AGENDA FOR THE EDNAP MEETING

INNSBRUCK – 30 OCTOBER 2018

Expected duration: 09.00 - 17.00

Coffee: 11.00-11.30 – Lunch: 13.00-14.00 – Coffee: 15.45-16.00

Host: Walther Parson and Richard Scheithauer
Chairman: Niels Morling

Welcome	Richard Scheithauer
The first collaborative EDNAP exercise	Peter Schneider
A brief history of EDNAP - The first ten years	Peter Gill
Regards from previous EDNAP members	Niles Morling
Update on activities	
Methylated DNA and age exercise	David Ballard
Exercise no. 2 on mRNA typing with MPS	C. Haas/G. Duro
mtDNA quantification exercise - update	Arnoud Kal
Updates from other groups	
ENFSI	Sander Kneppers
EMPOP (DNA.BASES)	Walther Parson
STRidER (DNA.BASES)	Walther Parson
The DNASEQEX project	Walther Parson
The VISAGE project	Walther Parson
ISFG	Walther Parson
Other activities	
Update on Euroformix	Peter Gill
Massive on-line open course in interpretations of DNA results	Tacha Hicks/P. Gill
Future activities	
Collaborative exercise on detection of mtDNA heteroplasmy by MPS	Walther Parson
EDNAP meeting in the spring of 2019	Niels Morling
Any other business	Niels Morling

Dr. David Ballard
Forensic and Analytical Science
King's College London
Franklin Wilkins Building
Waterloo
SE1 9NH London
UK
Tel:
Fax:
E-mail: david.ballard@kcl.ac.uk

Dr. Regine Banemann
KT31
Bundeskriminalamt
Thaerstrasse 11
D-65193 Wiesbaden
Germany
Tel: +49 61155 16053
Fax: +49 611 5545 089
E-mail: regine.banemann@bka.bund.de

Dr. Ingo Bastisch
KT31
Bundeskriminalamt
Thaerstrasse 11
D-65193 Wiesbaden
Germany
Tel: +49 61155 16030
Fax: +49 611 5545 089
E-mail: ingo.bastisch@bka.bund.de

Professor Denise Syndercombe Court
Forensic and Analytical Science
King's College London
Franklin Wilkins Building
Waterloo
SE1 9NH London
UK
Tel: +44 20 7848 4155
Fax: +44 20 7848 4129
E-mail: Denise.syndercombe-court@kcl.ac.uk

Dr. Guro Dörum
Institut für Rechtsmedizin
Universität Zürich-Irchel
Winterthurerstrasse 190
CH-8057 Zürich
Switzerland
Tel: +41 764321797
Fax:
E-mail: guro.dorum@irm.uzh.ch

Dr. Christina Forsberg
National Laboratory of Forensic Science
S-58194 Linköping
Sweden
Tel: +46 105628131
Fax:
E-mail: christina-u.forsberg@polisen.se

Professor Peter Gill
Department of Forensic Biology
National Institute of Public Health
PO Box 4404
Nydalen
N-0403 Oslo
Norway
Tel:
Fax:
E-mail: peterd.gill@gmail.com

Dr. Theresa Gross
Institute of Legal Medicine
University of Cologne
Melatenguertel 60-62
D-50823 Cologne
Germany
Tel: +49 221 478 89447
Fax:
E-mail: theresa.gross@uk-koeln.de

Dr. June Guiness
Home Office
Forensic Science Regulator Unit
5 St. Philips Place, Colmore Row
B3 2PW Birmingham
UK
Tel: +44 121 200 3830
Fax:
E-mail: june.guiness@homeoffice.gsi.gov.uk

Dr. Cordula Haas
Institut für Rechtsmedizin Zurich
Winterthurerstr. 190
CH-8057 Zurich
Switzerland
Tel: +41 44 635 5656
Fax: +41 44 635 6858

E-mail: cordula.haas@irm.uzh.ch

Dr. Arnoud Kal
Department of Human Biological Traces
Netherlands Forensic Institute
Laan van Ypenburg 6
24 97 GB The Haque
The Netherlands
Tel: +31 708 886 729
Fax: -
E-mail: a.kal@nfi.minvenj.nl

Dr. Alexander Kneppers
Department of Human Biological Traces
Netherlands Forensic Institute
Laan van Ypenburg 6
24 97 GB The Haque
The Netherlands
Tel: +31629623036
Fax:
E-mail: s.kneppers@nfi.minvenj.nl

Professor Maviky Lareu
Forensic Genetic Unit
Department of Legal Medicine
University of Santiago de Compostela
San Francisco, s/n
E-15705 Santiago de Compostela
Spain
Tel: +34 98158 2327
Fax: +34 98158 0336
E-mail: apimllar@usc.es

Professor, dr.med. Niels Morling
Section of Forensic Genetics
Department of Forensic Medicine
Faculty of Health Sciences
University of Copenhagen
Frederik V´s Vej 11
DK-2100 Copenhagen
Denmark
Tel: +45 3532 6115
Fax: +45 3532 6270
E-mail: niels.morling@sund.ku.dk

Dr. Fabrice Noël
National Institute of Forensic Science
98-100 Chaussée de Vilvorde
B-1120 Bruxelles
Belgium
Tel: +32 2243 4604
Fax: +32 2240 0501
E-mail: fabrice.noel@just.fgov.be

Prof. Dr. Walther Parson
Institute of Legal Medicine
Medical University of Innsbruck
Müllerstrasse 44
A-6020 Innsbruck
Austria
Tel: +43 512 9003 70640
Fax: +43 512 9003 73640
E-mail: walther.parson@i-med.ac.at

Dr. Vania Pereira
Section of Forensic Genetics
Department of Forensic Medicine
Faculty of Health Sciences
University of Copenhagen
Frederik V´s Vej 11
DK-2100 Copenhagen
Denmark
Tel:
Fax:
E-mail: vania.pereira@sund.ku.dk

Dr. Chris Phillips
Forensic Genetic Unit
Department of Legal Medicine
University of Santiago de Compostela
San Francisco, s/n
E-15705 Santiago de Compostela
Spain
Tel: +34 98158 2327
Fax: +34 98158 0336
E-mail: c.phillips@mac.com

Prof. Dr.med. Richard Scheithauer
Institute of Legal Medicine
Medical University of Innsbruck
Müllerstrasse 44
A-6020 Innsbruck
Austria
Tel: +43512 9003 70600
Fax: +43 512 9003 73600
E-mail: richard.scheithauer@i-med.ac.at

Prof.Dr. Peter M. Schneider

Institute of Legal Medicine
University of Cologne
Melatenguertel 60-62
D-50823 Cologne
Germany
Tel: +49 221 4788 8345
Fax: +49 221 4788 8370
E-mail: peter.schneider@uk-koeln.de

Dr. Bo Simonsen
Section of Forensic Genetics
Department of Forensic Medicine
Faculty of Health Sciences
University of Copenhagen
Frederik V´s Vej 11
DK-2100 Copenhagen
Denmark
Tel: +45 3532 6136
Fax: +45 3532 6270
E-mail: bo.simonsen@sund.ku.dk

Dr. Livia Zatkalikova
Institute of Forensic Science
Slovenská L'upca
Priboj 560
976 13
Slovak Republic
Tel: +421 961 60 6333
Fax: +421 961 60 6309
E-mail: livia.zatkalikova@minv.sk

EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

Innsbruck, Austria

30 October 2018

Host: Walther Parson and Richard Scheithauer
Chairman: Niels Morling

A list of participants is attached.

Welcome
Richar Scheithauer welcomed members to Innsbruck.

Update on exercises

The first collaborative exercise Peter Schneider
Peter Schneider gave a short review of the first collaborative EDNAP exercise on investigations of DNA with restriction fragment analysis and single locus DNA probes for forensic genetic investigation (presentation attached).

A brief history of EDNAP - The first ten years Peter Gill
Peter Gill gave a review of the activities of EDNAP during the first 10 years (presentation attached).

Regards from previous EDNAP members Niels Morling
Niels Morling presented regards from previous EDNAP members and selected photos from the first years of EDNAP (presentation attached)

Second exercise on methylated DNA and age David Ballard
David Ballard presented an update of the results of the second collaborative EDNAP exercise on age estimation by means of measurements of methylation of selected DNA positions. A draft of a manuscript is expected to be circulated before the end of 2018 (presentation attached).

Exercise no. 2 on mRNA typing with NGS Cordula Haas/Guro Dörum
Cordula Haas presented the results of the second collaborative EDNAP exercise on discrimination between various tissues and body fluids with mRNA determined with MPS. Cordula Haas reported new preliminary results on coding region SNPs (cSNPs) detected on transcripts of RNA from various tissues. Guro Dörum presented preliminary results on evaluation of the mRNA results. A draft of a manuscript is expected to be circulated before the next EDNAP meeting in spring 2019 (presentation attached).

mtDNA quantification exercise Arnoud Kal
Arnoud Kal updated about the planned collaborative exercise concerning quantification of mtDNA. Arnoud Kal on 29 Oct 2018 circulated the attached email. Please contact Arnoud Kal if you want to participate. The plan is to send out the exercise within the next few weeks (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).

TThe DNASEQEX project Walther Parson

Walther Parson gave an update on the DNASEQEX project (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

Update on Euroformix Peter Gill

Peter Gill gave an update on Euroformix (presentation attached).

Free on-line course in interpretations of DNA results Tacha Hicks/Peter Gill

Peter Gill presented on behalf of Tacha Hicks Champod, Formation continue UNIL-EPFL & Ecole des sciences criminelles, Lausanne, France a free on-line course of interpretation of forensic science data. The course is intended as an awareness course. The course will be announce soon. A trailer is found at https://www.youtube.com/watch?v=UNewyg5xV5Q (presentation attached).

Future activities

mtDNA quantification exercise Arnoud Kal

Arnoud Kal and colleagues will send out the samples etc. for the exercise within the next few weeks. Laboratories are expected to return the results before 15 January 2019. The plan is to present preliminary results at the EDNAP meeting in the spring of 2019.

Collaborative exercise on detection of mtDNA heteropasmy by MPS Walther Parson

Walther Parson suggested to perform a collaborative exercise on mtDNA heteroplasmy. The Innsbruck laboratory plans to send out DNA extracted from individuals with point and/or length heteroplasmy. Participants are kindly asked to inform Walther Parson before 30 November 2018 if they will be able to contribute with interesting samples for circulation. The participating laboratories are invited to contribute to this exercise by providing sequence data of these samples using their preferred mtDNA sequencing method(s). This includes conventional Sanger and Massive Parallel Sequencing technologies. Walther Parson plans to send the experimental plan and samples around 15 January 2019. The plan is to invite participants to submit results no later than 15 March 2019, so that the preliminary results can be presented at the EDNAP meeting in the spring of 2019 (presentation enclosed).

Next exercise on mRNA typing with NGS Cordula Haas

Cordula Haas will be considering to present a suggestion for a new exercise at the EDNAP meeting in the spring of 2019.

Next meetings Niels Morling

The next EDNAP meeting will take place in Madrid in association with the CODIS and ENFSI DNA Working Group – most likely on 7 May 2019.

Any other business Niels Morling

There was no other business.

Closing of the meeting Niels Morling

The meeting closed with sincere thanks to Walther Parson, Richard Scheithauer, and all colleagues, who helped organise the meeting.

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

- Agenda
- Group photo
- List of participants
- Presentations
  - Peter Schneider: The first collaborative EDNAP exercise
  - Peter Gill: A brief history of EDNAP - The first ten years
  - Niels Morling: Regards from previous EDNAP members
  - David Ballard: Report on methylated DNA and age determination
  - Cordula Haas: Report on the second collaborative exercise on mRNA NGS
  - Arnoud Kal: Information on the planned mtDNA quantification exercise
  - Sander Kneppers: Report from the ENFSI DNA Working Group
  - Walther Parson: EMPOP report (DNA.BASES)
  - Walther Parson: STRidER report (DNA.BASES)
  - Walther Parson: The DNASEQEX project
  - Walther parsons: The VISAGE project
  - Walther Parson: ISFG report
  - Peter Gill: Update on Euroformix
  - Tacha Hicks: Free on-line course in interpretations of DNA results
  - Walther Parson: Collaborative exercise on detection of mtDNA heteroplasmy by MPS.

30 years EDNAP
1988-2018

1989

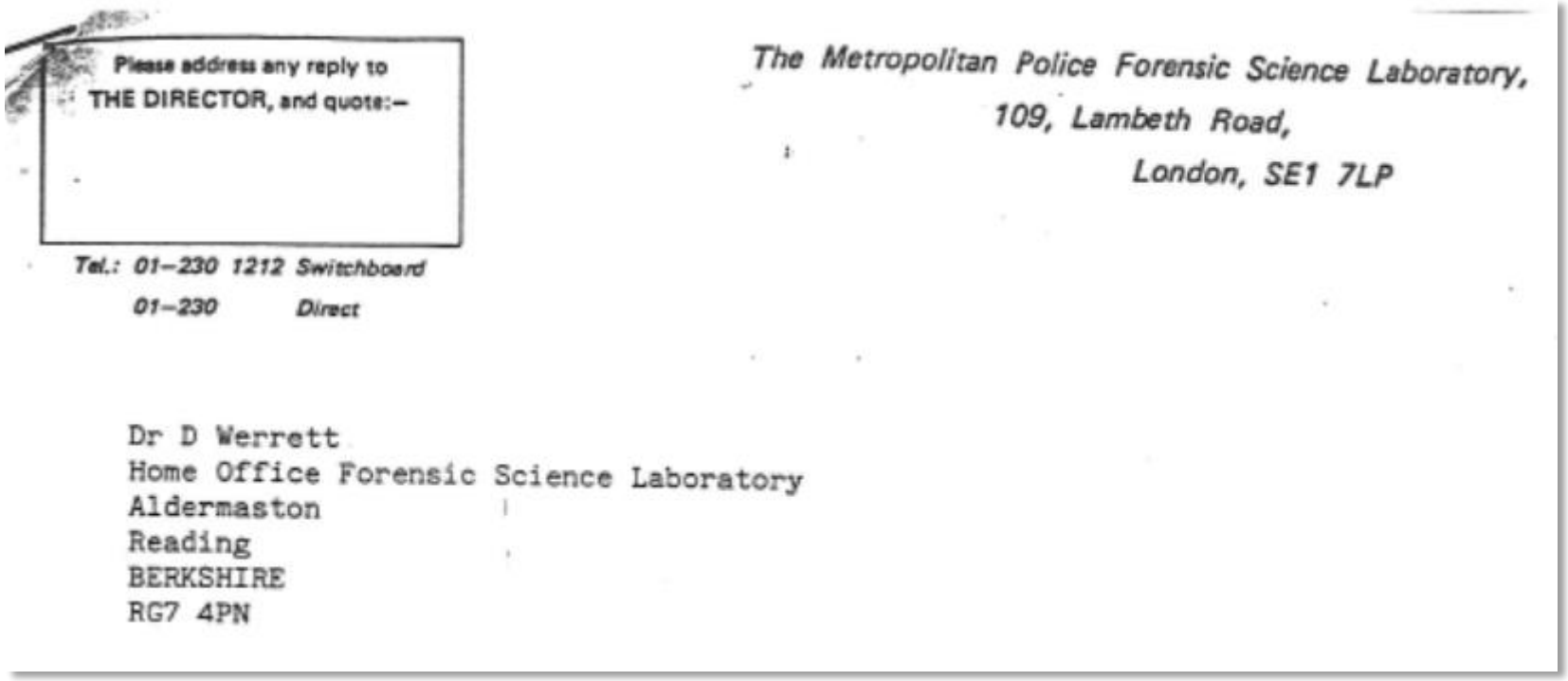
On the way to the first collaborative exercise

Peter Schneider (Innsbruck, 30.10.2018)

MEETINGS OF THE EDNAP GROUP

A selection of meeting pictures can be found in the Gallery

No.	Date	Place	Special Topic	Minutes
1.	16 10.1988	Sunbury, UK	First Meeting	
2.	04.02.1989	Münster, Germany	First collaborative exercise on SLPs decided, acronym 'EDNAP' introduced	
3.	06.10.1989	Den Haag, Netherlands	First results of first SLP exercise	PDF Minutes



Please address any reply to
THE DIRECTOR, and quote:–

Tel.: 01–230 1212 Switchboard
01–230 Direct

The Metropolitan Police Forensic Science Laboratory,
109, Lambeth Road,
London, SE1 7LP

Dr D Werrett
Home Office Forensic Science Laboratory
Aldermaston
Reading
BERKSHIRE
RG7 4PN

4 January 1989

Dear Dave

With a further meeting now imminent on European standardisation in DNA profiling I thought it worth summarising where we left off at the Sunbury meeting. Points needing further consideration were listed as follows:

1. Flexible alternatives to DNA profiling. Traditional grouping should not be abandoned
2. Core package of probes
3. Standardisation of restriction enzyme
4. Standard molecular weight markers
5. Population data
6. Records for comparative purposes
7. QA and proficiency testing - QC of commercial products
8. Legal constraints
9. New technology
10. Reporting probabilities

Please let us know if you need any further input from the MPFSL. By copy of this letter I will remind those who attended the Sunbury meeting to let the study group have any comments they wish to make if they have not already done so. In return, although I realise time is passing, I hope that those attending the February meeting in Munster will receive copies of the report a day or two ahead of the meeting itself.

With best wishes

Yours sincerely

Brian

Dr B Sheard
Director

Copies:

Dr B Eriksen
Prof B Brinkmann
[redacted]
Dr H Schmitter
Prof U Rossi
Prof E D'Aloia
Dr A D Kloosterman
Dr H Lochtenberg
Dr W Baer
Mr V Emerson
Mr B H Parkin
Mr P D Martin

METROPOLITAN POLICE

FORENSIC SCIENCE LABORATORY

14 February 1989

Dear Colleague

I have enclosed a synopsis of the recent meeting in Munster and I am sure that you would wish to join me in thanking Prof. Brinkmann and Mr Rand for their work in providing the venue for the meeting and also making the social arrangements.

The synopsis includes the details of the research exercise to determine standard deviations of bands usng Hinf I and YNH 24.

There appears to have been general agreement that the meeting was a great success and on behalf of the CRSE and Metropolitan Police Laboratory we thank you sincerely for your co-operation in this attempt to bring the Europeans closer together.

Very best wishes

P.D. Martin

Peter D Martin
Deputy Director

DNA Profiling - European Integration
Munster - 4th February 1989

Chairman: Prof B Brinkmann

A list of participants is appended.

Synopsis of the proceedings

Prof Brinkmann opened the meeting by welcoming all of the participants to the second gathering of this group. He outlined a case which had been examined in Germany and involved the rape and murder of an English woman by an English man. The DNA grouping of the semen showed a frequency of 1 in 60 million, and the British Army SIB felt that the suspect may have been responsible for other such crimes in the UK. This illustrates the value of inter-European databases.

Bruce Budowle (FBI) is investigating small alleles for PCR and also discrete alleles because of the problem of formulating band-sharing statistics. If this is successful and PCR is the choice for the future the subject of the use of enzymes would disappear.

There are difficulties with PCR/Dot blotting at present especially as dilution will increase the danger of losing alleles.

Small alleles are amplified preferentially over the larger ones. Contamination is still the biggest problem.

In the discussion which followed members were reminded that Cetus hold the patent for PCR and that each laboratory would have to be licensed separately.

A limited amount of research on PCR has been carried out at CRSE. It was thought that the use of PCR for routine casework is still some way into the future.

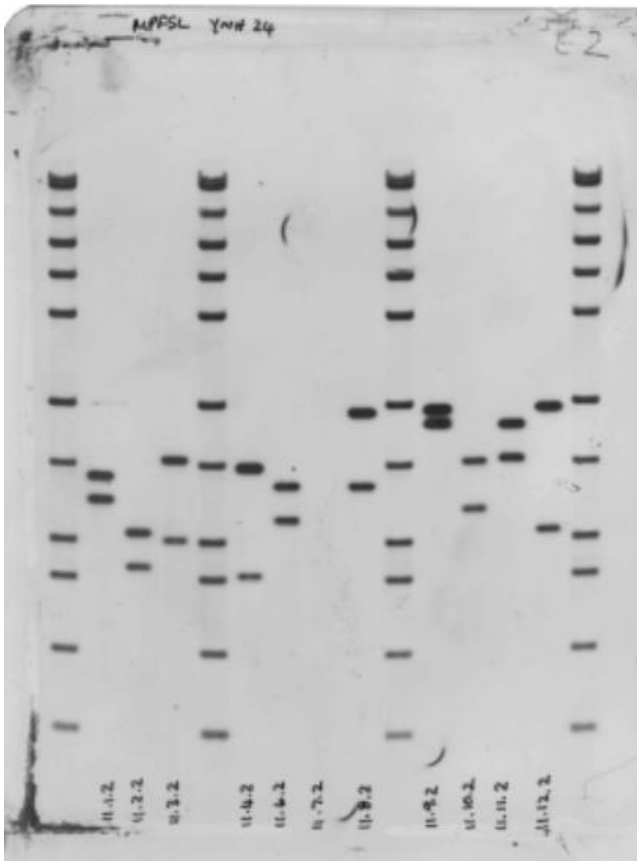
The conditions of the exercise were then presented to the meeting.

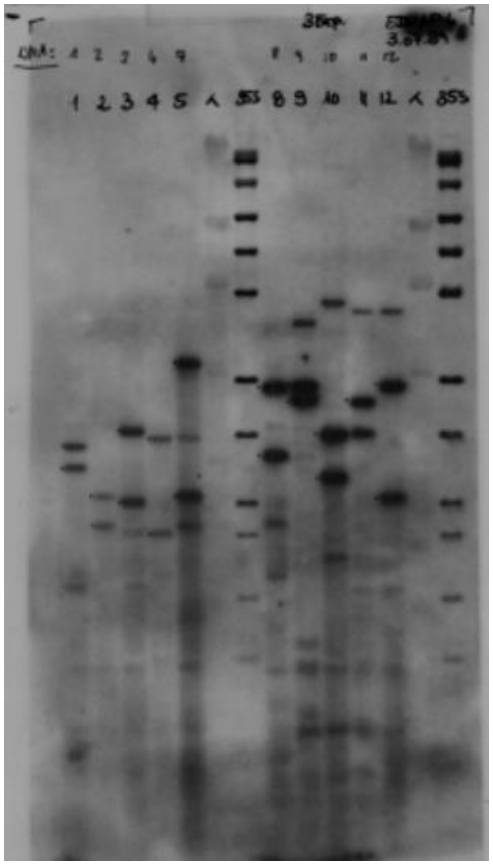
Each of the 12 labs will prepare enough DNA for a least 50µg to be sent to each of the participants.
The DNA should be in TE buffer and the concentration stated.
Samples should be distributed by 15 March 1989.
Werrett will supply each lab with YNH24 and ask Amersham to distribute ^{35}S markers by 15 March 1989.
Each lab should check their DNA preparation for high m.wt concentration.
The source and batch number of Hinf I should be stated.

The length of the gel should be stated and should be greater than 20 cm.
Each sample should be run ten times.
The concentration and volume added to each track should be stated.
Measurements of bands (as per Schneider) should be made, and those together with autoradiographs (or good copies) sent to Prof Rittner and Dr Werrett.
Results should be despatched by 31 July 1989.
(If a laboratory has not completed the whole exercise, whatever results are available should be sent by 31 July 1989.)

Mr Kloosterman and Mr Lochtenberg agreed to host the next meeting in The Hague on 6 October 1989.

Prof Brinkmann thanked all present and after a concensus wished the participants to be known as the European DNA Profiling Group, he closed the meeting.


MPPSL YNH 24
E2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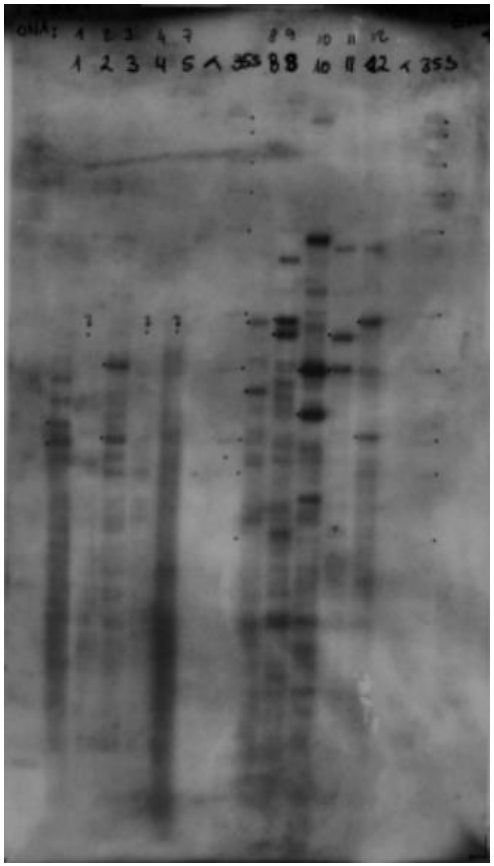
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Invited Paper

REPORT OF A EUROPEAN COLLABORATIVE EXERCISE COMPARING DNA TYPING RESULTS USING A SINGLE LOCUS VNTR PROBE

P.M. SCHNEIDER[a,*], R. FIMMERS[b], S. WOODROFFE[c], D.J. WERRETT[c], W. BÄR[d], B. BRINKMANN[e], B. ERIKSEN[f], S. JONES[g], A.D. KLOOSTERMAN[h], B. MEVÅG[i], V.L. PASCALI[j], C. RITTNER[a], H. SCHMITTER[k], J.A. THOMSON[l] and P. GILL[c]

[a]*Institute of Legal Medicine, University of Mainz (F.R.G.),* [b]*Institute for Medical Statistics, University of Bonn (F.R.G.),* [c]*CRSE, Home Office Forensic Science Service, Aldermaston (U.K.),* [d]*Institute of Legal Medicine, University of Zürich (Switzerland),* [e]*Institute of Legal Medicine, University of Münster (F.R.G.),* [f]*Institute of Forensic Genetics, Copenhagen (Denmark),* [g]*Metropolitan Police Forensic Science Laboratory, London (U.K.),* [h]*Forensic Science Laboratory, Rijswijk (The Netherlands),* [i]*Institute of Forensic Medicine, University of Oslo (Norway),* [j]*Institute of Legal Medicine, Catholic University, Rome (Italy),* [k]*Bundeskriminalamt, Wiesbaden (F.R.G.) and* [l]*Department of Haematology, London Hospital Medical College (U.K.)*

MINUTES OF THE GENERAL ASSEMBLY OF THE INTERNATIONAL SOCIETY FOR FORENSIC HAEMOGENETICS (ISFH) ON SEPTEMBER 20, 1991 IN THE KURFÜRSTLICHE SCHLOSS ZU MAINZ (17.00 - 18.20)

TOP 4 Report of the Executive Committee and the Auditors of the Account

4.1. Report of the President
B. Brinkmann reported that the Executive Committee had handled its business since the last congress in only 4 sessions.
He presented the wish of the EDNAP (European DNA Profiling Association) to become a Working Party of the ISFH. This request was accepted unanimously.


isfh
14TH CONGRESS
INTERNATIONAL SOCIETY
FOR FORENSIC HAEMOGENETICS
September 18 - 21, 1991

A brief history of EDNAP
The first ten years

Peter Gill

The first collaboration

- First meeting in Sunbury, UK in 1988.
- From the beginning the focus was on single locus probe rather than multi-locus probes, because the latter were to complex to interpret and there were limitations to reproducibility and scoring data for comparison purposes
- Single locus probes were discovered in 1982 by Ray White, some years before the Jeffreys probes were launched.
- The procedure was slow and tedious and involved using dangerous amounts of radioactivity, along with copious amounts of carcinogens like ethidium bromide

1989, Holland

- Topics included the thorny subject of biostatistics
- SLPs were ‘continuous’ which means that there were hundreds of overlapping alleles present in a population.
- We identified SLPs relative to their size measured next to ladder markers – much like today except the standard use was from a viral DNA marker
- Much discussion about how to apply frequency estimates to alleles and essentially there were two alternative methods termed the ‘binning method’ vs the ‘floating binning’ method

Questions addressed

Dr Werrett followed his presentation by posing 4 pertinent questions which should be discussed at the next meeting.

1. Is it possible to compare the results from one laboratory with those from another?

2. Is it possible to combine data-bases from various laboratories for population frequencies?

3. Should laboratories be accredited for their competence to perform DNA profiling?

4. What are the parameters necessary for the interpretation of autoradiographs?

Purpose and adoption of core probes as European standards

The aim of this group was restated:

"European integration using common technology with reference to stain grouping."

Finally it was agreed that MS43A should be the second probe adopted by EDNAP for all participating laboratories. This means that we now have a consensus to use Hinf 1 as the restriction enzyme and YNH 24 and MS43A as our core probes and the Amersham ^{35}S control markers.

1990 Bramshill, UK

- 4x SLPs had been introduced into casework in the UK
- MLPs gradually phased out of use
- Standardisation exercise:

It was unanimously agreed that we should all participate in an exercise whereby all laboratories used a standard method. The following points were considered:-

1.	Ladder markers	-	'Cold' Amersham ladder to be used.
2.	Ground control	-	Kloosterman will supply this from his immortal cell-lines.
3.	Agarose	-	CRSE will provide
4.	Buffer	-	Details will be supplied by CRSE.
5.	Ethidium bromide	-	This will only be used in the λ Hind III track.
6.	Samples	-	CRSE will supply 3 pre-extracted DNA samples and the same samples already restricted.

