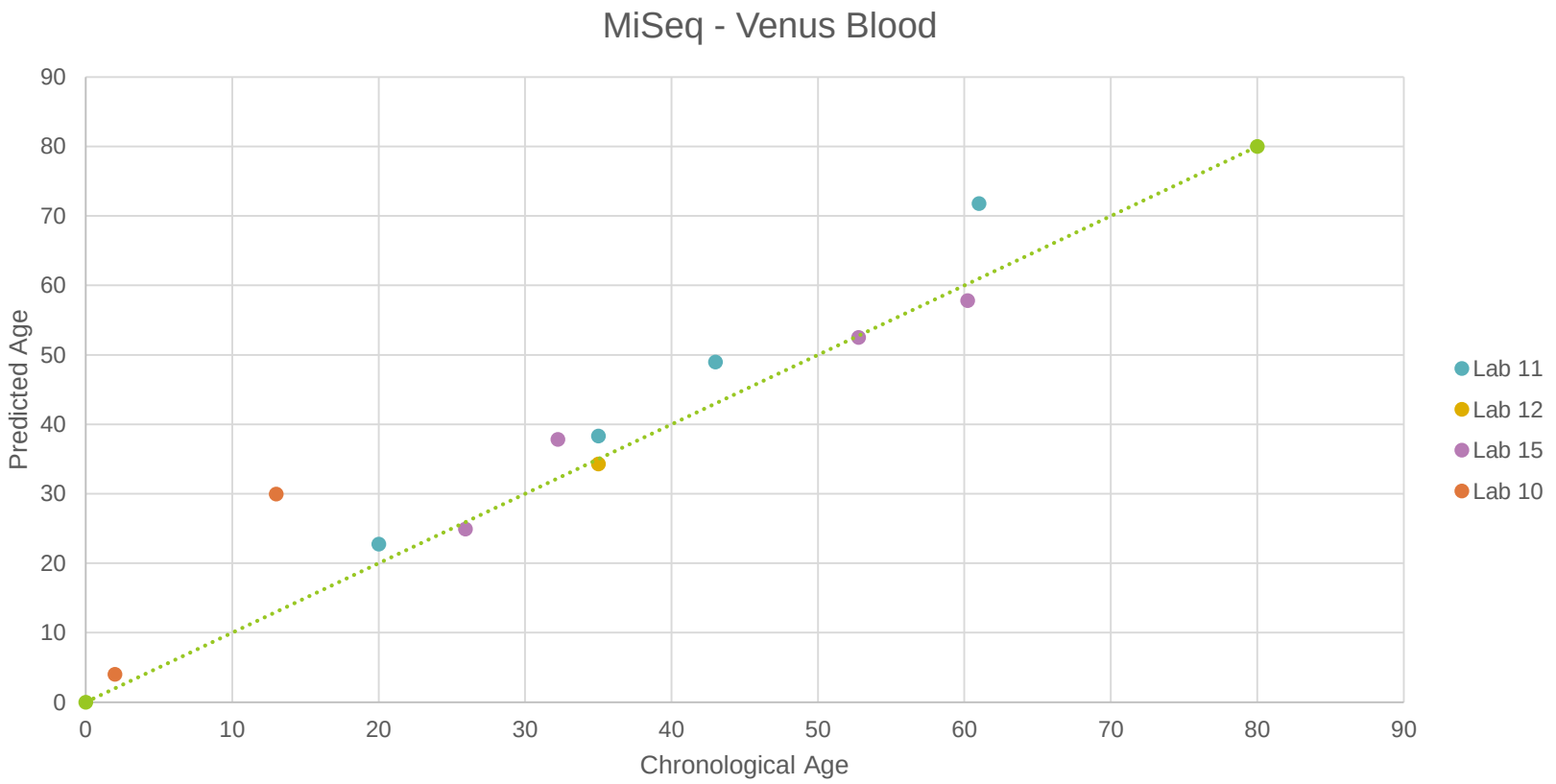
  - Blind prediction samples

Blind age predictions of extra MiSeq results



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  - Blind prediction samples
- Discussion/conclusion

Acknowledgments

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EUROFORGEN / EDNAP mRNA NGS exercise 2

Assay for body fluid/tissue identification and assignment to donor(s)

Cordula Haas / Sabrina Ingold / Guro Dørum
Erin Hanson / Jack Ballantyne

17. April 2018, Rome

EUROFORGEN Network of Excellence

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Forensic Science International: Genetics 34 (2018) 206–221

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research paper

Messenger RNA biomarker signatures for forensic body fluid identification revealed by targeted RNA sequencing

E. Hanson[a], S. Ingold[b], C. Haas[b], J. Ballantyne[a,c,*]

Targeted mRNA sequencing: Proof of concept

Forensic Science International: Genetics 34 (2018) 37–48

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research paper

Predicting the origin of stains from next generation sequencing mRNA data

Guro Dørum[a,1], Sabrina Ingold[a,1,*], Erin Hanson[b], Jack Ballantyne[b], Lars Snipen[c], Cordula Haas[a]

Probabilistic model

Collaborative exercise mRNA NGS part 1

Forensic Science International: Genetics 34 (2018) 105–115

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research paper

Body fluid identification using a targeted mRNA massively parallel sequencing approach – results of a EUROFORGEN/EDNAP collaborative exercise

S. Ingold[a,*], G. Dørum[a], E. Hanson[b], A. Berti[d], W. Branicki[e], P. Brito[f], P. Elsmore[g], K.B. Gettings[h], F. Giangasparo[i], T.E. Gross[j], S. Hansen[k], E.N. Hanssen[k], M.-L. Kampmann[l], M. Kayser[m], F.-X. Laurent[n], N. Morling[l], A. Mosquera-Miguel[o], W. Parson[p,q], C. Phillips[o], M.J. Porto[f], E. Pośpiech[e], A.D. Roeder[g], P.M. Schneider[j], K. Schulze Johann[r], C.R. Steffen[h], D. Syndercombe-Court[i], M. Trautmann[s], M. van den Berge[t], K.J. van der Gaag[t], J. Vannier[n], V. Verdoliva[d], A. Vidaki[m], C. Xavier[p], J. Ballantyne[b,c], C. Haas[a]

EUROFORGEN Network of Excellence

Collaborative exercise mRNA NGS part 2

- only MiSeq laboratories (1/2 library kit left from exercise 1)
- 2 separate assays
  1) targeted mRNA NGS approach for the identification of blood, saliva, semen, vaginal secretion, menstrual blood, skin
  2) cSNPs assay to associate specific mRNA transcripts to an individual
- RNA extraction (manual or kit), DNase treatment, quantification
- Protocols and primerpools were provided
- Laboratories analyzed 16 samples provided by UZH
- Results (FASTQ files) were collected and evaluated by UZH

Targeted mRNA NGS approach for body fluid/tissue identification and assignment to donor(s)

Body fluid/tissue	Gene	mRNA 33plex	cSNPs 35plex
Blood	ALAS2		
	ANK1		4
	SPTB		
	CD3G		
	CD93		3
	AMICA1		2
Semen	PRM1		
	PRM2		
	TGM4		4
	SEMG1		
	SEMG2		2
	KLK3		
Saliva	HTN3		
	HTN1		
	STATH		
	PRB3		
	PRB4		
	PRH2		
	MUC7		2
Vaginal	CYP2B7P1		
	DKK4		
	FAM83D		
	CYP2A6		
	CYP2A7		2
Menstrual	MMP10		2
	LEFTY2		
	MMP7		
	MMP11		
	SFRP4		
Skin	LCE1C		2
	CCL27		
	IL37		
	SERPINA12		
	KRT77		2
	COL17A1		3

Participating laboratories

Copenhagen, Denmark

Innsbruck, Austria

London, UK

Lyon, France

NFI, Netherlands

NIST, USA

Orlando, Florida, USA

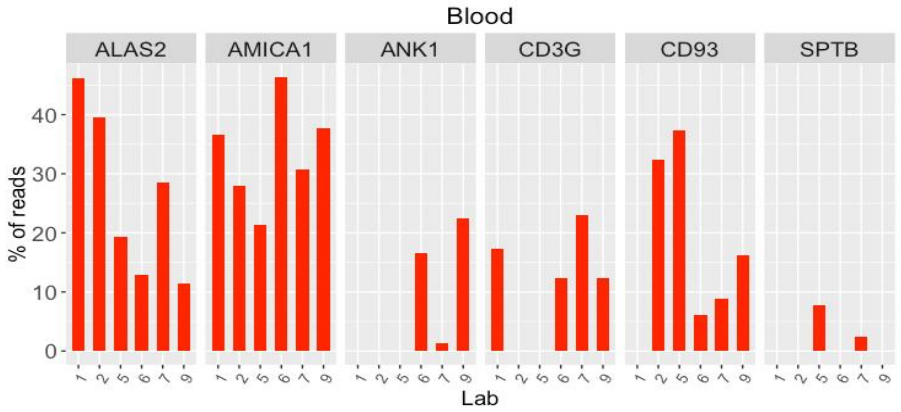
Rome, Italy

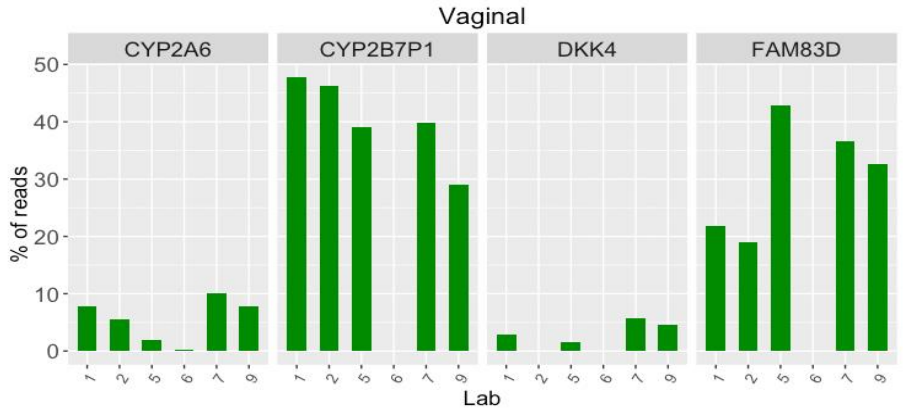
Rotterdam, Netherlands

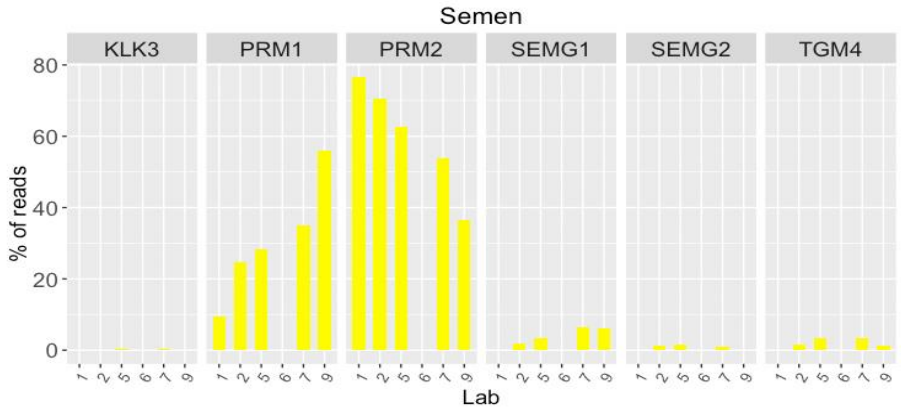
Zurich, Switzerland

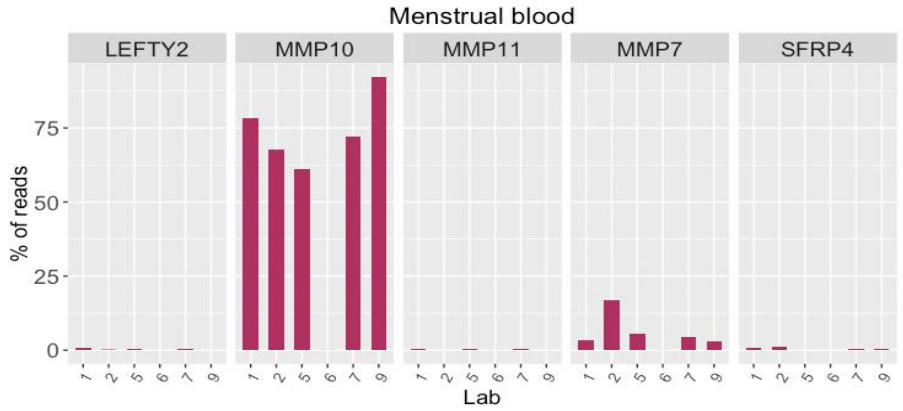
→ 1x analysis failed
→ 1x no data yet

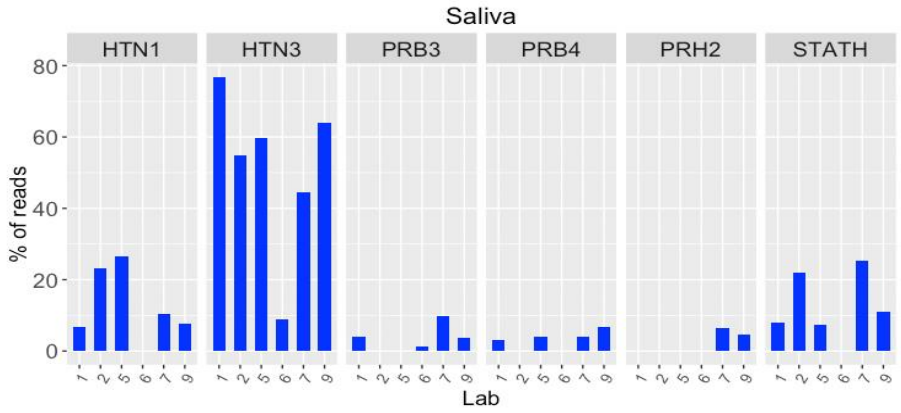
Comparison among laboratories (mRNA assay, single stains)

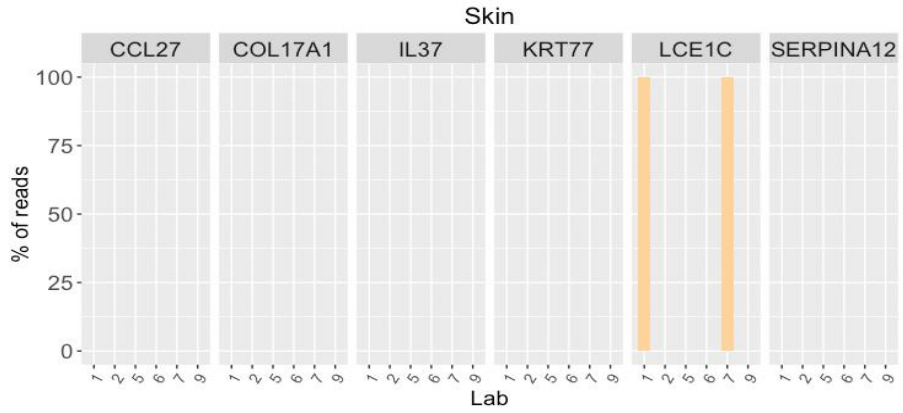


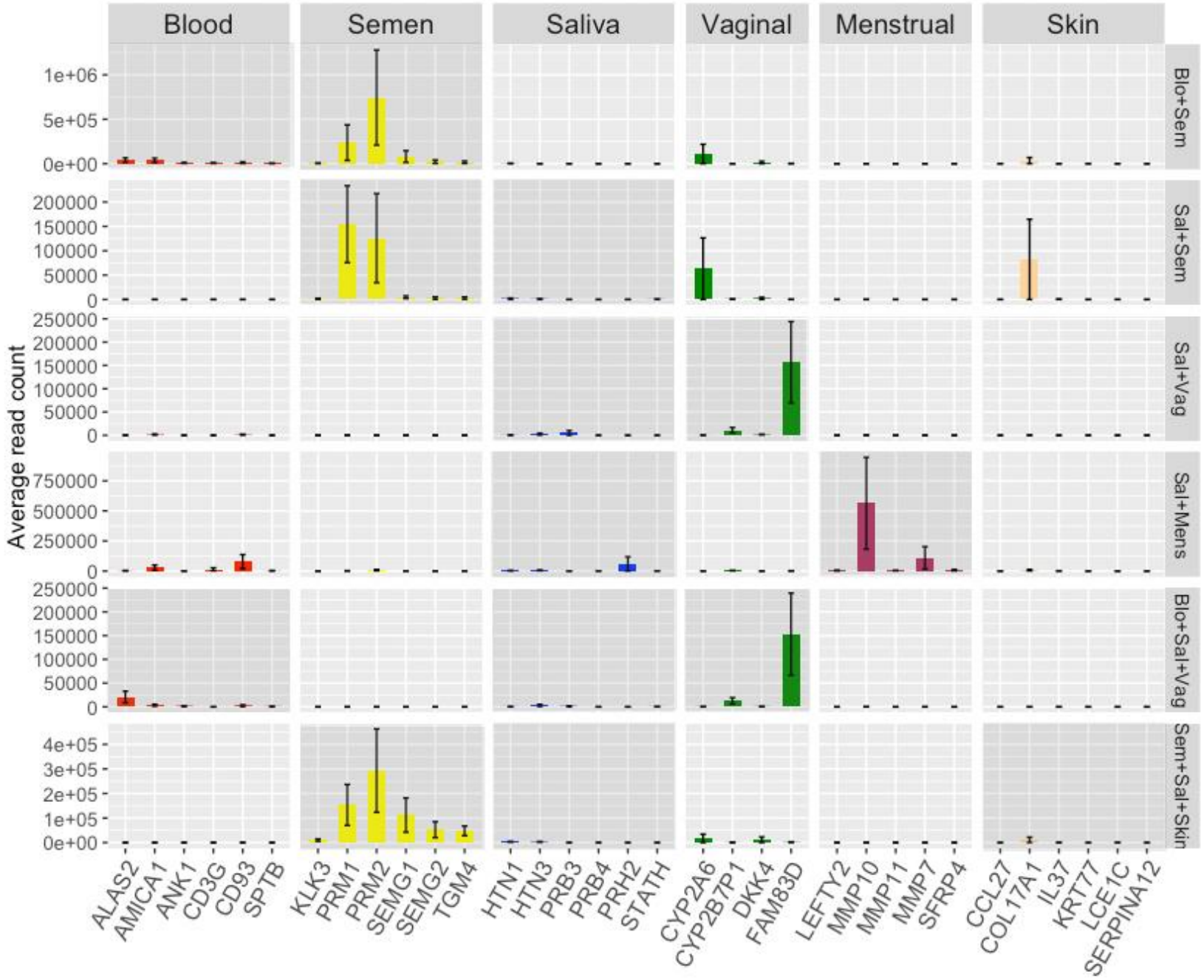











Blood
Semen
Saliva
Vaginal
Menstrual
Skin
Blo+Sem
Sal+Sem
Sal+Vag
Sal+Mens
Blo+Sal+Vag
Sem+Sal+Skin
Average read count
ALAS2
AMICA1
ANK1
CD3G
CD93
SPTB
KLK3
PRM1
PRM2
SEMG1
SEMG2
TGM4
HTN1
HTN3
PRB3
PRB4
PRH2
STATH
CYP2A6
CYP2B7P1
DKK4
FAM83D
LEFTY2
MMP10
MMP11
MMP7
SFRP4
CCL27
COL17A1
IL37
KRT77
LCE1C
SERPINA12

Results of mixed stains

Stain number 8: saliva (♂) – semen mixture (saliva 2 SNPs, semen 6 SNPs, remaining 1 saliva and 2 semen SNPs not discriminatory for this example)

Sample/ Marker	MUC7_1	MUC7_2	SEMG1	SEMG2_1	SEMG2_2	TGM4_1	TGM4_3	TGM4_4
Donor 1 (♂) (DNA)	CC	CC	TT	CC	AA	TT	GC	AA
Donor 2 (DNA)	CT	CG	TA	CA	AG	GG	GG	GG
saliva-semen mix (RNA)	CC C: 50’613 T: 59	CC C: 69’115 G: 14	TA T: 48’537 A: 35’771	CA C: 25’139 A: 11’172	AG A: 3625 G: 9056	GG G: 39’208 T: 5	GG G: 20’696 C: 1	GG G: 63’476 A: 100

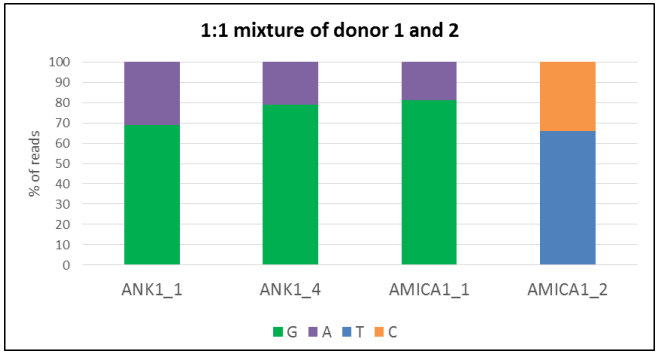
Results of mixed stains

Stain number 13: blood – blood mixture

(4 SNPs, remaining 7 SNPs not discriminatory for this example)

Sample/ Marker	ANK1_1	ANK1_4	AMICA1_1	AMICA1_2
Donor 1 (DNA)	GG	GA	GA	TT
Donor 2 (DNA)	GA	GG	GG	TC
blood-blood mix (RNA)	GA G: 32’825 A: 15’081	GA G: 20’650 A: 5613	GA G: 29’210 A: 6737	TC T: 93’229 C: 47’364

Sample/Marker	ANK1_1		ANK1_4		AMICA1_1		AMICA1_2	
Donor 1 (DNA)	G	G	G	A	G	A	T	T
Donor 2 (DNA)	G	A	G	G	G	G	T	C



Manuscripts in preparation

1) Association of a body fluid with a DNA-profile by targeted RNA/DNA deep sequencing - Proof of concept

- 188 DNA reference samples
  DNA cSNP data

- 45 samples
  DNA cSNP data
  RNA: TOP6, cSNPs

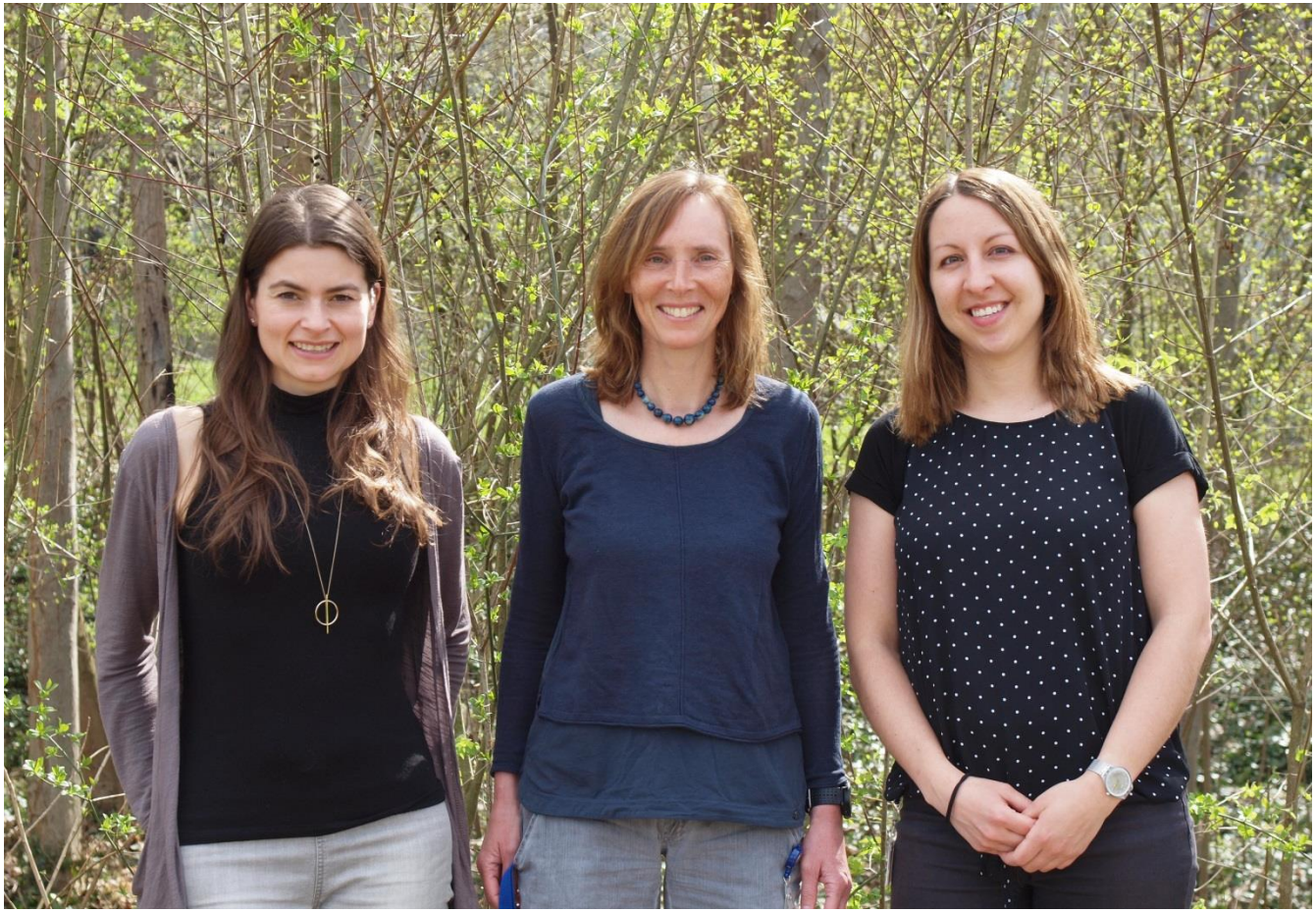
- 18 mixture samples
  DNA STRs: NGM Detect, ForenSeq
  DNA cSNP data
  RNA: TOP6, cSNPs

Manuscripts in preparation

2) Collaborative exercise mRNA NGS part 2

- 10 labs, 9 data sets
- 16 samples
- TOP6 and cSNP data
- Statistics

Thank you for your attention!



