

# EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

Riga, Latvia

23 October 2019

Host: Izanda Puncule  
Chairman: Niels Morling

A list of participants is attached.

## Welcome

Izanda Puncule welcomed members to Riga.

## Update on exercises

*Second exercise on methylated DNA and age*

David Ballard

David Ballard informed members that a manuscript will be circulated before the end of October 2019.

*Exercise no. 2 on mRNA and cSNP typing using Illumina MiSeq*

Cordula Haas

Cordula Haas informed members that the submitted manuscript got benevolent comments from the reviewers. An updated manuscript that takes the comments of the reviewers into consideration will be circulated as soon as possible (presentation attached).

*mtDNA quantification exercise*

Arnoud Kal

Arnoud Kal informed members that a PowerPoint with the results will be circulated within six months (presentation attached).

## Updates from other groups

*ENFSI*

Sander Kneppers

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

*EMPOP (DNA.BASES)*

Walther Parson

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

*STRidER (DNA.BASES)*

Walther Parson

Walther Parson gave a short update on STRidER (presentation attached).

*The VISAGE project*

Walther Parson

Walther Parson gave an update on the VISAGE project (presentation attached).

*ISFG*

Walther Parson

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

## Other activities

### DNAXS - DNASStatX

Sander Kneppers

Sander Kneppers presented a new software suite for data management and probabilistic interpretation of DNA profiles. There will be workshops on 27-28 February 2020 in The Hague, The Netherlands and 21-22 April 2020 in Ljubljana, Slovenia (presentation attached).

## Future activities

### *Collaborative exercise on detection of mtDNA heteroplasmy by MPS* Walther Parson

Walther Parson gave an update on the mitochondrial DNA collaborative exercise on Length (LHP) and Point Heteroplasmy (PHP). Two combined studies are planned: (1) A technical study that includes high-quality reference samples exhibiting LHP/PHP, and (2) a study of somatic mutations in hair shafts. The samples have been prepared in Innsbruck and will be distributed at the end of October 2019. Laboratories should submit the results to Innsbruck before the end of January 2020.

### *New collaborative exercise on mRNA and cSNP typing using TFS S5* Cordula Haas

Cordula Haas and colleagues will present an updated proposal of a collaborative exercise on identification of donors of body fluids by means of mRNA and cSNPs with an IonTorrent S5 assay in 2020.

### *New collaborative exercises on transfer of DNA*

Bas Kokshoorn

Bas Kokshoorn presented the framework of the project that was circulated to members (attached). The series of collaborative exercises will be organised by Bas Kokshoorn, The Netherlands, and Roland van Oorschot, Australia. The EDNAP members welcomed the proposal, and the great majority of the EDNAP members indicated interest in active participation. Laboratories affiliated with EDNAP and the DNA Working Group of ENFSI will be invited to participate before the end of November 2019 (presentation attached).

### *Biogeographical Ancestry - Current status and way forward*

C. Phillips/L. Roewer

Lutz Roewer reported on the results of two exercises on estimation of ancestry and discussed the current status (presentation attached).

Chris Phillips discussed the current status of estimation of biographical ancestry (presentation attached).

## Next meetings

Niels Morling

The next EDNAP meeting will take place on 5 May 2020 in Lisbon in association with the meetings of CODIS (5 May 2020) and the ENFSI DNA Working Group (6-8 May 2020).

## Any other business

Niels Morling

There was no other business.

## Closing of the meeting

Niels Morling

The meeting closed with sincere thanks to Izanda Puncule and all colleagues, who helped organising the meeting.

## The minutes and attachments are found at the EDNAP website:

<http://www.isfg.org/EDNAP/Meetings>, including:

- Agenda
- List of participants
- Presentations

- Cordula Haas: Update on the second collaborative exercise on mRNA NGS
- Arnoud Kal: Update on the mtDNA quantification exercise
- Sander Kneppers: Report from the ENFSI DNA Working Group
- Sander Kneppers: DNAXs – DNAStatistix - Invitation to collaborate
- Walther Parson: EMPOP report (DNA.BASES)
- Walther Parson: STRidER report (DNA.BASES)
- Walther Parson: The VISAGE project
- Walther Parson: ISFG report
- Kokshoorn/van Oorschot: Series of exercises relating to DNA transfer
- Bas Kokshoorn: Proposal for exercises on DNA transfer
- Lutz Roewer: Biogeographical Ancestry
- Chris Phillips: Biogeographical Ancestry

# AGENDA FOR THE EDNAP MEETING

**RIGA – 23 OCTOBER 2019**

Expected duration: 09.00 - 17.00

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

Host: Izanda Puncule

Chairman: Niels Morling

Welcome	Izunda Puncule
Update on activities	
Methylated DNA and age exercise	David Ballard
Exercise no. 2 on mRNA typing with MPS	Cordula Haas
mtDNA quantification exercise?	Arnoud Kal
Updates from other groups	
ENFSI	Sander Kneppers
EMPOP (DNA.BASES)	Walther Parson
STRidER (DNA.BASES)	Walther Parson
The VISAGE project	Walther Parson
ISFG	Walther Parson
Future activities	
Collaborative exercise on detection of mtDNA heteroplasmy by MPS	Walther Parson
Proposition for a new series of exercises relating to DNA transfer	Baas Kokshoorn
Proposition for an exercise on identification of donors of body fluids by means of cSNPs	Cordula Haas
Biogeographical Ancestry: Current status and way forward	Chris Phillips/Lutz Roewer
Next EDNAP meeting 5 May 2020 in Lisbon (ENFSI 6-8 May 2020)	Niels Morling
Any other business	Niels Morling



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# EDNAP mRNA NGS exercises

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

Cordula Haas / Guro Dørum  
Erin Hanson / Jack Ballantyne

23. October 2019, Riga



## 1. Collaborative exercise mRNA NGS part 2

- 2 separate MiSeq assays:
  - 1) targeted mRNA NGS approach for the identification of blood, saliva, semen, vaginal secretion, menstrual blood, skin
  - 2) cSNPs assay to associate specific mRNA transcripts to an individual
- 9 Laboratories analyzed 16 samples provided by UZH

### cSNP discussion:

- Analysis of RNA/cSNP in stains is challenging
- Combining evidence – DNA, RNA and cSNPs
- Need more suitable markers → Simultaneous identification of individual and body fluid



# 1. Collaborative exercise mRNA NGS part 2

## 1) cSNP proof of concept paper

- 12 single source samples, 51 mixtures
- STRs, mRNA, cSNPs
- Statistics

→ rejected by FSI Genetics

→ submitted to IJLM

## 2) Collaborative exercise mRNA NGS part 2

Body fluid identification and assignment to donors using a targeted mRNA  
massively parallel sequencing approach – results of a second  
EUROFORGEN / EDNAP collaborative exercise

S. Ingold<sup>a</sup>, G. Dørum<sup>a</sup>, E. Hanson<sup>b</sup>, D. Ballard<sup>d</sup>, A. Berti<sup>e</sup>, K.B. Gettings<sup>f</sup>, F.  
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J. Ballantyne<sup>h,k</sup>, C. Haas<sup>a\*</sup>

→ submitted to FSI Genetics,  
benevolent reply from reviewers





## 2. Thermofisher cSNP assay

- all primer pairs are designed to be mRNA-specific
- some marker overlap between the MiSeq cSNP assay and TF assay
- cSNP amplicons are useful for body fluid identification
- blood, semen, saliva, (vaginal, menstrual and skin)

21 cSNPs in body fluid specific mRNA transcripts:

- 7 blood (3 genes)
- 8 semen (4 genes)
- 6 saliva (4 genes)

In addition mRNA markers for vaginal secretion (2), menstrual blood (2), skin (3)

→ identification of all forensically relevant body fluids and skin,  
as well as cSNP sequencing for blood, semen and saliva samples



## 2. Thermofisher cSNP assay

Forensic Science International: Genetics Supplement Series XXX (XXXX) XXX–XXX



Contents lists available at ScienceDirect

Forensic Science International: Genetics Supplement Series

journal homepage: [www.elsevier.com/locate/bsigss](http://www.elsevier.com/locate/bsigss)



Assigning forensic body fluids to DNA donors in mixed samples by targeted RNA/DNA deep sequencing of coding region SNPs using ion torrent technology

Erin Hanson<sup>a</sup>, Sabrina Ingold<sup>b</sup>, Guro Dorum<sup>b</sup>, Cordula Haas<sup>b</sup>, Rob Lagace<sup>c</sup>, Jack Ballantyne<sup>a,d,\*</sup>

- Ongoing work: include cSNP marker for vaginal secretion, menstrual blood and skin
- Considering all the collaborative exercises going on at the moment, we suggest to postpone the TF cSNP assay exercise to 2020/2021, including additional cSNP markers



### 3. FoRNAP - Forensic RNA Profiling Grüppli

- Exchange on RNA profiling applied to casework
- Methods, Validation, Interpretation, Cases, Recommendations, etc.
- Online Platform to exchange / collect information
- 1. Meeting: 22./23. March 2018, Zurich
- 2. Meeting: 22. Feb. 2019, Jena
- 3. Meeting: 10. Sept. 2019, Prague
- 12 laboratories: Kiel, Ulm, Munich, Ljubljana, Zurich, Cologne, NFI, BKA, LKA Rheinland-Pfalz, ESR, OUS, UCF
- 2019: Collaborative exercise: 16 stains, use own RNA only or RNA/DNA co-analysis methods, CE/MPS
- 2019/2020: Questionnaire on own experience / success of RNA profiling in casework



Netherlands Forensic Institute  
*Ministry of Justice*

# EDNAP Exercise mtDNA quantification

Kris van der Gaag  
Natalie Weiler  
Titia Sijen  
Arnoud Kal



# EDNAP exercise mtDNA quantification

- Home made assay (cheap!)
- Quantification of autosomal, Y and mtDNA
- Long and short mt probes

DNA	Probe	Bp	Dye
Total DNA	Alu Ya5	127 bp	VIC
Y DNA	DYZ5	137 bp	FAM
mtDNA	16533-180	217 bp	JUN
mtDNA	2502-2571	70 bp	ABY



## 21 Labs

- 16 x Europe
- 1 x Asia
- 4 x USA

NFI provides:

- Primers and probes
- Challenging samples
- Protocols

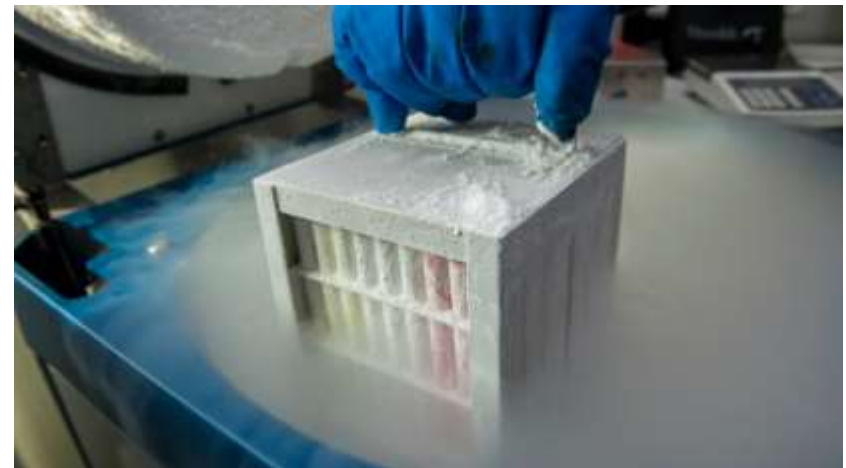
Labs provide:

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



# Challenging Samples

- Control DNA
- Sperm
- Unbalanced mixture male:female
- Fragmented DNA
- Oligo short mt amplicon
- Humic acid inhibited sample





## Analysis of the results

- Analysis started but delayed
- Variable results: effect of transit time?
- Unexplained results – outliers
- Data from 2 labs excluded
  
- Some examples in the next slides





## Sample #6

Sample = 10 ng 2800M control DNA (male)

Expected results:

Quant value >0 for ALU, Y, mt short and mt long





## Sample #7

Sample = 50 pg control DNA 9947A (female)

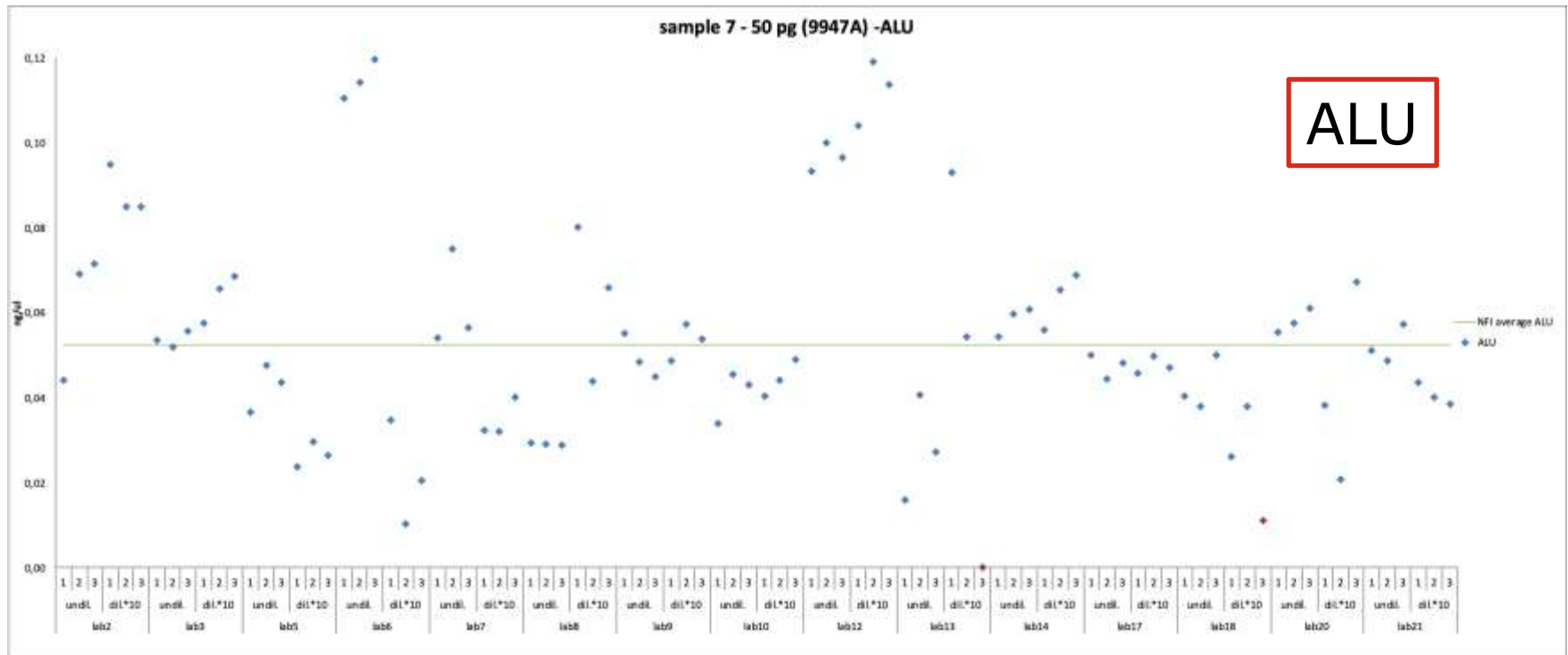
Expected results

Quant value  $>0$  for ALU, mt short and mt long

Quant value = 0 for Y



## Sample #7 Control DNA 9947A – female



Similar results for mt short and mt long





## Sample #5

Sample = oligo for the short mtDNA amplicon

Expected results:

Quant value >0 for mt short

Quant value = 0 for ALU, Y and mt long









## Sample #3

Sample = male DNA + inhibitor humic acid

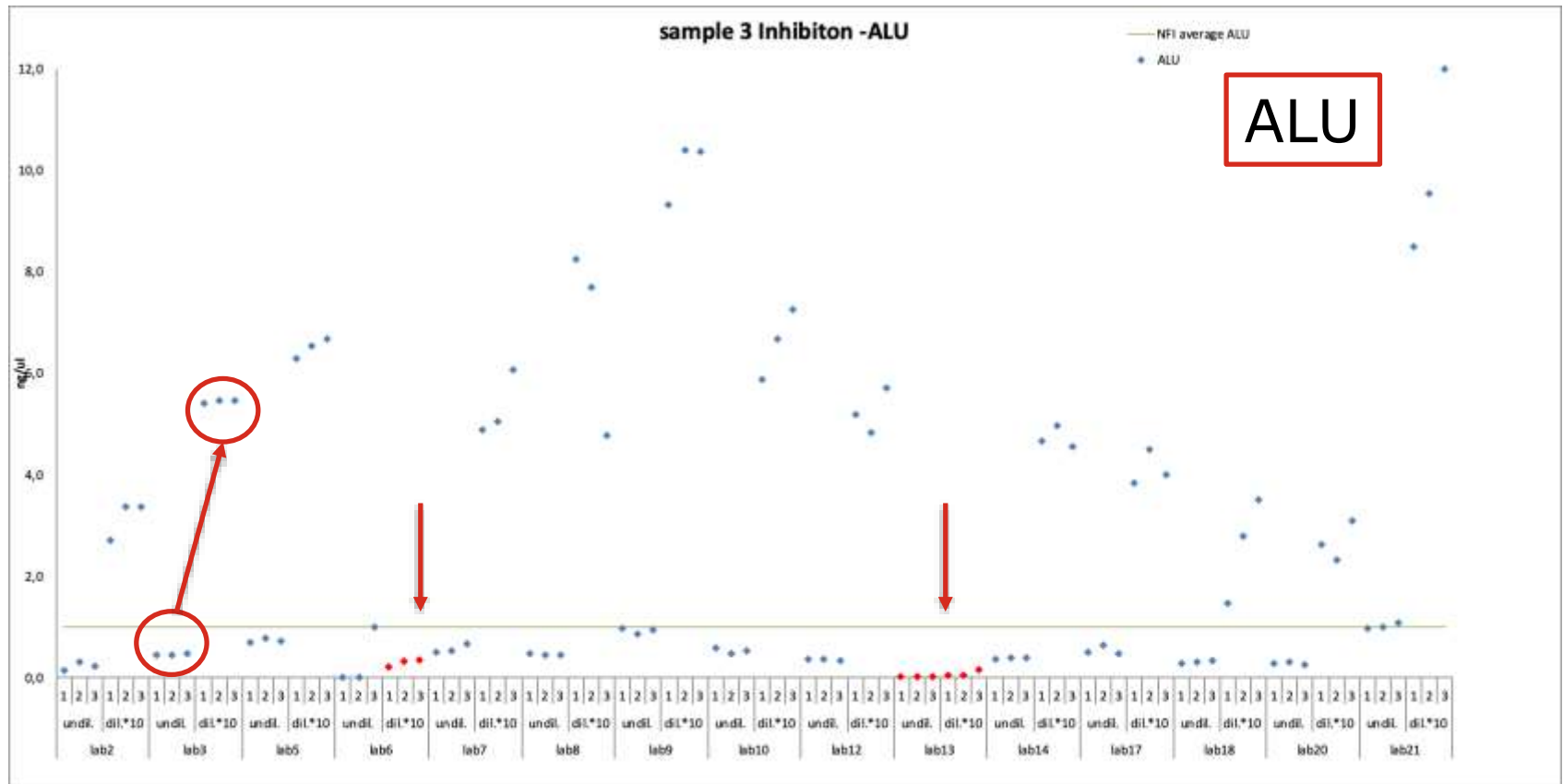
Expected results:

Quant value  $>0$  for ALU, Y, mt short and mt long

Quant value higher for diluted sample vs undiluted sample



## Sample #3 Male DNA + humic acid





## Next steps

- Further analysis of the data
- Preparation of a Powerpoint presentation of results for labs to review
- Decide on publication, journal
- Update at the next EDNAP meeting



# Update ENFSI DNA Expert Working Group activities

Alexander Kneppers  
Chair ENFSI DNA Expert  
Working Group

NFI Division Biological Traces

ENFSI update EDNAP Riga October 2019



# 71 MEMBERS IN 38 COUNTRIES



## New member institutes

- State Criminal Office of Rhineland-Palatinate (LKA Mainz), Forensic Science Institute, Permanent Representative Rainer Wenzel
- Malta Police Forensic Science Service Laboratory (MPFSL), Permanent Representative Charlot Casha (already a member since 2018)





# SYNERGY IN NETWORKING



> **1000**

**Forensic  
Experts**



**17 WORKING GROUPS**



ENFSI | EUROPEAN NETWORK OF FORENSIC SCIENCE INSTITUTES

enfsi.eu

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## Welcome to ENFSI!

The European Network of Forensic Science Institutes (ENFSI) was founded in 1995 with the purpose of improving the mutual exchange of information in the field of forensic science. This, as well as improving the quality of forensic science delivery in Europe have become the main issues of the network. Besides the general work in the fields of quality and competence management, research and development and education and training, different forensic expertizes are dealt with by 17 different **Expert Working Groups**. ENFSI therefore has been recognized as the monopoly organization in the field of forensic science by the European Commission.

Members [RESET MAP](#)

Search in members



**EUROPOL** PLATFORM FOR EXPERTS

**EUROPEAN NETWORK OF FORENSIC SCIENCE INSTITUTES**

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**Welcome!**

The ENFSI - **European Network of Forensic Science Institutes** is an interactive platform intended to bring together the forensic experts in Europe, allowing them to get involved with the content displayed. All users are advised to subscribe to every **Blog** and **Forum** in order to be timely notified on new posts/entries. It is possible to subscribe to the entire Forum, to a category or to a specific thread.

To get familiar with EPE please have a look [here](#).

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For any help please contact the site administrators Elisabeth Pickersgill or Isabelle Jopp from the ENFSI Secretariat by sending an e-mail to [secretariat@enfsi.eu](mailto:secretariat@enfsi.eu)

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## Strategic plan 2018 - 2019

1. Contribute to the establishment of a European Forensic Science Area 2020 through the implementation of the Action Plan.
  1. Facilitate the establishment and sharing of BPMs
  2. Facilitate the establishment and sharing of Forensic Databases.
  3. Facilitate the establishment of new Proficiency Tests and Collaborative Exercises
  4. Guide the coordinators of several actions of the EFSA2020 Action Plan



## Strategic plan 2020 - 2023

Three workshops during annual meeting in Rome with directors of institutes

*In which direction do you want the Board to emphasize the next Strategic Plan?*

Para

	Percentage	Number
Developing ENFSI by strengthening the voluntary network	54,55%	30
Developing ENFSI by increasing the permanent man-power	36,36%	20
Abstention	9,09%	5
<b>Sum</b>	<b>100%</b>	<b>55</b>

voluntary

basic world?



# Monopoly 2016 STEFA

Work Package 1	Benchmarking forensic laboratories for strategic planning purposes
Work Package 2	A fitted work tool for analytical data interpretation in forensic chemistry by multivariate analysis (chemometrics)
Work Package 3	Collaborative exercise covering the forensic disciplines of DND, document examination, fingerprint examination and handwriting examination
Work Package 4	Development of specialist fingermark visualisation training courses (FV Training)
Work Package 5	Development of a training and education concept for forensic hair and fibre experts
Work Package 6	IT forensic tools test and validation database (Valid)
Work Package 7	Empowering forensic genetic DNA databases for the interpretation of next generation sequencing profiles (DNA.bases)
Work Package 8	Best Practice Manual – Forensic examination of digitally captured signatures and handwritten entries
Work Package 9	Best Practice Manual – Forensic comparison of soils
Work Package 10	Best Practice Manual – Fingermark visualisation at the scene of crime
Work Package 11	Management and Coordination of the Action



# **STEFA Project Work Package 3**



**Collaborative exercise covering the forensic disciplines of DNA, document examination, fingerprint examination and handwriting examination**



# Introduction



Historically Proficiency Tests (PT) and Collaborative Exercises (CE) have

- Been carried out within a single discipline
- Tended to only cover the examination and interpretation aspects of the individual forensic processes.

However the “real” world is normally more complex than single examination types, and in many instances forensic material must be examined for a number of different evidence types.



# Team members



Two representatives from each of

- European Network of Forensic Handwriting Experts (ENFHEX),
- European Document Experts Working Group (EDEWG),
- European Fingerprint Working Group (EFP-WG) and
- ENFSI DNA working group (DNA-WG)



# Proposal



Within the four forensic discipline involved the project will:

- determine current availability and process for development of CEs
- determine the practicality of developing a multi-discipline CE
- prepare guidelines for the development of a multi-discipline CE
- develop, run and evaluate a multi-discipline CE covering the four areas of forensic science and the laboratory management processes

**ISFP-2016-AG-IBA-ENFSI**



# Time lines



34 labs have enlisted to participate for all 4 disciplines

Exhibits and questionnaires sent out in May

Results back by end of August

Results obtained from 27 laboratories

Data analyses coming months

Project extended to 31<sup>st</sup> May 2020 (6 of 10 STEFA projects extended)





## Horizon 2018 funding opportunities

- “Accreditation of Forensic Laboratories in Europe” (AFORE)
  - kick off meeting AFORE planned in Oslo on the 16th and 17th January 2020

Work Package 1	Management and Coordination of the Action
Work Package 2	Accreditation Model for Crime Scene Investigation
Work Package 3	Training Forensic Personnel in Accreditation Matters
Work Package 4	Training of Technical Experts
Work Package 5	Production of New and Updated BPMs

- BPM on Digital Image Authentication
- BPM on Forensic Examination on Fibres
- BPM on Forensic Examination of Gunshot Residues
- BPM on Forensic Handwriting Examination
- BPM on Forensic Voice Comparison
- **BPM on Human DNA Analysis (Application for funding (40K EUR))**
- BPM on Glass or BPM on Paint



## Future grant possibilities

- ISF-P funding program 2021 – 2025 Direct Grants options for ENFSI
- “Horizon Europe” which will be operational 2021-2027

Board will not write recommendation letters of grant applications

> Board doesn't see itself in the position to judge over the value of different projects

> ENFSI may provide a platform of communication by forwarding project information to relevant parties within ENFSI.



# GDPR

DDI (Deutsches Datenschutz Institut) 24 months contract

professional implementation of GDPR requirements comprising the aim to give guidance to all ENFSI Expert Working Groups on how to act in line with GDPR legal requirements



# Training

## CEPOL:

Brought forward by ENFSI member institutes, the following two training courses on forensics are planned for 2020:

Forensic investigation in CBRN contaminated environment

- Chip-off and ISP to recover data from damaged or protected devices

## EJTN

- The European Judicial Training Network
- draft of Memorandum of Understanding (MoU) in September

## DNA Expert Working group

- Ideas to facilitate training for working group member labs
- Complex mixture interpretation



## Connection to European bodies

Ideas how and which forensic issues to bring to the LEWP and the European Council.

> Acknowledgement by Law Enforcement Working Party (LEWP) for ENFSI as being an important advisory body in the field of forensic science and to be directly addressed and involved in relevant discussions

### White papers

The Board's current communication with our stakeholders like the EU commission, CEPOL, EJTN, LEWP reveals the need to have these trends – where is forensic science going to and what has to be done to fill the gaps to meet the needs of our customers



# OSAC

Overseas Security Advisory Council > US department of State

ENFSI BPMs and Guidelines acknowledged by OSAC

proper, balanced and agreed content of these documents for the target groups (forensic community), a transparent and documented, public reviewing process is needed > practicable procedure for public review of ENFSI documents

OSAC requirement that only documents which went through an SDO assessment (standardizing body like ASTM or ISO) will be listed in the OSAC registry



## Forensic databases

- ENFSI has created and already runs a series of databases (libraries, reference databases).
- MP2014 Direct Grant project (TDPEDFS) planned that ENFSI databases find a common external platform which enables controlled access, maintenance etc.
- All past negotiations with potential external providers (like Europol, JRC, LISA, etc.) failed.
- Inventory of existing databases (based on reporting cycle working groups)
- identify the number and character of databases which need an external sustaining provider and maintenance.



## Meetings

Joint Meeting (board/EWG)

Krakow, Poland on 27th to 29th November 2019

EDNAP meeting

Lisbon 5 May 2020

DNA working group annual meeting

Lisbon 6-8 May 2020

EAFS meeting

30 August to 3 September 2021 Stockholm





## ENFSI DNA Working group Steering Committee

Chair	Sander Kneppers NFI, the Netherlands
Vice chair	Livia Zatkalikova, Ministry of Interior, Slovakia
Secretary	Astrid Quak, NFI, the Netherlands
Treasurer	Ingo Bastisch, BKA, Germany
QCLG	June Guinness, FSR Home Office UK
R&D	Shazia Khan, MP UK
E&T	Paula di Simone, Italian National Police
Webmaster	Fabrice Noel, NICC Belgium
EDNAP	Niels Morling, Univ. Copenhagen, Denmark Peter Schneider, ILM, Univ. Cologne, Germany

5 subgroups with subgroup chairs



## DNA working group subgroups

Group A: Quality Assurance

Tom Heylen/Annick Delaire

Group B: DNA Analysis Methods & Interpretation

Antonio Alonso/Walther Parson

Group C: DNA Database and Legislation

Izanda Puncule/Emilia Lindberg

Group D: Automation & Expert Systems

Christina Forsberg/Shazia Khan

Group E: Forensic Biology and casework

Ricky Ansell/Arnoud Kal



## (Associate) Members DNA working group

- Currently 75 labs are participating in the DNA working group
  - 46 member laboratories
  - 29 associate member laboratories

# 4 task forces



Task force	Project title	Project description	Task Force member	Task Force leader
1	BPM DNA pattern recognition and comparison	Revising of the existing BPM	Ricky Ansell Peter Gill Walther Parson	Ela Zaimi
2	BPM Human DNA Analysis	Writing of BPM to cover the general accepted procedures and workflows for the processing of human DNA; from the collection of evidence to the reporting of findings	Ricky Ansell June Guinness (editorial) Ate Kloosterman Astrid Quak Tacha Hicks Paula di Simone Ela Zaimi Tom Heylen Aikaterini Kondili	Stavroulla Xenophontos
3A	Guideline: Training of DNA staff	A: Update the Guideline for the training of staff working in the forensic DNA laboratory	Heli Autere Paola Di Simone June Guinness	Tom Heylen
3B	Guideline: ENFSI Quality Assurance Program for DNA Laboratories	B: Update the Guideline regarding the ENFSI Quality Assurance Program for DNA Laboratories	June Guinness Elisabetta Mei Maria Vouropoulou Task Group 3a or 3B up to task force leader: Kua Guo Wei Coro Fernández Oliva	Tom Heylen
4	Guideline: 'Minimum Criteria for the Validation of Various Aspects of the DNA Profiling Process'	Update the Guideline regarding the minimum criteria for the validation of various aspects of the DNA profiling process	Christina Forsberg Ron Gafny Aikaterini Kondili Wong Hang Yee	Annick Delaire



## Contact ENFSI DNA Working group

Chair

Sander Kneppers

NFI, the Netherlands [s.kneppers@nfi.nl](mailto:s.kneppers@nfi.nl)

Vice chair

Livia Zatkalikova

Ministry of Interior, Slovakia, [livia.zatkalikova@minv.sk](mailto:livia.zatkalikova@minv.sk)

Secretary

Astrid Quak

NFI, the Netherlands, [a.quak@nfi.nl](mailto:a.quak@nfi.nl)





## EMPOP Update

Dr. Walther Parson

a. Prof. Institute of Legal Medicine, Medical University of Innsbruck, Austria  
adj. Prof. Forensic Science Program, Penn State University, PA, USA

[walther.parson@i-med.ac.at](mailto:walther.parson@i-med.ac.at)

**EMPOP v4 is fully  
phylogenetic**

rCRS-coded: C16188T T16189C A263G -315+C



ATGGCTCGGATCTCGATACACATCCGGGCTTCGATT

QUERY

ALIGNMENT

HAPLOGROUPING



before 2010

before 2018

before 2018

16188T 16189C

16188- 16193+C

28 matches

0 matches

performed manually  
Phylotree

performed manually /  
software (**alignment**)

EMPOP v3 (2010)

EMPOP v4 (2018)

EMPOP v4 (2018)

**SAM**

**SAM2**

**EMMA/SAM2**

16188T 16189C

16188- 16193+C

28 matches

28 matches

EMPOP V3 R11; N = 34,617



Alignment-Free Query

Röck et al 2011 **FSIG**  
Huber et al 2018 **FSIG**

Phylogenetic Alignment

Parson et al 2014 **FSIG**  
Dür et al in prep

Haplogrouping

Röck et al 2013 **FSIG**  
Dür et al in prep



# EMPOP 4 workflow

## Query

Query Result Details Neighbors Alignment Haplogrouping

Sample ID

Ranges

Profile

## Result of database search

Query Result Details Neighbors Alignment Haplogrouping

Sample ID (none specified)

Ranges 16024-576

Profile 16183- 16217C 16519C 263G 315+C 499A

Entire Database	Frequency	Clopper Pearson CI
2/33447	5.9796e-5	[7.2416e-6, 2.1599e-4]

## Phylogenetic alignment

Query Result Details Neighbors Alignment Haplogrouping

Phylogenetic alignment

Input Profile	263G	315.1C	499A	16183-				16217C	16519C
Phylogenetic alignment	263G	315.1C	499A		16183C	16186T	16189C	16193-	16217C 16519C

## Haplogrouping result

Query Result Details Neighbors Alignment Haplogrouping

Haplogrouping

Rank 1: MRCA: **B4b** 1.88

Source	Haplogroup	Range	Diagnostic Mutations	Cost
PT17	B2 B2b B2b3 B2c B2c1 B2c1a B2c1b B2c1c B2e B2f B2i B2n B2p B2q B2r B4b	16024-576	16183M 16189C 16217C 16519Y 73G 263G 315.1C 499A	1.88
PT17	B2b4	16024-576	16183M 16189C 16217C 16239Y 16353Y 16519Y 73G 159Y 195Y 263G 315.1C 499A	1.88

Rank 2: MRCA: **B2b+152** 2.26

Source	Haplogroup	Range	Diagnostic Mutations	Cost
PT17	B2b+152	16024-576	16183M 16189C 16217C 16519Y 73G 152C 263G 315.1C 499A	2.26

EMPOP meeting at SWGDAM, Woodbridge, VG, USA, Jul 07 2018

EMPOP workshop at ISFG Summer School, Catanzaro, ITA, Sep 03 2018

EMPOP workshop at GHEP, Araraquara, BRA, Sep 13 2018

EMPOP workshop at ISFG ESWG Meeting, St. Petersburg, RUS, Sep 13 2018

NGS workshop at ISHI, Phoenix, AZ, USA, Sep 24 2018

Targeting Mitochondria, Berlin, GER, Oct 24 2018

EMPOP Meeting at SAGF, Buenos Aires, ARG, Nov 21 2018

EMPOP workshop at GEDNAP, Jena, GER, Feb 21 2019

# mtDNA/EMPOP publications

May 2018 - April 2019

Simao et al (2018) *Forensic Sci Int Genet* **34**: 97-104

Parson et al (2018) *Forensic Sci Int Genet* **36**: 148-151

Huber et al (2018) *Forensic Sci Int Genet* **37**: 204-214

Simao et al (2019) *Forensic Sci Int Genet* **39**: 66-72

Lutz-Bonengel and Parson (2019) *Proc Natl Acad Sci USA* doi: 10.1073/pnas.1820533116