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Invited Paper

REPORT OF A EUROPEAN COLLABORATIVE EXERCISE COMPARING DNA TYPING RESULTS USING A SINGLE LOCUS VNTR PROBE

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[a]*Institute of Legal Medicine, University of Mainz (F.R.G.),* [b]*Institute for Medical Statistics, University of Bonn (F.R.G.),* [c]*CRSE, Home Office Forensic Science Service, Aldermaston (U.K.),* [d]*Institute of Legal Medicine, University of Zürich (Switzerland),* [e]*Institute of Legal Medicine, University of Münster (F.R.G.),* [f]*Institute of Forensic Genetics, Copenhagen (Denmark),* [g]*Metropolitan Police Forensic Science Laboratory, London (U.K.),* [h]*Forensic Science Laboratory, Rijswijk (The Netherlands),* [i]*Institute of Forensic Medicine, University of Oslo (Norway),* [j]*Institute of Legal Medicine, Catholic University, Rome (Italy),* [k]*Bundeskriminalamt, Wiesbaden (F.R.G.) and* [l]*Department of Haematology, London Hospital Medical College (U.K.)*

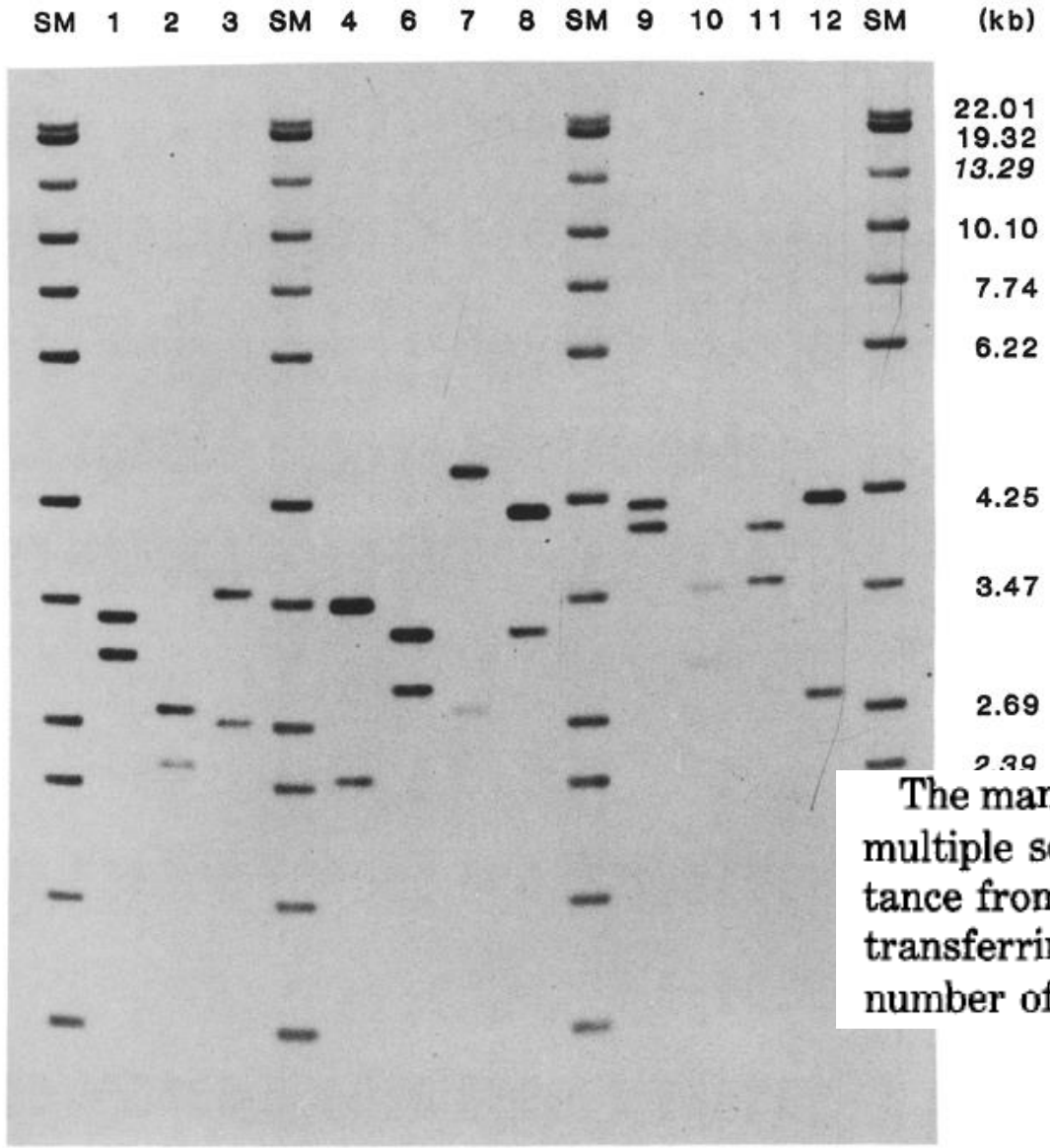

Fig. 2. Example for EDNAP exercise blot from laboratory 11 hybridized with ^{32}P-labelled YNH24. All 11 DNA samples of the exercise are included, the respective sample numbers are indicated above. The sizes of the ^{35}S-labelled marker fragments (SM) are given on the right side with the reference points for gel run length and RBP method printed in italics.

The manual reading of fragment positions using a ruler was problematic, since multiple sources of error were observed: (1) reading the correct migration distance from the ruler, (2) writing these measurements on a coding sheet and (3) transferring the data from the coding sheet to the computerized database. A number of these errors were only detected, when the printed data (see Figs. 3

The second collaborative exercise

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A REPORT OF AN INTERNATIONAL COLLABORATIVE EXPERIMENT TO DEMONSTRATE THE UNIFORMITY OBTAINABLE USING DNA PROFILING TECHNIQUES

P. GILL[a], S. WOODROFFE[a], W. BÄR[b], B. BRINKMANN[c], A. CARRACEDO[d], B. ERIKSEN[e], S. JONES[f], A.D. KLOOSTERMAN[g], B. LUDES[h], B. MEVAG[i], V.L. PASCALI[j], M. RUDLER[k], H. SCHMITTER[l], P.M. SCHNEIDER[m] and J.A. THOMSON[n]

[a]Home Office Central Research and Support Establishment, Aldermaston (U.K.), [b]Institute of Legal Medicine, University of Zurich (Switzerland), [c]Institute of Legal Medicine, University of Münster (Germany), [d]Department of Legal Medicine, University de Santiago de Compostela (Spain), [e]Institute of Forensic Genetics, Copenhagen (Denmark), [f]Metropolitan Police Forensic Science Laboratory, London (U.K.), [g]Forensic Science Laboratory, Rijswik (The Netherlands), [h]Institute of Legal Medicine, University of Strasbourg (France), [i]Institute of Forensic Medicine, University of Oslo (Norway), [j]Institute of Legal Medicine, Catholic University, Rome (Italy), [k]Laboratoire de Police Scientifie de Paris, Ministere de L'Interier (France), [l]Bundeskriminalamt, Wiesbaden (Germany), [m]Institute of Legal Medicine, University of Mainz (Germany) and [n]Department of Haematology, London Hospital Medical College (U.K.)

(Received July 15th, 1991)
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(Accepted December 20th, 1991)

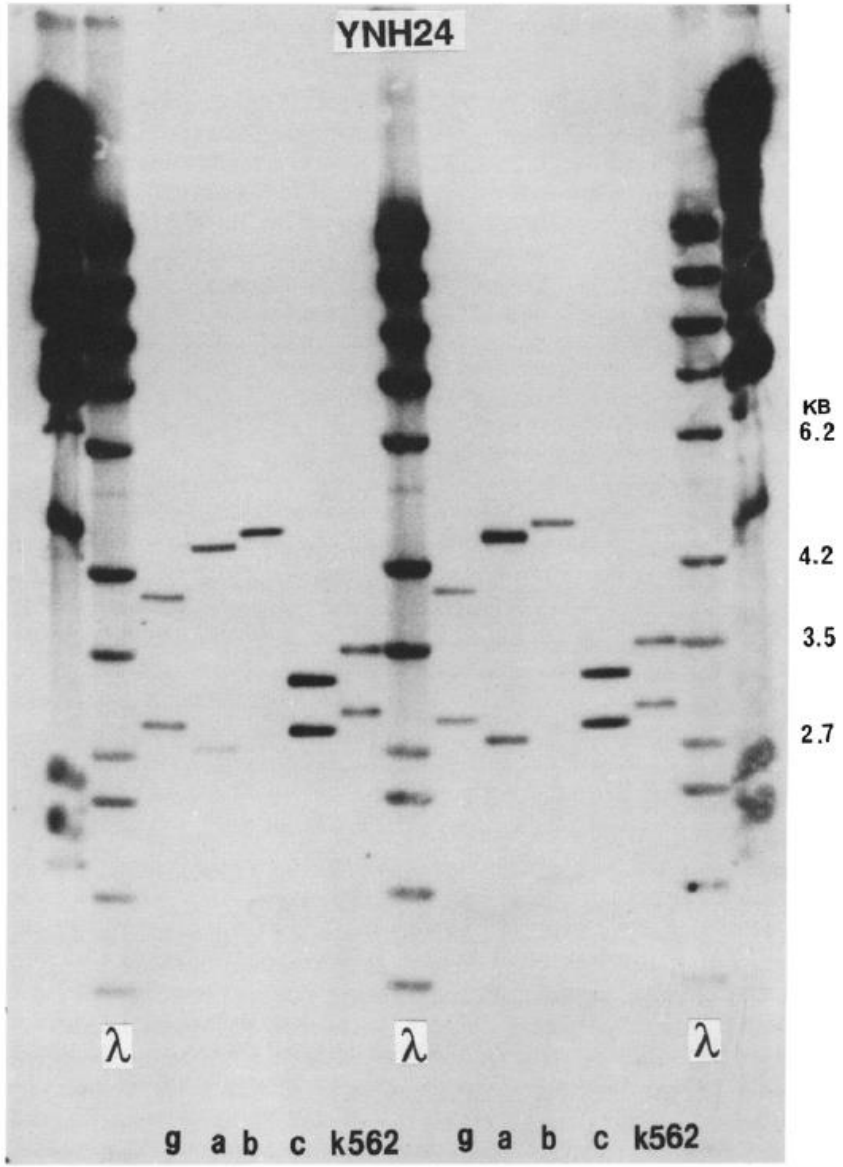

Fig. 1. Photograph of an autoradiograph showing the experimental design. The left hand side of the autoradiograph consisted of samples restricted by CRSE whereas the right hand side consisted of samples restricted by the participating laboratory. λ, ladder marker; g, genomic control (not used in this exercise); a, sample a; b, sample b; c, sample c; k, K562.

On a European level the following appear to be important:-

(a) Common enzyme
(b) Common probes
(c) Marker ladder
(d) Genomic control
(e) Electrophoretic system

The method of calculating the allelic (fragment) size is also important.

A uniform system has advantages:-

(a) Band sizes, determined in different labs, are directly comparable.
(b) The same statistical methodology can be used for interpretation.

suspect/victim. For this determination to be made each laboratory must have:-

(a) reproducible day-to-day methodology
(b) population frequency distributions
(c) genomic control which falls within a 'window'

For YNH 24

achieved > 99% agreement at a 2.8% match criterion
achieved 100% agreement at a 3.6% match criterion

Quality control and standards (discussed at the Rome meeting 1991

CRSE would provide a Quality Assurance programme which could either be used as proficiency test or as part of a Quality Management system.

For the first exercises laboratories would be given the results with no comments on performance.

In the discussion which followed Steve Rand suggested that some laboratories might join the scheme solely to avoid adverse criticism. He said that he could see many problems with the QA programme.

He felt that there were issues to be addressed beyond the integration process and that we should select objectives which are compatible with the EDNAP aims. Initially the group could become a European repository for population data and this would have the immediate advantages of

(a) an extended array of frequencies
(b) a deeper insight into population genetics

There was a unanimous decision at the meeting in Rome on 19 April, to make an application to the International Society for Forensic Haematologics to become a working group called EDNAP.

Aims: To meet the original aims of EDNAP

(a) To determine accurately alleles using single locus probing
(b) To facilitate the exchange of data between countries.
(c) The group will only study DNA extracted from stained material.

Further aims:

(i) Set and maintain standards within DNA profiling for European laboratories engaged in stain work.

(ii) To provide a forum for the exchange of population frequency databases and collaboration in research.

Oslo 1992 – towards STRs

- EDNAP became a working group of the ISFH

4. Short Tandem Repeats (STRs) **M.Greenhalgh, MPFSL**

PCR is still in the research stage at the MPFSL but, having identified several problems associated with PCR-based VNTRs, a programme of collaborative work with CRSE has been agreed to progress work on STRs.
There are several reasons for choosing STRs in preference to other PCR-based VNTRs, among them being:-

resistance to degradation
ease of multiplexing
general robustness
short repeats - less chance of deletions

MPFSL and CRSE have agreed to concentrate on SE33 and HumTHO 1 in the initial stages.

There was some considerable discussion on the ethical problems on the use of polymorphisms from non-coding regions which have some linkage association with disease problems related to coding parts of the genome. It was decided that we should keep a watching brief on this situation and return to the subject at our next meeting.

The following examples were used to illustrate the choice available:-

name	core repeat	No of alleles
HumTHO 1	(TCAT)n	7
SE33	(AAAG)n AA(AAAG)n	21

The third collaborative exercise

6. Further collaborative exercises

Peter Gill proposed that the next collaborative exercise should be based on STRs and that the group should have STRs as the linking systems for future casework.
In the first instance HumTHO 1 and SE33 should be used and form the basis of the collaborative exercise. This is without prejudice as other STRs may be identified at a later date which have better characteristics for casework.

This was accepted unanimously.

CRSE will prepare the trial and Peter Schneider offered to make the primers. Details will be sent to members of the group.

ELSEVIER SCIENCE
IRELAND

Forensic Science International
65 (1994) 51–59

Forensic
Science
International

Report of the European DNA profiling group (EDNAP) — towards standardisation of short tandem repeat (STR) loci

P. Gill*[a], C. Kimpton[a], E. D'Aloja[b], J.F. Andersen[c], W. Bar[d], B. Brinkmann[e], S. Holgersson[f], V. Johnsson[g], A.D. Kloosterman[h], M.V. Lareu[i], L. Nellemann[j], H. Pfitzinger[k], C.P. Phillips[l], H. Schmitter[m], P.M. Schneider[n], M. Stenersen[o]

Allelic ladders

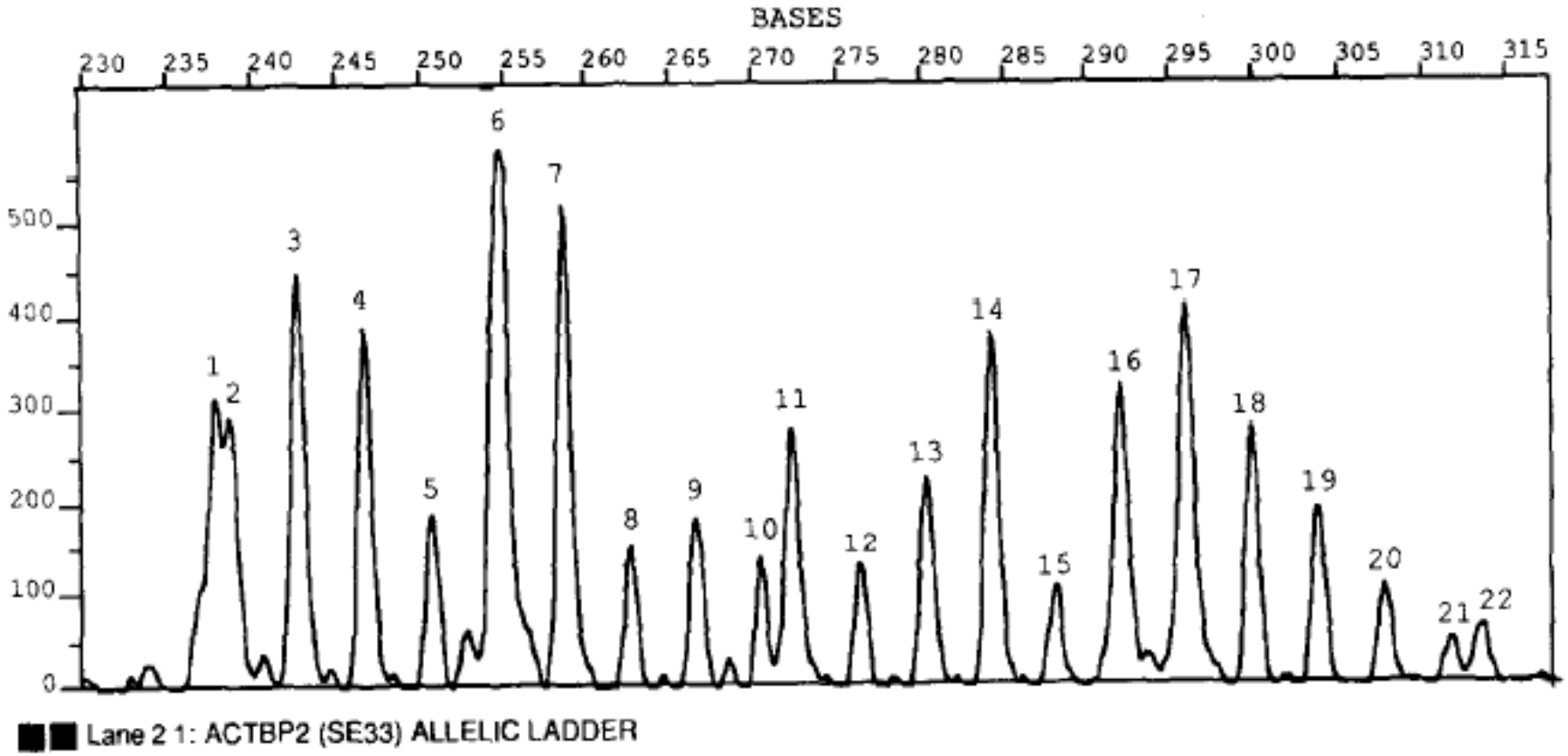

Fig. 2. Allelic ladder for HUMACTBP2 (SE33) analysed using the ABI automated sequencer. Allelic designation shown above each peak.

To summarise, the EDNAP exercise demonstrated that simple STR systems such as HUMTH01 are suitable candidates for standardisation even where diverse electrophoretic systems are utilised by different laboratories. Further work is required to characterise complex AT-rich loci (>30 alleles) because of difficulties encountered with the reproducibility of migration rates in different electrophoretic systems. For these systems, standardisation may not be possible unless common ladder markers and electrophoretic systems are used.

1993. The ISFH meeting at Venezia, Italy
The quadplex was introduced and an exercise carried out

Coimbra Portugal 1994 – the quadplex and first use of Fst. Variation between populations

Peter Gill presented some population information generated from the quadruplex of loci THO1, VWA, F13 and FES. Among fifteen populations obtained from various laboratories, he reported that the biggest variation in allelic frequency between different ethnic populations was in THO1. He reported a similar variation in allelic frequency between White Caucasian populations. Although the FSS and MPFSL routinely use a default frequency value of 5.0%, they also include an Fst kinship factor according to the formula produced by Nicholls and Balding:

$$P = C + (1-C)p$$ where C is the inbreeding coefficient

First discussions on national DNA databases (1994) and first mention of CE

3. Capillary electrophoresis

Ate Kloosterman reported that Beckmann have made considerable advances with the development of capillary electrophoresis for DNA analysis. It is now possible to obtain STR separations with a single base pair resolution and a throughput of 2 to 3 samples per hour. In order to increase the throughput work is continuing on automated sampling with multi-channel analysis. Each column is capable of 1000-2000 sample separations but each new column must be separately calibrated. He considered that it will be 1 to 2 years before the technology is sufficiently advanced for use in forensic science. **Peter Gill** informed members that ABI are also developing the technology.

Third collaborative exercise (1996)

ELSEVIER

Forensic Science International
78 (1996) 83–93

In summary, the results obtained from this collaborative EDNAP exercise demonstrate that the simple STR loci VWA and THO1 are good candidates for standardisation on an international scale. It is anticipated that the use of these systems will expedite cross-border exchange of information.

Report on the third EDNAP collaborative STR exercise

Julia Andersen[a,*], Peter Martin[a], A. Carracedo[b], M. Dobosz[c], B. Eriksen[d], V. Johnsson[e], C. Kimpton[f], A. Kloosterman[g], C. Konialis[h], A. Kratzer[i], P. Phillips[j], B. Mevåg[k], H. Pfitzinger[l], S. Rand[m], B. Rosén[n], H. Schmitter[o], P. Schneider[p], M. Vide[q]

[a] *Metropolitan Police Forensic Science Laboratory, 109,*
[b] *Institute of Legal Medicine, Santiago*
[c] *Catholic University, Ro*
[d] *Department of Forensic Genetics, Institute of Forensi Copenhagen, Denm*
[e] *National Bureau of Investigation Crime La*
[f] *Service Development, Forensic Science S*
[g] *Forensic Science Laboratory, Rijsw*
[h] *Metrogen SA, Athens,*
[i] *Institute of Legal Medicine, University of*
[j] *London Hospital Medical Colleg*
[k] *Rettsmedisinsk Institut, Rikshopit*
[l] *Institute of Legal Medicine, University of St*
[m] *Institute of Legal Medicine, University of*
[n] *Statens Kriminaltekniska Laboratorium, Linkoping, Sweden*
[o] *Bundeskriminalamt, Wiesbaden, Germany*
[p] *Institute of Legal Medicine, University of Mainz, Mainz, Germany*
[q] *Instituto de Medicina Legal, Coimbra, Portugal*

All EDNAP laboratories participating in this exercise successfully typed the DNA from five stains, including two of mixed origin, at the VWA and THO1 loci. This was achieved despite variation in the amplification, electrophoresis and detection systems utilised by individual laboratories. The results demonstrate that these simple STR loci are ideal for standardisation in the forensic community, where different laboratories have varying resources.

London, UK 1996. D21S11 and HUMFIBRA

- Nomenclature discussed

The discussion centred on the difficulties associated with the loci in which there is more than one repeating unit. There were differing recommendations offered to the meeting but the one which found universal favour concerned the production of an overall number for each fragment based on comparison with a known, defined allele. In order to achieve this it is necessary to have a comprehensive agreed internal ladder which would be used by all laboratories.

ELSEVIER

Forensic Science International
87 (1997) 185–192

Forensic Science International

Considerations from the European DNA profiling group (EDNAP) concerning STR nomenclature

Peter Gill[a], B. Brinkmann[b], E. d'Aloja[c], J. Andersen[d], W. Bar[e], A. Carracedo[f], B. Dupuy[g], B. Eriksen[h], M. Jangblad[i], V. Johnsson[j], A.D. Kloosterman[k], P. Lincoln[l], N. Morling[h], S. Rand[b], M. Sabatier[m], R. Scheithauer[n], P. Schneider[o], M.C. Vide[p]

[a]*Service Development, Forensic Science Service, Birmingham UK*
[b]*Institute of Legal Medicine, University of Münster, Münster, Germany*
[c]*Catholic University, Rome, Italy*
[d]*Forensic Science Service, London, UK*
[e]*Institute of Legal Medicine, University of Zurich, Zurich, Switzerland*
[f]*Department of Legal Medicine, University de Santiago de Compostella, Santiago de Compostella, Spain*
[g]*Rettsmedisinsk Institut, Ricshospitalet 0027, Oslo, Norway*
[h]*Department of Forensic Genetics, Institute of Forensic Medicine, University of Copenhagen, Copenhagen, Denmark*
[i]*SKL-National Laboratory of Forensic Science, Linkoping, Sweden*
[j]*National Bureau of Investigation, Helsinki, Finland*
[k]*Netherlands Forensic Science Institute, Volmerrlanan 17, Rijswijk, Netherlands*
[l]*Department of Haemotology, St. Bartholomews and the Royal London School of Medicine and Dentistry, London, UK*
[m]*Laboratorie de Police Scientifique, Toulouse, France*
[n]*Institute of Forensic Medicine, University of Innsbruck, Innsbruck, Austria*
[o]*Institute of Legal Medicine, University of Mainz, Mainz, Germany*
[p]*Instituto de Medicina Legal, Largo da se Nova, 3000, Coimbra, Portugal*

Brussels 1997 - STADNAP

- D21S11 and HUMFIBRA exercise reported
- The Oslo triplex – SE33, APOAII and D11S554
- Mitochondrial DNA – the first discussion and exercise
- The second and third generation multiplexes

A new combination of STR systems, TGM (third generation multiplex), is going to be introduced. The TGM will be used for further investigations when a match has been found in the intelligence database. The power of discrimination (PD) of the SGM is 10^{-7} and the combined PD of the SGM and TGM is 10^{-15}.

Herman Schmitter informed that, in Germany, it seems as if there is a growing interest in intelligence DNA databases.

- Relationship with ENFSI discussed
- The NRCII report

Rome 1998

- More on mitochondrial DNA
- First exercises on Y-chromosome DNA

ELSEVIER

Forensic Science International
97 (1998) 165–170

Forensic Science International

Reproducibility of mtDNA analysis between laboratories: a report of the European DNA profiling group (EDNAP)

A. Carracedo[a,*], E. D'Aloja[b], B. Dupuy[c], A. Jangblad[d], M. Karjalainen[e], C. Lambert[f], W. Parson[g], H. Pfeiffer[h], H. Pfitzinger[i], M. Sabatier[j], D. Syndercombe Court[k], C. Vide[l]

[a]*Institute of Legal Medicine, Faculty of Legal Medicine, University of Santiago de Compostela, 15705 Santiago de Compostela, Spain*
[b]*Catholic University, Rome, Italy*
[c]*Rettsmedisinsk Institutt, Oslo, Norway*
[d]*SKL-National Laboratory of Forensic Science, Linköping, Sweden*
[e]*National Bureau of Investigation, Helsinki, Finland*
[f]*Service Development, Forensic Science Service, Birmingham, UK*
[g]*Institute of Forensic Medicine, University of Innsbruck, Innsbruck, Austria*
[h]*Institute of Legal Medicine, University of Munster, Munster, Germany*
[i]*Codgene, University of Strasbourg, Strasbourg, France*
[j]*Laboratorie de Police Scientifique, Toulouse, France*
[k]*Department of Haematology, St. Bartholomew's and The Royal London School of Medicine and Dentistry, London, UK*
[l]*Instituto de Medicina Legal, Coimbra, Portugal*

Conclusion

- The EDNAP collaboration was instrumental in driving forward the adoption of DNA profiling technology throughout Europe and beyond
- Standardisation
- Nomenclature
- Population genetics
- Expansion to new markers like STRs, MtDNA, Y chromosome

REGARDS FROM PREVIOUS EDNAP MEMBERS

30 YEARS

OCTOBER 2018

INNSBRUCK

NIELS MORLING

SECRETARY OF EDNAP

1st EDNAP MEETING 1988

Meeting on

The Science of DNA Profiling: Needs for European Integration

London 15-16 October 1988

PROGRAMME

Saturday, 15 October

Chairman: Mr. V. Emerson

2.00 pm	Introduction	Dr. B. Sheard
2.20	Review of the technical alternatives for DNA profiling	Mr. B. Parkin
3.00	The introduction of DNA profiling into Forensic Casework	Mr. D. Werrett
3.40	Tea	
4.00	Summaries of progress to date:	
	Denmark	Dr. Birthe Eriksen
	Federal Republic of Germany	
	University of Munster	Prof. B. Brinkmann
	Bundeskriminalamt, Wiesbaden	Dr. H. Schmitter
	Italy	Prof. Rossi
	Netherlands	Dr. A. Kloosterman
	Switzerland	Dr. W. Bäer
	UK	Mr. B. Parkin/Mr. D. Werrett
6.00	Free time	
7.00	Drinks	
7.30	Dinner	

Sunday, 16 October

Chairman: Dr. B. Sheard

9.00 am	Company Presentations:	
	Amersham	
	Cellmark	
	Cetus	
	Collaborative Research	
	Fresenius	
	Lifecodes	
10.30	International compatibility and Databases: a review of the issues	Mr. P. Martin
11.00	Coffee	
11.30	Panel Discussion (panel to be decided later)	
1.00	Lunch	
2.00	Discussion Session (forensic scientists only)	

1st EDNAP MEETING 1988

AGENDA

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1st EDNAP MEETING 1988

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EDNAP - FOUNDERS

Viv Emerson

Brian Sheard

Brian Parkin

Dave Werrett

Birthe Eriksen

Bernd Brinkmann

Herman Schmitter

Prof. Rossi

Ernesto **d'Aloija**

Ate Kloosterman

Walter Bär

PETER MARTIN

Thank you so much for the invitation to the EDNAP etc meeting. It is with regret that I will not be able to attend. There are many reasons but the major difficulty is getting from our home (in the middle of the French countryside) to Innsbruck which would require at least one extra overnight. This of course makes it very costly.

So , while I would love to meet some old friends and hear about progress I must decline your kind invitation.

Very best wishes for an excellent meeting.

Pete

DAVE WERRETT

ATE KLOOSTERMAN

BERND BRINKMANN

thank you for your mail.
Since I am a real founding member of the EDNAP and the creation of the name happened in Münster, I would have loved to come to the meeting in the alps.
Unfortunately I am unable to come, because I am only back from a journey to South Africa on November 5th.
Therefore, please understand, that I cannot.
In my professional life, the EDNAP and all the surrounding activities were really some of the greatest successes.
Of course, I would have loved to have seen you again and some of the old gang.

Many greetings
Yours
Bernd

WALTER BÄR

Thank you very much for the invitation. It would have been nice to remember the good old times but unfortunately I have a serious schedule problem which I cannot resolve. So I will let you celebrate „alone“ but I remember the successful times we had together.
I hope that everything goes well at Copenhagen and for yourself.
Very best wishes
Walter

HERMAN SCHMITTER

congratulations to EDNAP and its very successful 30 years. For I have‘nt lost contacts to my former collegues at BKA, i knew EDNAP still is working fine and I‘m sure it will do so for the following decades.
It‘s a pity, but for private reasons I‘m not able to follow your very kind invitation. Thus, it‘s left to me just to wish you a good and successful meeting -as EDNAP meetings allways used to be- and much pleasure by celebrating the 30 years anniversary.