Netherlands Forensic Institute
Ministry of Justice

EDNAP mini-Exercise proposal mtDNA quant

Kris van der Gaag
Natalie Weiler
Titia Sijen
Arnoud Kal

Benefits of a good mtDNA quantification

- Establish if sufficient mtDNA is present in the sample

- Optimize the input for your favourite typing method
  - Sanger (mini-mito)
  - MPS (equalize input for multiple samples in one run)

  Note: mtDNA copy number varies for cell types, individuals

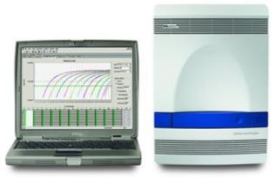
Strategy

Real time multiplex PCR assay:

- 40 cycles
- total & male based on Nicklas and Buel 2006
- Buffer system: TaqPath Multiplex Master Mix
- NEW: short mtDNA amplicon

DNA	Probe	Bp	Dye	Sensitivity
Total DNA	Alu Ya5	127 bp	VIC	0,5 pg/µl
Y DNA	DYZ5	137 bp	FAM	4 pg/µl
mtDNA	16533-180	217 bp	JUN	?
mtDNA	2502-2571	70 bp	ABY	?

! Need a 7500 !



Current status @NFI

Primers and probes total/Y/mt1 

Primers and probes mt2 

PCR protocol 

Optimizing primer concentrations 

Are you interested?

NFI provides:

- Primers and probes
- Challenging samples
- Protocols

You provide:

- Your own favourite sample
- Your own total/Y/mtDNA quantification method

Invitation sent out via email by Niels

Or email a.kal@nfi.minvenj.nl

Santiago	Chris Philips
Lausanne	Vincent Castella
Zurich	Cordula Haas
Insbruck	Walther Parson
NIST	Erica Romsos
Rotterdam	Athina Vidaki
Coimbra	Maria João Porto
Athene	Maria Vouropoulou
Kopenhagen	Niels Morling
Penn State Univ	Mitchell Holland

Netherlands Forensic Institute
Ministry of Justice

Update ENFSI DNA Expert Working Group activities

Alexander Kneppers
Chair ENFSI DNA Expert Working Group

NFI Division Biological Traces

ENFSI update EDNAP meeting Rome April 2018

ENFSI member/partner organisations

68 member organisations from 36 countries

2 standing committees

Quality and competence

Research and development

DNA working group

63 ENFSI (associate) member organisations from 37 countries

ENFSI Board news

- Best practice manual and guidelines templates > approved by board
- Amended constitution for presentation to the membership (Budapest/May)
- Education and Training Taskforce
  - QCC and RC members Bart Nys and Chanda Lowther-Harris
  - Michal Bovens board liaison
- Action plan 2018-2019
- Direct Grant 2018?

DNA working group

Annual ENFSI board meeting with workgroup chairs

Two steering group meetings DNA working group

Annual meeting of the working group (together with the European CODIS meeting and EDNAP meeting (European DNA Profiling group))

Group A: Quality Assurance

Group B: DNA Analysis Methods & Interpretation

Group C: DNA Database and Legislation

Group D: Automation & LIMS

Group E: Forensic Biology

Documents DNA working group

- DNA database management
- DNA Contamination prevention guidelines
- Minimum validation guidelines in DNA profiling
- Recommendations for the training of DNA staff
- Quality Assurance Program for DNA Laboratories
- DNA Pattern recognition and comparison (BPM)
- Internal validation of probabilistic software to undertake DNA mixture interpretation (BPM)

Surveys DNA working group

- DNA databases in Europe
- ENFSI Kit, automation and LIMS
- Rapid DNA

Rome 18-20 April 2018
Annual meeting ENFSI DNA Expert Working Group

Host Scientific Police Service of the Italian National Police

>100 persons attending
80 participants
20 company representatives
12 companies
31 countries represented
>60 speakers

New associate members

Division of Identification and Forensic Sciences, Israel	**Ron Gafny**
National Bureau of Expertises, Armenia	**Marine Baghdasaryan**
Forensic Institute of the Albanian State Police	**Ela Zaimi**

QA-subgroup

chairs Annick Delaire (France), Tom Heylen (Belgium)

- BPM:
  - Discussion at Spring meeting 2018
  - Scope to be drafted 2018
  - Different chapters shall be written by the different subgroups according to their expertise
- Update documents
  - Minimum Criteria for the Validation of Various Aspects of the DNA Profiling Process
  - Training of DNA staff

Methods analysis and interpretation sub group

chairs Peter Gill (Norway) and Walther Parson (Austria)

- Software analysis tools
  - DNAxs
  - EuroForMix with link to STRidER
  - TOASTR
  - Case Solver
- Activity level (ISFG recommendations)
- MPS updates

DNA database and legislation subgroup
chairs Dyan Daly (Ireland) and Izanda Puncule (Latvia)

- DNA Database Management Document
  - Feedback on audit
- Software tools
  - CODIS/Pedigree searching
  - Pedigree trees/ Rank Manager
  - STRmix
- Discussion on EU Data protection directive

Automation and LIMS subgroup

Chairs: Christina Forsberg (Sweden) and Shazia Khan (UK)

- Solutions for lab automation
  - MPS workflow approaches
  - Forensic Sample Workflows
- Rapid DNA
  - Questionnaire
  - User experiences
- LIMS solutions
- ENFSI Kit, Automation and LIMS inventory list
- Preparation for workshop on Rapid DNA London 21/22 May 2018

Forensic Biology and Casework subgroup
chairs Arnoud Kal (Netherlands) and Ricky Ansell (Sweden)

- Routine casework examples in and beyond DNA testing
- Scene, sampling and analytical strategies
- mRNA-profiling applications in casework

Direct Grant 2016 projects (Monopoly)

Period 2018- 2019

- Empowering Forensic Genetic DNA Databases for the Interpretation of Next Generation Sequencing Profiles (DNA.bases) N7 M2016
  - Ingo Bastisch

- Preparation of a Collaborative Exercise covering the forensic disciplines of DNA, document examination, fingerprint examination and handwriting examination
  - Jonathan Morris (Chair ENFHEX EWG)

Updates

- SWGDAM
- ISFG activities
- DNA SEQ EX
- STRbASE>STRidER STR Population Database
- GEDNAP > Proficiency testing
- EDNAP > collaborative exercises
- Update from ICMP, DVI and Missing Migrants

Contact ENFSI DNA Expert Working Group

Website

www.enfsi.eu

Chair
Sander Kneppers
NFI, the Netherlands, s.kneppers@nfi.minvenj.nl

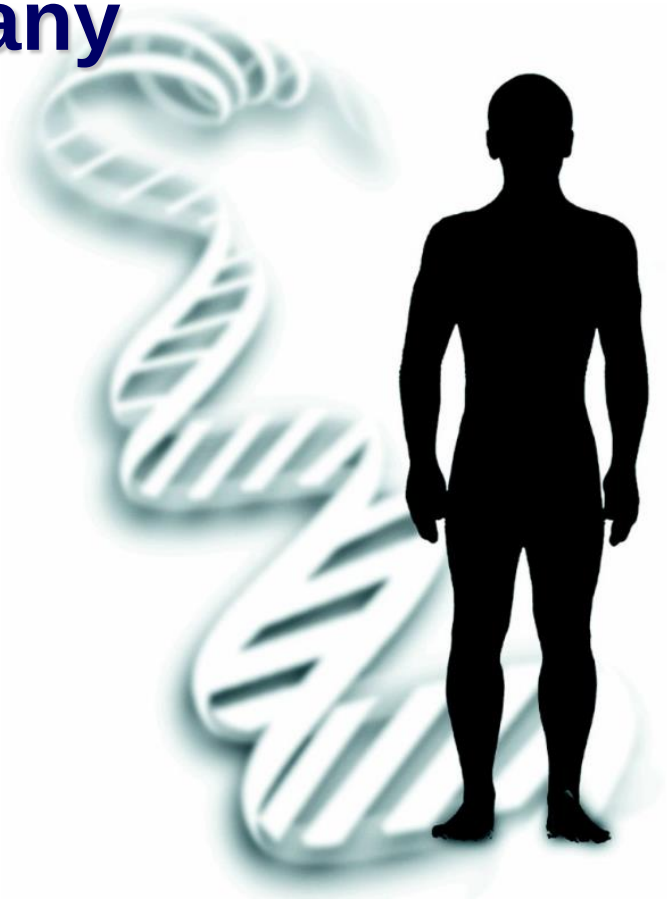
Vice chair
Livia Zatkalikova
Ministry of Interior, Slovakia, livia.zatkalikova@minv.sk

Secretary
Astrid Quak
NFI, the Netherlands, a.quak@nfi.minvenj.nl

UNIKLINIK KÖLN
Institut für Rechtsmedizin

Update on the introduction of forensic DNA phenotyping in Germany

Peter M. Schneider



EDNAP Meeting

Rome, April 17, 2018

FDP Implementation in Europe

- Three legislative scenarios for the use of FDP methodology criminal casework in Europe
  - The use of FDP is explicitly forbidden, mainly for reasons of privacy protection
  - The use of FDP is legally possible within a framework defined by laws
  - The use of FDP is not regulated
- Several countries are considering to create a legal basis for implementing FDP at present
  - Switzerland, Germany

Germany

- A 19-year-old student was raped and killed in Oct. 2016
- In December 2016, a young refugee from Afghanistan was arrested who confessed to the crime.
- Local police officials as well as the State Minister of Interior called for an amendment of the law to allow DNA-based prediction of externally visible traits.
- Subsequently, a group of critical social scientists published numerous articles raising concerns about the reliability and value of DNA phenotyping and the risk of discriminating against ethnic minorities

Issues raised against introducing FDP

From: Staubach, Nature (Correspondence) 545: 30, 2017

- The high probabilities for predicting visible traits and … ancestry … do not take into account the prevalence of (observed) traits in the German population.
- Probabilities for rare characteristics can drop to less than 50% when adjusted for prevalence.
- FDP-guided investigations will focus on rare characteristics: a positive test for dark skin … could much more efficiently (even if wrongly) narrow down an investigation than could a positive result for the common trait of light skin.
- Biogeographical testing is less reliable for individuals with mixed ancestry or for those from regions that are undersampled in the reference databases.
- Ancestry, ethnicity and appearance are therefore at risk of becoming conflated in policing practice.

Article in Press

Predictive values in Forensic DNA Phenotyping are not necessarily prevalence-dependent

Amke Caliebe, Michael Krawczak
Institute of Medical Informatics and Statistics, Kiel University, Brunswiker Strasse 10, 24105 Kiel, Germany
Manfred Kayser
Department of Genetic Identification, Erasmus MC University Medical Center Rotterdam, PO Box 2040, 3000 CA Rotterdam, The Netherlands

DOI: http://dx.doi.org/10.1016/j.fsigen.2017.11.006

- The relationship between the predictive values of FDP and the prevalence of the investigated appearance phenotype depends upon
  - (i) the extent to which causal factors (genetic and environmental) of the phenotype are accounted for by the prediction model used, and
  - (ii) the degree of inter-population variability, if any, of the remaining causal factors.
- If all causal factors are accounted for by the prediction model, then the predictive values would be the same in every population, irrespective of the prevalence of the phenotype in question.

The current legal basis of forensic DNA analysis in Germany

- The German Code of Criminal Procedures does not allow the investigation of DNA coding regions
- Section 81e:
  - (1) Material *(i.e. reference samples)* may also be subjected to molecular genetic examinations, insofar as such measures are necessary to establish descent or to ascertain whether traces found originate from the accused or the aggrieved person; in so doing the gender of the person may also be determined by examination. … Findings on facts other than those referred to in the first sentence shall not be made; examinations designed to establish such facts shall be inadmissible.
    Examinations admissible pursuant to subsection (1) may also be carried out on trace materials which have been found, secured or seized.

Current proposals to introduce FDP

- Several German Federal States have made proposals in the Federal Council
  - February 2017: Amendment to include only pigmentation markers (eye, hair, skin colour)
  - March 2017: Extension of the amendment to include also biogeographic ancestry and age prediction
  - The draft amendment included the need to provide funding of NGS equipment and consumables
  - The proposed amendment was rejected to allow further discussions in the parliamentary commissions
- 2018: The new federal government has plans to reconsider a revision of the Code to include some level of FDP (currently only pigmentation markers and age)
- A more general definition for the use of FDP in casework would be highly desirable!

Identification of the deceased?

§ 88 – Identification

(1) The identity of the deceased person shall be established before the autopsy. In particular, persons who knew the deceased person may be questioned to this end and measures of forensic identification applied. In order to establish identity and gender, cell tissue may be removed and subjected to a molecular and genetic examination. Section 81f subsection (2) shall apply *mutatis mutandis* to the molecular and genetic examination.

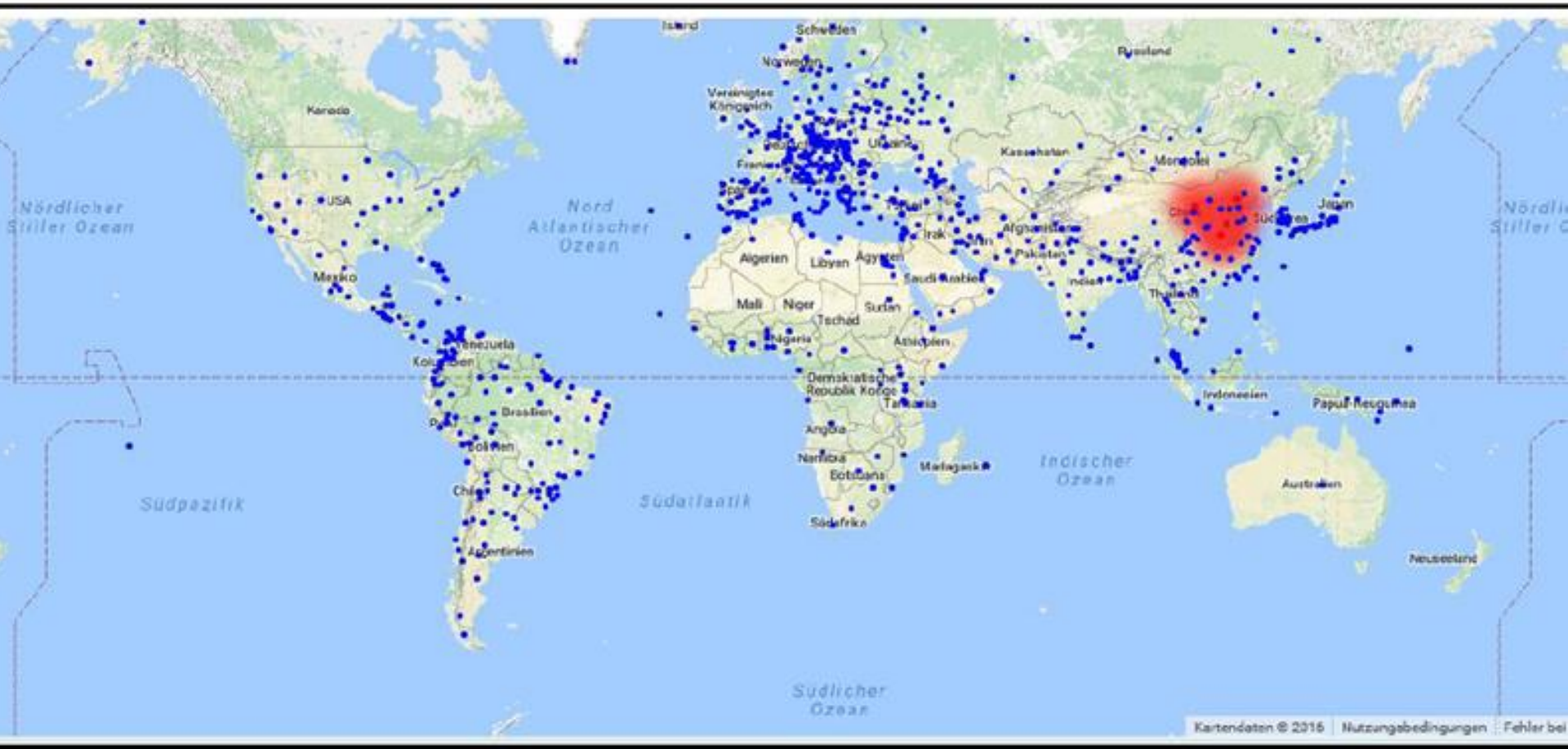
→ No reference is made to Section 81e (1) (restricting the scope of forensic genetic analyses)

The Rhine River Torso

- A male torso was discovered in July 2016 at the Rhine river banks carefully packaged in plastic bags together with clothing and shoes
  - STR profile for the DNA database and the missing persons / unidentified human remains database
  - Y-STR typing and YHRD search
  - mtDNA HV 1-3 sequencing and EMPOP search
  - Autosomal ancestry SNPs for biogeographic ancestry
  - Methylation analysis for age prediction
- Forensic STR typing of items from plastic bag

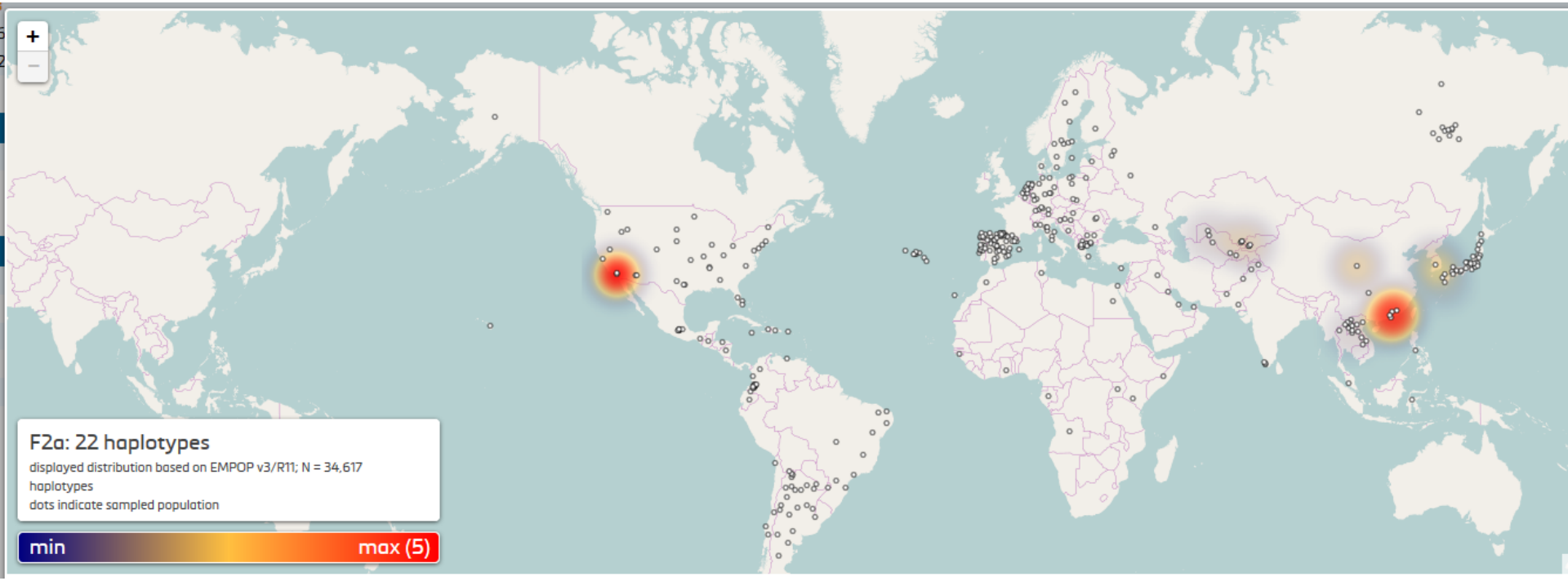
Torso case: Y-STR Analysis

- YHRD: haplotype of torso observed 7x among 29,900 East-Asian haplotypes, not observed in other metapopulations (n=188,200)



Torso case: mtDNA Analysis

- Haplotype from haplogroup **F2a**
- EMPOP: 22x among 34.600 haplotypes
  - All 22 haplotypes were from East Asia or Southern California (from persons with asian ancestry)

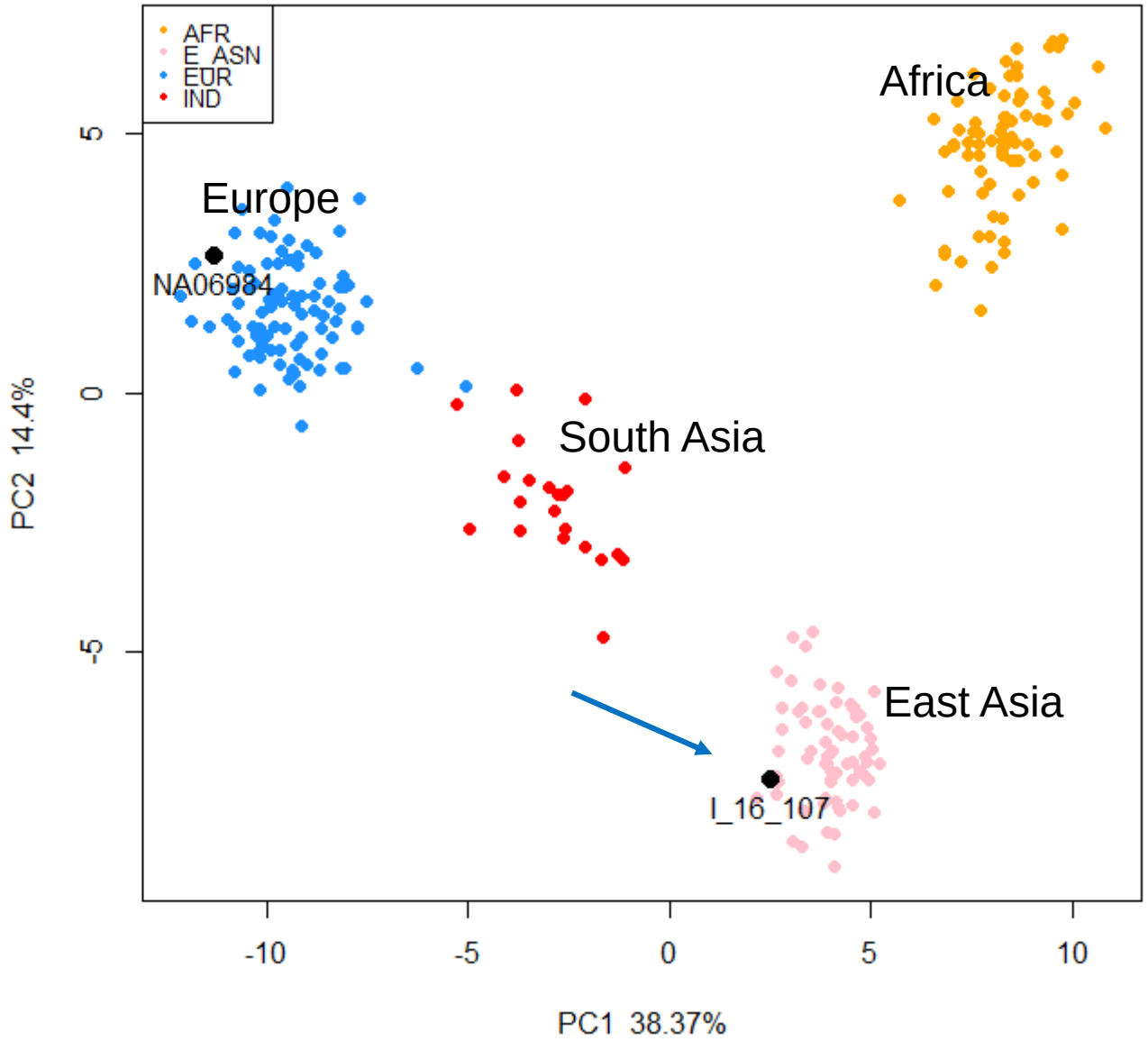


Torso case: autosomal AIM SNPs

- Analysis of 34plex and Eurasiaplex (23 SNPs) using SNaPshot
- Reference populations from Europe, Africa, South & East Asia using SNIPPER
- This profile is 444,900,402 times more likely EAST_ASIA than SOUTH_ASIA, and more than a billion (10^9) times more likely EAST_ASIA than AFRICA

→ These findings clearly support the Y-STR and mtDNA results

Torso case: autosomal AIM SNPs



Torso Case: Methylation analysis

- Post mortem blood sample from pericardium
- Sequence analysis of 5 CpG sites after bisulfite conversion from 10ng DNA,
  - ASPA, PDE4C, ITGA2B (Weidner et al. 2014)
  - ELOVL2 (Zbiec-Piekarska et al. 2015)
- Predicted age: 48-56 years (+/- 10 years)
- Potential pitfalls
  - decomposition of blood
  - No reference data for East Asian ancestry
  - Individual variance

  - Predicted age turned out to be incorrect by 20 years

The Rhine River Torso: case resolved

- The missing body parts were discovered in May 2017 in a small forest near a cemetry
  - STR typing confirmed that these remains were from the torso
- March 2018: remains identified
- 28-years old man from Central China, reported missing by his parents in 2017
  - Information request by Chinese Consulate in Germany to German police arriving 1.5 years after the body was discovered

Application scenario for FDP in casework

Which sample types are suitable for DNA phenotyping?

