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EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

Athens, Greece

25 October 2017

Host: Maria Vouropoulou Chairman: Peter Schneider A list of participants is attached.

Welcome

Penelope Miniati welcomed members to Athens on behalf of the Hellenic Police. Peter Schneider forwarded greetings to all participants from Niels Morling who is unfortunately unable to attend the meeting.

<u>Presentation of the Hellenic Police Laboratory</u> Katerina Kondili gave an overview on the structure and workflow of the DNA laboratory of the Hellenic police forensic science division (presentation attached).

Update on exercises

<u>Second exercise on methylated DNA and age prediction</u> 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 (presentation attached).

Exercise on mRNA typing with NGS, part 2

Cordula Haas presented the results of the second collaborative EDNAP exercise on NGS based study of discrimination between various body fluids, including the sequence analysis of coding SNPs (presentation attached).

mtDNA quantification collaborative exercise

Arnoud Kal presented an update on the preparation of a new exercise on mtDNA quantification by real time PCR. An invitation to participate will be sent out after the meeting, the results will be presented at the next meeting in April 2018 (presentation attached).

Updates from other groups

Forensic science and humanitarian identification - DNA analysis and centralized DNA database

Penelope Miniati presented the current situation of migrants entering Greece via the Aegean Sea and the problems with the identification of deceased refugees, as well as the reunification of separated families (presentation attached).

ArnoudKal

PenelopeMiniati

CordulaHaas

High quality DNA sequence database - STRidER

Walther Parson informed about the update of the website, https://strider.online. Colleagues are invited to submit population genetic data to the database. STRidER will be used as a quality control tool and data repository for population genetic studies to Forensic Science International: Genetics. The website will be ready very soon to provide instructions about the submission format and procedure (presentation attached).

EMPOP update

Walther Parson gave a short update about new analytical tools (SAM2) for mitochondrial DNA data alignment. The new software will be implemented into the EMPOP website in Q1 of 2018 (presentation attached).

ISFG report

Walther Parson gave an update of the activities of the ISFG and the outcome of the recent congress in Seoul (presentation attached). Two new calls for short term fellowships are planned for March and October 2018.

The EU-funded project 'VISAGE'

The project has started on 1st May 2017 and will run for 4 years. Analytical tools using SNP and methylation markers to predict appearance, age and ancestry by using MPS technology will be developed and validated by extensive user testing. In this context, there may also be a role for EDNAP at a later stage (presentation attached).

EUROFORGEN-NoE – Final outcome and activities

Peter Schneider gave a final update on the project that is now associated with the ISFG. There will be a Forensic Genetics Summer School in the years between the international congresses. The first Summer School will take place in September 2018 in Southern Italy in connection with the Italian speaking DNA working group meeting (Ge.F.I.) and will have a format similar to the pre-congress workshops at the ISFG congresses (presentation attached).

<u>Plans in Germany to introduce forensic DNA phenotyping</u> Peter Schneider Peter Schneider gave an overview on the origin and current situation to introduce legislation allowing the application of forensic DNA phenotyping using ancestry, pigmentation markers and age prediction in criminal investigations. It can be expected that the legislative process will be resumed next year, after the new German government has been formed (presentation attached).

ENFSI Update

Sander Kneppers gave a presentation on the structure of ENFSI and the recent and future activities of the EDNAP DNA Working Group (presentation attached).

Other activities

Verbal scale discussions

Peter Gill gave a presentation on the use of verbal scales to express the strength of evicence following likelihood ratio calculations. We participates in a SWGDAM commission representing ENFSI which is addressing this issue for the forensic DNA community in the US (presentation attached).

Database searches revisited.

Peter Gill gave a presentation addressing the biostatistical treatment of hits following DNA database searches. He points out that different propositions have to be considered within an ongoing criminal investigation compared to the courtroom scenario (presentation attached).

Walther Parson e outcome of the

WaltherParson

Peter Schneider

Walther Parson

SanderKneppers

PeterGill

PeterGill

Walther Parson

Future activities

No other collaborative exercises are planned in addition to the exercise on mtDNA quantification in preparation by the NFI (Arnoud Kal, see above).

Next meetings

At the ENFSI Steering Group meeting, the colleagues from Rome informed the group that they are planning the EDNAP/CODIS/ENFSI meeting in 2018 during the week 16-20 April 2018.

For the second meeting in 2018 an invitation was extended by Walther Parson to come to Innsbruck. The planned dates will be October 30th for the ENFSI Steering Group meeting and October 31st for the EDNAP meeting. At this occasion, the 30th birthday of the EDNAP group will be celebrated.

Any other business

There was no other business.

Closing of the meeting

The meeting closed with sincere thanks to Maria Vouropoulou and all other colleagues, who helped to organise the meeting.

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

- Agenda
- List of participants
- Presentations
 - Katerina Kondili: Presentation of the Hellenic Police Forensic Science Division
 - o David Ballard: Report on methylated DNA and age determination
 - o Cordula Haas: Results from the second collaborative exercise on mRNA by NGS
 - Arnoud Kal: Update on the collaborative exercise on mtDNA quantification
 - Penelope Miniati: Forensic DNA analysis and humanitarian identification in Greece
 - Walther Parson: STRidER report
 - Walther Parson: EMPOP report
 - o Walther Parson: ISFG report
 - Walther Parson: VISAGE update
 - o Peter Schneider: Report on EUROFORGEN-NoE
 - Peter Schneider: Plans for FDP legislation in Germany
 - Sander Kneppers: ENFSI Update
 - Peter Gill: Verbal scale discussions
 - Peter Gill: Database searches

Peter Schneider

Peter Schneider

Peter Schneider

C/N	53rd EDNAP MEETING		OITY	COUNTRY			
5/N	PARTICIPANT'S NAME	ADDRE55	CITY	COUNTRY	EWAIL		
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FORENSIC SCIENCES DIVISION

Subdivision of Biological and Biochemical Examinations and Analyses

> Aikaterini Kondili PhD Police Colonel-Biochemist



Subdivision of Biological and Biochemical Examinations and Analyses

Administration Section Biological Materials Analysis Section

National DNA Database Section Scientific Support Section



INCOMING CASES 2007-2017





type of cases 2017 (JAN-SEP)











61 people personnel

- 26 DNA experts
 - 15 hold PhD
- 16 technicians
- 19 Police officers



Training programme

- ENFSI DNA WG / Quality Assurance Programme, ------
- ENFSI DNA WG / Concept training document, Nov. 2010



- Public servants
- CEPOL webinars, on line modules



performance

per year

- 20.000 pieces of evidence
- 30.000 PCR
- 3.000 reports
- 5.000 profiles searched
- 600 matches

equipment

- 2 AB 3500xl Genetic Analyzers
- 3 Qiagen Biorobot Universal
- 3 9700 GeneAmp PCR
- 3 AB Veriti 96 PCR
- 1 AB 7500 RT-PCR

Where we want to go :

- Fully automated lab for DNA typing of reference samples
- LIM system
- > One fully automated line for DNA typing in the casework lab
- Massively Parallel Sequencing technology in the Subdivision



Where we are :

> Fully automated line for DNA typing of reference samples

Internal validation on BRU platforms using the Globalfiler express PCR amplification kit for the direct amplification of reference samples





Where we are :

> LIM system

To be installed in the 1st Section by the end of October

- > One fully automated line for DNA typing in the casework lab
- > Massively Parallel Sequencing technology in the Subdivision

Submission to Ministry of Interior a Proposal for funding from the European Internal Security Program





Thank you



Methylated DNA & Age Exercise



EDNAP, Athens 2017



EDNAP Exercise

- 15 laboratories participated
 - 8 MiSeq only
 - o 5 PGM only
 - 2 MiSeq and PGM/S5
- Part 1 7 Methylation standards between 0-100% sent out to all labs
- Part 2 7 blood stains sent out to laboratories to test. Also optional submission of extra blood samples. Age prediction by ANN from methylation values at 12 markers.

Combined prediction results from EDNAP labs for samples A-G



Average Prediction Error Per Laboratory

Blind age predictions of extra MiSeq results

MiSeq - Venus Blood



Blind age predictions of extra PGMresults

PGM - Venus Blood





Blood/saliva samples



Saliva samples normalised to blood data



Next steps

• Paper is being written up, will be circulated when ready.