Best wishes and kindest regards to all the collegues and friends of the EDNAP group.

Hermann

ERNESTO **d'ALOJA** 1993

BIRTHE ERIKSEN

PATRICK LINCOLN

30 years of EDNAP….it seems a long time ago in many ways…so many things happen/change but I remember so well us all sitting in a room and trying with Christian Rittner to come up with a name for the group and him saying EDNAP, EDNAP yes that sounds good!!
Thank you very much for your kind invitation to attend the Dinner and EDNAP meeting in Innsbruck in October. We really don't travel abroad these days, age I fear is catching up one way and another so I don't think I shall be accepting your invitation to attend. Sorry about that, I'm sure it will be a great occasion and certainly Innsbruck is a lovely place to have it not to mention the very attractive looking hotel!!
With warmest good wishes
Patrick

CHRISTIAN RITTNER

MAINZ 1991

VINCE PASCALI

VENICE 1993

MAUREEN SMYTH

As a former working forensic scientist, I want to acknowledge the invaluable contribution EDNAP made to the field. It was the arena where theories could be formulated, tested and argued before being let loose on the world!!

A toast to you all.

Best regards

Maureen

BRAMSHILL 1990

SANTIAGO DE COMPOSTELA 1992

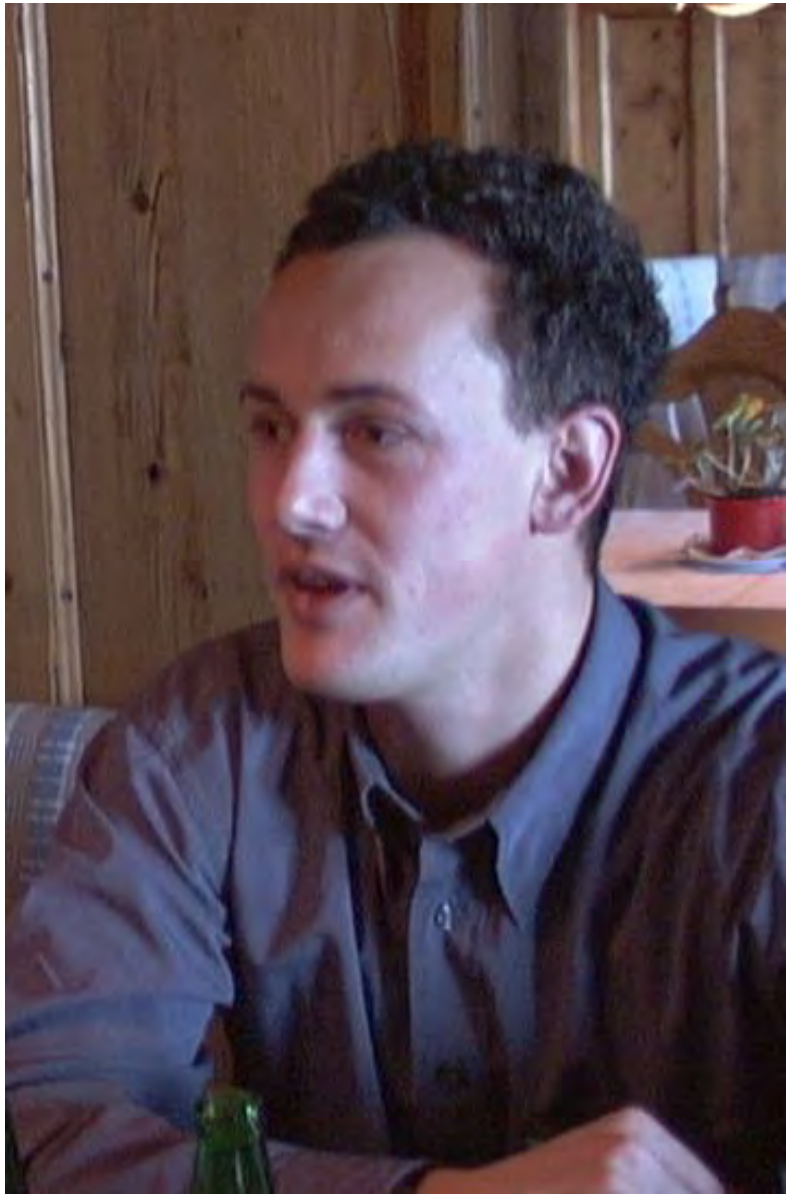
OSLO 1992

COIMBRA 1994

INNSBRUCK 1998

INNSBRUCK ENFSI 1999

INNSBRUCK ENFSI 1999



DUBLIN 2000

Methylated DNA & Age Exercise

David Ballard

EDNAP, Innsbruck 2018

KING'S College LONDON

EDNAP Exercise

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

Manuscript

Figure 1 – Laboratory prediction of samples A-F

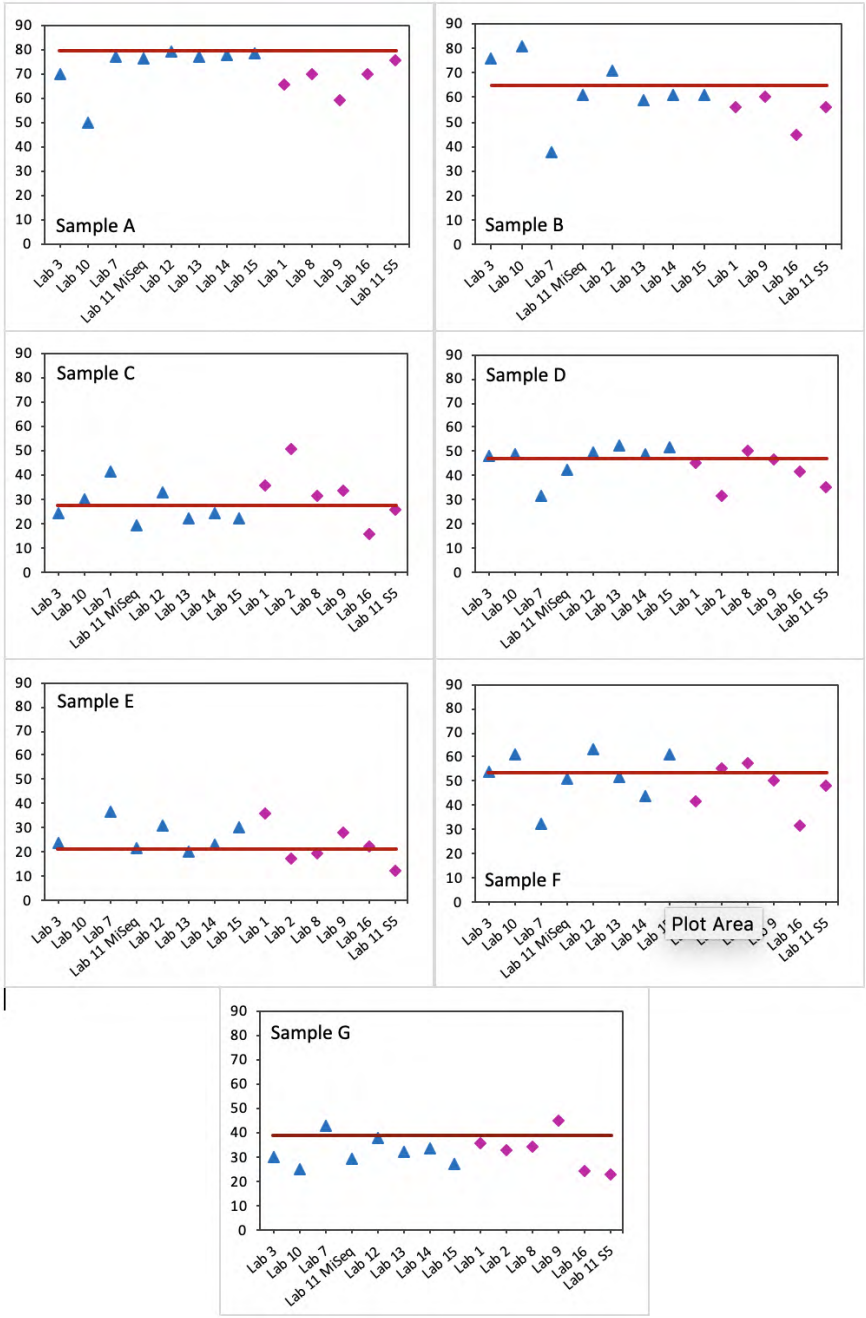
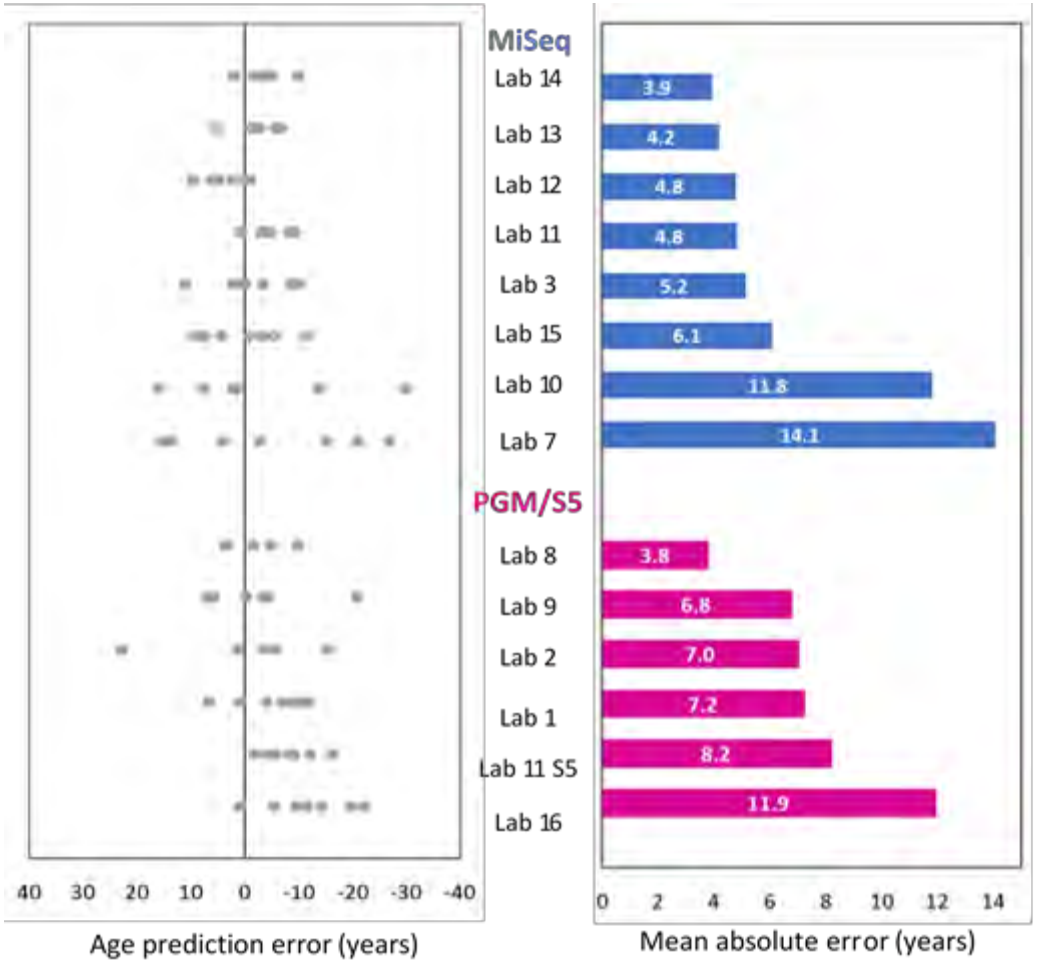


Figure 2 – Laboratory prediction of samples A-F

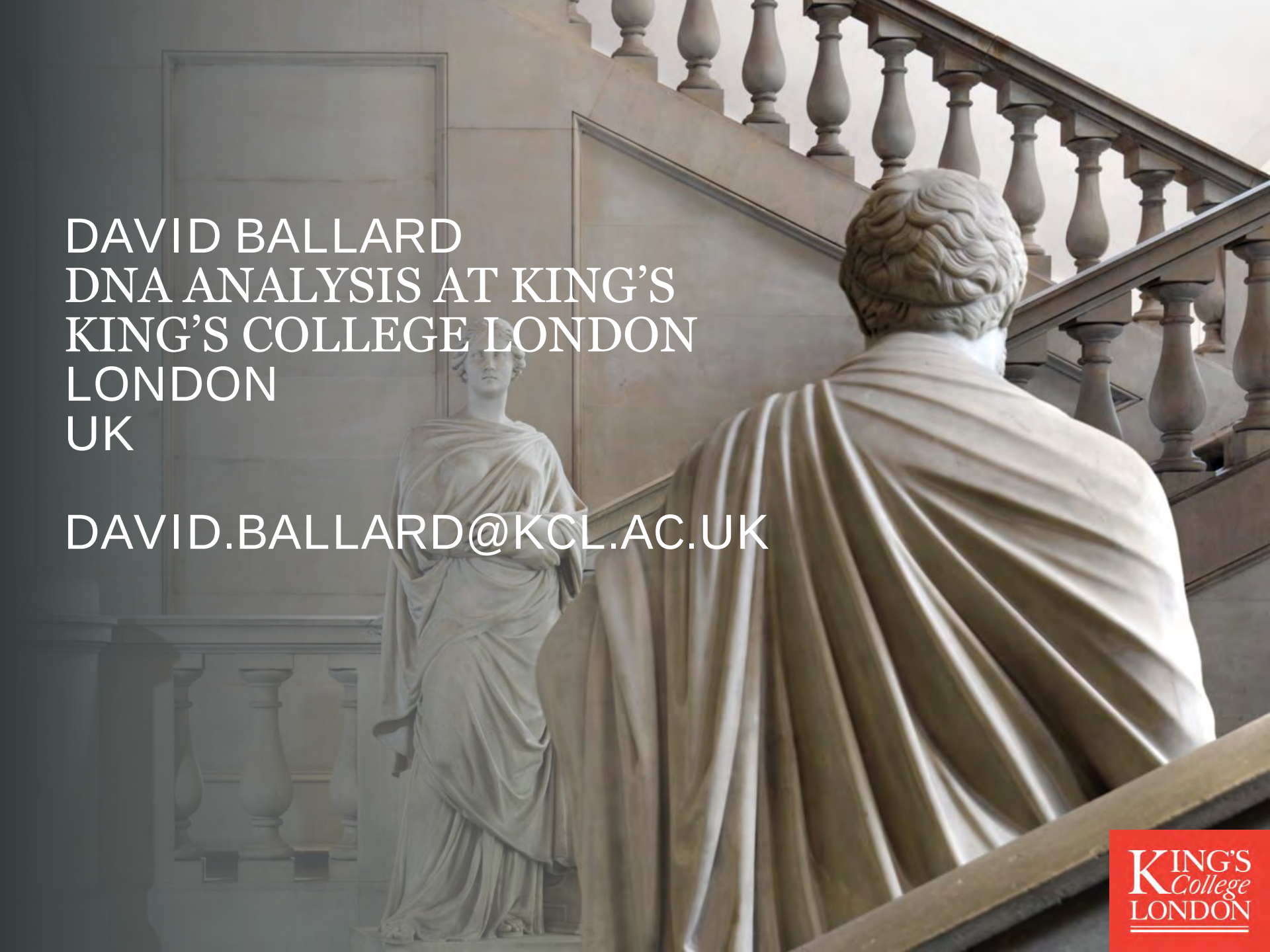


Manuscript

- Introduction
- Methods
- Results Phase 1
  - Methylation control reproducibility per lab and marker
  - Methylation control differences MiSeq vs PGM/S5
- Results Phase 2
  - Age prediction reproducibility per lab
  - Blind prediction samples
- Discussion/conclusion

Acknowledgments

- Anastasia Aliferi
- Athina Vidaki
- Leon Barron
- Denise Syndercombe Court



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

DAVID.BALLARD@KCL.AC.UK

KING'S College LONDON

University of Zurich[UZH]

Zurich Institute of Forensic Medicine

EUROFORGEN / EDNAP mRNA NGS exercise 2

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

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

30. October 2018, Innsbruck

EUROFORGEN Network of Excellence

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

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

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research paper

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

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

Targeted mRNA sequencing: Proof of concept

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

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research paper

Predicting the origin of stains from next generation sequencing mRNA data

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

Probabilistic model

Collaborative exercise mRNA NGS part 1

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

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research paper

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

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

EUROFORGEN Network of Excellence

Collaborative exercise mRNA NGS part 2

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

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

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

Collaborative exercise mRNA NGS part 2

Provided samples

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

Collaborative exercise mRNA NGS part 2

10 participating laboratories: → 1x no data

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

Collaborative exercise mRNA NGS part 2

Summary Questionnaire:

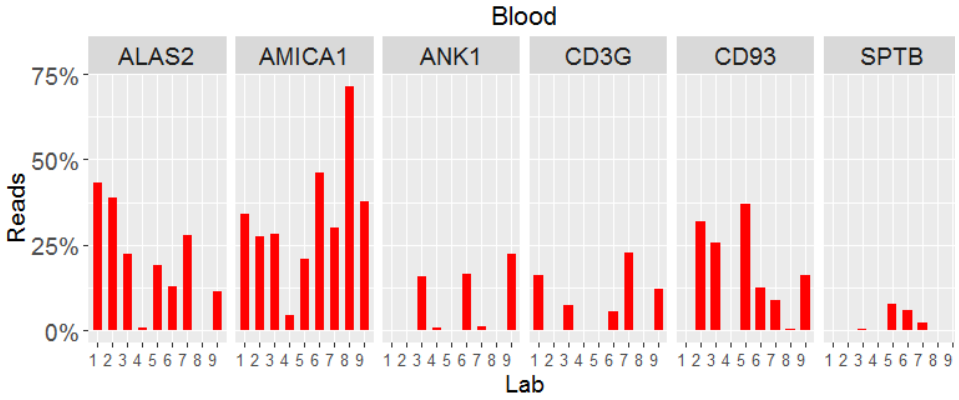
- Delivery time Fedex (samples+primers): all labs within 1-2 days
- 3x manual RNA extraction, 6x RNA extraction kit (RNeasy, mirVana)
- 8x RNA quantification (Qubit, RiboGreen, Quantus, NanoDrop), 1x no quant

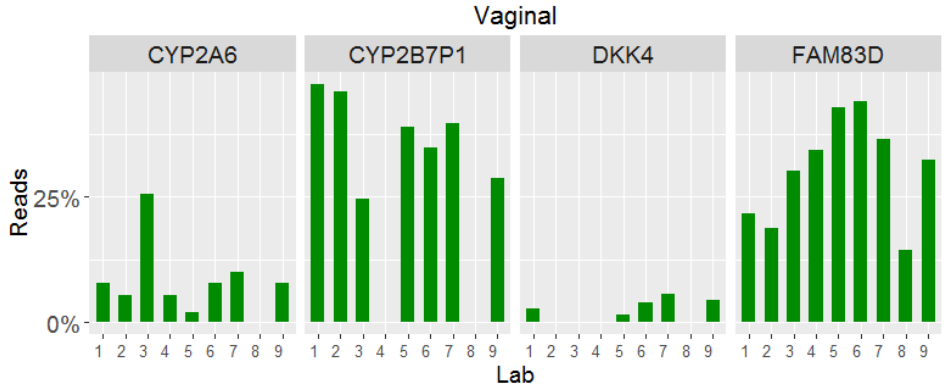
Collaborative exercise mRNA NGS part 2

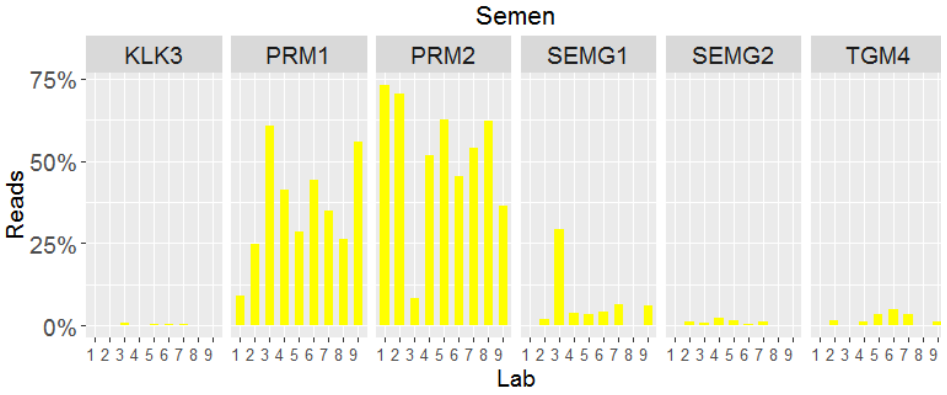
RNA-Quantification

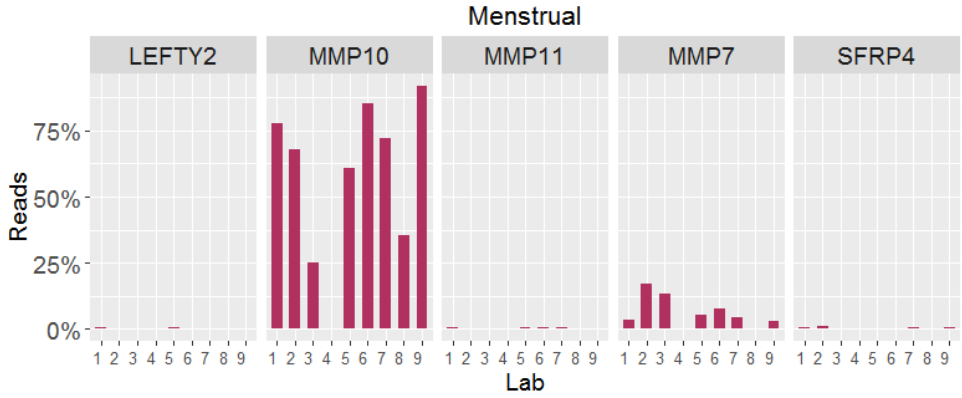
[ng/µL]	extraction method	quantification method	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16
Lab_1	Rneasy Mini	Qubit	<	<	<	292	62	<	<	<	90.4	25.6	49.8	7.2	<	4.4	122	15.7
Lab_2	Rneasy Mini	Qubit	<	<	<	>	38	<	<	<	55	29	31	<	<	<	8.67	<
Lab_3	manual	Nanodrop	81.6	390.8	64.3	624.8	109.6	70	119.3	122.1	717.2	302.6	528.6	194.5	64.3	54.6	742.6	344.9
Lab_4	Rneasy Mini	Qubit	<	<	<	<	<	<	<	<	<	<	<	<	<	<	<	<
Lab_5	Rneasy Mini	Qubit	<	<	2.4	85	7.9	<	<	<	58	34.4	25.7	2.1	<	<	87	10.7
Lab_6	manual	Quantus	22	21	18	270.5	131.5	0.97	22	8.8	251	89.85	566.5	35.75	18	11	242	68.75
Lab_7	manual	Quant-iT RiboGreen	5.4	19.7	5.8	202.5	42.8	<	14	1	205.7	129.1	210.9	11.1	8	2.6	168	139
Lab_8	mirVana	no quant	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Lab_9	Rneasy Mini	Qubit	<	<	<	94	13	<	<	<	52	7	4	<	<	2.3	198	24.6

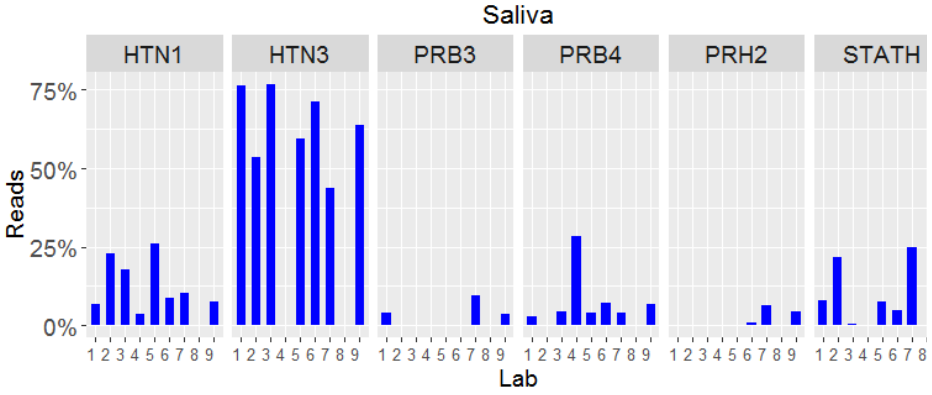
Comparison among laboratories (mRNA assay, single stains)

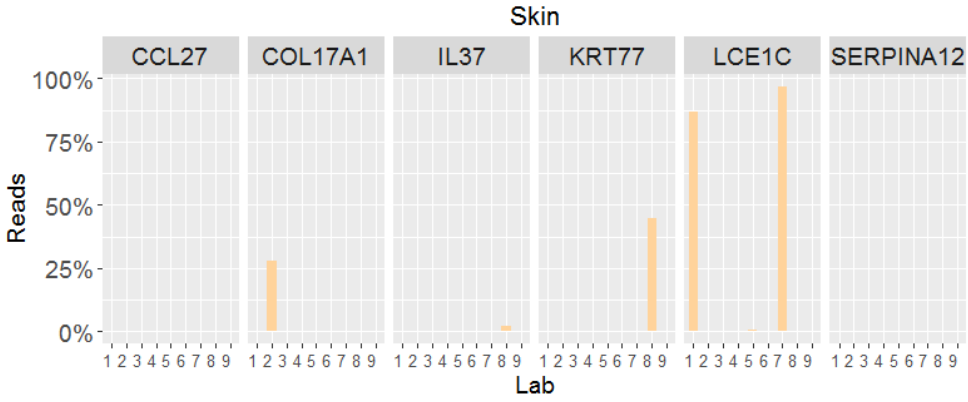


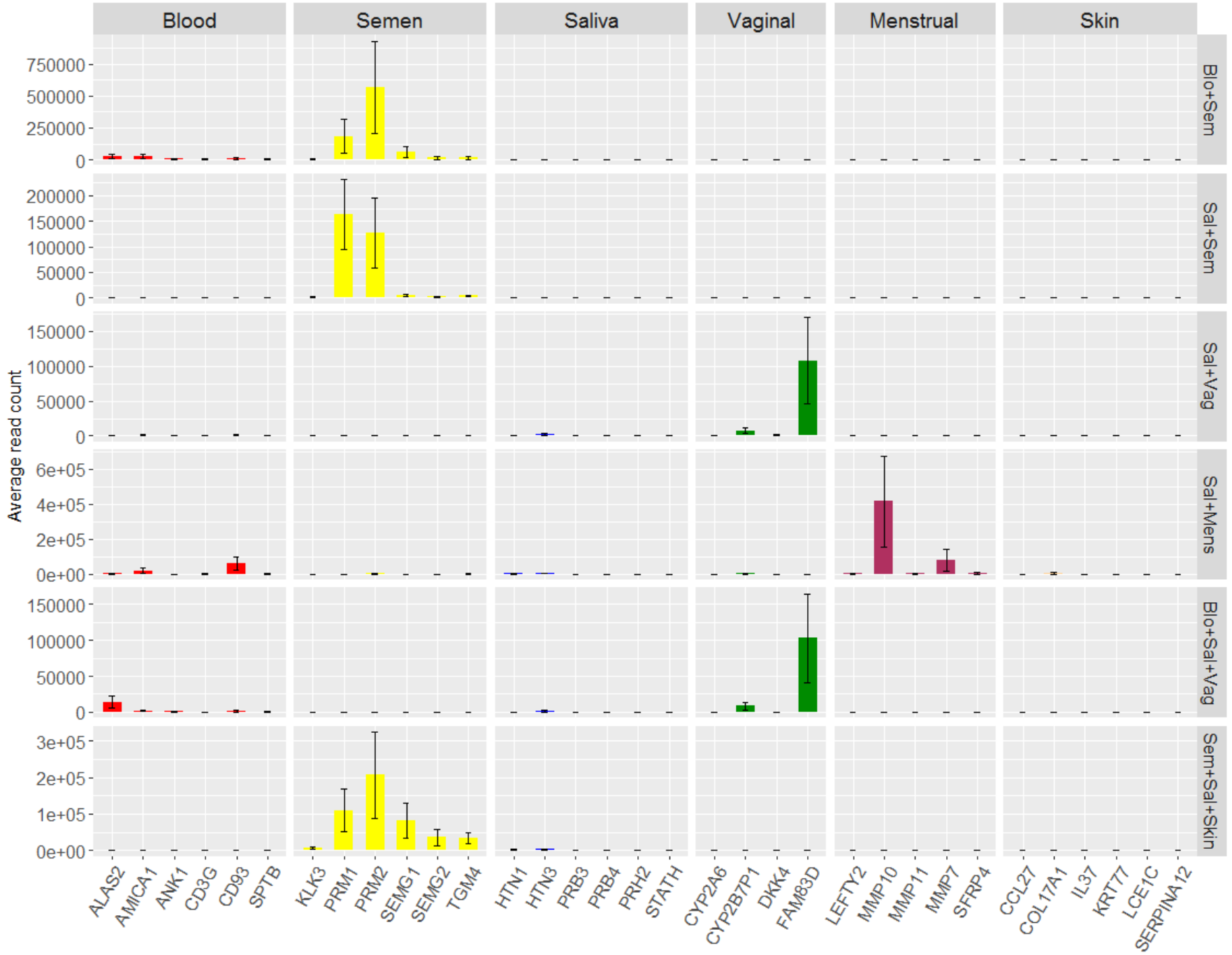




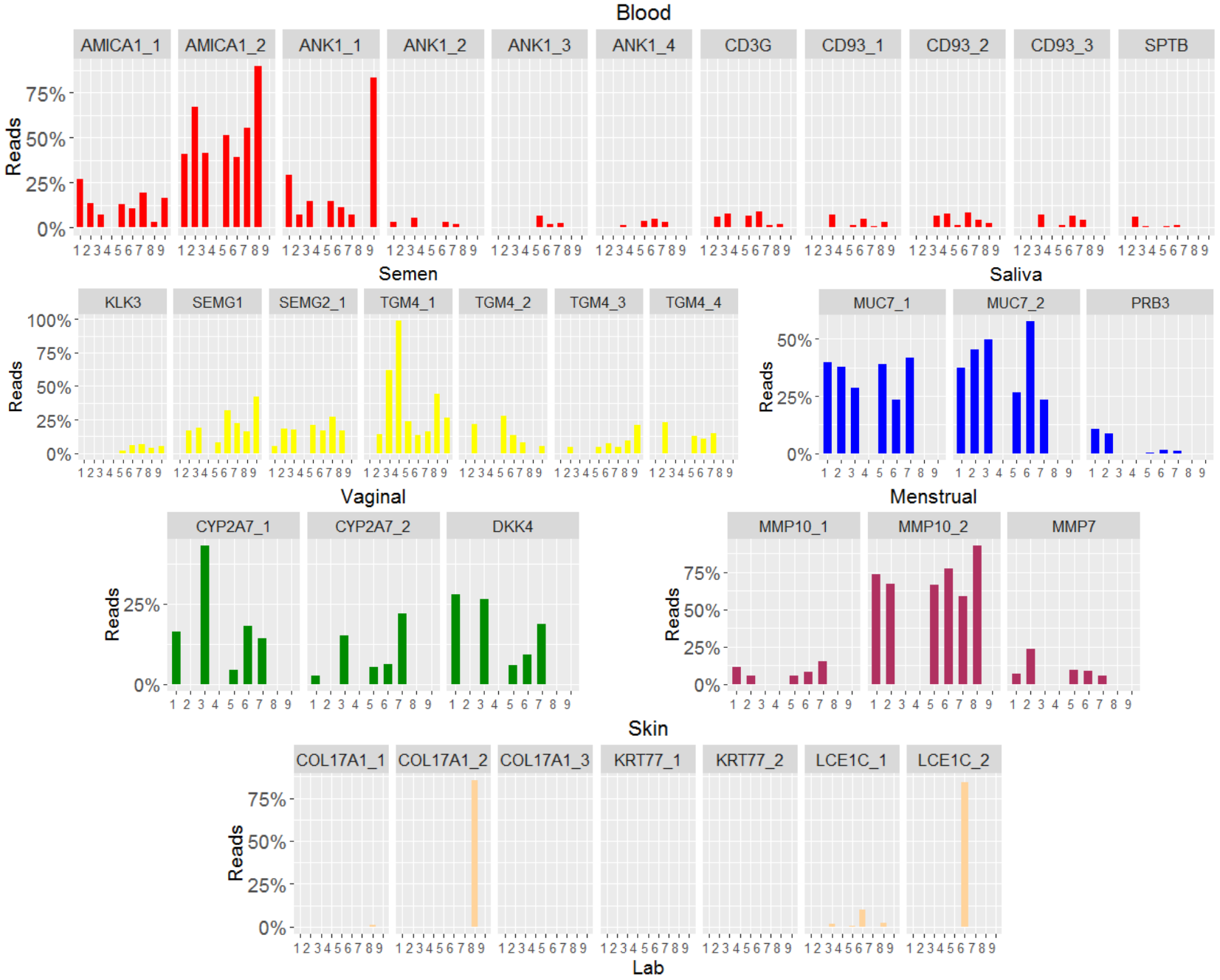







Blood
Semen
Saliva
Vaginal
Menstrual
Skin
Blo+Sem
Sal+Sem
Sal+Vag
Sal+Mens
Blo+Sal+Vag
Sem+Sal+Skin
Average read count
ALAS2
AMICA1
ANK1
CD3G
CD93
SPTB
KLK3
PRM1
PRM2
SEMG1
SEMG2
TGM4
HTN1
HTN3
PRB3
PRB4
PRH2
STATH
CYP2A6
CYP2B7P1
DKK4
FAM83D
LEFTY2
MMP10
MMP11
MMP7
SFRP4
CCL27
COL17A1
IL37
KRT77
LCE1C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

Comparison among laboratories (cSNP assay, single stains)



Overview - forensic stain analysis

DNA (STR profiling)

who is the donor of the DNA?