- The STR profile has the first priority – the national DNA database is the best chance for providing investigative leads
- Single source or major/minor profiles are best, multi-person mixtures are not suitable
- There must be some DNA left for testing
- The DNA sample in question should be relevant for the case
- Severity of the crime?

The hierarchy of propositions and evaluative reporting

An introduction

Principles of interpretation

1. To consider the uncertainty of any given hypothesis it is necessary to consider at least one alternative hypothesis

2. Scientific interpretation is based on evaluating the probability of the evidence given the hypothesis

3. Scientific interpretation is conditioned by the framework of circumstances within which the competing hypotheses are to be evaluated, i.e. the non-scientific evidence

$$LR = \frac{\Pr(E|Hp, I)}{\Pr(E|Hd, I)}$$

Propositions should be set before results of tests are known

References

CAI

Cook R, Evett IW, Jackson G, Jones PJ, and Lambert JA. A model for case assessment and interpretation. Science and Justice (1998) 38: 151-156

Cook, R, Evett IW, Jackson G, Jones PJ & Lambert JA. Case pre-assessment and review in a two-way transfer case. Science & Justice (1999) 39: 103–111

Evett IW, Jackson G, & Lambert JA. More on the hierarchy of propositions: exploring the distinction between explanations and propositions. Science & Justice (2000) 40: 3–10

Hierarchy of **issues**

Level		Example
3	**Offence**	**Role of the jury / court - truth of the proposition**
2	**Activity**	**How did the DNA get there?**
1	**Source**	**What body fluid has the DNA originated from?**
0	**Sub-source**	**Origin of DNA - Whose DNA is it?**

Investigative vs Evaluative reporting

Forensic scientists have a dual role as 'investigator' and 'evaluator':

a) To provide investigative leads, typically by searching a national DNA database (no suspect yet).

b) To provide evidence of the value of a comparison in the context of the case when a suspect is found.

From the ENFSI guideline:

'Evaluative reports for use in court should be produced when two conditions are met:

1. The forensic practitioner has been asked by a mandating authority or party to examine and/or compare material (typically recovered trace material with reference material from known potential sources).

2. The forensic practitioner seeks to evaluate results with respect to particular competing propositions set by the specific case circumstances or as indicated by the mandating authority.'

Roles

EVALUATIVE

Level 3 Did he commit the offence?

Level 2 Did he do the activity?

Level 1 Is he/the item the source of the trace material?

Sublevel 1 Is he/the item the sub-source of the test result?

INVESTIGATIVE

Level 3 What offence has been committed?

Level 2 What activity took place?

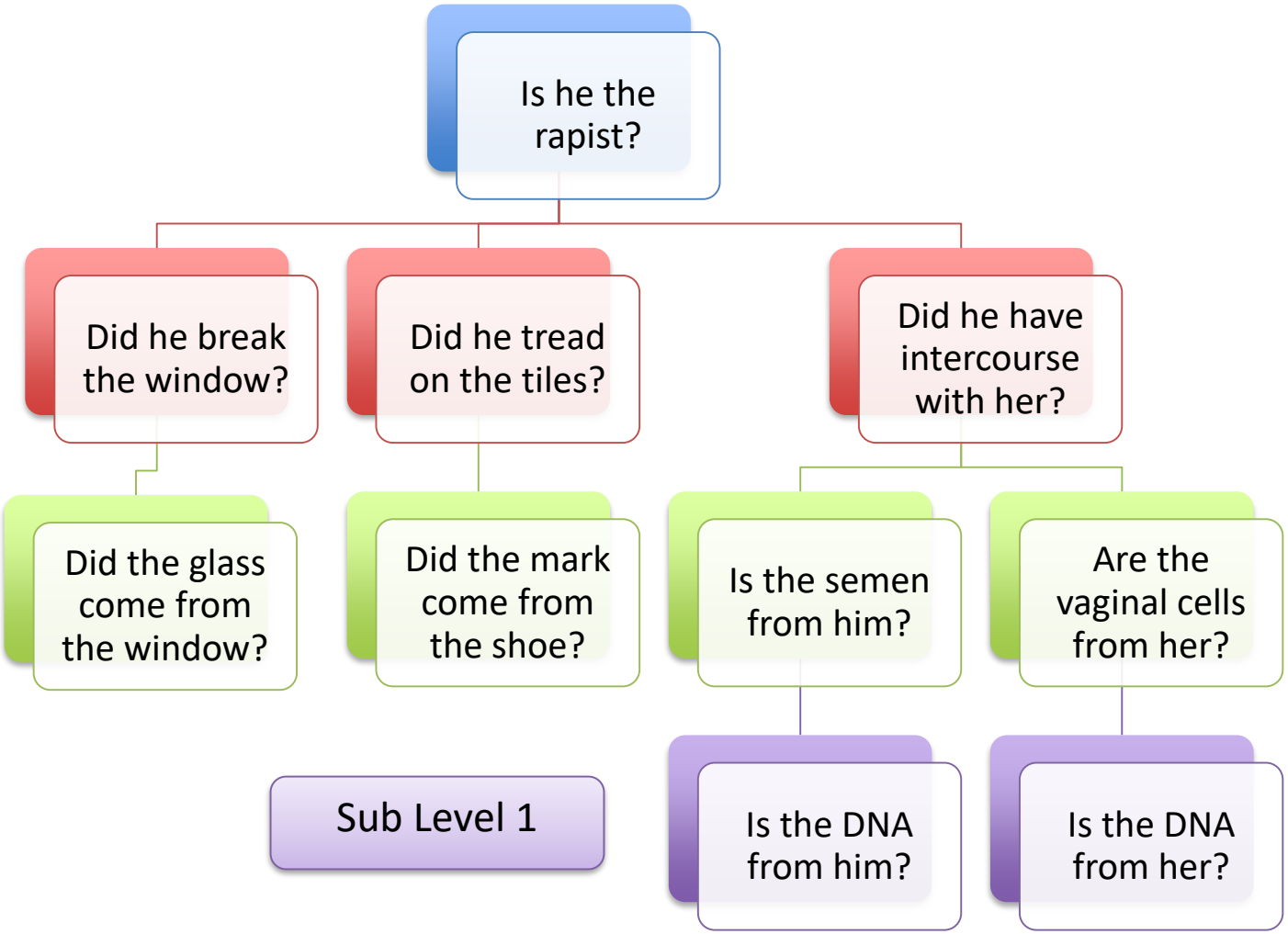
Level 1 What is the source of the trace material?

Sublevel 1 What is the sub-source of the test result – is there a match on the national DNA database?

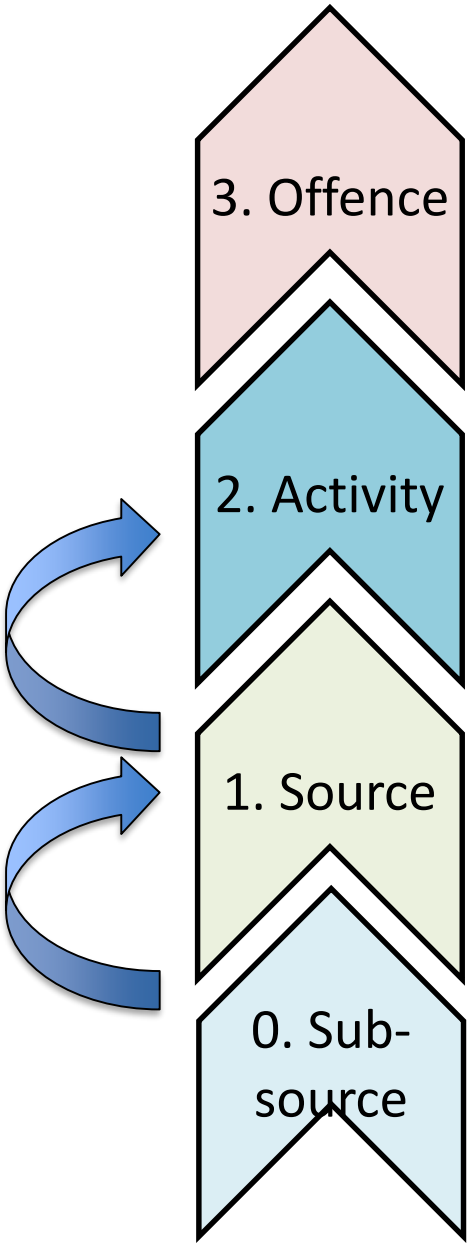
Hierarchy of **propositions**

Level		Example
3	**Offence**	**Guilt or innocence. e.g. *Hp* "The suspect raped the victim"** ***Hd*: Some other man raped the victim**
2	**Activity**	**e.g. *Hp* "The POI had intercourse with the complainant"** ***Hd*: Some other man had intercourse with the complainant**
1	**Source**	**e.g. *Hp* "The semen came from the POI"** ***Hd*: The semen came from some other man**
0	**Sub-source**	**e.g. *Hp* "The DNA in the sample came from the POI"** ***Hd*: The DNA in the sample came from another man**

A Case Example



Hierarchy of propositions



The **offence level** is usually not the realm of the forensic scientist. This is for the court to decide based on all the available information presented

There may be occasions when a reporting scientist can address the **activity level**, based on their experience and available literature – involves issues of DNA transfer and persistence, background levels of DNA and 'contamination'

In some instances it may be possible to step up to body fluid attribution & hence report at the **source level** – lab tests

A DNA reporting analyst will spend most of their time at the **sub-source level**

Hierarchy of propositions (rules)

- The likelihood ratio given at sub-source level (i.e. the strength of evidence if a DNA profile has come from a suspect) cannot be carried over to another level in the hierarchy e.g. to source or activity level

- If we do not evaluate activity level propositions then there is an implicit danger that a court will confuse the strength of evidence given at sub-source with that at activity level

Activity Level Reporting

- Adds value to the court process
- Ensures most informed individuals are providing an opinion e.g. on issues of transfer and persistence, background levels of DNA , contamination
- Reduces risk of mis-interpretation e.g. associate source evaluation with activity

Activity data

Mr A carried out the activity

- Studies on transfer of trace evidence
- Studies on persistence of trace evidence

Some other person carried out the activity

- Background levels of trace evidence
- Dataset on types of trace evidence
- Dataset on secondary transfer

Challenges and discussion points

- Activity level reporting is dependent upon data (we should not rely on raw ‘experience’)
- Although there is a body of literature – we need to make sure that the data are relevant to the case
  - Can results obtained with method (a) be applied to method (b)?
  - Complete transparency is needed
  - Knowledge bases are needed
- Define the limitations
- Define educational requirements

LUS nomenclature with respect to utility for probabilistic genotyping

Peter Gill and Jodi Irwin

Forensic Science International: Genetics 34 (2018) 197–205

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Use of the LUS in sequence allele designations to facilitate probabilistic genotyping of NGS-based STR typing results

Rebecca S. Just*, Jodi A. Irwin

DNA Support Unit, Federal Bureau of Investigation Laboratory, 2501 Investigation Parkway, Quantico, VA, 22135, USA

Sequence reads instead of peak height

Forensic Science International: Genetics 31 (2017) 105–110

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research paper

Open source software EuroForMix can be used to analyse complex SNP mixtures

Øyvind Bleka[a], Mayra Eduardoff[b], Carla Santos[c], Chris Phillips[c], Walther Parson[b,d], Peter Gill[a,e,*]

[a] Department of Forensic Sciences, Oslo University Hospital, Norway
[b] Institute of Legal Medicine, Medical University of Innsbruck, Austria
[c] Forensic Genetics Unit, Institute of Forensic Sciences, University of Santiago de Compostela, Spain
[d] Forensic Science Program, The Pennsylvania State University, PA, USA
[e] Department of Clinical Medicine, University of Oslo, Norway

The LUS principle

- Advantages
  - Simple
  - A natural nomenclature that can be derived from knowledge of the sequence.

Table 1
LUS length reference regions for the STR loci typed by the ForenSeq assay.

Locus	Example Alleles Showing LUS Length Reference Regions[a]	CE/Repeat Unit Designation	LUS Concept Allele Designation
CSF1PO	(ATCT)7 ACCT (ATCT)3	11	11_7
D10S1248	(GGAA)6 GTAA (GGAA)7	14	14_7

  - Stutters are calculated as -1_-1 entities

- Disadvantages
  - Doesn’t take account of all sequence variation, especially with complex STRs like D21S11, D2S1338
  - In addition, some alleles produce stutter that is not consistent with -1_-1 if it arises from outside the LUS region. Ie the stutter repeat is -1_0 compared to parent allele.
  - However, these stutter variants are low level and don’t seem to have much impact

A simple nomenclature to take account of all sequence variation

- D21S11 has four variants at allele 32_12

Allele Sequence[a]	*LUS*[b] *Length*	LUS Concept Allele Designation	CE/Repeat Unit Designation
(TCTA)7 (TCTG)6 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)11	*11*	32_11	32
(TCTA)8 (TCTG)5 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)11			
(TCTA)9 (TCTG)5 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)10	*10*	32_10	
(TCTA)8 (TCTG)6 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)10			
(TCTA)5 (TCTG)6 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)13	*13*	32_13	
(TCTA)6 (TCTG)5 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)13			
(TCTA)6 (TCTG)6 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)12	*12*	32_12	
(TCTA)7 (TCTG)5 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)12			
(TCTA)5 (TCTG)7 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)12			
(TCTA)8 (TCTG)4 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)12			

- The nomenclature can be expanded to subdivide LUS genotypes further: 32_12_1; 32_12_2; 32_12_0 and 32_12_0

What about stutters?

These alleles fall in stutter positions, but of course , by observation in a case stain we still don't know if they are alleles or stutters even if we sequence them

(TCTA)7 (TCTG)5 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)11	*11*	31_11
(TCTA)6 (TCTG)6 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)11		

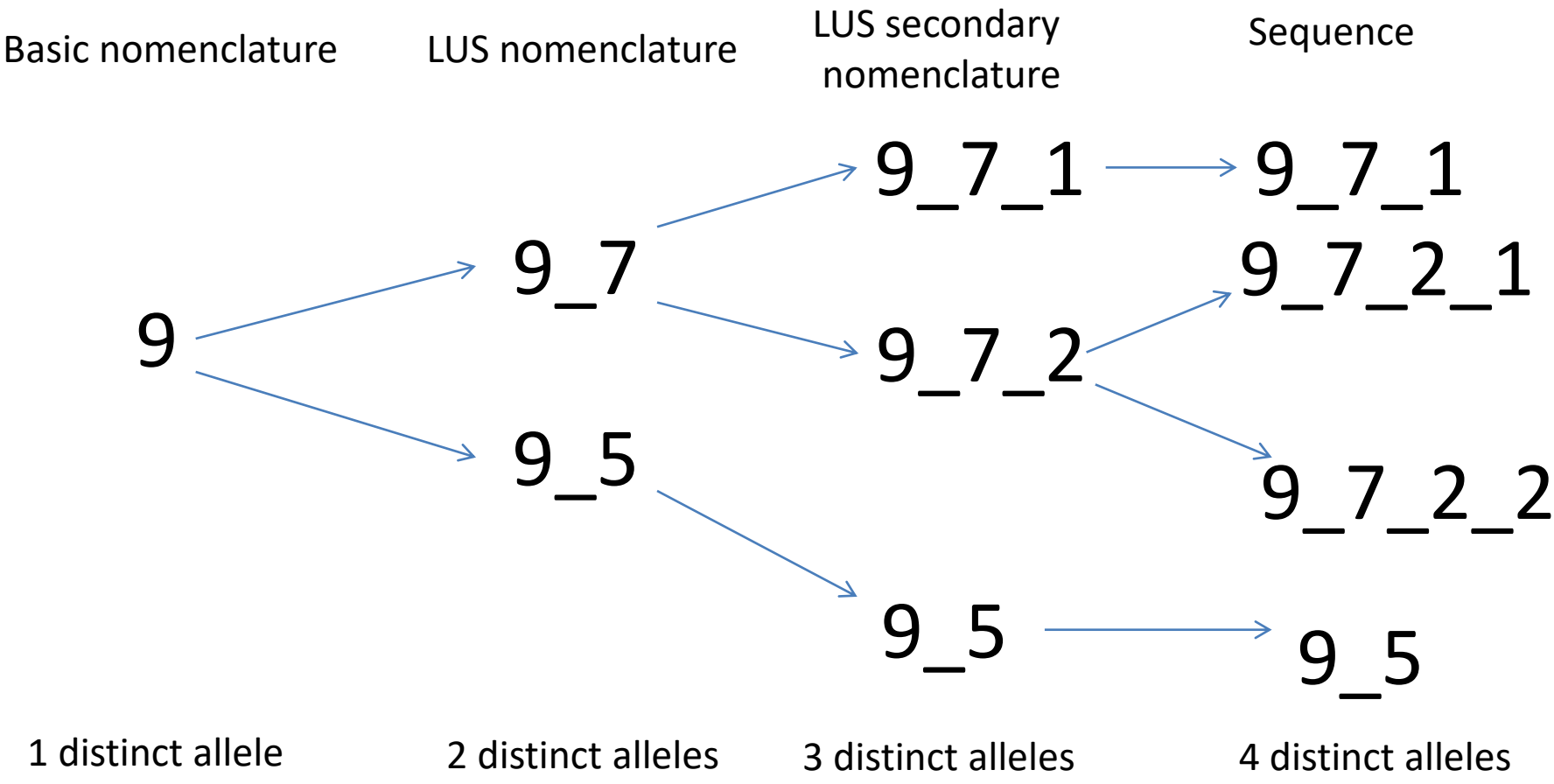
We can sum the reads that comprise 31_11

Allele Sequence[a]	*LUS*[b] *Length*	LUS Concept Allele Designation	CE/Repeat Unit Designation
(TCTA)7 (TCTG)6 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)11	*11*	32_11	32
(TCTA)8 (TCTG)5 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)11			
(TCTA)9 (TCTG)5 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)10	*10*	32_10	
(TCTA)8 (TCTG)6 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)10			
(TCTA)5 (TCTG)6 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)13	*13*	32_13	
(TCTA)6 (TCTG)5 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)13			
(TCTA)6 (TCTG)6 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)12	*12*	32_12	
(TCTA)7 (TCTG)5 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)12			
(TCTA)5 (TCTG)7 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)12			
(TCTA)8 (TCTG)4 (TCTA)3 TA (TCTA)3 TCA (TCTA)2 TCCA TA (TCTA)12			

Nomenclature

- A more precise nomenclature can be achieved using the LUS designation with a third or more identifiers.
- This will improve discriminating power further.
- There are considerable attractions to using a simple nomenclature that is ‘natural’
- It’s doubtful any nomenclature is perfect
- We don’t need a perfect system so long as it is consistent
- A programmable method should be achievable

Hierarchical nomenclature



Predominantly natural nomenclature

- LUS is straightforward
- Decisions on secondary, tertiary quaternary methods need to decided per locus (but once decision is made the nomenclature is natural)
- Some sequences may need an artificial non-natural designator, however.
- Whole process of nomenclature can be automated to take account of new sequences – but this needs to be centralised if non-natural designations are used for sequences
- Don’t forget all stutters need allelic designations too even if true alleles are never observed!
- Doesn’t have to be a perfect system for probabilistic genotyping.