Acknowledgments

Anastasia Aliferi Athina Vidaki Leon Barron Denise Syndercombe Court

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DAVID BALLARD DNA ANALYSIS AT KING'S KING'S COLLEGE LONDON LONDON UK

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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

25. October 2017, Athens



targeted mRNA NGS approach for the identification of blood, saliva, semen, vaginal secretion, menstrual blood, skin

- > 14/17 labs successfully implemented the mRNA NGS approach
- MiSeq-assay satisfying, PGM-assay needs some optimization
- PGM protocol is more lab-work
- manual RNA-extraction!
- Manuscript under review

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^I, M. Kayser^m, F.-X. Laurentⁿ, N. Morling^I, 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 Berge¹, K.J. van der Gaag¹, J. Vannierⁿ, V. Verdoliva^d, A. Vidaki^m, C. Xavier^q, J. Ballantyne^{b,c}, C. Haas^a





- only **MiSeq** laboratories (1/2 library kit left from exercise 1)
- targeted mRNA NGS approach for the identification of blood, saliva, semen, vaginal secretion, menstrual blood, skin and cSNPs assay to associate specific mRNA transcripts to an individual (separate assays)
- RNA extraction (manual or kit), DNase treatment, quantification
- Protocols and primerpools were provided
- Laboratories analyzed 12 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	Gana	mRNA	cSNPs
fluid/tissue	Gene	33plex	35plex
	ALAS2		
	ANK1		4
Blood	SPTB		
bioou	CD3G		
	CD93		3
	AMICA1		2
	PRM1		
	PRM2		
Formore	TGM4		4
Semen	SEMG1		
	SEMG2		2
	KLK3		
	HTN3		
	HTN1		
	STATH		
Saliva	PRB3		
	PRB4		
	PRH2		
	MUC7		2
	CYP2B7P1		
	DKK4		
Vaginal	FAM83D		
	CYP2A6		
	CYP2A7		2
	MMP10		2
	LEFTY2		
Menstrual	MMP7		
	MMP11		
	SFRP4		
	LCE1C		2
	CCL27		
Chin	IL37		
SKIN	SERPINA12		
	KRT77		2
	COL17A1		3



Provided stains

stain number		composition	primer pool 1 (mRNA)	primer pool 2 (cSNPs)		
1	5	0 μL blood on swa	х	х		
2	50	<mark>Ο μL semen on swa</mark>	х	х		
3	5	0 μL saliva on swa	х	х		
4	1/4	vaginal secretion s	х	х		
5	1/4	menstrual blood s	х	х		
6		skin swab	х	х		
7	25 μL blood	25 μL semen		х	х	
8	25 μL saliva	25 μL semen		х	х	
9	12.5 μL saliva	1/4 vaginal swab		х		
10	12.5 μL saliva	1/4 mens. swab		х		
11	7 μL blood	7 μL saliva	1/4 vaginal swab	х		
12	25 μL semen	25 μL saliva	skin swab	х		
13	25 μL blood	25 μL blood			х	
14	25 μL blood	25 μL saliva			х	
15	12.5 μL blood	1/4 vaginal swab			х	
16	12.5 μL blood	1/4 mens. swab			x	



Participating laboratories

Copenhagen, Denmark Innsbruck, Austria London, UK* Lyon, France NFI, Netherlands* NIST, USA Orlando, Florida, USA Rome, Italy Rotterdam, Netherlands* Zurich, Switzerland



Summary Questionnaire

- Delivery time Fedex (samples+primers): all labs within 1-2 days
- 2x manual RNA extraction, 6x RNA extraction kit (RNeasy, mirVana)
- 5x RNA quantification (Qubit, RiboGreen, Quantus), 2x no quant
- RT: 8x ProtoScript II Reverse Transcriptase



RNA-Quantification

[ng/µL]	extraction method	quantification method	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16
Lab_1	Rneasy Mini	Qubit	undet	undet	undet	292	62	undet	undet	undet	90.4	25.6	49.8	7.2	undet	4.4	122	15.7
Lab_2	Rneasy Mini	Qubit	undet	undet	undet	>	38	undet	undet	undet	55	29	31	undet	undet	undet	8.67	undet
Lab_3	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?
Lab_4	Rneasy Mini	no quant	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Lab_5	Rneasy Mini	Qubit	undet	undet	2.4	85	7.9	undet	undet	undet	58	34.4	25.7	2.1	undet	undet	87	10.7
Lab_6	manual	Quantus	22	21	18	270.5	131.5	0.97	22	8.8	251	89.85	566.5	35.75	18	11	242	68.75
Lab_7	manual	Quant-iT RiboGreen	5.4	19.7	5.8	202.5	42.8	undet	14	1	205.7	129.1	210.9	11.1	8	2.6	168	139
Lab_8	mirVana	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?
Lab_9	Rneasy Mini	no quant	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Lab_15	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?

Comparison among laboratories (mRNA assay, single stains)












Comparison mRNA-cSNP assays













Stain number 8: saliva (3) – 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)	СТ	CG	TA	CA	AG	GG	GG	GG
saliva-semen mix (RNA)	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

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	ТС
blood-blood	G: 32'825	G [.] 20'650	G [.] 29'210	TC
	A: 15'081	A: 5613	A: 6737	C: 47'364

Sample/Marker	ANK1_1		ANK1_4		AMICA1_1		AMICA1_2	
Donor 1 (DNA)	G	G	G	А	G	Α	Т	Т
Donor 2 (DNA)	G	Α	G	G	G	G	Т	С



Stain number 14: blood – saliva mixture

(blood 4 SNPs, saliva 0 SNPs, remaining 7 blood not discriminatory for this example, 2 saliva SNPs no coverage, 1 saliva SNP not discriminatory)

- \rightarrow Blood can be assigned to donor 1
- \rightarrow 98% of reads align to blood amplicons
- → More saliva SNPs needed

Sample/ Marker	AMICA1_1	AMICA1_2	ANK1_1	ANK1_4
Donor 1 (DNA)	GG	TC	GA	GG
Donor 2 (DNA)	AG	ТТ	GG	GA
blood-saliva mix (RNA)	GG G: 16'532 A: 18	TC T: 24'984 C: 39'451	GA G: 9996 A: 8861	GG G: 17'007 A: 5613

Gene Name	Stain number 14
AMICA1	16562
AMICA1	62972
ANK1	6714
ANK1	18835
ANK1	17033
ANK1	2433
CD3G	3508
CD93	6398
CD93	4489
CD93	5742
SPTB	3322
KLK3	0
SEMG1	0
SEMG2	0
SEMG2	0
TGM4	0
PRB3	0
MUC7	0
MUC7	1506
CYP2A7	0
CYP2A7	0
DKK4	0
MMP10	2
MMP10	4
MMP7	0
KRT77	0
KRT77	0
LCE1C	2
LCE1C	4
COL17A1	0
COL17A1	0
COL17A1	0

Stain number 15: blood (\bigcirc) – vaginal secretion mixture

(blood 1 SNP, vaginal secretion 1 SNPs, remaining 10 blood SNPs not enough coverage, remaining 2 vaginal secretion SNPs not discriminatory for this example)

- \rightarrow Inconclusive result
- \rightarrow More vaginal secretion SNPs needed

→ Skin marker LCE1C supports assignment of vaginal secretion to donor 2

Sample/ Marker	AMICA1_1	CYP2A7_1	LCE1C_1
Donor 1 (♀) (DNA)	AG	CC	AA
Donor 2 (DNA)	GG	AC	GG
blood - vag mix (RNA)	GG? G: 1131 A: 0	AC A: 402 C: 1993	GG G: 1011 A: 1

Gene Name	Stain number 15
AMICA1	1133
AMICA1	79
ANK1	638
ANK1	21
ANK1	3
ANK1	4
CD3G	5
CD93	4
CD93	4
CD93	1
SPTB	5
KLK3	2
SEMG1	29
SEMG2	10
SEMG2	8
TGM4	0
TGM4	2
TGM4	8
TGM4	7
PRB3	0
MUC7	0
MUC7	0
CYP2A7	2097
CYP2A7	2462
DKK4	2437
MMP10	0
MMP10	3
MMP7	0
KRT77	0
KRT77	1
LCE1C	1025
LCE1C	0
COL17A1	0
COL17A1	0
COL17A1	0

Stain number 16: blood (♀) – menstrual blood mixture

(blood 5 SNP, menstrual blood 0 SNPs, remaining 2 blood SNPs not discriminatory / 4 SNP not enough coverage, remaining 3 menstrual blood SNPs not discriminatory for this example)

 \rightarrow More menstrual blood SNPs needed

 \rightarrow 3 of 5 blood SNPs show alleles from blood and menstrual blood

 \rightarrow 2 of 3 vaginal secretion SNPs discriminatory \rightarrow assignment of menstrual blood still possible

Sample/ Marker	ANK1_1	ANK1_4	CD93_1	CD93_2	CD93_3	CYP2A7_1	CYP2A7_2
Donor 1 (♀) (DNA)	GA	GG	AG	ТТ	GA	AA	ТС
Donor 2 (DNA)	GG	GA	GG	CC	AA	CC	ТТ
blood – mb mix (RNA)	GA G: 2526 A: 3450	GG G: 1153 A: 1	(A)G A: 273 G: 3024	CT T: 1629 C: 1352	AA G: 20 A: 5832	C: 905 A: 0	TT T: 1750 C: 1



Zurich Institute of Forensic Medicine

Thanks for participating!





Netherlands Forensic Institute Ministry of Justice

EDNAP mini-Exercise proposal mtDNA quant

24 October 2017, Athens

Kris van der Gaag 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	Вр	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

Standard reference material

in progress
 in progress
 in progress



Are you interested?

NFI provides:

- •Primers and probes
- •Challenging samples
- Protocols

You provide:

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

We will send you an invitation via email soon!





Forensic Science and Humanitarian Identification

DNA Analysis & Centralized DNA Database

Athens, 25 October 2017

Dr. Penelope Miniati, Ph.D.