- **Enzymatic and immunological tests (presumptive tests)**
- **mRNA profiling**
- **miRNA profiling**

what type of cellular material is present?

- **cSNP profiling**

who contributed which body fluid in mixtures?

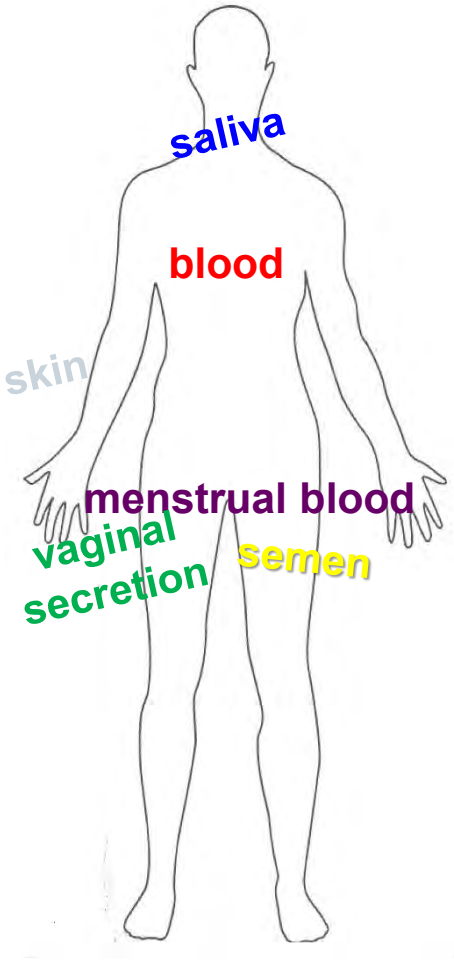
cSNPs = coding region SNPs

Body fluid / tissue specific SNPs

- In body fluid specific RNA we looked for SNPs that discriminate European individuals the most
- 34 off-the shelf cSNPs (MAF from 0.48 – 0.06)
- 3-11 markers per body fluid

cSNPs give a direct link between donors and body fluids

→ Identification of individual and body fluid



cSNP overview

Allele frequencies estimated from 188 European individuals

Body fluid	SNPs	PM
Blood	11	3.88E-4
Semen	7	2.77E-3
Skin	7	2.66E-3
Saliva	3	2.58E-1
Vaginal	3	1.76E-1
Menstrual	3	1.36E-1

Match probability per body fluid

cSNP profile – toy example

Blood - saliva mixture

cSNP stain profile (from RNA) only shows genotypes for blood and saliva markers

cSNP1	cSNP2	cSNP3	cSNP4	cSNP5	cSNP6	cSNP7	cSNP8	cSNP9	cSNP10
AA	AG	GT	CG	AT	GG				

Stain

cSNP reference profiles (from DNA) show genotypes for all markers

cSNP1	cSNP2	cSNP3	cSNP4	cSNP5	cSNP6	cSNP7	cSNP8	cSNP9	cSNP10
AA	AG	GT	GT	AA	CG	AG	CT	CC	GT

Donor 1

cSNP1	cSNP2	cSNP3	cSNP4	cSNP5	cSNP6	cSNP7	cSNP8	cSNP9	cSNP10
AG	AA	GG	CG	AT	GG	AG	CT	CC	GT

Donor 2

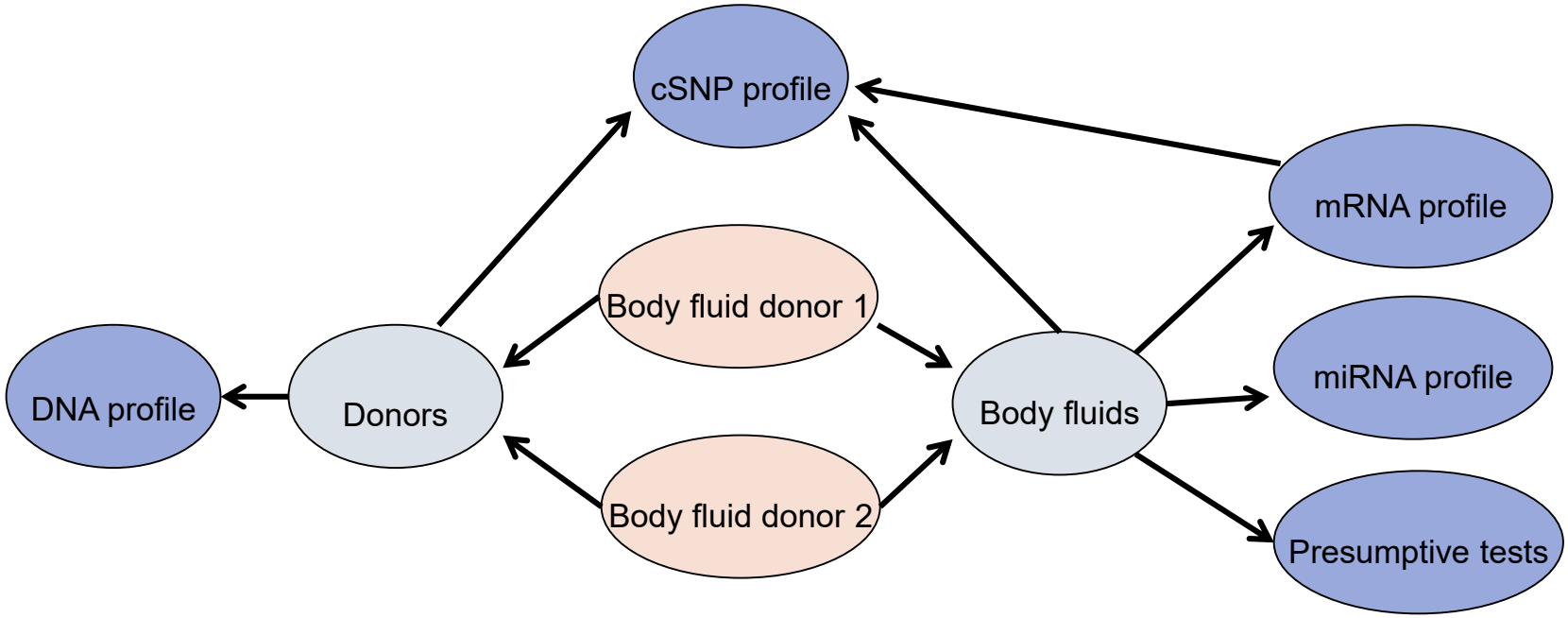
cSNP profile – a real data example

Blood – saliva mixture (blood 5 SNPs, saliva 1 SNP, remaining 6 blood and 2 saliva SNPs not discriminatory for this example)

Sample/ Marker	ANK1_2	ANK1_3	ANK1_4	AMICA1_1	CD3G	MUC7_2
Donor 1 (DNA)	GG	CT	GG	AA	AA	CG
Donor 2 (DNA)	GA	CC	GA	AG	AG	CC
blood-saliva mix (RNA)	GG G: 947 A: 4	CT C: 1604 T: 2940	GG G: 8619 A: 4	AA A: 9489 G: 32	AA A: 6522 G: 13	CC C: 2073 G: 1

Combine evidence on different levels with a Bayesian network

- DNA profile assists in identifying the donors
- RNA profile/presumptive tests assist in identifying the body fluids
- cSNP profile helps to link a donor to a specific body fluid
- Additional case related information



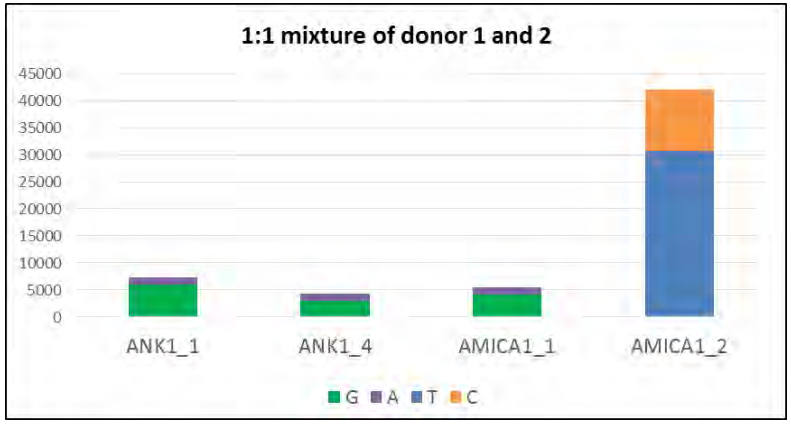
cSNP profile – mixture of same body fluid

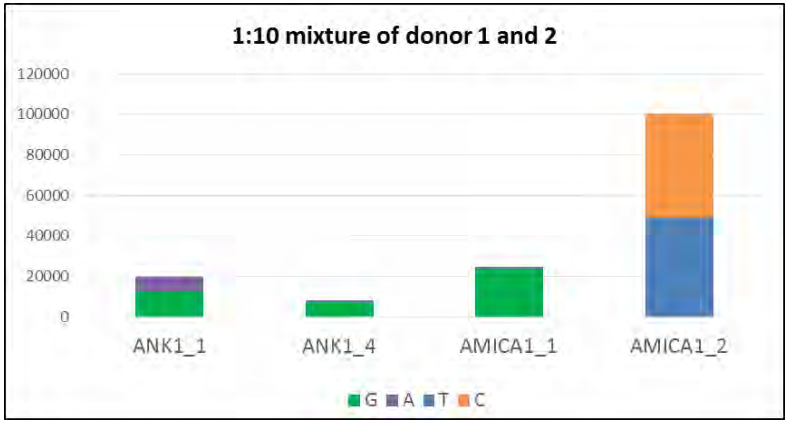
Real data example: blood – blood mixture

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

	Sample/ Marker	ANK1_1		ANK1_4		AMICA1_1		AMICA1_2	
1x	Donor 1 (DNA)	G	G	G	A	G	A	T	T
10x	Donor 2 (DNA)	G	A	G	G	G	G	T	C

Read counts reflect the mixture proportions





With only one donor we would expect a 1:1 ratio for heterozygous SNPs

cSNP discussion

- Analysis of RNA/cSNP in stains is challenging
  - RNA easily degrades - some alleles drop out
  - Residual DNA in RNA extract - markers for body fluids not present “drop in”
  - Heterozygote read count ratio can deviate from an expected 1:1 ratio, due to stochastic amplification processes with low template targets
  - → Inconsistencies between reference profiles and stain/mixture profile
  - → Not useful for mixture deconvolution
- Combining evidence – DNA, RNA and cSNP
- Need more suitable markers → Simultaneous identification of individual and body fluid

Manuscripts in preparation

1) cSNP proof of concept paper
   - 12 single source samples, 51 mixtures
   - STRs, mRNA, cSNPs
   - Statistics

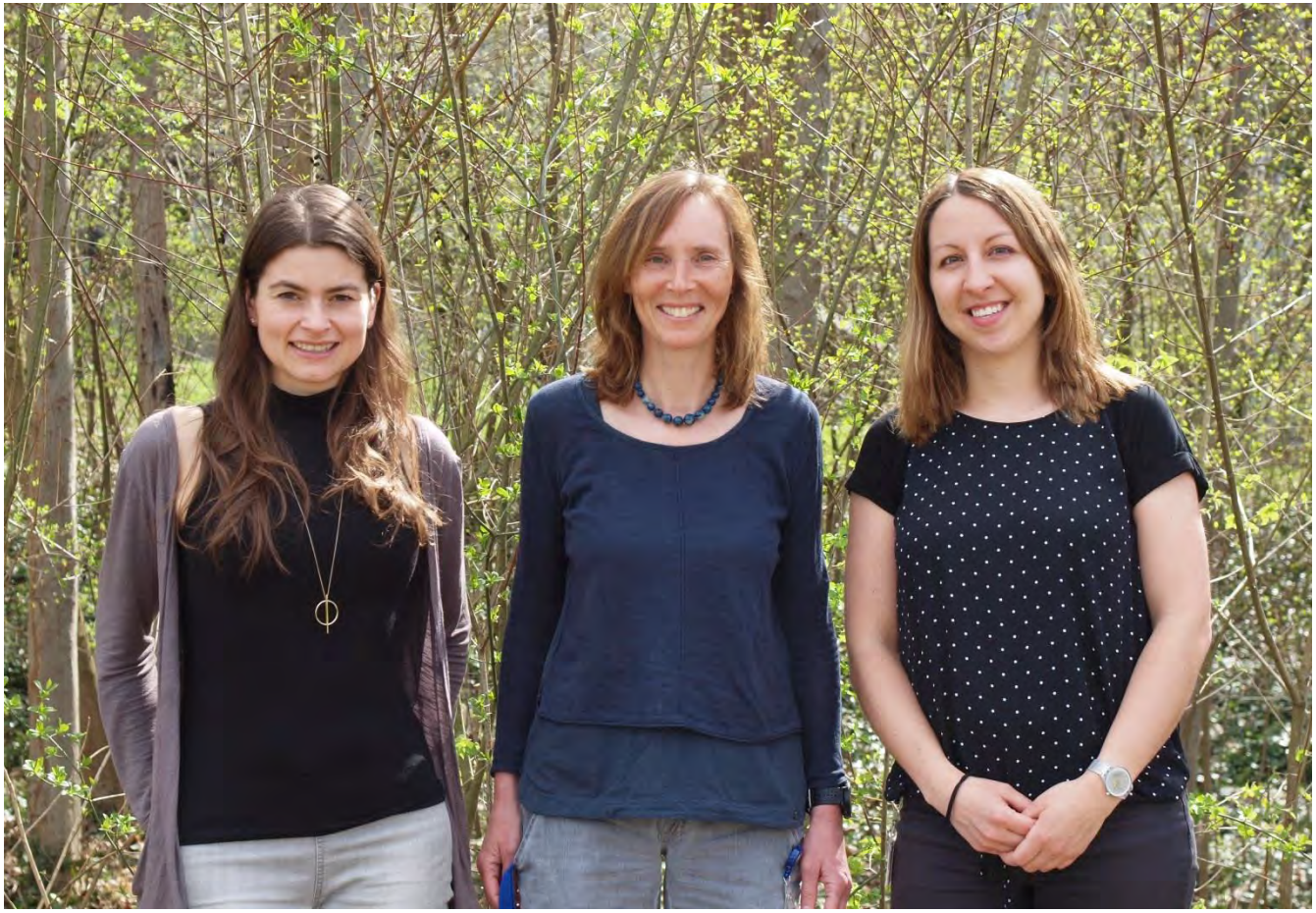
2) Collaborative exercise mRNA NGS part 2
   - 10 labs, 9 data sets
   - 16 samples
   - TOP6 and cSNP data
   - Statistics

Thermofisher cSNP assay

- blood, semen, saliva, vaginal, menstrual and skin
- cSNP amplicons are useful for body fluid identification
- some marker overlap between the MiSeq cSNP assay and TF assay
- first trial experiment → genotypes can be distinguished for most of the body fluids, even amongst a small number of donors → good discrimination at the cSNP level

→ Possible collaborative exercise in 2019

Thank you for your attention!



Netherlands Forensic Institute
Ministry of Justice

EDNAP mini-Exercise proposal mtDNA quant

Kris van der Gaag
Natalie Weiler
Titia Sijen
Arnoud Kal

Calibration of equipment

The exercise assay uses four colors, you will need to calibrate your equipment for the following labels as shown below. Unfortunately we cannot supply these reagents.

Label	Catalog nr. ThermoFisher Scientific	Description
FAM / VIC	4349180 (usually calibrated during service)	7500 Real Time PCR Systems Spectral Calibration Kit I
ABY	A24738	ABY™ Dye Spectral Calibration Plate for Multiplex qPCR, 96-well
JUN	A24737	JUN™ Dye Spectral Calibration Plate for Multiplex qPCR, 96-well
Mustang Purple	4461599	MUSTANG PURPLE™ Dye Spectral Calibration Plate, 96-well

What we need from you

We will need one contact person for each lab that will join the exercise. Please send us the postal address and the email address of your contact person via email to a.kal@nfi.minvenj.nl, k.van.der.gaag@nfi.minvenj.nl and n.weiler@nfi.minvenj.nl . Your contact person will be notified when the samples are shipped.

Questions?

Please do not hesitate to ask us if anything is unclear.

Best wishes,

Arnoud, Kris, Natalie and Titia

Dear Friends,

The mini-exercise mtDNA quantification has been delayed for several reasons. My apologies to all of you who have been waiting for news and answers to questions. Fortunately we can give you more detailed information now. We are planning to send out the exercise within the next few weeks.

Setup of the assay

We developed a DNA-quantification assay using four different targets:

- autosomal DNA
- Y-chromosomal DNA
- mitochondrial DNA (long fragment 217 bp)
- mitochondrial DNA (short fragment 70 bp)

We will provide primers, probes, protocols and interesting samples. You can also test your own favorite samples and your own quantification method(s).

Setup of the exercise

In the exercise, you will test the following samples with the exercise protocol and reagents and also with your own protocol and reagents:

- 8 samples provided by the NFI (in triplicate)
- 8 standards, provided by the NFI (in duplicate)
- 4 samples, provided by you

We will also ask you for your interpretation of the quantification results. What do these results tell you about the sample?

Shipment of samples

To save costs, we will send you the samples and reagents via regular mail instead of frozen on dry ice. Templates are stable at high concentration, you will be asked to prepare dilutions in your own lab.

Netherlands Forensic Institute
Ministry of Justice

Update ENFSI DNA Expert Working Group activities

Alexander Kneppers

Chair ENFSI DNA Expert Working Group

NFI Division Biological Traces

ENFSI update EDNAP Innsbrück 2018

ENFSI Permanent Members

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EXECUTIVE

QCC

R&D

ADVISORY

Working Groups

Task Forces

SCIENTIF. & OPER.

EAFS

OOSs

EVENTS


1992
MUTUAL
INTEREST
1993
1ST
MEETING
11 LABS
OCT. 1995
FOUNDING
MEETING
1997
1ST EAFS
MEETING
1999
CONSTITU
-TION
WEBSITE
2002
SECRETARY
MEMBERS
FEE
2009
MONOPOLY
STATUS IN
EUROPE
2016
LEGAL
ENTITY

69 MEMBERS IN 36 COUNTRIES

EVERY MEMBER HAS TO:

ORIGINATE FROM A COUNCIL OF EUROPE MEMBER STATE

COVER 50 % OF THE WORKING GROUPS

CREDIBLE STATUS IN THE COUNTRY

MORE THAN 25 REPORTING OFFICERS

ACCREDITATED AGAINST ISO17020 / ISO17025 OR …

… HAVE THREE YEARS TO ACHIEVE THIS

SYNERGY IN NETWORKING

Zeno Geradts
The Netherlands
Forensic Information Technology

Jan Eric Grunwald
Germany
Textile and Hair

Michael Braune
Germany
Marks

Aldo Mattei
Italy
Fingerprint

Sander Kneppers
The Netherlands
DNA

Dagmar Boss
Germany
Forensic Speech and Audio Analysis

Irene Kuiper
The Netherlands
Animal Plant and Soil Traces

Camilla Lilleng
Norway
Fire and Explosion

Andreas Rippert
Switzerland
Documents

Florin Rusitoru
Romania
Road Accident Analysis

Chris Moynehan
UK
Firearms/GSR

Matthew Beardah
UK
Explosives

Irene Breum
Denmark
Drugs

Jonathan Morris
Scotland, UK
Handwriting

Fernando Viegas
Portugal
Scene of Crime

Ulrich Simmross
Germany
Paint and Glass

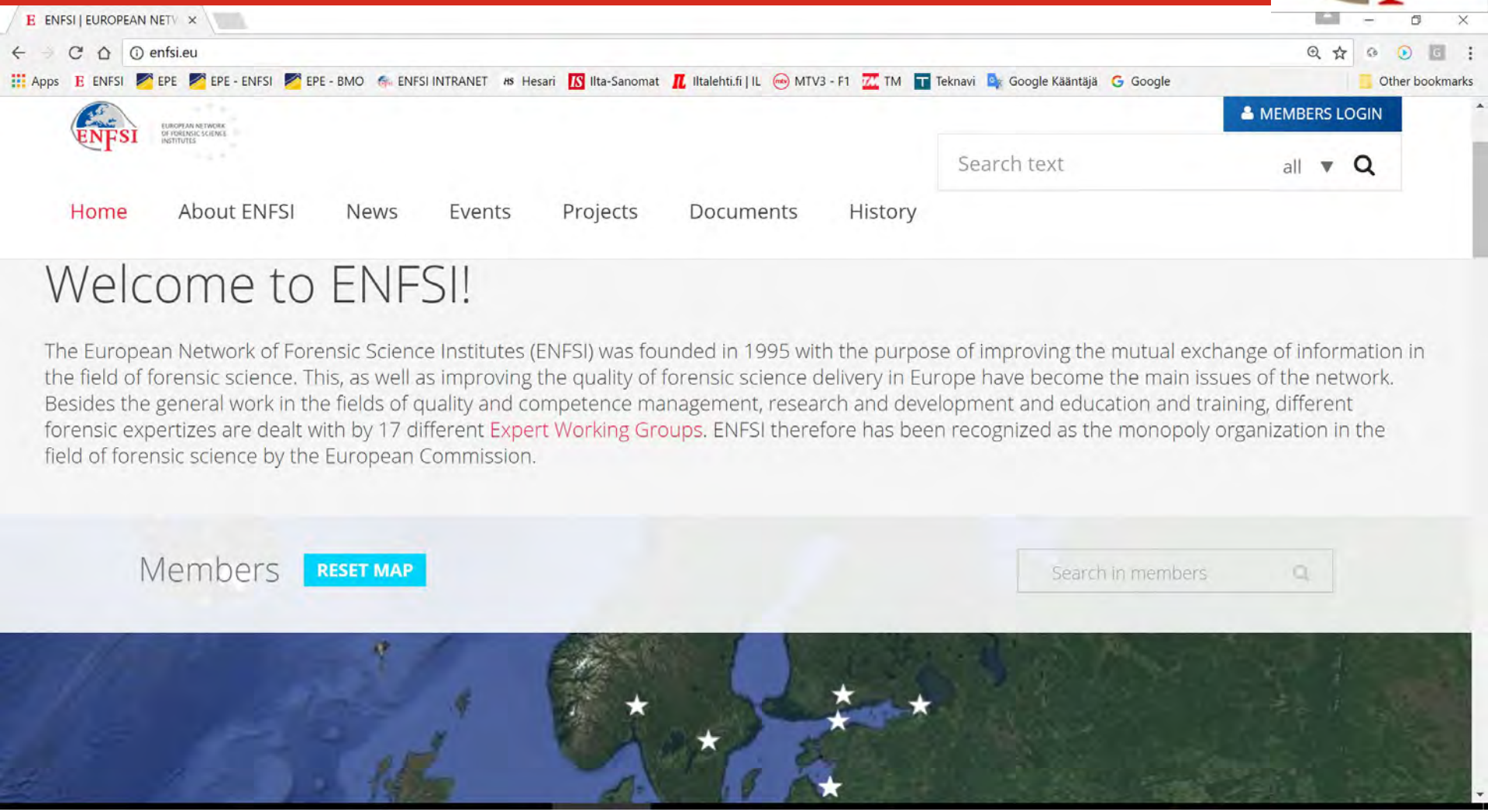
Martin Wermuth
Switzerland
Digital Imaging

> 1000

Forensic Experts

17 WORKING GROUPS

COMMUNICATION



EXTERNAL: WWW.ENFSI.EU

COMMUNICATION

EUROPOL PLATFORM FOR EXPERTS | EUROPEAN NETWORK OF FORENSIC SCIENCE INSTITUTES

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- Monopoly Programmes
- Forensic Wiki
- Wiki
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- User Directory
- ENFSI Public Site

Animal, Plant and Soil Traces EWG

Digital Imaging WG

DNA EWG

Documents EWG

Drugs EWG

ENFSI - Permanent Members

ENFSI - European Network of Forensic Science Institutes

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.

Quick Navigation

ENFSI main site - Permanent Members

Standing Committees: Quality and Competence SC - Research & Development SC

Working Groups: Animal, Plant and Soil Traces - Digital Imaging - DNA - Documents - Drugs -Explosives - Fingerprint - Fire and Explosions Investigation - Firearms/GSR - Forensic Information Technology - Handwriting - Marks - Paint & Glass - Road Accident Analysis - Scene of Crime - Speech and Audio Analysis - Textile and Hair

Contact

For any help please contact the site administrators Isabelle Jopp or Bianca Benisch from the ENFSI Secretariat by sending an e-mail to secretariat@enfsi.eu

Quick Links

- Reimbursement rules & Claim form
- Access Policy of the ENFSI intranet & Annex I (user rights)

INTERNAL: EPE.EUROPOL.EUROPA.EU

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 Guiness, 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 meetings

- Annual meeting of the working group in association with
  - the European CODIS meeting
  - EDNAP meeting (European DNA Profiling group)
- Annual ENFSI board meeting with workgroup chairs
- Two steering committee meetings DNA working group

DNA working group subgroups and co-chairs

Group A: Quality Assurance

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

Group B: DNA Analysis Methods & Interpretation

> Antonio Alonso (Spain) and Walther Parson (Austria)

Group C: DNA Database and Legislation

> Dyan Daly (Ireland) and Izanda Puncule (Latvia)

Group D: Automation & LIMS

> Christina Forsberg (Sweden) and Shazia Khan (UK)

Group E: Forensic Biology and casework

> Arnoud Kal (Netherlands) and Ricky Ansell (Sweden)

(Associate) Members DNA working group

Returning guests > associate members

Currently 80 labs are participating in the DNA working group

- 58 member laboratories
- 22 associate member laboratories

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

Host Scientific Police Service of the Italian National Police

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

BPM/guideline principles ENFSI

Official BPM and guideline templates approved by the board (12/2017)

- BPM: field specific document describing the principles of the methods used, instrumentation, QA … written in general terms, aimed at practitioners
- Guideline: gives recommendations or clarifications, aimed at practitioners

BPM Human DNA Analysis

Scope agreed in 2017:

This document provides guidance on general accepted procedures and workflows for processing human DNA evidence; starting from the collection to the presentation of the findings, including requirements related to personnel, material and environment. It also refers to generally applied methods, their validation and quality control.