Tiny or very large numbers

- The Forenseq/LUS system gives a match probability for a full profile in the region 10^{-35} or a LR=10^{35}
- There are 7.4 billion people on the planet
- i.e. $(7.4 \times 10^{9})^{2}/2$ pairwise comparisons
- = 2.74×10^{19} which is 16 orders of magnitude less than 10^{35}
- Although relatives are more likely to match

ISFG Update

President: Walther Parson, Innsbruck • **Vice President**: Mechthild Prinz, New York • **Secretary**: Peter M. Schneider, Cologne
Treasurer: Leonor Gusmão, Rio de Janeiro • **Representative of the Working Parties**: John Butler, Gaithersburg

EDNAP Meeting, Rome, Italy, Apr 17 2018

27th ISFG World Congress Seoul
Aug 28 - Sep 02 2017

705 participants (excluding local staff and company representatives)

from 68 countries

56 oral presentations

482 poster presentations

11 workshops

Status of ISFG Working Groups

https://www.isfg.org/Working%20Groups

Working Group	Working Group Chair (Location)	Recent Activities
German	**Uta-Dorothee Immel** (Halle)	Met June 2016 (Bielefeld), June 2017 (Münster)
English (ESWG-ISFG)	**Andreas Tillmar** (Linköping)	Met Sept 2016 (Budapest), Aug 2017 (Seoul); conducted 2016 and 2017 Relationship Testing Workshops
French	**Vincent Castella** (Lausanne)	Met May 2016 (Predore), May 2017 (Paris)
Italian (Ge.F.I.)	**Susi Pelotti** (Bologna)	Met June 2016 (Rimini)
Spanish & Portuguese (GHEP-ISFG)	**Ulises Toscanini** (Buenos Aires)	Met Sept 2016 (Bayahíbe); published 7 articles since 2015; active proficiency test program
Chinese	**Yiping Hou** (Sichuan)	Met Aug 2017 (Seoul)
Korean	**Soong Deok Lee** (Seoul)	Organized ISFG 2017 (and Asian DNA WG here)
Japanese	--	*Not currently active in ISFG*
DNA Commission	**Peter Gill** (Oslo)	Published 4 articles since 2015
EDNAP	**Niels Morling** (Copenhagen)	Actively meet twice a year; 2 articles since 2015

Proposal for a Polish Speaking Working Group

Support expressed from researchers at

1. Jagiellonian University
2. Institute of Forensic Research
3. Pomeranian Medical University
4. Warsaw Medical University
5. Nicolaus Copernicus University
6. Medical University of Gdańsk
7. Wroclaw Medical University
8. Poznan University of Medical Science
9. Medical University of Lublin
10. Central Forensic Laboratory of the Police Biology Department
11. Medical University of Lodz
12. Medical University of Bialystok

Kraków, 18th April 2017

Letter of Intent

Dear President of the International Society for Forensic Genetics
Dear Professor Walther Parson,

By signing this Letter of Intent we hereby declare our intention to initiate a Polish Speaking Working Group within the International Society for Forensic Genetics.

The undersigned institutions collaborate in Poland promoting the exchange of scientific experiences in the field of forensic genetics. The initiative is strongly supported by the local organizers of the 26th ISFG Congress in Krakow and organizers of the successful local meeting – Symposium of Forensic Genetics with the associated workshops co-organized by the EUROFORGEN-NoE consortium. We plan to meet regularly at national level continuing the idea of the Symposium of Forensic Genetics.

We are convinced that formation of the Polish Speaking Working Group will contribute not only to better promotion of forensic genetics in Poland, but also to propagation of standards, sharing best practices and more effective collaboration of Polish forensic DNA laboratories at national and international levels.

Canine DNA Profiling (CaDNAP) Group

Canine DNA profiling group

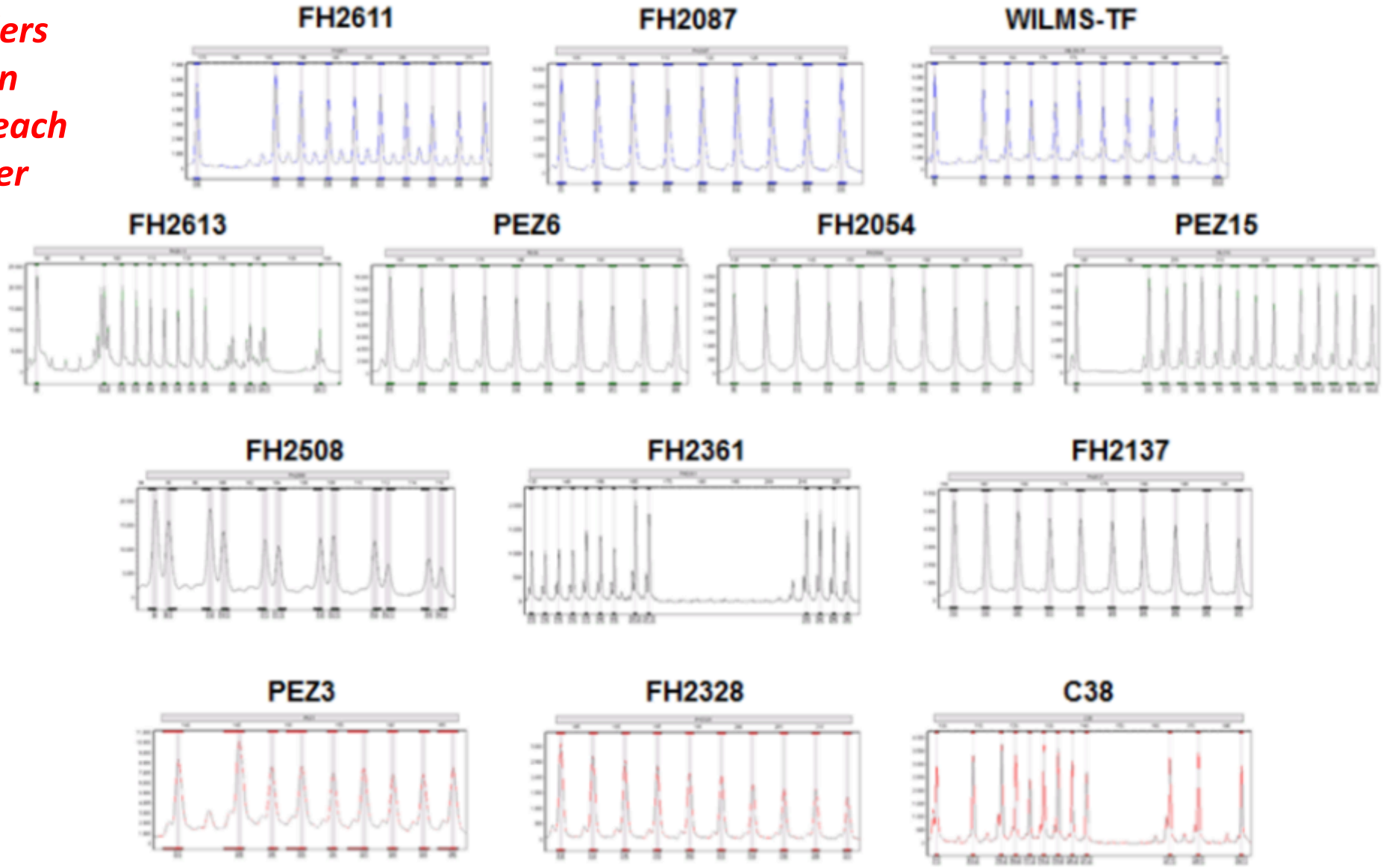
B. Berger presenting at ISFG 2017

- The **Canine DNA Profiling (CaDNAP) group** was founded in 2003 as collaboration of the **Institute of Legal Medicine Innsbruck** with the **German Federal Criminal Police Office** (Bundeskriminalamt Wiesbaden; BKA).
- In 2008 the **Institute of Veterinary Pathology, Justus-Liebig-University, Giessen** joined the group,
- and in 2015 the **Institute of Forensic Medicine, University of Zurich** joined

- **Objectives:**
- selection, characterization and validation of suitable canine STR markers
- establishment of a harmonized STR nomenclature (based on repeats)
- setting quality standards comparable to human identification (allelic ladders, etc.)
- application in forensic casework

Canine DNA Profiling (CaDNAP) group

Allelic ladders have been created for each STR marker



Canine DNA Profiling (CaDNAP) group

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- Eichmann,C.; Berger,B.; Parson,W.; A proposed nomenclature for 15 canine-specific polymorphic STR loci for forensic purposes. Int J Legal Med 2004 118(5):249-266
- Eichmann,C.; Berger,B.; Reinhold,M.; Lutz,M.; Parson,W.; Canine-specific STR typing of saliva traces on dog bite wounds. Int J Legal Med 2004 118(6):337-342

Forensic Science International: Genetics

Journal Status Report

John M. Butler

Associate Editor

ISFG General Assembly – Seoul, South Korea

31 August 2017

Current *FSI Genetics* Editorial Board

Angel Carracedo (Editor-in-Chief)

Peter Schneider (Associate Editor)
John Butler (Associate Editor)
Adrian Linacre (Associate Editor)
Leonor Gusmão (Associate Editor)
Walther Parson (Associate Editor)

8 individuals were added to the editorial board since 2013

Walther Parson became an Associate Editor

http://www.fsigenetics.com/edboard

Cintia Alves, Portugal
Charles Brenner, USA
John Buckleton, New Zealand
Bruce Budowle, USA
Michael Coble, USA
Thore Egeland, Norway
Rolf Fimmers, Germany
Peter Gill, UK
Manfred Kayser, The Netherlands
James Lee, Taiwan
Bertrand Ludes, France
Wolfgang Mayr, Austria
Niels Morling, Denmark
Walther Parson, Austria → moving to Associate Editor
Tom Parsons, Bosnia & Herzegovina (now The Netherlands)
Vince Pascali, Italy
Chris Phillips, Spain
Mecki Prinz, USA
Lutz Roewer, Germany
Antti Sajantila, Finland
Antonio Salas, Spain
Titia Sijen, The Netherlands
Keji Tamaki, Japan
Andreas Tillmar, Sweden
Peter Vallone, USA

Latest *FSI Genetics* Impact Factors

For 2016 (released June 2017)

3.911 based on 3,366 citations

Generally we have experienced an increasing number of citations and a rising impact factor

Year	# Citations	Impact Factor
2012	1,461	3.861
2013	1,656	3.202
2014	2,388	4.604
2015	3,424	4.988
2016	**3,366**	**3.911**

FSI Genetics is the #1 Journal

in the Forensic Science & Legal Medicine Category

Rank	Journal	2016 Impact Factor
1	***Forensic Science International: Genetics***	**3.911**
2	*International Journal of Legal Medicine*	2.382
3	*Regulatory Toxicology and Pharmacology*	2.221
4	*Science & Justice*	1.992
5	*Forensic Science International*	1.989
6	*Forensic Science, Medicine, & Pathology*	1.842
7	*Legal Medicine*	1.276
8	*Journal of Law Medicine & Ethics*	1.223
9	*Journal of Forensic and Legal Medicine*	1.135
10	*Journal of Forensic Sciences*	1.127

2017 Population Data Requirements

Forensic Science International: Genetics 30 (2017) 160–163

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Editorial

Revised guidelines for the publication of genetic population data

- **Introduces a quality check of autosomal STR population data using STRidER**
- **Provides new requirements for MPS generated population data**
- **Provides updates on the minimum number of samples and markers required for autosomal, Y-chromosomal, and X-chromosomal population data**

Updated (2017) Requirements for Population Data Submissions

		Minimum requirements		
		Data[a]	No. samples[c]	Submission to:
Data type	Au-STRs	15 STRs	500	STRidER
	X-STRs	12 STRs	500 (males)	–
	Y-STRs	23 STRs	400	YHRD
	Au-SNPs/ InDels	30 SNPs/InDels	500	–
	X-SNPs/Indels	20 SNPs/InDels	500 (males)	–
	Y-SNPs/Indels	[b]	300	YHRD
	mtDNA_Sanger	Full CR	200	EMPOP
	mtDNA_Sanger	Full mtDNA molecule	100	EMPOP
	mtDNA-SNPs	SNPs in coding region[b]	200	EMPOP
	MPS	MPS–strings	50	–

Gusmao et al. (2017) *Forensic Sci Int Genet* 30: 160-163

Achievements 2015 - 2017

CONGRESS PROCEEDINGS

Freely available

Conference Volumes of the International Society for Forensic Genetics

Progress in Forensic Genetics 16
26th Congress of the International Society for Forensic Genetics,
Kraków, Poland, 2015
Edited by: W. Branicki, T. Kupiec and M. Prinz
Forensic Science International Genetics Supplement Series, Vol. 5, No. 1, 2015

Progress in Forensic Genetics 15
25th Congress of the International Society for Forensic Genetics,
Melbourne, Australia, 2013
Edited by: A. Linacre and N. Morling
Forensic Science International Genetics Supplement Series, Vol. 4, No. 1, 2013

Progress in Forensic Genetics 14
24th Congress of the International Society for Forensic Genetics,
Vienna, Austria, 2011
Edited by: N. Morling
Forensic Science International Genetics Supplement Series, Vol. 3, No. 1, 2011

Achievements 2015 - 2017

Forensic Science International: Genetics 22 (2016) 54–63

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Massively parallel sequencing of forensic STRs: Considerations of the DNA commission of the International Society for Forensic Genetics (ISFG) on minimal nomenclature requirements

Walther Parson[a,b,*], David Ballard[c], Bruce Budowle[d,e], John M. Butler[f], Katherine B. Gettings[f], Peter Gill[g,h], Leonor Gusmão[i,j,k], Douglas R. Hares[l], Jodi A. Irwin[l], Jonathan L. King[d], Peter de Knijff[m], Niels Morling[n], Mechthild Prinz[o], Peter M. Schneider[p], Christophe Van Neste[q], Sascha Willuweit[r], Christopher Phillips[s]

Achievements 2015 - 2017

Forensic Science International: Genetics 24 (2016) 97–102

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Recommendations of the DNA Commission of the International Society for Forensic Genetics (ISFG) on quality control of autosomal Short Tandem Repeat allele frequency databasing (STRidER)

Martin Bodner[a], Ingo Bastisch[b], John M. Butler[c], Rolf Fimmers[d], Peter Gill[e,f], Leonor Gusmão[g,h,i], Niels Morling[j], Christopher Phillips[k], Mechthild Prinz[l], Peter M. Schneider[m], Walther Parson[a,n,*]

Achievements 2015 - 2017

Forensic Science International: Genetics 25 (2016) 191–197

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research paper

DNA Commission of the International Society for Forensic Genetics: Recommendations on the validation of software programs performing biostatistical calculations for forensic genetics applications

M.D. Coble[a,*], J. Buckleton[b,c], J.M. Butler[d], T. Egeland[e], R. Fimmers[f], P. Gill[g,h], L. Gusmão[i,j,k], B. Guttman[l], M. Krawczak[m], N. Morling[n], W. Parson[o,p], N. Pinto[j,k,q,r], P.M. Schneider[s], S.T. Sherry[t], S. Willuweit[u], M. Prinz[v]

Achievements 2015 - 2017

Forensic Science International: Genetics 29 (2017) 269–275

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research Paper

DNA Commission of the International Society for Forensic Genetics (ISFG): Guidelines on the use of X-STRs in kinship analysis

Andreas O. Tillmar[a,b,*], Daniel Kling[c], John M. Butler[d], Walther Parson[e,f], Mechthild Prinz[g], Peter M. Schneider[h], Thore Egeland[i,1], Leonor Gusmão[j,1]

Member statistics

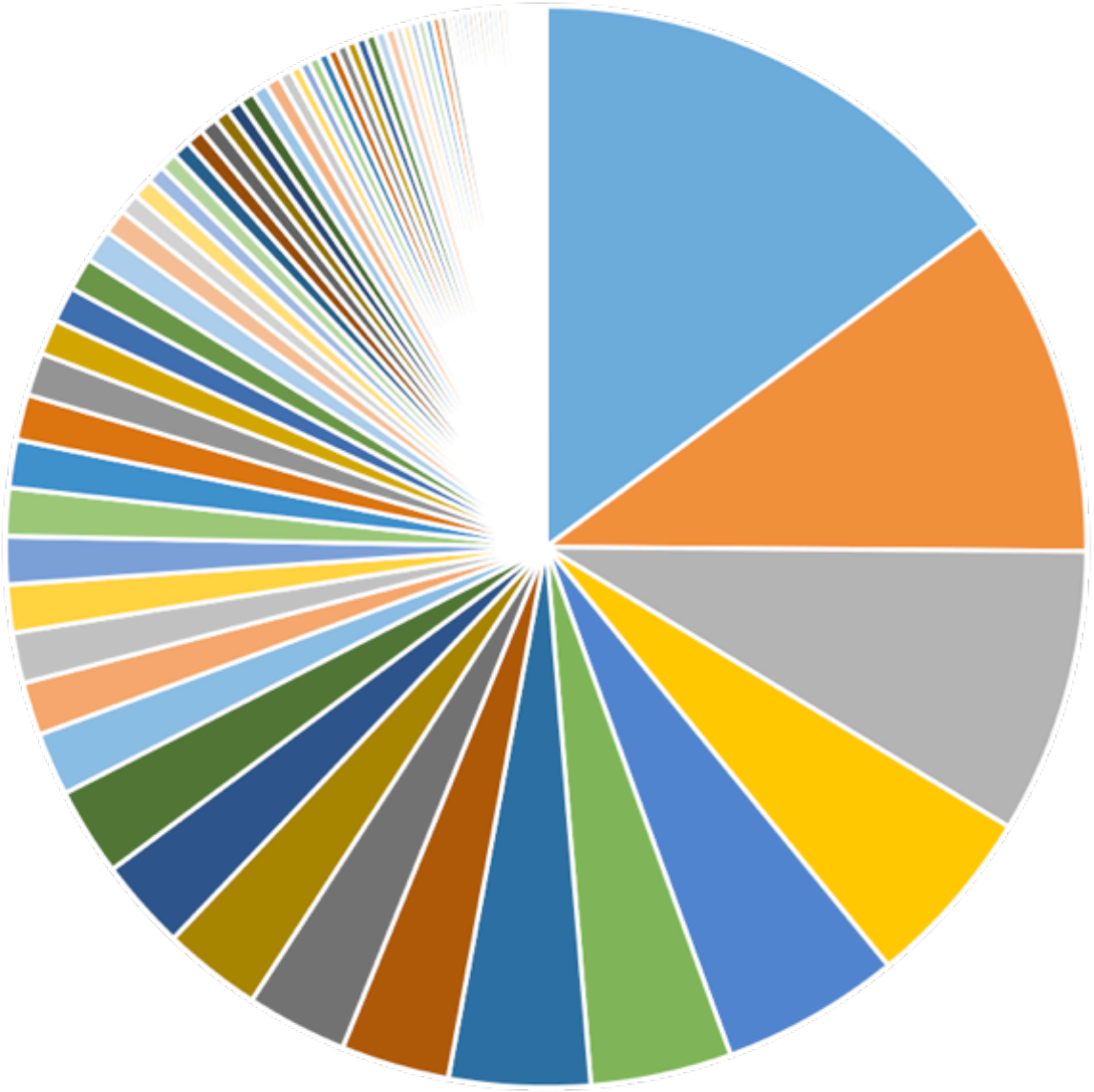
Total number (August 2017):	1325
Countries of origin:	84

Membership has increased by 150 persons since 2015

Changes 2015-2017:

New members accepted:	278
Members removed:	128

Member statistics by country



- United States
- Germany
- Spain
- United Kingdom
- Australia
- Italy
- Korea, Republic of
- Argentina
- Denmark
- Switzerland
- Brazil
- China
- Belgium
- Portugal
- United Arab Emirates
- Austria
- Colombia
- France
- Mexico
- Netherlands
- Poland
- New Zealand
- Norway
- Peru
- Sweden
- Japan

7th ← Korea, Republic of

57

Congress Travel Bursaries

Purpose: To support young scientists presenting at an ISFG congress

- We received 48 applications.
- The initial announcement was for 10 awards. The board reviewed all applicants and spontaneously decided to give out 18 awards

- These will be made available again for the congress in Prague 2019
- For current Terms of Reference, see
  - https://www.isfg.org/files/ISFG_Bursaries_Nov2016.pdf
- We will update this document for the next round

Short-term fellowships

- Purpose: **To support transnational exchange visits between collaborating research groups for specific projects related to forensic genetics**
- For Terms of Reference, see
  - **https://www.isfg.org/files/ISFG_Fellowships_Nov2016.pdf**
- Announcement was made via the November 2016 ISFG newsletter
- Financial support for travel and accommodations for **up to 1000 euros (within continent)** and **2000 euros (between continents)**
- Application rounds: (1) April 2017, (2) October 2017, (3) April 2018, (4) October 2018 – see https://www.isfg.org/Members+Area/Short+Term+Fellowships
- Selection committee included the Working Group chairs and was chaired by John Butler from the ISFG Executive Board
- Will be renewed annually depending on available funding

Second round of short-term travel fellowships

Second round of short-term fellowships (30 applications)

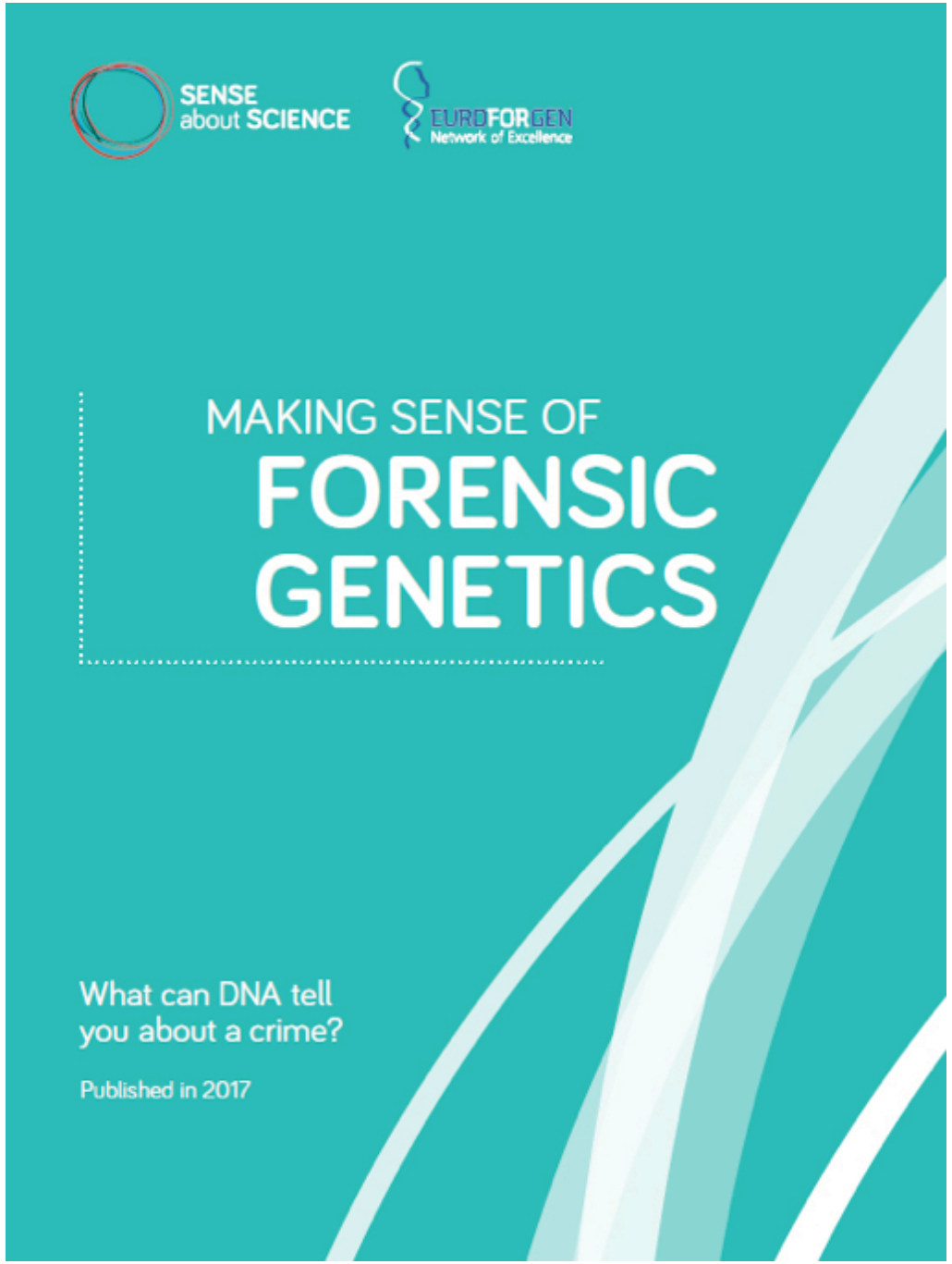
(previous recipients not considered this time)

apply by sending to **fellowships@isfg.org**

first submission deadline October 15th 2017 for 10 applications

second submission deadline March 1st - April 15th 2018 for 10 applications

third submission deadline September 1st - October 15th 2018 for 10 applications



German

Italian

Spanish

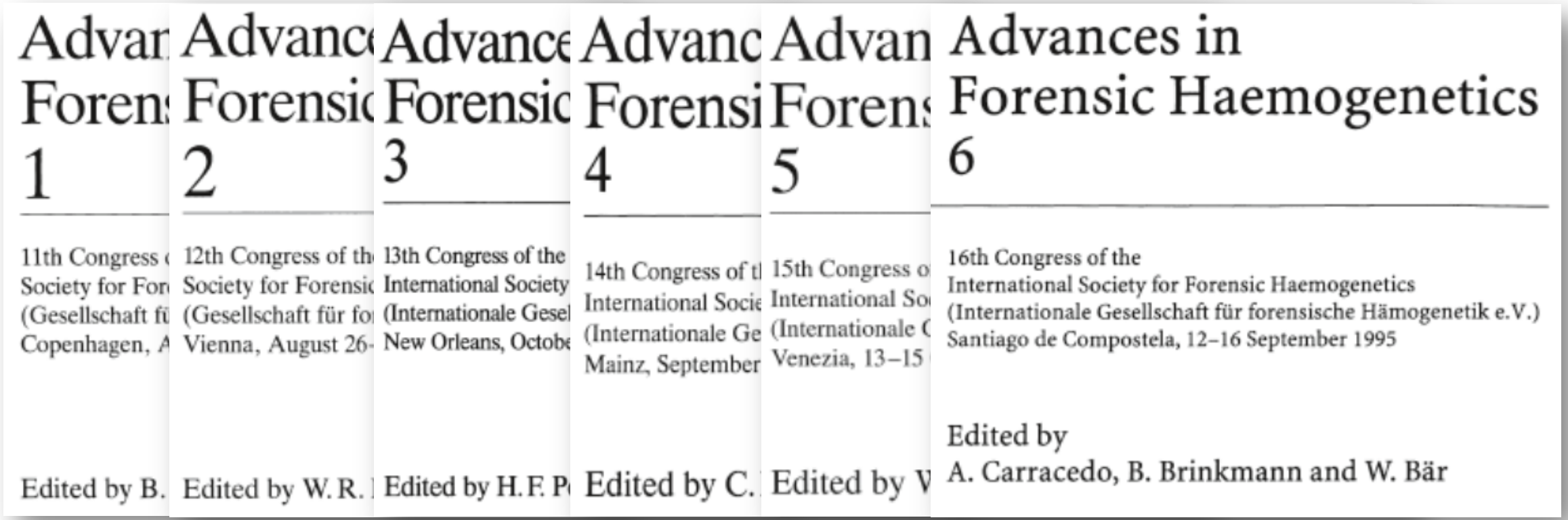
Portuguese

Polish?