HELLENIC POLICE – FORENSIC SCIENCE DIVISION



HELLENIC POLICE – FORENSIC SCIENCE DIVISION







FORENSIC SCIENCE DIVISION (F.S.D.) is:

✓ the National Forensic Service of Greece

 One of the five Central Divisions of the Hellenic Police, that reports directly to the Chief of the Hellenic Police Forces

Accreditations

- Certification: ISO 9001
- Accreditation : ELOT-EN ISO/IEC 17025 / 17020



- 6. Chemical and Physical Examinations
- 7. Digital Examinations
- 8. Photography / Voice & Video Analysis
- 9. Statistical Data
- 10. Archives

Furthermore the F.S.D. supervises the Forensic Science Subdivision of Northern Greece (F.S.S.N.G.), as well as 11 Forensic Science Sections (F.S.S.) and 53 Forensic Science Offices (F.S.O.)



DNA in Hellenic Police



Greece has been a popular transit route for migrants and refugees many decades now – Meeting point of two/ three continents DNA analysis for identification purposes has been used in Greece since the mid '90s



Evros borders

• Large influx of non paper migrants / refugees



Evros Fence



12.5 km barbed-wire fence
along the land border with
Turkey succeeded in blocking
one of the most popular
transit routes for migrants
and refugees seeking to make
their way to the West

Aegean islands borders



The sharp drop in
undocumented migrants/
refugees entering Greece
through Evros has been
accompanied by a tremendous
increase in the influx via the
islands of the Aegean Sea.

During 2015 – 2016 more than one million people have travelled through Greece to different European countries







"HELIOS" Airplane Crash



"HELIOS" Airplane Crash



92 DNA Identifications

Extensive Fires in Peloponnese



53 victims identified through DNA

The impressive speed and accuracy in the identification of victims at the foretold Mass Disasters (Helios Airplane Crash, Large Fires in the Pelononnese)

proved that DNA was a very "useful method" for the identification of deceased - Disaster Victim Identification.

Identification needs - DNA analysis

*****Identification of Deceased Migrants / Refugees

*Family reunification

• Non-accompanied Children - Determination of Familial Relationships:

- A case of a 14-year old refugee

a future challenge

"Unidentified deceased refugees / migrants abroad"

Interpol/Europol Requests

DNA Database Search

Interpol/Europol




President of Interpol's DVI Steering Committee Carlos Eduardo Palmares Machado





Ημερίδα με θέμα: Ορθές πρακτικές ταυτοποίησης νεκρών αγνώστων στοιχείων, με έμφαση στη διαχείριση δεδομένων

Roundtable on best practices for the identification

of unidentified bodies, with emphasis on data management



Aθήνα 4 Οκτωβρίου 2016 Athens 4 October 2016 Best Practices in Human Identification with emphasis data collection – Organized by ICRC and FSD Athens, 4 October, 2016



These Meetings resulted

•High Officials such as the Attorney General, the General Secretary of the Ministry of Interior, the Chief of Police etc. in supporting the cause

•High Officials from all Ministries involved and from the Data Protection Agency working together not only to solve every day problems but also to standardize the procedures.



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Athens DNA lab helps trace those lost at sea on Aegean crossing

Mediterranean crisis sees DNA lab in Athens go from dealing with about 30 cases a year on average to as many as 70 in two days.

By: Maria Petrakis, eds: Andrew Dobbie/Jonathan Clayton | 5 April 2016 | Français | عربي |



TOP NEWS

Wed Aug 17, 2016 | 4:46 PM EDT

Naming the nameless: experts struggle to identify drowned migrants



Aug 17, 2016 | 01:23 Forensic teams in Greece struggle to identify drowned migrants



euronews.c



Η δύσκολη δουλειά της ταυτοποίησης των μεταναστών που πνίγονται στο Αιγαίο

The use of Centralized Databases

The example of centralized DNA Database

National DNA Database





National DNA Database

According to the Greek Law (recently modified):

• There is a **centralized data base**.

• "...All the state and institutional laboratories, performing DNA analyses following orders of judicial or investigate authorities, submit the analyses results to the National DNA Database..."

Hellenic DNA Database includes

- 700 DNA profiles from Unidentified Human Remains (UHR) – the majority of them refer to refugees / immigrants
- >250 reference samples
- >186 pedigrees

<u>Reference Material cannot be accepted by non-</u> <u>governmental agencies or individuals</u>

- Police and Coast Guard (Judicial Order)
- Embassies
- Interpol Offices

All ante-mortem information are important for identification purposes and non-governmental agencies play a crucial mediator role between the families that are searching for their loved ones and the governmental agencies

Shipwreck between the islands of Kalymnos and Kalolimnos (29-10-2015, 02:50)

- 160 people on board
- 139 people saved
- 19 people drowned
- 2 people were missing





Syrian citizen declared to the Police Authorities of Orestiada that her husband went missing, after embarking on a boat heading from the Turkish coast to the island of Lesvos (28-10-2015)

The couple's two children ptovided DNA samples to the Police Authorities in Orestiada (20-12-2016)

HIT to the database with the DNA profile of a deceased person found in the sea region between the islands of Lesvos and Samos in 2015 (DNA profile submitted by the DNA lab of the Forensic Medical Service of Athens 16-05-2016)



 It is proven once more that the use of centralized DNA Databases is not limited only in providing valuable information to the law enforcement, but also providing valuable information to families that have lost their loved ones, so the grieving process will start for them.





HELLENIC POLICE FORENSIC SCIENCE DIVISION

e-mail: p.miniati@hellenicpolice.gr

Thank you!!!

ΕΔΝΑΠ Μεετιγγ, Άθενς, Γρεεχε, Όχτοβερ 25 2017



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	

ΣΤΡιδΕΡ Ύπδατε

Δρ. Ωάλθερ Παρσον άσσοχ. Προφ. Ίνστιτυτε ὀφ Λεγαλ Μεδιχινε, Μεδιχαλ Ύνιψερσιτυ ὀφ Ίννσβρυχκ, Αὐστρια αδϑ. Προφ. Φορενσιχ Σχιενχε Προγραμ, Πενν Στατε Ύνιψερσιτυ, ΠΑ, ΎΣΑ walther.parson@i-med.ac.at



HOME QUERY BATCH QUERY ABOUT FREQUENCIES FORMULAE STR SEQUENCE NOMENCLATURE CONTACT TERMS OF USE

Welcome to STRIdER!

STRidER (STRs for Identity ENFSI Reference Database) is the expanded and enhanced version of the ENFSI STRbASE (2004-2016). This curated anline high quality STR allele frequency population database enables scientifically reliable STR genotype probability estimates and provides quality control of autosamal STR data. A suite of software taols has been developed at the Institute of Legal Medicine, Medical University of Innsbruck (UINK: https://gerichtsmedizin.at/) to scrutinize STR population data and thus increase the quality of datasets to ensure reliable allele frequency estimates. STRidER acts as frequency database and software platform for the development of novel tools for STR data QC and other forensic analyses.

STRidER serves the STR community in forensics and beyond in inter-related ways:

- The high-quality autosomal STR allele frequency database can be directly queried
- Allele frequency tables of STR laci from diverse populations can be downloaded and used for third party saftware
- · Centralized STR data quality control is offered prior to publication
- Accepted datasets will become rapidly available anline and receive a unique and traceable STRidER accession number
- Allele frequencies and forensic/population genetic parameters are calculated from datasets
- Individual STR genatypes are not accessible on STRidER to comply with privacy regulations



STRidER in the field of forensic STR typing (from Bodner et al. 2016)

The concept of STRidER has been developed together with the DNA Commission of the ISFG) and is outlined in Bodner M, Bastisch I, Butler JM, Fimmers R, Gill P, Gusmãa L, Morling N, Phillips C, Prinz M, Schneider PM, Parsan W (2016) 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); Forensic Sci Int Gen 24:97-102.

The STRidER online platform is work in progress. Additional datasets and features will continuously become available. To receive periodic news and stay updated about STRidER, register here for the STRidER newsletter.

Please consider citing STRidER [https://www.isfg.org/Publication;Bodner2016] when using it with your research

new URL https://strider.online/



CrossMark

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 Prinz¹, Peter M. Schneider^m, Walther Parson^{a,n,*}

STRidER newsletter

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,*}

Content

- Positioning STRidER relative to other existing databases (STRbase, ALFRED, pop STR, popAffiliator, ALLST*R); important element of QC
- II) Rationale, concept and workflow of QC via STRidER
- **III)** Benefits to forensic and other scientific community
- IV) Transparency, traceability and protection of data
- V) Outloook: **STR sequence data** in STRidER (MPS)





STRICER STRs for identity ENFSI Reference database, v2

ABOUT



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QUALITY CONTROL

(iSFG)

STR SEQ NOMENCLATURE CONTACT

STRICER STRs for identity ENFSI Reference database, v2

HOME BATCH OUERY ABOUT FREQUENCIES FORMULAE OUERY

OUALITY 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 OC 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 OC 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.

STRIDER STRs for identity ENFSI Reference database, v2



BATCH QUERY QUERY ABOUT STR SEQ NOMENCLATURE HOME FREQUENCIES FORMULAE QUALITY CONTROL CONTACT **CE** datasets submitted to STRidER for QC since 8/2017 **MPS** submitted sample no sample no cumulative 50 submitted sample no -----cumulative sample no

Errors found during STRidER QC

i i i i i i i i i i i i i i i i i i i	1	2	3	4	5	6	7	8	9
submitted sample no	550	1008	504	1220	100	100	101	102	102
sample no cumulative	550	1558	2062	3282	3382	3482	3583	3685	3787

QC finished

Note: QC is in progress

80 definitive errors in 3787 samples

2.1% of dataset profiles contained errors

doublets single alleles incomplete profiles wrong alleles (raw data inspection) non ascending alleles nomenclature errors wrong locus names

Note: statistics include errors found in datasets that were retracted - possible more errors, but QC was stopped

NCBI BioProject—STRseq

Mission: To provide high-confidence STR allele sequence records with uniform annotation, facilitating exchange of information across forensic laboratories.