4 task forces

Task force	document(s)	Task
1	BPM DNA pattern recognition and comparison	Revising of the existing BPM to a guideline
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
3	Guideline: Training of DNA staff Guideline: ENFSI Quality Assurance Program for DNA Laboratories	Update the Guideline for the training of staff working in the forensic DNA laboratory Update the Guideline regarding the ENFSI Quality Assurance Program for DNA Laboratories
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

DNA EWG documents

Document Name	Responsible Group	Current Version Date	Next Review Start Date	Task force	Comments
Best Practice Manuals					
Human DNA Analysis	DNA EWG	new		2	
Validation of mixture interpretation software	DNA Analysis Methods and Interpretation	2017	2019	Subgroup B	Update and change to guideline
DNA pattern recognition and comparison	DNA EWG	2016	2018	1	Update and change to guideline
Guidelines					
Concept Training Document	QA group (input from other groups)	nov-10	2018	3A	
Quality Assurance Program for DNA Laboratories	QA group	apr-10	2017	3B	
Recommended Minimum Criteria for the Validation of Various Aspects of the DNA Profiling Process	QA group	nov-10	2017	4	
Contamination prevention guidelines	QA group	apr-17	2019	QA-group	Update to guideline
Document on DNA Database Management	DNA Database group	2017		Subgroup C	Update to guideline

EU Direct Grants - Update 2018

Dr Richard Gill

ENFSI Monopoly Programme Manager

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

Business Meeting

Annual Meeting – Budapest
18 May 2018

Current Situation – May 2018

EU Direct Grants to ENFSI

	Grant Agreement
2009	€ 499,973
2010	€ 582,113
2011	€ 646,931
2012	€ 537,982
2013	€ 645,649
2014	€ 1,425,821
2016	€ 1,500,000

> € 5.8 million

New signed Grant Agreement Implementation Started 1st Jan 2018 →

Direct Grant Programme Themes Selected by ENFSI Board

MP2009

Sustainable Quality Within European Forensic Science (SQWEFS)

MP2010

Strengthening the Evaluation of Forensic Results across Europe (STEOFRAE)

MP2011

Improving Forensic Methodologies across Europe (IFMAE)

MP2012

Towards European Forensic Standardisation through Best Practice Manuals (TEFSBPM)

MP2013

Towards the Vision for European Forensic Science 2020 (TVEFS-2020)

MP2014

Towards the Development of Pan-European Databases in Forensic Science (TDPEDFS)

MP2016

Steps Towards a European Forensic Science Area (STEFA)

Direct Grant 2016 projects (Monopoly)

Period 2018- 2019

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

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

New proposals

European Commissions invite to ENFSI

ISF Police Annual Work Programme 2018

Proposals for actions 5 and 6 of the EFSA2020 Action Plan

- Stimulate accreditation of forensic service providers and competence of forensic personnel on a voluntary basis.
- Stimulating exchange of forensic data via Prüm and improving its quality.

- Total amount € 1 500 000.
- Projects max 24 months.
- Outline for projects/team leads/members 15 November 2018 > ENFSI board
- Project submission by 21 February 2019 > ISF

ENFSI board proposals

A. Accreditation of Digital Forensics and Scene of Crime Services
B. Training of Forensic Personnel in Accreditation Matters
C. Awareness Training of Stake Holders
D. Training of Technical Assessors
E. Production of New and/or Updated Best Practice Manuals (BPMs)
F. Proficiency Testing Across Europe
G. Other Work Packages?

Next meetings (proposal)

Madrid (host Guardia Civil; Victor Esteban)

7 May 2019

EDNAP meeting

European CODIS meeting

8-10 May 2019

ENFSI DNA Working group annual meeting

Contact ENFSI DNA Working group

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

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

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

EMPOP Update

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

EMPOP

17 August 1999 San Francisco, CA, USA

18th ISFH World Congress (G. Sensabaugh)

759 members
Name change to ISFG

Elected board:

President: A. Carracedo
Vice president: B. Olaisen
Secretary: W. Mayr
Treasurer: N. Morling
Representative of the Working Groups: P. Schneider

Chris Phillips

"The DNA commission had a meeting in San Francisco and will publish in due course recommendations concerning mt-DNA" (Carracedo et al 2000)

Around this time N. Morling suggested the development of a centrally curated forensic mtDNA database

8 May 2000 Dublin, Ireland

5. Suggestion for an EDNAP mtDNA database Walther Parson

A draft description of the project is attached at Annex 7

Two mtDNA databases are already in existence; one from the FBI and the other from Germany. The proposal is that there should be an open database on the Internet that is maintained by EDNAP. The acronym EMPOP was suggested for 'The EDNAP mitochondria DNA Population Database Project'.

The pilot project, which has been set up in Innsbruck, uses software that can check input data to ensure that it is a sensible result. Statistical data on major ethnic types and sub-affiliations is included. The software also records who has accessed the database and what has been searched.

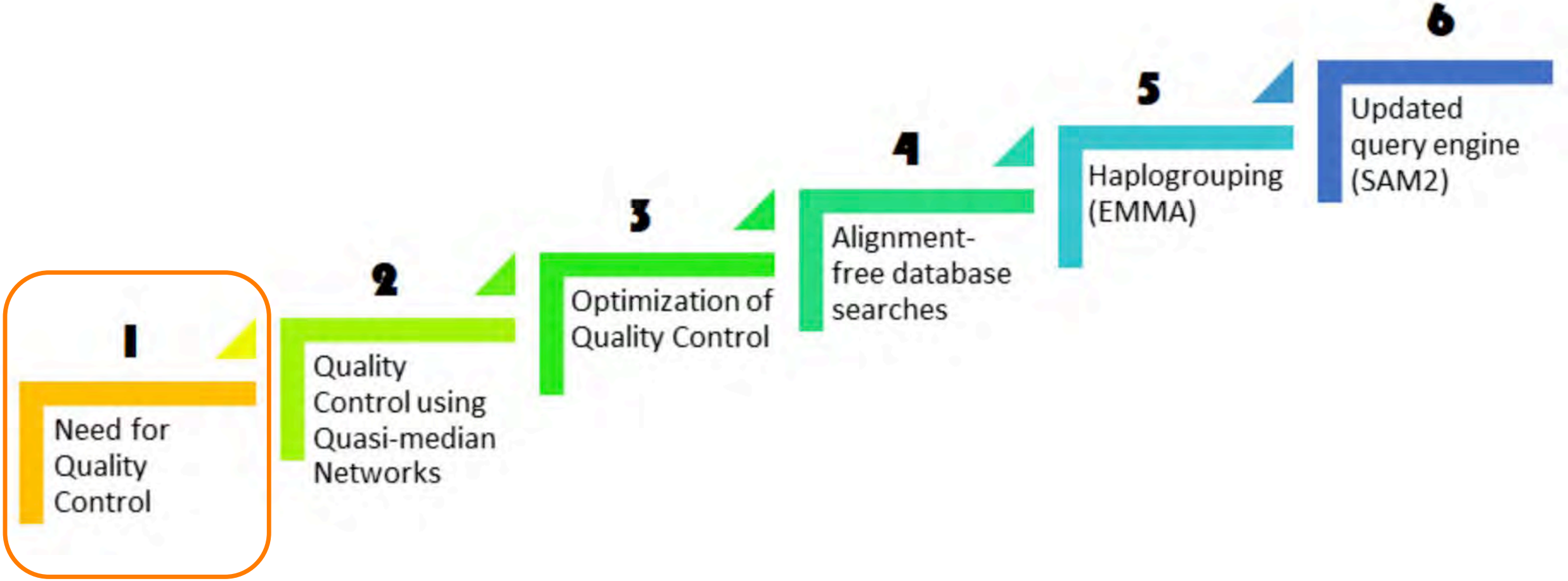
8 May 2000 Dublin, Ireland

Richard Scheithauer guarantied that the Innsbruck laboratory can maintain a mtDNA database for at least 5 years.

The members agreed that they would be happy to have EMPOP under the EDNAP umbrella.

As it is essential to maintain quality standards it might be necessary to have proficiency testing on those who will input data.

History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Optimization of Quality Control
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Alignment-free database searches
5
Haplogrouping (EMMA)
6
Updated query engine (SAM2)

High Observed Error Rate in mtDNA Data

ELSEVIER

Available online at www.sciencedirect.com

SCIENCE DIRECT®

Forensic Science International 139 (2004) 215–226

Forensic Science International

www.elsevier.com/locate/forsciint

The EDNAP mitochondrial DNA population database (EMPOP) collaborative exercises: organisation, results and perspectives

Walther Parson[a,*], Anita Brandstätter[a], Antonio Alonso[b], Nathalie Brandt[c], Bernd Brinkmann[d], Angel Carracedo[e], Daniel Corach[f], Olivier Froment[g], Ivana Furac[h], Tomasz Grzybowski[i], Karin Hedberg[j], Christine Keyser-Tracqui[k], Tomasz Kupiec[l], Sabine Lutz-Bonengel[m], Bente Mevag[n], Rafal Ploski[o], Hermann Schmitter[p], Peter Schneider[q], Denise Syndercombe-Court[r], Eric Sørensen[s], Heather Thew[t], Gillian Tully[u], Richard Scheithauer[a]

Error rate 10% - 70% clerical error

Lab-specific mutation processes

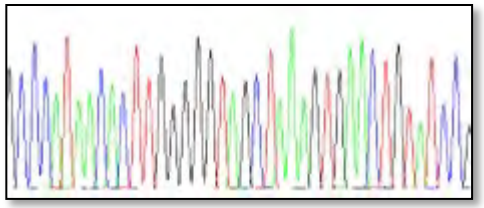
Bandelt H-J, Kivisild T, Parik J, Villems R, Bravi C, Yao Y-G, Brandstätter A, Parson W

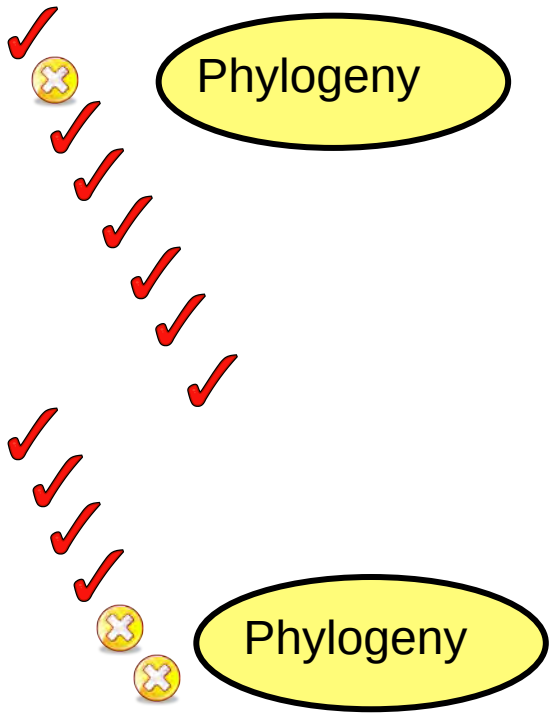
In: ***Human mitochondrial DNA and the evolution of Homo sapiens***

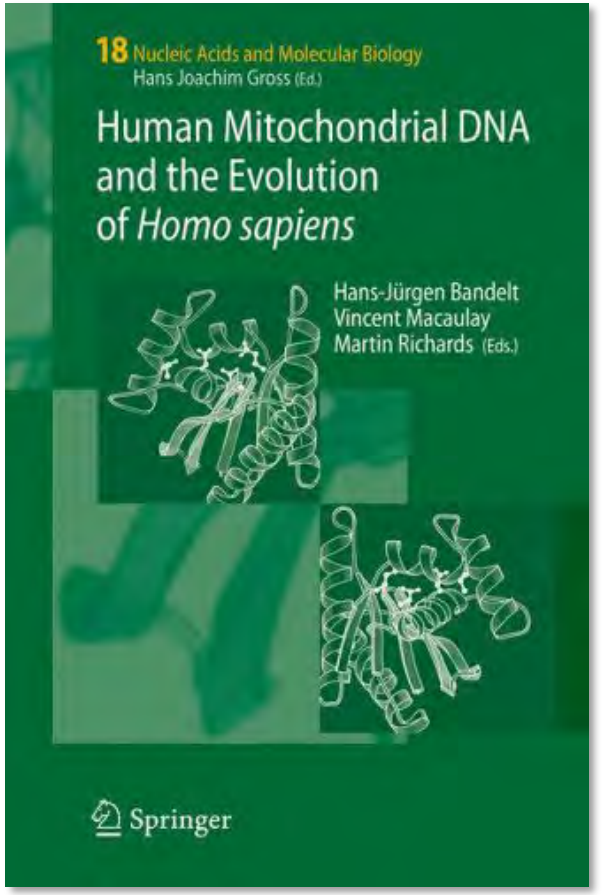
Springer-Verlag eds. Hans-Jürgen Bandelt, Vincent Macaulay, Martin Richards (2006)

Table 1 Error classification "Stage– Cause– Phenotype"

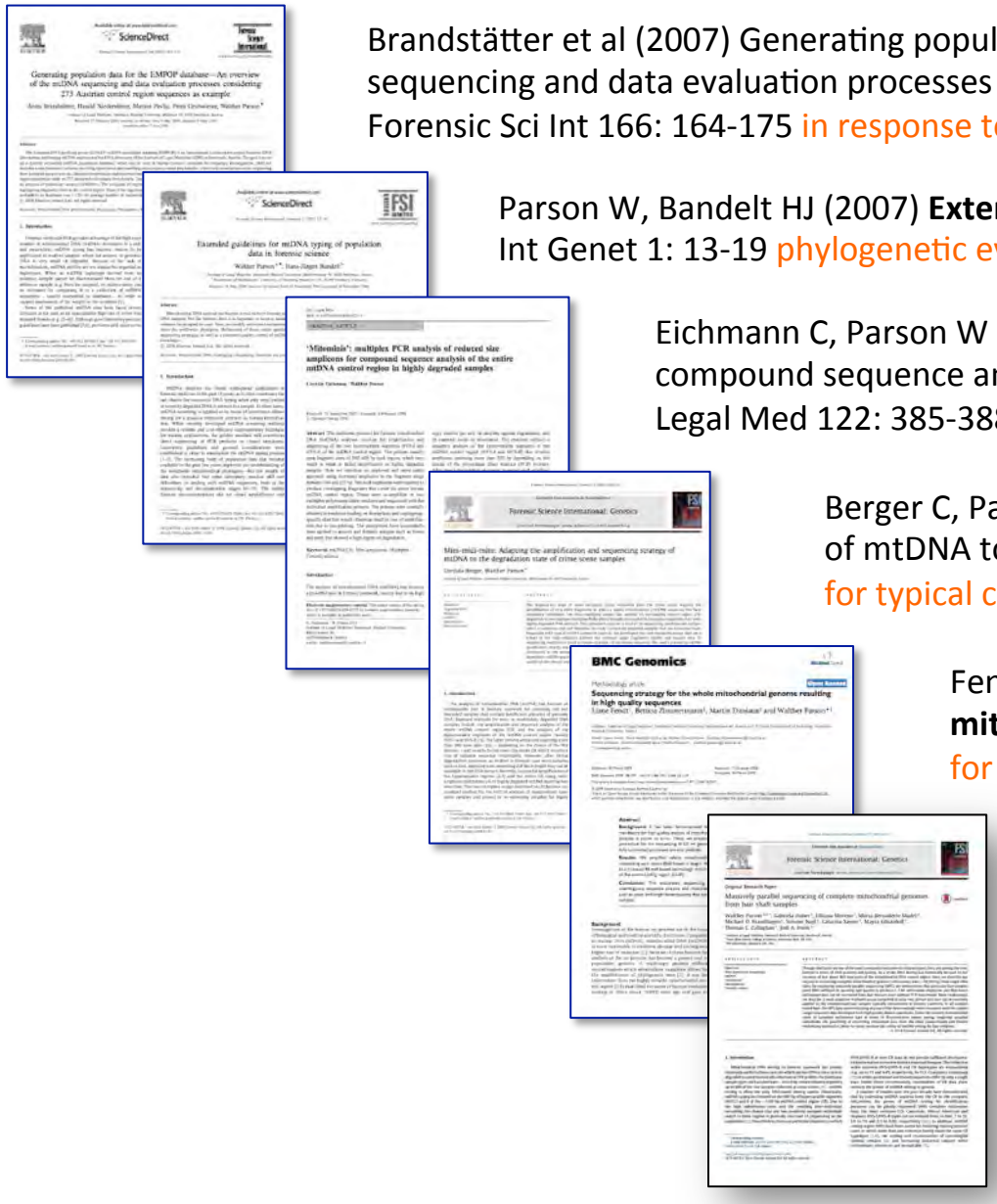
Stage	Cause	Phenotype	Process
1			DNA extraction
2			PCR
3			Sequencing
4			Electrophoresis
5			Interpretation and documentation
	C		Contamination
	M		Sample mix-up
	A		Sequencing artefact
	S		Sample manipulation and bias
	L		Misalignment or incorrect reference sequence
	B		Basecall misinterpretation
	N		Nomenclature violation
	T		Transcription error
		I	Base shift
		II	Reference bias
		III	Phantom mutation
		IV	Base misreporting
		V	Artificial recombination
		VI	Skewed variation







Updated CR/mitogenome protocols



Brandstätter et al (2007) Generating population data for the **EMPOP database** - an overview of the mtDNA sequencing and data evaluation processes considering 273 Austrian control region sequences as example. Forensic Sci Int 166: 164-175 in response to high error rate due to phantom mutations

Parson W, Bandelt HJ (2007) **Extended** guidelines for mtDNA typing of population data in forensic science. Forensic Sci Int Genet 1: 13-19 phylogenetic evaluation of phantom mutations

Eichmann C, Parson W (2008) 'Mitominis': multiplex PCR analysis of **reduced size** amplicons for compound sequence analysis of the entire mtDNA control region in highly degraded samples. Int J Legal Med 122: 385-388 for severely degraded DNA

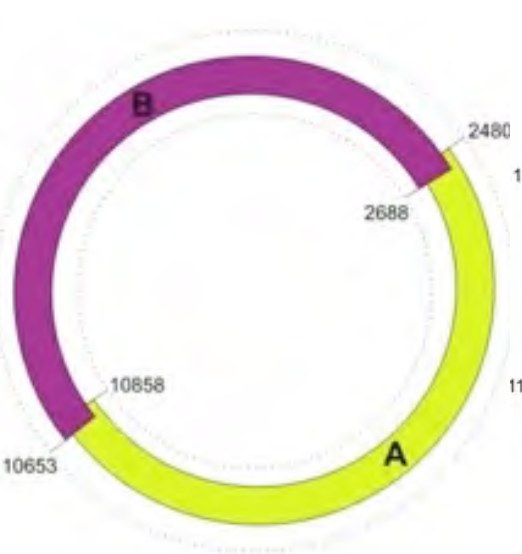
Berger C, Parson W (2009) **Mini-midi-mito**: Adapting the amplification and sequencing strategy of mtDNA to the degradation state of crime scene samples. Forensic Sci Int Genet 3: 149-153 for typical casework samples

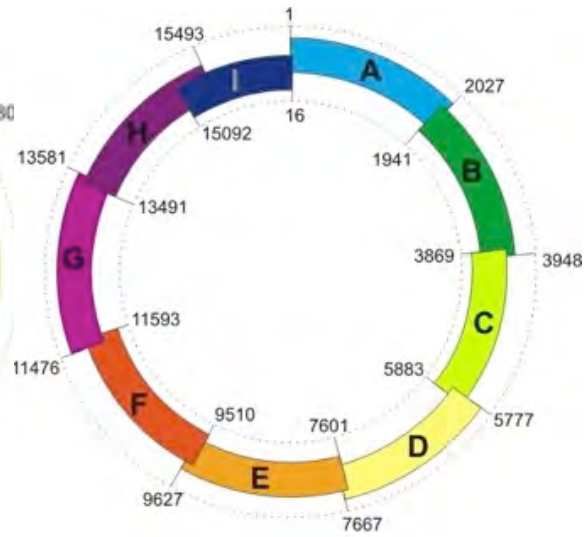
Fendt L, Zimmermann B, Daniaux M, Parson W (2009) Sequencing strategy for the **whole mitochondrial genome** resulting in high quality sequences. BMC Genomics 10: 139 for high quality DNA

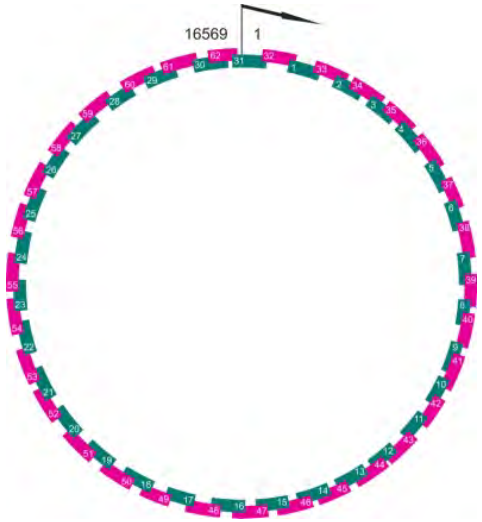
Parson W et al. (2015) Massively parallel sequencing of complete **mitochondrial genomes** from hair shaft samples." Forensic Sci Int Genet **15**: 8-15. for casework samples

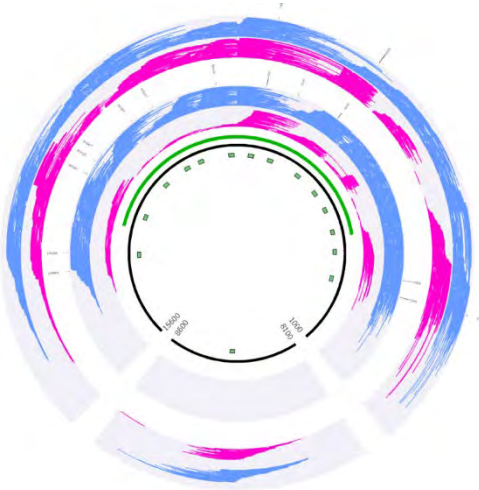
Strobl C et al. (2018) Evaluation of the **precision ID whole MtDNA genome** panel for forensic analyses." Forensic Sci Int Genet **35**: 21-25. for casework samples and aDNA

mtDNA amplification/enrichment strategies









“maxi”

“midi”

“mini”

“micro”

Fendt et al 2009
mtG ca. 8.5 kbp

GMI modified
mtG ca.2 kbp

Parson et al 2015
CR/mtG ca. 300 bp

Precision-ID (TFS)
CR/mtG ca. 175 bp

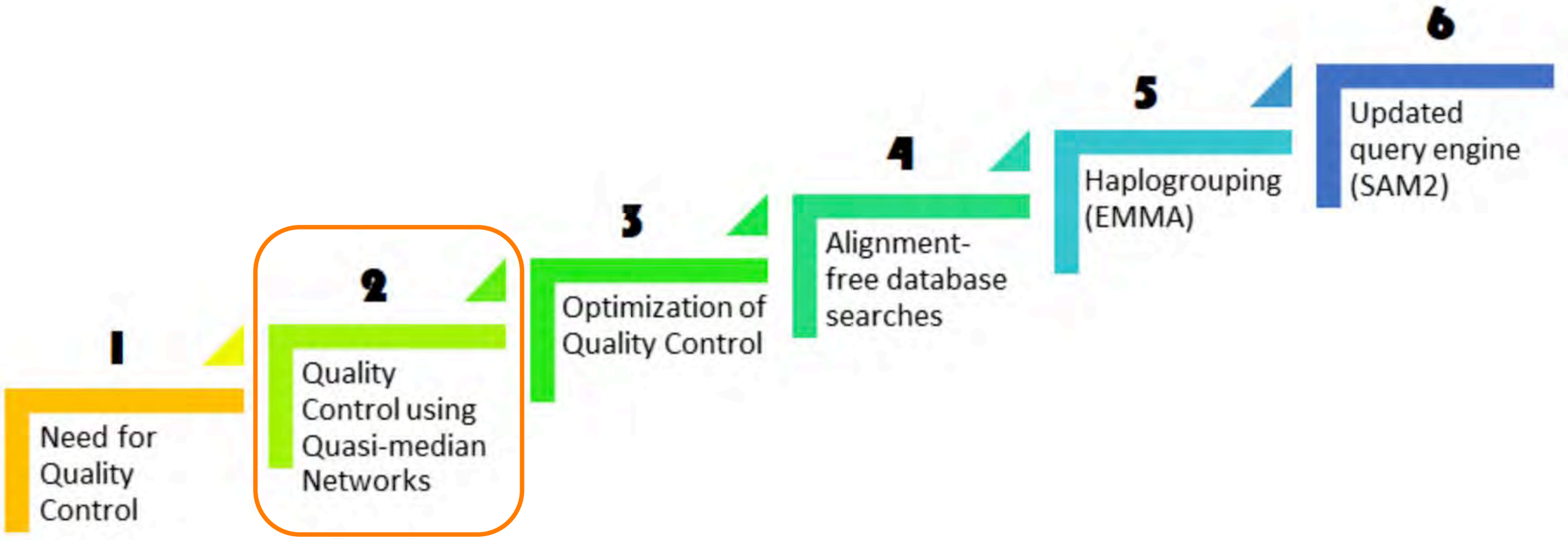
Eduardoff et al 2017
PEC CR/mtG < 70 bp

PGM

Ion Chef/Ion S5

MiSeq FGx®

History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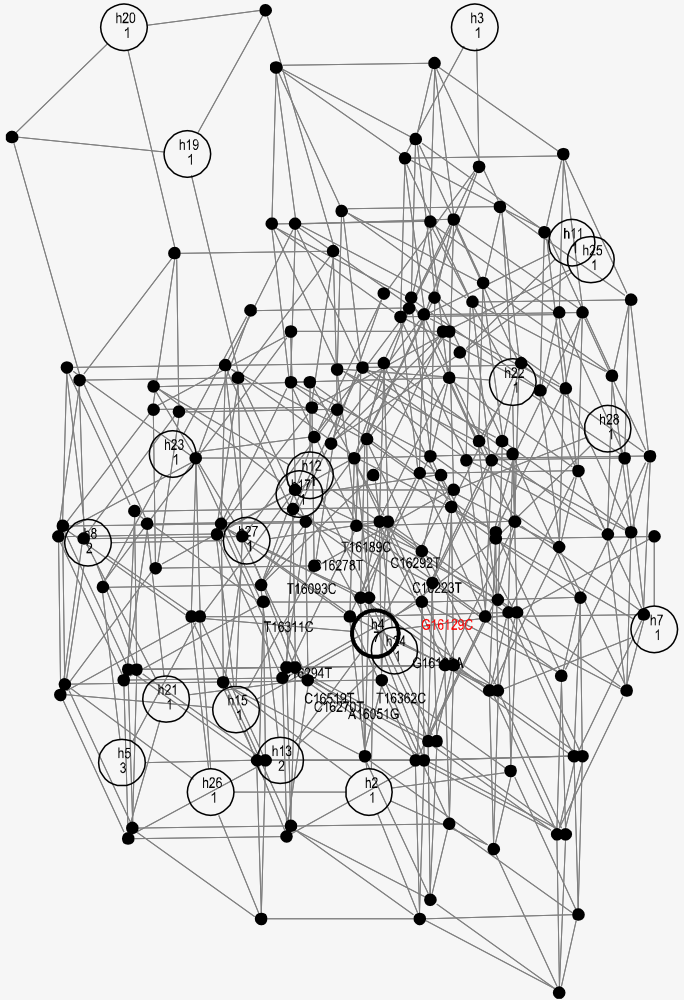
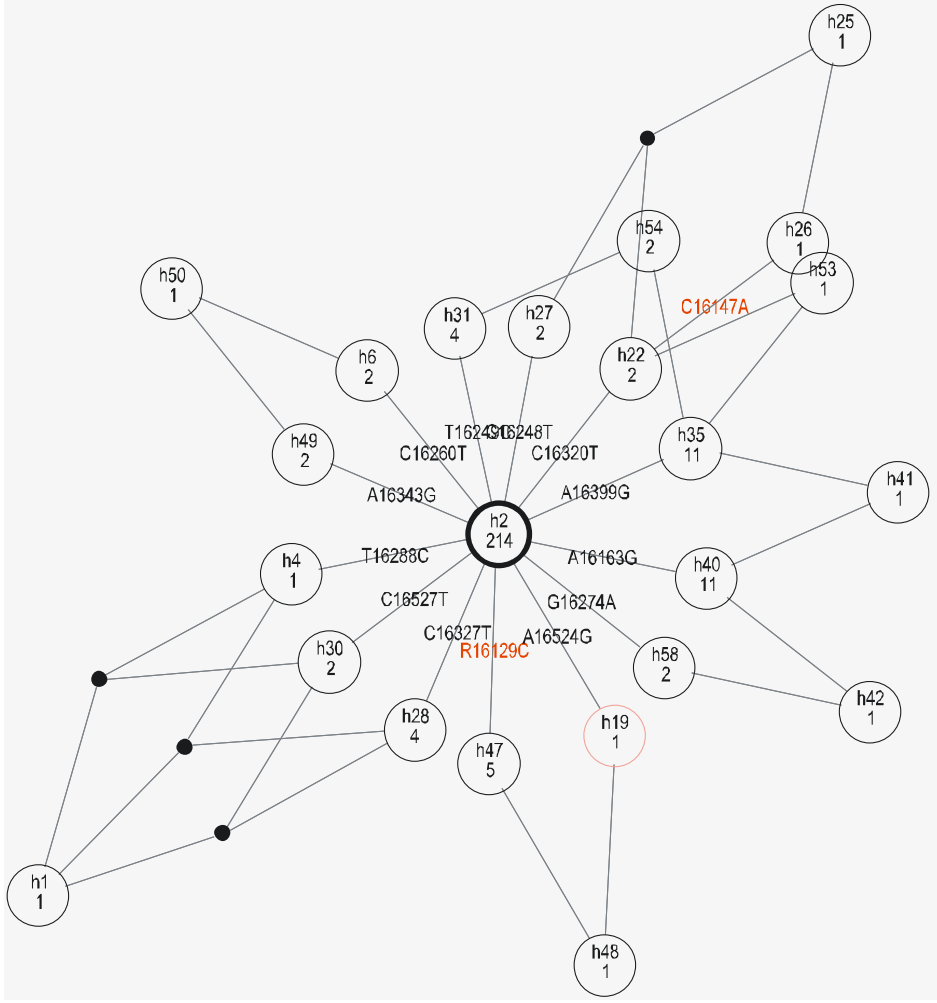
Quasi-Median Networks

unfiltered (N=100; 16024-16340)