Hungarian?

ISFG Website

- Purchase of Springer “Advances in Forensic Haemogenetics” Vol 1-6 (Congress Proceedings 1985 – 1995)
- Are available online as a searchable PDF archive



ISFG Summer School

ISFG Summer School 2018

- Official announcement with registration information coming soon
- Fees between EUR 30 for ½ day to EUR 120 for 2 day-courses

Day	Tract 1	Tract 2	Tract 3
September 3 Morning Session	**DNA interpretation in criminal casework (including LRmix and EuroForMix)** Peter Gill Lourdes Prieto Corina Benschop Oyvind Bleka	**Paternity testing (including the use of Familias, and FamLink-X)** Thore Egeland Daniel Kling	**mtDNA/ EMPOP** Walther Parson
September 3 Afternoon Session			**Population genetics, MPS and Structure software** Chris Phillips Leonor Gusmao Walther Parson
September 4 Morning session			
September 4 Afternoon session			**ISO17025 procedures and DNA database management** Renato Biondo

ISFG Newsletter

4 newsletters published 2016-2017

NEWSLETTER 04-2016

WELCOME

In this edition of o
like to share inform
ISFG World Congre
new board membe
meeting, the activiti
other activities.

NEWSLETTER 11-2016

WELCOME

In this edition of o
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NEWSLETTER 07-2017

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NEWSLETTER 12-2017

INTERNATIONAL SOCIETY FOR FORENSIC GENETICS
http://www.isfg.org

THE 28TH CONGRESS OF THE INTERNATIONAL SOCIETY FOR FORENSIC GENETICS

ISFG
PRAGUE
9–14TH SEPTEMBER 2019
WWW.ISFG2019.ORG

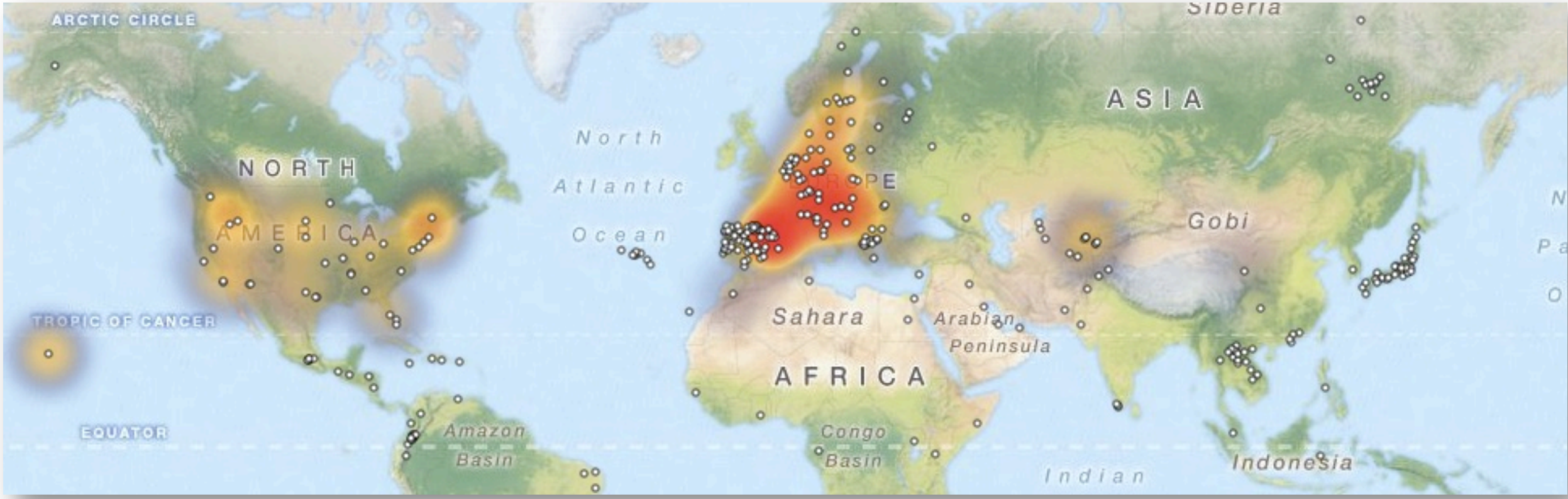
THE 28th CONGRESS OF THE INTERNATIONAL SOCIETY FOR FORENSIC GENETICS

PRAGUE, 9–14TH SEPTEMBER 2019
CZECH REPUBLIC, PRAGUE CONGRESS CENTRE

How to become an ISFG member?

- Go to www.isfg.org and click link for membership
- Enter your personal details
- Nominate two reference persons (ISFG members) that support your membership (good to ask them first, if not known, ask board member)
- Have 60 Euro/year ready to spend (online payment)

Executive committee approves application



https://empop.online/

EMPOP Update

Dr. Walther Parson
assoc. Prof. Institute of Legal Medicine, Medical University of Innsbruck, Austria
adj. Prof. Forensic Science Program, Penn State University, PA, USA
walther.parson@i-med.ac.at

EMPOP

Publications

Meetings

New alignment software

1. Rathbun, M. M., J. A. McElhoe, W. Parson and M. M. Holland (2017). "Considering DNA damage when interpreting mtDNA heteroplasmy in deep sequencing data." Forensic Sci Int Genet **26**: 1-11.
2. Weiler, N. E., K. Baca, D. Ballard, F. Balsa, M. Bogus, C. Borsting, F. Brisighelli, J. Cervenakova, L. Chaitanya, M. Coble, V. Decroyer, S. Desmyter, K. J. van der Gaag, K. Gettings, C. Haas, J. Heinrich, M. Joao Porto, A. J. Kal, M. Kayser, A. Kudelova, N. Morling, A. Mosquera-Miguel, F. Noel, W. Parson, V. Pereira, C. Phillips, P. M. Schneider, D. Syndercombe Court, M. Turanska, A. Vidaki, P. Wolinski, L. Zatkalikova and T. Sijen (2017). "A collaborative EDNAP exercise on SNaPshot-based mtDNA control region typing." Forensic Sci Int Genet **26**: 77-84.
3. Gomes, S. M., M. van Oven, L. Souto, H. Morreira, S. Brauer, M. Bodner, B. Zimmermann, G. Huber, C. Strobl, A. W. Rock, F. Corte-Real, W. Parson and M. Kayser (2017). "Lack of gene-language correlation due to reciprocal female but directional male admixture in Austronesians and non-Austronesians of East Timor." Eur J Hum Genet **25**(2): 246-252.
4. Eduardoff, M., C. Xavier, C. Strobl, A. Casas-Vargas and W. Parson (2017). "Optimized mtDNA Control Region Primer Extension Capture Analysis for Forensically Relevant Samples and Highly Compromised mtDNA of Different Age and Origin." Genes (Basel) **8**(10).
5. Simao, F., A. P. Ferreira, E. F. de Carvalho, W. Parson and L. Gusmao (2018). "Defining mtDNA origins and population stratification in Rio de Janeiro." Forensic Sci Int Genet **34**: 97-104.
6. Strobl, C., M. Eduardoff, M. M. Bus, M. Allen and W. Parson (2018). "Evaluation of the precision ID whole MtDNA genome panel for forensic analyses." Forensic Sci Int Genet **35**: 21-25.

Meetings 2017/2018

SAM2 - SWGDAM Meeting, Fredericksburg, VA, Jan 2017
EMPOP workshop - GEDNAP Meeting, Giessen, Germany, Feb 2017
EMPOP-QC/SAM2 - mitoDB meeting, MPI Jena, Germany, Feb 2017
EMPOP-Update - EDNAP, Vilnius, Lithuania, Apr 2017
EMPOP workshop - Rio de Janeiro, Brazil, May 2017
EMPOP-QC/SAM2 - Genome Variants, Santiago de Compostela, Spain, Jun 2017
EMPOP workshop - ISFG World Conference, Seoul, South Korea, Aug 2017
EMPOP training - NFI, The Hague, Netherlands, Sep 2017
EMPOP-Update - EDNAP, Athens, Greece, Oct 2017

SAM2 - SWGDAM Meeting, Woodbridge, VA, Jan 2018
EMPOP training - HSA, Singapore, Jan 2018
EMPOP QC - Monopoly 16 project dna.bases, Innsbruck, Austria, Jan 2018
EMPOP workshop - GEDNAP Meeting, Basel, Switzerland, Feb 2018
11th Haploid Marker Meeting, Bydgoszcz, Poland, May 2018

SAM 2

Development of software for automated phylogenetic alignment of mitochondrial DNA sequences

SAM2 - Motivation

Consistent alignment is difficult to achieve manually and should be supported by software

Alignment-free searches introduced in EMPOP v2 (**SAM**, Röck et al 2011)

Database searches independent from alignment

But did not address alignment problem

Goal: Extend SAM to produce consistent alignment and mitotype nomenclature from FASTA-like strings

Code programming is accomplished

Internal and external testing is accomplished

Manuscript in preparation

Adaptation of EMPOP in preparation

‘Inferring Ancestry from DNA’

Invited speakers
Christopher Phillips
Mark Jobling
Chris-Tyler Smith

44 oral presentations
64 posters

Bydgoszcz [ˈbɨdgɔʃʧ] ‘bid’ ‘gosh’

Acknowledgements

EUROFORGEN Network of Excellence FP7-SEC-2011-285487

DNA SeqEx STR Sequencing & Exchange Home/2014/ISFP/AG/LAWX/4000007135
DNA-STR Massive Sequencing & International Information Exchange

VISAGE VISIBLE ATTRIBUTES THROUGH GENOMICS 740580
Visual Attributes Through Genomics

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Maximizing mtDNA Testing Potential with the Generation of High-Quality mtGenome Reference Data

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EMPOP–an innovative human mtDNA database

Research project P22880-B12
Genetic discovery of an early medieval Alpine population

EMPOP STRidER

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dna.bases Monopoly 2016, STEFA, 779485
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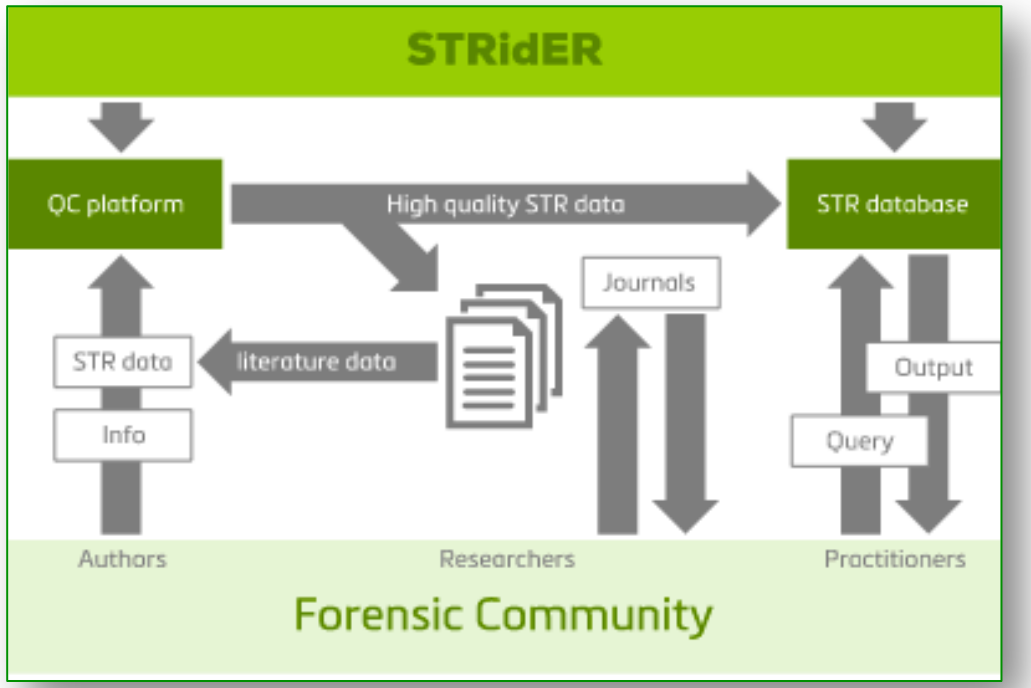


D8S1179

Allele	AUSTRIA	BELGIUM	BOSNIA AND HERZEGOWINA	CZECH REPUBLIC	DENMARK	FINLAND	FRANCE	GERMA
	222	206	171	200	200	230	208	
8	1.8018e-2	7.2816e-3	5.8480e-3	7.5000e-3	1.5000e-2	1.7391e-2	2.4039e-2	1.2840
9	1.8018e-2	1.2136e-2	8.7719e-3	5.0000e-3	1.0000e-2	8.6956e-3	9.6154e-3	1.2840
10	9.4595e-2	8.7379e-2	5.8479e-2	5.5000e-2	9.7500e-2	8.2609e-2	8.4135e-2	8.7613
11	1.0135e-1	9.7087e-2	3.2164e-2	1.0000e-1	8.0000e-2	1.3261e-1	8.8942e-2	7.7795
12	1.6216e-1	1.5049e-1	1.8713e-1	1.5250e-1	1.3000e-1	1.3261e-1	1.3462e-1	1.4199
13	2.9054e-1	3.1311e-1	3.4210e-1	3.5000e-1	3.4500e-1	3.5217e-1	3.1490e-1	3.1269
14	1.9144e-1	1.6990e-1	2.1637e-1	2.1250e-1	2.0750e-1	1.8478e-1	2.0433e-1	1.9864
15	1.0360e-1	1.2379e-1	1.1403e-1	9.7500e-2	8.5000e-2	5.6522e-2	1.0336e-1	1.1933
16	1.8018e-2	3.3981e-2	3.2164e-2	2.0000e-2	2.0000e-2	1.5217e-2	3.6058e-2	3.0211
17	2.2522e-3	4.8544e-3	2.9240e-3		5.0000e-3	1.0870e-2		6.0423

https://strider.online/

STRidER Update

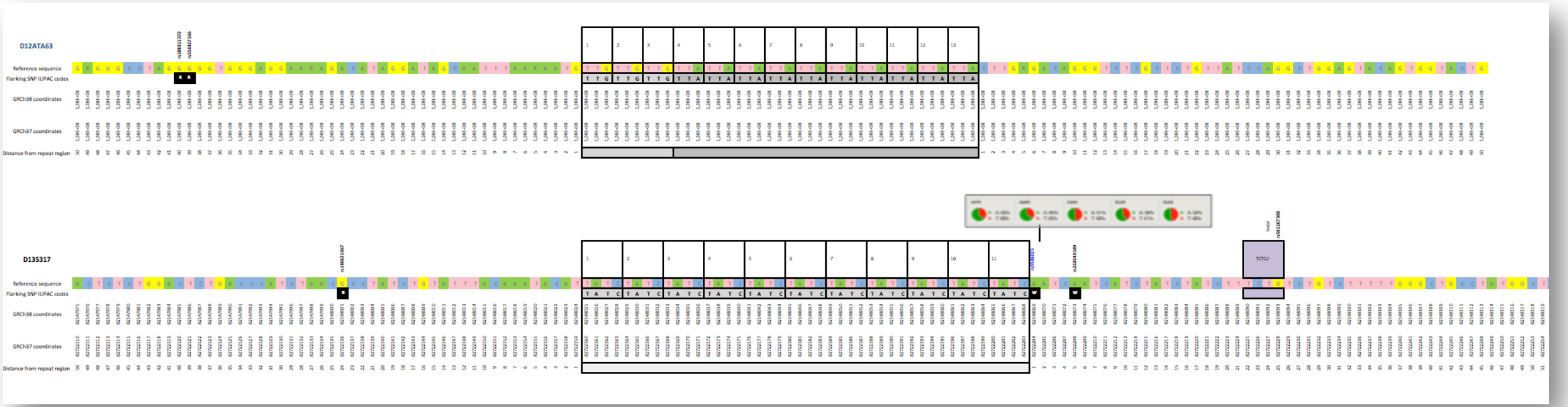
Dr. Walther Parson
assoc. Prof. Institute of Legal Medicine, Medical University of Innsbruck, Austria
adj. Prof. Forensic Science Program, Penn State University, PA, USA
walther.parson@i-med.ac.at

STRidER

Recommendations of the DNA Commission of the International Society for Forensic Genetics (ISFG) on quality control of autosomal Short Tandem Repeat allele frequency databasing (STRidER)

Martin Bodner[a], Ingo Bastisch[b], John M. Butler[c], Rolf Fimmers[d], Peter Gill[e,f], Leonor Gusmão[g,h,i], Niels Morling[j], Christopher Phillips[k], Mechthild Prinz[l], Peter M. Schneider[m], Walther Parson[a,n,*]

+ **STR Sequence Guide** as static ESM with alignment examples



STRidER

“The devil’s in the detail”: Release of an expanded, enhanced and dynamically revised forensic STR Sequence Guide

C. Phillips[a,*], K. Butler Gettings[b], J.L. King[c], D. Ballard[d], M. Bodner[e], L. Borsuk[b], W. Parson[e,f]

[a] *Forensic Genetics Unit, Institute of Forensic Sciences, University of Santiago de Compostela, Spain*
[b] *National Institute of Standards and Technology, Biomolecular Measurement Division, Gaithersburg, MD, USA*
[c] *Institute of Applied Genetics, Department of Molecular and Medical Genetics, University of North Texas Health Science Center, Fort Worth, TX, USA*
[d] *King’s Forensics, King’s College London, Franklin-Wilkins Building, London, UK*
[e] *Institute of Legal Medicine, Medical University of Innsbruck, Innsbruck, Austria*
[f] *Forensic Science Program, The Pennsylvania State University, University Park, PA, USA, USA*

+ **revised** STR Sequence Guide as **dynamic** document at STRidER

HOME | QUERY | BATCH QUERY | ABOUT | FREQUENCIES | FORMULAE | QUALITY CONTROL | STR SEQ NOMENCLATURE

STR Sequence Nomenclature

The **‘Forensic STR Sequence Structure’ file** is an updated set of forensic STR sequences that was originally published as *Supplementary File S1* in the article:

The most recent version of this permanently curated and updated Forensic STR sequence structure file containing updated information is available for download here. The updates since the last version are reported in a change log contained in the file. To receive information on new releases of the Forensic STR sequence structure file and to stay updated about STRidER, register here for the STRidER newsletter.

	A	B	C	D	
1	**Change Log**				
2					
3	New in online version	Date	Sheet	Locus	Description of change
77	2	22.Nov.17	A-STRs	D2S441	Added a mobility shift nucleotide substitution/
78	2	22.Nov.17	A-STRs	D3S1358/D19S433/D21S11	Repeat region summary sequence structures r
79	2	22.Nov.17	XY-STRs	DYS19/DXS10103	Repeat region summary sequence structures r
80					
81	2	29.Nov.17	All STRs	Multiple	Repeat region summary sequence structures f
82					
83	2	06.Dez.17	A-STRs	D7S820	Pie chart added for flanking region SNP rs7789
84					
85	3	08.Dez.17	All-STRs	Multiple	Repeat region summary sequence structures f
86					
87	3	11.Feb.18	A-STRs	D9S1122	2-NT Indel [TG/-] rs754976988 creating X.2 alle
88					
89	3	20.Feb.18	XY-STRs	DYS458	Repeat region summary sequence structure co
90					
91	3	22.Feb.18	XY-STRs	DYS612	Revised repeat region description confirmed a
92					

S1A. Common Use A-STRs | S1B. Common Use XY-STRs | S1C. Additional A-STRs | S1D. Additional XY-STRs | Sequence strings for all STRs | **Change log** | +

“The devil’s in the detail”: Release of an expanded, enhanced and dynamically revised forensic STR Sequence Guide

C. Phillips[a,*], K. Butler Gettings[b], J.L. King[c], D. Ballard[d], M. Bodner[e], L. Borsuk[b], W. Parson[e,f]

[a] *Forensic Genetics Unit, Institute of Forensic Sciences, University of Santiago de Compostela, Spain*
[b] *National Institute of Standards and Technology, Biomolecular Measurement Division, Gaithersburg, MD, USA*
[c] *Institute of Applied Genetics, Department of Molecular and Medical Genetics, University of North Texas Health Science Center, Fort Worth, TX, USA*
[d] *King’s Forensics, King’s College London, Franklin-Wilkins Building, London, UK*
[e] *Institute of Legal Medicine, Medical University of Innsbruck, Innsbruck, Austria*
[f] *Forensic Science Program, The Pennsylvania State University, University Park, PA, USA, USA*

Audit of GRCh38 reference genome builds released between 2013 - 2017

Revised repeat region sequence structure summaries

Inverted multiple allele Y-STRs, mobility shift SNPs, flank indels, …

+ 34 aSTRs (total of 71 aSTRs)

+ 22 Y-STRs (total of 47 Y-STRs)

STRidER STRs for identity ENFSI Reference database, v2

HOME QUERY BATCH QUERY ABOUT FREQUENCIES FORMULAE QUALITY CONTROL STR SEQ NOMENCLATURE

Quality Control

STRidER provides quality control of autosomal STR data. A suite of software tools has been developed to scrutinize STR population data and thus increase the quality of datasets to ensure reliable allele frequency estimates. The board of the International Society of Forensic Genetics (ISFG) and the editors of Forensic Science International: Genetics invited STRidER to logistically organize and perform quality control (QC) of autosomal STR population data in the course of manuscript preparations for the journal [1]. Before STR population papers are put forward to the editors for review, the authors are requested to submit the data to STRidER. After positive evaluation, the authors will be contacted with the respective STRidER accession numbers that serve as indicator of successful QC for the editors and reviewers. The necessary steps for submission of STR data to STRidER are outlined below.

Step 1

Prepare your **STR data file** as shown in the example file that can be downloaded and used as template. It is a tab delimited text file that can be created using standard text software or MS Excel (then, save file under .txt format). The minimum requirements for population datasets are 16 autosomal STR loci typed in 500 samples (for exceptional populations, the latter number can be smaller, please contact STRidER before submission).