- Collaborators with large datasets "seed" the BioProject
- NIST evaluates raw sequence data with agnostic bioinformatic pipeline
- GenBank record for all unique sequences
- BioProject searchable by string (BLAST), locus, allele...

Katherine Gettings

LOCUS DEFINITION ACCESSION	123456 196 bp DNA omo sapien microsatellite 5123456	linear PRI 19-Jan-2017 D21S11 sequence						
VERSION	/123456.1							
DBLINK	oProject: PRJNA12345							
ORGANISM	omo sapien							
REFERENCE	(bases 1 to 196)							
AUTHORS	ttings, K.B., Kiesler, K	.M., Steffen, C.R., Borsuk, L.A., and P.M. Vallone.						
TITLE	S. Population Sequence Data for 27 Autosomal STR Loci, 24 YSTR Loci and 7 XSTR Loci							
JOURNAL	rensic Science Internati	onal: Genetics						
COMMENT	Annotation ("bracketing") of the repeat region is consistent with the guidance of the ISFG (International Society for Forens Genetics), PMID: 26844919. Lower case letters in the bracketed repeat region below (rpt_unit_seq) denote uncounted bases.							
	ie given length-based all llele between individuals	ele value was determined using the designated length-based technology. Variation in the length-bas ; or assays can result from indels in flanking regions.						
	is information is provid prensic DNA community. Th maracterization, facilita echnology. For guestions	ed as part of the STR Sequencing Project (STRseq), a collaborative effort of the international e mission of this Project is to provide high-confidence STR allele sequence data and uniform ting exchange of information across forensic laboratories and compatibility with preceding or feedback, please contact strseq@nist.gov. Allele frequency data can be accessed in the						
	rider.online database.							
##humanSTR-STAR	ŕ							
Sample source	:: Genomic DNA	•						
Sequencing tech	logy :: MiSeq Foren	Seq						
Coverage	:: >30X							
Length-based al	21e :: 28							
Length-based te	1. :: ABI3500x1 G	lobalFiler						
STR locus name	:: D21511							
STR locus alt.	ime ::							
Chromosomal loc	ion :: 21q21.1							
GRCh38 coordina	es :: CHR21:19181	953-19182149						
GRCh38 repeat r	gion :: CHR21:1918)	1973-19182099						
##humanSTR-END#								
FEATURES	cation/Qualifiers							
variation	1010							
	/db_xref="dbSNP:rs1	123456″						
	/note="SNP A/C"							
repeat_region								
	rpt_type="tandem"							
	rpt_unit_seq= "[TC	CTA]4 [TCTG]6 [TCTA]3 ta [TCTA]3 tca [TCTA]2 tccata [TCTA]10"						
	satellitemicrosate]	llite="D21S11"						
variation	182182							
	/db_xref="dbSNP:rs1	12345″						
	/note="SNP C/T"							
ORIGIN								
ORIGIN 1 ATTCCCC#	G TGAATTGCCT TCTATCTATC 1	FATCTATCTG TCTGTCTGTC TGTCTGTCTG						
ORIGIN 1 ATTCCCCF 61 TCTATCTF	G TGAATTGCCT TCTATCTATC 1 C TATATCTATC TATCTATCAT (FATCTATCTG TCTGTCTGTC TGTCTGTCTG TTATCTATCC ATATCTATCT ATCTATCTAT						
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ORIGIN 1 ATTCCCC 61 TCTATCTA 121 CTATCTA 181 TGTCTCT	G TGAATTGCCT TCTATCTATC : C TATATCTATC TATCTATCAT (T ATCTATCTAT CTATCGTCTA 1 A GAACA	IAICIAICE TOTEICIEIC TETCIEICE JIAICIAICE AIAICIAICI AICIAICIAI ICIAICCAET CIAICIACCI CCIAITAEIC						

NCBI BioProject STRseq

Publication containing sequence

Description of STRseq

Metadata

Annotation

Sequence String

LOCUS DEFINITION DEFIN	AF123456 196 bp DNA linear PRI 19-Jan-2017 Homo sapien microsatellite D21511 sequence AF123456 AF123456.1 BioProject: PRJNA12345 Homo sapien 1 (bases 1 to 196) Gettings, K.B., Kiesler, K.M., Steffen, C.R., Borsuk, L.A., and P.M. Vallone. U.S. Population Sequence Data for 27 Autosomal STR Loci, 24 YSTR Loci and 7 XSTR Loci Forensic Science International: Genetics Annotation ("bracketing") of the repeat region is consistent with the guidance of the ISFG (International Society for Forensis							
	The given ler allele betwee This informat forensic DNA characterizat technology. E strider.onlir	ngth-based allele value was en individuals or assays ca tion is provided as part of community. The mission of tion, facilitating exchange for questions or feedback, y the database.	determined using the designated length-based technology. Variation in the length-bas n result from indels in flanking regions. the STR Sequencing Project (STRseq), a collaborative effort of the international this Project is to provide high-confidence STR allele sequence data and uniform of information across forensic laboratories and compatibility with preceding please contact strseq@nist.gov. Allele frequency data can be accessed in the					
##humanSTR-STAR	T##							
Sample source Sequencing tech: Coverage	:: nology :: ::	Genomic DNA MiSeq ForenSeq >30X 29	Kit / Instrument / Quality					
Length-based tec STR locus name STR locus alt.	ch. :: :: name ::	ABI3500x1 GlobalFiler D21511	Length-based allele					
Chromosomal loca GRCh38 coordina GRCh38 repeat_ro ##humanSTR-END#	ation :: tes :: egion :: #	21q21.1 CHR21:19181953-19182149 CHR21:19181973-19182099	Location on reference genome					
FFATURES	Location/Oual	ifiers						
variation	101 /db_x /note	0 ref="dbSNP:rs123456" ="SNP A/C"	SNPs or InDels					
repeat_region 2	21144 rpt_t rpt_u	ype="tandem" nit_seq= ``[TCTA]4 [TCTG]6 litemicrosstellite="D21511"	[TCTA]3 ta [TCTA]3 tca [TCTA]2 tccata [TCTA]10"					
variation	182 /db_x /note	1102 182 ref="dbSNP:rs12345" ="SNP C/T"	Bracketed Sequence					
ORIGIN 1 ATTCCCCA 61 TCTATCTA 121 CTATCTAT 181 TGTCTCTG //	AAG TGAATTGCC ATC TATATCTAT TCT ATCTATCTA GGA GAACA	Т ТСТАТСТАТС ТАТСТАТСТБ ТСТ С ТАТСТАТСАТ СТАТСТАТСС АТА Т СТАТСБТСТА ТСТАТССАБТ СТА	EGTETGTE TGTETGTETG ATETATET ATETATETAT ATETACET EETATTAGTE					

NIST Katherine Gettings



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 STRs for identity ENFSI Reference database, v2



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STR Sequence Nomenclature

The file 'STR Sequence Structure (Supplementary File S1)' is an updated set of forensic STR sequences that accompany the article:

Parson W, Ballard D, Budowle B, Butler JM, Gettings KB, Gill P, Gusmão L, Hares DR, Irwin JA, King JL, de Knijff P, Morling N, Prinz M, Schneider PM, Van Neste C, Willuweit S, Phillips C: Massively parallel sequencing of forensic STRs: Considerations of the DNA commission of the International Society for Forensic Genetics (ISFG) on minimal nomenclature requirements. Forensic Science International Genetics 2016, 22: 54-63 (doi: 10.1016/j.fsigen.2016.01.009; available at http://www.isfg.org/Publication;Parson2016).

Major changes to this file are currently ongoing. When the review process is finished, a new version of Supplementary File S1 containing updated information will be available for download here. To stay updated about STRidER, register here for the STRidER newsletter.