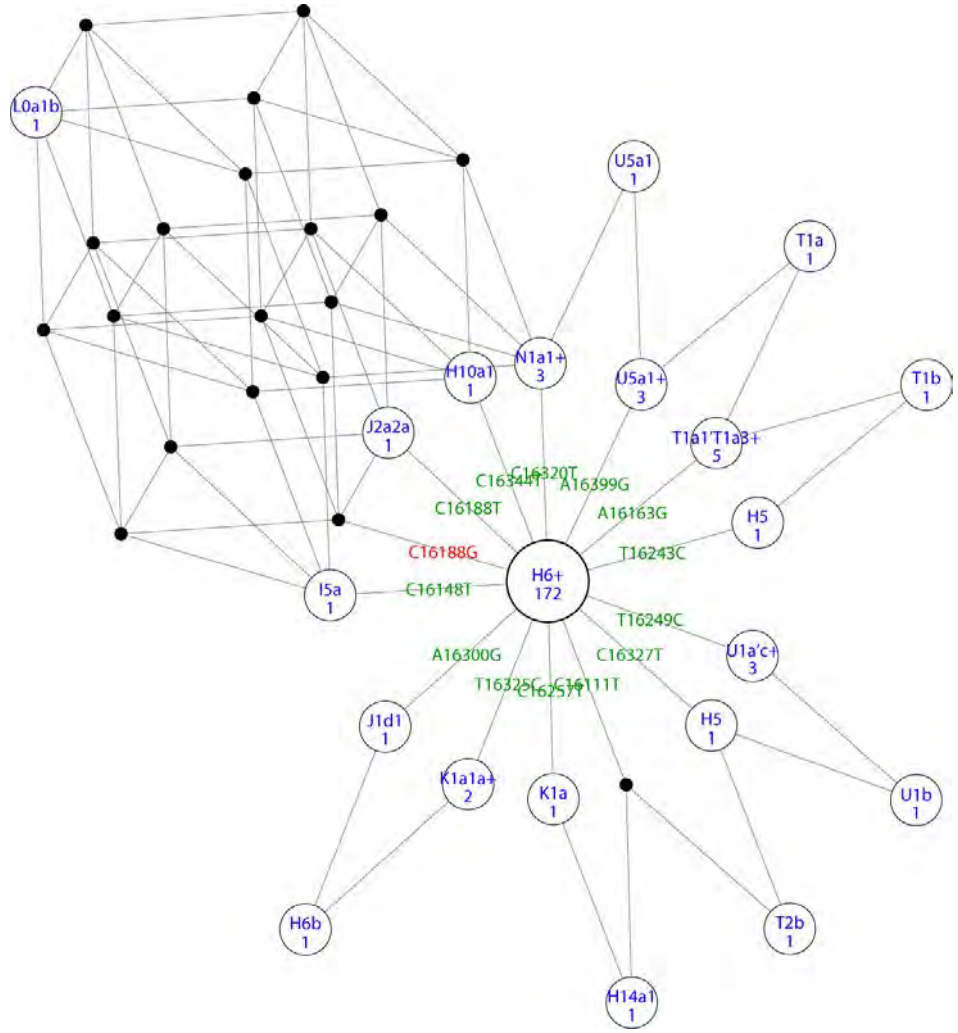
Brandstätter et al IJLM 2006

speedyWE (N=273; 16024-16569)

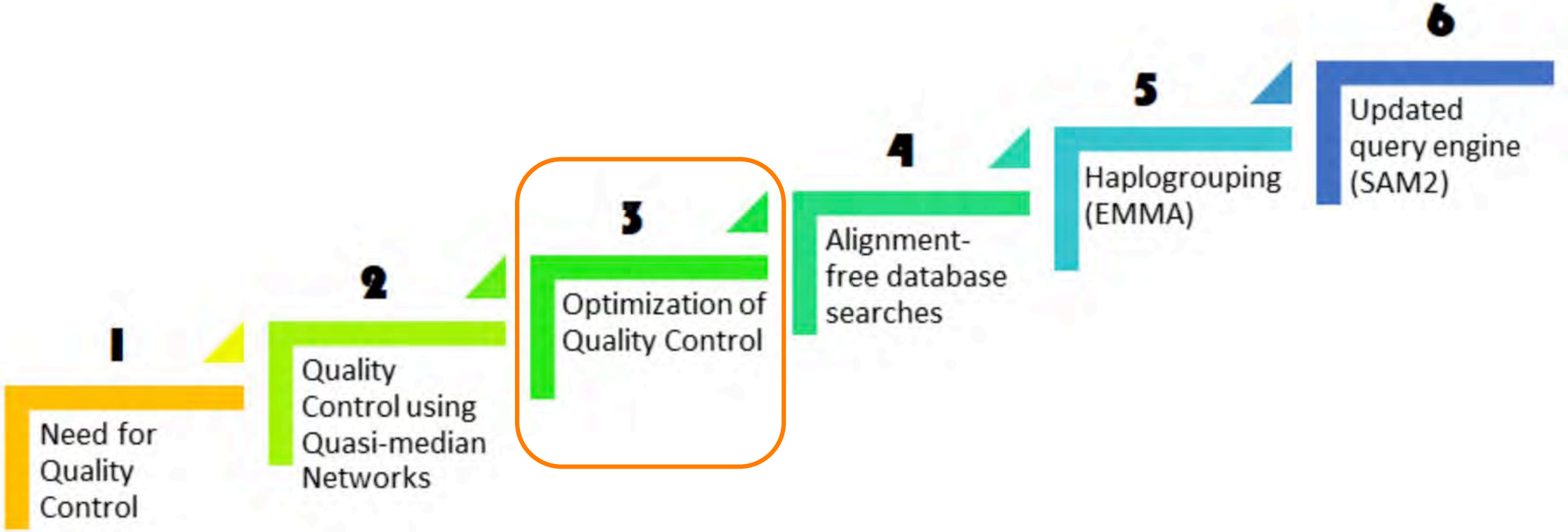


Zimmermann et al FSI:G 2011

QMN - distant phylogenies



History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Quality Control in Forensic mtDNA Analysis

GHEP-EMPOP

CC-GHEP 2006

Muestra	Tasa de error
M1-M2-M4	14%
M3	15%
M5	31%!!!

CC-GHEP 2008

Muestra	Tasa de error
M1	13%
M2-M5	7.5%
M3	13%
M4	13%

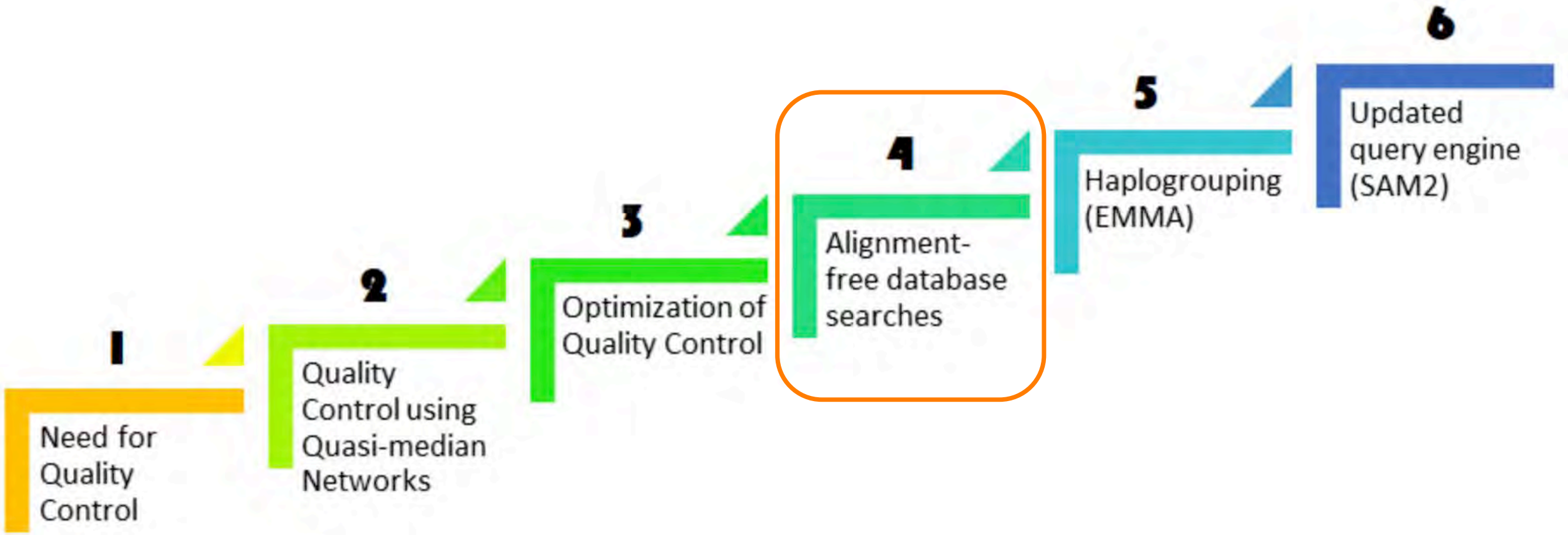
CC-GHEP 2010

Muestra	Tasa de error
M1	5%
M2-M3	6%
M4	5%
M5	2%

Prieto et al ***FSIG*** (2011)

slide by L. Prieto, Spain

History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Alignment-immune searches in EMPOP

Forensic Science International: Genetics 5 (2011) 126–132

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

SAM: String-based sequence search algorithm for mitochondrial DNA database queries

Alexander Röck [a], Jodi Irwin [b], Arne Dür [a], Thomas Parsons [c], Walther Parson [d,*]

[a] Institute of Mathematics, University of Innsbruck, Technikerstrasse 13, 6020 Innsbruck, Austria
[b] The Armed Forces DNA Identification Laboratory, 1413 Research Blvd., Rockville, MD 20850, USA
[c] The International Commission on Missing Persons, Alipašina 45 A, 71000 Sarajevo, Bosnia and Herzegovina
[d] Institute of Legal Medicine, Innsbruck Medical University, Müllerstrasse 44, 6020 Innsbruck, Austria

Queries of unaligned sequences guarantee that a haplotype is not missed in a database search

SAM on EMPOP since V2.0 (04/2010)

Solving the database issue

Search method	16188T 16189C	16188- 16193+C
rCRS-coded	28 matches	0 matches

EMPOP V3 R11; N = 34,617

Search method	16188T 16189C	16188- 16193+C
SAM	28 matches	28 matches

EMPOP V3 R11; N = 34,617

SAM on EMPOP since V2.0 (04/2010)

History


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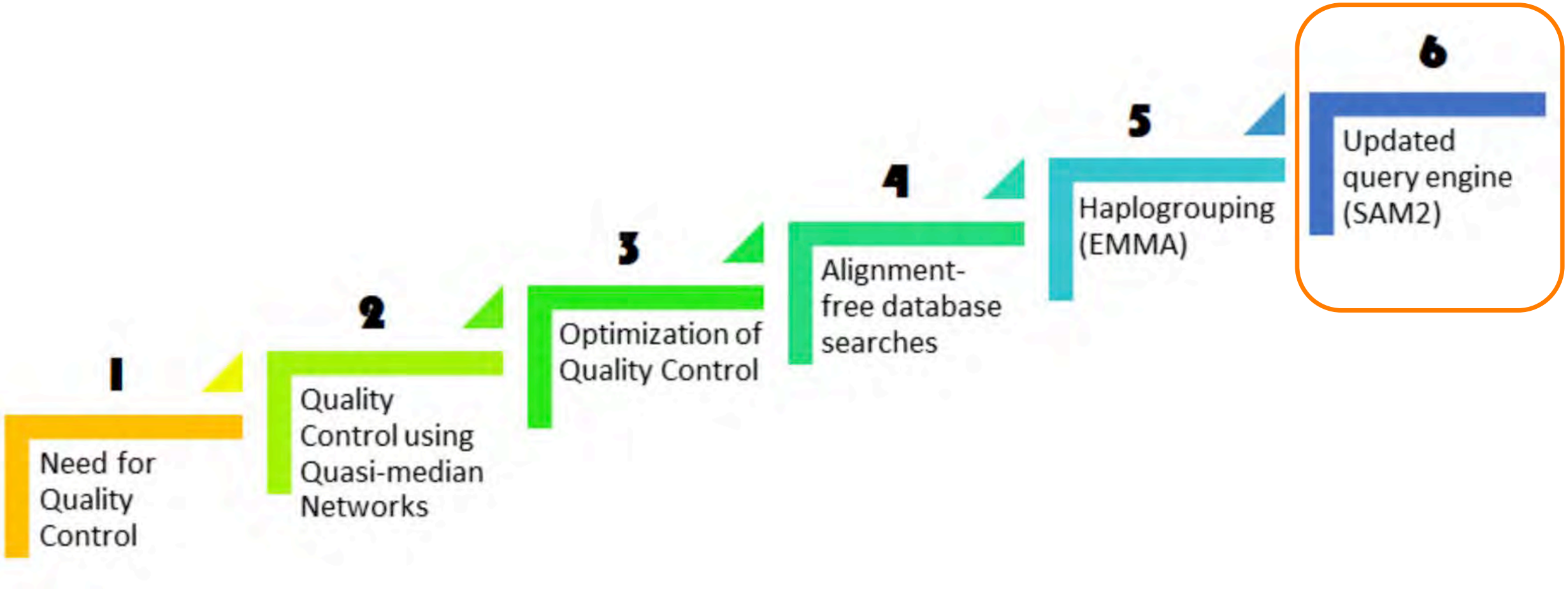
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SAM 2

Forensic Science International: Genetics 37 (2018) 204–214

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research paper

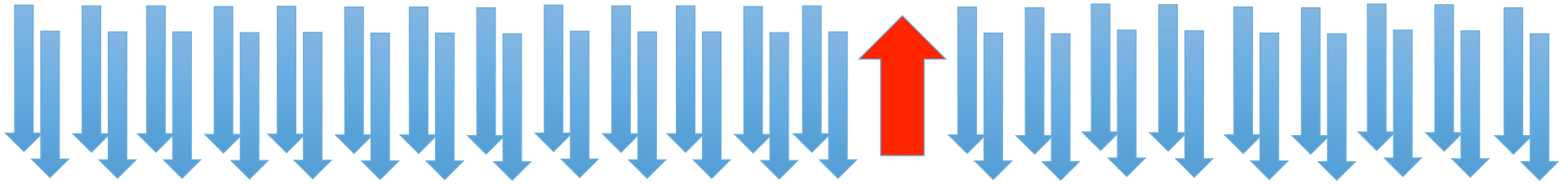
Next generation database search algorithm for forensic mitogenome analyses

Nicole Huber[a], Walther Parson[a,b,*], Arne Dür[c]

[a] Institute of Legal Medicine, Medical University of Innsbruck, Innsbruck, Austria
[b] Forensic Science Program, The Pennsylvania State University, University Park, PA, USA
[c] Institute of Mathematics, University of Innsbruck, Austria

Alignment

16024 TTCTTTCATGGGGAAGCAGATTTGGGTACCACCCAAGTATTGACTCACCCATCAACAACCGCTATGTATTTCGTACATTACTGCCAGCCACCATGAATATTGTACGGTACCATAA
ATACTTGACCACCTGTAGTACATAAAAACCCAATCCACATCAAAACCCCCCCCCCCCATGCTTACAAGCAAGTACAGCAATCAACCCTCAACTATCACACATCAACTGCAACTCCA
AAGCCACCCCTCACCCACTAGGATACCAACAAACCTACCCACCCTTAACAGTACATAGTACATAAAGCCATTTACCGTACATAGCACATTACAGTCAAATCCCCTCTCGCCCCCA
TGGATGACCCCCCTCAGATAGGGGTCCCTTGACCACCATCCTCCGTGAAATCAATATCCCGCACAAGAGTGCTACTCTCCTCGCTCCGGGCCCATAACACTTGGGGGTAGCTA
AAGTGAACTGTATCCGACATCTGGTTCCTACTTCAGGGCCATAAAGCCTAAATAGCCCACACGTTCCCCTTAAATAAGACATCACGATGGATCACAGGTCTATCACCCTATTAAC
CACTCACGGGAGCTCTCCATGCATTTGGTATTTTCGTCTGGGGGGTATGCACGCGATAGCATTGCGAGACGCTGGAGCCGGAGCACCCTATGTCGCAGTATCTGTCTTTGATTC
CTGCCTCATCCTATTATTTATCGCACCTACGTTCAATATTACAGGCGAACATACTTACTAAAGTGTGTTAATTAATTAATGCTTGTAGGACATRATAATAACAATTGAATGTCTGC
ACAGCCGCTTTCCACACAGACATCATAACAAAAAATTTCCACCAAACCCCCCCTCCCCCCGCTTCTGGCCACAGCACTTAAACACATCTCTGCCAAACCCCAAAAACAAAGAACC
CTAACACCAGCCTAACCAGATTTCAAATTTTATCTTTTGGCGGTATGCACTTTTAACAGTCACCCCCCAACTAACACATTATTTTCCCCTCCCACTCCCATACTACTAATCTCATCAA
TACAACCCCCCGCCCATCCTACCCAGCACACACNN--CGCTGCTAACCCCATACCCCGAACCAACCAAACCCCAAAGACACCCCCCCCACA 576



953,110

(24+1 mutations)

16181C 16182C 16183C 16189C 16213A 16217C 16242T 16261T 16292T 16301T 16519C

61A 62A 73G 183G 263G 309.1C 309.2C 309.3C 315.1C 323N 324N 523Del 524Del

16181C 16182C 16183C 16189C 16213A 16217C 16242T 16261T 16292T 16301T 16519C

61A 62A 73G 183G 263G 308.1C 309.1C 309.2C 315.1C 323N 324N 523Del 524Del

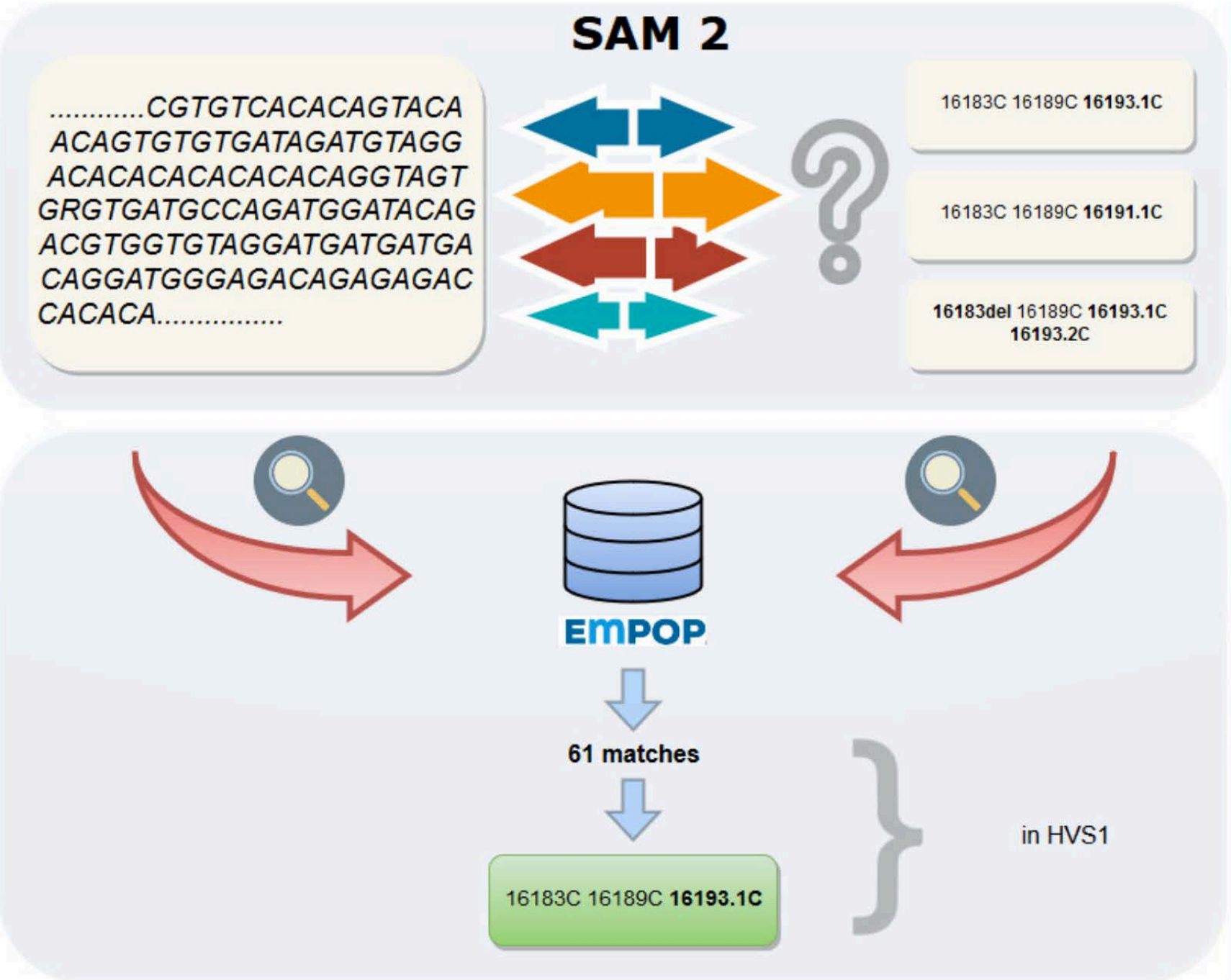
Alignment

16024 TTCTTTCATGGGGAAGCAGATTTGGGTACCACCCAAGTATTGACTCACCCATCAACAACCGCTATGTATTTCGTACATTACTGCCAGCCACCATGAATATTGTACGGTACCATAA
ATACTTGACCACCTGTAGTACATAAAAACCCAATCCACATCAAAACCCCCCCCCCCCATGCTTACAAGCAAGTACAGCAATCAACCCTCAACTATCACACATCAACTGCAACTCCA
AAGCCACCCCTCACCCACTAGGATACCAACAAACCTACCCACCCTTAACAGTACATAGTACATAAAGCCATTTACCGTACATAGCACATTACAGTCAAATCCCCTCTCGCCCCCA
TGGATGACCCCCCTCAGATAGGGGTCCCTTGACCACCATCCTCCGTGAAATCAATATCCCGCACAAGAGTGCTACTCTCCTCGCTCCGGGCCCATAACACTTGGGGGTAGCTA
AAGTGAACTGTATCCGACATCTGGTTCCTACTTCAGGGCCATAAAGCCTAAATAGCCCACACGTTCCCCTTAAATAAGACATCACGATGGATCACAGGTCTATCACCCTATTAAC
CACTCACGGGAGCTCTCCATGCATTTGGTATTTTCGTCTGGGGGGTATGCACGCGATAGCATTGCGAGACGCTGGAGCCGGAGCACCCTATGTCGCAGTATCTGTCTTTGATTC
CTGCCTCATCCTATTATTTATCGCACCTACGTTCAATATTACAGGCGAACATACTTACTAAAGTGTGTTAATTAATTAATGCTTGTAGGACATRATAATAACAATTGAATGTCTGC
ACAGCCGCTTTCCACACAGACATCATAACAAAAAATTTCCACCAAACCCCCCCTCCCCCCGCTTCTGGCCACAGCACTTAAACACATCTCTGCCAAACCCCAAAAACAAAGAACC
CTAACACCAGCCTAACCAGATTTCAAATTTTATCTTTTGGCGGTATGCACTTTTAACAGTCACCCCCCAACTAACACATTATTTTCCCCTCCCACTCCCATACTACTAATCTCATCAA
TACAACCCCCGCCCATCCTACCCAGCACACACNN--CGCTGCTAACCCCATACCCCGAACCAACCAAACCCCAAAGACACCCCCCCCACA 576

SAM 2

16181C 16182C 16183C 16189C 16213A 16217C 16242T 16261T 16292T 16301T 16519C

61A 62A 73G 183G 263G 309.1C 309.2C 309.3C 315.1C 323N 324N 523Del 524Del



Alignment-free query engine for unbiased search results and pyhlogenetic alignment of mtDNA sequences

EMPOP uses SAM2, a string-based search algorithm that converts query and database sequences into alignment-free nucleotide strings and thus guarantees that a haplotype is found in a database query regardless of its alignment.

Furthermore, SAM2 provides the phylogenetic alignment of mtDNA sequences to harmonize alignment and notation of mtDNA sequences.

Harmonizing Alignment

EMPOP 4

Query | Result | Details | Neighbors | **Alignment** | Haplogrouping

Phylogenetic alignment

Input Profile	263G	315.1C	523-	524-	16183-			
Phylogenetic alignment	263G	315.1C	523-	524-	16183C	16188T	16189C	16193-

Alignment was estimated using SAM 2.0 on the basis of 5,440 haplogroup motifs (Phylotree, Build 17) following the phylogenetic concept and the recommendations of the ISFG and was derived from haplogroup

HV18 | H1+16189 | H1f | H1g | H1g1 | H1g2 | H1y | H1aa | H1aa1 | H1ab | H1ac | H1ad | H1j9 | H3+16189

in range 16024-576 by the following transcript with cost 1.40:

[M16183C(0.00)] C16188T(0.80) 16193delC(0.20) [Y16519T(0.00)] 523-524delAC(0.40)

Huber et al 2018

130 peer reviewed original articles on mtDNA /EMPOP since 1998

Forensic Science International: Genetics 13 (2014) 134–142

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

DNA Commission of the International Society for Forensic Genetics: Revised and extended guidelines for mitochondrial DNA typing

W. Parson [a,b,*], L. Gusmão [c,d], D.R. Hares [e], J.A. Irwin [e], W.R. Mayr [f], N. Morling [g], E. Pokorak [e], M. Prinz [h], A. Salas [i], P.M. Schneider [j], T.J. Parsons [k]

Haploid Marker Meetings

Education - EMPOP conferences and courses

The EMPOP project and database arose from collaborative studies that were established and discussed at scientific meetings. We continue this tradition to inspire and facilitate research.