The initial lines (identified using the "#" symbol) specify details of the dataset and origin of the samples. Line 1 must contain a description of population(s) reported (e.g., the title of the study), number of samples, geographic origin, and the number of STR loci. Line 2 must indicate the contact author's name with email address. Further text lines marked with "#" can be included for comments or description of the detailed geographic background and the appropriate metapopulation affiliation of the genotypes. Lines below these text lines list the original STR genotypes. Allele nomenclature criteria are applied as described in the "About" tab of this website. The order of loci does not matter. Alleles for the same locus have to be reported in adjacent columns. Loci names must not contain spaces. Report both alleles for homozygous loci. Use "." instead of "," for incomplete alleles, e.g. "9.3" not "9,3". Note that only complete genotypes are accepted. It is imperative that STR genotypes are reported individually and unshuffled using a unique identifier for each genotype in the dataset. The names are necessary for correspondence.

Also prepare an **accompanying STR information file** per population containing additional information on the dataset as outlined in the example information file. This information might be necessary for evaluation of the dataset. Keep raw data files available for any later inquiries.

Step 2

Submit your files to STRidER by email (see contact). The genotype data should be submitted as a file containing the following notation: Author_country_number of samples.txt (e.g. Parson_AUT_573.txt), the accompanying file should be named Author_country_number of samples_Info.xls or .xlsx (e.g. Parson_AUT_573_Info.xls). The data will be quality checked as outlined in [2] using in-house software.

Step 3

After STRidER evaluation, communication with respect to individual genotypes may follow. Once your data passed QC you will receive the STRidER accession number(s) for your data together with allele frequencies and forensic/population genetic parameters calculated from the dataset(s). Please provide accession number(s) to the journal editor and cite STRidER [2] in your manuscript.

Forensic Science International: Genetics 31 (2017) 111–117

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research paper

STRSeq: A catalog of sequence diversity at human identification Short Tandem Repeat loci

Katherine Butler Gettings[a,*], Lisa A. Borsuk[a], David Ballard[b], Martin Bodner[c], Bruce Budowle[d,e], Laurence Devesse[b], Jonathan King[d], Walther Parson[c,f], Christopher Phillips[g], Peter M. Vallone[a]

NCBI BioProject—STRseq and **STRidER**
Collaboration in QC and exchange of data

STRidER

Acknowledgements

R. Scheithauer

Monopoly 2010

Monopoly 2014

Monopoly 2016

Co-funded by the Internal Security Fund of the European Union

dna.bases

Monopoly 2016, STEFA, 779485
Steps Towards a European Forensic Science Area; WP7; Empowering Forensic Genetic DNA Databases for the Interpretation of Next Generation Sequencing Profiles (dna.bases)

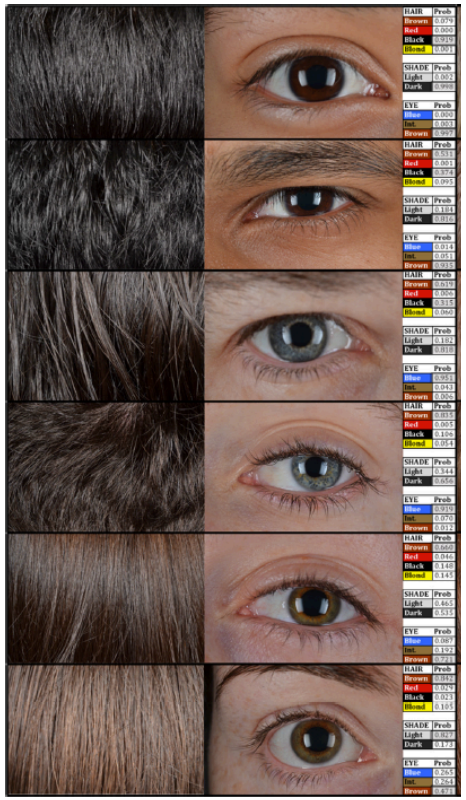
ISFG Commission on MPS of STRs
ISFG Commission on STRidER

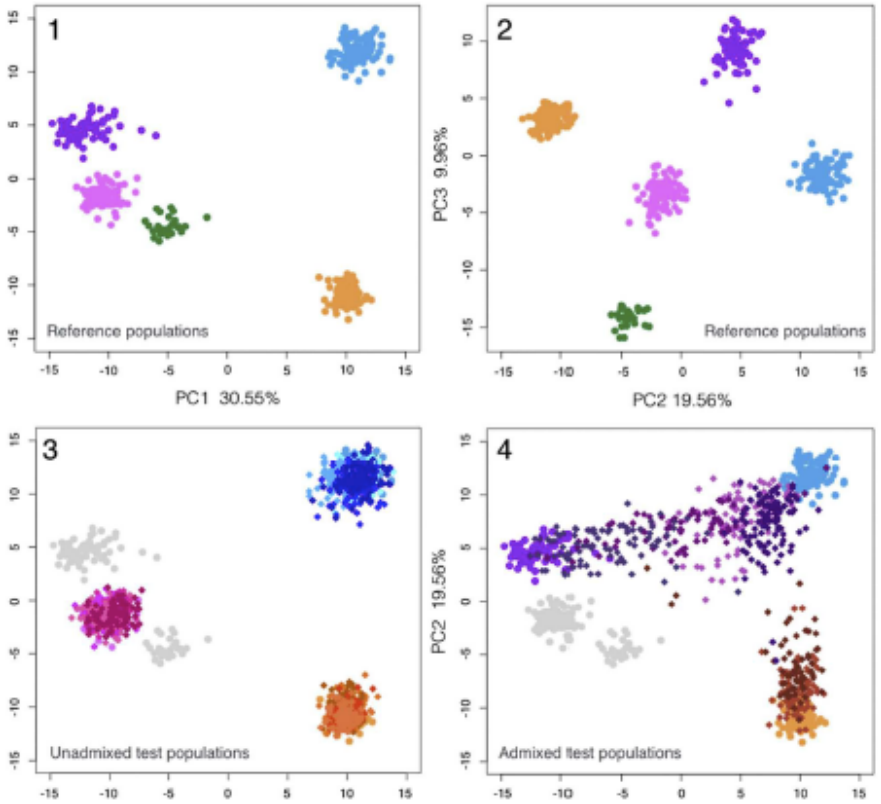
ENFSI laboratories

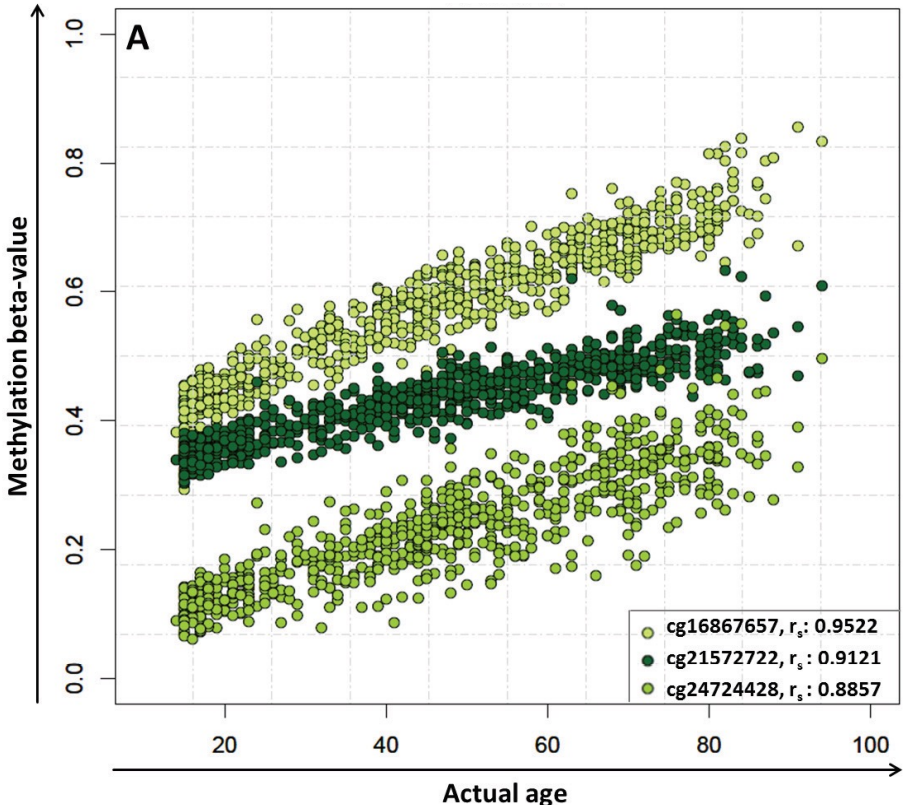
Peter Gill
Ingo Bastisch
David Ballard
Chris Phillips
Katherine Gettings
Jonathan King
Martin Bodner

STRidER

DNA SeqEx
STR Sequencing & Exchange







Update VISAGE

Prof. Dr. Walther Parson[1,2]
[1]Institute of Legal Medicine, Innsbruck Medical University, Innsbruck, Austria
[2]Forensic Science Program, The Pennsylvania State University, PA, USA

VISAGE
VISIBLE ATTRIBUTES THROUGH GENOMICS

Cases without database match

STR typing → NO MATCH

Phenotypic markers → Composite Sketch → 

- appearance
- ancestry
- age

Manfred Kayser (Coordinator) Rotterdam, NED
Wojciech Branicki Krakow, POL
Chris Phillips, Angel Carracedo S. de Compostela, ESP
Walther Parson Innsbruck, AUT
Michael Nothnagel Cologne, GER
Barbara Prainsack Vienna, AT
Peter M. Schneider Cologne, GER
Ingo Bastisch Wiesbaden, GER
François-Xavier Laurent Lyon, FRA
Titia Sijen The Hague, NED
Johannes Hedman Linkoping, SWE
Shazia Khan London, UK
Magdalena Spólnicka Warsaw, POL

VISAGE

VISIBLE ATTRIBUTES THROUGH GENOMICS

www.visage-h2020.eu

This project received funding from the European Union's Horizon 2020 Research and Innovation programme under grant agreement No 740580 05/2017 - 04/2021

VISAGE – Main Goals

1) **Known markers + explore new** markers on appearance, age and ancestry

2) **Design, develop and validate** prototype tools

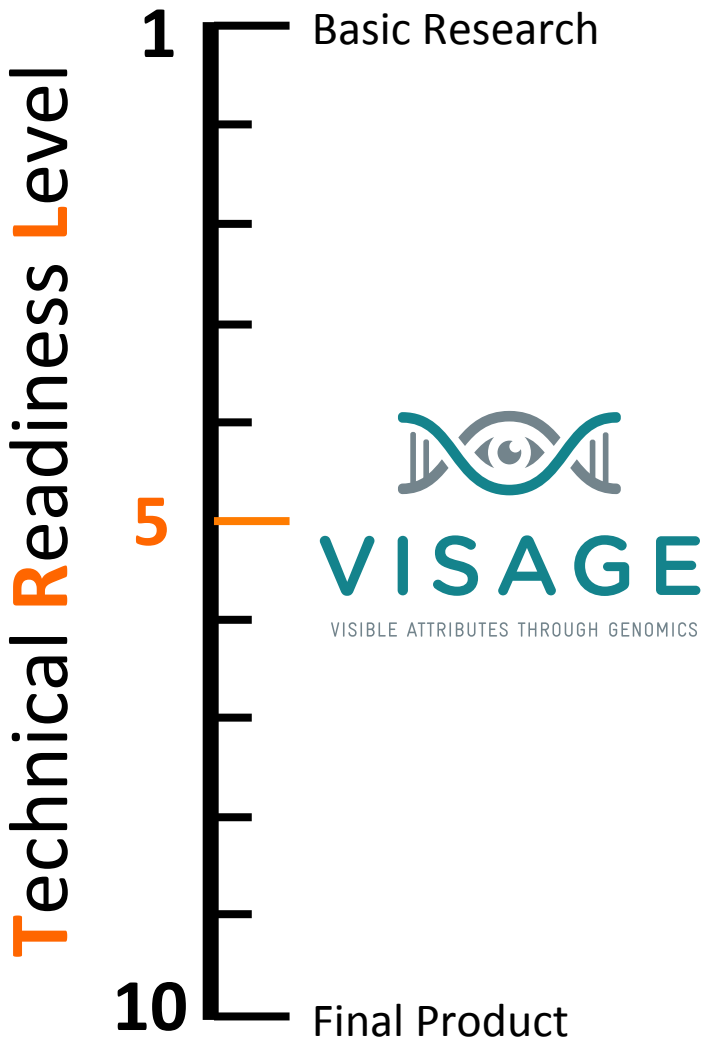
3) **Design interpretation software** considering combined stats of all information

4) Identify **ethical issues** and make recommendations

5) Implement in **routine casework**

6) **Training and dissemination**

Tool development in VISAGE



VISAGE
VISIBLE ATTRIBUTES THROUGH GENOMICS

The **VISAGE** - Consortium is developing genotyping and statistical prototype tools, forensically validate and implement them into forensic practice for predicting **appearance**, **age**, and **ancestry** from DNA traces and study its ethical, societal & regulatory dimensions (period: 05/2017-04/2021).

Tool 1: Appearance & Ancestry (SNP multiplex)

Tool 2: Age (quantitative methylation)

Primer design / PCR optimization

Primer design

Structure

Dimer formation

No of primer pools

Reaction conditions

Efficiency

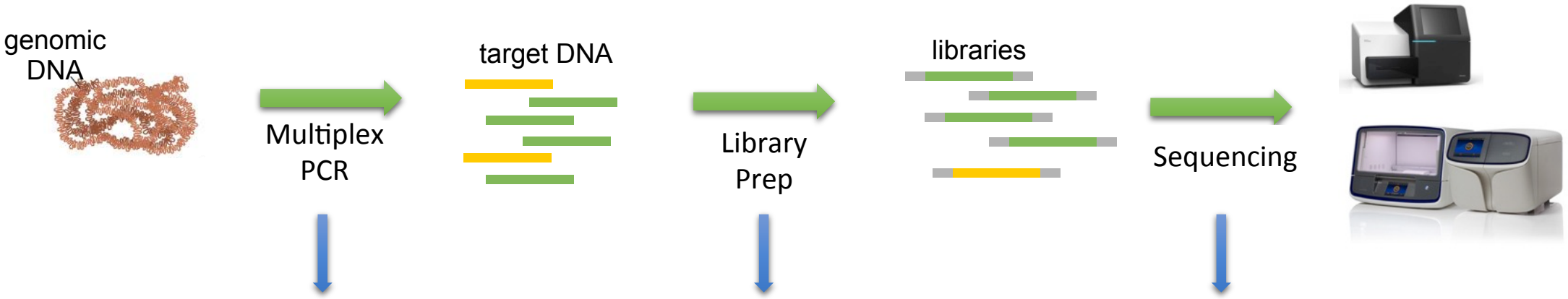
Sensitivity

Multiplex design/ optimization

Check with

Final evaluation

Ancestry & Appearance Tool

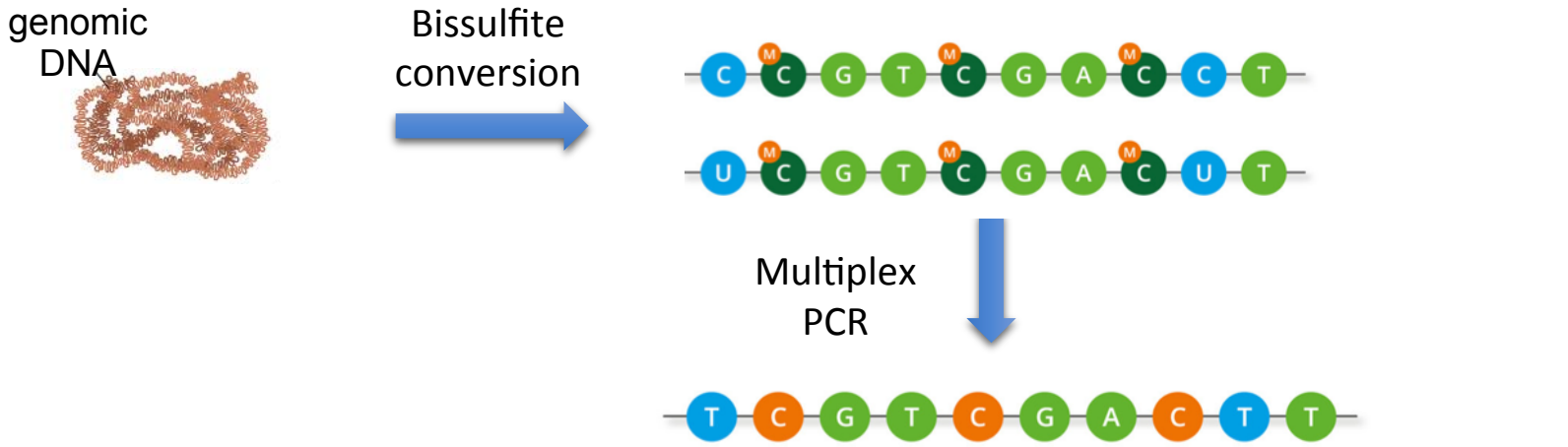


Multiplex design/ optimization
primer pools
how many reactions
optimize PCR

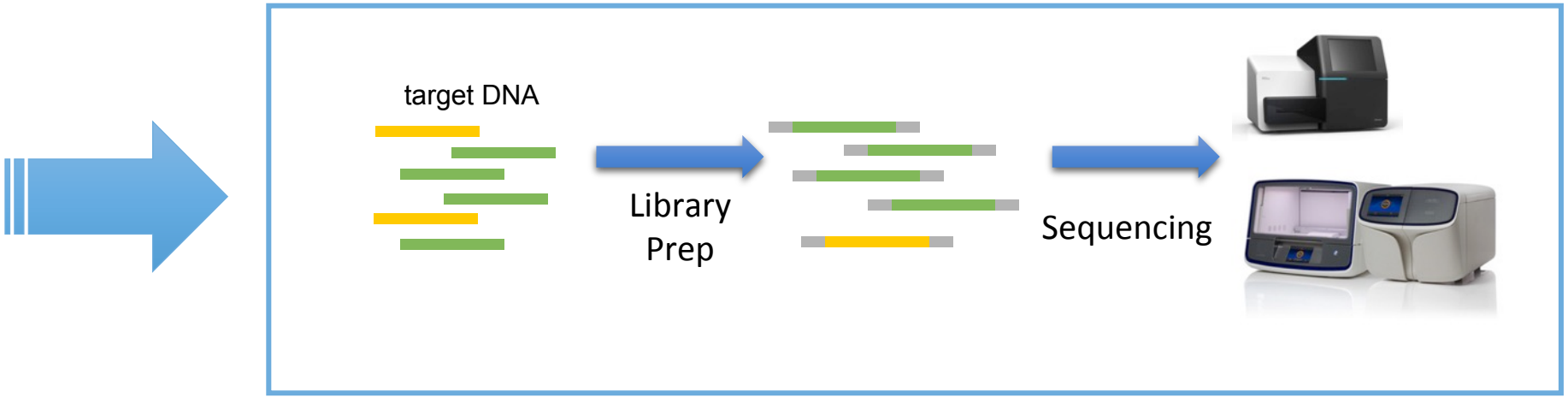
Adapt to different library prep commercial products

How many samples per chip
Overall performance
Coverage requirements

Age estimation by quantitative methylation



https://www.gatc-biotech.com



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dna.bases Monopoly 2016, STEFA, 779485
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Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

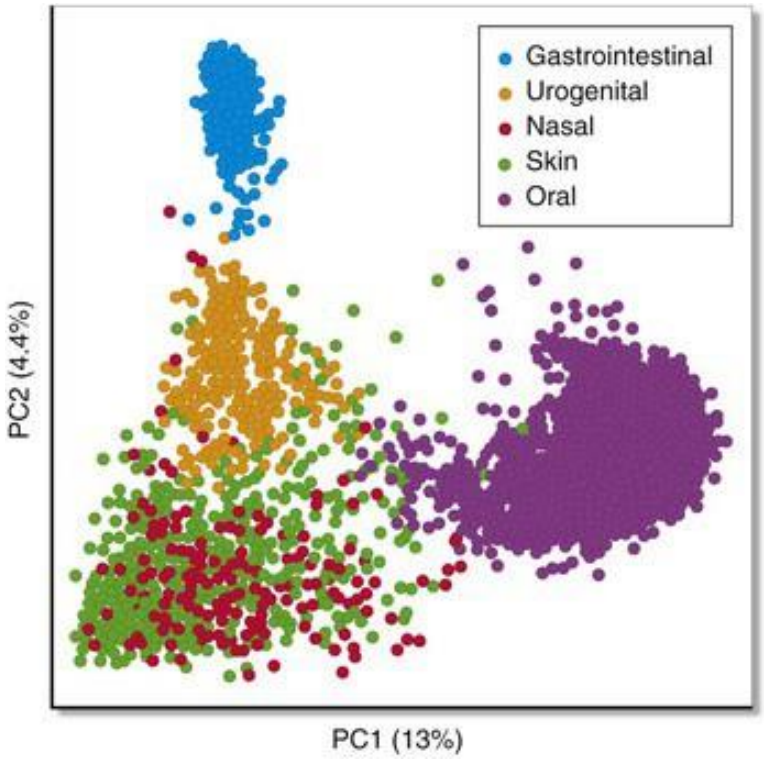
Research paper

Body fluid prediction from microbial patterns for forensic application

Eirik Nataas Hanssen[a,b,*], Ekaterina Avershina[c], Knut Rudi[c], Peter Gill[a,b], Lars Snipen[c,**]

[a] Department of Forensic Biology, Oslo University Hospital, Oslo, Norway
[b] Department of Forensic Medicine, University of Oslo, Oslo, Norway
[c] IKBM, Norwegian University of Life Sciences, Aas, Norway

The healthy human microbiota

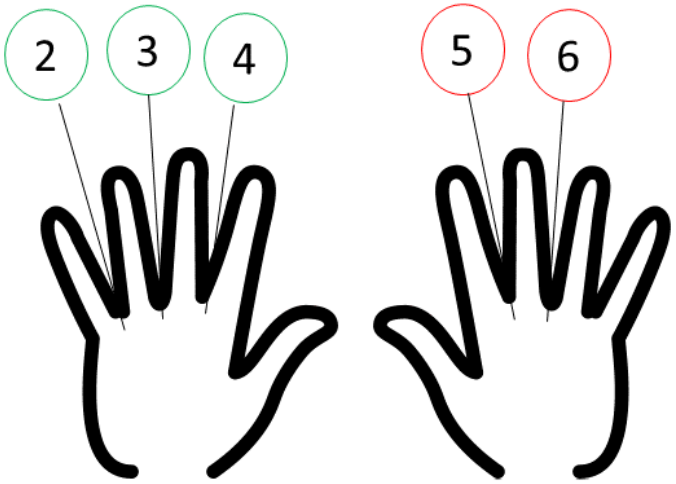


C Huttenhover et al, *Structure, function and diversity of the healthy human microbiome*. Nature, 2012. 486(7402): p. 207-14