The updates since the last version are:



20

- ٠
- •

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



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



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



Updated **STR Sequence Structure file** from ISFG considerations will be available on STRidER













UNIVERSITÄTSMEDIZIN BERLIN



DNA-STR Massive Sequencing & International Information Exchange (HOME/2014/ISFP/AG/LAWX/4000007135)



Objectives

Promote the implementation of MPS technology for improved STR profiling and international data exchange

Evaluate the impact of STR sequencing on National DNA databases (EU Prüm, CODIS)

Facilitate and standardize forensic STR sequence allele nomenclature



Acknowledgements

R. Scheithauer



Monopoly 2010



Monopoly 2014



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







ΕΔΝΑΠ Μεετιγγ, Άθενς, Γρεεχε, Όχτοβερ 25 2017



ΕΜΠΟΠ Ύπδατε

Δρ. Ωάλθερ Παρσον ἀσσοχ. Προφ. Ἰνστιτυτε ὀφ Λεγαλ Μεδιχινε, Μεδιχαλ Ύνιψερσιτυ ὀφ Ἰννσβρυχκ, Αὐστρια αδϑ. Προφ. Φορενσιχ Σχιενχε Προγραμ, Πενν Στατε Ύνιψερσιτυ, ΠΑ, ΎΣΑ walther.parson@i-med.ac.at

https://empop.online/

SMI EMPOP







EDNAP initiative (ISFH 1999) Create open and freely accessible mtDNA database

EDNAP MtDNA Population Database



1999




SAM 2

Development of software for automated phylogenetic alignment of mitochondrial DNA sequences

Alignment



16181C 16182C 16183C 16189C 16213A 16217C 16242T 16261T 16292T 16301T 16519C 61A 62A 73G 183G 263G 309.1C 309.2C 309.3C 315.1C 323N 324N 523Del 524Del 16181C 16182C 16183C 16189C 16213A 16217C 16242T 16261T 16292T 16301T 16519C 61A 62A 73G 183G 263G 308.1C 309.1C 309.2C 315.1C 323N 324N 523Del 524Del



576

Phylogenetic alignment

Bandelt and Parson (2008)

Consistent treatment of length variants in the human mtDNA control region: a reappraisal Int J Legal Med 122:1-21

Rule 1. Phylogenetic rule

Rule 2. Anchor 16189 and 310

Rule 3. 3' alignment

adopted by SWGDAM (2013) and ISFG (2014)

DOI: 10.1017/v00414-006-0151-5	
ORIGINAL ARTICLE	
Consistent treatment of length va mtDNA control region: a reappr	ariants in the human aisal
IIJ. Bandek - W. Parson	
Reseived: 20 March 2006/Accepted: 13 December 2006 / Published on © Springer Verlag 2007	line 9 March 2017
Abstract In foremic science, as well as in molecular anthropology and multical genetics, human minchoodrial DNA (set2NA) variation is being recorded by aligning mENA sequences to the revised Cambridge reference response (sCRS). This take is similghtforward for the vari majority of nucleoridu positions but appears to be difficult for scene short sequence structure, namely, in regions aunique alignment relied on binary alignment to a similar sequence (the sCRS) and used additional periodry rules for resolving ambiguities. It turns out, however, that these rules have not been applied rigorously and led to inconsistent resolving ambiguities. It turns out, however, that these rules have not been applied rigorously and led to inconsistent resonable way because binary alignment to a simularit sequence (the based to predice artificial alignments that may face sequences separated by a single multice at mismisch distance larger than 1. To runnedy the situation, we propose a phylogenetic approach for multiple alignment and result- ing neurains. Keywards Mitochondrial DNA - Hapingroup - Alignment Phylogeny	Introduction The postation of mition-broading DNA (mtDNA) variation is requester (xD3; [2]), which is the corrected variation of the function of the state of the state of the state of the contential value of the correction second particle by guided has to standardize the atDNA reporting precess [4, 9, 23] in particular by providing general rules, for sequence alignment. In a statebor of cases, however, specification of the value of the polycytosine stretchen of the mtDNA particular by providing general [2, 23] attempting by binding formal rules for most gassinonicos sequence is and the stretcher of the mtDNA durations (10, 10, 10, 10, 10, 10, 10, 10, 10, 10,
HJ. Bandel: Department: of Mathematics, University of Hamburg, Bandroadraffs 33, 2014d Bandroac, Garmann	Algoment and neuronlature
e-coal: bandwijimet.uni-bamburg.de W. Parun (52) Institute of Lagal Medicine, Inschrack Madical University, Millertresses 44, 5020 Inschreck, Austria e-mail: walken generolity end as al	In general, the alignment of human mfDNA sequences does not pose serious problems except for the vicinity of polycytosias or disuctionide mars: that are both protect to considerable length heteroplasmy. Occasionally, however, one faces some nul obtacles for unloss alignment is some hort sequences involved one to multiple multipless.





> Manual sequence alignment difficult, ambiguous and error-prone

SAM 2

> We developed new software to perform

unbiased sequence searches

phylogenetic alignment according to ISFG recommendations haplogrouping of mtDNA haplotypes

- Started external testing in July 2017
- Very positive comments

discussing observations/ amending suggested changes

- Publication in preparation
- Change SAM to SAM 2 (Q1 2018)

Empop_{mtDNA database, v3/R11}



Query	Result	Details	Neighbors	Alignment	Haplogrouping
Sample ID					
Ranges	16024	4-576			
Profile	16188	9- 16193.1C	16356C 16519	IC 263G 315.1C	

Query	Result	Details	Neighbors	Alignment			
Sample ID	(none s	pecified)					
Ranges	16024-	576					
Profile	16188-	16193.1C 1635	6C 16519C 263G	315.1C			
Entire Data	base					Frequency	Clopper Pearson Cl
					2/26127	7.6549e-5	[9.2706e-6, 2.7649e-4]

Empop_{mtDNA database, v3/R11}



16519C

16519C

Query	Result	Details	Neigl	hbors	Alignm	ent H	laplogroupin	g			
Sample ID											
Ranges	16024	i-576									
Profile	16188	- 16193.10	2 16356	C 16519)C 263G 3	315.1C					
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Empop_{mtDNA database, v3/R11}



Query	Result	Details	Neighbors	Alignment	Haplogrouping
Sample ID					
Ranges	16024	i-576			
Profile	16188	- 16193.1C	16356C 16519)C 263G 315.1C	

Query	Result	Details	Neighbors	Alignment	Haplogrouping	_			
Haplog	groupii	ng							
Rank 1: MRCA: H1b 0.80-0.80									
Source		Haplogroup			Cost	Missing Mutations	Private Mutations		
PT17					0.80	none	C16188T		

SAM 2 schedule

- Expose SamCost to **external testing** (SWGDAM, ISFG WGs,...)
- Feb 23-24 2017 mtDNA workshop at MPI Jena, Germany presentation to population genetics, anthropology (W. Haak, M. Richards, A. Torroni, A. Achilli, M. vanOven)
- Apr 25 2017 EDNAP-Meeting Vilnius, Lithuania
- Apr 26-28 2017 ENFSI-Meeting Vilnius, Lithuania
- Jun 06-08 2017 Human Variome Santiago de Compostela, Spain
- Aug 28-Sep 02 2017 ISFG World Conference Seoul, South Korea
- May 2018 Haploid Marker Meeting Bydgoszcz, Poland







R. Scheithauer



CR and mitogenome



EMPOP database

Der Wissenschaftsfonds.

ISFG Commission on mtDNA EMPOP collaborators

Arne Dür (Univ. of Innsbruck) EMPOP Team Innsbruck IT Team Innsbruck vxWEB Nicole Huber





Bridging East & West ISFG 2017

27th Congress of the International Society for Forensic Genetics August 28 - September 2, 2017 Coex, Seoul, Republic of Korea



ΊΣΦΓ Υπδατε 2017

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

Άθενς, **Ό**χτ 25 2017

ISFG Board Meeting in Sept 2016 (held in Minneapolis, MN with ISHI)



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 5 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 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

Letter of Intent



Kraków, 18th April 2017

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 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

• "Forensic canine DNA profiling" Meetings

- 12 meetings since 2003
- alternating organized by the member institutes
- next meeting: September 2017 in Wiesbaden, Germany, organized by BKA

• biannual CaDNAP Proficiency Test

- first PT: 2014/15 (CaDNAP intern only)
- second PT: 2016/17 (open to all interested laboratories)
- first papers have been published in 2004
- In 2011 members of the CaDNAP group were invited as co-authors to publish the recommendations of the ISFG regarding the use of non-human (animal) DNA in forensic genetic investigations [Linacre et al. 2011].
- In 2014 the CaDNAP group published protocols for the analysis of dog specific STRs, assembled in two multiplex reactions that were validated according to the above mentioned ISFG guidelines [Berger et al. 2014].



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Forensic canine DNA profiling Meeting 2015 in Innsbruck



Forensic Science International: Genetics Journal Status Report

John M. Butler Associate Editor

ISFG General Assembly – Seoul, South Korea 31 August 2017

Journal Editors



Peter Schneider (Associate Editor) Angel Carracedo (Editor-in-Chief) John Butler (Associate Editor)

Adrian Linacre (Associate Editor) **Leonor Gusmão** (Associate Editor)

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 will become 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 \rightarrow 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

Impact Factors for Forensic Genetics Journals

6,000 🗉



2017 Population Data Requirements

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



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)	77.0		
		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

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



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 Morlingⁿ, Mechthild Prinz^o, Peter M. Schneider^p, Christophe Van Neste^q, Sascha Willuweit^r, Christopher Phillips^s



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





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)



GENETIC

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,*}



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



GENETI

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. Guttman¹, M. Krawczak^m, N. Morlingⁿ, W. Parson^{o,p}, N. Pinto^{j,k,q,r}, P.M. Schneider^s, S.T. Sherry^t, S. Willuweit^u, M. Prinz^v



 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):1325Countries 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 7th Korea, Republic of 57 Argentina Denmark Switzerland Brazil China Belgium Portugal United Arab Emirates Austria Colombia France Mexico Netherlands Poland New Zealand Norway Peru Sweden 🗏 Japan



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



Travel Bursary Award Winners 2017





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)
- Applications were received in February and March 2017 with decisions made in April 2017
- 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



Nine (9) short-term fellowships awarded in 2017





Rui PereiraMPortugal → BrazilSwitz

Miriam Ender Switzerland → Norway



Ana Freire-Aradas Spain → Poland



Lode Sibben Belgium → UK



Runa Daniel Australia \rightarrow Spain



Carlo Robino Italy → Spain



Filipa Simao Brazil → Austria



Fabio Oldoni Switzerland → USA



Dragana Zgonjanin Serbia → United Arab Emirates



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 (open now) 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



Portuguese



·

MAKING SENSE OF FORENSIC GENETICS

What can DNA tell you about a crime?