Scientific Working Group on  
DNA Analysis Methods

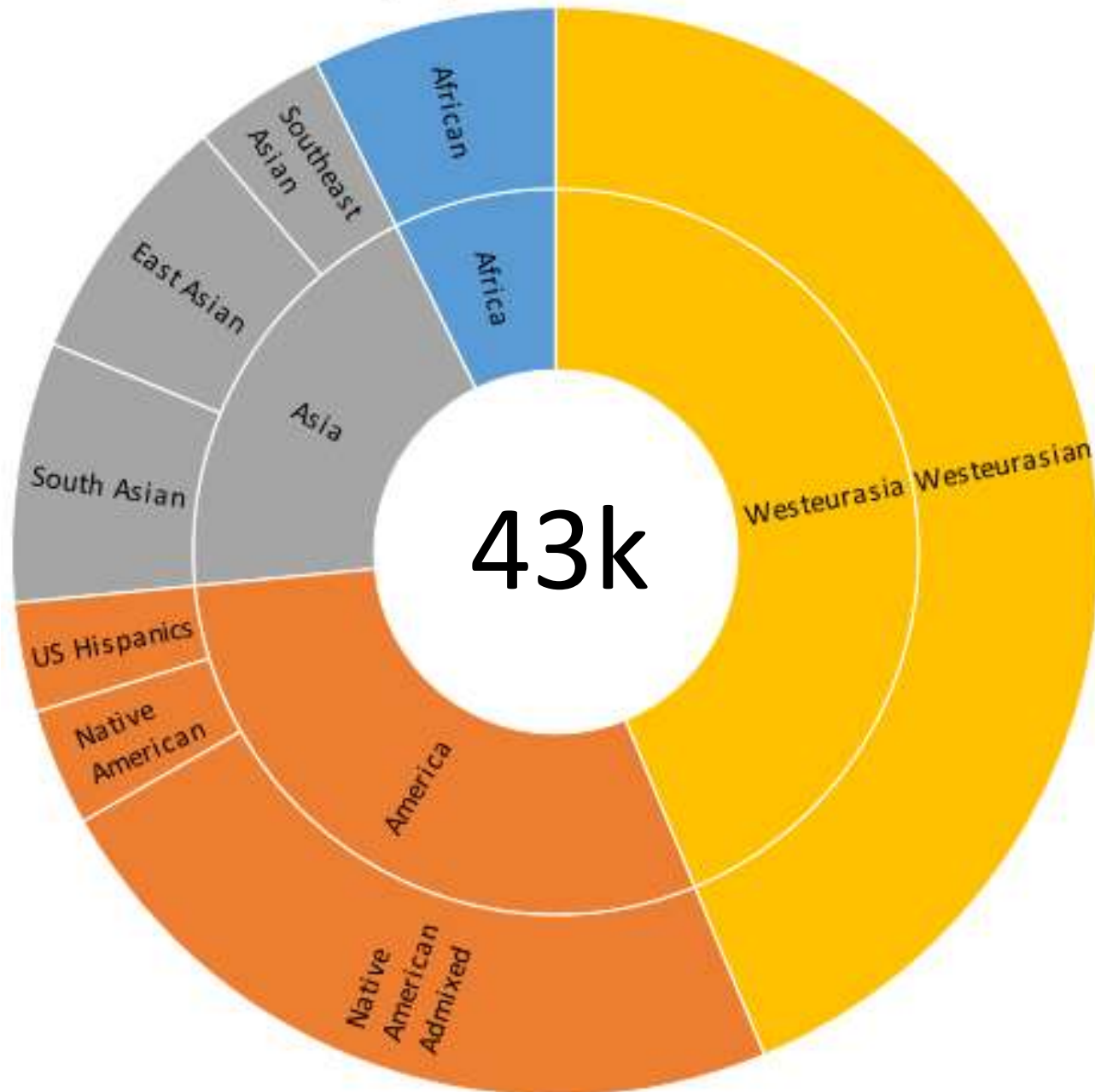
Interpretation Guidelines for  
Mitochondrial DNA Analysis  
by Forensic DNA Testing  
Laboratories



**approved April 23 2019**  
alignment and nomenclature  
based on phylogenetic  
considerations in EMPOP

<https://www.swgdam.org/publications>

## Metapopulation Distribution



## EMPOP Release 12

ORIGIN		
Continent	Country	# samples
Africa	Guinea-Bissau	80
	Angola	5
America	Argentina	899
	United States of America	871
	Bolivia	720
	Brazil	511
	Paraguay	123
Asia	Pakistan	636
	South Korea	515
	Timor-Leste	323
	United Arab Emirates	168
	China	107
	Indonesia	1
Europe	Netherlands	664
	Spain	656
	Romania	563
	Bulgaria	313
	Slovakia	291
	Portugal	256
	Hungary	164
	Greece	150
	Serbia	136
	Ukraine	57
	Norway	16
	Kosovo	1
	Montenegro	1
SUM		8227

## Project aims

- **Improved database query engine**  
SAM2 (Huber et al 2018)
- **Additional markers and populations**  
Extension to mitogenomes and R12 (R13)
- **Tools for QC**  
New data submission tool, extended QC capabilities
- **User friendly access and mobile devices**  
Improved layout, new pdf reports, basic mobile version

# Acknowledgements

## **Development, code programming, testing**

Nicole Huber, Arne Dür (Innsbruck)

## **IT (Innsbruck)**

Stefan Troger, Martin Pircher, vxweb

## **EMPOP 4 testers (international)**

Lara Adams, Kimberly Andreaggi, Laura Catelli, Constance Fisher, Mario Gysi, Douglas Hares, Jodi Irwin, Rebecca Just, Hwan-Young Lee, Sabine Lutz-Bonengel, Charla Marshall, Dixie Peters, Dirk Sauer, John Tonkyn

## **EMPOP analysts (Innsbruck)**

Christiane Bauer, Cordula Berger, Martin Bodner, Mayra Eduardoff, Liane Fendt, Theresa Harm, Antonia Heidegger, Gabriela Huber, Anita Kloss-Brandstätter, Anna König, Simone Nagl, Harald Niederstätter, Daniela Niederwieser, Jannika Oeke, Alexander Röck, Lisa Schnaller, Filipa Simao, Christina Strobl, Catarina Xavier, Bettina Zimmermann

## **Richard Scheithauer (Director)**

**Big THANK YOU to all EMPOP contributors worldwide**

EU 779485 — STEFA — ISFP-2016-AG-IBA-ENFSI





# MONOPOLY 2016 - STEFA - WP G7

Empowering forensic genetic DNA databases for the interpretation of next generation sequencing profiles (**dna.bases**)

STRidER & EmPOP

Jan 2018 - Dec 2019

Sequence alignments

Increase sample size

Increase markers/regions

Further develop QC tools

User-friendly access



**STRidER**

**dna.bases**

**EmPOP**





# Save the Date



YHRD.ORG

**EMP20P**  
YEARS



**HIFS**  
HUNGARIAN INSTITUTE  
FOR FORENSIC SCIENCES



# 12<sup>th</sup> Haploid Markers **2020**

May 13-16, 2020 | Budapest, Hungary

Jan 17 2020 abstract deadline; Mar 06 2020 publication of program

<http://www.hm2020.hu/>





# STRiDER

Martin Bodner<sup>1</sup>, Walther Parson<sup>1,2</sup>

<sup>1</sup> Institute of Legal Medicine, Medical University of Innsbruck, Austria

<sup>2</sup> Forensic Science Program, Penn State University, PA, USA



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

# MONOPOLY 2016 - STEFA - WP G7

Empowering forensic genetic DNA databases for the interpretation  
of next generation sequencing profiles (**dna.bases**)

## Project aims

### ➤ **Improved database query engine**

STRAND developments

### ➤ **Additional markers and populations**

Extension to all marker panels currently available, more pop-data

### ➤ **Tools for QC**

New data submission tool, extended QC capabilities

### ➤ **User friendly access and mobile devices**

Improved layout, new pdf reports, basic mobile version

Final consortium meeting Nov 11-13 2019, Innsbruck



# MONOPOLY 2016 - STEFA - WP G7

Empowering forensic genetic DNA databases for the interpretation of next generation sequencing profiles (**dna.bases**)

STRidER & EmPOP

Jan 2018 - Dec 2019

Sequence alignments

Increase sample size

Increase markers/regions

Further develop QC tools

User-friendly access



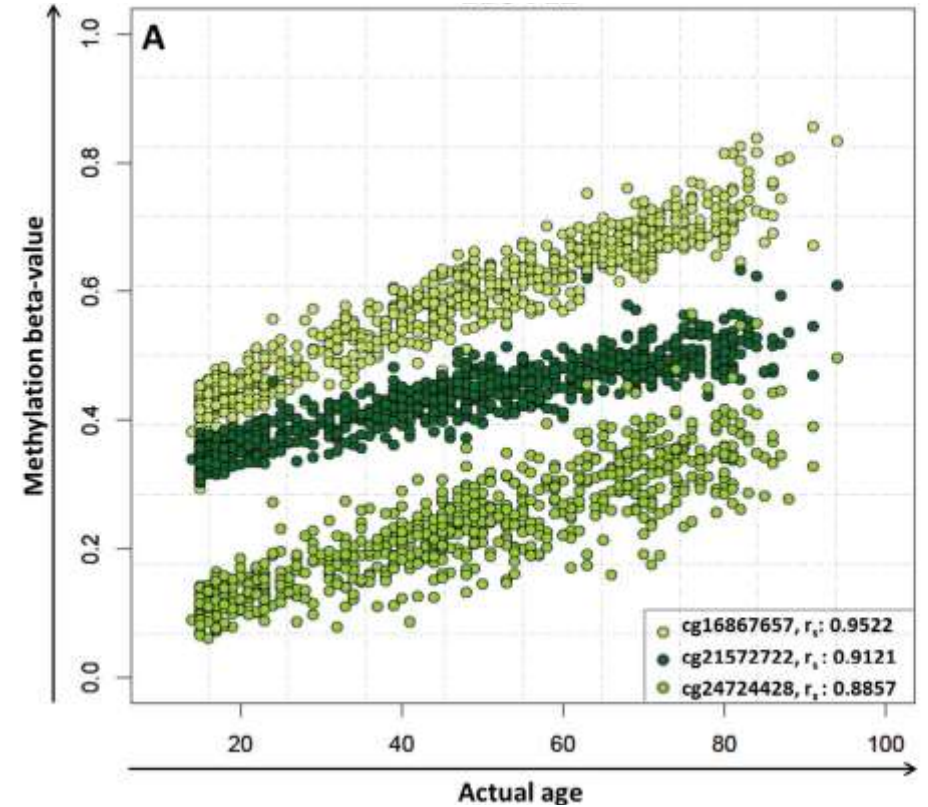
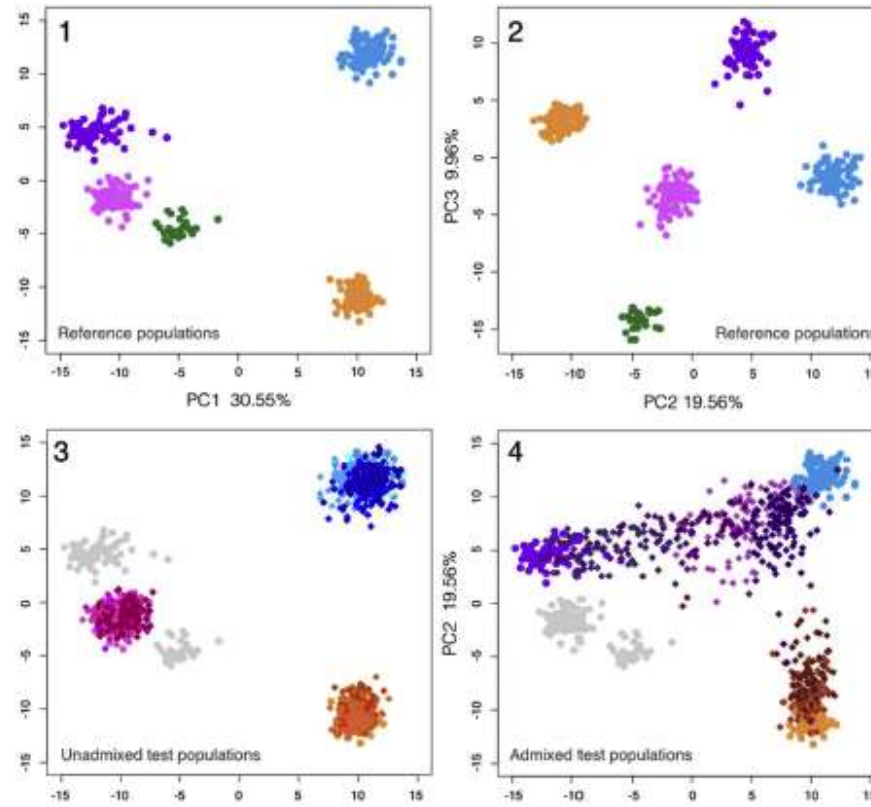
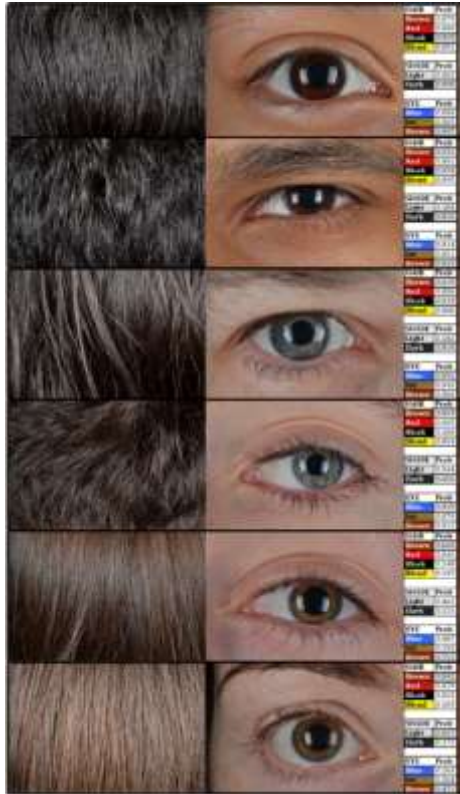
**STRidER**

**dna.bases**

**EmPOP**







# Update VISAGE

Catarina Xavier<sup>1</sup>, Walther Parson<sup>1,2</sup>

<sup>1</sup> Institute of Legal Medicine, Medical University of Innsbruck, Austria

<sup>2</sup> Forensic Science Program, Penn State University, PA, USA

# WP2 MARKER DISCOVERY

## D2.1 Markers for **Basic** Prototype Tool (M3)

Appearance - 41 SNPs (EMC)  
Ancestry - 116 SNPs (USC)  
Age (blood/saliva) - 5 genes / 32 CpG sites (JU)

} 154 SNPs

### Appearance

Hair  
Eye  
Skin

### Ancestry

Continental

## D2.2 Markers for **Enhanced** Prototype Tool (M18)

Appearance - 211 SNPs (EMC)  
Ancestry - 206 SNPs + 22MHTs (USC)  
Age (blood/saliva) - 8 genes / 42 CpG sites (JU)  
Age (semen) - 13 genes / 13 CpG sites (JU)

} 524 SNPs

### Appearance

Hair  
Eye  
Skin  
Eyebrow  
Hair shape  
Freckles  
MPB

### Ancestry

Continental  
Middle East  
North Africa  
X and Y-SNPs



# WP3 PROTOTYPE TOOL DEVELOPMENT

Started in M1 (May 2017) - MUI

D3.1 (M12) Report on new MPS prototype tool(s) for constructing **basic** composite sketches from DNA

D3.2 (M24) Report on forensic developmental validation of new MPS prototype tool(s) for constructing **basic** composite sketches from DNA



# WP3 PROTOTYPE TOOL DEVELOPMENT

Started in M1 (May 2017) - MUI

D3.1 (M12) Report on new MPS prototype tool(s) for constructing **basic** composite sketches from DNA

D3.2 (M24) Report on forensic developmental validation of new MPS prototype tool(s) for constructing **basic** composite sketches from DNA

D3.3 (M24) Report on new MPS prototype tool(s) for constructing **enhanced** composite sketches from DNA

# WP4 STATISTICAL MODELLING

Started in M1 (May 2017) - UoK

D4.1 (M24) Report on new integrative statistical framework for combined appearance, age, and ancestry prediction from DNA

D4.2 Develop prototype software for constructing composite sketches from DNA (M36)

# WP5 ETHICS

Started in M1 (May 2017) - KCL

D5.1 (M12) Report on current and expected legal and regulatory landscape at

(a) EU level and

(b) specific for each participant country

D5.2 (M24) Report on challenges to the implementation of FDP in an ethical and societally responsible manner, with special emphasis on privacy and data protection

# WP6 IMPEMENTATION

Starting in M25 (May 2019) - BKA

D6.1 Implementation of MPS prototype tools for constructing basic composite sketches from DNA (M36)

# WP7 EDUCATION AND TRAINING

Started in M19 (Nov 2018) - UKK

D7.1 (M24) Report on European-wide inquiry on training needs among stake holders and end users

D7.2 Develop training curricula tailor-made for the different target groups on technical, societal, regulatory challenges of constructing composite sketches from DNA in forensic practice (M36)

# VISAGE - Consortium Meeting Lyon, France, 10-11 Sep 2018



[www.visage-h2020.eu](http://www.visage-h2020.eu)



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**illumina**<sup>®</sup>

**VEROGEN**



  
**VISAGE**  
VISIBLE ATTRIBUTES THROUGH GENOMICS





# Acknowledgements



**Catarina Xavier**  
**Antonia Heidegger**  
**Harald Niederstätter**  
**Maria de la Puente**  
**Walther Parson**









# The 28<sup>th</sup> Congress of the International Society for Forensic Genetics

PRAGUE, 9 – 13<sup>TH</sup> SEPTEMBER 2019, THE CZECH REPUBLIC, PRAGUE CONGRESS CENTRE



Alchemy of for

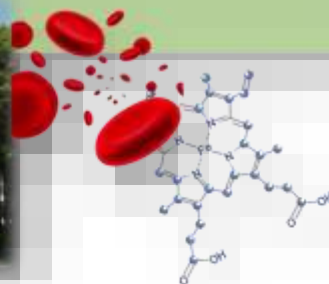
**ISFG Update**

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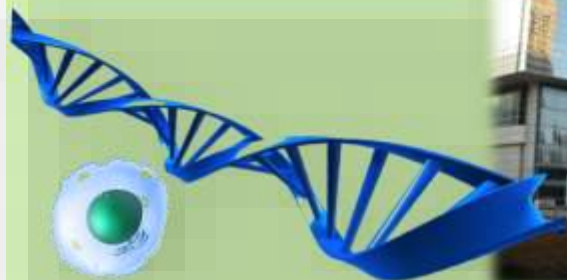
**President:** Walther Parson, Innsbruck • **Vice President:** Mechthild Prinz, New York • **Secretary:** Peter M. Schneider, Cologne  
**Treasurer:** Leonor Gusmão, Rio de Janeiro • **Representative of the Working Parties:** John Butler, Gaithersburg

---

Riga, Oct 23 2019



**50 Years**  
**International Society**  
**for Forensic Genetics**  
**1968 - 2018**



# 28<sup>th</sup> ISFG Congress Prague

**1,017** registered participants

from **64** countries

**753** submitted abstracts

**66** oral presentations (incl. 6 lectures)

**637** posters up

**14** workshops - **473** participants

Germany	105
USA	86
China	65
UK	58
Italy	51
Netherlands	40
Poland	35
Czeck Rep	32
Spain	32
Australia	28

**Thank you** to all workshop coordinators and presenters



# ISFG DNA Commission publication

## Recommendations of the DNA Commission

Gill P., Hicks T., Butler JM., Connolly E., Gusmão L., Kokshoorn B., Morling N., Van O., Parson W., Prinz M., Schneider PM., Sijen T., Taylor D. (2018), 'DNA commission of the ISFG: Assessing the value of forensic biological evidence - Guidelines highlighting the importance of propositions: Part I: evaluation of DNA profiling comparisons given (sub-) source propositions.', *Forensic Science International: Genetics* **36**, 189-202 ([↗REF](#) Gill et al. 2018)

# ISFG DNA Commission publication

DNA Commission of the International Society for Forensic Genetics:  
Assessing the value of forensic biological evidence - guidelines highlighting the importance of propositions. Part II: Evaluation of biological traces considering activity level propositions

Peter Gill et al

# ISFG DNA Commissions in preparation

DNA Commission of the International Society for Forensic Genetics:  
“Interpretation of Y-STR analysis”

Lutz Roewer et al in preparation

# CONGRESS PROCEEDINGS

# Freely available

## Conference Volumes of the International Society for Forensic Genetics

### ✚ **Progress in Forensic Genetics 17**

27<sup>th</sup> Congress of the International Society for Forensic Genetics,  
Seoul, Republic of Korea 2017

Edited by: S.H. Lee, S.D. Lee, H.Y. Lee and M. Prinz

Forensic Science International Genetics Supplement Series, Vol. 6, No. 1, 2017

### ✚ **Progress in Forensic Genetics 16**

26<sup>th</sup> 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**

25<sup>th</sup> 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



# FSIG and FSIR



IF (2018)= 4.884





# Member statistics

Total number (September 2019): 1393

Countries of origin: 84

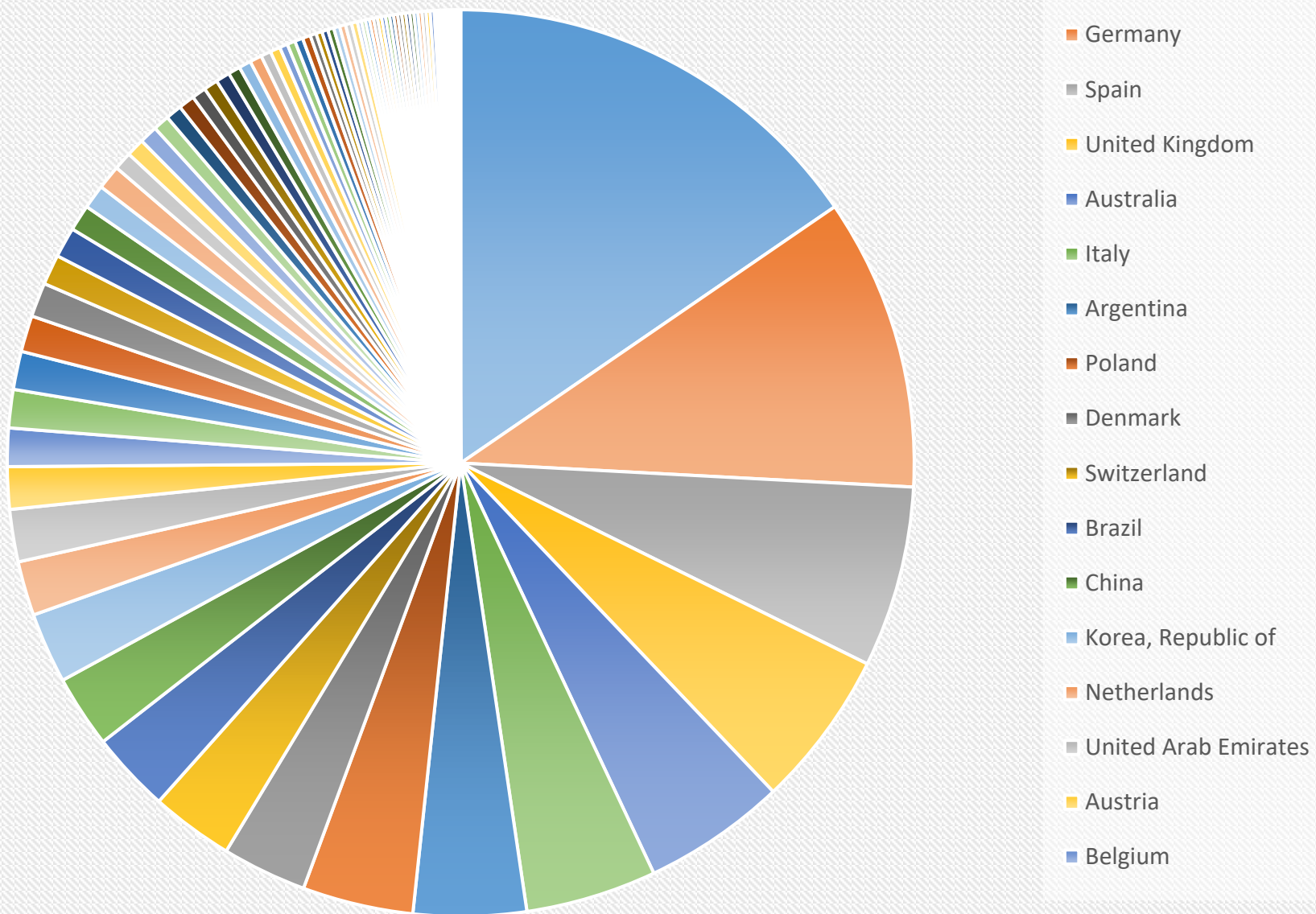
Membership has increased by 68 persons since August 2017

Changes 2017-2019:

New members accepted: 330

Members removed: 246

## 1393 ISFG members from 84 countries



# Congress Travel Bursaries

Support young scientists presenting at an ISFG congress



# Status of ISFG Working Groups



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

Working Group	Working Group Chair (Location)	Recent Activities
German	<b>Uta-Dorothee Immel</b> (Mainz)	Met June 2018 (Freiburg), June 2019 (Cologne), <b>Sept 2019 (Prague)</b>
English (ESWG-ISFG)	<b>Andreas Tillmar</b> (Linköping)	Met Sept 2018 (St. Petersburg), <b>Sept 2019 (Prague)</b> ; conducted 2018 and 2019 Relationship Testing Workshops
French	<b>Diane Séguin</b> (Montréal)	Met June 2018 (Crete), June 2019 (Brussels), <b>Sept 2019 (Prague)</b>
Italian (Ge.F.I.)	<b>Loredana Buscemi</b> (Ancona)	Met Sept 2018 (Catanzaro), <b>Sept 2019 (Prague)</b> ; hosted ISFG Summer School in Sept 2018; <i>prepared recommendations for ID</i>
Spanish & Portuguese (GHEP-ISFG)	<b>Ulises Toscanini</b> (Buenos Aires)	Met Sept 2018 (Araraquara), <b>Sept 2019 (Prague)</b> ; published 3 articles since 2017; active proficiency test program
Chinese	<b>Yiping Hou</b> (Sichuan)	Met Aug 2017 (Seoul) with Asian DNA WG, <b>Sept 2019 (Prague)</b>
Korean	<b>Soong Deok Lee</b> (Seoul)	Organized ISFG 2017, <b>Sept 2019 (Prague)</b>
Polish	<b>Wojciech Branicki</b> (Krakow)	Met Nov 2017 (Krakow), <b>Sept 2019 (Prague)</b>
DNA Commission	<b>Peter Gill</b> (Oslo)	Published 2 articles since 2017
EDNAP	<b>Niels Morling</b> (Copenhagen)	Actively meet twice a year; published 2 articles since 2017
CaDNAP	<b>Cordula Berger</b> (Innsbruck)	Met <b>Sept 2019 (Prague)</b> ; organizing bi-annual proficiency tests

# Proposal for an Arabian Speaking Working Group



- There are 73 Arabian speaking members of ISFG (as of March 2019)
- Members come from 11 countries: United Arab Emirates, Oman, Qatar, Kuwait, Iraq, Bahrain, Algeria, Jordan, Lebanon, and Kingdom of Saudi Arabia
- Proposal provided by Dr. Rashed Alghafri, Head of Biology and DNA Section, Dubai Police, United Arab Emirates
- >50 Arabian speaking members have expressed support for the idea of initiating this working group
- Goal is to exchange experience, quality control and stimulate cooperation between members



# ISFG Newsletter

4 newsletters published since past meeting



## NEWSLETTER 7-2019

INTERNATIONAL SOCIETY FOR FORENSIC GENETICS

<http://www.isfg.org>

### WELCOME

In this edition of our ISFG newsletter, we provide information about the upcoming ISFG Congress in Prague, on new short-term fellowships for collaborative research & travel, about new publications as well as about the ISFG Congresses in 2021 and 2023.

annex to this newsletter. Please bring a copy of this agenda to the meeting to identify yourself as a member of our society.

At the meeting, **elections will be held** for the positions of **President, Treasurer** and **Representative of the Working Parties**. To save time, we will introduce a new "streamlined" election procedure where all three roles will be on the same ballot sheet.



# Thank you Mecki

## **2003 ISFG Meeting Arcachon, France**

Elected as Representative of Working Parties

## **2011 ISFG Meeting Vienna, Austria**

Elected as President

**Vice-president since 2015**



**Mecki Prinz, New York**

# Membership Fee

We decided to keep the membership fee at Euro 60 / year



# Best Poster Presentation @ISFG Prague

**Piyamas Kanokwongnuwut** (Detection of cellular material within handprints)



# Best Oral Presentation @ISFG Prague

**Sofie Claerhout** (MPS panel with 12,523 Y-SNP markers)



# ISFG Scientific Award

**Christopher Phillips** (AIMs, SNP+STR software)



**Thore Egeland** (paternity statistics software)





# New Honorary Members



**Hermann Schmitter & Ate Kloosterman  
EDNAP Members in Bramshill 1990  
Honorary ISFG members in 2019**

# Election of Board members



**President**  
John Butler



**WG Representative**  
Leonor Gusmao



**Treasurer**  
Marielle Vennemann



# iSFG 2021

Washington, D.C. // AUGUST 23-28, 2021

More information coming soon.





Netherlands Forensic Institute  
*Ministry of Justice*

## DNAXs DNAStatistX



A new software suite for data  
management and probabilistic  
interpretation of DNA profiles

Alexander Kneppers  
NFI Division Biological Traces

ISFP-2017-AG-FORENSIC - **DNAXs2.0**

EDNAP October 2019





# History

- NFI experience with development of:
  - Automation solution for the laboratory process
  - Automation solution for the storage of samples
  - Software tools used in DNA case work
    - Bonaparte/Napoleon
    - LOCIM tool
    - LRmix/LRmix Studio
    - SmartRank
    - MixCal







## Key projects within NFI strategy

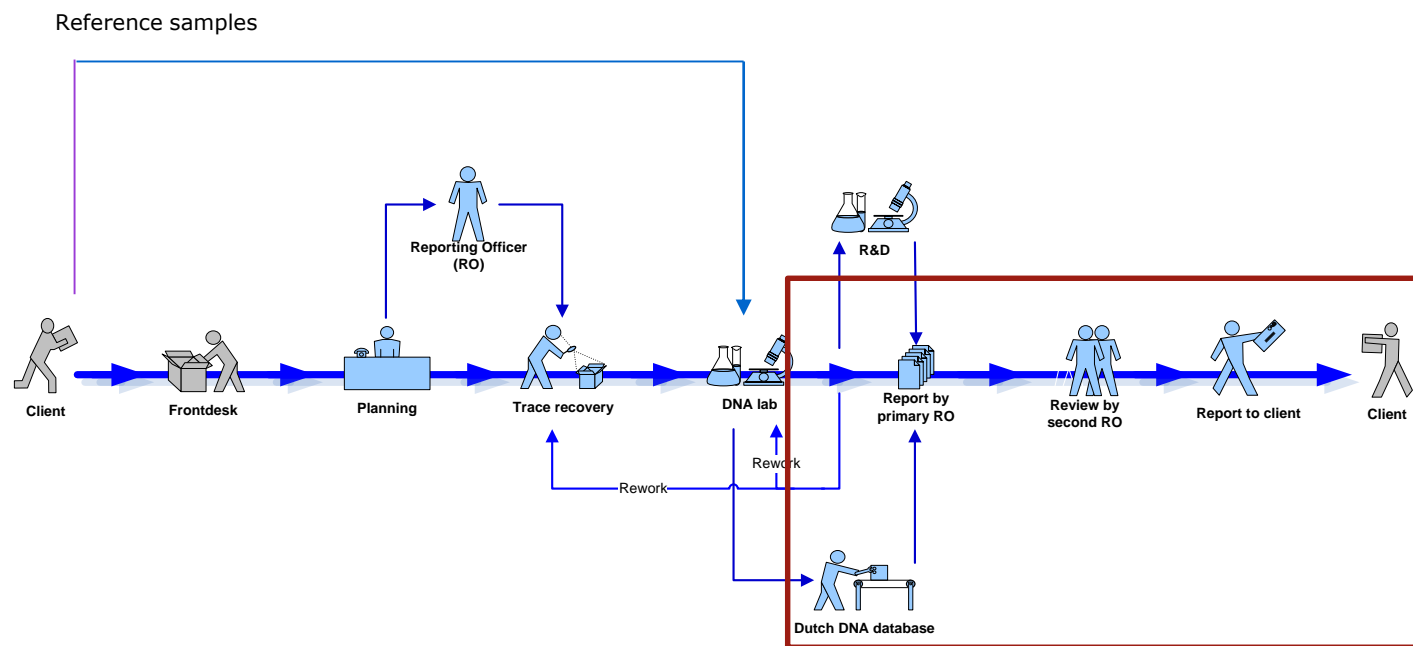
Projects to

- Allow more capacity for more casework and more traces per casework
- Allow a faster workflow and fast answers in the Police investigation process > investigative leads
- Allow a reduction in costs in DNA profiling
- Enhance evidential value by gathering more information from traces by development and implementation of
  - Molecular tools
  - Analytical and Statistical tools





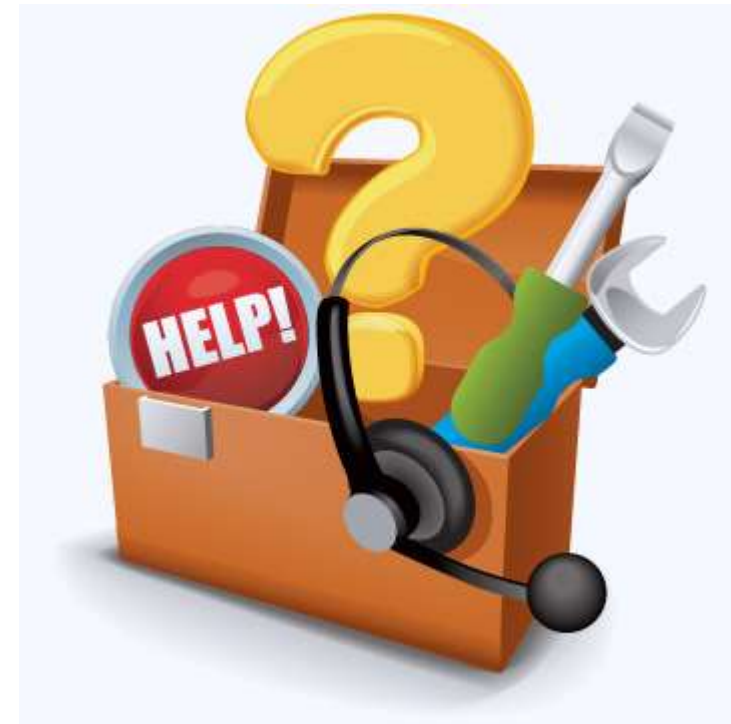
# Workflow: Following a case

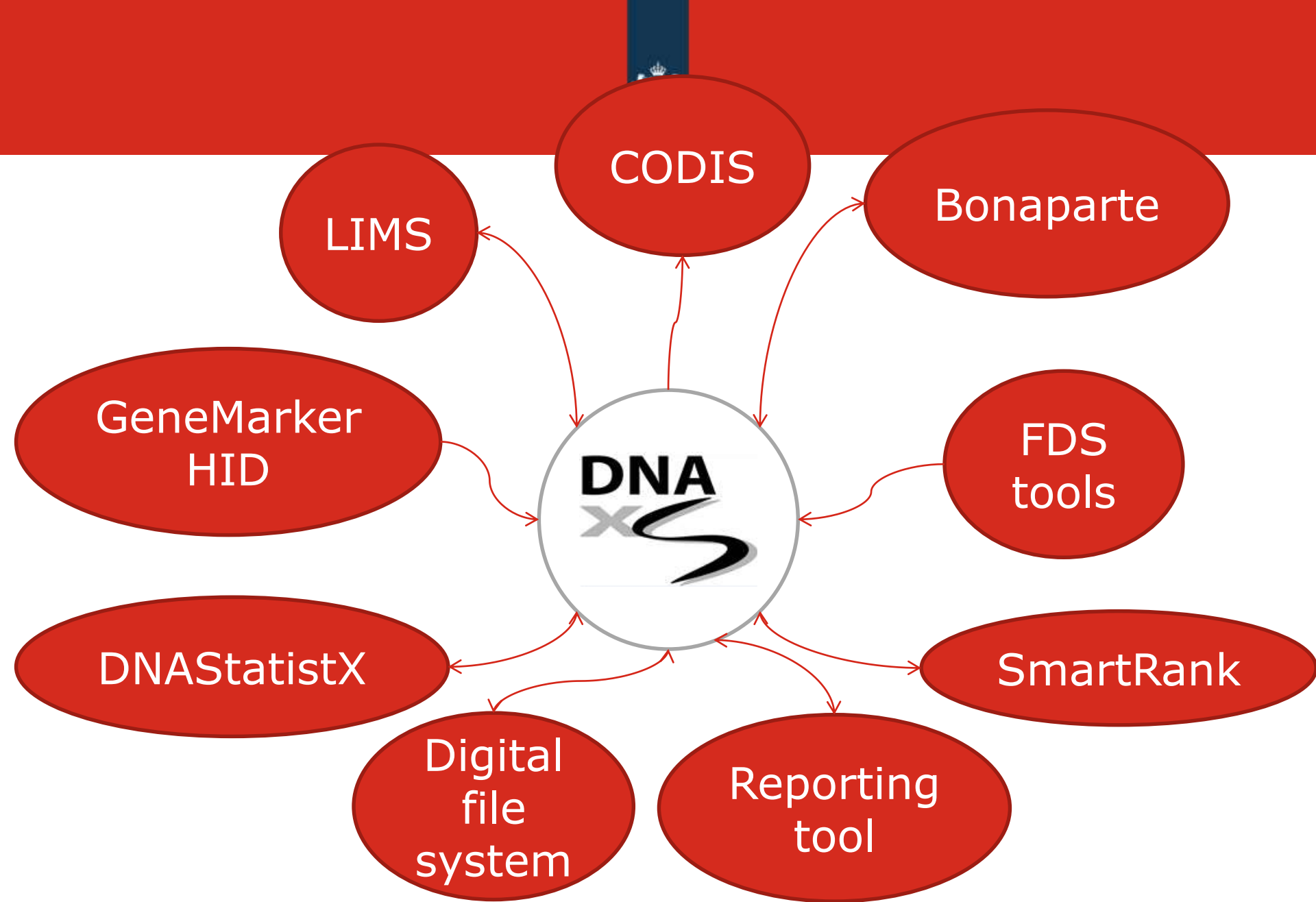




# Support tools in casework interpretation

- Growing number of markers in profiling systems
  - Global STR marker systems available
  - Standard kit the PPF6C kit (27 loci)
- DNA-profile comparison therefor increasingly
  - complex
  - time-consuming
  - error-prone
- Statistical support integrated in casework workflow