Challenges

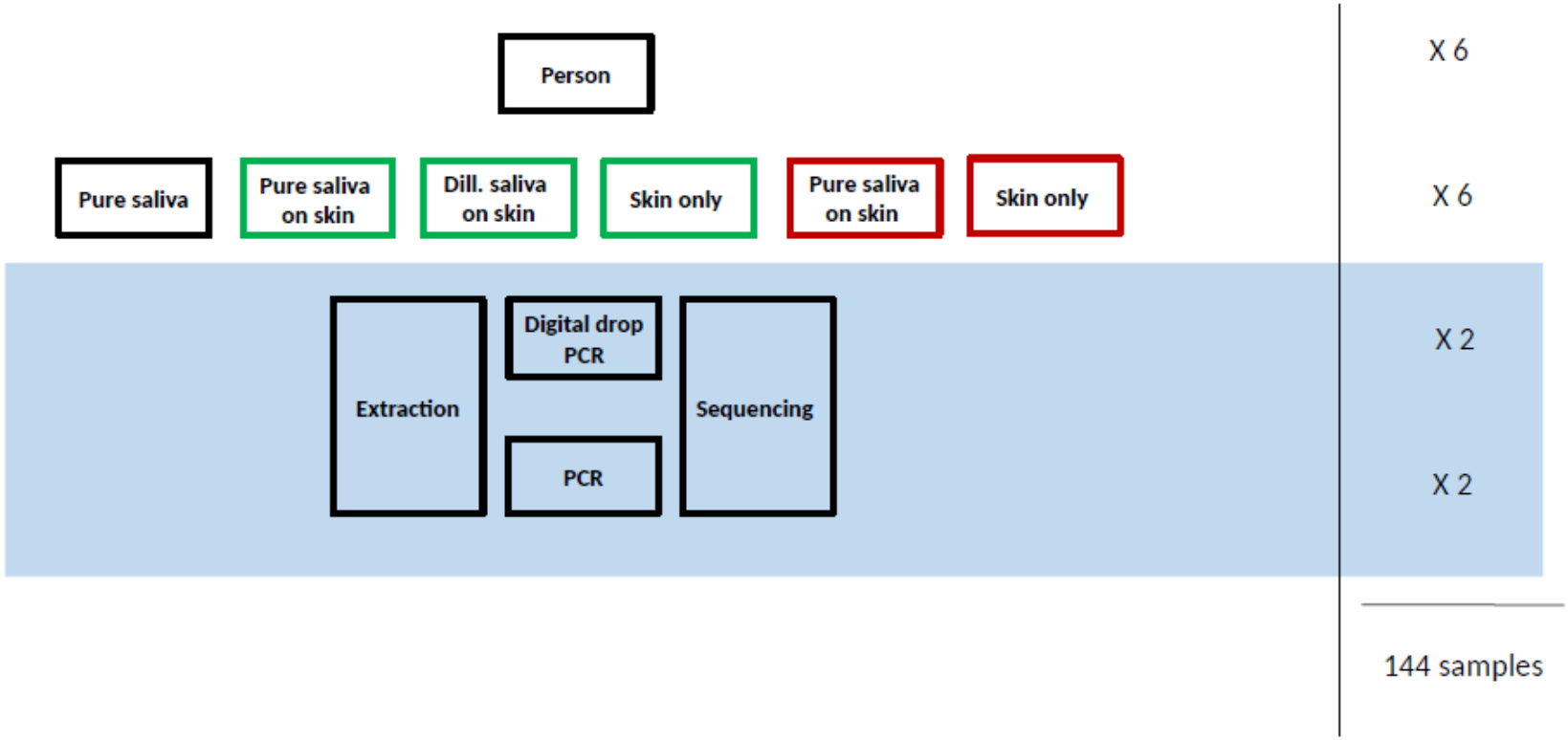
- Sampling technique
- Detection level
- Technical bias (extraction , primers, amplification)
- Method precision
- Mixtures of different body fluids
- Data interpretation tools (explore vs pattern recognition)

Experimental setup - sampling

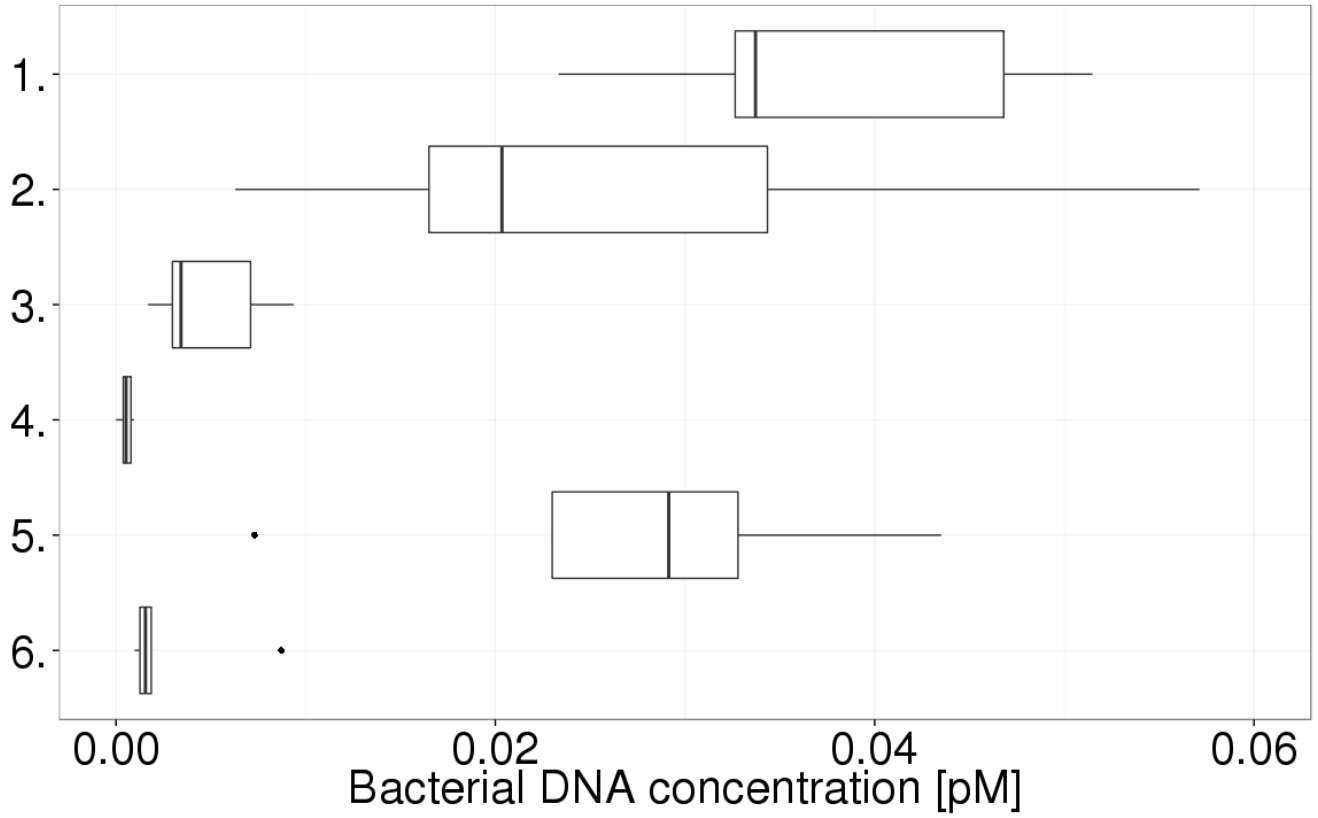


1. Pure saliva from mouth
2. Saliva deposited on skin, collected with tape
3. Diluted saliva on skin, collected with tape
4. Pure skin, collected with tape
5. Saliva deposited on skin, collected with swab
6. Pure skin, collected with swab

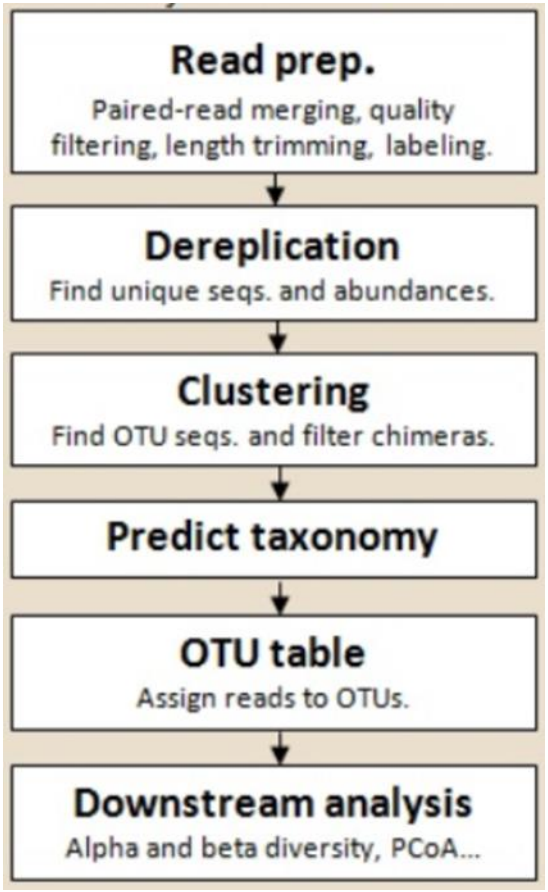
Experimental setup - Lab



Content of bacterial DNA

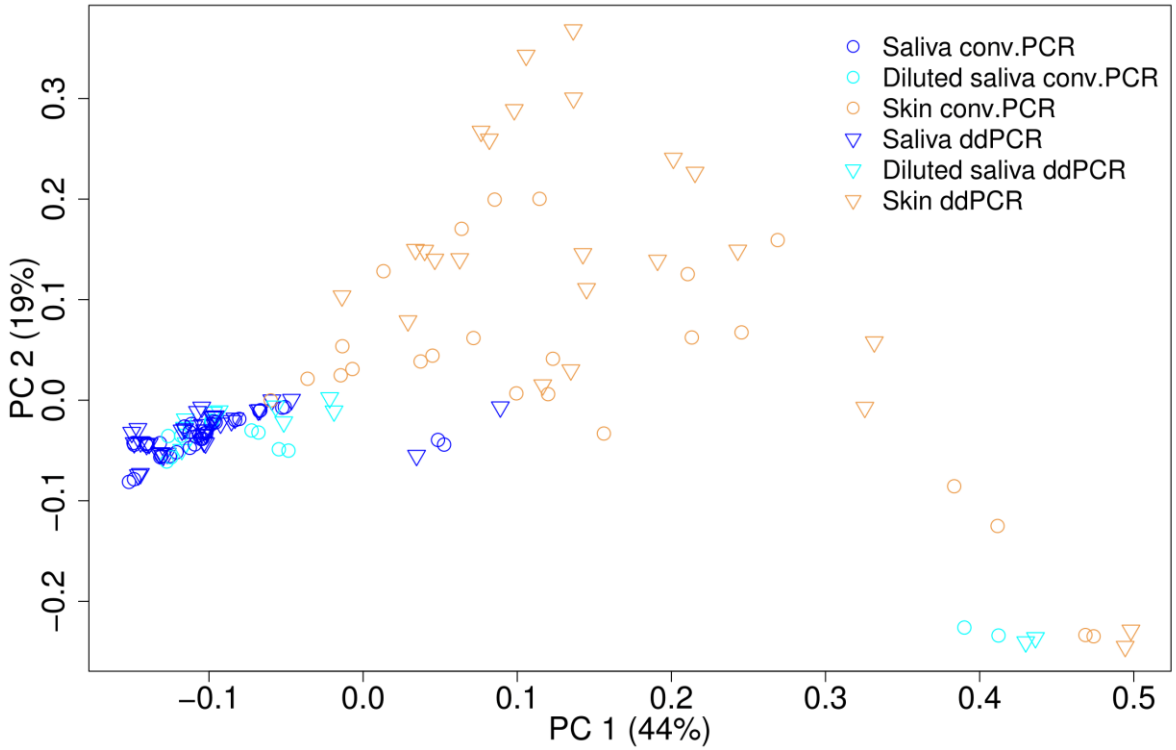


Data workflow

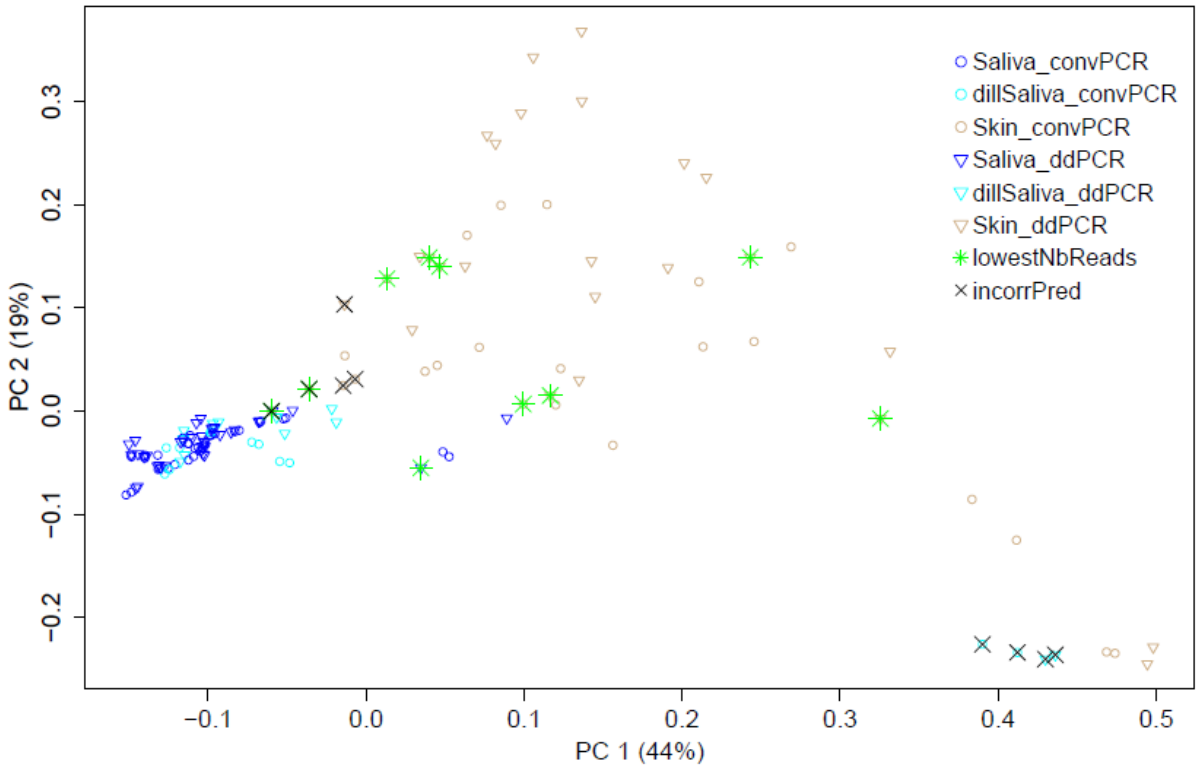


\+ PCA/LDA

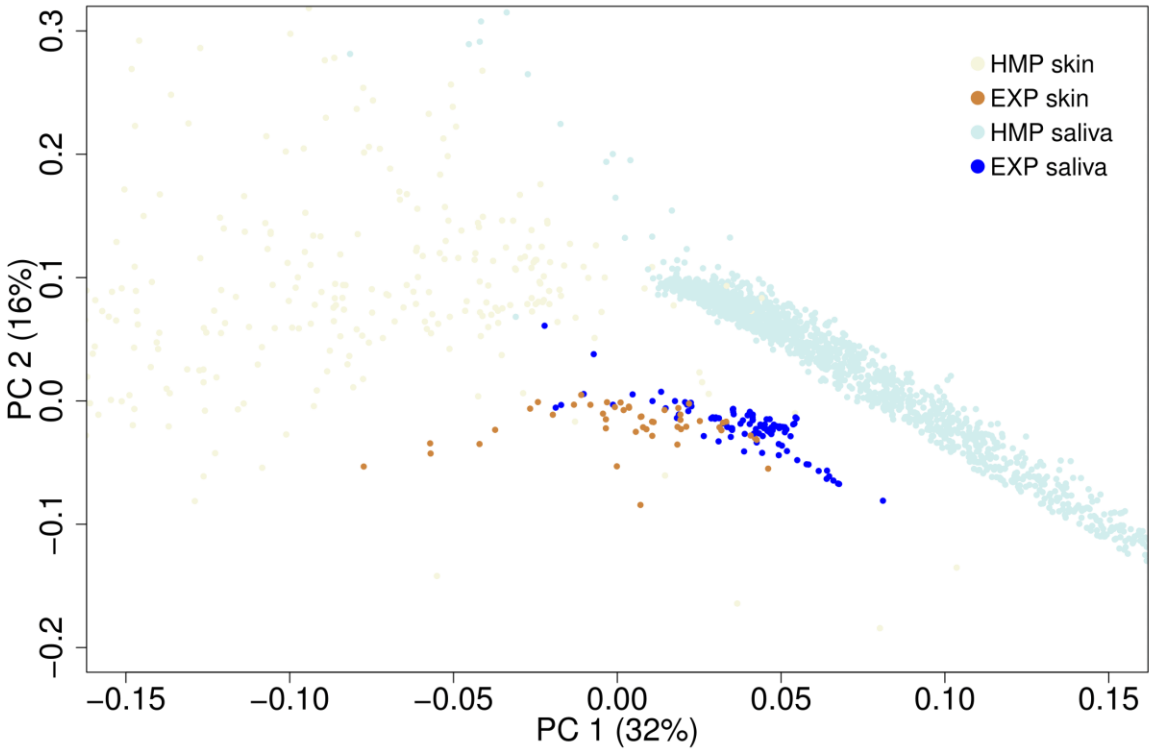
PCA – effect of factors



PCA – accuracy



PCA – comparison to HMP data



Optimizing body fluid recognition from microbial taxonomic profiles

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Customizing model

- PCA -> PLS
- Adjustment of calculation settings
  - Taxa resolution
  - Dimension reduction of data
  - Data transformation
  - Variable selection
- OTU -> direct assigning reads to taxa

- Conclusion -> robust method with high accuracy (>96%)

Thanks

Aims

- Evaluate the effect from the mentioned experimental factors on the measured bacterial composition
- How well can we recognize body fluids
- Building a costumized method for bodyfluid recognition

Different trace material

- Blood
- Semen
- Saliva
- Urine
- Hair
- Bones
- Tissue
- Feces
- Skin
- Tears

- Vaginal
- Nasal

Different trace material

- Blood
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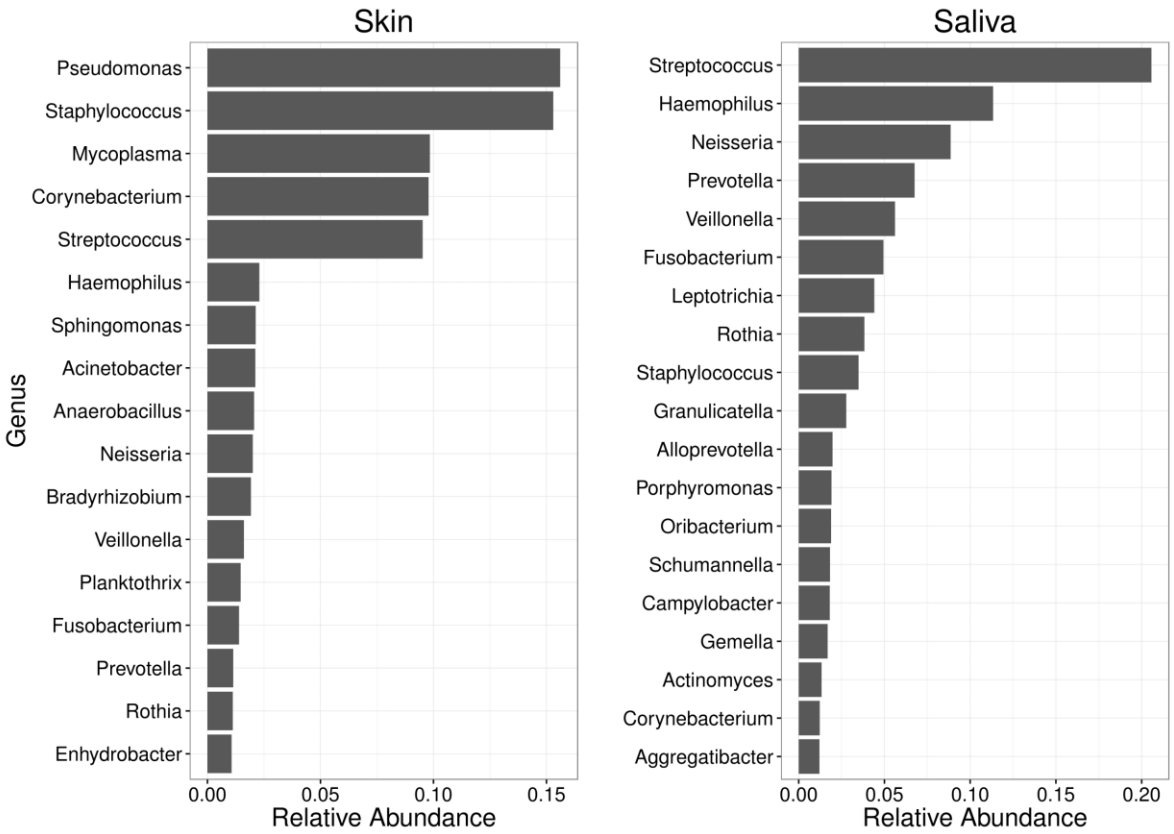
- Nasal

Bacterial composition



MicroFLOQ Direct for Direct Amplification

Rapid Processing of Crime Exhibits

Standard GF
~ 5 hrs

Maxwell 16
1 hr

Quantitation
2 hrs

GF amp
1.5 hr

3500xL CE
0.75 hr

RapidHIT
~ 2 hrs

RapidHIT
2 hrs

DNA Profile Interpretation

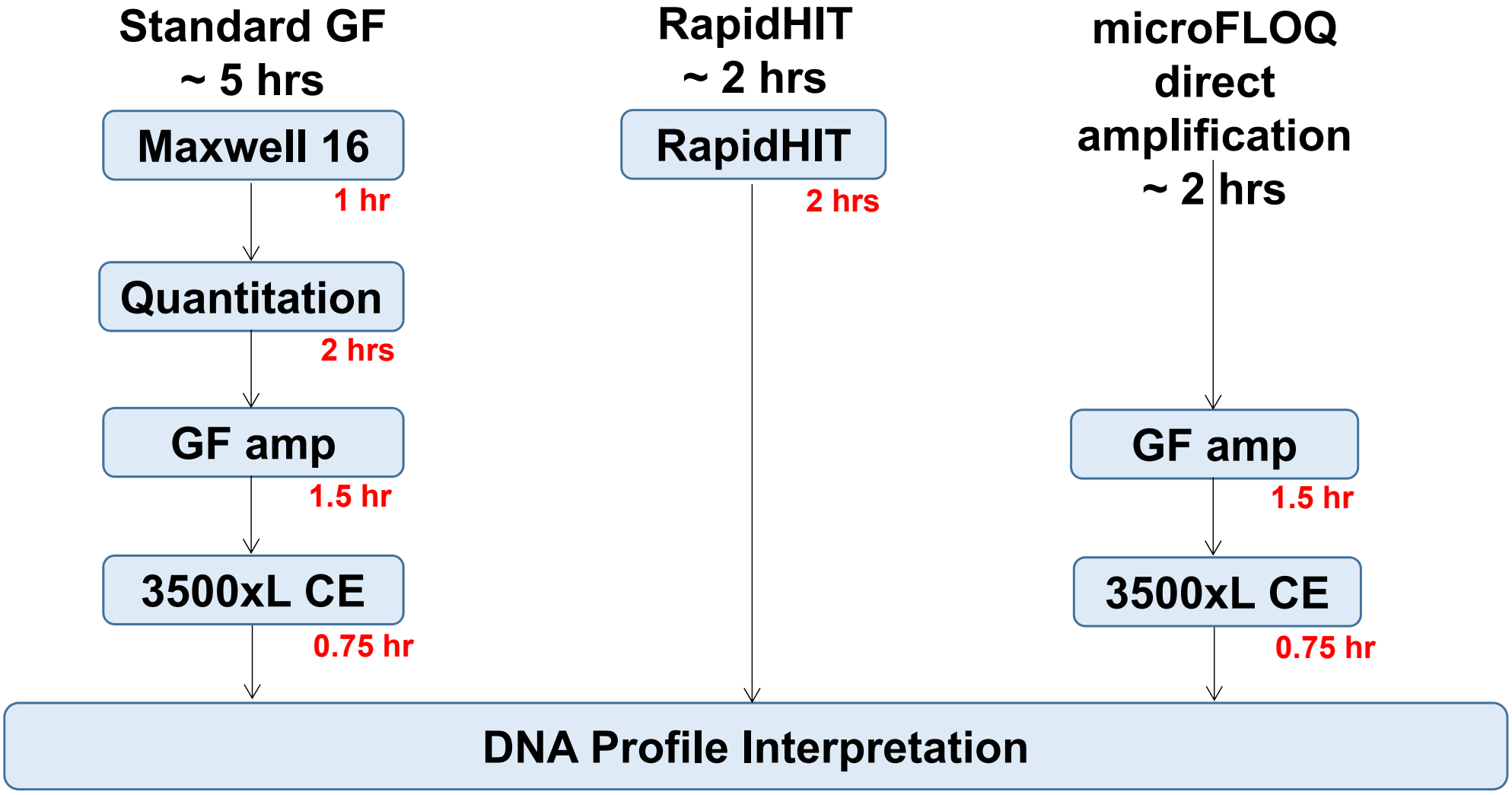
Rapid Processing of Crime Exhibits

RapidHIT

- Low sensitivity
  - Buccal swab – 80% success rate (based on ≥ 50% alleles detected)
  - 50 µL blood on swab – 94% success rate
  - Variability between RapidHIT units
- Multiple technical issues
  - Faulty capillary board
  - Faulty USB ports *(Data cannot be transferred out)*
  - Faulty in sensor of the motor *(MAXTRAVEL error)*
  - Faulty software issue *(Timeout error)*
  - Faulty capillary bundle

	RapidHIT						Standard protocol	
	Unit 1			Unit 2				
Volume of Blood (µl)	Alleles Called (%)	Mean Peak Height (RFU)	Peak Height Ratio (%)	Alleles Called (%)	Mean Peak Height (RFU)	Peak Height Ratio (%)	Alleles Called (%)	Peak Height Ratio (%)
50	98 – 100	11,608	68 – 100	100	8,286	88 – 90	100	87 – 92
1	78 – 100	635	28 – 99	93 – 100	919	59 – 74	100	87 – 93
0.5	39 – 100	431	23 – 95	68 – 93	433	64 – 71	100	80 – 92
0.125	17 – 67	85	22 – 95	11 – 52	120	56 – 74	78 – 100	72 – 88

Rapid Processing of Crime Exhibits



- Easy integration into a laboratory's workflow
  - ➢ PCR machine
  - ➢ CE machine

Overview of MicroFLOQ™ Workflow

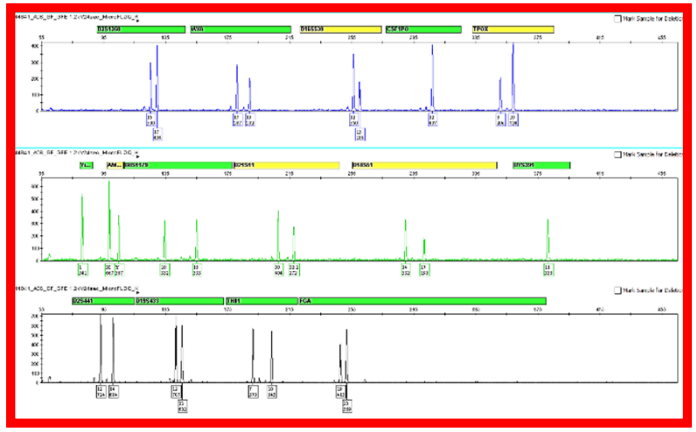
SAMPLE → **Whole process within ~2 hours** → **DNA RESULTS**

Sample collection using microFLOQ™ direct (~5 - 10 minutes)

1 mm tip coated with short nylon fibers that are treated with a lysis agent that allows cell lyse for direct DNA amplification.