Published in 2017



ISFG Website

- Purchase of Springer "Advances in Forensic Haemogenetics"
 Vol 1-6 (Congress Proceedings 1985 1995)
- Will be made available online as a searchable PDF archive

Advar Advance Advance Advance Advan Advances in Foren: Forensic Forensic Forensi Forensi Forensic Haemogenetics 1 2 4 5 6

11th Congress (12th Congress of the Society for For (Gesellschaft fü Copenhagen, A Vienna, August 26-New Orleans, Octobe (Internationale Gesel New Orleans, Octobe (Internationale Gesel Mainz, September Venezia, 13–15

16th Congress of the International Society for Forensic Haemogenetics (Internationale Gesellschaft für forensische Hämogenetik e.V.) Santiago de Compostela, 12–16 September 1995

Edited by B. Edited by W.R. Edited by H.F.P. Edited by C. Edited by V A. Carracedo, B. Brinkmann and W. Bär



ISFG Newsletter

3 newsletters published 2016-2017

other activities.

isce	NEV			
	(ISFR)	NEWSLE		
WELCOME		(ISFG)	NEWSLETTER 07-201	.7
like to share informat ISFG World Congress	WELCOME		http://www.isfg.org	ENETICS
new board members meeting, the activities	In this edition of our IS like to share information	WELCOME	accommodation of up to B	EUR 1,000 for visits

ISFG World Congress, ti the Executive Board, th In this edition of our ISFG newsletter we would Working Parties, and oth like to share information about the upcoming 27th ISFG World Congress in Seoul, the activities and

plans of the Executive Board, the recent meetings of our Working Parties, and other news and views. accommodation of up to EUR 1,000 for visits within the same continent, and EUR 2,000 for visits from continent to continent. The successful applicants and their projects will be announced at the General Assembly in Seoul.



ISFG Scientific Award - Manfred Kayser







Announcements

28th ISFG Congress, Prague We already started the preparations for the next congress



HER SHE SOF THE INTERNATION HER SOF THE IN

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?

- Goto ISFG webpage and click link for membership
- Enter your details
- Nominate 2 reference persons (ISFG members) that support your membership (good to ask them first)
- Have 60 Euro/year ready to spend

Executive committee discusses application



Why should I become a member?

- Because it is cool
- Reduced fee for conferences
- Free access to Forensic Sciences International Genetics





ΕΔΝΑΠ Μεετιγγ, Άθενς, Γρεεχε, Όχτοβερ 25 2017



ΨΙΣΑΓΕ Ύπδατε

Δρ. Ωάλθερ Παρσον ἀσσοχ. Προφ. Ἰνστιτυτε ὀφ Λεγαλ Μεδιχινε, Μεδιχαλ Ύνιψερσιτυ ὀφ Ἰννσβρυχκ, Αὐστρια αδϑ. Προφ. Φορενσιχ Σχιενχε Προγραμ, Πενν Στατε Ύνιψερσιτυ, ΠΑ, ΎΣΑ walther.parson@i-med.ac.at The <u>VISible Attributes Through GEnomics</u> - VISAGE - Consortium will establish new scientific knowledge, develop 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).

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, AUT Peter M. Schneider Cologne, GER Ingo Bastisch Wiesbaden, GER François-Xavier Laurent Lyon, FRA *Titia Sijen* The Hague, NED Johannes Hedmann Linkoping, SWE Shazia Khan London, UK Magdalena Spólnicka Warsaw, POL



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

www.visage-h2020.eu

The VISAGE Consortium

Home

Summary Objectives

Partners

Scientific Advisory Board

Ethics and Societal Impact Advisory Board

Media

Contact

VISAGE

VISIBLE ATTRIBUTES THROUGH GENOMICS

About the VISAGE Consortium.

The VISible Attributes Through GEnomics - VISAGE - Consortium aims to overcome the general limitation of current forensic DNA analysis by broadening forensic DNA evidence towards constructing composite sketches of unknown perpetrators from as many biological traces and sources and as fast as possible within current legal frameworks and ethical guidelines. The VISAGE Project will establish new scientific knowledge, develop prototype tools for DNA analysis and statistical interpretation, validate and implement these tool in forensic practice,



ResearchGate

Show details

Active project Updates quarterly

VISAGE - Visible Attributes through Genomics

😪 Walther Parson · 🔘 Manfred Kayser · 🌍 Christopher P Phillips · Show all 8 collaborators

Goal: Unknown perpetrators of crime cannot be identified with the current forensic use of DNA. The VISAGE Project aims to overcome this major limitation by developing, validating, and implementing in the relevant forensic DNA service environment a set of prototype tools for

 Updates
 2 0 new

 Recommendations
 1 0 new

 Followers
 40 12 new

 Reads
 239 76 new



F Visage Horizon2020



Visage Horizon2020 @visageh2020

Startseite





Final Report on the European Forensic Genetics Network of Excellence

Prof. Dr. Peter M. Schneider Institute of Legal Medicine University Hospital of Cologne



EDNAP Meeting, Athens, Greece, 25th October 2017



The EC requires

- Fourth periodic report
- Final report
- Financial statement
- Review meeting

The current status

- All reports have been accepted after 3 rounds of revisions
- All financial statements have been accepted (except for some minor expenses for travel and subsistence) by the end of September
- A major loss due to the bankruptcy of our previous management partner GABO:mi was covered by the EC Guarantee Fund
- Despite all the painful negotiations, it was worth the effort!



The Project at CORDIS



• The final report can be retrieved from the CORDIS server

- http://cordis.europa.eu/result/rcn/203192_en.html

×.	*	CORDIS						
Europe Commi	an ission	Community Research and Development Information Service						
pean Commiss	sion > CORDIS > Proj	iects and Results > Final Report Summary - EU	IROFORGEN-NOE (EUROPEAN FORE	NSIC GENETICS Network	of Excellence)			
				Search	L Sign in			
	EWS & EVEN	TS PROJECTS & RESULTS	RESEARCH*EU MAG	GAZINES				
EUROFORGEN-NOE Report Summary								
PERCENTER	Project ID: <u>28</u> Funded under: Country: Germ	5487 FP7-SECURITY hany						

Final Report Summary - EUROFORGEN-NOE (EUROPEAN FORENSIC GENETICS Network of Excellence)





- A European network of forensic DNA labs
 - connecting >200 members
 - Dissemination via email and social media (Facebook, Twitter)
- A website with lots of resources such as
 - Publications & newsletters
 - Videos
 - Education and training resources
- Integration into the ISFG Working Group activities
 - Short term fellowship program
 - Summer school educational workshops
 - Support for post graduate curriculum in forensic genetics
- "Making Sense of Forensic Genetics"





- The ISFG is offering travel fellowships for scientists to support transnational exchange visits between collaborating research groups for projects related to forensic genetics.
- Each fellowship includes financial support for travel and accommodation of up to EUR 1,000 for visits within the same continent, and EUR 2,000 for visits from continent to continent.
- Applicants must be ISFG members and have to submit a written application.
- See https://www.isfg.org/Members+Area/Overview







- New call with second round of short-term fellowships for 3x 10 applications in 2017-2018
- Apply by sending to <u>fellowships@isfg.org</u>
 (previous applicants will not be considered again)
- First submission deadline October 15th 2017
- Second submission deadline April 15th 2018
- Third submission deadline October 15th 2018
- All details on ISFG website in Members' Area







- The EUROFORGEN ISFG Summer School will be organized bi-annually
 - 2 days of educational workshops in locations not visited by ISFG congresses, with topics such as at pre-ISFG congress workshops
 - ISFG will cover expenses for travel and honorarium of faculty, participants pay for local arrangements (hotel, venue, catering)

The first Summer School 2018

- In connection with the Italian Ge.F.I. meeting in Calabria
- First half of September 2018
- 2 full days with 6-9 different topics
- Details to be published soon!







Details published in FSI Genetics 28 (2017) e12-e13



- Supported by more than 8 universities across Europe
- Introduced at the University of Roma Tor Vergata
 - Coordinated and supervised by Prof. Emiliano Giardina and Prof. Giuseppe Novelli
 - Inscription at http://mastergeneticaforense.it/



"Making Sense of Forensic Genetics"







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

Social media presence







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

Slide no 10



UNIKLINIK Institut für Rechtsmedizin

Plans for the introduction of forensic DNA phenotyping in Germany

Peter M. Schneider

KÖLN



The murder case Maria L.

- A 19-year-old student was raped and killed in Freiburg in October 2016.
- In December 2016, a suspect was arrested. He was identified due due an unusually dyed black-blond hair that could be assigned to him based on CCTV video.
- The suspect was a 18-year-old refugee from Afghanistan who apparently had a previous criminal record from his time in Greece.
- In the context of this case, local police officials as well as the State Minister of Interior called for an amendment of the law to allow DNA-based prediction of hair, eye and skin colour.
- This has started a general debate about "enhanced DNA typing methods" in Germany.