# What is DNAXs



- NFI developed DNA eXpert System
  - Automatic comparison of sets of DNA-profiles
  - Summary statistics on allele numbers and genotype reproducibility
  - Mixture interpretation
  - Statistical Analysis (March 2019 release)
- In house built (Java)
- Web application (browser)
- Server based
- Validated according to ISO 17025 and NFI standards
- In use since December 2017
- Three releases per year



# Functionality of DNAXs

- View profiles
  - Overview of runs and peak heights
  - Bar graphs visualizing alleles/peaks heights/read counts
  - Electropherograms, link to pdf of EPG
- Match profiles
  - Trace vs person
  - Trace vs trace
  - Match matrix
- Derive profiles
  - LoCIM inference of major profile, consensus and composite profile
- Statistics
  - DNASStatistX module
  - Summary statistics (TAC/MAC/type I/II/III loci)
- Supports several NFI profiling workflows (from HVC to complex/severe cases)
- Connectivity to other software LIMS/CODIS/SmartRank/DNASStatistX
- Audit trail





# Quality control

- Internal validation according to ISO 17025 standard and internal procedures
  - Validation plans
  - Validation reports
- Q-procedure and software manual
- Internal audit
- External audit
- Integration testing





## DNAStatistX

- Based on EuroForMix R code
- As a separate module within DNAXs
- MLE method
- Degradation module included
- Stutter module not included
- Dye Specific detection thresholds
- Tool for number of contributors



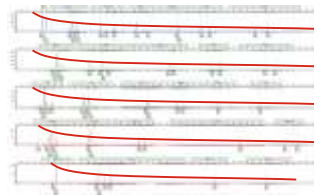
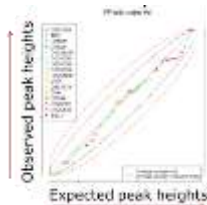
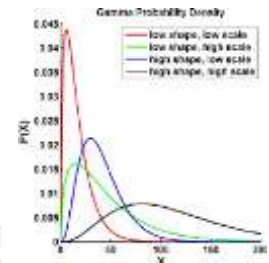
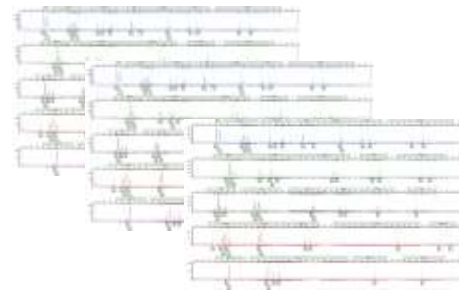




# DNASTatistX features

$$LR = \frac{P(E|Hp)}{P(E|Hd)} = \frac{\sum w(E, Gp|\beta p)P(Gp|Hp)}{\sum w(E, Gd|\beta d)P(Gd|Hd)}$$

- MLE method
- Up to four contributors
- Can handle multiple replicates
- Degradation model
- Model validation



- Aim for a maximum run time of 24h for a four-person mixture with three replicates and four unknowns under Hd





# From EuroForMix to DNASTatistX: What's the same?

LR calculation using maximum likelihood estimate (MLE)

User:

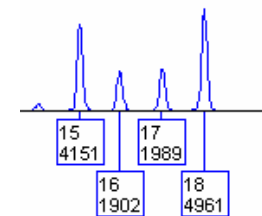
- Define hypotheses

Likelihood computation (under  $H_p$  and  $H_d$ ):

1. Estimate parameters (using optimizer, trial and error)
  - Mixture proportions
  - Peak height expectation
  - Peak height variance
  - Degradation slope
2. Determine possible genotype combinations
3. Calculate genotype probabilities (incl. drop-in)
4. Calculate peak height probabilities (incl. drop-in/-out) for each genotype combination
5. Compute profile likelihood

LR calculation:

- Likelihood  $H_p$  / likelihood  $H_d$



15	16	&	17	18
15	17	&	16	18
15	18	&	16	17



# From EuroForMix to DNAMix: What's different?

- Parts of the EuroForMix code implemented in DNAMix
  - Maximum Likelihood Estimate (MLE)

	<b>EuroForMix</b>	<b>DNAMix</b>
Code	R and C++	Java
Optimizer	nlm	CMA-ES
Model validation	AdaptIntegrate	TrapezoidIntegrator
Rare allele frequency	Lowest frequency at particular locus	$1/(2 \times \text{size of population})$
Detection threshold	Overall	Dye (locus) specific



## Which EuroForMix features in DNASTatistX?

### ➤ Degradation model

- NGM profiles sometimes showed degradation for research samples and often for casework samples
- All PowerPlex Fusion 6C (PPF6C) profiles showed degradation to some extent

Configurable in DNASTatistX, ON by default



### ➤ Stutter model

- Types of stutter:
  - GeneMapper/GeneMarker etc: -2, -1, -0,5, +0,5, +1 repeat unit
  - EuroForMix: -1 repeat unit
- Less specific than profile analysis software, very time consuming

NOT (YET) in DNASTatistX

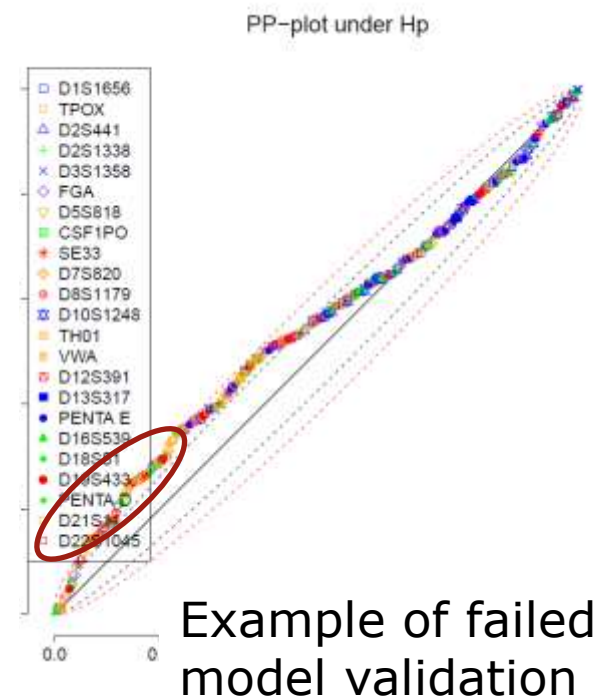
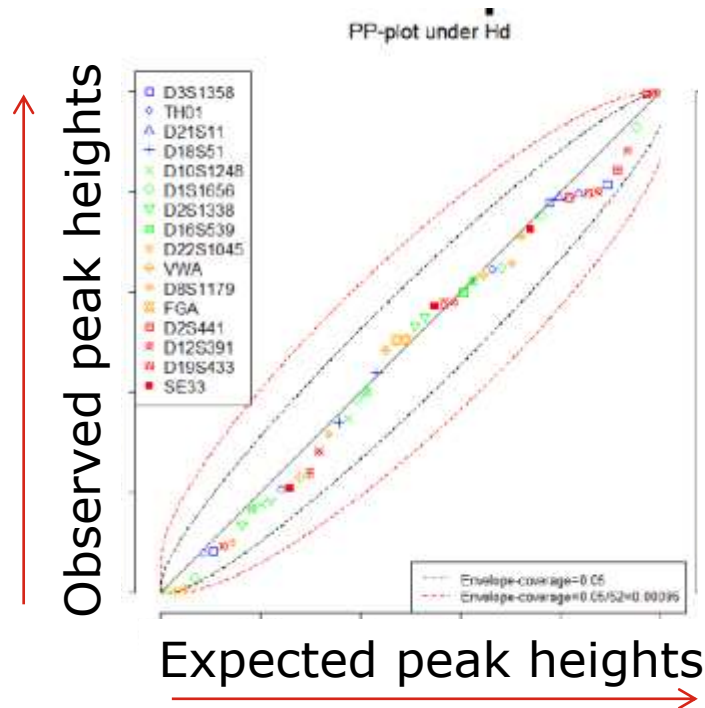




# Which EuroForMix features in DNASStatistX?

## ➤ Model validation

- Important quality check: Do observed PHs follow model's expected PHs



Implementation in DNASStatistX, for every analysis





## Developmental validation of DNASTatistX

- Accuracy:
  - Comparison to analyses using ground truth parameters
  - Comparison to EuroForMix
- Precision:
  - Repeated analyses
  - Optimizer iterations
- Robustness:
  - Analyses that should fail
- Sensitivity:
  - True positives/ false negatives (Type I errors)
- Specificity:
  - True negatives / false positives (Type II errors)

Using a range of case type samples



## Collaboration with international partners

- Additional funding for research and development
- Enhance quality of software by incorporating integration testing
- Develop DNAXs in a multi-lab tool for profile comparison/evaluation/interpretation
- Across laboratory validation
- Possibility to disseminate software to other forensic institutes

ISFP-2017-AG-FORENSIC - DNAXs2.0



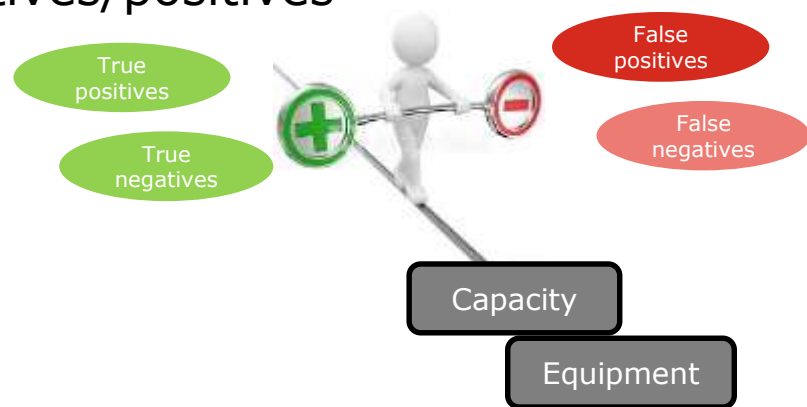




## Create guidelines by

### ➤ Examining:

- True positives/negatives - False negatives/positives
- Effect replicates
- Effect number of contributors
- Effect number of drop-outs
- Etc.



### ➤ Defining:

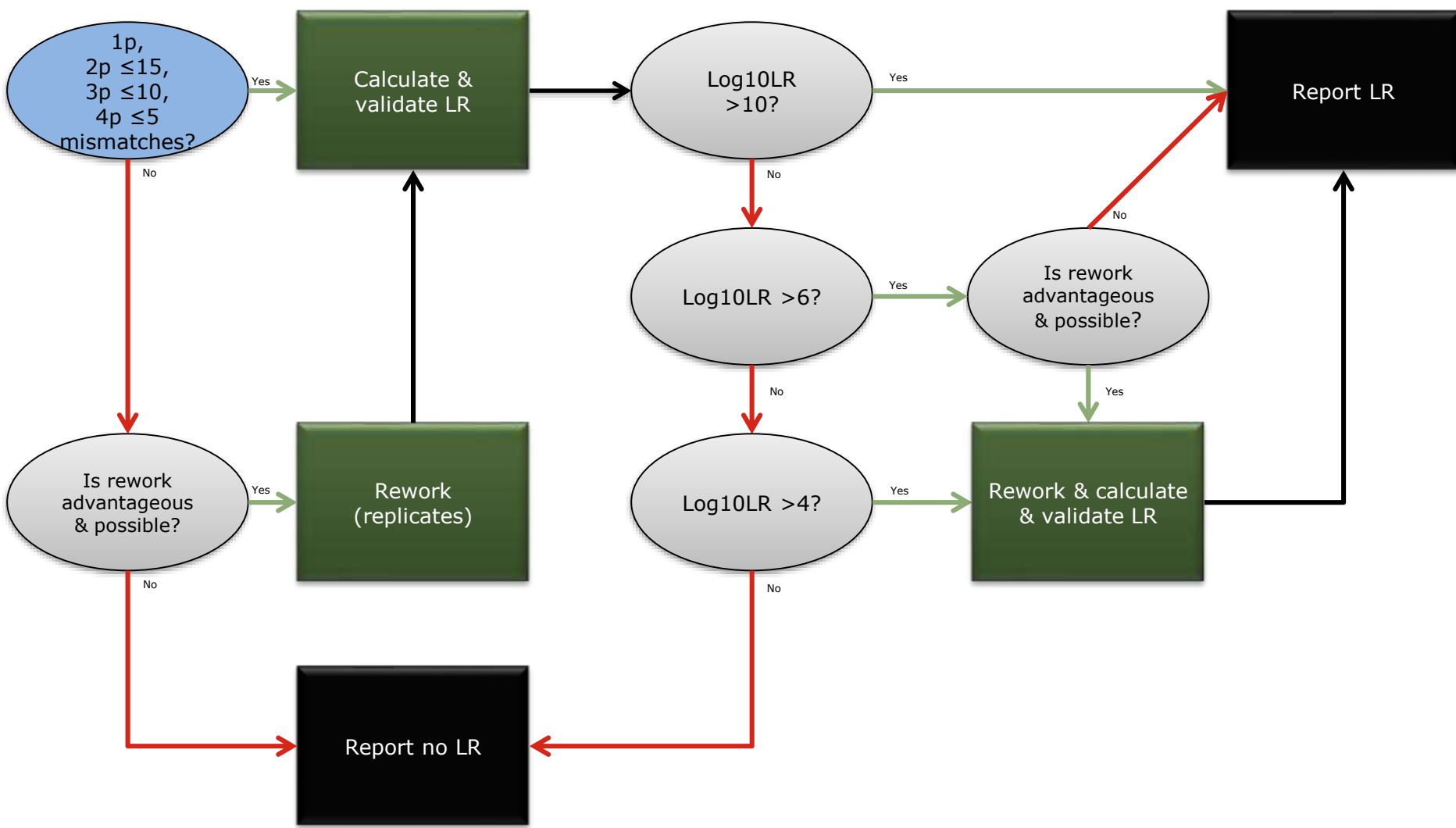
- Sample types/hypotheses for which LR calculations can be informative
- LR threshold
- What to consider when examining results
- What to do if model validation fails
- Etc.



## Define guidelines for use in forensic DNA casework to:

- Ensure chance of obtaining 'false-positive' results is close to zero
- Minimise the number of false-negative results
- Perform LR calculations when regarded useful
- Aim for uniformity among reporting officers







## Future functionalities

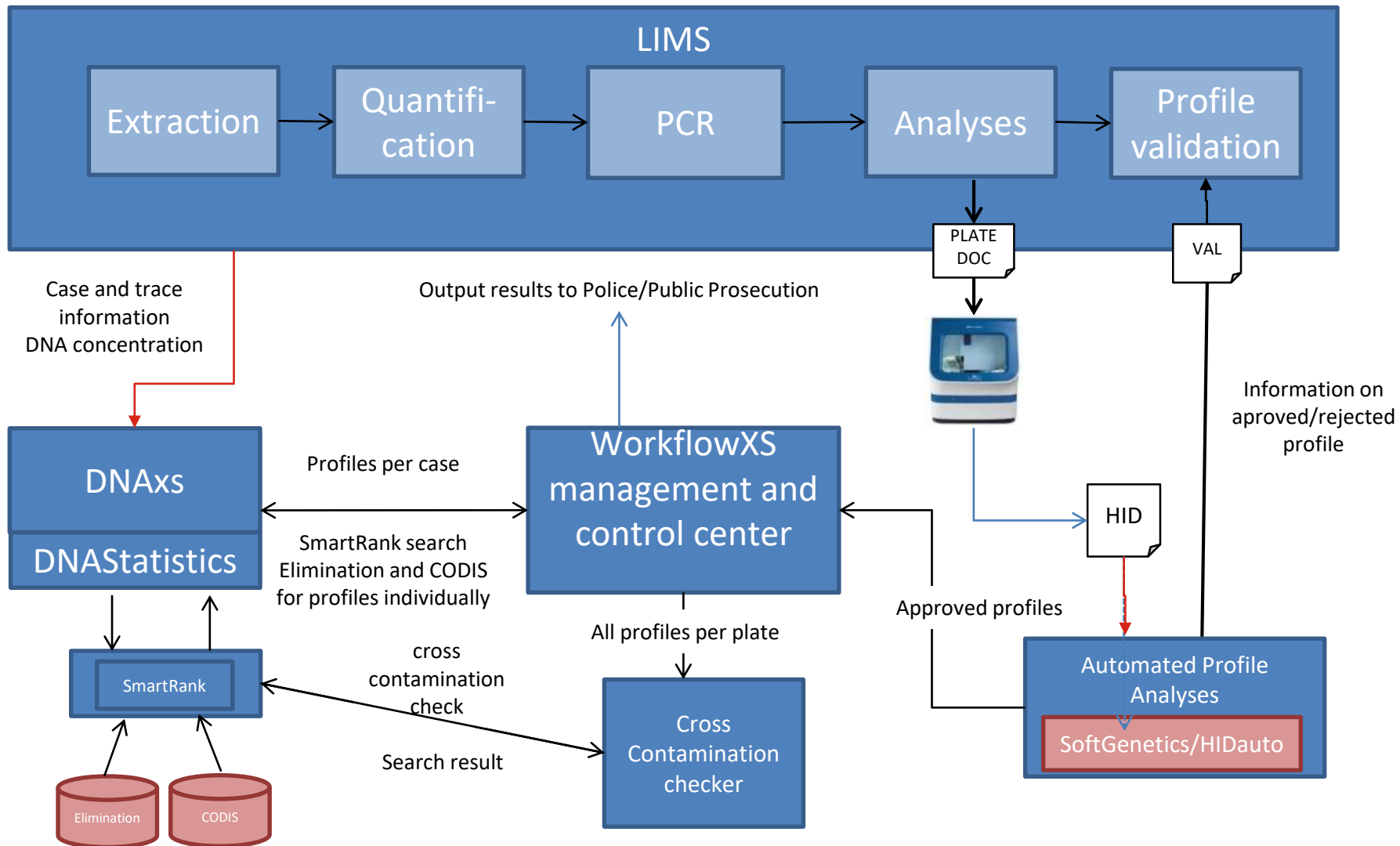
- MPS data; first module for mtDNA
  - only accessible with mtDNA data
  - Sequential matching
  - Release planned for mid 2019
  - EMPOP searches
  - mtDNA matchbox
  - CODIS export
- Followed with STR MPS data
- Under research investigation
  - Stutter model inclusion vs use of laboratory stutter filtered data only
  - Implementation method to estimate number of contributors
  - Deconvolution of all mixed profiles followed by LR computation



## Fast identification workflow

- Fast profile and matching results.
- Speed up of tracing process in the early stages of investigation by having investigative leads.
- Automation of analyses of trace profiles
- Automated SmartRank search
- Automated determination of major donor
- Possibility to switch route to manual process for non processable data
- Automated feedback of match search results to customer IT systems (National Police/Public Prosecution)

# Fast identification workflow





## Time lines

- Kick off meeting February 2019
- Pre validation meeting October 2019
- Validation period October 2019 – Q1-2020
- Dissemination workshops February/April 2020
  - Netherlands
  - Slovenia



**27<sup>th</sup> & 28<sup>th</sup> FEBRUARY 2020**

**The Hague, The Netherlands**

**21<sup>st</sup> & 22<sup>nd</sup> APRIL 2020**

**Ljubljana, Slovenia**

### Workshop details

The workshops will include a series of scientific presentations regarding the functionalities, development, validation, testing and implementation of the DNAXs software. A demonstration will be given after which various hands-on exercises will be performed. The DNAXs software will be provided to the participants for use after the workshop and can be used thereafter within their own laboratories.

### Learning outcomes

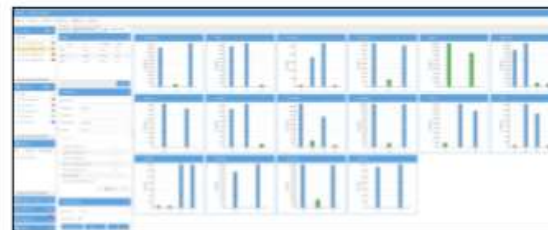
- Get familiarized with the DNAXs software.
- Gain hands-on experience using the DNAXs software.
- Be able to use the software on own cases.
- Know what actions are required for validation and implementation into own laboratory.

### Intended audience

The workshop is intended for laboratories and scientists active in forensic DNA analysis in Europe who are interested in using DNAXs to aid in profile comparisons and evaluation of mixed DNA profiles. The presentations will be aimed towards an audience with experience in DNA profile interpretation, statistical calculations and population genetics with basic computer skills.

### Registration deadline: 6<sup>th</sup> December 2019

Applicants can register their interest for one of the two locations (The Hague or Ljubljana) and will be contacted on 13<sup>th</sup> December 2019 to confirm whether they can participate or not.



**Costs:** no workshop fee nor software costs required. An amount of 100 euro p.p. is required to cover lunch, dinner and software distribution costs. Travel and accommodation at own expense.

The DNAXs2.0 project is funded by the European Union's International Security Fund – Police (Proposal Number: 820838, Proposal Acronym: DNAXs2.0)



**For further information and registration please visit**

<https://www.forensicinstitute.nl/research-and-innovation/european-projects/dnaxs>



## Short video with highlights functionalities DNAXs





For more information on  
DNAxs and to pre register  
for the workshop see

[www.forensicinstitute.nl/research-and-innovation/european-projects/dnaxs](http://www.forensicinstitute.nl/research-and-innovation/european-projects/dnaxs)



## Acknowledgements

### **Software development**

- Christophe Creeten
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- Raymond Parag
- Martin Slagter
- Jennifer van der Linden
- Jennifer Verdier

### **Oslo University Hospital**

- Peter Gill
- Øyvind Bleka

### **Reporting officers**

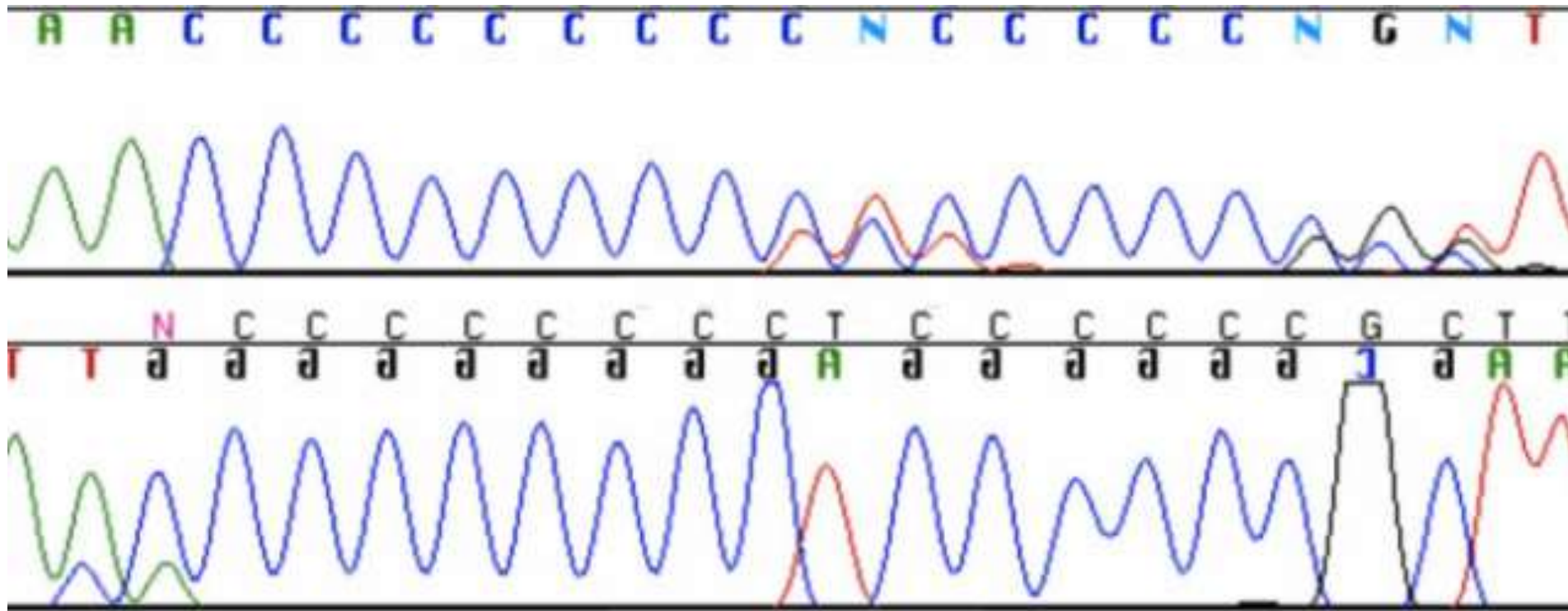
- Patrick Dieltjes
- Jord Nagel
- Heidi van Paassen
- Klaas Slooten
- Kristy Steensma

### **Information technology/network**

- Arie Koppelaar

### **Research**

- Corina Benschop
- Titia Sijen



# mtDNA heteroplasmy exercise

# Dr. Walther Parson

assoc. Prof. Institute of Legal Medicine, Medical University of Innsbruck, Austria

adj. Prof. Forensic Science Program, Penn State University, PA, USA

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# Suggestion for an EDNAP Heteroplasmy exercise

Compare PHP/LHP between CE and MPS on reference samples

**25 labs agreed to participate (+GMI)**

would be a waste of time and money for a **technical** study only

add a **somatic mutation rate study** using hair shafts

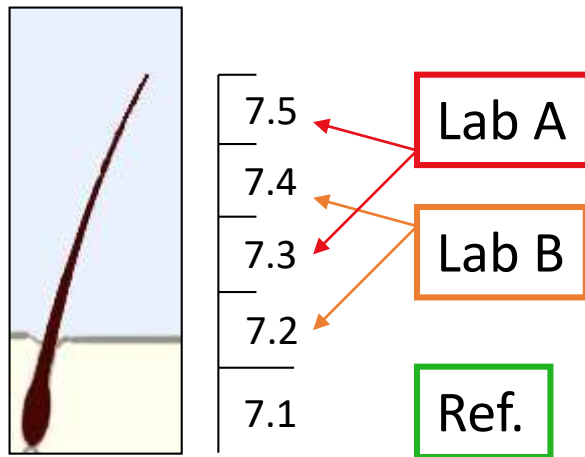
- learn about progression of PHP/LHP along the hair shaft

- estimate size of bottleneck

- provide scientific basis for forensic interpretation of evidence

- (current guidelines are old)

## EDNAP study 55 hair shafts by 10 laboratories



### Results

Different segregation of 16234Y at varying ratios

Also at 16093 and HV2 stretch

16129 transition in one hair

16195 PHP in one hair segment

16304 PHP in one hair segment

Donor's haplotype (blood)

16093C 16129A 16162G 16172C **16234Y** 16304C

73G 249DEL 263G 309.1C 315.1C (hg F1a1)

results confirmed later by independent studies (e.g. Desmyter et al 2016)

Tully et al (2004)



# Somatic mutation rate study

Ask 3 volunteers with long hair

- no heteroplasmy in buccal cells

- moderate heteroplasmy in buccal cells

- severe heteroplasmy in buccal cells

Protocol for hair sample preparation

- excise root

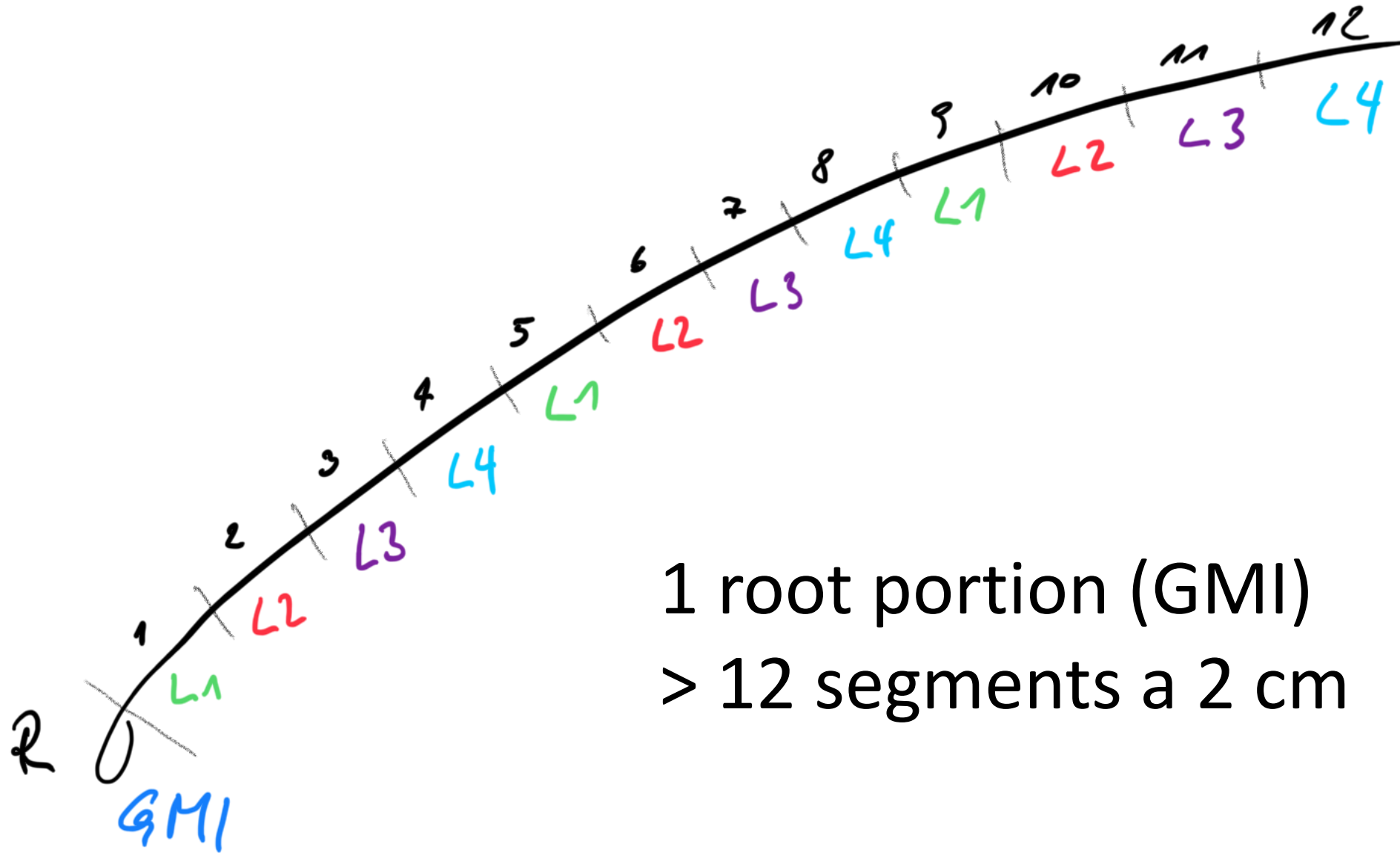
- clean hair shafts (manually)

- cut 2 cm segments

- perform mild lysis (w/o DDT; digest any remaining DNA)

- perform DNA extraction

# Sampling scheme



# Laboratory equipment / method<sup>a</sup>

Method	n
PGM	1
S5	12 <sup>b</sup>
MiSeq	10 <sup>b</sup>
CE/Sanger	8 <sup>b</sup>
<b>total</b>	<b>31<sup>b</sup></b>

CR / HVS-I/II	Mitogenome
31	16

<sup>a</sup> according to the survey (Aug 20 2019)

<sup>b</sup> double specifications

# Hair samples

## Hair samples prepared @GMI

A total of > 650 hair segments

420 DNA extractions

- 360 segments for MPS (Illumina/Ion)

- 60 segments for Sanger (CE)

225 purified hair segments

- (to be extracted by labs)

- 180 segments for MPS (Illumina/Ion)

- 45 segments for Sanger (CE)



Gabriela Huber

# Experimental setup

## Illumina/Ion

### Extracted DNA from hair shafts

Segment	PA001	PA002	PA003	PA004	PA005	PA006	PA007	PA008	PA009	PA010	PB001	PB002	PB003	PB004	PB005	PB006	PB007	PB008	PB009	PB010	PC001	PC002	PC003	PC004	PC005	PC006	PC007	PC008	PC009	PC010
Root	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI
S1	L1	L2	L5	L6	L9	L10	L13	L14	L17	L18	L3	L4	L7	L8	L11	L12	L15	L16	L19	L20	L1	L2	L5	L6	L9	L10	L13	L14	L17	L18
S2	L2	L3	L6	L7	L10	L11	L14	L15	L18	L19	L4	L1	L8	L5	L12	L9	L16	L13	L20	L17	L2	L3	L6	L7	L10	L11	L14	L15	L18	L19
S3	L3	L4	L7	L8	L11	L12	L15	L16	L19	L20	L1	L2	L5	L6	L9	L10	L13	L14	L17	L18	L3	L4	L7	L8	L11	L12	L15	L16	L19	L20
S4	L4	L1	L8	L5	L12	L9	L16	L13	L20	L17	L2	L3	L6	L7	L10	L11	L14	L15	L18	L19	L4	L1	L8	L5	L12	L9	L16	L13	L20	L17
S5	L1	L2	L5	L6	L9	L10	L13	L14	L17	L18	L3	L4	L7	L8	L11	L12	L15	L16	L19	L20	L1	L2	L5	L6	L9	L10	L13	L14	L17	L18
S6	L2	L3	L6	L7	L10	L11	L14	L15	L18	L19	L4	L1	L8	L5	L12	L9	L16	L13	L20	L17	L2	L3	L6	L7	L10	L11	L14	L15	L18	L19
S7	L3	L4	L7	L8	L11	L12	L15	L16	L19	L20	L1	L2	L5	L6	L9	L10	L13	L14	L17	L18	L3	L4	L7	L8	L11	L12	L15	L16	L19	L20
S8	L4	L1	L8	L5	L12	L9	L16	L13	L20	L17	L2	L3	L6	L7	L10	L11	L14	L15	L18	L19	L4	L1	L8	L5	L12	L9	L16	L13	L20	L17
S9	L1	L2	L5	L6	L9	L10	L13	L14	L17	L18	L3	L4	L7	L8	L11	L12	L15	L16	L19	L20	L1	L2	L5	L6	L9	L10	L13	L14	L17	L18
S10	L2	L3	L6	L7	L10	L11	L14	L15	L18	L19	L4	L1	L8	L5	L12	L9	L16	L13	L20	L17	L2	L3	L6	L7	L10	L11	L14	L15	L18	L19
S11	L3	L4	L7	L8	L11	L12	L15	L16	L19	L20	L1	L2	L5	L6	L9	L10	L13	L14	L17	L18	L3	L4	L7	L8	L11	L12	L15	L16	L19	L20
S12	L4	L1	L8	L5	L12	L9	L16	L13	L20	L17	L2	L3	L6	L7	L10	L11	L14	L15	L18	L19	L4	L1	L8	L5	L12	L9	L16	L13	L20	L17

### Prepared hair segments

Segment	PA011	PA012	PA013	PA014	PA015	PB011	PB012	PB013	PB014	PB015	PC011	PC012	PC013	PC014	PC015
Root	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI	GMI
S1	L1	L5	L9	L13	L17	L1	L5	L9	L13	L17	L1	L5	L9	L13	L17
S2	L2	L6	L10	L14	L18	L2	L6	L10	L14	L18	L2	L6	L10	L14	L18
S3	L3	L7	L11	L15	L19	L3	L7	L11	L15	L19	L3	L7	L11	L15	L19
S4	L4	L8	L12	L16	L20	L4	L8	L12	L16	L20	L4	L8	L12	L16	L20
S5	L1	L5	L9	L13	L17	L1	L5	L9	L13	L17	L1	L5	L9	L13	L17
S6	L2	L6	L10	L14	L18	L2	L6	L10	L14	L18	L2	L6	L10	L14	L18
S7	L3	L7	L11	L15	L19	L3	L7	L11	L15	L19	L3	L7	L11	L15	L19
S8	L4	L8	L12	L16	L20	L4	L8	L12	L16	L20	L4	L8	L12	L16	L20
S9	L1	L5	L9	L13	L17	L1	L5	L9	L13	L17	L1	L5	L9	L13	L17
S10	L2	L6	L10	L14	L18	L2	L6	L10	L14	L18	L2	L6	L10	L14	L18
S11	L3	L7	L11	L15	L19	L3	L7	L11	L15	L19	L3	L7	L11	L15	L19
S12	L4	L8	L12	L16	L20	L4	L8	L12	L16	L20	L4	L8	L12	L16	L20

ION

Illumina

# Experimental setup

## Sanger

### Extracted DNA from hair shafts

Segment	PA016	PB016	PC016
Root	GMI	GMI	GMI
S1	L21	L23	L25
S2	L22	L24	Reserve
S3	L23	L25	Reserve
S4	L24	L21	Reserve
S5	L25	L22	Reserve
S6	L21	L23	Reserve
S7	L22	L24	Reserve
S8	L23	L25	Reserve
S9	L24	L21	Reserve
S10	L25	L22	Reserve
S11	L21	L23	Reserve
S12	L22	L24	Reserve

### Prepared hair segments

Segment	PA018	PB018	PC018
Root	GMI	GMI	GMI
S1	L21	L23	L25
S2	L22	L24	Reserve
S3	L23	L25	Reserve
S4	L24	L21	Reserve
S5	L25	L22	Reserve
S6	L21	L23	Reserve
S7	L22	L24	Reserve
S8	L23	L25	Reserve
S9	L24	L21	Reserve
S10	L25	L22	Reserve
S11	L21	L23	Reserve
S12	L22	L24	Reserve

# MitoMetrics

**MitoMetrics** is an adhoc-forming group of scientists and practitioners working with **mtDNA** in the field of **forensic genetics**.