EMPOP training HSA Singapore

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

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

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**)

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

Short Tandem Repeats

DNASeqEx - **DNA**-STR Massive **Seq**uencing & International Information **Ex**change

2 years (2016-2018)

Madrid

Berlin

DNA SeqEx

STR Sequencing & Exchange

Interlaboratory studies

Concordance
Sensitivity
Reproducibility
Casework

EA Applied Biosystems Precision ID Globalfiler Mixture ID™ & Globalfiler NGS STR Panels for the Ion S5™

Concordance
Sensitivity
Reproducibility
Mixtures

ForenSeq® DNA Signature Prep Kit (Verogen)

Innsbruck

Additional Testing

Qiagen

Promega aSTR/mito

Promega aSTR/Y-STRs

SEQforSTRs

Seq for STRs

< Previous Article | July 2017 Volume 29, Pages e23–e25 | Next Article >

European survey on forensic applications of massively parallel sequencing

Antonio Alonso
National Institute of Toxicology and Forensic Sciences, Madrid Department, Spain
Petra Müller
Institute of Legal Medicine, Medical University of Innsbruck, Austria
Lutz Roewer, Sascha Willuweit
Institute of Legal Medicine and Forensic Sciences, Charité–Universitätsmedizin Berlin, Germany
Bruce Budowle
Walther Parson

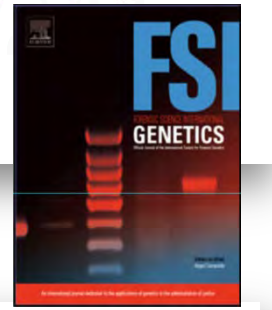
PlumX Metrics
DOI: https://doi.org/10.1016/j.fsigen.2017.04.017

Research paper

Inter-laboratory validation study of the ForenSeq™ DNA Signature Prep Kit

Steffi Köcher[a,*,1], Petra Müller[b,1], Burkhard Berger[b], Martin Bodner[b], Walther Parson[b,c], Lutz Roewer[a], Sascha Willuweit[a], The DNASeqEx Consortium

[a] *Institute of Legal Medicine and Forensic Sciences, Charité – Universitätsmedizin Berlin, Germany*
[b] *Institute of Legal Medicine, Medical University of Innsbruck, Austria*
[c] *Forensic Science Program, The Pennsylvania State University, PA, USA*



Research paper

Systematic evaluation of the early access applied biosystems precision ID Globalfiler mixture ID and Globalfiler NGS STR panels for the ion S5 system

Petra Müller[a], Antonio Alonso[b], Pedro A. Barrio[b], Burkhard Berger[a], Martin Bodner[a], Pablo Martin[b], Walther Parson[a,c,*], The DNASEQEX Consortium

[a] *Institute of Legal Medicine, Medical University of Innsbruck, Innsbruck, Austria*
[b] *National Institute of Toxicology and Forensic Sciences, Madrid Department, Las Rozas de Madrid, Spain*
[c] *Forensic Science Program, The Pennsylvania State University, PA, USA*

ELECTROPHORESIS

Review

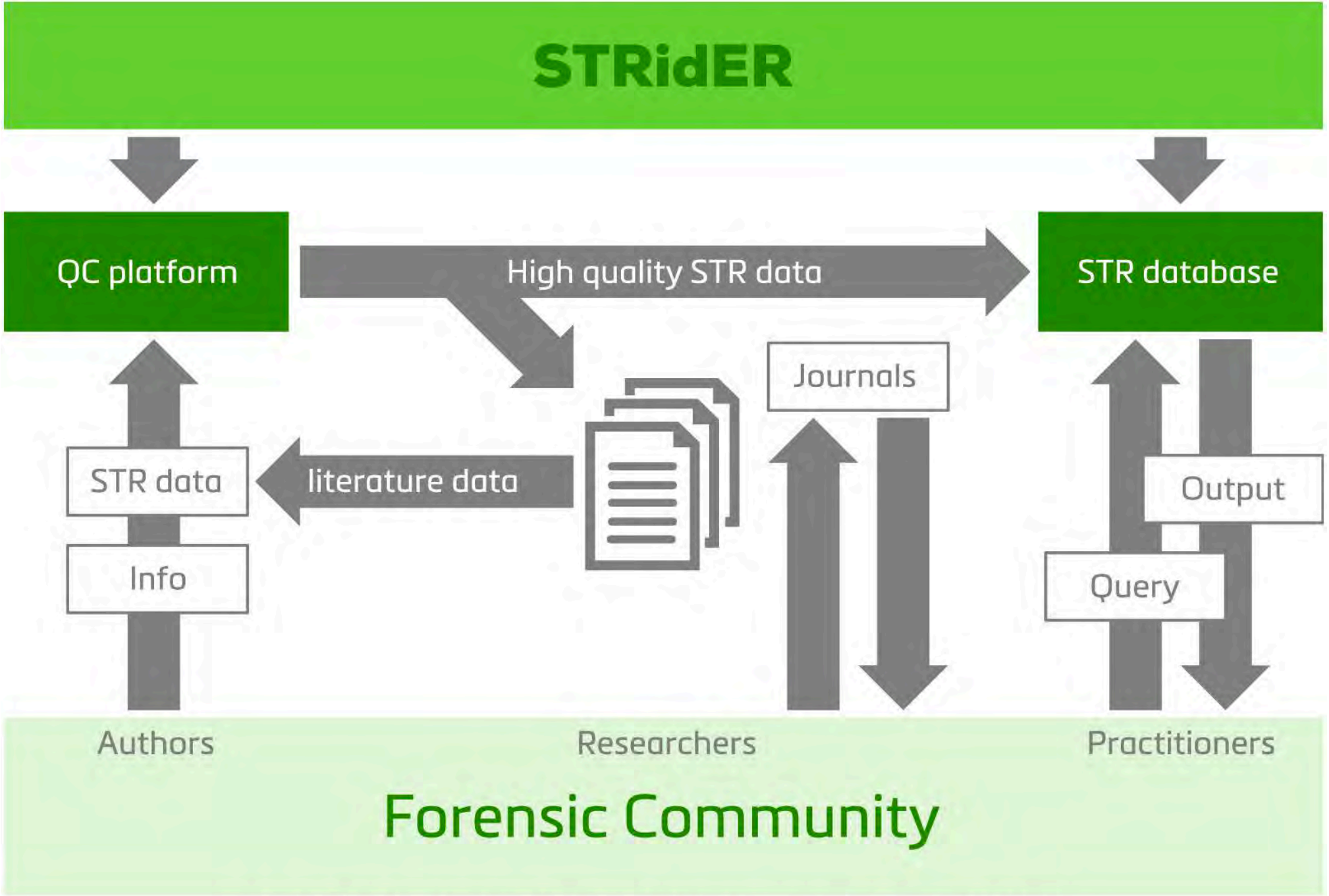
Current state-of-art of STR sequencing in forensic genetics

Antonio Alonso, Pedro Alberto Barrio, Petra Müller, Steffi Köcher, Burkhard Berger, Pablo Martin, Martin Bodner, Sascha Willuweit, Walther Parson, Lutz Roewer, Bruce Budowle

DNA SeqEx
STR Sequencing & Exchange

STRidER

former STRbASE ENFSI DNA WG STR Population Database, v2



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

STRidER

STRs for identity ENFSI Reference database, v2

https://strider.online/

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

Frequencies

These tables include allele frequencies and number of samples (n) from the most recent database release sorted by marker and country.
In these tables, „1" represents all rare alleles shorter than the accepted allele categories. The value „99" represents all rare alleles longer than the accepted categories.

This data can be downloaded as XML file.

VWA

Allele	AUSTRIA	BELGIUM	BOSNIA AND HERZEGOWINA	CZECH REPUBLIC	DENMARK	FINLAND	FRANCE	GERMANY	GREECE	HUNGARY	IRELAND	MONTENEGRO	NORWAY	POLAND	SLOVAKIA	SLOVENIA	SPAIN	SWEDEN
	222	206	171	200	200	230	208	662	208	224	304	200	202	206	247	207	449	424
11								7.5529e-4										
12									4.8077e-3									
13			1.1696e-2					2.2659e-3	2.4038e-3	2.2321e-3			2.4753e-3		2.0243e-3	2.4155e-3	6.6815e-3	1.1792e-3
14	1.0586e-1	1.0680e-1	1.1111e-1	1.0000e-1	7.0000e-2	1.3043e-1	8.6539e-2	9.7432e-2	9.3750e-2	1.1161e-1	1.1349e-1	1.4500e-1	8.6634e-2	7.7670e-2	1.1943e-1	1.0145e-1	1.1024e-1	9.4340e-2
15	9.2342e-2	1.2136e-1	1.2573e-1	9.7500e-2	9.7500e-2	5.2174e-2	1.2740e-1	1.0347e-1	7.9327e-2	1.1384e-1	1.0197e-1	9.0000e-2	9.9010e-2	8.4951e-2	1.1943e-1	1.2077e-1	1.2361e-1	8.9623e-2
16	1.7568e-1	1.9903e-1	2.0468e-1	1.7500e-1	2.6000e-1	1.7609e-1	2.4038e-1	2.2130e-1	1.6827e-1	2.0536e-1	2.1875e-1	1.7500e-1	2.2277e-1	2.2330e-1	1.9231e-1	1.8599e-1	2.4276e-1	2.0991e-1
17	2.8604e-1	2.7185e-1	2.3977e-1	3.1250e-1	2.3000e-1	2.7174e-1	2.3317e-1	2.5453e-1	3.1731e-1	3.0134e-1	2.7138e-1	2.8750e-1	2.8960e-1	2.7670e-1	2.7530e-1	2.8985e-1	2.7171e-1	2.6533e-1
18	2.5901e-1	2.0146e-1	2.1053e-1	2.2750e-1	2.4000e-1	2.0435e-1	2.1154e-1	2.2054e-1	2.4279e-1	1.7634e-1	1.9243e-1	2.1250e-1	1.9802e-1	2.4757e-1	2.0445e-1	2.1739e-1	1.7038e-1	2.4174e-1
19	7.2072e-2	8.0097e-2	9.0643e-2	7.2500e-2	8.2500e-2	1.3696e-1	8.6539e-2	8.6103e-2	7.4519e-2	7.1429e-2	9.3750e-2	7.2500e-2	8.6634e-2	8.0097e-2	7.6923e-2	5.5556e-2	6.1247e-2	7.9009e-2
20	9.0090e-3	1.9418e-2	5.8480e-3	1.5000e-2	1.7500e-2	2.1739e-2	1.4423e-2	1.2840e-2	1.4423e-2	1.5625e-2	8.2237e-3	1.7500e-2	1.4852e-2	9.7087e-3	1.0122e-2	2.1739e-2	1.3363e-2	1.6509e-2
21					2.5000e-3	6.5217e-3		7.5529e-4	2.4038e-3	2.2321e-3						4.8309e-3		2.3585e-3

TH01

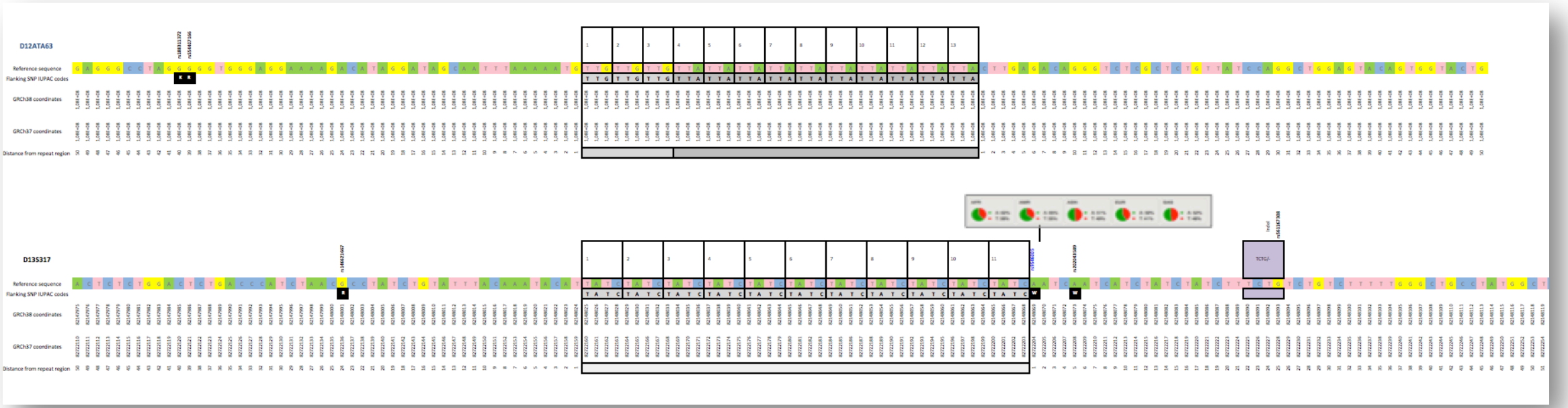
Allele	AUSTRIA	BELGIUM	BOSNIA AND HERZEGOWINA	CZECH REPUBLIC	DENMARK	FINLAND	FRANCE	GERMANY	GREECE	HUNGARY	IRELAND	MONTENEGRO	NORWAY	POLAND	SLOVAKIA	SLOVENIA	SPAIN	SWEDEN
	222	206	171	200	200	230	208	662	208	224	304	200	202	206	247	207	454	425

MPS Nomenclature

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

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

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



STRidER

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

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

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

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

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STR Sequence Nomenclature

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

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

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

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

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

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

[a] *Forensic Genetics Unit, Institute of Forensic Sciences, University of Santiago de Compostela, Spain*
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[c] *Institute of Applied Genetics, Department of Molecular and Medical Genetics, University of North Texas Health Science Center, Fort Worth, TX, USA*
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[e] *Institute of Legal Medicine, Medical University of Innsbruck, Innsbruck, Austria*
[f] *Forensic Science Program, The Pennsylvania State University, University Park, PA, USA, USA*

Audit of GRCh38 reference genome builds released between 2013 - 2017

Revised repeat region sequence structure summaries

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

+ 34 aSTRs (total of 71 aSTRs)

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

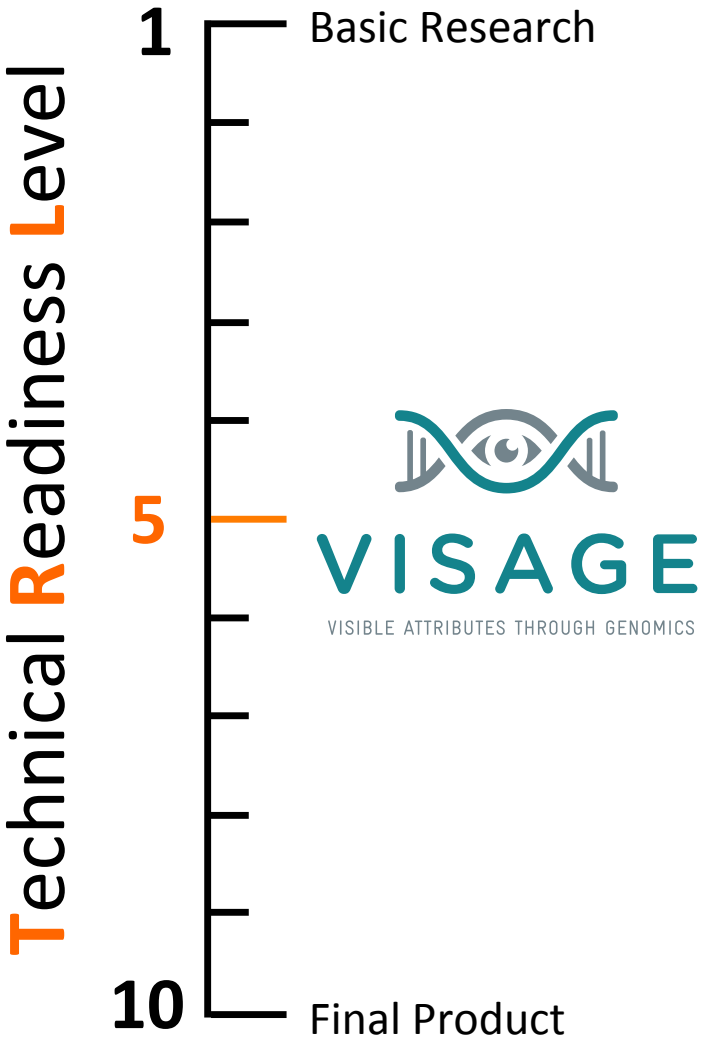
VISAGE - Visual Attributes Through Genomics

Manfred Kayser (Coordinator) Rotterdam, NED
Wojciech Branicki Krakow, POL
Chris Phillips, Angel Carracedo S. de Compostela, ESP
Walther Parson Innsbruck, AUT
Michael Nothnagel Cologne, GER
Barbara Prainsack Vienna, AT
Peter M. Schneider Cologne, GER

Ingo Bastisch Wiesbaden, GER
François-Xavier Laurent Lyon, FRA
Titia Sijen The Hague, NED
Johannes Hedman Linkoping, SWE
Shazia Khan London, UK
Magdalena Spólnicka Warsaw, POL

www.visage-h2020.eu

Tool development in VISAGE

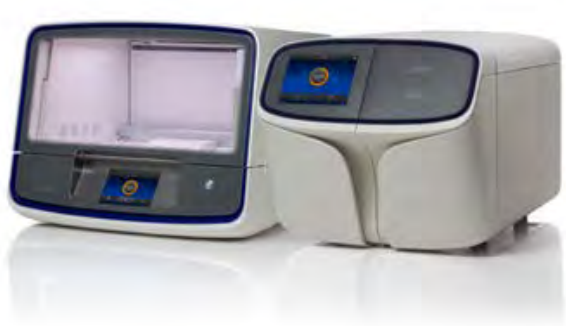


VISAGE
VISIBLE ATTRIBUTES THROUGH GENOMICS

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

Tool 1: Appearance & Ancestry (SNP multiplex)

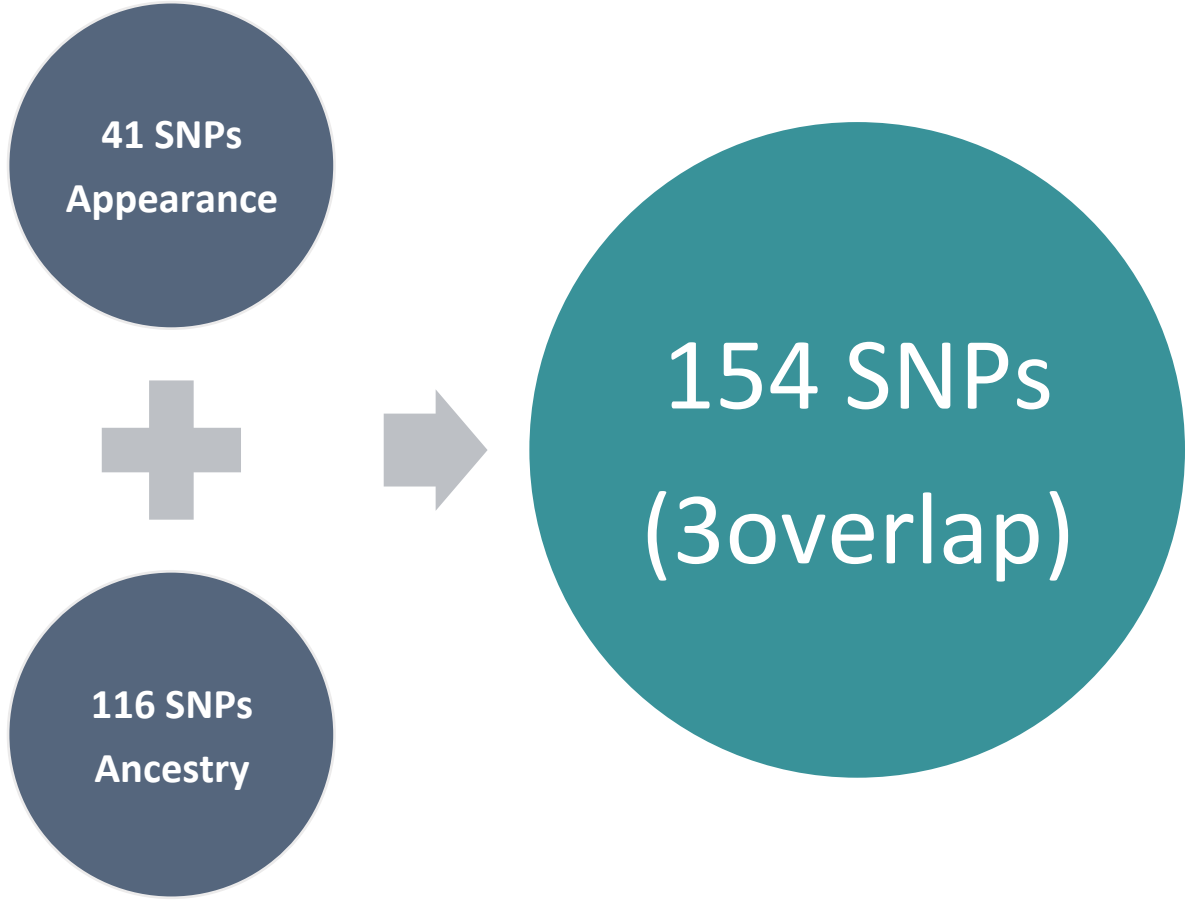
Tool 2: Age (quantitative methylation)



VISAGE Basic Tool

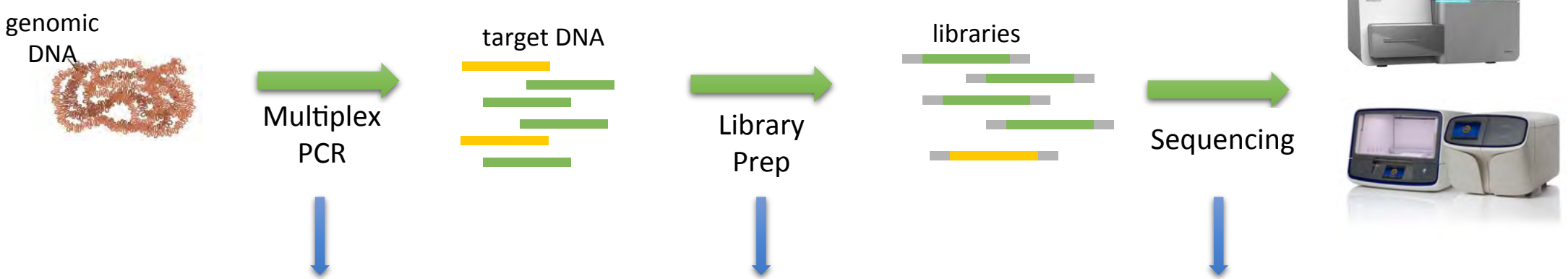
Tool 1: Appearance & Ancestry (SNP multiplex)

Known markers
+ explore new markers



Tool 1: Appearance & Ancestry (SNP multiplex)

Design, develop and validate prototype tools

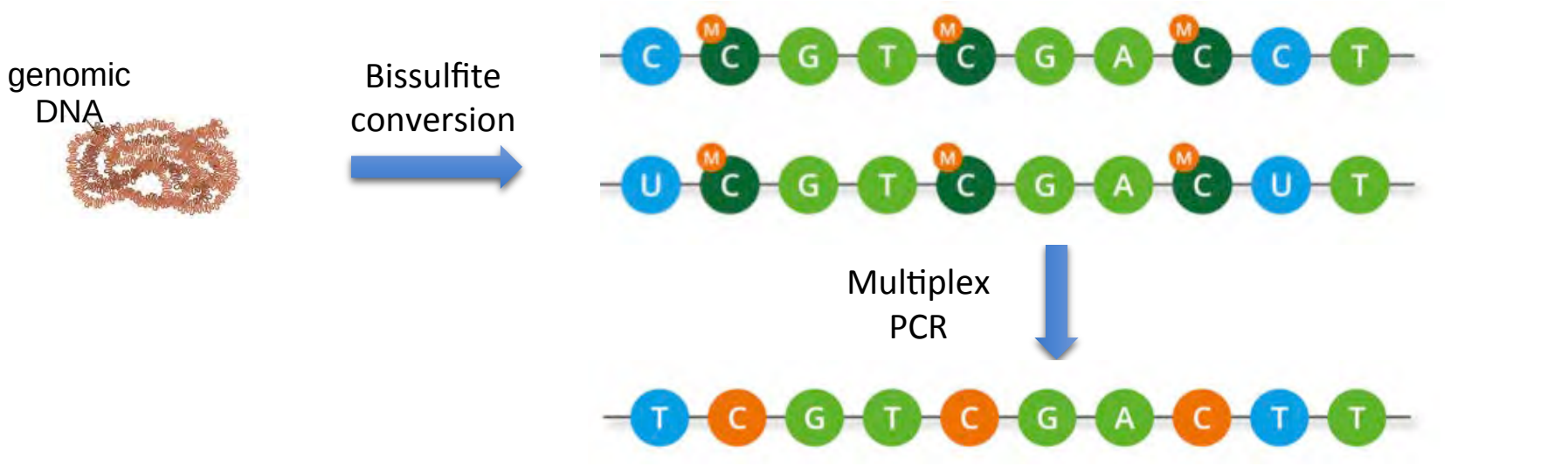


Multiplex design/ optimization
primer pools
how many reactions
optimize PCR

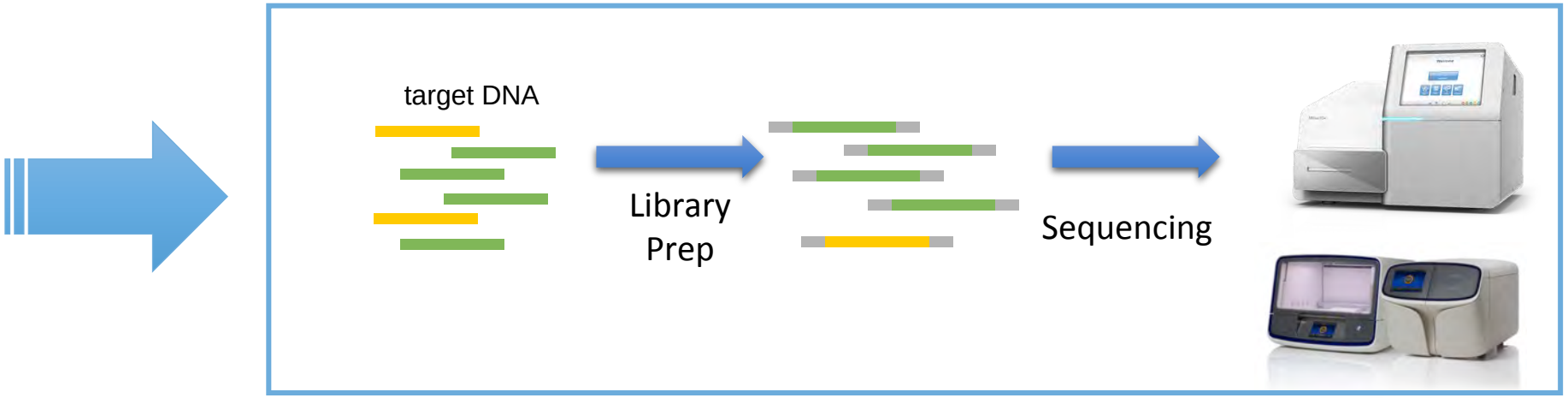
Adapt to different library prep commercial products

How many samples per chip
Overall performance
Coverage requirements

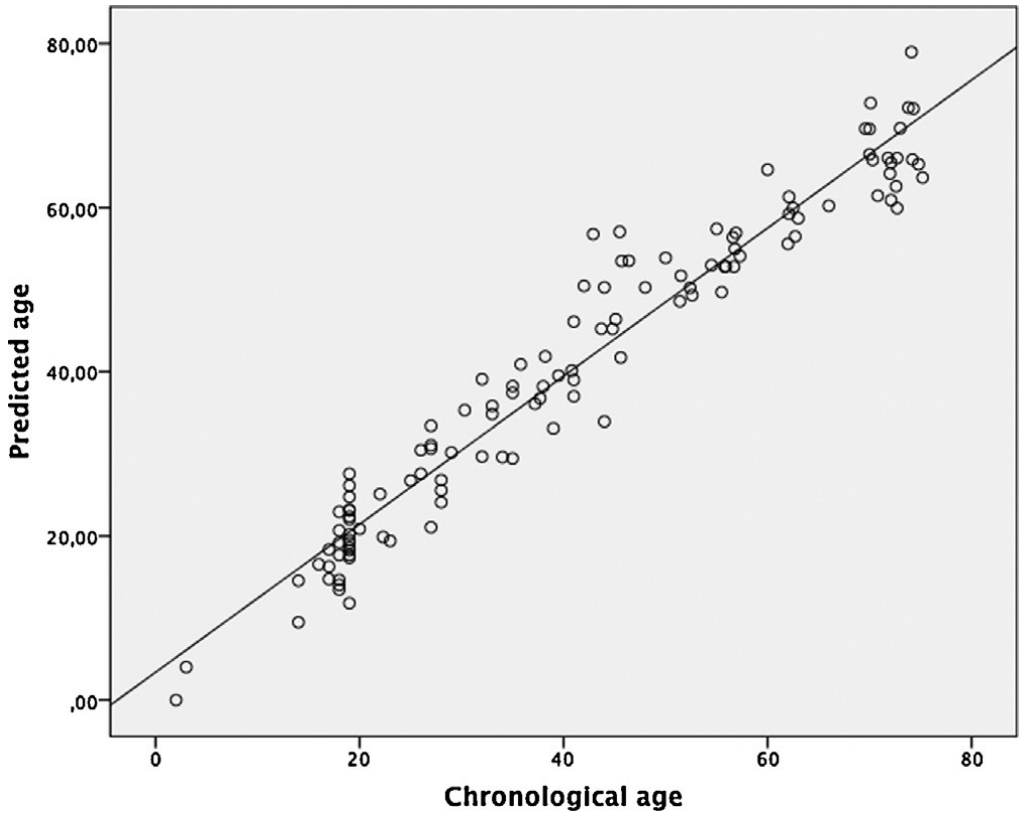
Age estimation by quantitative methylation



https://www.gatc-biotech.com



Age estimation – VISAGE basic tool



Methods

- 5 marker for **blood**
- Singleplex assays for pyrosequencing

Results

- MAD testing set (N=120): 3.9 years
- R^2 = 0.9444
- Correct overall prediction (±5 yrs): 71.7%

Development of a forensically useful age prediction method based on DNA methylation analysis

Renata Zbieć-Piekarska [a], Magdalena Spólnicka [a], Tomasz Kupiec [b], Agnieszka Parys-Proszek [b], Żanetta Makowska [a], Anna Pałeczka [a], Krzysztof Kucharczyk [c], Rafał Płoski [d], Wojciech Branicki [b,e,*]