The public debate

- After these political demands were made public, a group of social scientists from the University of Freiburg published an "open letter" raising concerns about the reliability and value of DNA phenotyping, as well as the risk of discriminating against ethnic minorities.
 - From these publications and from several newspaper articles it became clear that there is an apparent confusion and lack of knowledge about FDP methods and their scientific background.
- The German Stain Commission published a statement in December to address these issues, and to make clear what can be done, and where limitations exist.
- In March 2017, a public symposium was held in Berlin at the Federal Ministry of Justice to proceed with the debate

The Berlin Symposium (March 2017)



The Berlin Symposium

Speakers on March 21st 2017

- Heiko Maas (Minister of Justice)
- Peter Schneider
- Manfred Kayser
- Ingo Bastisch
- Marion Albers (legal scholar on public law)
- Barbara Praisack (forensic and medical ethics, King's College)
- Veronika Lipphardt, Matthias Wienroth (social scientists, Freiburg and Newcastle)
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.
- Minority groups could therefore become overrepresented in police investigations.

Issues raised against introducing FDP

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

- 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.
- The use of these technologies also requires a balanced framework of governance, including judicial, ethical and regulatory oversight by independent governmental bodies

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 change the code

- Several German Federal States have made proposals in the Federal Council
 - <u>February:</u> Amendment to include only pigmentation markers (eye, hair, skin colour)
 - <u>March</u>: Extension of the amendment to include also biogeographic ancestry and age prediction
- The draft proposal included the need to provide funding of NGS equipment and consumables
- The proposal was rejected to allow further discussions in the parliamentary commissions
- It is expected that the proposal will be resubmitted next year after a new federal government has been formed



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

SWGDAM meeting Woodbridge July 2017





European Network of Forensic Science Institutes





ENFSI member/partner organisations

- 66 member organisations from 36 countries
- 67 partner organisations from 27 countries
- DNA working group46 ENFSI member organisationsfrom 35 countries21 partner organisations from 13
- countries







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 Programme 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
- Case management and resources
- Presumptive tests
- Mixture interpretation software
- Rapid DNA



Vilnius 26-28 April 2017 Annual meeting ENFSI DNA Expert Working Group

Host Lithuanian Police Forensic Science Centre

101 persons attending81 participants20 company representatives10 companies25 countries represented49 speakers

USA Singapore Australia New Zealand





QA-subgroup chairs Annick Delaire (France), Tom Heylen (Belgium)

Contamination prevention guidelines: approval of final version
 Proposal for new contamination survey next year; with questions regarding how labs detect contaminations, which software, only single source stains or not, partial profiles, definition of contamination etc.

- Overarching BPM:
 - Scope to be drafted by February 2018
 - Open for discussion at Spring meeting 2018
 - Different chapters shall be written by the different subgroups according to their expertise



QA-subgroup chairs Annick Delaire (France), Tom Heylen (Belgium)

O Update document 'Minimum Criteria for the Validation of Various Aspects of theDNA Profiling Process'

o Update Quality Assurance Program for DNA Laboratories

 A survey will be held among the ENFSI laboratories to benchmark contamination rates in different laboratories and to make an inventory of how labs detect contaminations (which software, which kind of profiles (single source, mixtures up to how many persons)



QCLG Meeting

QCC (ENFSI quality control committee) Quality managers of forensic labs QCLG liaisons of working groups (June Guiness, FSR, UK)

One day one topic seminar (OOS) on ILAC G19:2014

PT/CT scheme updates



Methods analysis and interpretation sub group chairs Peter Gill (Norway) and Walther Parson (Austria)

- Training workshop in STR mixture interpretation in conjunction with CEPOL
- EMPOP: Harmonization of mtDNA alignment and notation Develop, evaluate and validate software for harmonizing mtDNA alignment and notation across forensic laboratories
- STRidER: Quality control of STR fragment size and sequence data

Develop, evaluate and validate software for quality control of STR genotypes derived from CE and MPS technologies



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

European Database:

- Convicted not arrested Only 11 European Countries (less than 25%) have authorized arrestee testing
- Serious crimes DNA tested, sometimes not all crimes DNA tested
- Very few expansion efforts in last ten years
- Hit rates below 10% except The Netherlands 57% and UK at 60%



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

- Audit questionnaire: ENFSI DNA Database Management review and Recommendations document contains 34 recommendations > are to be filled in by the persons responsible for the database in their country and returned for compilation. Results presented at Spring meeting 2018
- Subgroup news bulletin on interesting facts for Databases



Update on the implementation of the EU-Prüm Council Decisions

Only 156 connections (33%) have been realized so far

The deadline for the implementation was 26 August 2011 !

The European Commission has written letters to countries that have not yet implemented the Prüm Council Decisions



Automation and LIMS subgroup Chairs: Christina Forsberg (Sweden) and Shazia Khan (UK)

Exchange knowledge on issues related to laboratory automation, including: Automated workflows for NGS Rapid DNA devices



Automation, LIMS and kit inventory list - Resource for sharing of knowledge

- <u>Automation</u> 44 laboratories added information 202 robotic systems listed
- <u>LIMS</u> 32 laboratories 17 have LIMS
- STR and quantification kits

22 laboratories STR/qPCR kits from three vendors plus home-brew qPCR

Updated yearly and put on EPE web site





RAPID DNA INSTRUMENT QUESTIONNAIRE

- 11 replies (ENFSI labs and guest)
 Answers predominantly from labs with interest in the topic
- Send out the questionnaire on rapid DNA again to collect more answers.





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

To address questions from:

- routine casework in and beyond DNA testing
- scene and analytical strategies
- stain characterization
- activity analysis
- case reporting

Two projects still ongoing: Presumptive test Study on Case Management and Resources



Updates

- SWGDAM
- ISFG activities
- DNA SEQ EX
- STRbASE>STRidER STR Population Database
- EUROFORGEN Network of Excellence
- GEDNAP > Proficiency testing
- EDNAP > collaborative exercises
- ICMP > Exclusion database





Evaluation of the GEDNAP Proficiency Tests 52 & 53

Carsten Hohoff, Katrin Schnöink & Bernd Brinkmann

Institute of Forensic Genetics, Münster (Germany)

ENFSI DNA WORKING GROUP MEETING: 16:35-17:00, 27 APRIL 2017, VILNIUS, LITHUANIA





GEDNAP participants

> 230 participants

university labs:
state labs:
private labs:

87 44
ENFSI
77 members
66



GEDNAP 52

39 497 typing events - 33 errors



99.92 % of all analyses are correct

ENFSI update 2017





INTERNATIONAL COMMISSION ON MISSING PERSONS

ICMP – EDB

ENFSI update 2017



Current statistics

Profiles stored (total 592)

- •US collection (Florida): 202
- ICMP collection: 15
- •FSS unsourced: 375

Users registered: 39 (last registration 29 Mar 2017)

Searches

- Searches performed: 232
- Matches found: 14



Data storage

- $_{\odot}~$ EDB data are stored at ICMP's HQ in The Hague
- ICMP as International Organization can guarantee the inviolability of data processed under ICMP's control
- Secured by Host State Agreement (which is an international treaty)



Current situation

- O Up to now no profiles from manufacturers or users
- Some users had issues with data stored in BiH (solved), DE will submit profiles
- o Manufacturers currently low interest
- ISO 18385 requires process for voluntary samples
- Companies (if following) prefer decentralized solutions



Way forward

- Please request consumables manufactured according to ISO 18385
- o Ask for proof of certification for ISO 18385
- Inventory of 'open issues' from both manufacturers and laboratories
- Meeting 2018 ICMP/ENFSI/companies in The Hague at ICMP



Netherlands Forensic Institute Ministry of Justice

DNAxs Expert System



DNAxs

- In house built (Java)
- Web application (browser)
- Server based



Functionality

- View profiles
 - Overview of runs and peak heights
 - Link to pdf of EPG
- Match profiles
 - Trace vs person
 - Trace vs trace
- Derive profiles
 - LoCIM inference
 - Consensus
 - Composite
- Match matrix










DNAxs - 2017.10.17.001

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Future functionality

- Integration of EuroForMix
 - Match mixed profiles using:
 - Maximum Liklihood
 - Deconvolved profiles
- CODIS, Bonaparte export formats
- Trigger based removal of profiles in cases



Time line

Validation finished October 2017 Implementation within NFI November 2017 Present software at next DNA working group meeting April 2018



Contact ENFSI DNA Expert Working Group

Website

www.enfsi.eu

Chair Sander Kneppers NFI, the Netherlands, <u>s.kneppers@nfi.minvenj.nl</u>

Vice chair Livia Zatkalikova Ministry of Interior, Slovenia, <u>livia.zatkalikova@minv.sk</u>

Secretary Astrid Quak NFI, the Netherlands, <u>a.quak@nfi.minvenj.nl</u>

Verbal Scale discussions

Peter Gill

Motivation

- Numerous enquiries about verbal scale.
- Believed that verbal scales may help Juries to better understand evidence.
- However, there is evidence that verbal scales might be misleading
 - Weak evidence effect
 - When evidence is described as weak in favour of the prosecution, it will be interpreted by a juror as favourable to the defence – ie evidence is underated

- Agreed that the number should always be provided.
- The verbal predicator is not compulsory

Universality

- The universal scale provided in the ENFSI guideline is intended to be universal across all evidence types.
- There is discussion about whether we need different scales for DNA, but in general this seems difficult to justify as the likelihood ratio is an absolute expression of strength of evidence – independent of method used

Proposed scale

Table 1. SWGDAM verbal scale for the expression of likelihood ratios

LR for H ^e Support and 1/LR for H ^e Support	Verbal Expression
1	Uninformative
2 – 99	Limited (or Weak) Support
100 – 9,999	Moderate Support
10,000 – 999,999	Strong Support
≥1,000,000	Very Strong Support

- 1) Similar to the ENFSI scale
- 2) A scale that mirrors support for Hp and Hd

A verbal scale to support Hd

- Question of whether it is necessary (or useful) to provide a verbal scale for Hd?
- Or is it sufficient to state that anything below LR=1 is support for the defence proposition of exclusion?