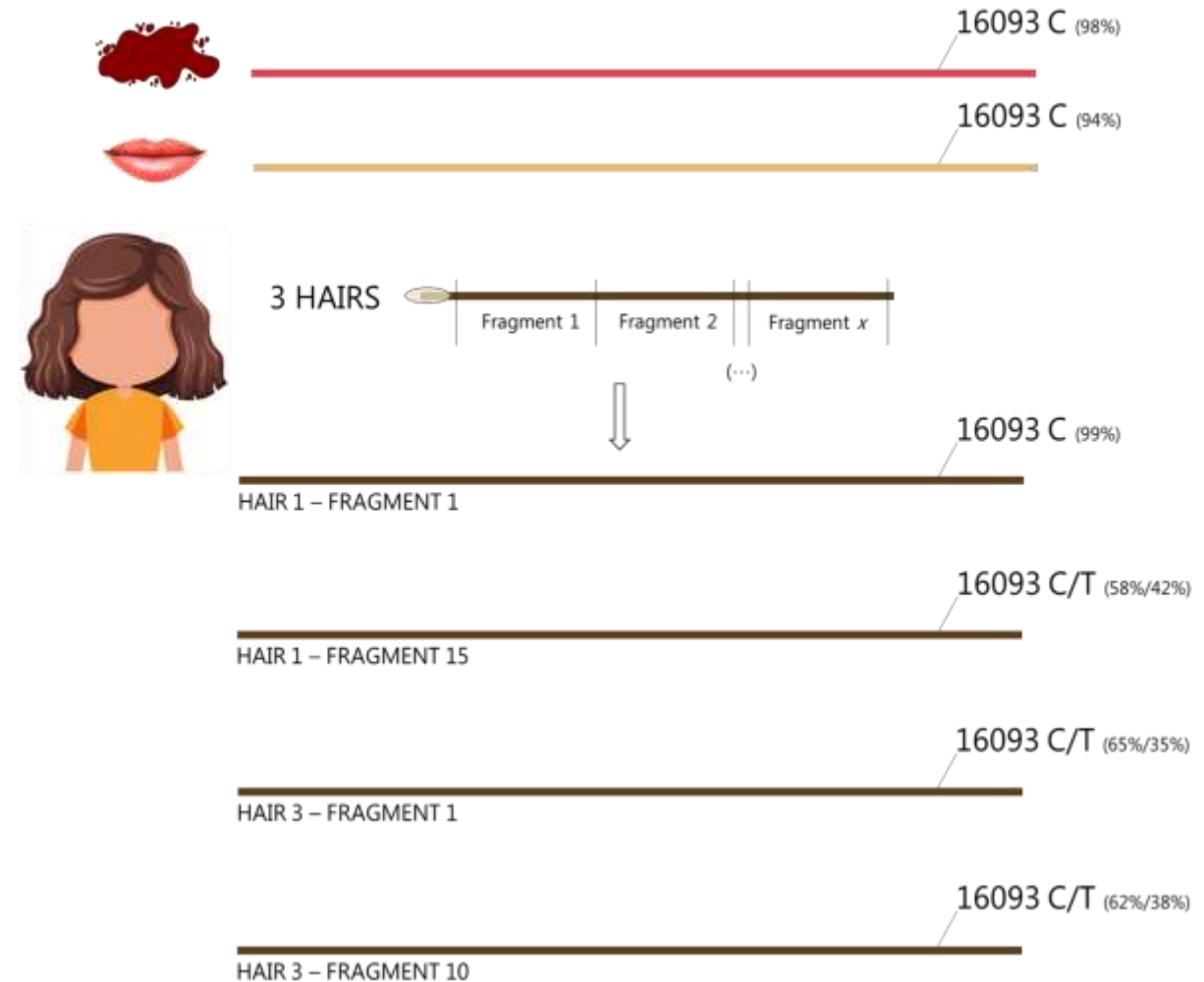
We plan to conduct a **world-wide study** to evaluate **heteroplasmy in hair** and provide a phylogenetically broad data basis to aid **interpretation** of mtDNA evidence in forensic genetics.

Vania Pereira (GHEP countries)

[Vania.Pereira@sund.ku.dk](mailto:Vania.Pereira@sund.ku.dk)

Walther Parson (other worldwide

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# Experiments / lab

SANGER CR	DNA	Hair
Technical study	5	
Somatic study	5	5

MPS (CR/mtG)	DNA	Hair
Technical study	5	
Somatic study	18	9

Shipment of samples - End of October (on a Monday)

Results send back - End of Jan 2020?

Could present at EDNAP Meeting Spring 2020

# Series of exercises relating to DNA transfer

## Proposal for new collaborative EDNAP exercises

By: Bas Kokshoorn (Netherlands Forensic Institute) - b.kokshoorn@nfi.nl.  
Roland van Oorschot (Victoria Police, Australia) - roland.vanoorschot@police.vic.gov.au.

Date: September 2019.

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### Why?

There is an increasing need for forensic scientists to provide guidance on the likelihoods of DNA from a person of interest being present on a specific collection site/location given alternative scenarios. Our capacity to provide such guidance is limited due to scarce availability of relevant data to determine probabilities of contribution of a person-of-interest within a profile of a sample taken from a particular type of item/surface, given a specific event. Further, we know that methodologies, procedures and threshold criteria applied from sampling area determination through to profile generation and interpretation, impact the profile outcomes used in these likelihood calculations. Our knowledge of the extent of these impacts is however limited, which impacts the application of the available DNA transfer related data.

In addition, there appears to be variation among scientists who should provide guidance on transfer of DNA in activity level assessments, more specifically with respect to: (1) understanding of factors affecting the transfer, persistence, prevalence and recovery (TPPR) of DNA; (2) sourcing of relevant data; means of addressing the questions and providing the guidance; and (3) levels/standards of training and proficiency testing on the interpretation and reporting aspects of activity level assessments.

We would like to conduct a series of exercises with the support from, and under the auspices of, EDNAP as they are uniquely established in part to promote and conduct such trials, have an excellent track record, link into a large diverse group of independent laboratories, and are enthusiastic participants.

### Aims

The proposed series of exercise aims to:

- Generate, and make accessible, relevant data to assist activity level assessment.
- Acquire a better understanding of the impact of different sets of methodologies, procedures and thresholds on DNA yields and profile types.
- Build awareness and knowledge associated with DNA transfer and activity level assessment.
- Become aware of the abilities and means of individuals to identify factors within scenarios relevant to the transfer of DNA, and address activity level questions associated with the transfer of DNA.
- Set a benchmark on the current status of 'activity level reporting' in forensic genetics. Depending on the outcome, this series of exercises may illustrate the need for development in certain areas, and will allow for targeted training and research.

## How

- A series of four separate exercises themed around a common/relevant set of items and scenarios.
- Participation in exercises to be offered to EDNAP laboratories plus other laboratories around the world.
- Participation can be for one or more exercises in the series.
- No dedicated funding will be available for this series of exercises. Resources will have to be made available by laboratories upon participation. Each exercise will be designed such that the work load and resources required by a participating lab are deemed acceptable for most interested participants.
- A reasonable period of time will be provided to complete each exercise.

## Type of exercises

Exercise A: *Case file data collection*. Lab participation. Paper/electronically based.

Exercise B: *Experimental data generation*. Lab participation. Laboratory & paper/electronically based.

Exercise C: *Case assessment*. Individual participation. Paper/electronically based.

Exercise D: *Evaluation of findings*. Individual participation. Paper/electronically based.

See attached for general details of draft plan for each of the four exercises A to D.

## When

- The intent is to commence the first exercise (Exercise A) in the first half of 2020.
- Exercise A will be followed by Exercises B, C and D.
- The exercises will run after each other (no overlap in respect to participant workloads), with the aim of running no more than 2 per year.

## Governance and reporting

- Program of exercises to be led by Bas Kokshoorn and Roland van Oorschot.
- Each exercise will also include one or more associated investigators who will co-lead an exercise.
- Potential participating labs will be invited to participate prior to each exercise with general details about the exercise including: aims, what is expected of the participants, anticipated workload, start and completion dates, and deadline to reply.
- The exercise leads will distribute the relevant materials to participants, be available to address questions, analyse the collated returns from participants, prepare a report, and disseminate findings.
- The findings will be reported back to participating laboratories.
- One or more of the exercise leads will attend at least one of the EDNAP meetings each year the program is running to provide status reports.
- Findings of exercises will also be prepared as manuscripts for submission to peer reviewed journals.
- Relevant data collected through exercises A and B will be made available / accessible to participants and the wider community through publications as supplementary materials of associated manuscripts in peer reviewed journals and/or other suitable platform.

- Participating laboratories (and individuals depending on the circumstances) will be identified as one of the participating laboratories in an exercise, but laboratories and individuals will be de-identified within the results and not associated with any specific results (key only known to exercise leads).

## Draft plan of exercises

*(Note, this is a brief outline of the exercises. Details will be discussed and communicated before exercises are finalized and started. Elements may change during further program development)*

### **Exercise A: Case file data collection**

A lot of useful data is stored in case files. Collation, comparison and sharing of case work data will: a) provide a bulk of data, which will be made available, that can be mined for data most suitable to the user's need; b) allow assessment and transparency of the impact of different sets of methodologies and procedures on profile types. This will benefit those conducting activity level assessments as well as those interested in comparing success rates to help drive potential improvement opportunities in respect to the methodologies and procedures they utilise as part of their service delivery. Additionally, questions will be asked to gain insight on how readily the requested information was able to be sourced within each laboratory.

Participants will be asked to collate as much available information as possible relating to 2 to 4 specific relevant item types. The requested information will relate to: item details; methodologies applied from sample identification and collection through to DNA extraction, DNA quantities and profiling; types profile generated. No case identifying information will be requested. The number of items per item type for which data is requested will be capped. A user friendly spreadsheet template, including drop-down options, will be supplied to participants to facilitate recording of requested data.

Cost: Time (approx. 36h per participating laboratory).

Associate exercise lead: Dr Bianca Szkuta (Deakin University, Australia).

### **Exercise B: Experimental data generation**

Data where the ground truth regarding the activity of interest is known is most valuable. However, it is costly and time consuming to generate and cannot be practically done for all situations arising. Further, data generated with one set of methodologies and procedures may have limitation when used where the casework data was generated using different methodologies.

Depending on the further development of the exercise, each lab will be sent a set of a), b) or c):

- a) A prepared set of samples of known ground truth.
- b) A specific set of items, with clear instruction on what activities are to carry out with them.
- c) A clear description of the items to acquire and the activities to conduct with them; and/or samples to be collected from known ground truth situations.

The participating lab will collect samples and process them to generate profiles as per their in-house methods and procedures. The total number of samples to be processed will not exceed 30 and may include reference samples. A user friendly spreadsheet template will be supplied for participating labs to enter their generated data.

Cost: Time (approx. 36h) + consumable and equipment use for up to 30 samples (per participating laboratory).

Associate exercise leads: To be decided.

***Exercise C: Case assessment***

Those performing (and those who may be asked to perform) activity level assessments will identify potential forensic examinations that may differentiate between relevant scenarios in a case. To this end, they need to be aware of the factors impacting DNA transfer, persistence, prevalence and recovery (DNA-TPPR), as well as be able to provide guidance to the requesting authority on the best courses of action. This exercise will test the proficiency of individuals to identify relevant examinations and DNA-TPPR factors of relevance.

This exercise is to be completed by individuals. Participating labs will be able to request multiple tests to be completed by individual staff members. To assist reflection of outcomes, questions will also be asked relating to participants' level of training, casework experience and court experience. Individual participants will remain anonymous. This will be a paper and/or electronic exercise.

Outcomes will inform us of the general level and variation of abilities of those providing (and those who may be asked to provide) activity level guidance, and if there is a need to enhance training efforts.

Cost: Time (approx. 12h per participant).

Associate exercise leads: To be decided.

***Exercise D: Evaluation of findings***

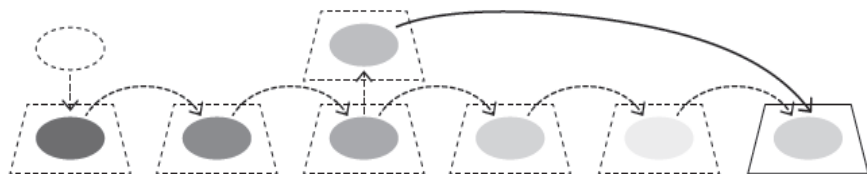
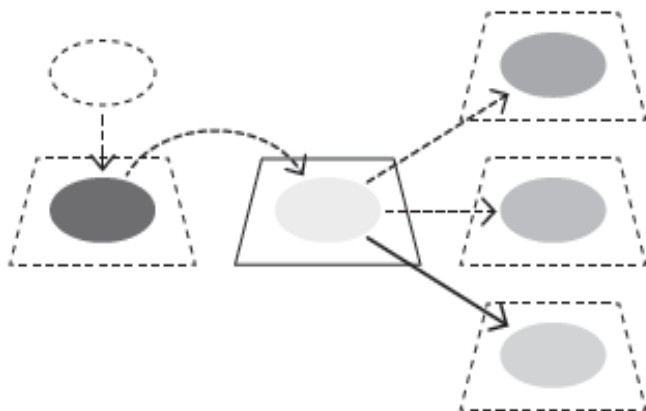
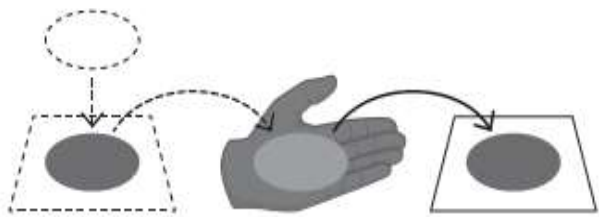
Those performing activity level assessments need not only be aware of the factors impacting DNA-TPPR and be able to recognise relevant factors within various scenarios, but also be able to provide the requested guidance on the matter at hand. This requires consideration of scenarios, sourcing of relevant data, conduct of analyses / assessment, and reporting the guidance. This exercise will test the proficiency by which relevant data is sourced and utilised. It will also collate and compare the current means employed to conduct assessments and provide the requested guidance.

This exercise is to be completed by individuals. Participating labs will be able to request multiple tests to be completed by individual staff members. To assist reflection of outcomes, questions will also be asked relating to the participants' level of training, casework experience and court experience. Individual participants will remain anonymous. This will be a paper and/or electronic exercise.

Outcomes will inform us of the general level and variation in abilities of those providing (and those who may be asked to provide) activity level guidance and the current state of means employed within and among laboratories in respect to reporting on activity level questions. It may also point to needs and directions of future training efforts.

Cost: Time (approx. 20h per participant).

Associate exercise leads: To be decided.



## EDNAP collaborative exercise on DNA transfer

### ***Proposal***

- *Roland A.H. van Oorschot*  
(Victoria Police, Australia)
- *Bas Kokshoorn*  
(NFI, the Netherlands)





# Proposal for series of exercises on DNA transfer

- Introduction to the topic of the series of exercises
- Briefly outline the series of exercises A to D
- Detailing of exercises A and B
- Discussion of all exercises (focus on A and B)

## Series of exercises relating to DNA transfer

### Proposal for new collaborative EDNAP exercises

By: Bas Kokshoorn (Netherlands Forensic Institute) - b.kokshoorn@nfi.nl.  
Roland van Oorschot (Victoria Police, Australia) - roland.vanoorschot@police.vic.gov.au.

Date: September 2019.

#### Why?

There is an increasing need for forensic scientists to provide guidance on the likelihoods of DNA from a person of interest being present on a specific collection site/location given alternative scenarios. Our capacity to provide such guidance is limited due to scarce availability of relevant data to determine probabilities of contribution of a person-of-interest within a profile of a sample taken from a particular type of item/surface, given a specific event. Further, we know that methodologies, procedures and threshold criteria applied from sampling area determination through to profile generation and interpretation, impact the profile outcomes used in these likelihood calculations. Our knowledge of the extent of these impacts is however limited, which impacts the application of the available DNA transfer related data.



## What is at issue?

- **Increased sensitivity and specificity of genetic testing**  
*excellent opportunities for intelligence and investigative purposes*
- **Relevance of biological traces (and trace DNA in particular) in connection to disputed activities increasingly debated**  
*Experts are expected to testify in court on DNA transfer issues*
- **Push from courts and experts alike to move from expertise only to evidence based opinions**  
*Data on DNA transfer, persistence, prevalence and recovery (DNA-TPPR) needed.*



## Sources of data

- 1) Perform experiments that mimic case circumstances to assign probabilities.
- 2) Use literature values from studies that represent similar properties to the case circumstances and outline the differences or limitations in the report.
- 3) Consider a range of reasonable values for the probability of interest and examine the sensitivity of the LR to it.
- 4) Assign a value based on the expert's experience or knowledge, preferably supported by structured analysis of similar case files, which can be justified by an argument, and be disclosed for review, (as required, for example, by the ENFSI guideline), even though the invoked expert knowledge cannot be directly ascribed to a particular study, experiment or validation.



# Availability of data

- Case specific experiments limited by constraints on time and resources
- Published data increasingly relevant and available  
*van Oorschot, R. A., Szkuta, B., Meakin, G. E., Kokshoorn, B., & Goray, M. (2018). DNA transfer in forensic science: a review. Forensic Science International: Genetics, 38, 140-166.*
- However difficult to mine and assign appropriate probabilities  
*Kokshoorn, B., Aarts, L. H., Ansell, R., Connolly, E., Drotz, W., Kloosterman, A. D., ... & van Oorschot, R. A. (2018). Sharing data on DNA transfer, persistence, prevalence and recovery: Arguments for harmonization and standardization. Forensic Science International: Genetics, 37, 260-269.*



## Replication versus iteration...

- **Mechanisms behind DNA transfer**
  - *strictly controlled*
  - *pre-cleaned items*
  - *replicate experiments*
- **Probabilities of DNA transfer close to case circumstances**
  - *semi-controlled*
  - *items with known history*
  - *number of iterations of activities*



## Relevance of published data to case?

- ISFG DNA commission (2019):

*Compilations of experiments are encouraged as a basic validation of use of data from other laboratories, with details of methods used in order to derive the probabilities that can be utilised. An ENFSI supported inter-laboratory example is outlined by Steensma et al. (2017).*



## ENFSI monopoly 2013 – ‘DNActivity’

- Steensma et al. (2018) *DNA transfer and persistence on cable ties*
- Szkuta et al. (2019) *Prevalence of DNA on upper garments*
- Szkuta et al. (2019 - submitted) *Transfer DNA to upper garments*
- Goray et al. (2019 - submitted) *Trace DNA dynamics in office spaces*
- Szkuta et al. (in prep.) *Transfer DNA to lower garments*
- Sjoukema et al. (in prep.) *Structured comparison of lab success rates*
- Kokshoorn et al. (2018) *On sharing DNA TPR data*

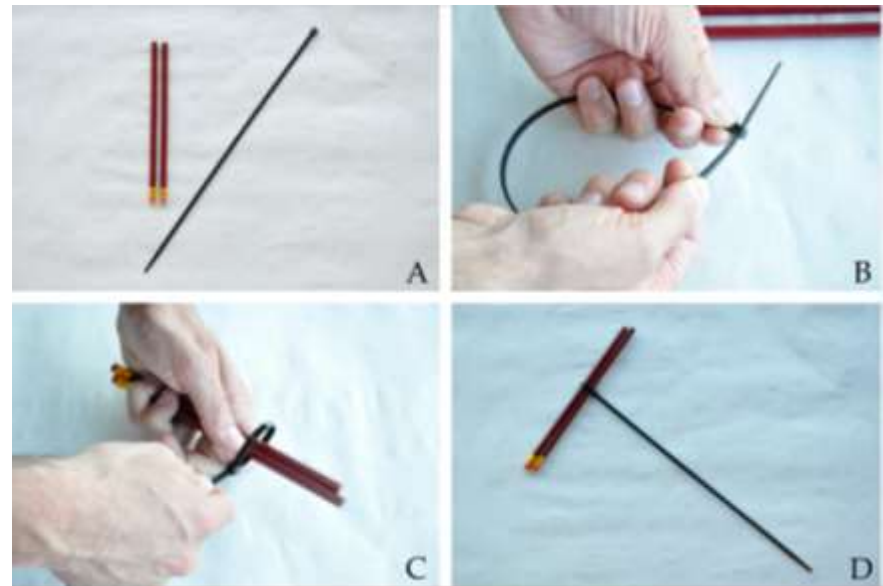


EUROPEAN NETWORK  
OF FORENSIC SCIENCE  
INSTITUTES



## Cable tie study sample collection

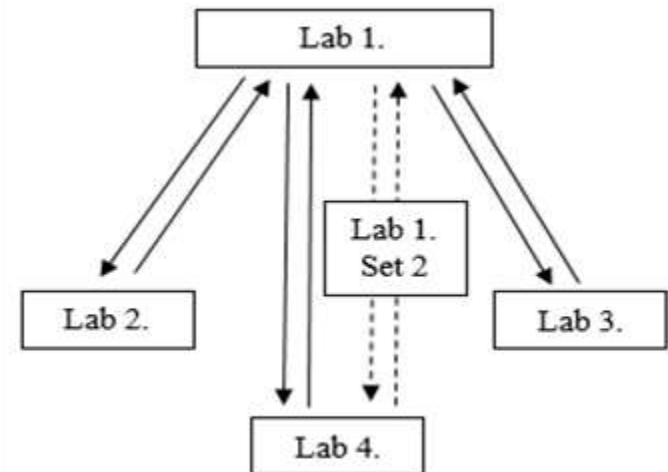
- Cable ties regularly encountered in criminal offences, easy to transport and takes friction and pressure to apply
- 20 participants (10 men and 10 women) each tied 5 cable ties around five pairs of pencils
- 3 hours in between repeat activity
- No instructions
- Questionnaire activity history



## Cable tie study sample set distribution

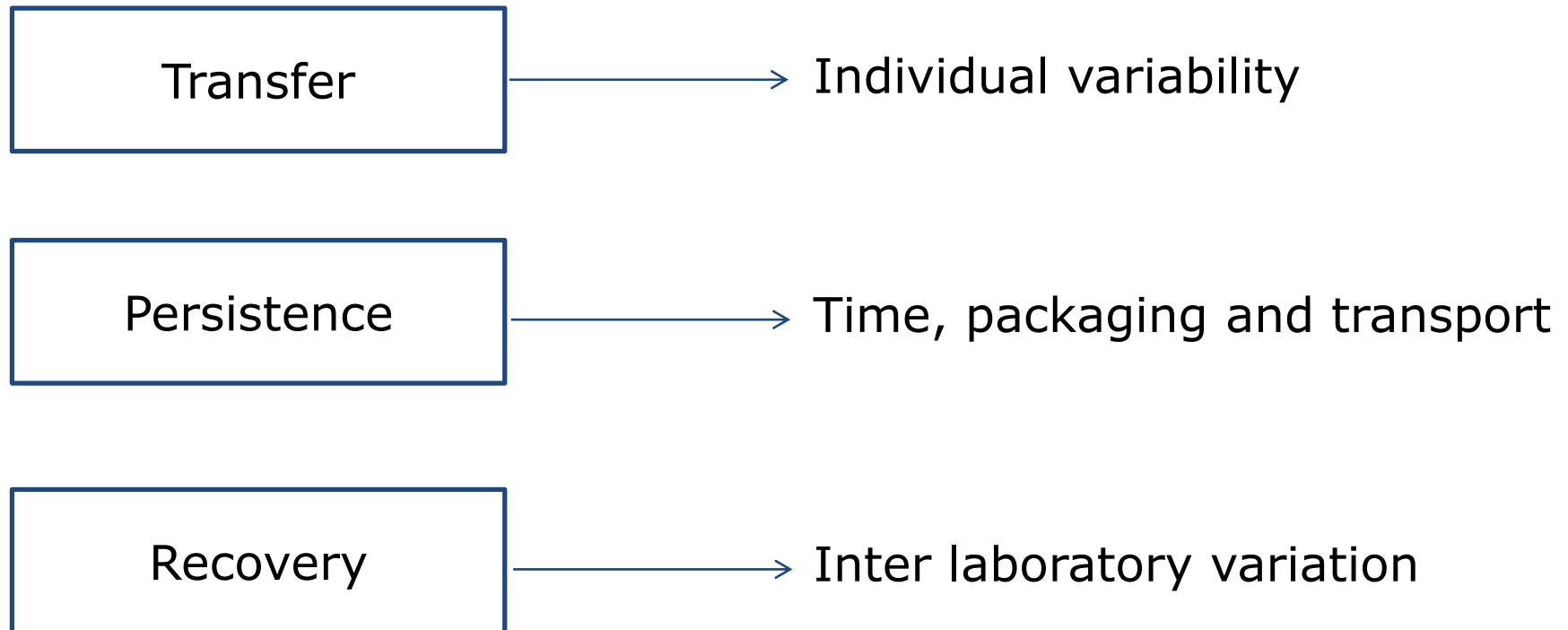
- 4 ISO accredited laboratories from 4 countries
- Cable ties separately packaged
- 21 cable ties sent to every laboratory
- Reference profiles of participants
- Sampled and analysed by laboratory specific protocols

Cable tie packaging



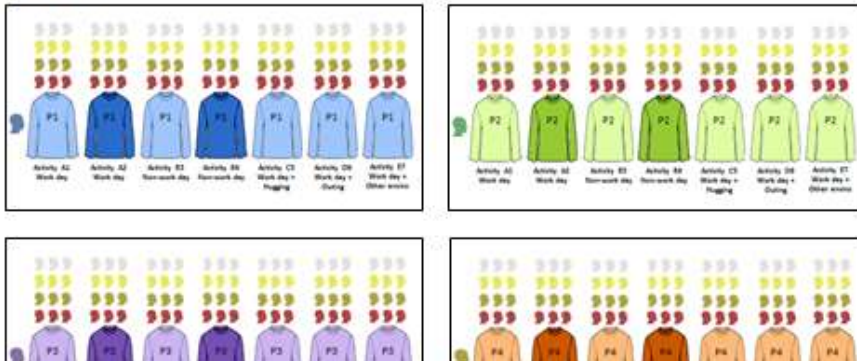


## Examined variables

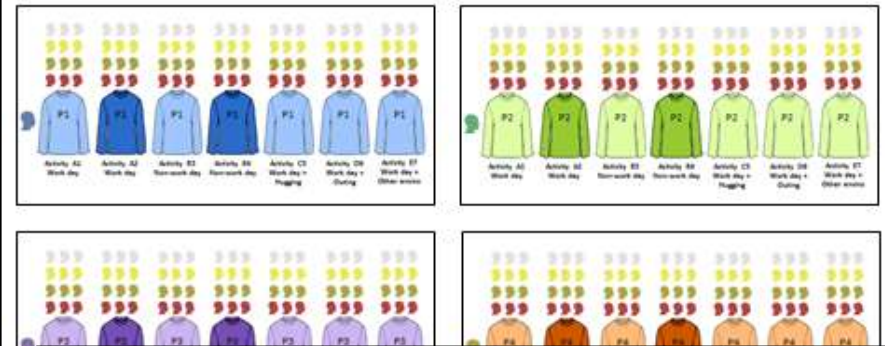


## Further inter laboratory studies

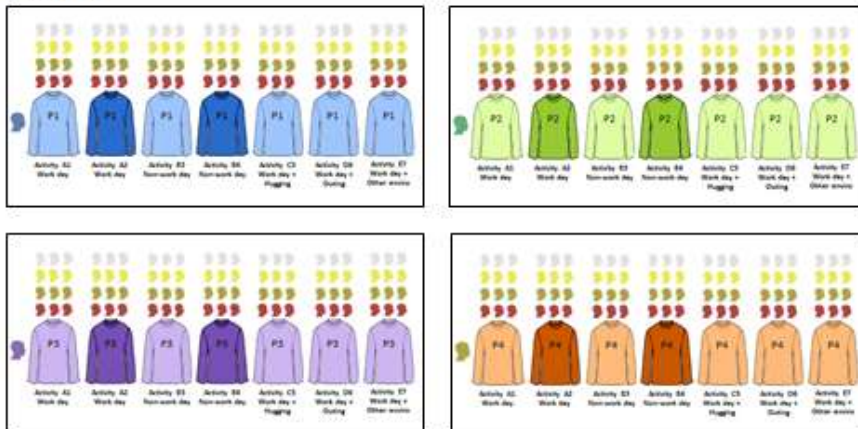
## Lab 1



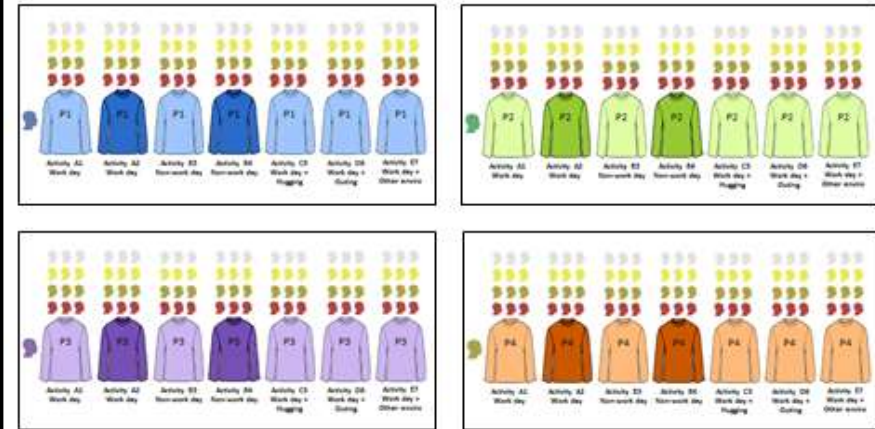
## Lab 2



## Lab 3



## Lab 4





## Outline of series of exercises

**Exercise A:** *Case file data collection.*

Lab participation. Paper/electronically based.

**Exercise B:** *Experimental data generation.*

Lab participation. Laboratory & paper/electronically based.

**Exercise C:** *Case assessment.*

Individual participation. Paper/electronically based.

**Exercise D:** *Evaluation of findings.*

Individual participation. Paper/electronically based.



## Exercise A – Case file data collection

- **Some data published**  
*Non-structured approach, comparison complex/impossible*
- **Relevance for case assessment**  
*Trace analysis success rates for triage*
- **Relevance for evaluation given activity propositions**  
*Can provide information to assign probabilities on DNA-TPPR*
- **Limitations to casefile data**  
*Ground truth generally unknown (but known or irrelevant for some factors) but data readily available*



# Exercise A – Case file data collection

## **Purpose**

First collaborative exercise on lab success rates to help drive potential improvement opportunities in respect to the methodologies and procedures they utilise as part of their service delivery.

Collate information on frequency of specific types of profiles given contextual information (item type, substrate etc.) and methodologies

Additionally, the exercise will gain insight on how readily the requested information was able to be sourced within each laboratory.





## Exercise A – Case file data collection

### **Selection of items to target**

- Items commonly analyzed in participating labs
- Limited variation in substrate
- Limited variation in use/item histories
- Limited variation in type of sample targeted (no body fluids)

## Exercise A – Case file data collection

### **Suggested items to target; two broad categories**

#### *Burglary related tools*



#### *Gloves*



## Exercise A – Case file data collection

- ✓ Items commonly analyzed in participating labs
- ✓ Limited variation in substrate (categorize)
- ✓ Limited variation in use/item histories (exclude exceptions)
- ✓ Limited variation in type of sample targeted (exclude body fluids)





## Exercise A – Case file data collection

### **Information to collect**

- Item type and known history
- Sampling method
- Sampling area and substrate
- Sample analysis methods
- Analysis and interpretation methods
- Resulting DNA profile composition
- Resulting inclusion/exclusion of Persons-of-Interest

*Use of tested, user-friendly spreadsheet*



## Exercise B: *Experimental data generation*

**Keep it simple!** Suggest to go for option (a) in document.

- Focus on sampling through to profiling methodologies  
*Have collaborators sample items and profile samples*
- Exclude impact of experimental execution  
*items Prepared at one location*
- Exclude impact of data analysis  
*DNA profile data analysed and interpreted at one location*
- Burden for ethics and privacy (mostly) on organizing laboratory

## Exercise B: *Experimental data generation*

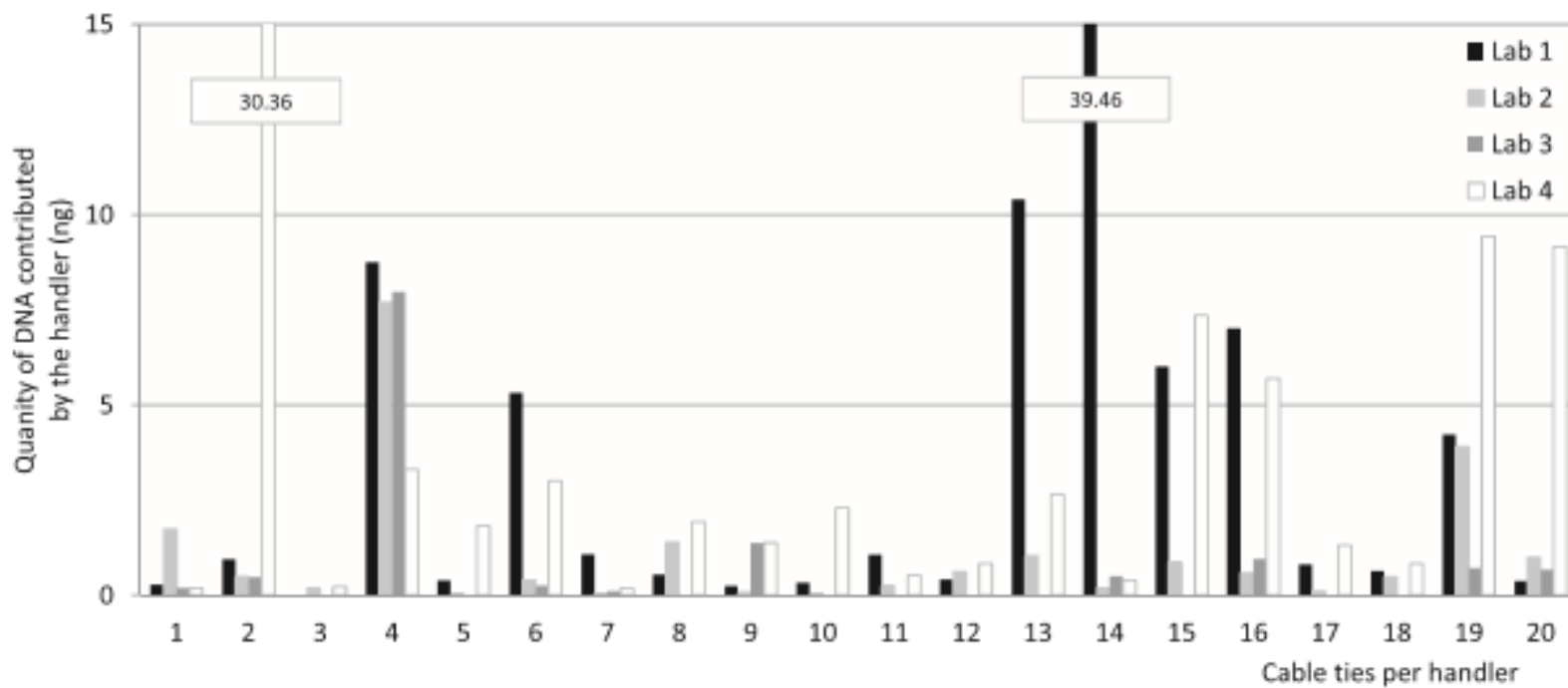
### **Suggested item to target**

- Standardized item
- Standardized activity/handling
- Standardized sampling (e.g. reduce impact of strategy)
- Allows for comparison to case file data (Exercise A; but...)