Break tip for PCR amplification (Globalfiler) (~1.5 hours)

Capillary electrophoresis & STR Analysis (~0.75 hour)



No ‘residue’ or DNA extract for retention

Our Approach towards MicroFLOQ™

FORENSIC SCIENCE INTERNATIONAL GENETICS | Official Journal of the International Society for Forensic Genetics

Direct PCR amplification of DNA from human bloodstains, saliva, and touch samples collected with microFLOQ® swabs

Angie Ambers, Rachel Wiley, Nicole Novroski, Bruce Budowle

Published Online: October 26, 2017

Open Access | PlumX Metrics

Published online Oct 2017.

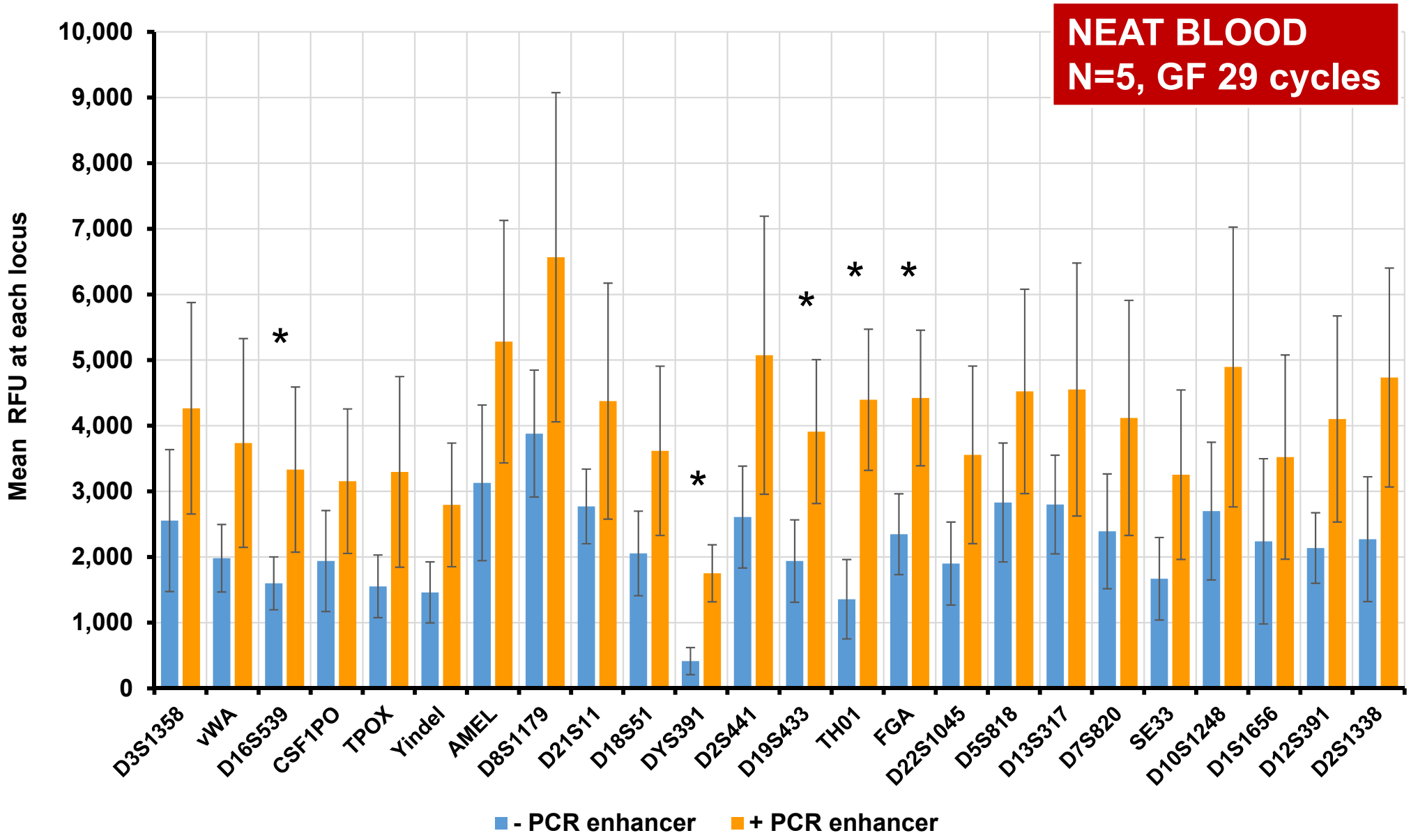
- Samples on sterile glass microscopic slides.
- Besides neat, various dilutions of samples were used.
- Compare microFLOQ with 4N6 FLOQSwabs.

Blood	Saliva	Substrate	Amp Kit microFLOQ	Status of fluid and swabbing methods
10ul of 10%, 5%, 1% blood	**10ul of 100%, 10%, 5%, 1% saliva**	**Microscopic glass slide**	**GFE 28 cycles**	**Dried fluid with wet or dry swab**

Our approach and areas of considerations

- Various stains (blood, semen, saliva) on various substrates that are commonly encountered for DNA casework

PCR Enhancer with MicroFLOQ™



* Significantly higher with PCR enhancer

DNA Collection Method

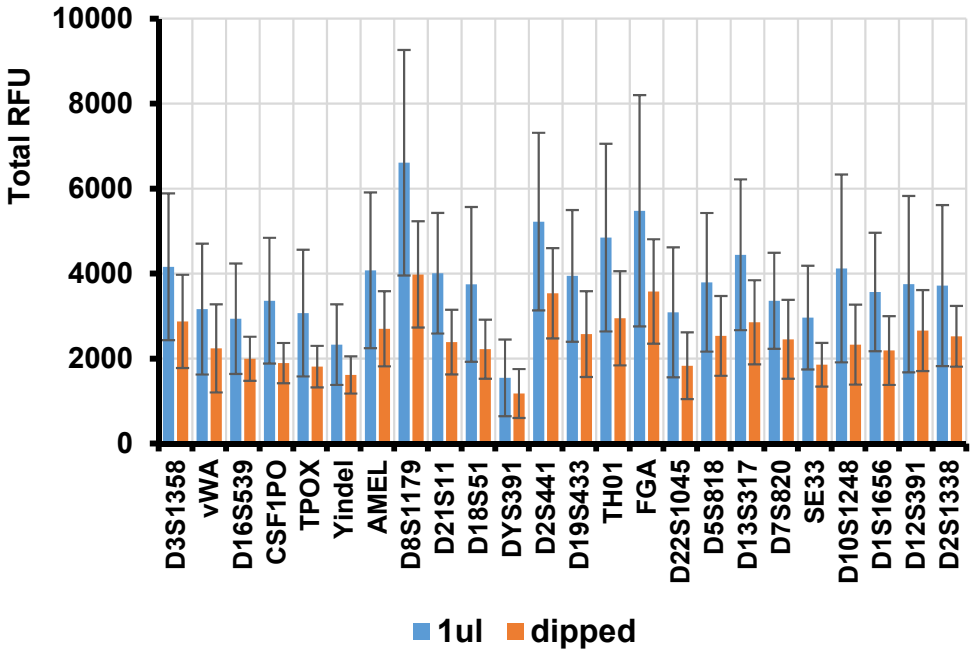
To collect dry traces, Copan recommended moistening the swab with 1ul of water

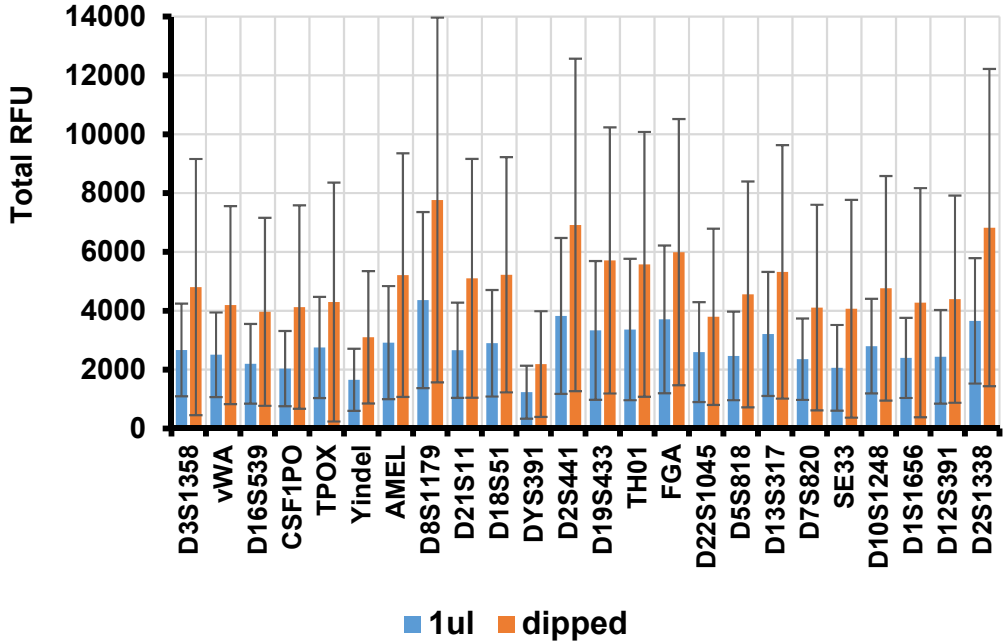
- Additional steps to prepare/pipette water onto the swab
- Not practicable to use this method at crime scenes

More steps might lead to increased chance of contamination

Investigated alternative method: contacting the swab with the surface of water

- No particular trend when comparing the 1ul and the alternative method

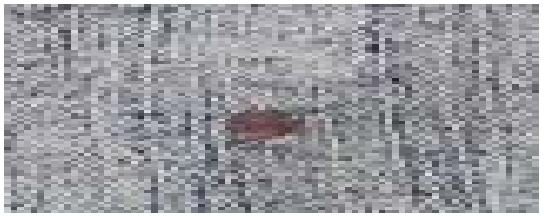


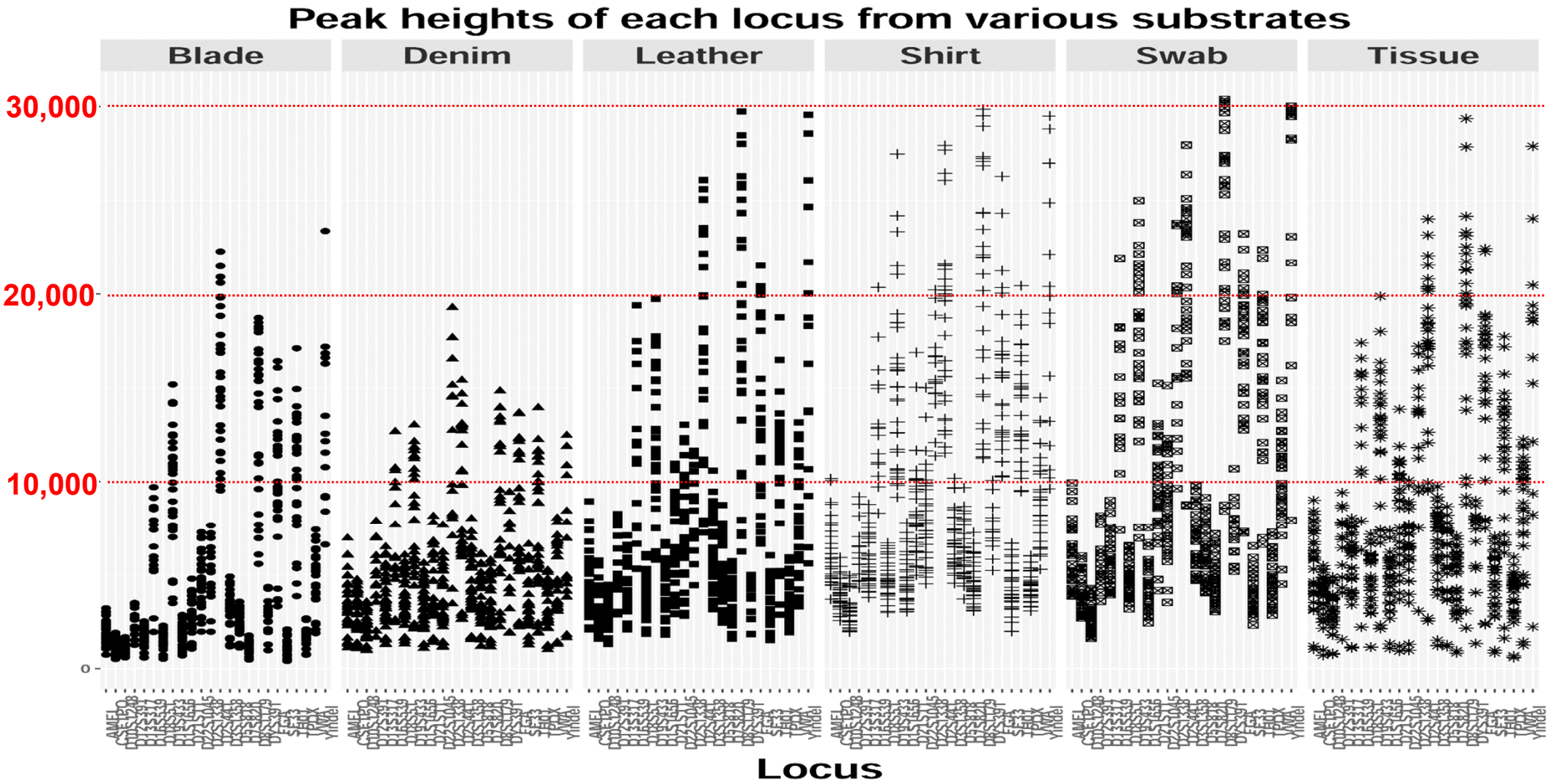


NEAT BLOOD

Use of MicroFLOQ™ on Blood-stained Substrates

- Blood-stained substrates (n=15 each)
- Lowest RFU obtained well above lab analytical and stochastic thresholds

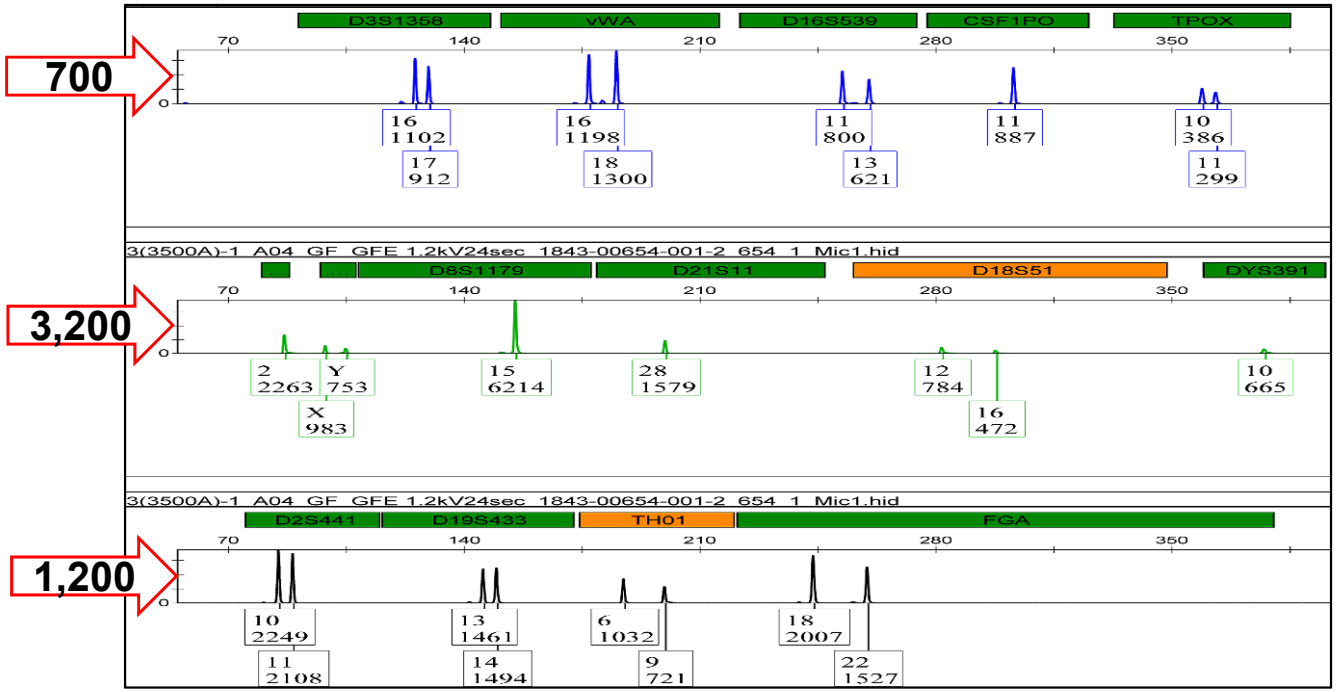





Case Study on Crime Casework

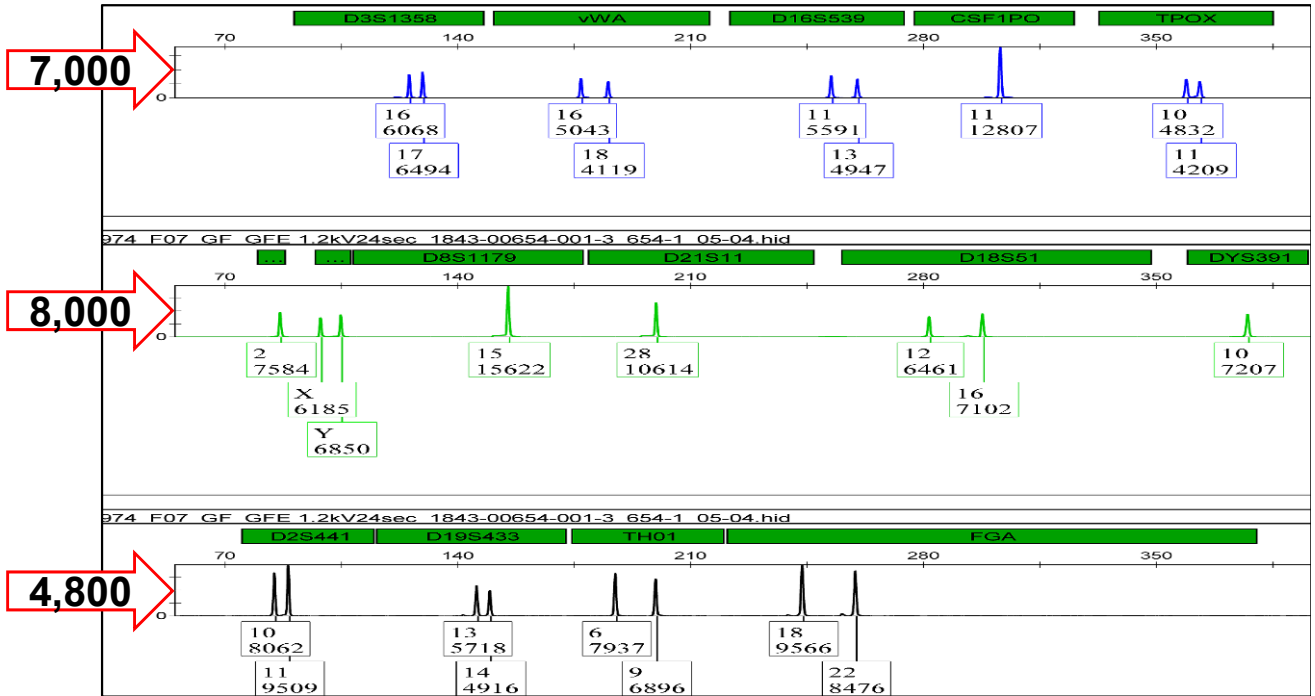
Results of swabbing a blood-stained cotton swab with **microFLOQ™ swab**

- Balanced PHR
- Although lower peak height than standard lab protocol
- No dropout



Results of processing the blood-stained cotton swab using **standard laboratory protocol**

- Balanced PHR
- Higher peak height than microFLOQ ™
- microFLOQ™ results are concordant with standard procedure



Ongoing Studies

Assess the suitability of using microFLOQ swab on

- different types of semen-stained substrates.
  - Swabs
  - Textiles (shirt and jeans)
  - Tissue paper
  - Condoms

- different types of saliva-stained substrates.
  - cigarette butts

- different types of touch samples.

EDNAP 30 Meeting

Innsbruck, Austria, Oct 30 2018

Book directly with hotel
(Ms. Tanja Federbusch)
- accommodation
- registration (incl. transport)

We reserved 20 + 20 rooms
until July 02 2018

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