The verbal scale and 'inconclusive' calls

- A suggestion to include an 'inconclusive category' in the verbal scale e.g. LR<1000 and LR>0.1.
- The range will be based upon experiments carried out with mixtures where the ground truth is known. Inconclusive is defined as a range of LRs where it has been recorded:
 - false positive LRs, i.e false inclusion
 - False negative LRs, i.e. false exclusion

Inconclusive

- Difficulties with defining an inconclusive zone
- It is dependent upon the sample size
- It is dependent upon the method used
- It is dependent upon the kind of sample analysed
- i.e. we would many different inconclusive zones for any method
- The LR is itself an expression of the uncertainty that we have why is anything else needed?

Non-contributor tests

- Advantage as that non-contributor tests can be used on a per sample basis to indicate the distribution of LRs provided by random individuals.
- Non-contributor tests are always compared to the LR actually achieved with the suspect
- The actual LR achieved should be a clear 'outlier'
- Non-contributor tests can be used to show if a model is not working properly.

Example – two suspect problem

•The crime-stain is from an epithelial swab taken from the female victim

•There are two suspects accused of sexual assault, S_1 and S_2 respectively; both deny the offence.

•This epg is classified as a low template of three or more individuals

•Consequently, the preliminary propositions are formulated as $Hp=V,S_1,S_2$ and Hd=V,U,U

•*log*₁₀*LR*=4.56

Sensitivity plot (evaluate one suspect at a time)





LRmix Studio S1 effect (1 million iterations)



Step 6: Non-contributor performance (Np) tests

•Np tests can be used to support the conclusion that evidence supporting S_1 is 'inclusionary' whereas evidence supporting S_2 is 'exclusionary'

	Three pe	erson mixture	Non-contributor performance		
Нр	Hd	Random man substituted	$\log_{10}(LR)$	percentiles	
S_1, S_2, V	<i>V,U,U</i>	S_1	4.5	(-23,-17,-9)	
S_1, S_2, V	<i>V,U,U</i>	S_2	4.5	(-3,+2.9,+7)	
S ₁ , <i>V</i> , <i>U</i>	<i>V,U,U</i>	S_{I}	6.4	(-11,-6,-1)	
S ₂ , <i>V</i> , <i>U</i>	<i>V,U,U</i>	S_2	-3.8	(-12,-6,-1)	

Database searches revisited the effect of database trawls on strength of evidence Peter Gill How is the strength of evidence affected by database searches

- Do multiple comparisons with a database diminish the strength of evidence?
- An early NRC report (1996), recommendation 5.1 stated that the match probability in the database search should be calculated as LR=1/Np where n=size of database and p=match probability.
- E.g. LR=1/(5,000,000 x 1/1bn) = 200

Np-rule

- 1/Np rule was proposed by Stockmarr (1999)
- The propositions are:
 - Hp: The donor of the crime stain is in the database
 - Hd: The donor of the crime stain is not in the database
- Note there is nothing wrong with the calculation
- But there was strong criticism all of which was based on the propositions.

What is the question?

- It was argued by Balding and others, that in fact the strength of evidence is actually increased as a result of a database search.
- Consider Mr X is found as a result of a database search size n.
- Propositions:
 - Hp: Mr X is the donor of the crime stain
 - Hd someone else is the donor of a crime stain

Likelihood ratio as a result of a database search

- The culprit is a member of a population size N and N>n.
- $LR = \frac{1}{p_a} \frac{N-1}{N-n}$
- So if we have N= 30million and n=5million
- Pa=1 billion, for example
- Then LR=(30m/25m)x 1bn
 - Which is slightly greater than 1bn
- So the conclusion was that a database search actually increases the strength of the evidence because it eliminates all but one individual

What are the conditions applied for this logic to be valid?

- From Balding (2002):
 - "1. that, to be convinced of a defendant's guilt, they must as a matter of logic be simultaneously convinced that every other individual on earthincluding each of the defendant's close relatives-is innocent, and
 - 2. that the scientific evidence must be combined with the nonscientific evidence to come to an overall view on guilt or innocence. "

Other conditions of the formula

- That only one suspect is found
- That all but one member of the database is eliminated
- For mixtures this is not true
 - You can have thousands of matches with LR>1
 - You may miss the donor because his DNA contribution is too low level,

From the jurors perspective

 Is it true that a juror must be convinced to the point that every other individual on earth, including relatives is innocent?

Patently this is not true

- The benchmark is 'reasonable doubt'
- Research on juror behaviour shows that they only need to be c. 95% sure of guilt
- DNA evidence tends to be overweighted in the investigator and juror mindset, such that other evidence (that points away from guilt) is underweighted). This is *confirmation bias*.

Classic example described in "sense about science" paper

- The problems are amply demonstrated with the case of Raymond Easton who spent several months in custody on the basis of a DNA 'match' resulting from a database search. The evidence was given as *sic*[37-million-to-one].
- This individual was charged with burglary, even though he lived 175 miles away from the crime scene, he was in the advanced stages of Parkinsons disease and was unable to walk more than 10m unaided
- OK it never went to trial because the defence managed to persuade the police to carry out further tests – but the point is that the *investigators* were subject to confirmation bias
- Other examples, that did go to trial, resulting in conviction include Farah Jamah (Australia)

DNA as sole evidence

• The second Balding condition is:

"that the scientific evidence must be combined with the nonscientific evidence to come to an overall view on guilt or innocence."

- What if there is none?
- In R v. Tsekiri[3], the defendant was found guilty of burglary. The only evidence was presence of DNA (mixture) on a door handle, and the court acknowledged "<u>the finding</u> <u>of DNA attributable to a defendant at the scene of a crime</u> <u>was the sole evidence against a defendant</u>". The jury was provided with a *sic* [match probability 1:1 billion].
- However the conviction was upheld
- In my view the court's reasoning was superficial and misses the point.

How should DNA as sole evidence be reported?

- Clear responsibility of investigators and scientists to make clear the implications of DNA as sole evidence by explaining the effect of priors on the posterior odds
- From Meester R, Sjerps M. 2003



Example of R v. Tsekiri

- There is no *a priori* reason (evidence) to suggest Mr Tsekeri is guilty
- Let's suppose any male in London is equally likely to have committed the offence and their priors are the same. N=4 million
- A number of these may be eliminated by the national DNA database. We could also eliminate other individuals – too old, too young, infirm etc. Suppose n= 700,000
- So the posterior odds = 1/(4m-700,000) x (1/1bn)
- =303

- The posterior odds of 303 are much lower than the LR of 1 bn.
- With the example of Raymond Easton.
 - LR=37m
 - N-n=30m (based on male population of UK)
 - Posterior odds are neutral

Conclusion

- In a case where the suspect is identified before the DNA test, there is already substantial non-DNA evidence to raise the priors.
 - The jury are in a good position to assess the entirety of the evidence
- When the DNA evidence is sole-plank, then the jury is not in a position to assess the evidence.
 - The fact of the database search is not disclosed so the jury never get to think about population size.
 - The priors (i.e the 'suspect' population size is never discussed in court)
 - i.e. the evidence can be misrepresented which invites confirmation bias

Roma 17-20 April 2018

CODIS Meeting – 17 April EDNAP Meeting – 17 April

ENFSI DNA WG 18-20 April

Location

• Scuola Superiore di Polizia

Address: Via Pier della Francesca, 3




Hotels*

 40 rooms Hotel Villa Glori **** (500mt from the Meeting Venue) €130 double for single use/€150 double including continental breakfast and free Wi-Fi



*Prices are intended excluding Local Taxes

Hotels*

 20-40 rooms Best Western Hotel Astrid**** (1 Km from the Meeting Venue) €120 double for single use including continental breakfast and free Wi-Fi



*Prices are intended excluding Local Taxes

Hotels*

 40 rooms Hotel Donna Laura Palace **** (1,5Km from the Meeting Venue) €130 double for single use/€150 double including continental breakfast and free Wi-Fi



*Prices are intended excluding Local Taxes

Web site will be ready in January 2018

- Registration
- Accomodation
- Tourist information
- Information about transfer from/to Airports (Fiumicino and Ciampino) and Railway Station

Registration fees

ENFSI regular participant: 320-350 €

EDNAP and CODIS partecipant: 125-140 €

Paola Di Simone Alessandra La Rosa