## Exercise B: *Experimental data generation*

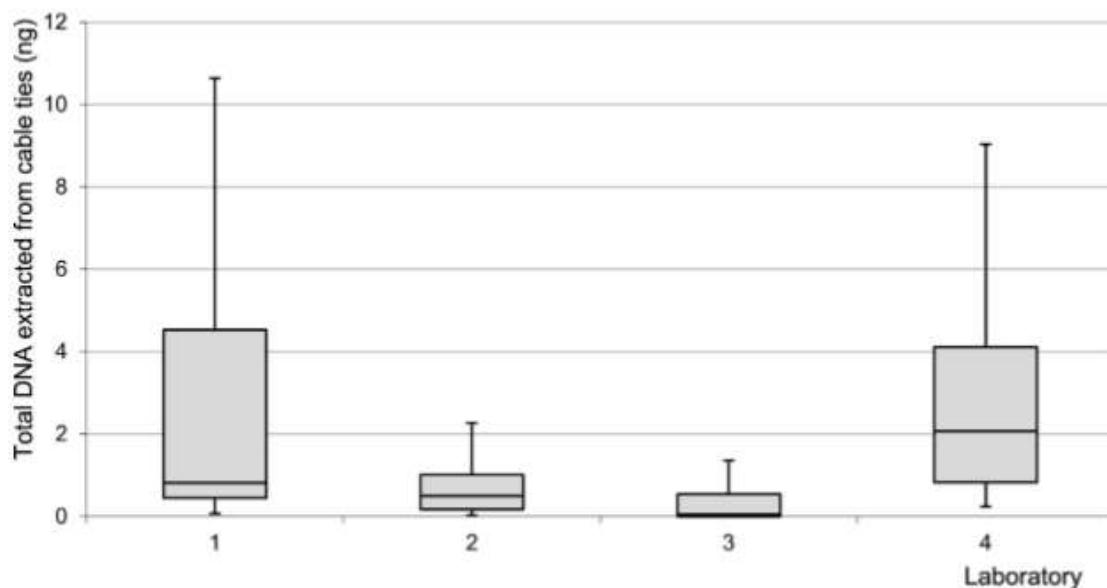
- Yes; individual variation volunteers!





## Exercise B: *Experimental data generation*

- Yes; individual variation volunteers!
- But; not limiting to assess inter-lab variation!



**Fig. 3.** Boxplot of the median quantity of total DNA (ng) extracted by the four participating laboratories from the twenty cable ties.



## Exercise B: *Experimental data generation*

### **Data collection**

The participating lab will collect samples and process items to generate profiles as per their in-house methods and procedures.

The total number of samples to be processed will not exceed 30.

A user friendly spreadsheet template will be supplied for participating labs to enter their generated data.

## Exercise C – Case assessment

### Benchmark on case assessment and triage

- Provide (mock) case
  - *Case issue*
  - *Case information*
- Purpose to compare;
  - *What info would expert use?*
  - *Which scenario's would be considered relevant?*
  - *What factors impacting on DNA-TPPR are being considered?*
  - *What examination strategies would be considered?*
  - *What would be the expected outcomes for examinations?*
    - > *based on which information/expertise?*
  - *What would the recommended strategy be?*



## Exercise D: *Evaluation of findings*

### Benchmark on reporting given activity propositions

- Provide (mock) case
  - *Case context*
  - *Case examination and profiling data*
- Purpose to compare;
  - *Formulating propositions*
  - *Management of case information*
  - *Structure of argument*
  - *Data sources used*
  - *Reporting structure*





## What can participants expect?

- Consistency of exercises; guarded by exercise leads
- Well prepared exercises, convenient item and data transfer
- Minimization of efforts needed
- Access to (anonymized) collated data to be mined for casework
- Communication of progress and findings
  - > at EDNAP meetings
  - > through scientific publication



## Practicalities

- Maximum of 1-2 exercises/year, no overlap  
*Reduce strain on participating labs and leads alike*
- If accepted, proposed start of exercise A in Q2 2020
- Additional exercise leads on each exercise  
*Bianca Szkuta (Deakin University) on exercise A*
- Participants will be invited for each exercise separately  
*opt-in for each exercise, no commitment for full series!*
- Invite goes out to EDNAP liased labs, ENFSI and AUS/NZ  
*Depending on number of committed labs, possibly extend*



## Feedback on exercises A and B and C and D

### **Some issues to consider**

- Are suggested items relevant? Other suggestions?
- Is experimental outline feasible?
- What number of iterations would be acceptable? (n=?)
- What number of participant laboratories should we expect/facilitate?



# Results of two ancestry exercises (2017, 2018) with participation of 11 European forensic labs

Roewer L<sup>1\*</sup>, Ballard D<sup>2</sup>, Syndercome-Court D<sup>2</sup>, Ansell R<sup>3</sup>, Bogus M<sup>4</sup>, Eduardoff M<sup>5</sup>, Freire-Aradas A<sup>6</sup>, Geppert M<sup>1</sup>, Groß T<sup>4</sup>, Haas C<sup>7</sup>, Lessig R<sup>8</sup>, Kayser M<sup>9</sup>, de Knijff P<sup>10</sup>, Lutz-Bonengel S<sup>11</sup>, Morling N<sup>12</sup>, Nagy M<sup>1</sup>, Parson W<sup>5</sup>, Phillips C<sup>6</sup>, Ralf A<sup>9</sup>, Rothe J<sup>1</sup>, Schmidt U<sup>11</sup>, Schneider PM<sup>4</sup>, Smidt-Mogensen H<sup>12</sup>, Tillmar AO<sup>13</sup>, Xavier C<sup>5</sup>, Immel U<sup>8</sup>

<sup>1</sup> Institut für Rechtsmedizin und Forensische Wissenschaften, Charité – Universitätsmedizin Berlin, Germany

<sup>2</sup> Department of Analytical, Environmental and Forensic Sciences, King's College London, UK

<sup>3</sup> Swedish National Laboratory of Forensic Science, Linköping, Sweden

<sup>4</sup> Institut für Rechtsmedizin, Universität Köln, Germany

<sup>5</sup> Institut für Gerichtliche Medizin, Medizinische Universität Innsbruck, Austria

<sup>6</sup> Institute of Forensic Sciences, University of Santiago de Compostela, Spain

<sup>7</sup> Forensische Genetik, Institut für Rechtsmedizin, Universität Zürich, Switzerland

<sup>8</sup> Institut für Rechtsmedizin, Universitätsklinikum Halle, Germany

<sup>9</sup> Department of Genetic Identification, Erasmus MC University Medical Center Rotterdam, The Netherlands

<sup>10</sup> Department of Human Genetics, Leiden University Medical Center, The Netherlands

<sup>11</sup> Institut für Rechtsmedizin, Universitätsklinikum Freiburg, Germany

<sup>12</sup> Section Forensic Genetics, Dept. Forensic Medicine, University of Copenhagen, Denmark

<sup>13</sup> National Board of Forensic Medicine, Linköping, Sweden



Universität  
Zürich<sup>UZH</sup>



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Medizinische Fakultät  
der Martin-Luther-Universität  
Halle-Wittenberg

# Design of exercises 2017 and 2018

- **Invited European** forensic labs who have prepared at least one “ancestry report” for the national police
- **Eleven** institutes participated in the two trials (10 university, 1 police)
- **Eight** countries
- **Deadline** 8 weeks
- Full reports in **customary** form and wording
- All reports are **circulated** between participants
- **Validation and publication (e.g. at HM2018)**

# Exercise 2017 (8 participants)

- Unknown male extracted DNA (blind sample)
- Person born in Nigeria,
- Belongs to Igbo ethnic group (ca. 20 Mio.)
- Language: Igbo (a Bantu language)
- Father/Mother: Igbo



# Results Exercise 2017

(stated in the „Summary“ of the reports)

**Male persons**  
**maternal/paternal ancestry is**

Subsaharan Africa	5/8
Africa	2/8
Without statement on ancestry	1/8

# Exercise 2018 (11 participants)

## Swab „I“

- Male person, born in Germany
- Mother: born in Germany
- Father: born in Nepal

## Swab „D“

- Male person, born in the Philippines (language: Tagalog)
- Language: Tagalog, Language Group: Malay
- Mother: born in the Philippines
- Father: born in the Philippines

# Results Exercise 2018 (1)

(taken from „Conclusion“)

The unknown male person „I“  
is of **admixed ancestry**, and **paternal ancestry is**

Nepal

<b>Indian subcontinent (with mentioning „Nepal“)</b>	<b>1/11</b>
<b>India, Iran</b>	<b>1/11</b>
<b>South Asia</b>	<b>1/11</b>
<b>Asia (with mentioning „Nepal“)</b>	<b>1/11</b>
East Asia	1/11
Eurasia	1/11
South-, Central-, Southwest Asia or Caucasus	1/11
Southern Europe/Middle East	1/11
Middle East	1/11
<b>Without statement on ancestry</b>	<b>2/11</b>

**and maternally**

Germany

<b>Western Eurasia</b>	<b>2/11</b>
<b>Eurasia</b>	<b>2/11</b>
Central South Asia or Europe	1/11
Middle East	1/11
<b>Without statement on ancestry</b>	<b>5/11</b>

# Results Exercise 2018 (2)

(taken from „Conclusion“)

The unknown male person's „D“  
paternal ancestry is

Philippines

Malay Archipelago, Southeast Asian mainland, East Asia (with mentioning „Philippines“)	1/11
Southeast Asia (with mentioning „Philippines“)	1/11
Southeast Asia, East Asia (with mentioning „Philippines“)	1/11
East Asia (2x with mentioning „Philippines“)	4/11
East Asia, Northeast-, South Asia (with mentioning „Philippines“)	1/11
Asia (1 x with mentioning „Philippines“)	3/11
Without statement on ancestry	0/11

and maternally

Philippines

Malay Archipelago	1/11
Insular Southeast Asia	2/11
Maritime Southeast Asia	1/11
Southeast Asia	4/11
East Asia	3/11
Without statement on ancestry	0/11



# Number of employed methods per lab

Institute	Exercise 2017	Exercise 2018
Berlin	2	3
Cologne	3	3
Freiburg	2	2
Innsbruck	5	3
Leiden	4	3
Linköping	3	2
Rotterdam	3	3
Santiago	3	3
London	-	3
Zurich	-	4
Copenhagen	-	1

# Most frequently employed methods

Method	Exercise 2017	Exercise 2018
mtDNA	6	9
Y-STRs	6	9
Y-SNPs	5	4
AIMs	5	8
Autosomal STRs	2	0
<b>Participants</b>	<b>8</b>	<b>11</b>

# Conclusions

## (verbal statements in the reports)

### Strong

- Test sample **is from** a donor who is ...
- Test sample is more than **a billion times more likely** ... than, so results in summary indicate a ... ancestry
- Test sample **most likely is from** a donor who is ...
- **Most probable** ancestry is..., also possible is..., but less probable..., ... ancestry is not excluded, but highly unlikely
- A ...descent of the ancestors of the sample donor is **most plausible**
- **Results point towards** a ... origin of the direct biological ancestors
- Give **some support** for the statement that the donors biogeographical ancestry is from a ... population, compared with other possible origins
- The detected DNA **belongs to** ... which is **mainly observed in...**

### Weak

# Questionnaire

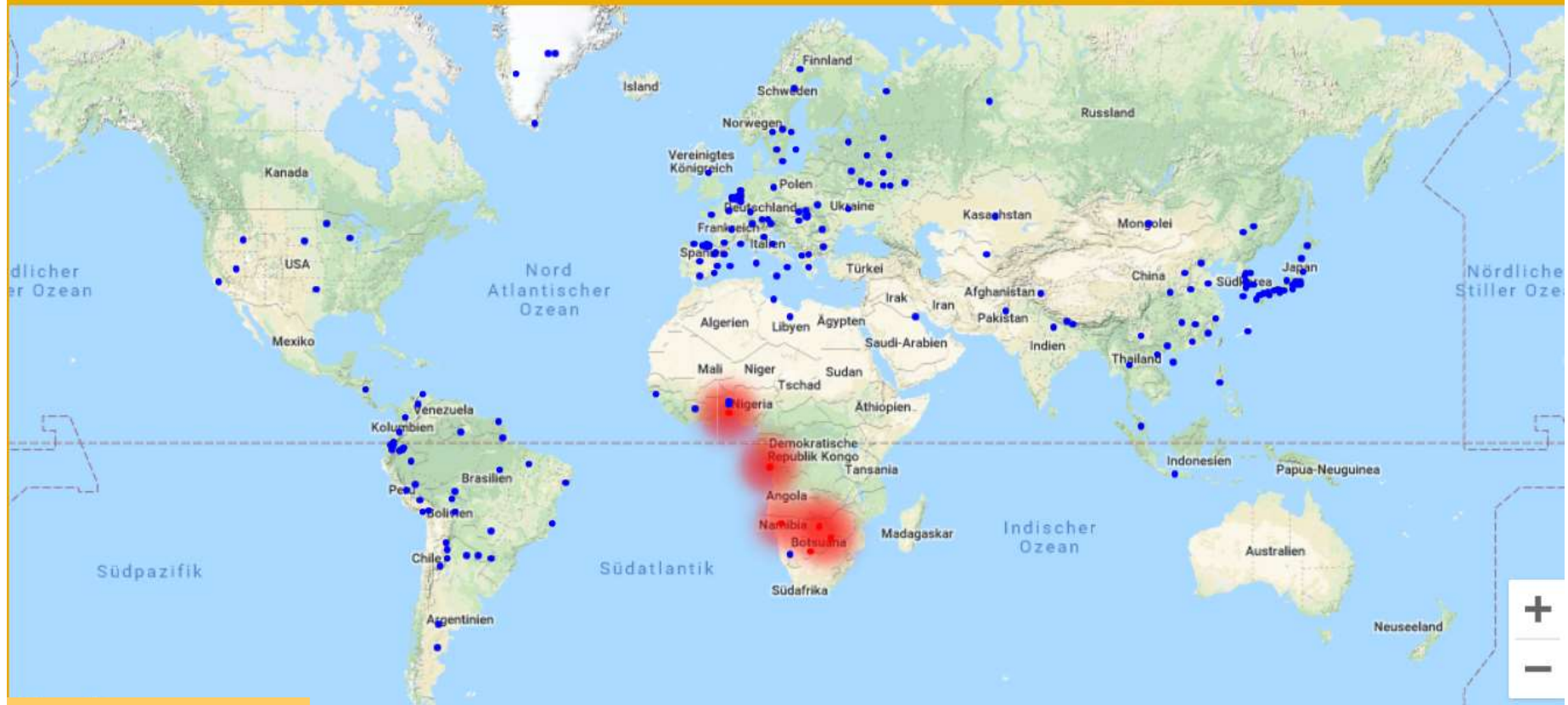
- **How much time does it take in practice?**
  - 1-3 weeks
- **Costs?**
  - 200-250 € / sample
- **How often ancestry is ordered by law enforcement?**
  - 1-10 x per year (for identification)

# Exercise 2017, sample 1 (Method Y-SNP, Ref. YHRD)

## Y-SNP Branch Information on B2a

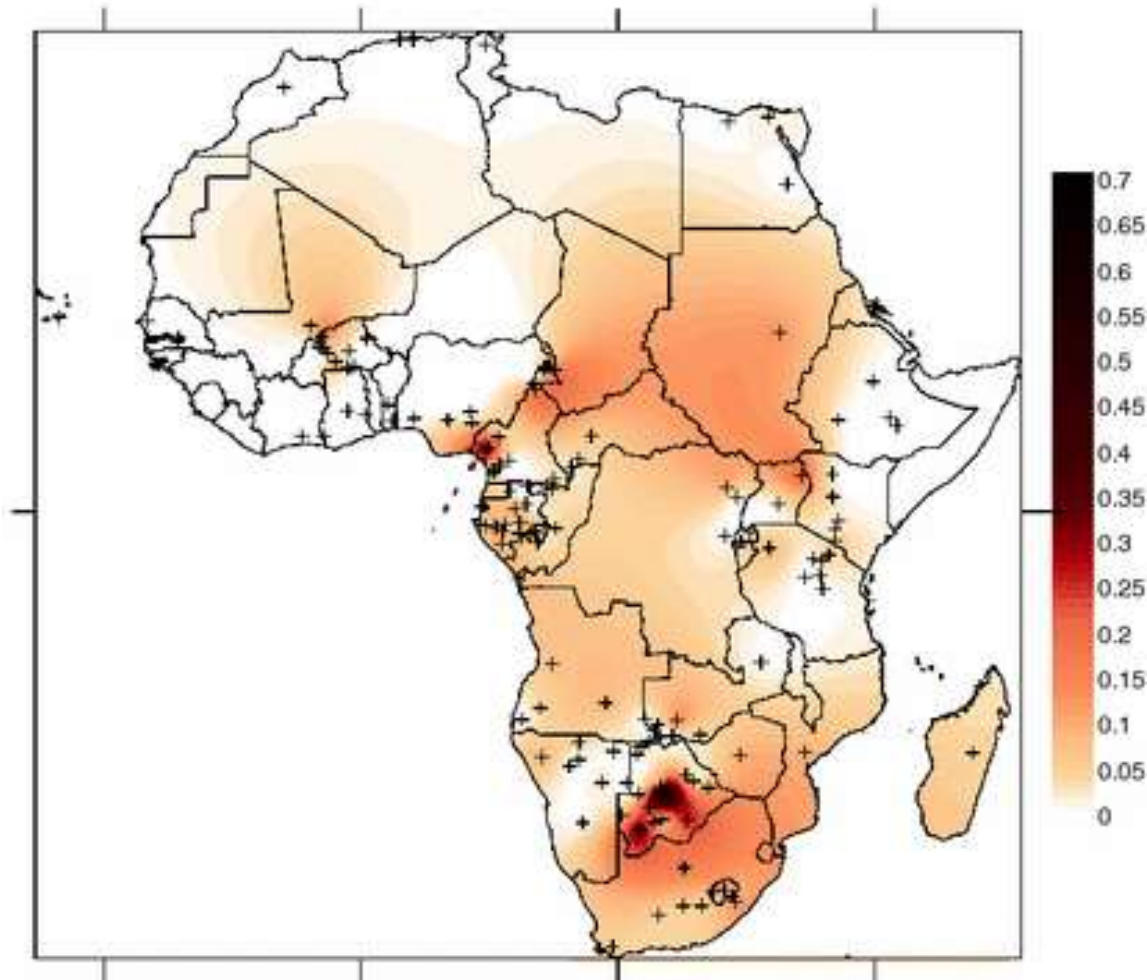
The Y-SNP branch B2a is defined by M150. Additionally, all downstream markers G1, M109, M152, M218, P32, Page60, V197, V227, V62, V75, V78 are defining this branch as well. For further information on a marker, please see [marker details at Phylotree](#) or consult the [Y-SNP tree](#) there.

Please note, that colours on the map only reflect a haplogroup distribution from the SNP-typed samples which were submitted to the YHRD.



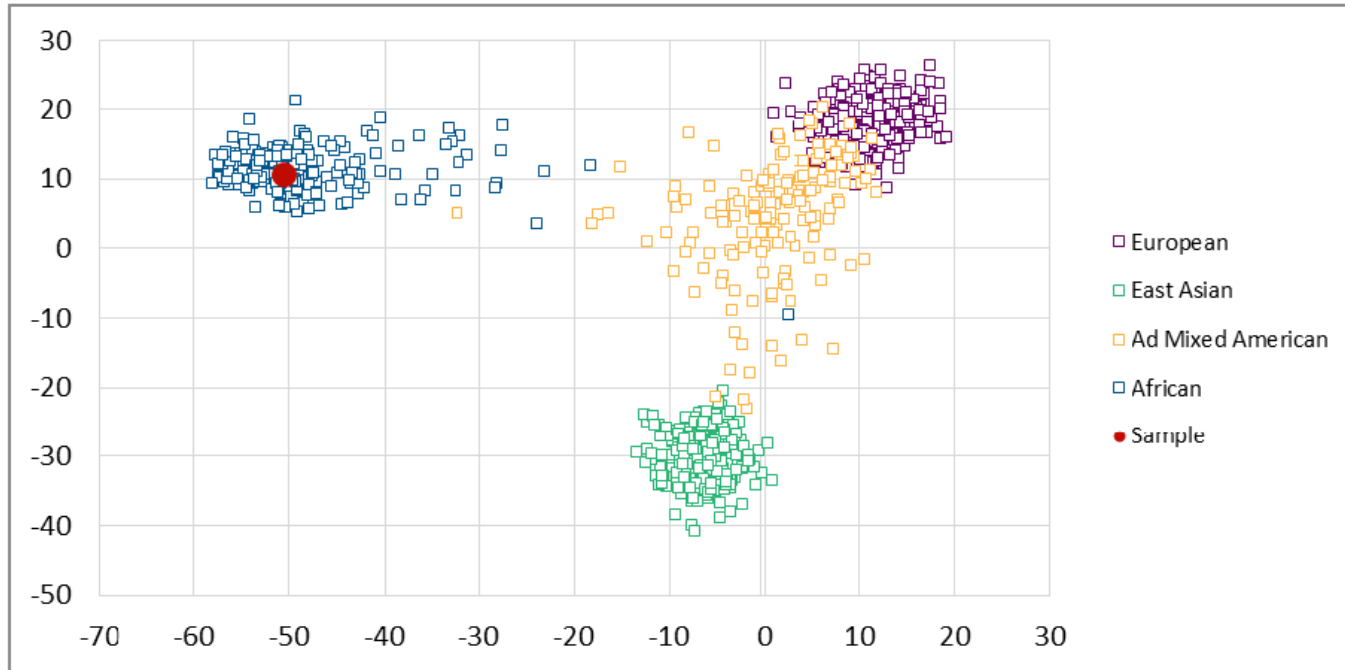
<https://yhrd.org>

# Exercise 2017, sample 1 (Method Y-SNP, Ref. literature)



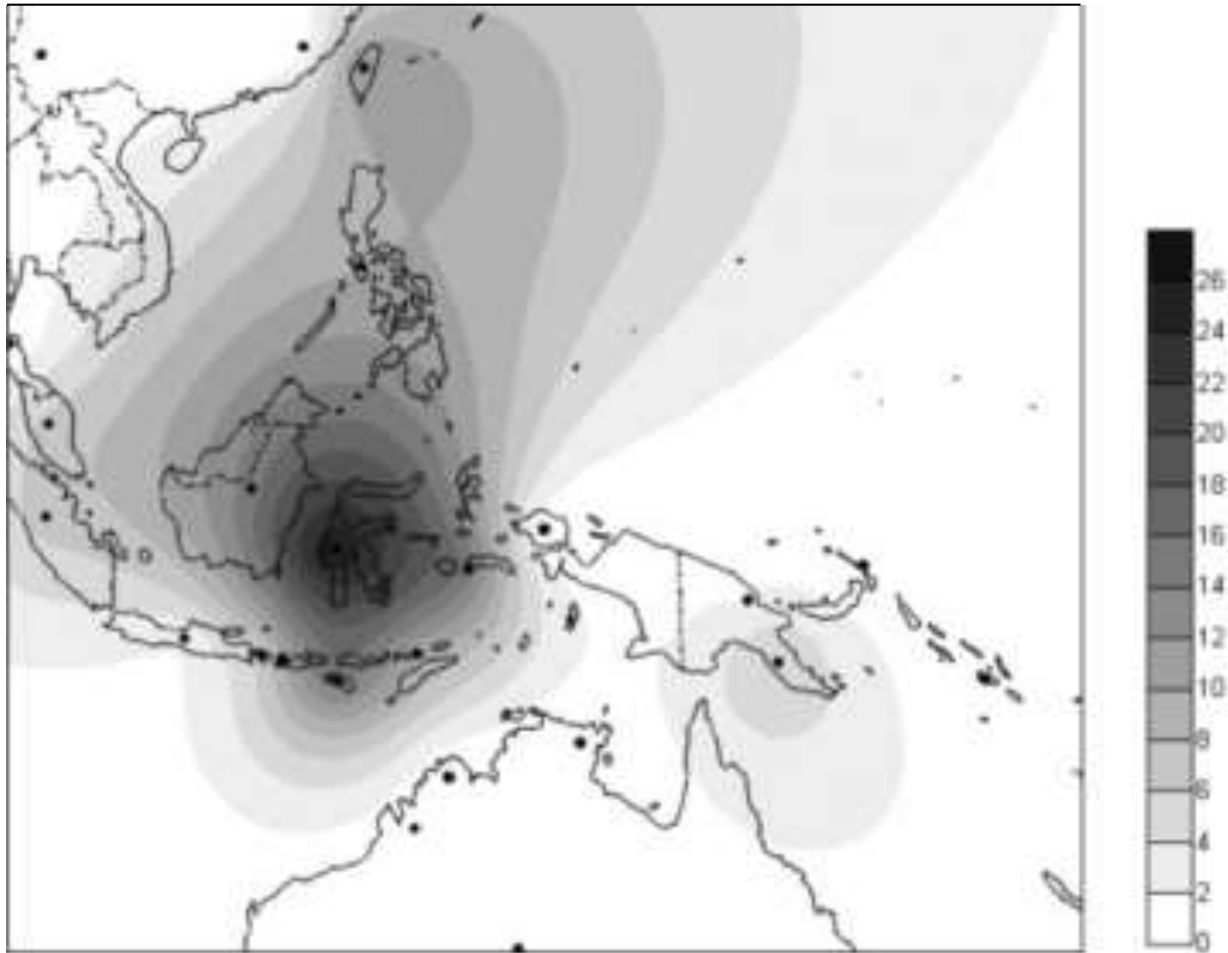
Barbieri C, Hübner A, Macholdt E, Ni S, Lippold S, Schröder R, Mpoloka SW, Purps J, Roewer L, Stoneking M, Pakendorf B (2016) Refining the Y chromosome phylogeny with southern African sequences. *Human Genetics* 135(5):541-53

# Exercise 2017, sample 1 (Method AIMs, ref. Verogen)



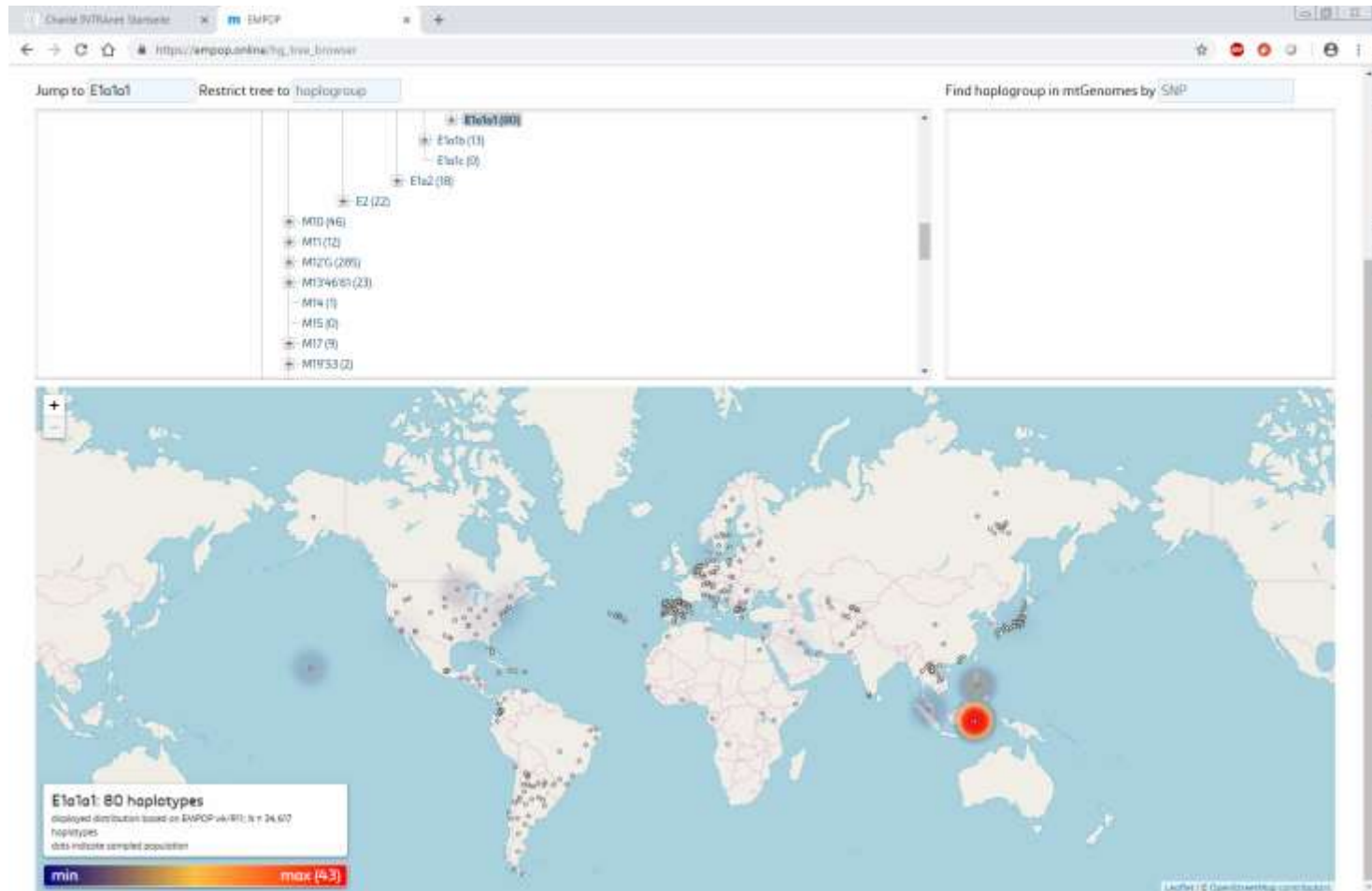
ForenSeq DNA Signature Prep Kit (Verogen) - 56 AIMs-DNA marker

## Exercise 2018, sample D (Method mtDNA, ref. literature)





# Exercise 2, sample D (method mtDNA, ref. EMPOP)



# Summary

- Ancestry (not residency) can be narrowed down using a combination of DNA methods “(Ancestry package”)
- In Germany for the **identification of unknowns** (StPO §88)
- For Identification of unknown suspects, mostly in “**cold cases**” - Legislative process is pending in several countries, including Germany
- Approved and regulated e.g. in the Netherlands
- **Blind tests, exercises and exchange** – this is currently the way to build expertise

Welcome note

General information

Preliminary programme

Registration and accommodation

About Budapest



YHRD.ORG

EMP20P  
YEARS



**HIFS**

HUNGARIAN INSTITUTE  
FOR FORENSIC SCIENCES



# 12<sup>th</sup> Haploid Markers 2020

May 13-16, 2020 | Budapest, Hungary

249 days 10 hours 34 minutes 18 seconds

ONLINE REGISTRATION

ABSTRACT SUBMISSION

[www.hm2020.hu](http://www.hm2020.hu)

# **Biogeographical Ancestry: Current Status and Way Forward**

# The 2014 EDNAP ancestry exercise



Bratislava, 2013

Forensic ancestry analysis with two capillary electrophoresis ancestry informative marker (AIM) panels: Results of a collaborative EDNAP exercise



C. Santos<sup>a,1</sup>, M. Fondevila<sup>a,1</sup>, D. Ballard<sup>b,1</sup>, R. Banemann<sup>c</sup>, A.M. Bento<sup>d</sup>, C. Børsting<sup>e,1</sup>, W. Branicki<sup>f,2</sup>, F. Brisighelli<sup>g</sup>, M. Burrington<sup>h</sup>, T. Capal<sup>i</sup>, L. Chaitanya<sup>j</sup>, R. Daniel<sup>k</sup>, V. Decroyer<sup>l</sup>, R. England<sup>m</sup>, K.B. Gettings<sup>n</sup>, T.E. Gross<sup>o,1</sup>, C. Haas<sup>p</sup>, J. Hartevelde<sup>q</sup>, P. Hoff-Olsen<sup>r</sup>, A. Hoffmann<sup>c</sup>, M. Kayser<sup>j</sup>, P. Kohler<sup>r,2</sup>, A. Linacre<sup>s</sup>, M. Mayr-Eduardoff<sup>t,1</sup>, C. McGovern<sup>m</sup>, N. Morling<sup>e,l,1</sup>, G. O'Donnell<sup>h</sup>, W. Parson<sup>t,u</sup>, V.L. Pascali<sup>g</sup>, M.J. Porto<sup>d</sup>, A. Roseth<sup>r</sup>, P.M. Schneider<sup>o,1</sup>, T. Sijen<sup>q</sup>, V. Stenzl<sup>i</sup>, D. Syndercombe Court<sup>b,1</sup>, J.E. Templeton<sup>s</sup>, M. Turanska<sup>v</sup>, P.M. Vallone<sup>n</sup>, R.A.H. van Oorschot<sup>k</sup>, L. Zatkalikova<sup>v</sup>, The EUROFORGEN-NoE Consortium, Á. Carracedo<sup>a,1</sup>, C. Phillips<sup>a,1,\*</sup>



# The 2014 EDNAP ancestry exercise



## Bratislava, 2013

Forensic ancestry analysis with two capillary electrophoresis ancestry informative marker (AIM) panels: Results of a collaborative EDNAP exercise



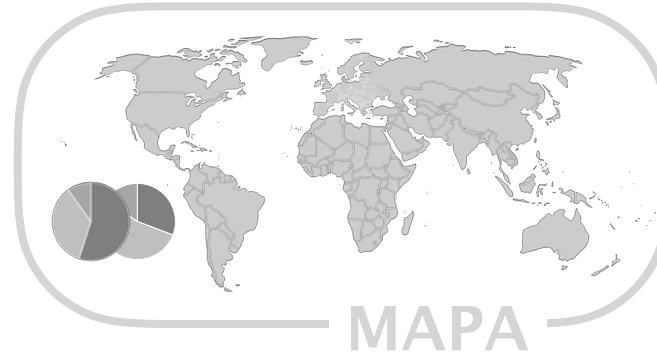
C. Santos<sup>a,1</sup>, M. Fondevila<sup>a,1</sup>, D. Ballard<sup>b,1</sup>, R. Banemann<sup>c</sup>, A.M. Bento<sup>d</sup>, C. Børsting<sup>e,1</sup>, W. Branicki<sup>f,2</sup>, F. Brisighelli<sup>g</sup>, M. Burrington<sup>h</sup>, T. Capal<sup>i</sup>, L. Chaitanya<sup>j</sup>, R. Daniel<sup>k</sup>, V. Decroyer<sup>l</sup>, R. England<sup>m</sup>, K.B. Gettings<sup>n</sup>, T.E. Gross<sup>o,1</sup>, C. Haas<sup>p</sup>, J. Hartevelde<sup>q</sup>, P. Hoff-Olsen<sup>r</sup>, A. Hoffmann<sup>c</sup>, M. Kayser<sup>j</sup>, P. Kohler<sup>r,2</sup>, A. Linacre<sup>s</sup>, M. Mayr-Eduardoff<sup>t,1</sup>, C. McGovern<sup>m</sup>, N. Morling<sup>e,1,1</sup>, G. O'Donnell<sup>h</sup>, W. Parson<sup>t,u</sup>, V.L. Pascali<sup>g</sup>, M.J. Porto<sup>d</sup>, A. Roseth<sup>r</sup>, P.M. Schneider<sup>o,1</sup>, T. Sijen<sup>q</sup>, V. Stenzl<sup>i</sup>, D. Syndercombe Court<sup>b,1</sup>, J.E. Templeton<sup>s</sup>, M. Turanska<sup>v</sup>, P.M. Vallone<sup>n</sup>, R.A.H. van Oorschot<sup>k</sup>, L. Zatkalikova<sup>v</sup>, The EUROFORGEN-NoE Consortium, Á. Carracedo<sup>a,1</sup>, C. Phillips<sup>a,1,\*</sup>

Focused on reliability and ease-of-use of CE-based tests for 34 SNPs and 46 Indels (for mixed DNA); use of *Snipper* for PCA and Bayes LR

In the MPS era, CE plays an important role in starting labs off into ancestry analysis, or where resources don't allow MPS (e.g. Ethiopia-Italy)

The ForenSeq MPS test has 55 AIMs  $\equiv$  80 CE SNPs/Indels in power

# MPS allows much larger SNP numbers



**Global AIMS:**  
127 SNPs  
(6 tri-allelic)

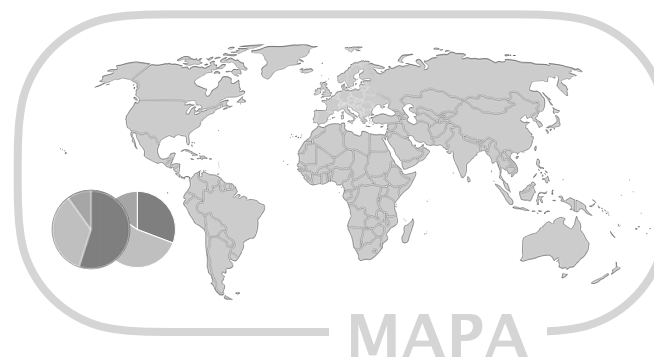


**MAPlex:**  
144 SNPs  
(28 tri-allelic)  
(2 tetra-allelic)  
20 microhaplotypes



**Basic tool:**  
113 ancestry SNPs  
(15 tri-allelic)  
38 EVC SNPs

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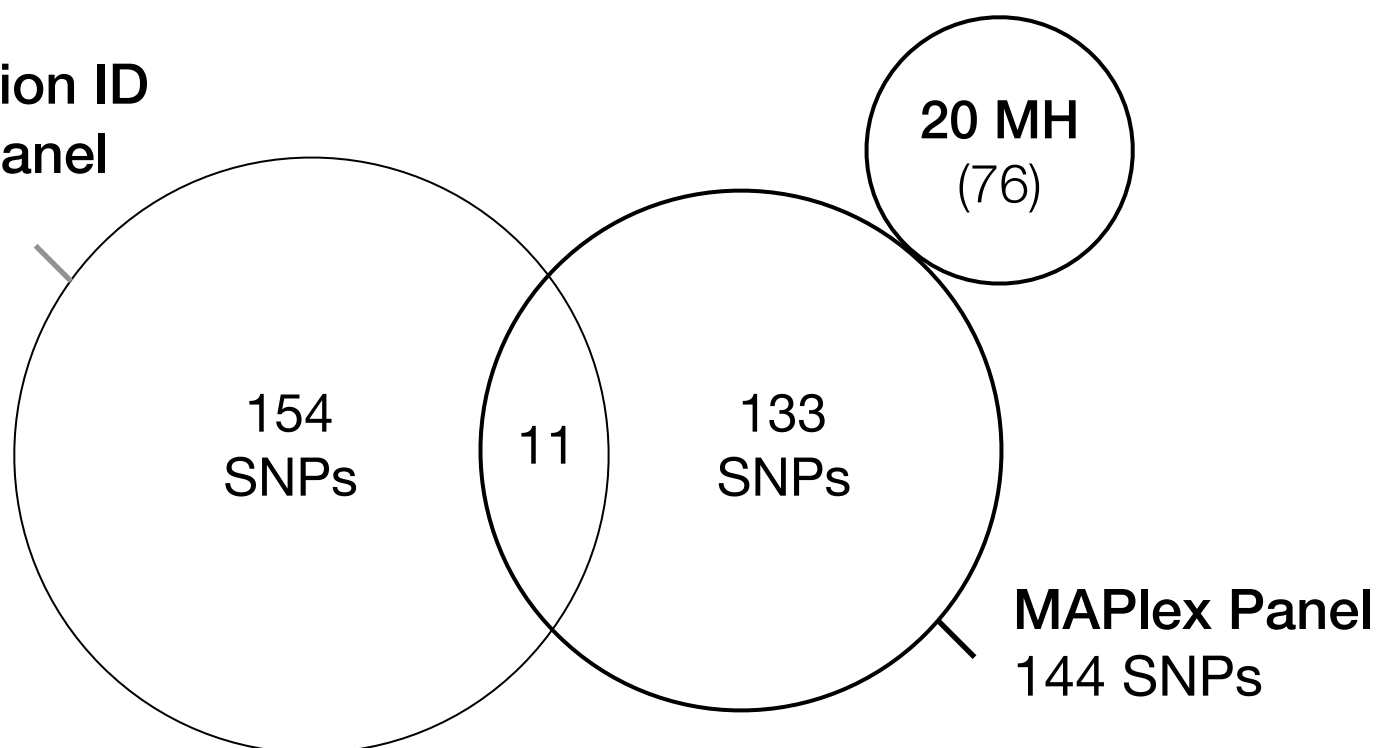


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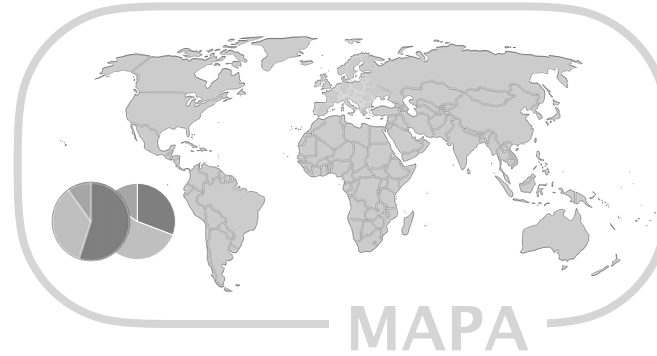
**TFS Precision ID  
Ancestry Panel**  
165 SNPs



2 MPS multiplexes: 318 ancestry markers



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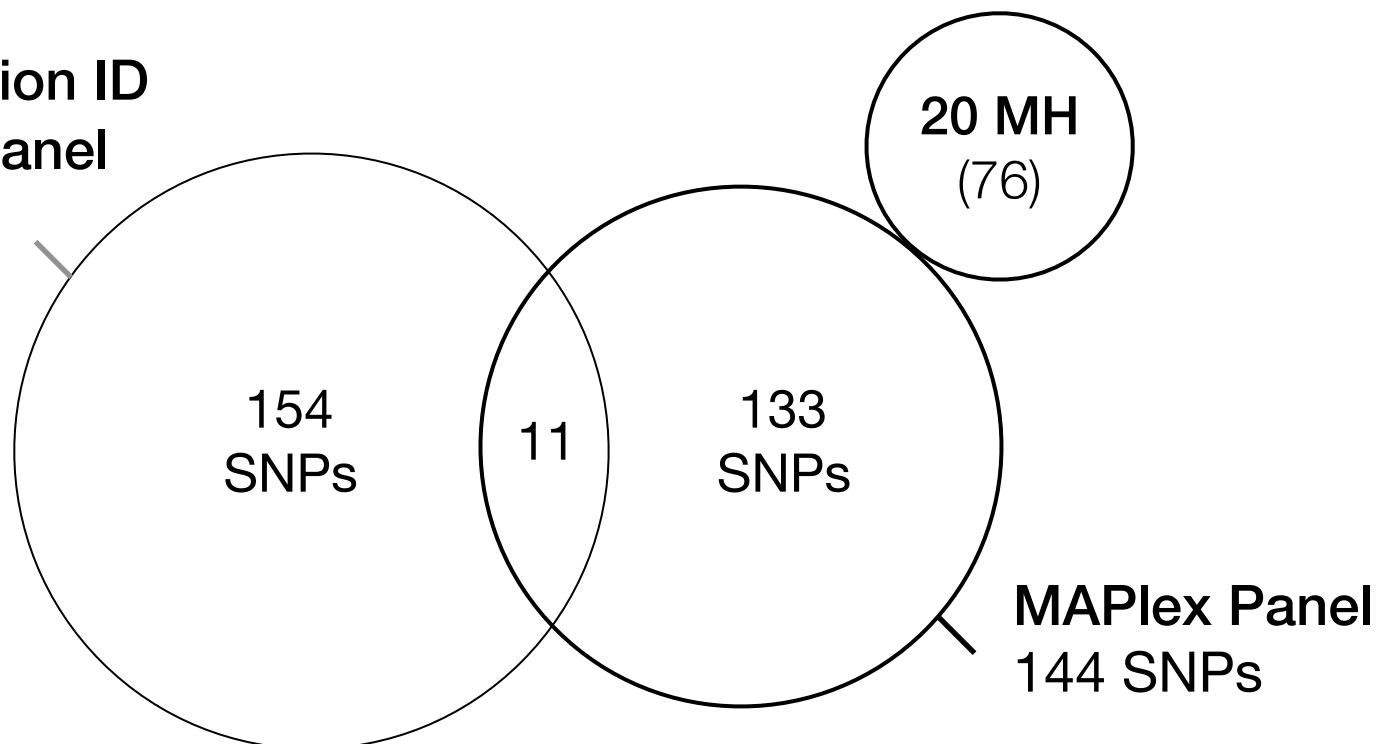


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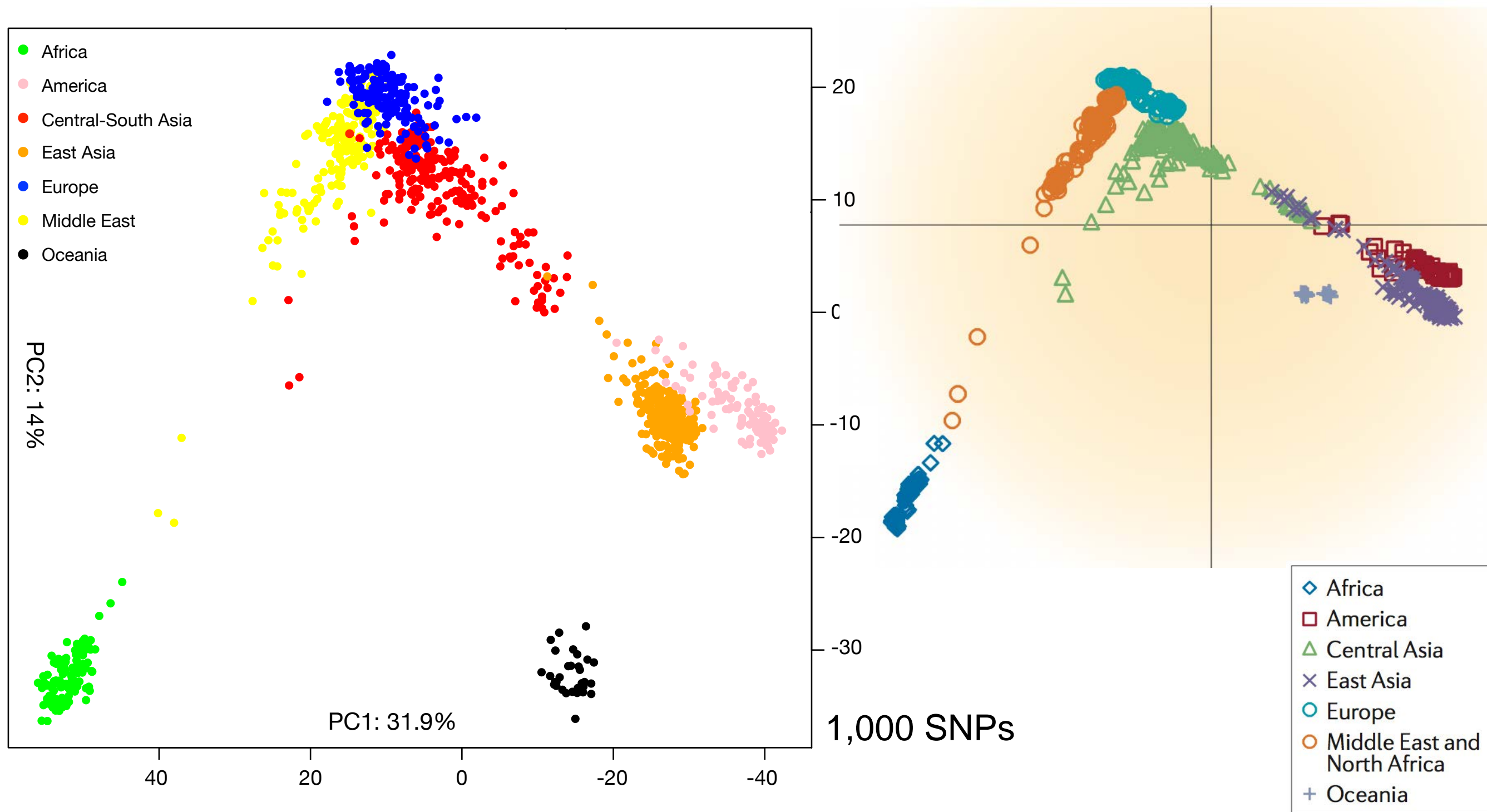
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**Enhanced tool:**  
205 ancestry SNPs  
(26 tri-allelic)  
194 EVC SNPs  
21 microhaplotypes

2 MPS multiplexes: 318 ancestry markers

# Eventually adding enough SNPs provides equivalent power to much larger marker densities



**But there are several major issues that are not necessarily fixed by adding more SNPs**

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**Most labs tend to 'think locally' and want forensic tests to provide fine scale differentiation of very closely related populations**

But there are several major issues that are not necessarily fixed by adding more SNPs

Most labs tend to 'think locally' and want forensic tests to provide fine scale differentiation of very closely related populations



A nested approach can focus on local needs

*Japaneseplex*: A forensic SNP assay for identification of Japanese people using Japanese-specific alleles

Isao Yuasa<sup>a,\*</sup>, Atsushi Akane<sup>b</sup>, Toshimichi Yamamoto<sup>c</sup>, Aya Matsusue<sup>d</sup>, Minoru Endoh<sup>a</sup>, Mayumi Nakagawa<sup>e</sup>, Kazuo Umetsu<sup>f</sup>, Takaki Ishikawa<sup>g</sup>, Morio Iino<sup>a</sup>



But there are several major issues that are not necessarily fixed by adding more SNPs

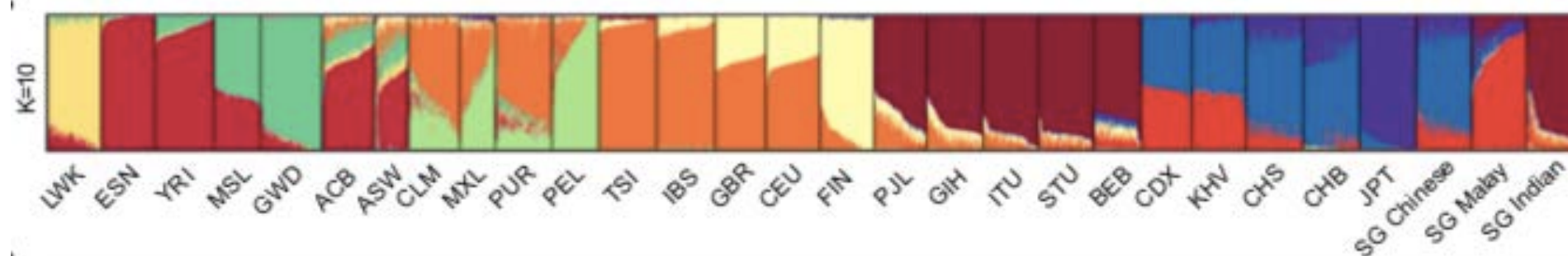
Most labs tend to 'think locally' and want forensic tests to provide fine scale differentiation of very closely related populations



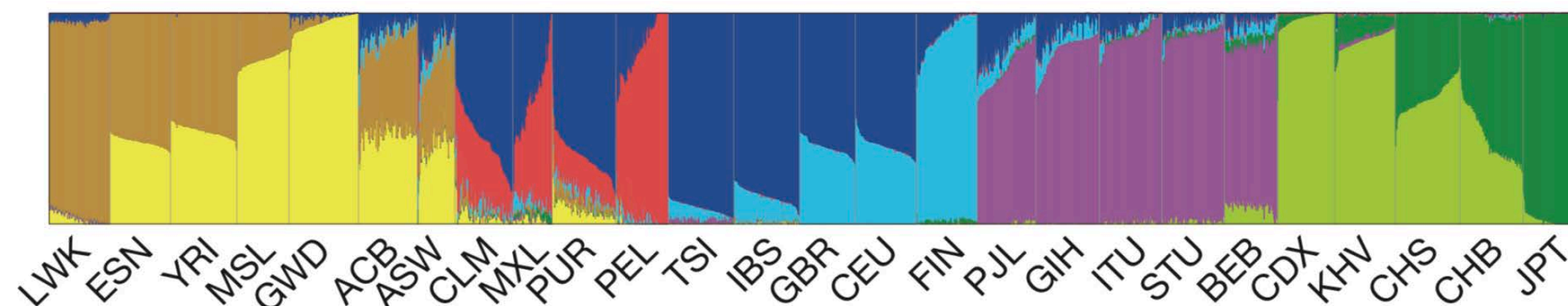
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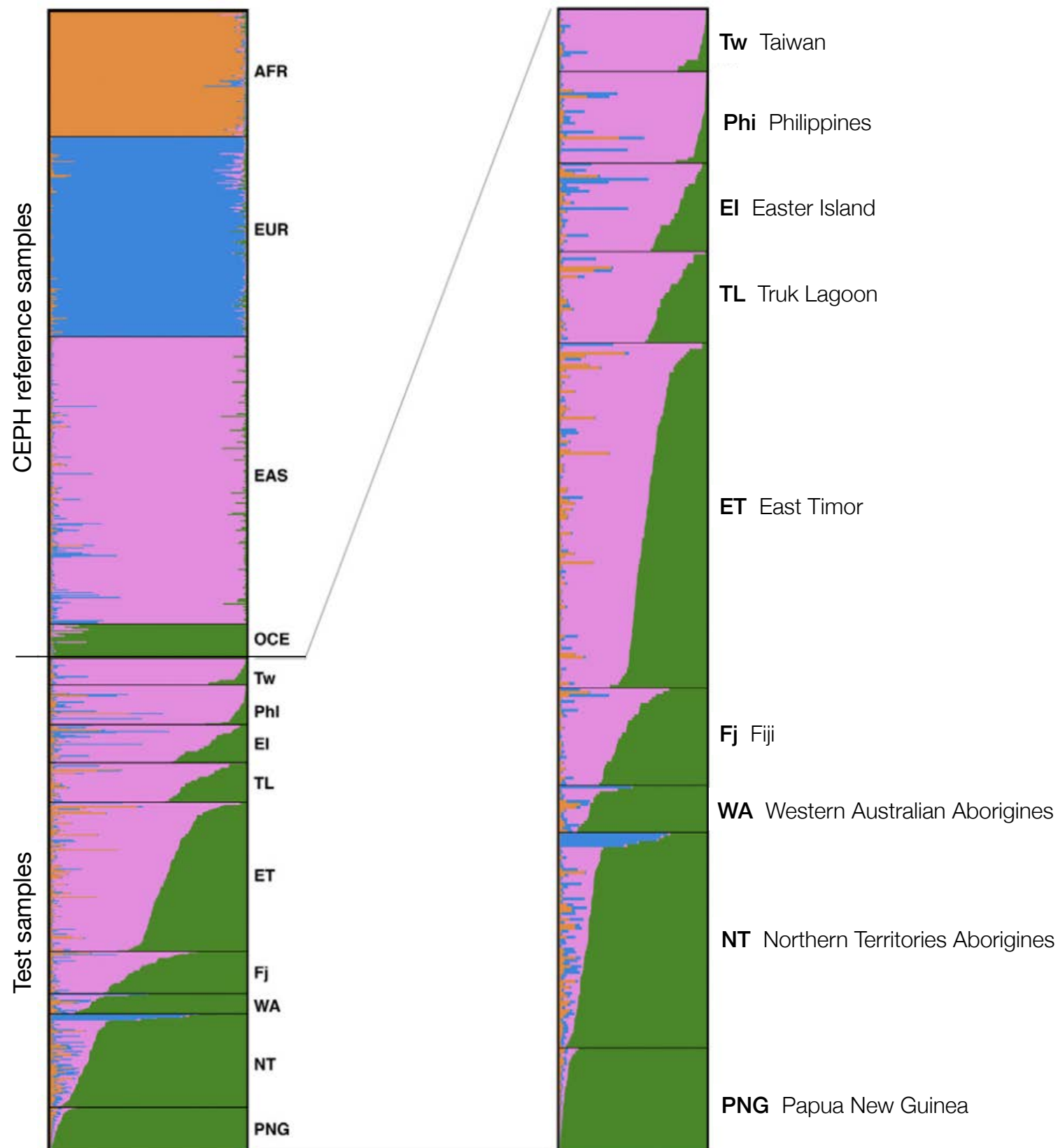
Singapore Genomes



1000 Genomes Phase III



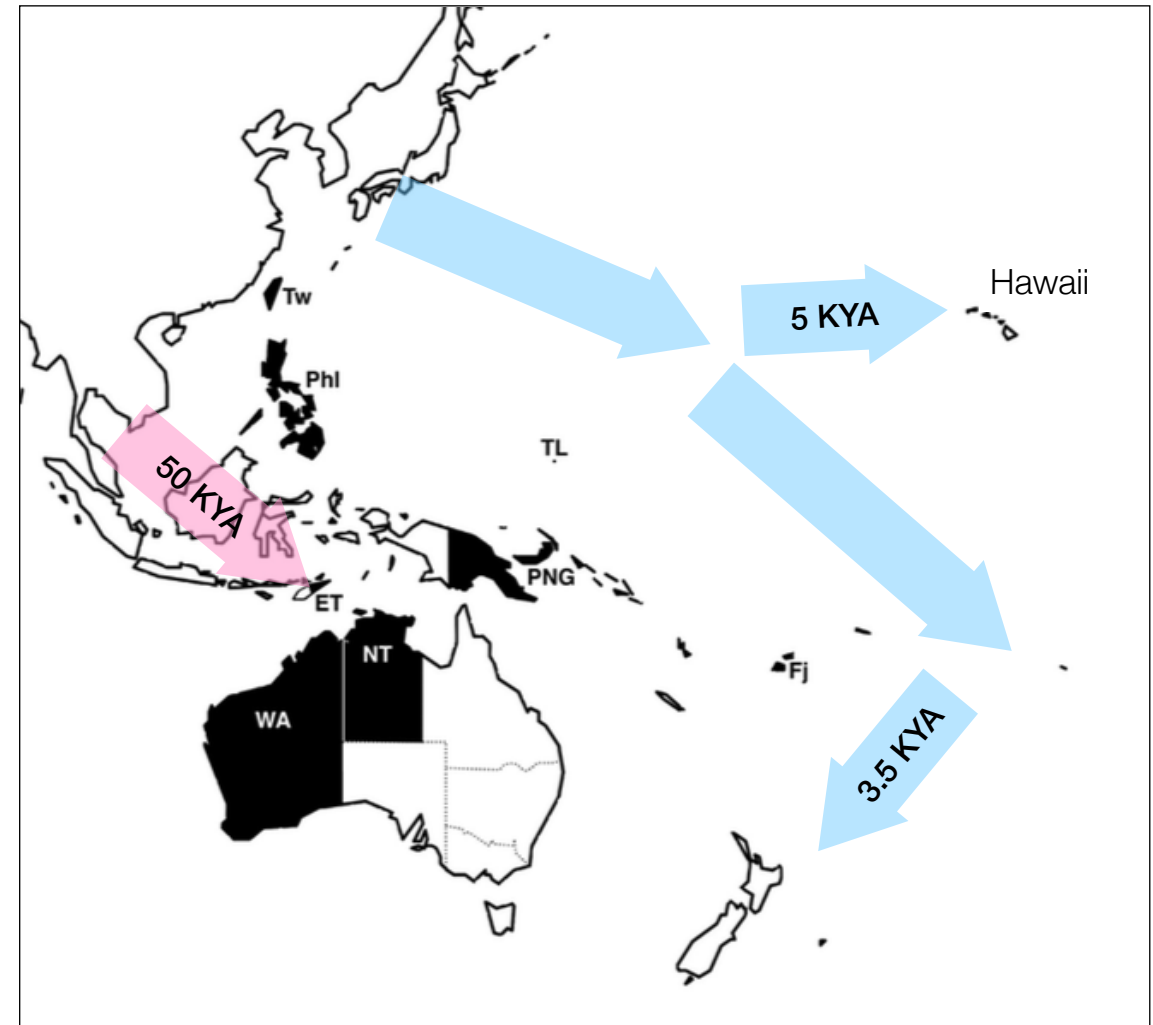
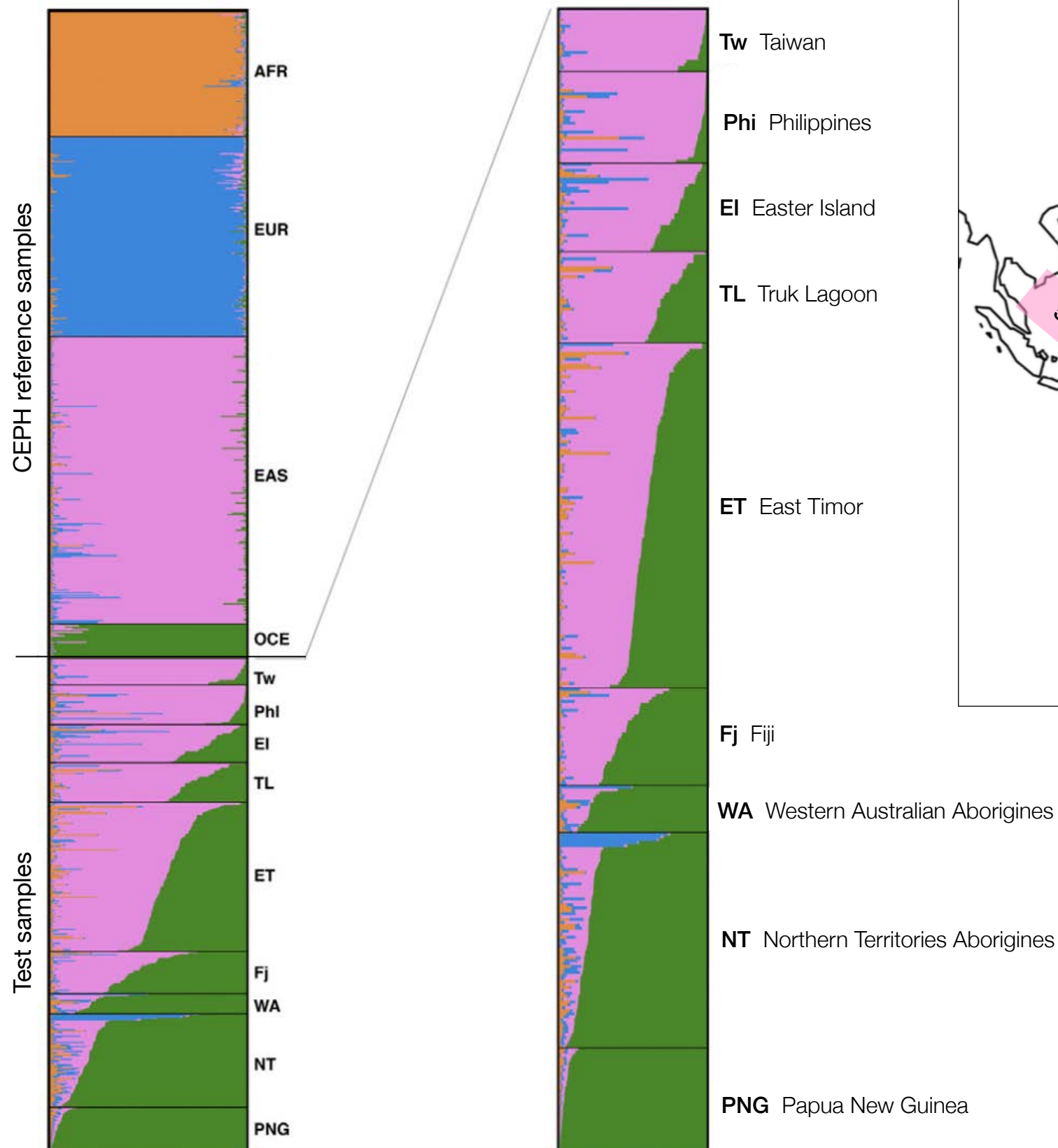
# *Pacifiplex*: devised for Australian local needs



- Do CEPH panel Papuans make a good proxy for Native Australians?



# Pacifiplex: population genetics can inform patterns



- Multiple migrations at different times
- Multiple Denisovan introgression with early Sahul-region migrants
- Do CEPH panel Papuans make a good proxy for Native Australians?
- What about the rest of the Pacific?

**But there are several major issues that are not necessarily fixed by adding more SNPs**

**Most labs tend to 'think locally' and want forensic tests to provide fine scale differentiation of very closely related populations**

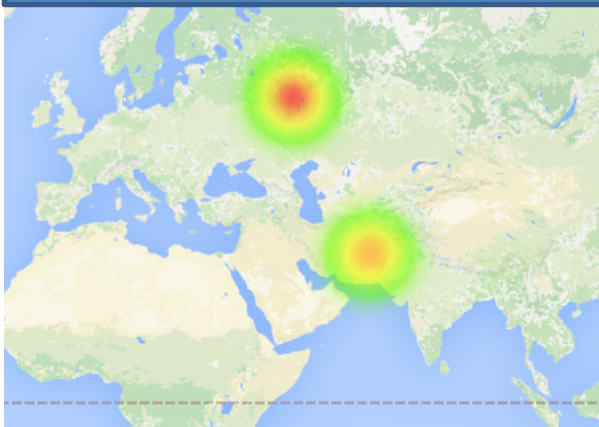
**Populations with admixture are indistinguishable from populations located at continental margins**

But there are several major issues that are not necessarily fixed by adding more SNPs

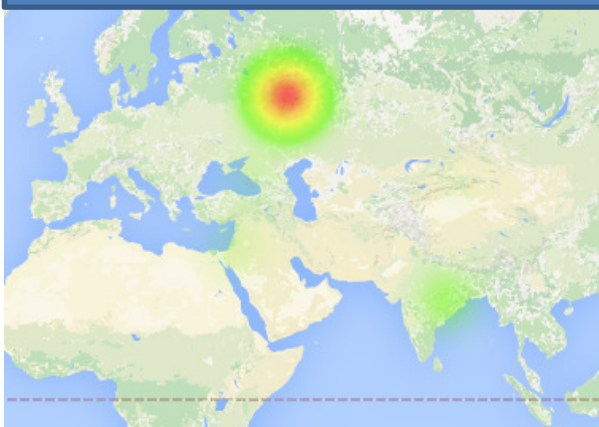
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Populations with admixture are indistinguishable from populations located at continental margins

PGM – Kidd 55

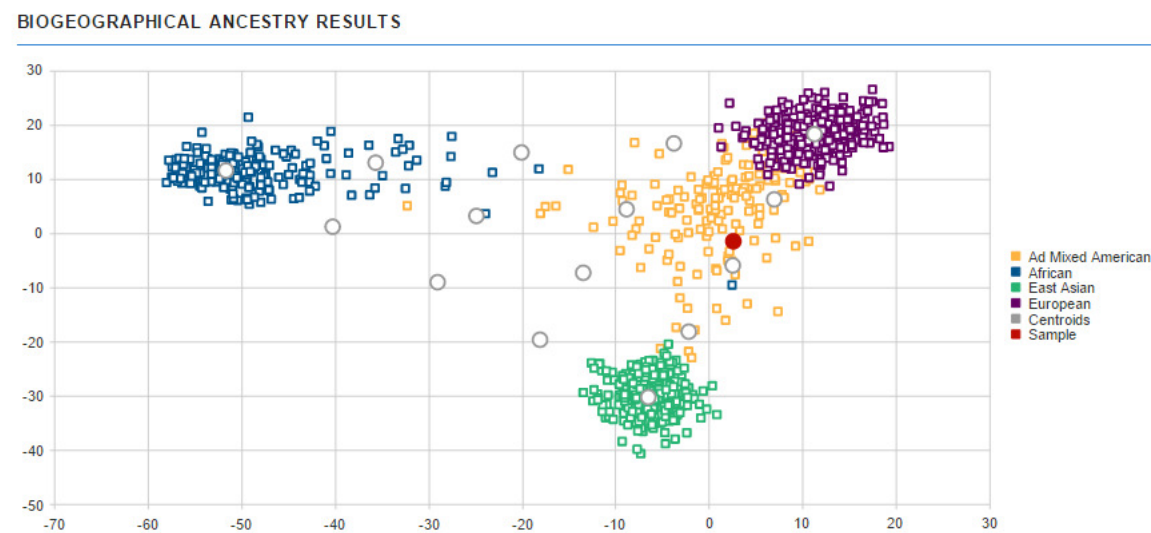


PGM – Seldin 128



Half European –  
Half East Asian Individual

ForenSeq – Kidd 55

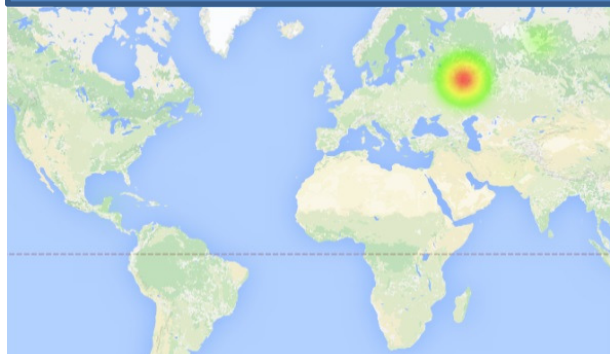


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PGM – Kidd 55



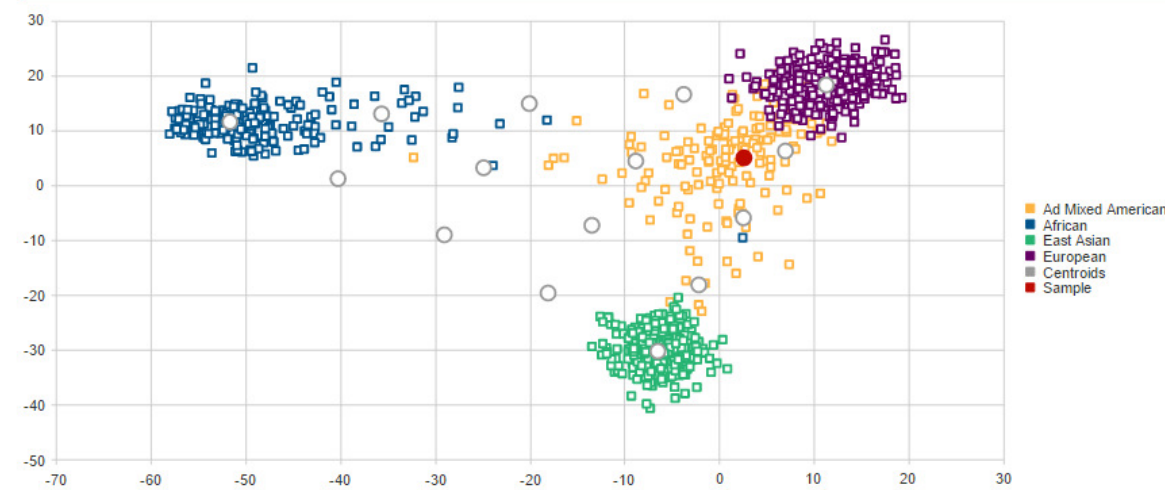
PGM – Seldin 128



Hispanic Individual

ForenSeq – Kidd 55

BIOGEOGRAPHICAL ANCESTRY RESULTS



Here the SNPs lack power to differentiate American variation effectively so the geographic imprecision becomes worse

# But there are several major issues that are not necessarily fixed by adding more SNPs

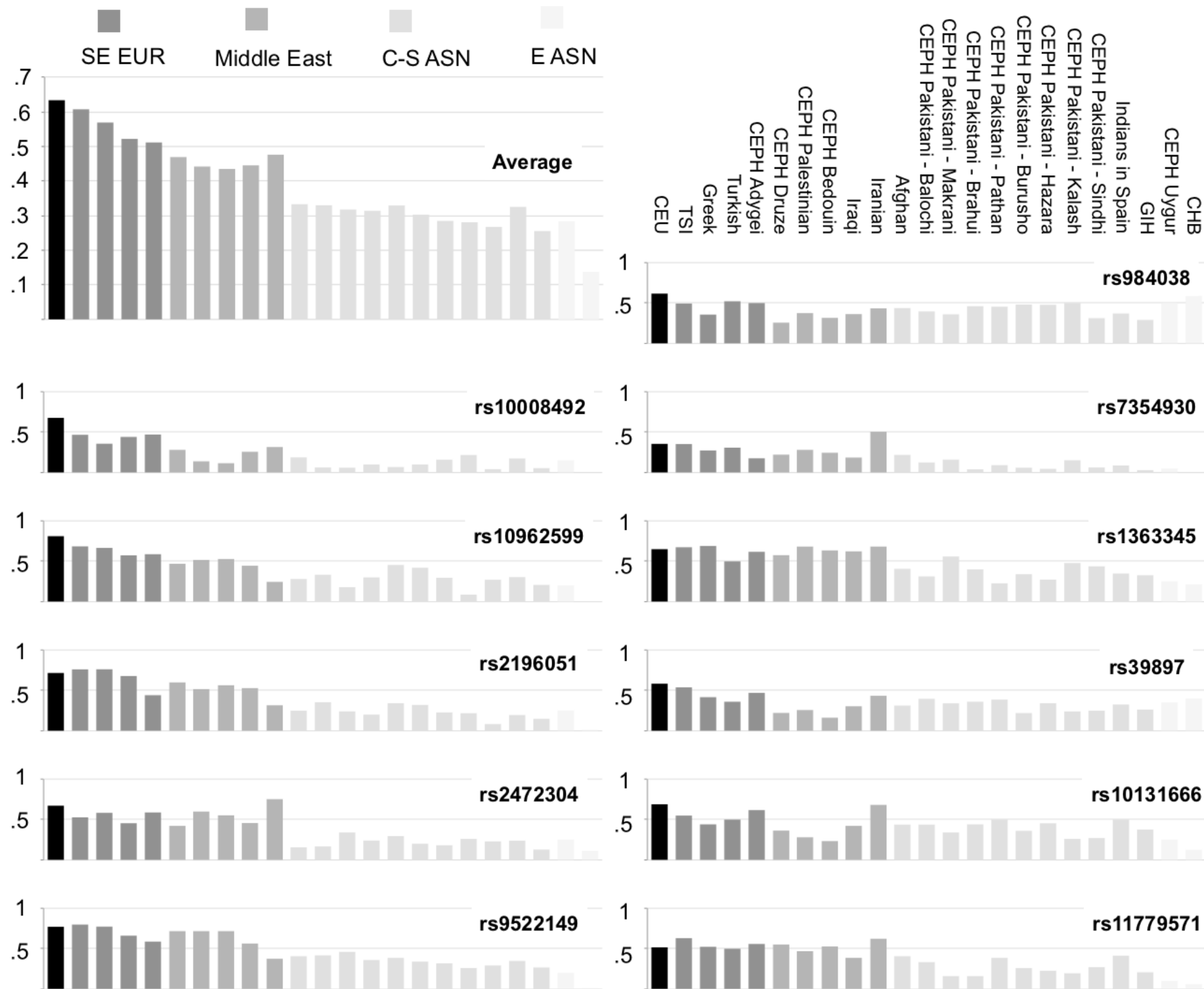
Most labs tend to 'think locally' and want forensic tests to provide fine scale differentiation of very closely related populations

Populations with admixture are indistinguishable from populations located at continental margins

Without geographic barriers, many of the world's populations form frequency clines - so apart from the ends, differentiation is limited

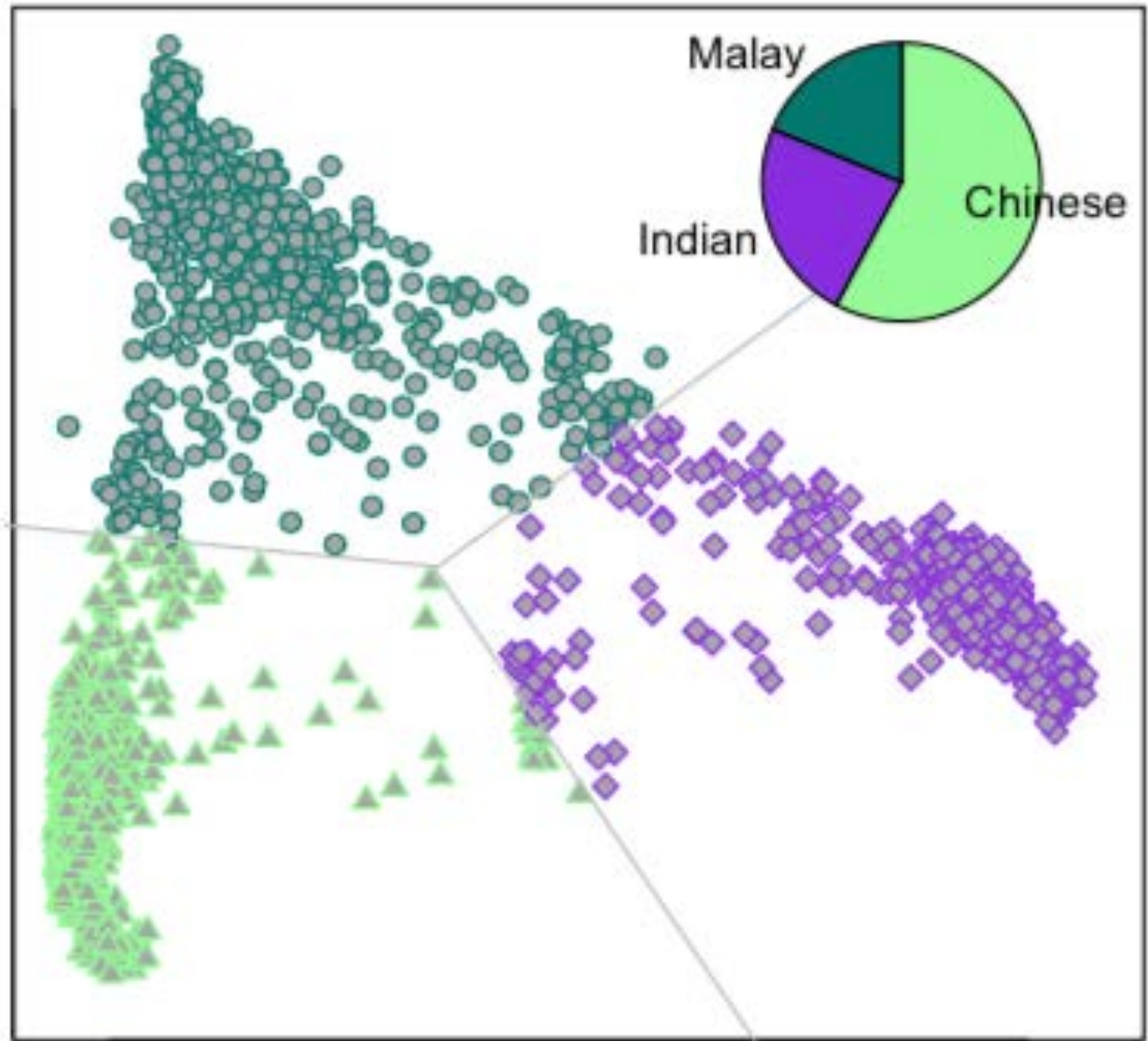
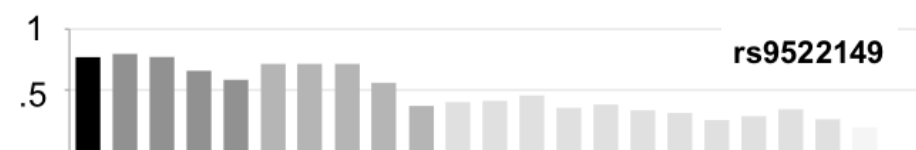


# Frequency clines 'blur the edges' between populations



Eurasiaplex

# Frequency clines 'blur the edges' between populations



Eurasiaplex



# But there are several major issues that are not necessarily fixed by adding more SNPs

Most labs tend to 'think locally' and want forensic tests to provide fine scale differentiation of very closely related populations

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It is increasingly difficult to obtain population samples that can fill gaps in geography

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North Native American

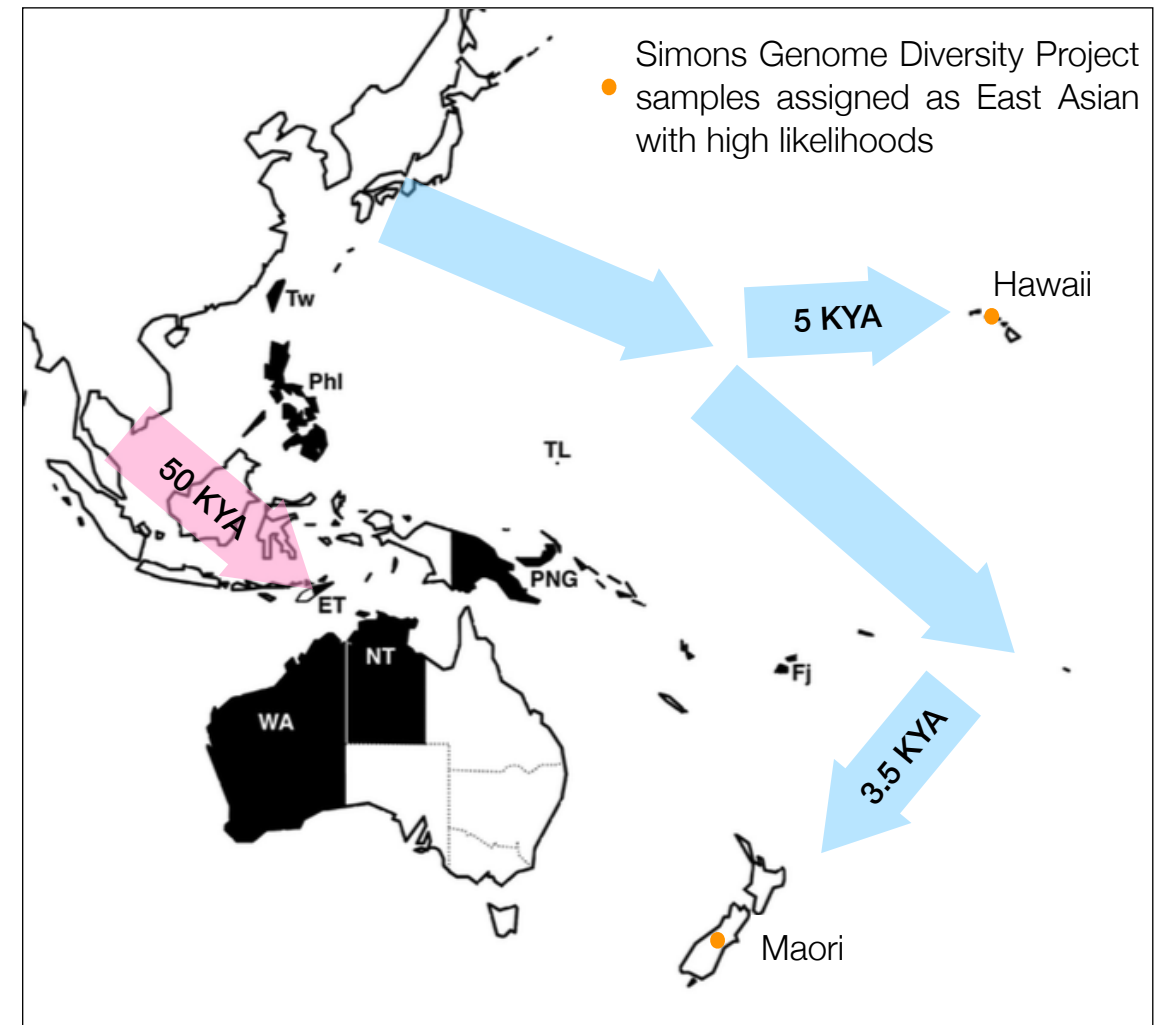
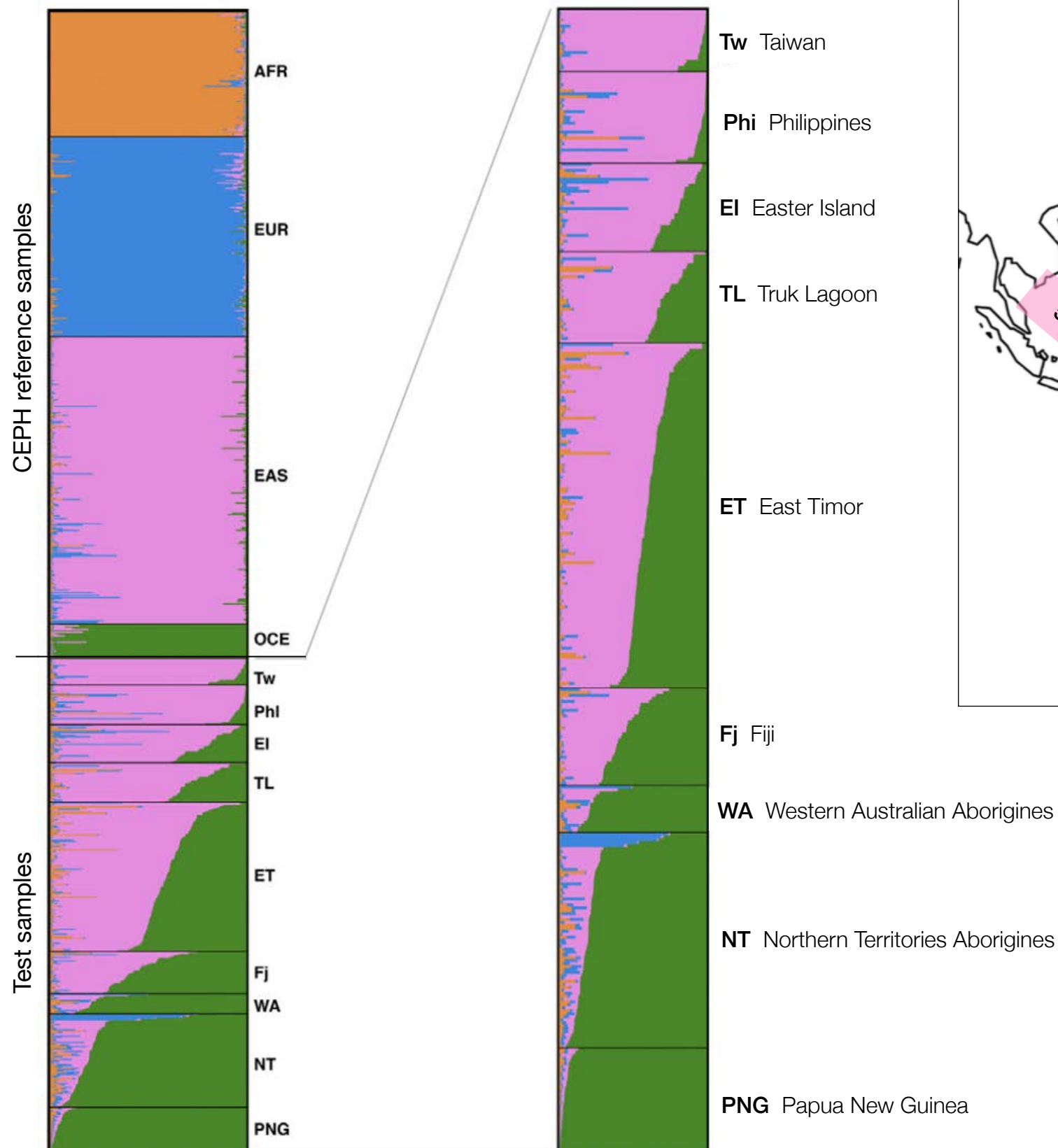
East African

East European

Far Oceania

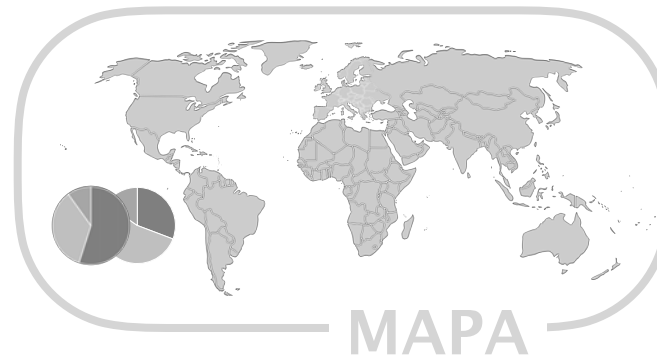
Roma/Jewish

# Pacifiplex: sampling problem for NZ and Hawaii



- Multiple migrations at different times
- Multiple Denisovan introgression with early Sahul-region migrants
- Do CEPH panel Papuans make a good proxy for Native Australians?
- What about the rest of the Pacific?

# Admixture: towards combining marker types in one test



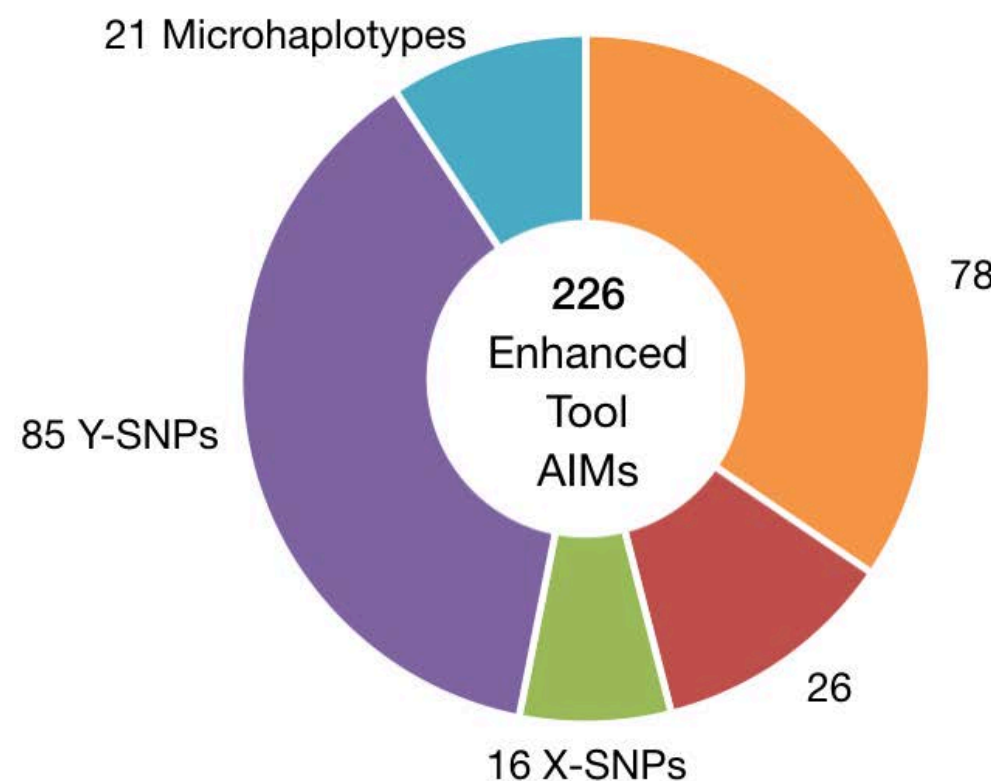
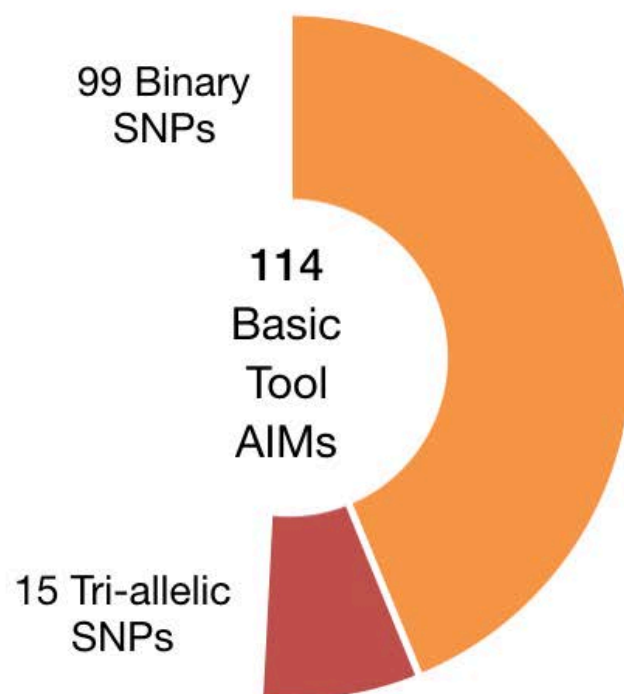
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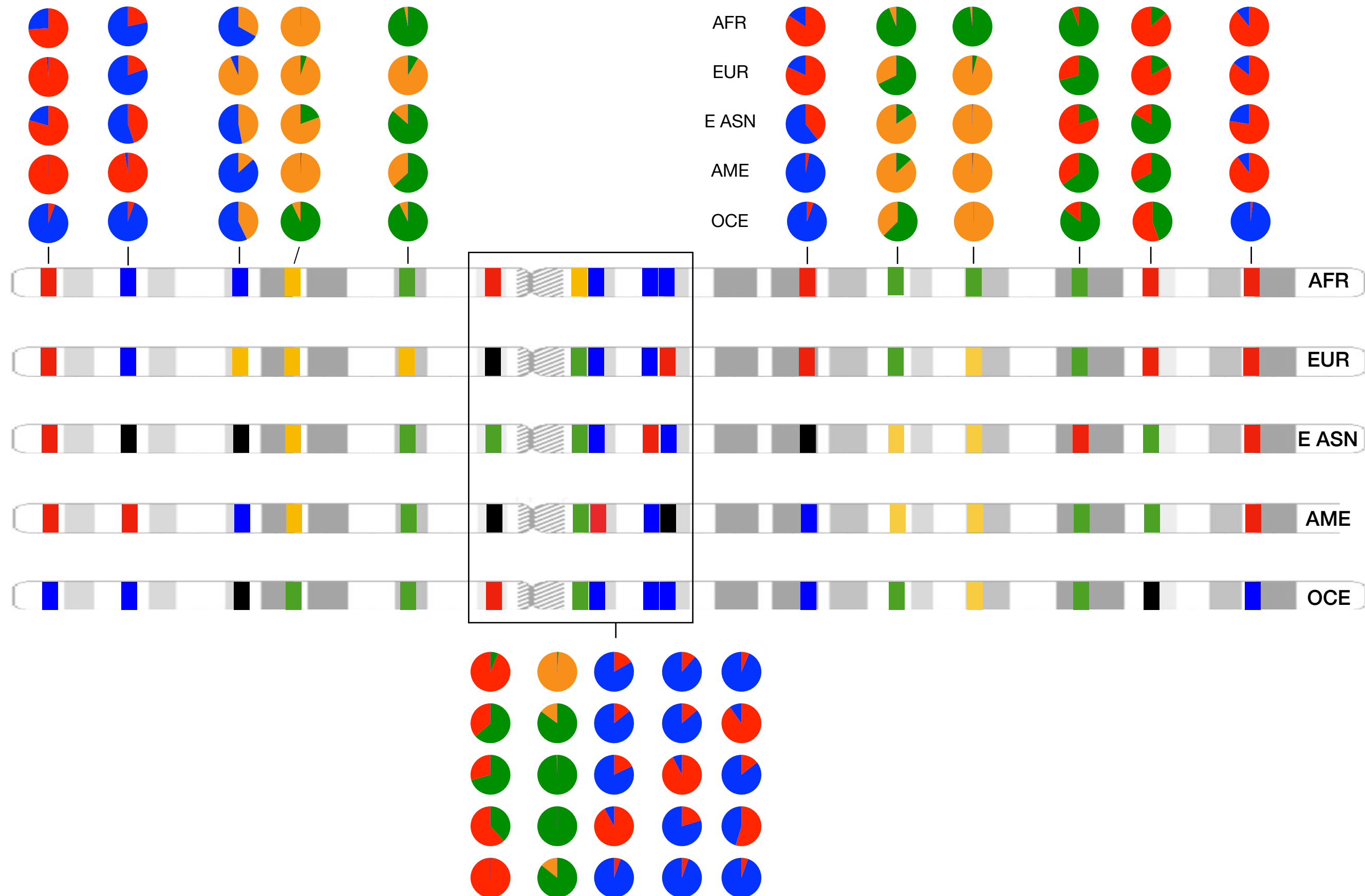
**Multiplex size tripled**

**Mix of autosomal markers**

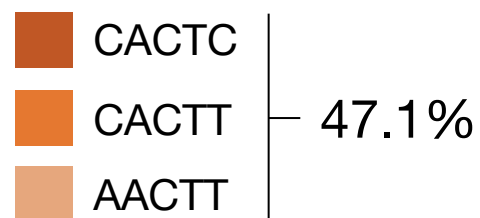
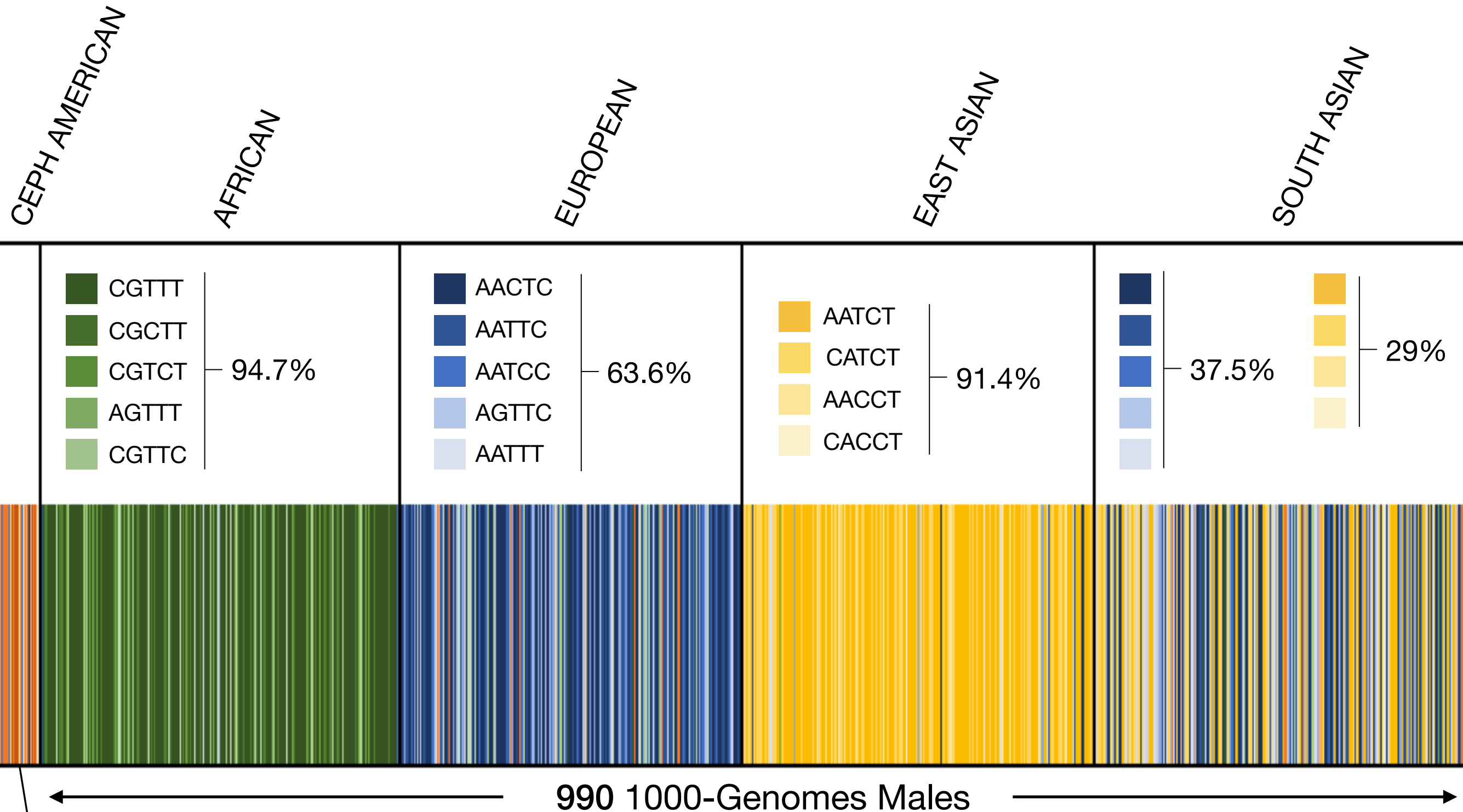
**SNP<sup>2</sup> : SNP<sup>3</sup> : MHs**

**AXY combinations**

# Admixture: combining X-SNPs and an X-haplotype



# The X-haplotype alone efficiently defines X ancestry





# Statistical approaches to admixture

- GenoGeographer assesses z-scores
- Genetic Distance Analysis can be as efficient as **STRUCTURE** and runs in real time - so it can flag data that needs more detailed testing

Generation % Admixture Ratio	STRUCTURE	GDA	% difference with true value
2 <sup>nd</sup> 1×1 50:50	2.0	12.7	
3 <sup>rd</sup> 1:1×1:1 50:50	3.4	12.4	
3 <sup>rd</sup> 1×1:1 75:25	3.3	10.0	
3 <sup>rd</sup> 1×1:1 50:25:25	4.8	9.0	
3 <sup>rd</sup> 1:1×1:1 50:25:25	5.4	8.5	
3 <sup>rd</sup> 1:1×1:1 25:25:25:25	7.9	6.5	

- HID SNP Genotyper ancestry module is also robust for many test samples analysed so far - the 10,000-step simulated admixture test creates a realistic range of likelihoods



Forensic Science International:  
Genetics

Available online 17 October 2019, 102154

In Press, Journal Pre-proof



Research paper

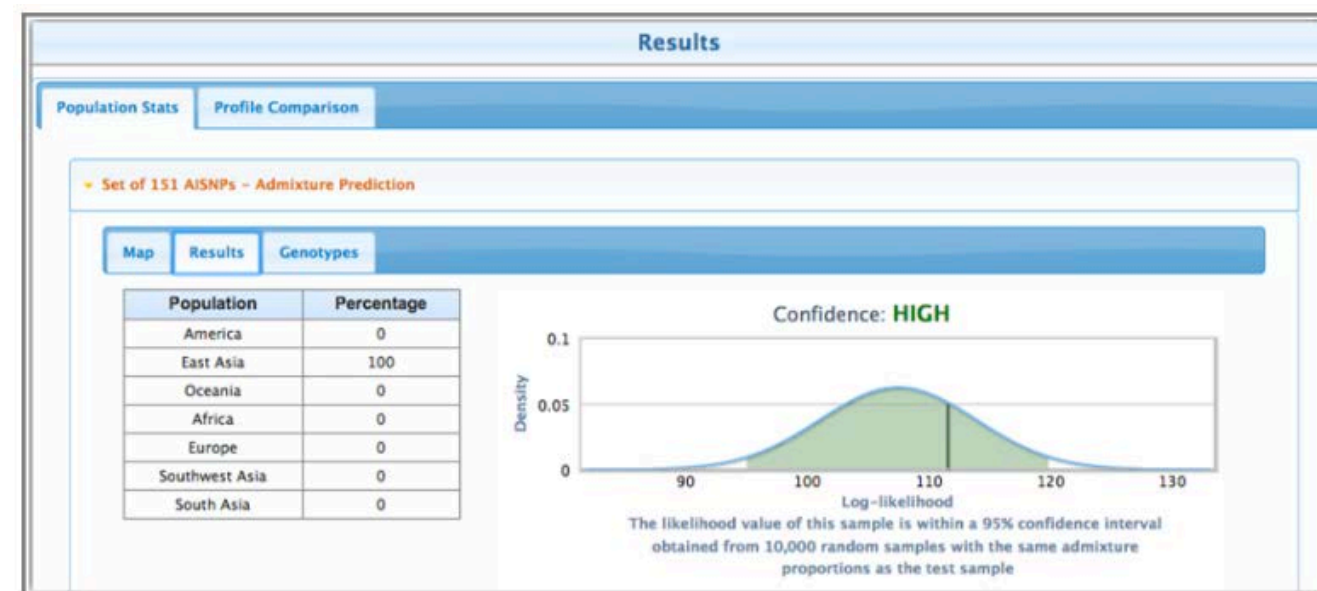
Ancestry prediction efficiency of the software *GenoGeographer* using a z-score method and the ancestry informative markers in the Precision ID Ancestry Panel

Int J Legal Med (2017) 131:901–912  
DOI 10.1007/s00414-016-1504-3

ORIGINAL ARTICLE

## Prediction of biogeographical ancestry from genotype: a comparison of classifiers

Elaine Y Y Cheung<sup>1</sup> • Michelle Elizabeth Gahan<sup>1</sup> • Dennis McNevin<sup>1</sup>

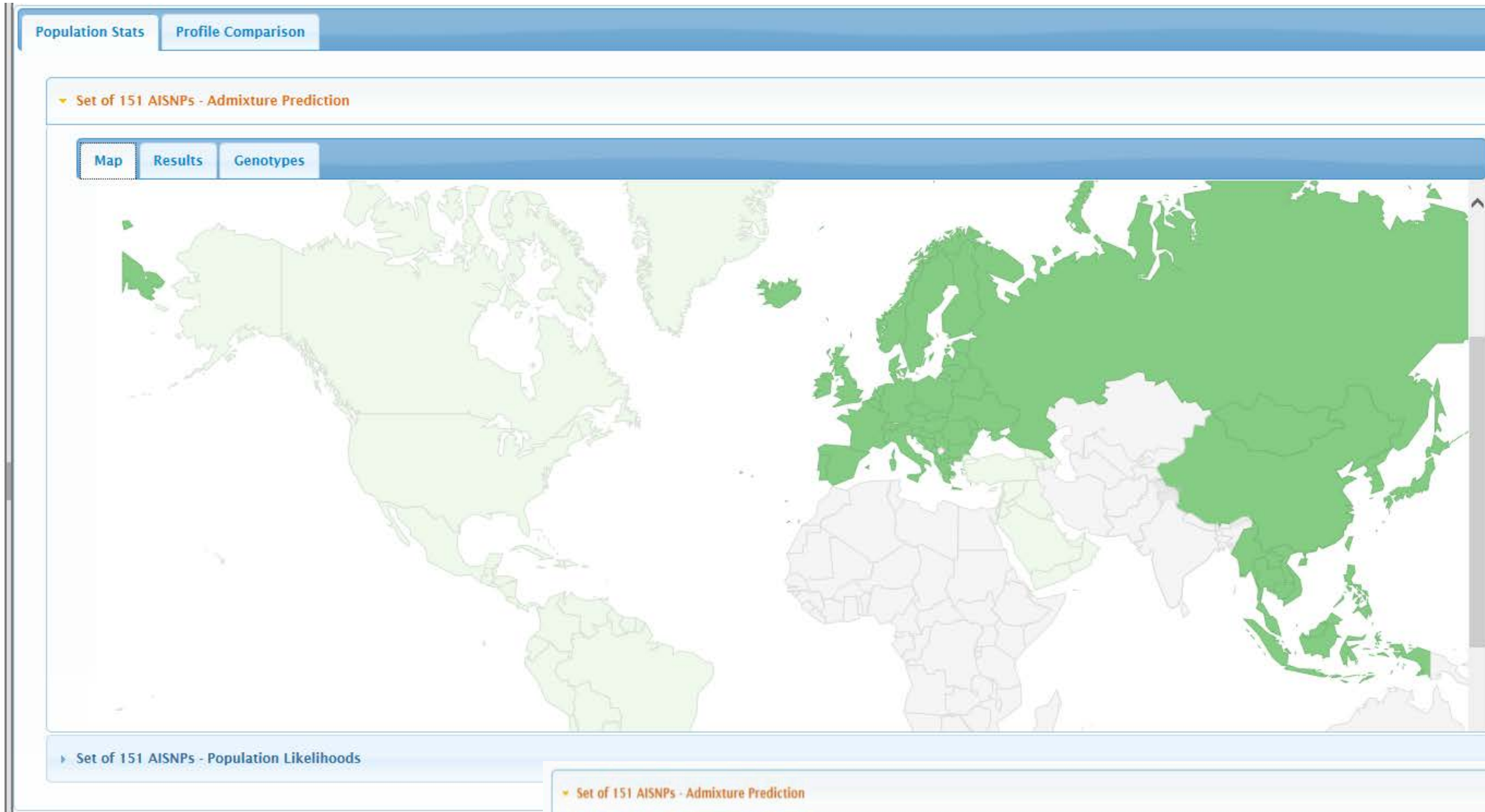




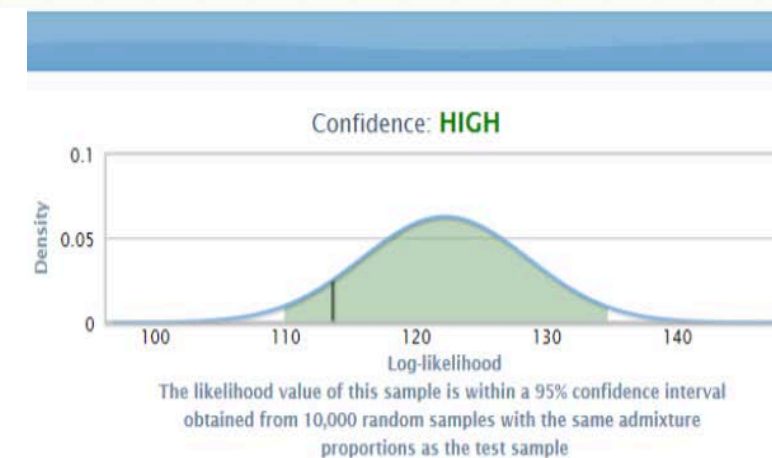
# Statistical approaches to admixture

Show genotypes

Sample Name	Barcode ID	Sample Data
PLPC170919	IonCode_0101	<a href="#">Download</a>
GAL17002-13-1	IonCode_0102	<a href="#">Download</a>
GAL17002-14-1	IonCode_0103	<a href="#">Download</a>
GAL17002-15-1	IonCode_0104	<a href="#">Download</a>
GAL17002-16-1	IonCode_0105	<a href="#">Download</a>
GAL17002-17-1	IonCode_0106	<a href="#">Download</a>
GAL17002-18-1	IonCode_0107	<a href="#">Download</a>
NLPC170919	IonCode_0108	<a href="#">Download</a>
PLPC170912	IonCode_0117	<a href="#">Download</a>
GAL17002-1-1	IonCode_0118	<a href="#">Download</a>
GAL17002-2-1	IonCode_0119	<a href="#">Download</a>
GAL17003-3-1	IonCode_0120	<a href="#">Download</a>
GAL17002-4-1	IonCode_0121	<a href="#">Download</a>
GAL17002-5-1	IonCode_0122	<a href="#">Download</a>
GAL17002-6-1	IonCode_0123	<a href="#">Download</a>
NLPC170912	IonCode_0124	<a href="#">Download</a>
PLPC170913	IonCode_0125	<a href="#">Download</a>
GAL17001-7-1	IonCode_0126	<a href="#">Download</a>
GAL17001-8-1	IonCode_0127	<a href="#">Download</a>
GAL17001-9-1	IonCode_0128	<a href="#">Download</a>
GAL17002-10-1	IonCode_0129	<a href="#">Download</a>
GAL17002-11-1	IonCode_0130	<a href="#">Download</a>
GAL17002-12-1	IonCode_0131	<a href="#">Download</a>
NLPC170913	IonCode_0132	<a href="#">Download</a>



1. Simulate 10,000 random samples that have the same predicted admixture proportions as the test sample.
2. Calculate the likelihood value for each simulated sample and the test sample.
3. Generate a distribution of log-likelihood values from the simulated samples, then compare the log-likelihood of the test sample to the distribution.



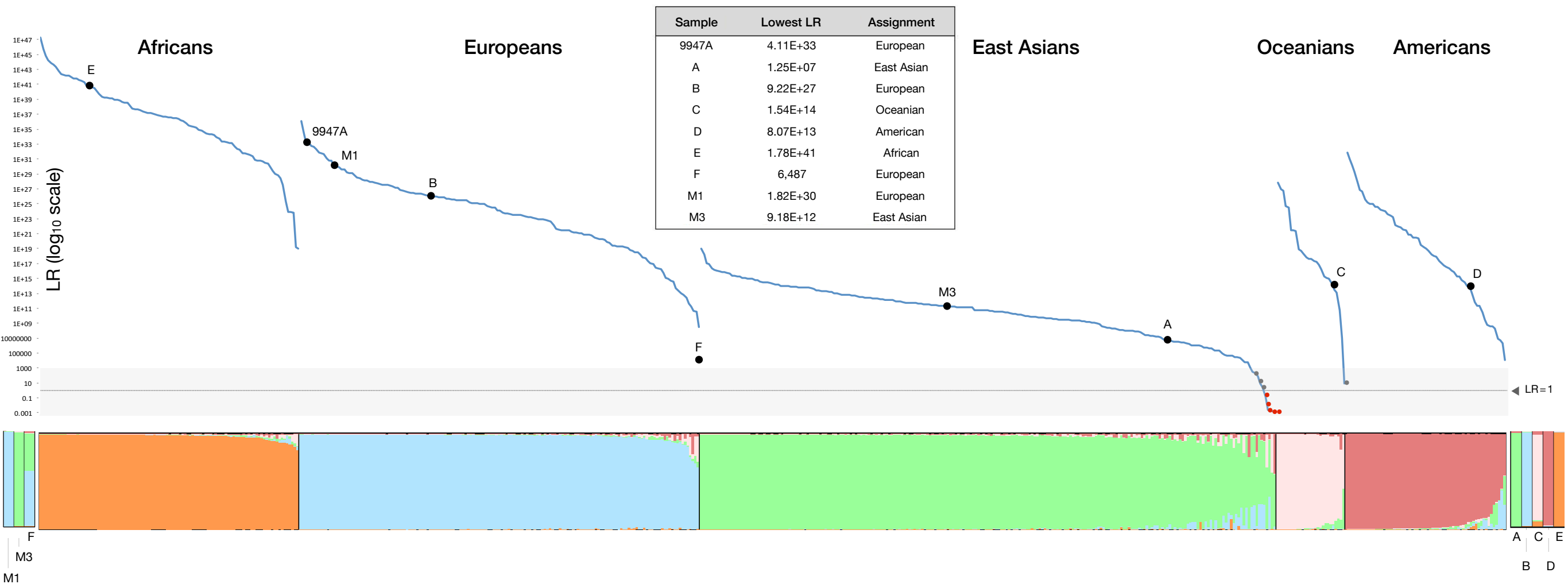
**Error: particularly important when closely related populations are compared**

# Error: particularly important when closely related populations are compared

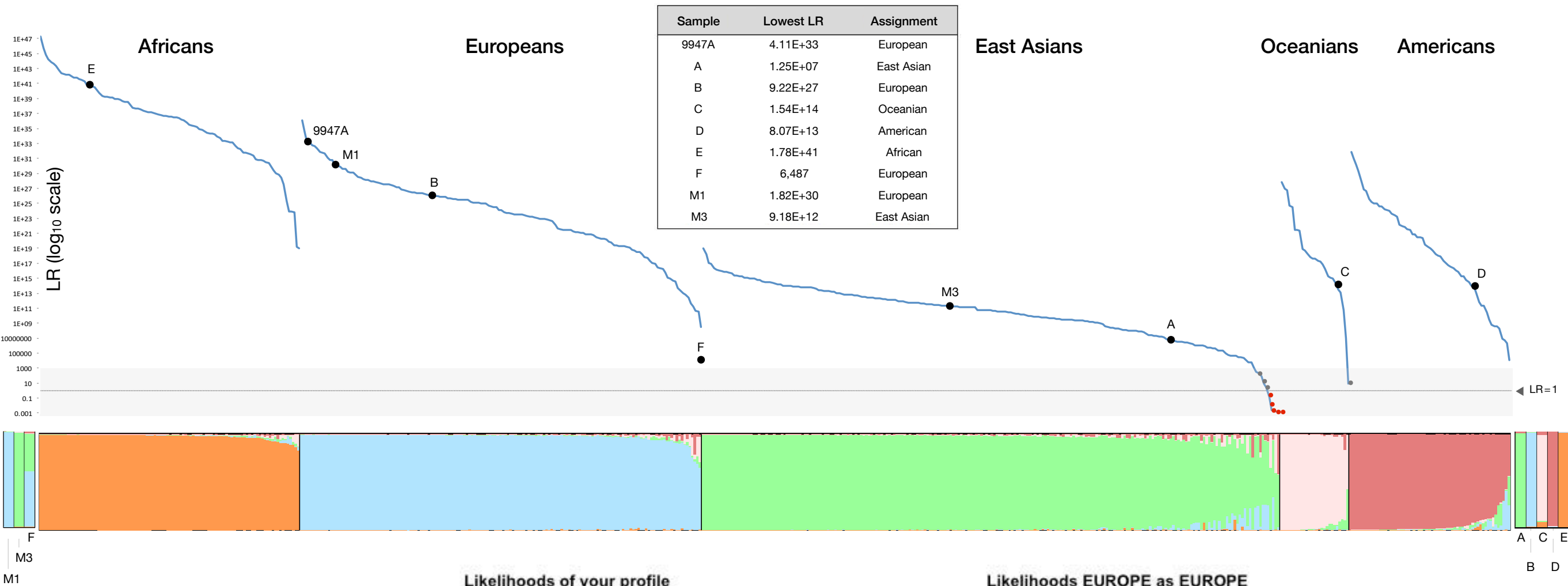
## Daubert's considerations for a new scientific technique

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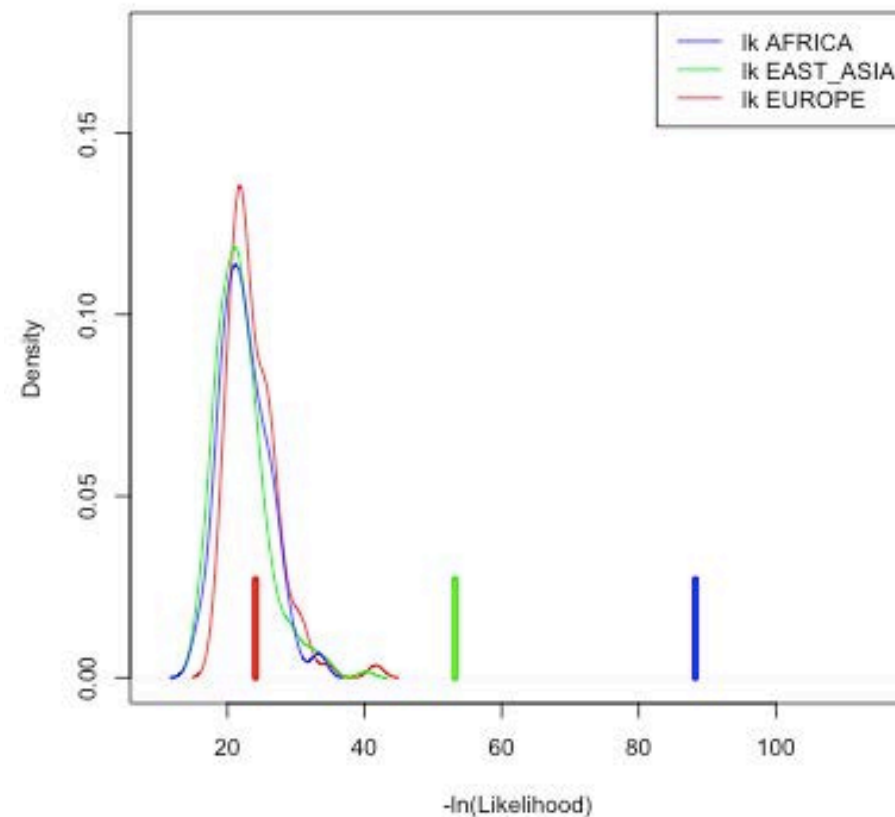
- (1) Can the theory or technique in question be tested and has it been tested?
- (2) Has the technique been subject to peer review and publication?
- (3) What is the known or potential rate of error?
- (4) Do standards exist for the control of the technique's operation?
- (5) Has the technique been generally accepted within the relevant scientific community?



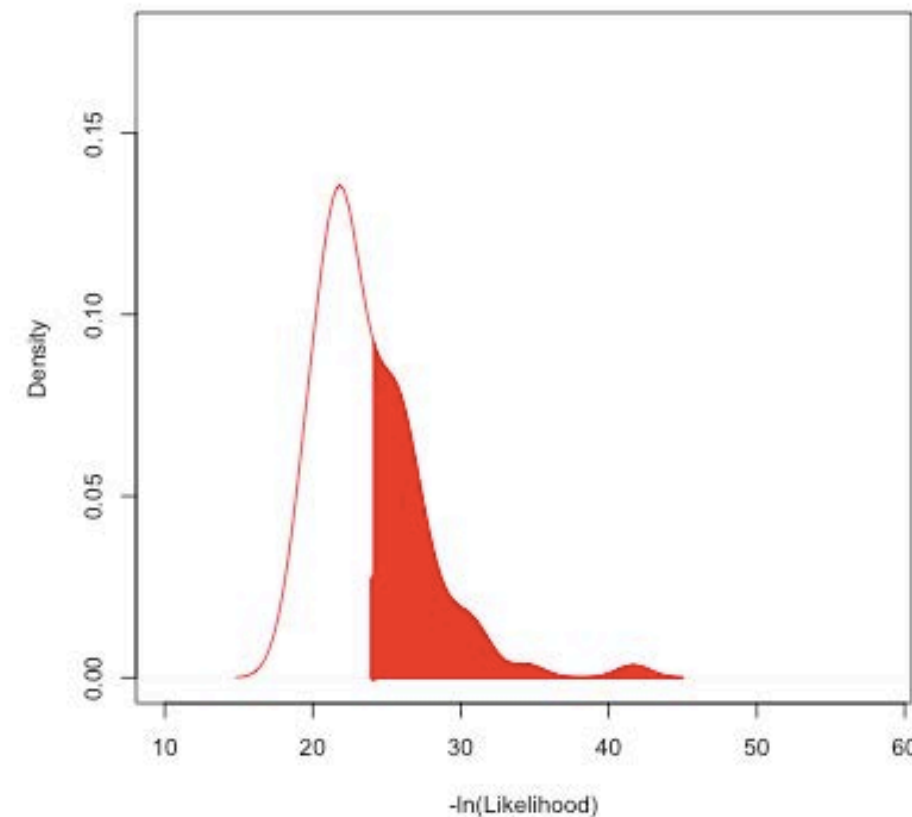
# Error: Reference data likelihood distribution plots



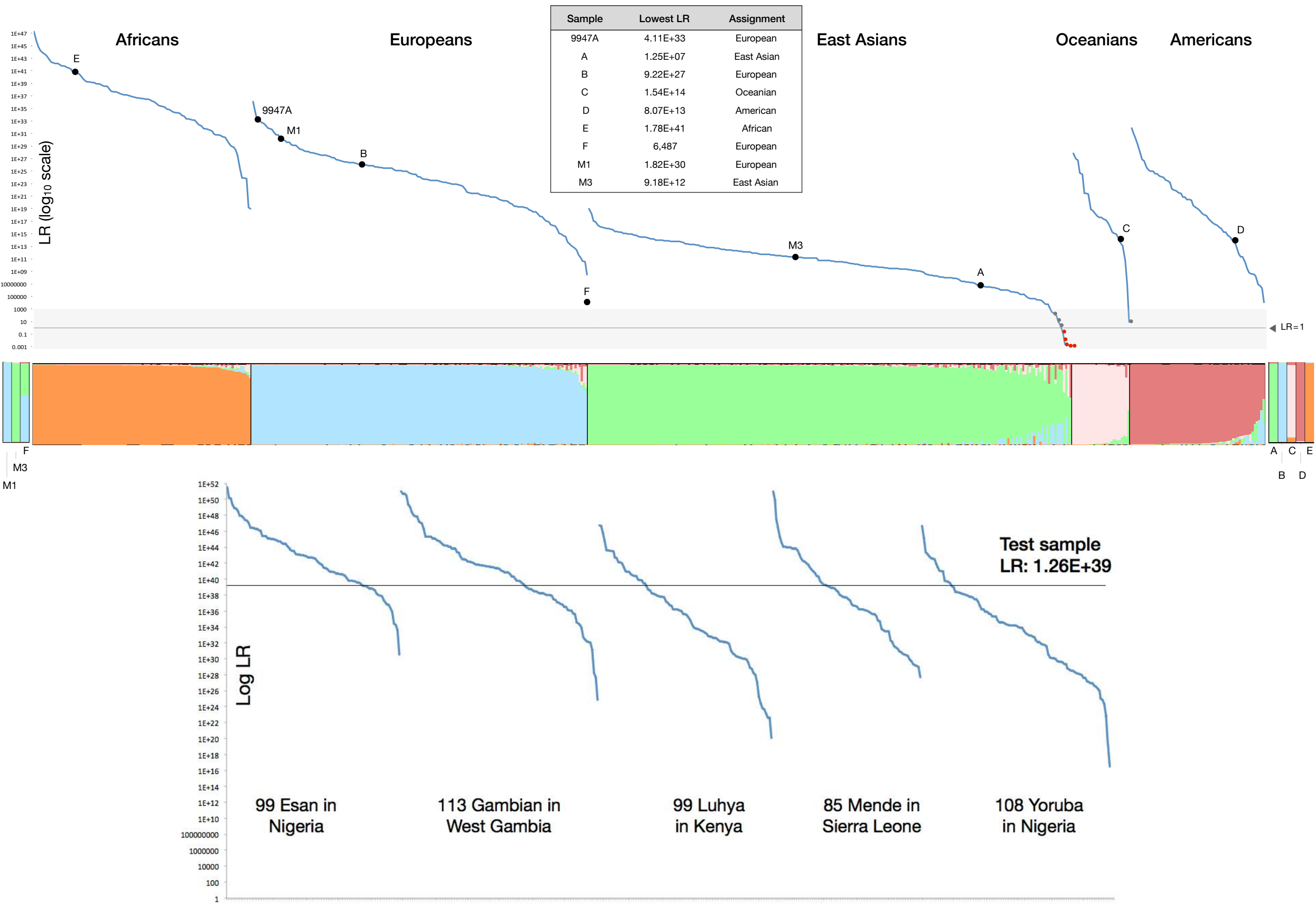
Likelihoods of your profile



Likelihoods EUROPE as EUROPE



*Snipper*





# Viewpoints for consideration, moving forensic ancestry analysis forward and reaching consensus



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*We need to manage expectations about what population differentiations are realistic*

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*We need to manage expectations about what population differentiations are realistic*



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- More detailed analyses made of physical characteristics suggested by the SNP data  
*Likely to have male pattern baldness (but age was estimated to be ~70 years)      Blue eyes*
- Ancestry analysis was much more detailed than is possible with current forensic tests  
*Closely related or directly descended from Italian emigres to the US*

# Viewpoints for consideration, moving forensic ancestry analysis forward and reaching consensus

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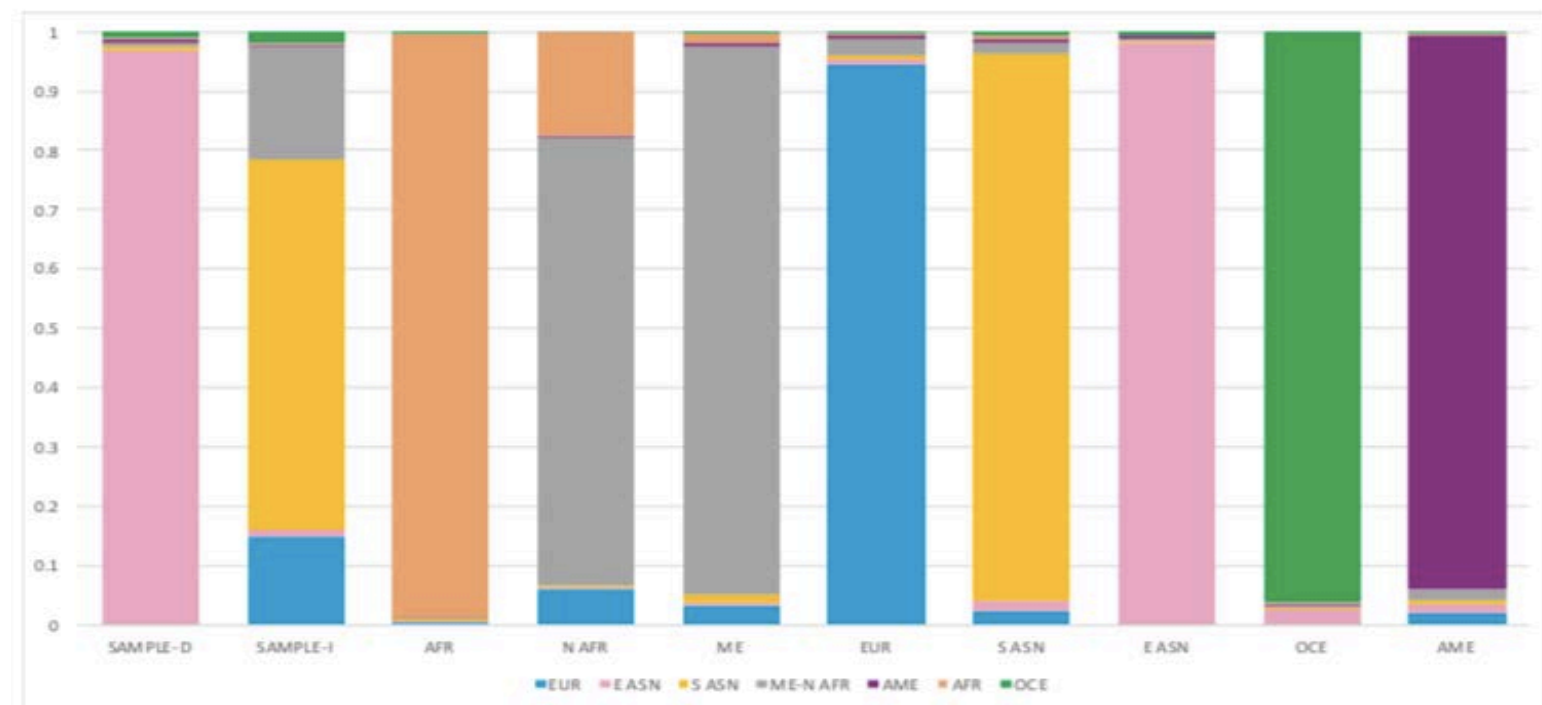
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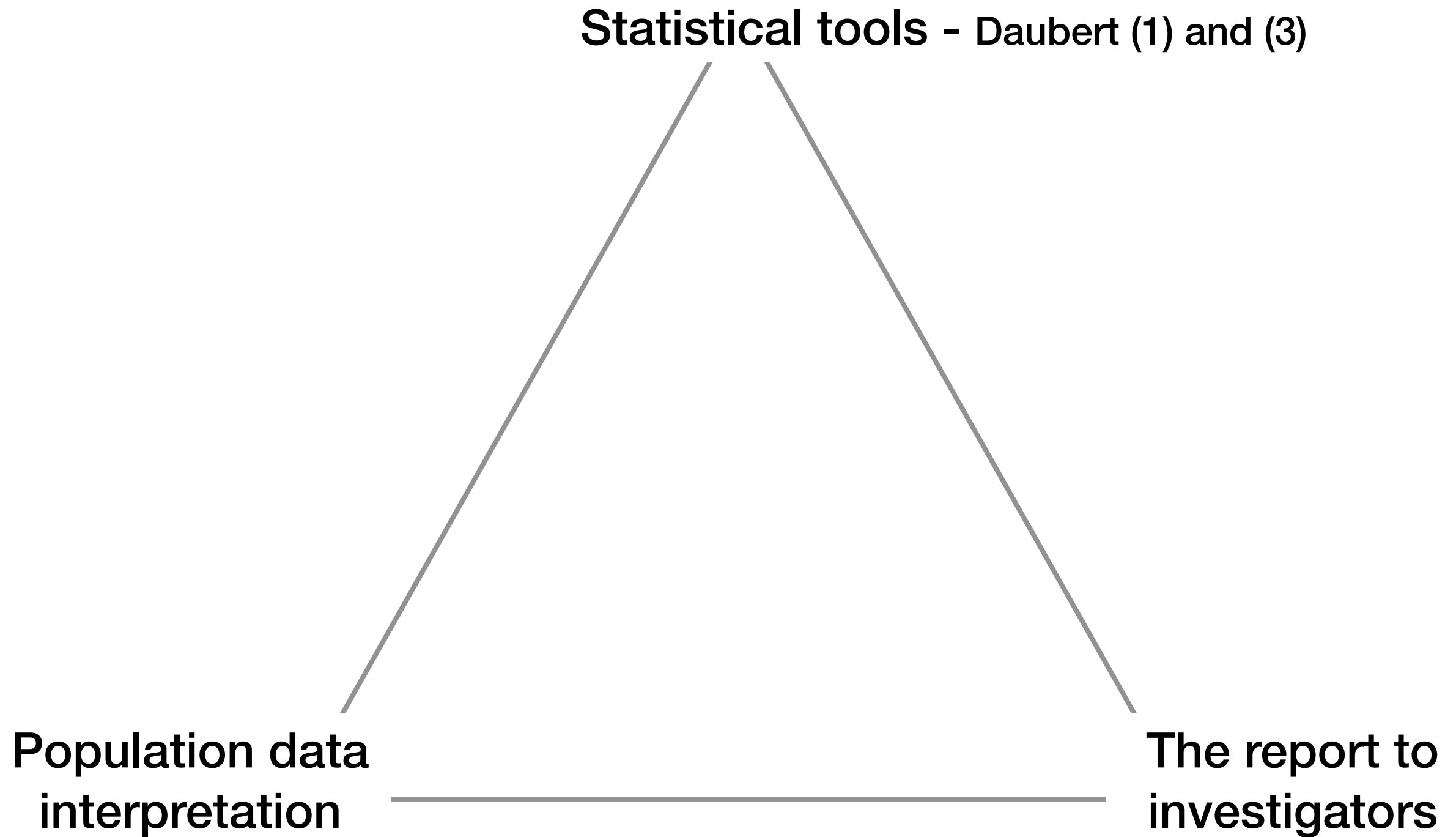
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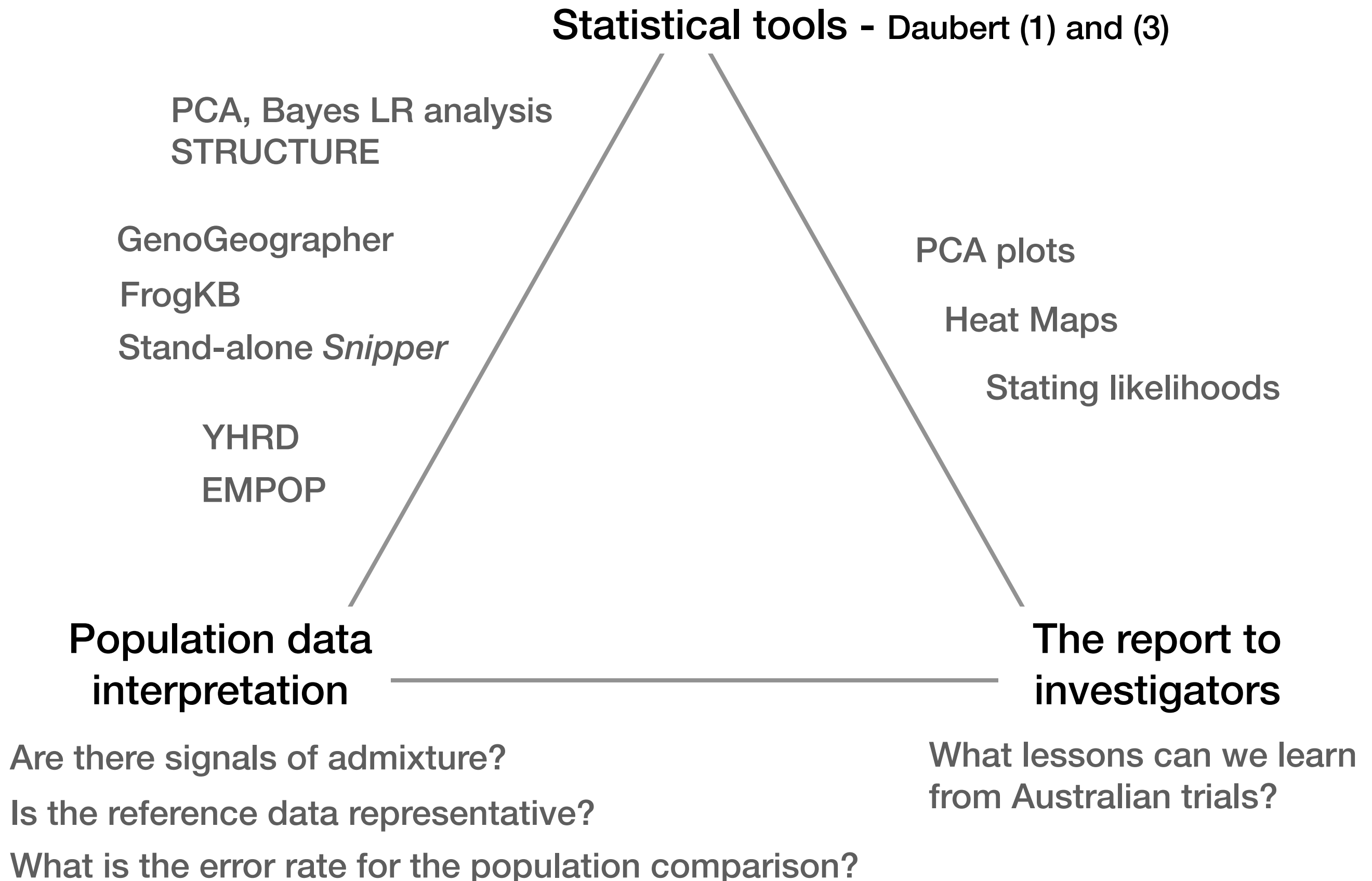
*A system to gauge ancestry assignment error is important - and can be developed around agreement on thresholds*

*Establishing agreed frameworks for statistical tools :  
interpretation : reporting to investigators*

# Aspects of ancestry analysis that will need to evolve:



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LD / allele frequencies

Statistical tools - Daubert (1) and (3)

PCA, Bayes LR analysis  
STRUCTURE

GenoGeographer

FrogKB

Stand-alone *Snipper*

YHRD

EMPOP

PCA plots

Heat Maps

Stating likelihoods

Population data  
interpretation

The report to  
investigators

Are there signals of admixture?

Is the reference data representative?

What is the error rate for the population comparison?

What lessons can we learn  
from Australian trials?

# Next steps in the short-to-medium term

Discussion paper - USC : EMPOP : YHRD

Round table discussions at a suitable forum

Consensus on and development of a toolbox

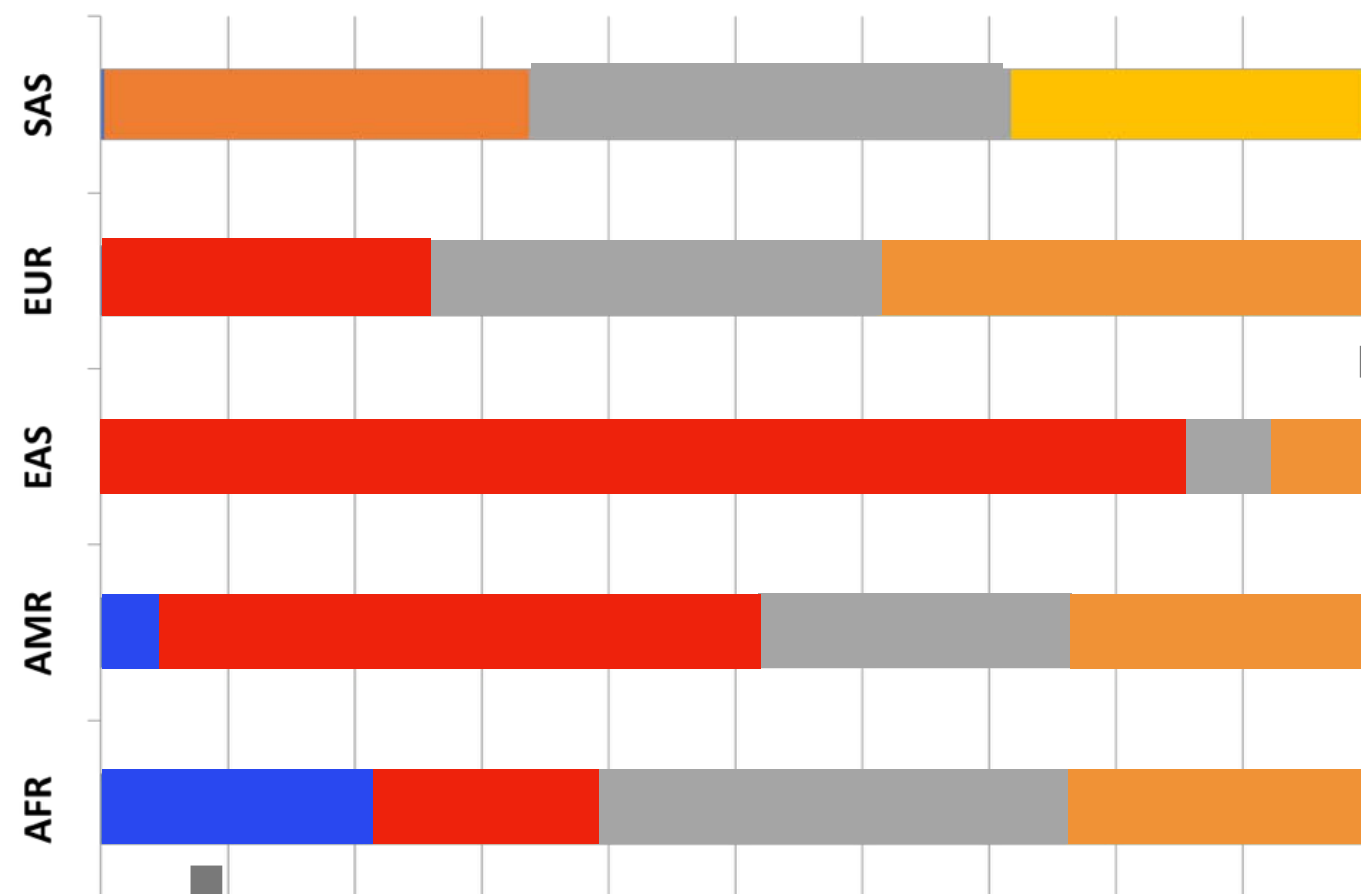
Training and response to user needs

build interpretation skills / rule sets



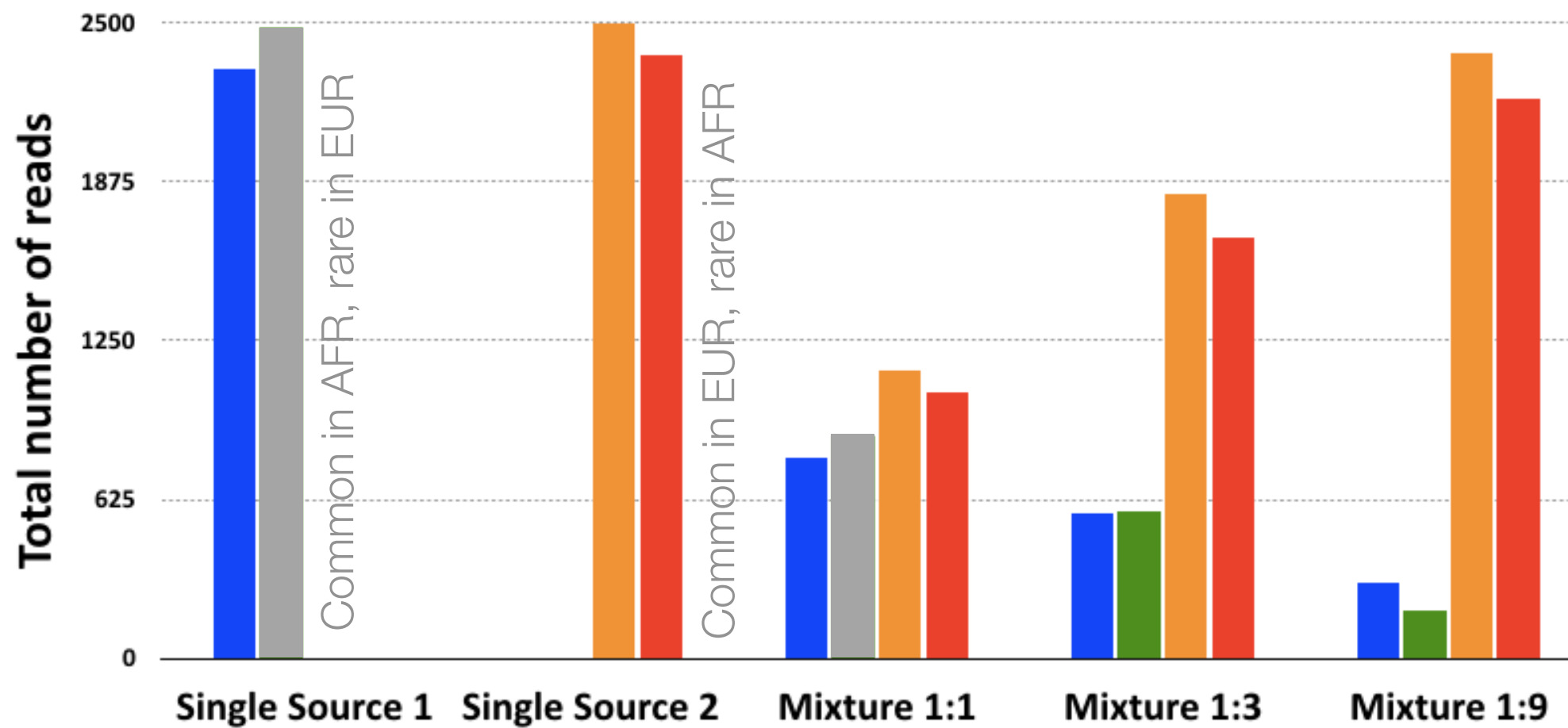






**Skewed  
frequencies**

**Population-  
specific alleles**





Contents lists available at [ScienceDirect](#)

## Forensic Science International: Genetics

journal homepage: [www.elsevier.com/locate/fsigen](http://www.elsevier.com/locate/fsigen)



Research paper

### Performance of ancestry-informative SNP and microhaplotype markers

Elaine Y.Y. Cheung<sup>a,\*</sup>, Christopher Phillips<sup>b</sup>, Mayra Eduardoff<sup>c</sup>, Maria Victoria Lareu<sup>b</sup>,  
Dennis McNevin<sup>d</sup>