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

www.visage-h2020.eu

ThermoFisher SCIENTIFIC

Promega

illumina®

VEROGEN

QIAGEN

VISAGE
VISIBLE ATTRIBUTES THROUGH GENOMICS

GMI

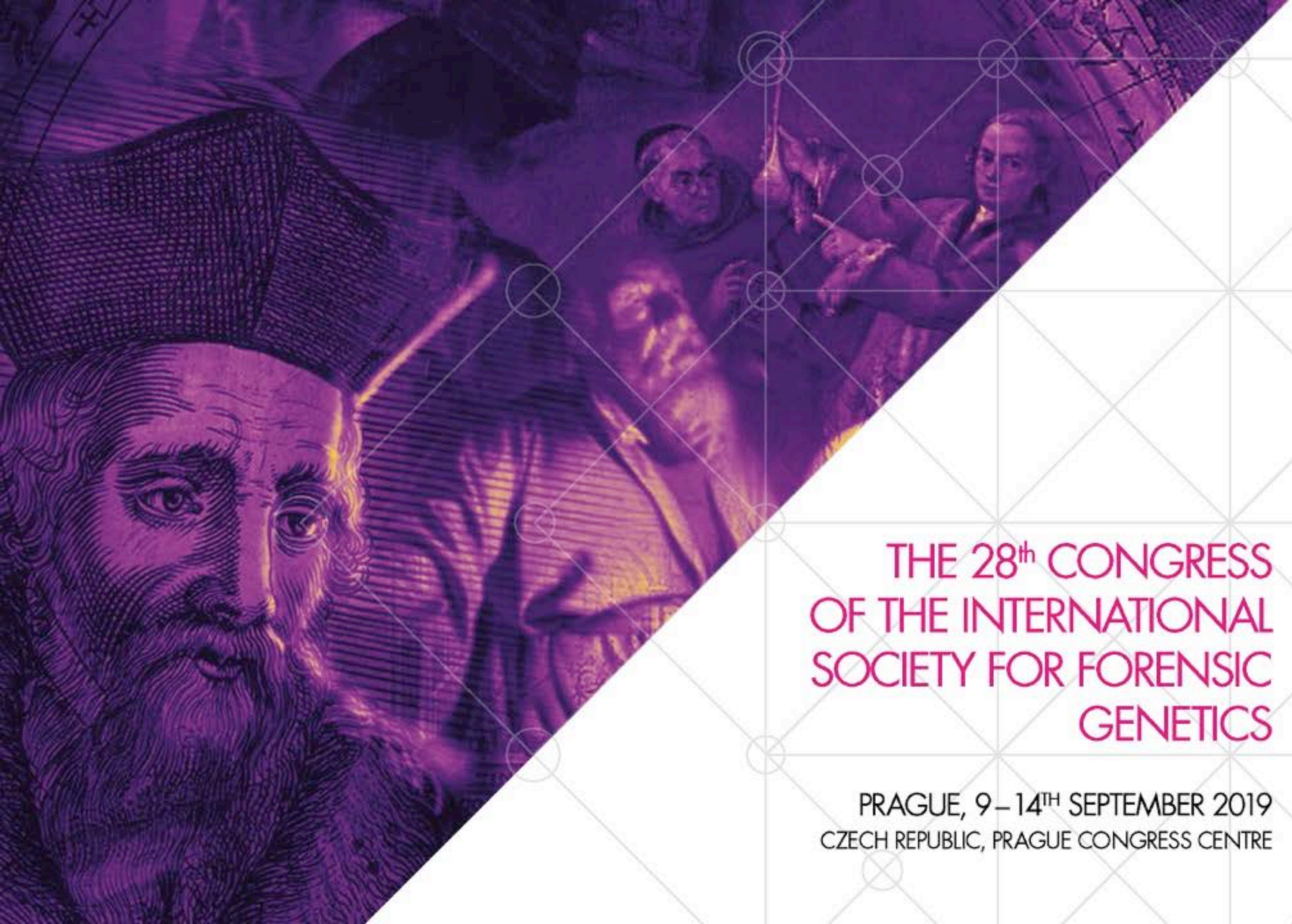
Acknowledgements

Catarina Xavier
Antonia Heidegger
Harald Niederstätter
Maria de la Punte
Walther Parson

ISFG Update

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

EDNAP Meeting, Buchen, Austria, Oct 30 2018



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

THE 28th CONGRESS OF THE INTERNATIONAL SOCIETY FOR FORENSIC GENETICS

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

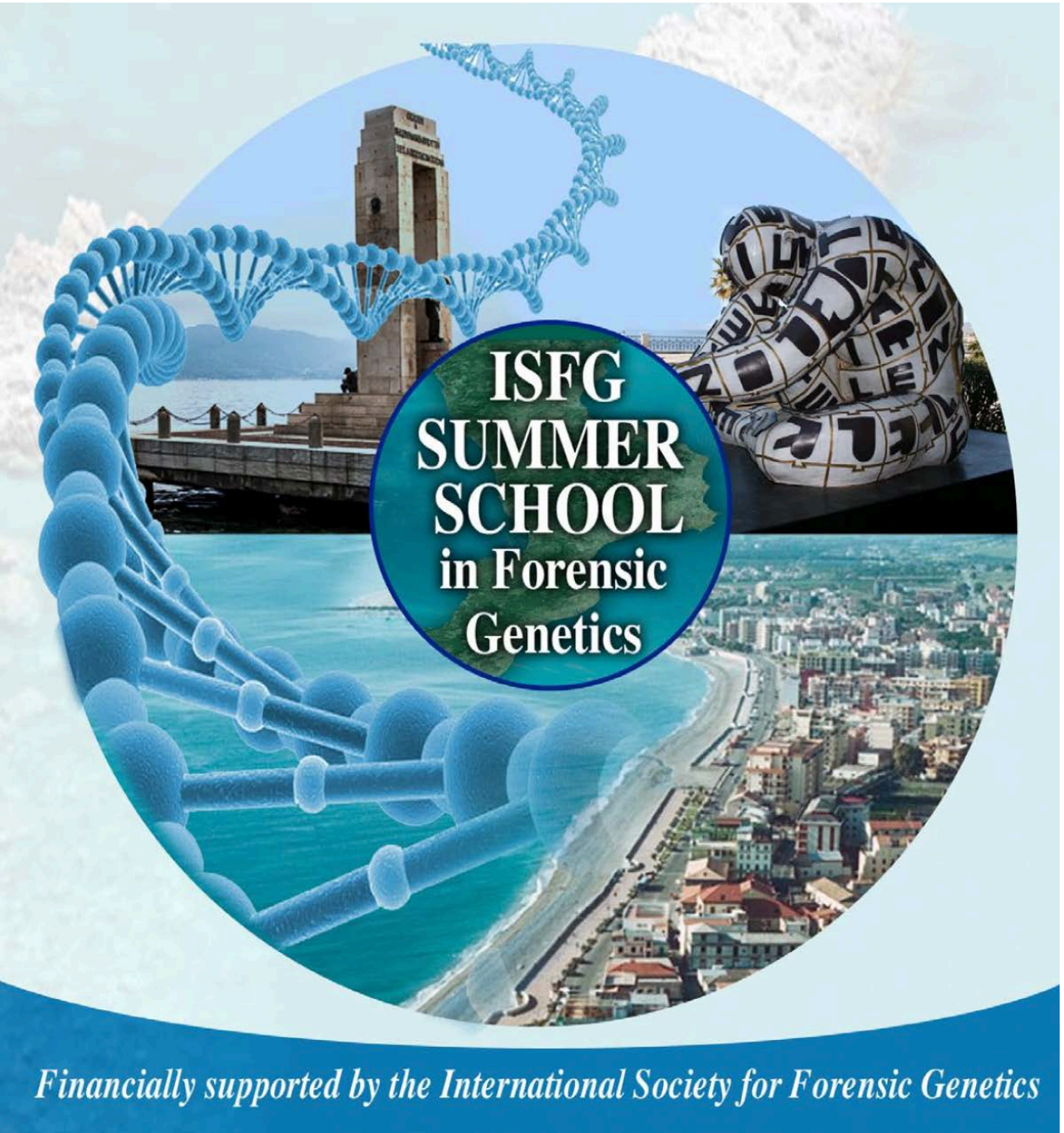
Congress Travel Bursaries

Purpose: To support young scientists presenting at an ISFG congress

- Travel bursaries will be made available again for the congress in Prague 2019
- For current Terms of Reference, see
  - https://www.isfg.org/files/ISFG_Bursaries_Nov2016.pdf

Short-term fellowships

- Purpose: **To support transnational exchange visits between collaborating research groups for specific projects related to forensic genetics**
- For Terms of Reference, see
  - **https://www.isfg.org/files/ISFG_Fellowships_Nov2016.pdf**
- Financial support for travel and accommodations for **up to 1000 euros (within continent)** and **2000 euros (between continents)**
- Application rounds: (1) April 2017, (2) October 2017, (3) April 2018, (4) October 2018 – see https://www.isfg.org/Members+Area/Short+Term+Fellowships
- Selection committee included the Working Group chairs and was chaired by John Butler from the ISFG Executive Board
- Two new calls planned for 2019



Ge.F.I.

UMG
Dipartimento di Medicina Legale

SIMEF

ISFG

ASSOCIAZIONE ITALIANA BIOLOGI

UMG

UMG School of PhD Programmes

3 - 4 Settembre 2018

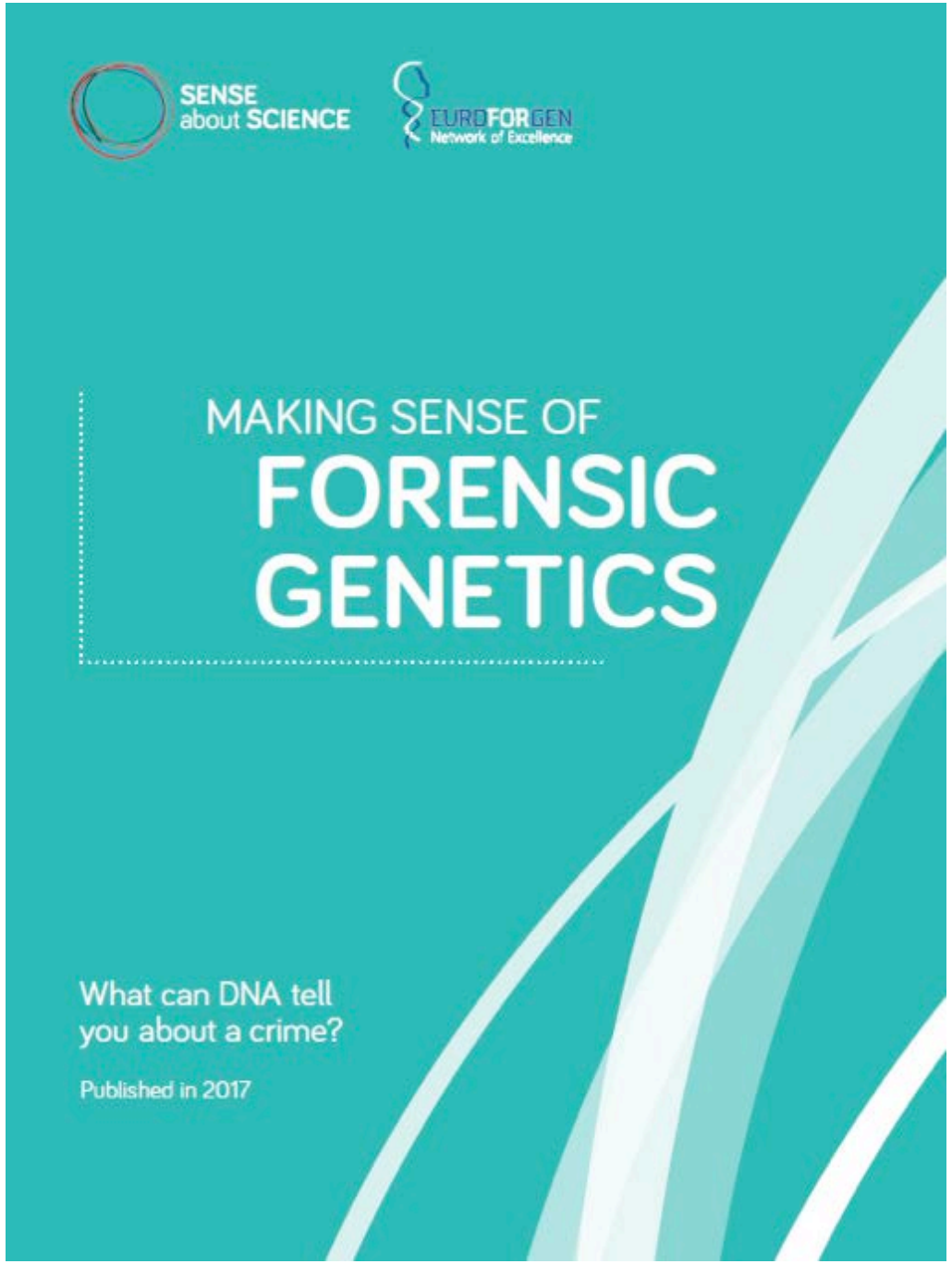
Grand Hotel Paradiso - Catanzaro

ISFG

ISFG Summer School Program

- Paternity and kinship testing including X-chromosomal markers
  - Thore Egeland, Daniel Kling (2 days)
- DNA interpretation in criminal casework using probabilistic software (LRmix Studio and Euroformix)
  - Peter Gill, Lourdes Prieto, Corina Benschop, Oyvind Bleka (2 days)
- Mitochondrial DNA analysis and interpretation using EMPOP
  - Walther Parson (0.5 days)
- Next generation sequencing and population studies using Snipper and Structure analysis
  - Christopher Phillips, Leonor Gusmao (1 day)
- ISO17025 accreditation procedures and DNA database management
  - Renato Biondo (0.5 days)


Suite



German

Italian

Polish

Portuguese

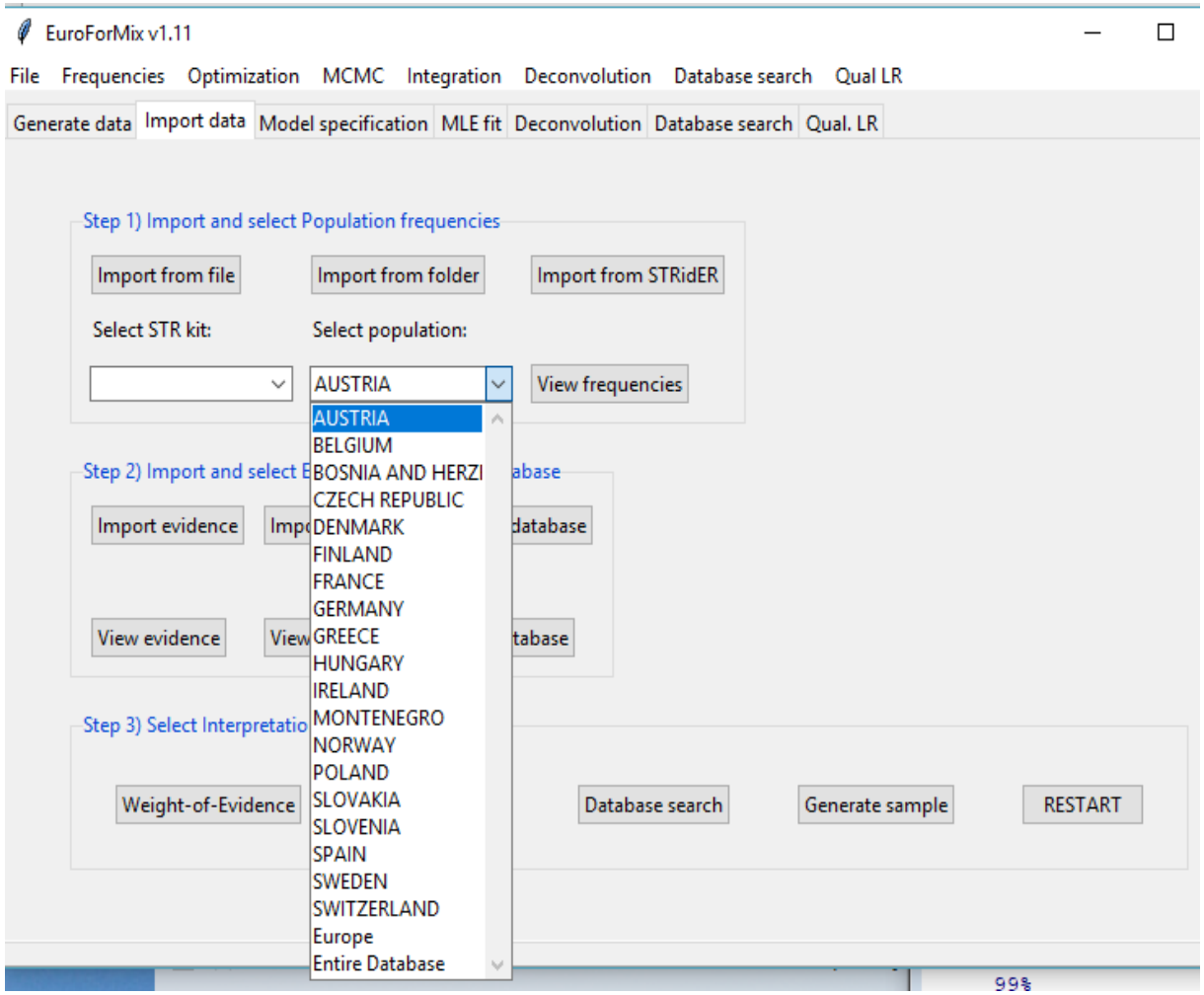
Spanish

Hungarian

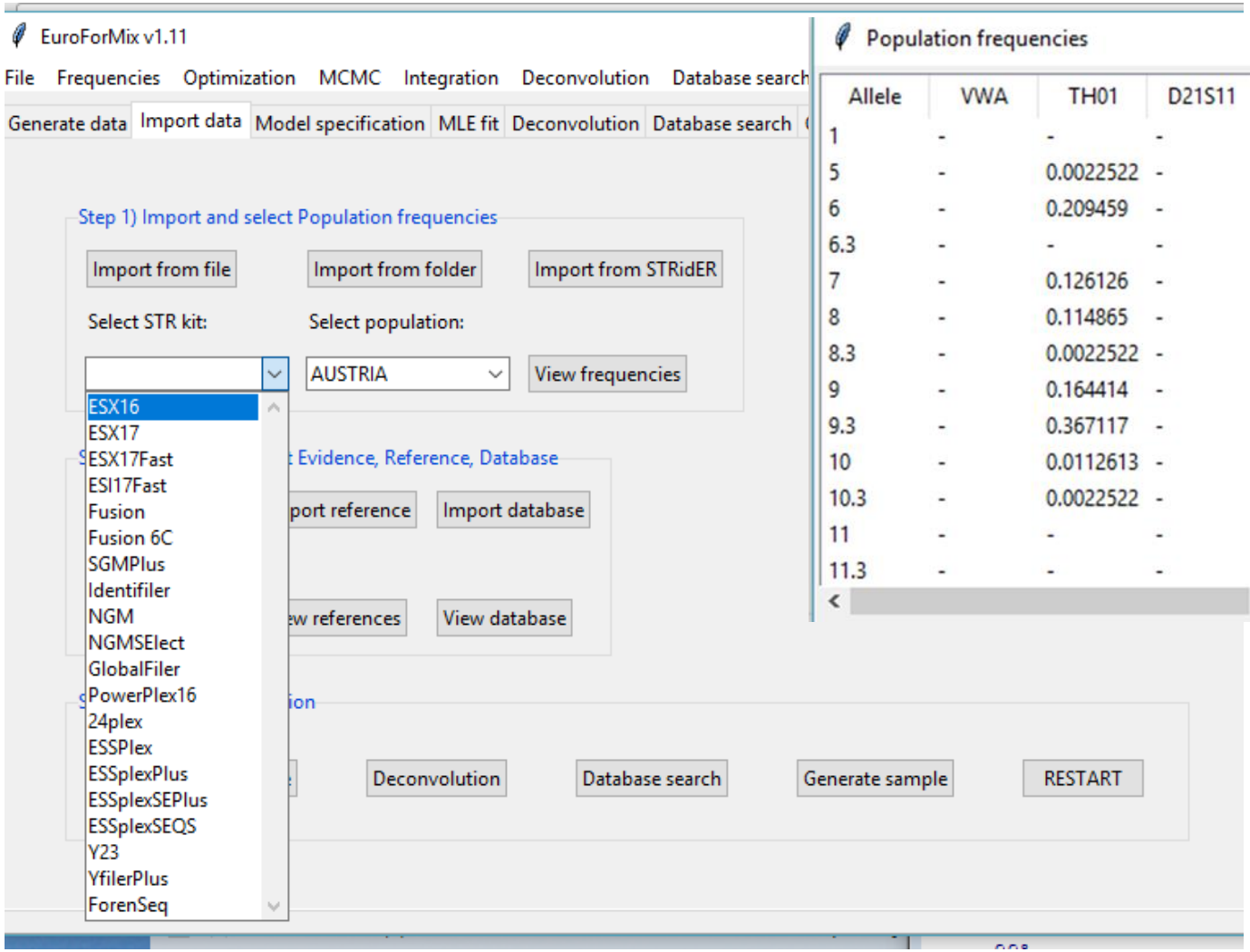
Euroformix update

Peter Gill, Oyvind Bleka

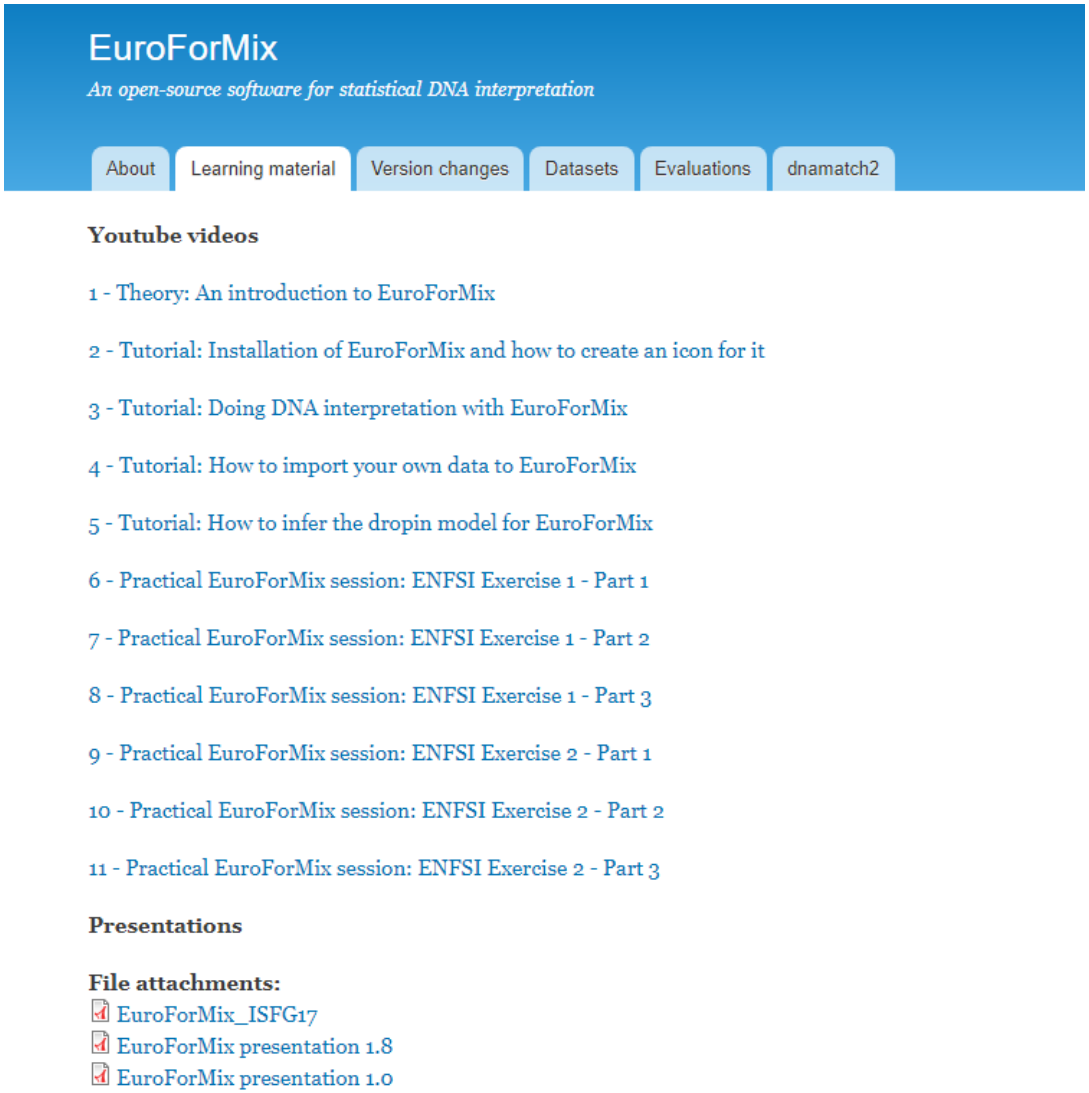
Integration with STRider



Kit selection



Teaching material available online



New developments

- Extension to MPS data
- SNP assays
- STR assays using LUS designations now possible.

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

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Research paper

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

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

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

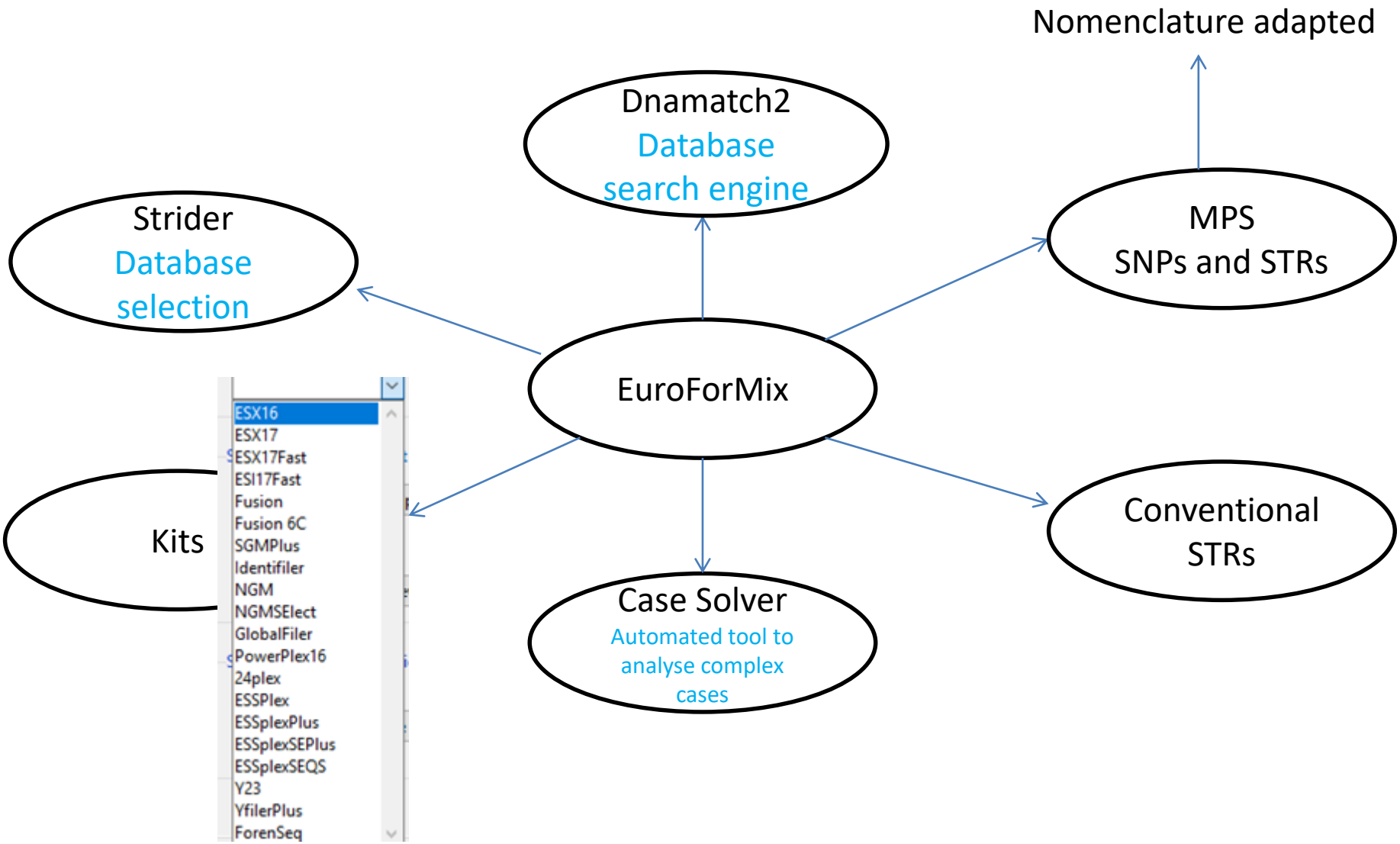
Development of a community of users

- Advantages:
  - Transparency
  - Available to all users without restriction
  - Peer review and standardisation
  - Enables adoption of (same)software by different providers
  - Encourages a community of users approach
  - Encourages cascade training of users because training materials are supplied
  - Central help desk
  - Support is provided to users for court-going purposes
  - A highly cost effective approach to adoption of fully validated methods that are supported by regular updates to programs
- From last count there are c. 40 labs using open-source in Europe.

Adoption by other providers

- DNA*xs* under development by NFI will use Euroformix as a core program to be re-written in Java
- Euroformix has been adopted by a US company ‘genetic technologies’ as “BulletProof”
- They have rewritten the gui and it has some nice features – of course the core program is the same
- Open-source has flexibility of approach that allows users to adopt the core program and to change the ‘wrapper’ according to local requirements.
- http://ednalims.com/probabilistic-genotyping/

Scope


Nomenclature adapted
Dnamatch2
Database
search engine
Strider
Database
selection
MPS
SNPs and STRs
EuroForMix
Kits
ESX16
ESX17
ESX17Fast
ESI17Fast
Fusion
Fusion 6C
SGMPlus
Identifiler
NGM
NGMSElect
GlobalFiler
PowerPlex16
24plex
ESSPlex
ESSplexPlus
ESSplexSEPlus
ESSplexSEQS
Y23
YfilerPlus
ForenSeq
Conventional
STRs
Case Solver
Automated tool to
analyse complex
cases

ISFG Summer school

Interpretation of complex DNA profile mixtures using open-source software including LRmix and EuroForMix

Teachers:

Peter Gill (University of Oslo Hospital)
Lourdes Prieto (University of Santiago de Compostela)
Oyvind Bleka (University of Oslo Hospital)
Corina Benschop (Netherlands Forensic Institute)

Aim: To provide participants with necessary skills to carry out probabilistic genotyping of complex mixtures using open-source programs to calculate the strength of evidence of complex mixtures.

Target Group: Law enforcement forensic experts – experienced reporting officers who deal routinely with DNA profiling evidence and are required to interpret complex mixtures in casework.

General Learning Outcomes:

- Understand the theory behind using likelihood ratios to interpret evidence
- Discuss the theory used to interpret complex mixtures of two or more contributors where the samples may be compromised - partial, degraded
- Describe best practice in relation to the ISFG DNA commission recommendations
- Be proficient in the use of open source software in order to carry out the calculations (LRmix Studio, EuroForMix, Case Solver)
- Write court going statements
- Describe the limitations of methods
- Demonstrate extension to Massive Parallel sequencing data for STRs and SNPs
- New developments will be disseminated, including DNAxs, a new package for reporting officers
- Participants will be provided with necessary tools to carry out cascade training at a national level

Software

Software

All software are open-source and freely available for users. Free support is provided to users via help desks found on the web-sites.

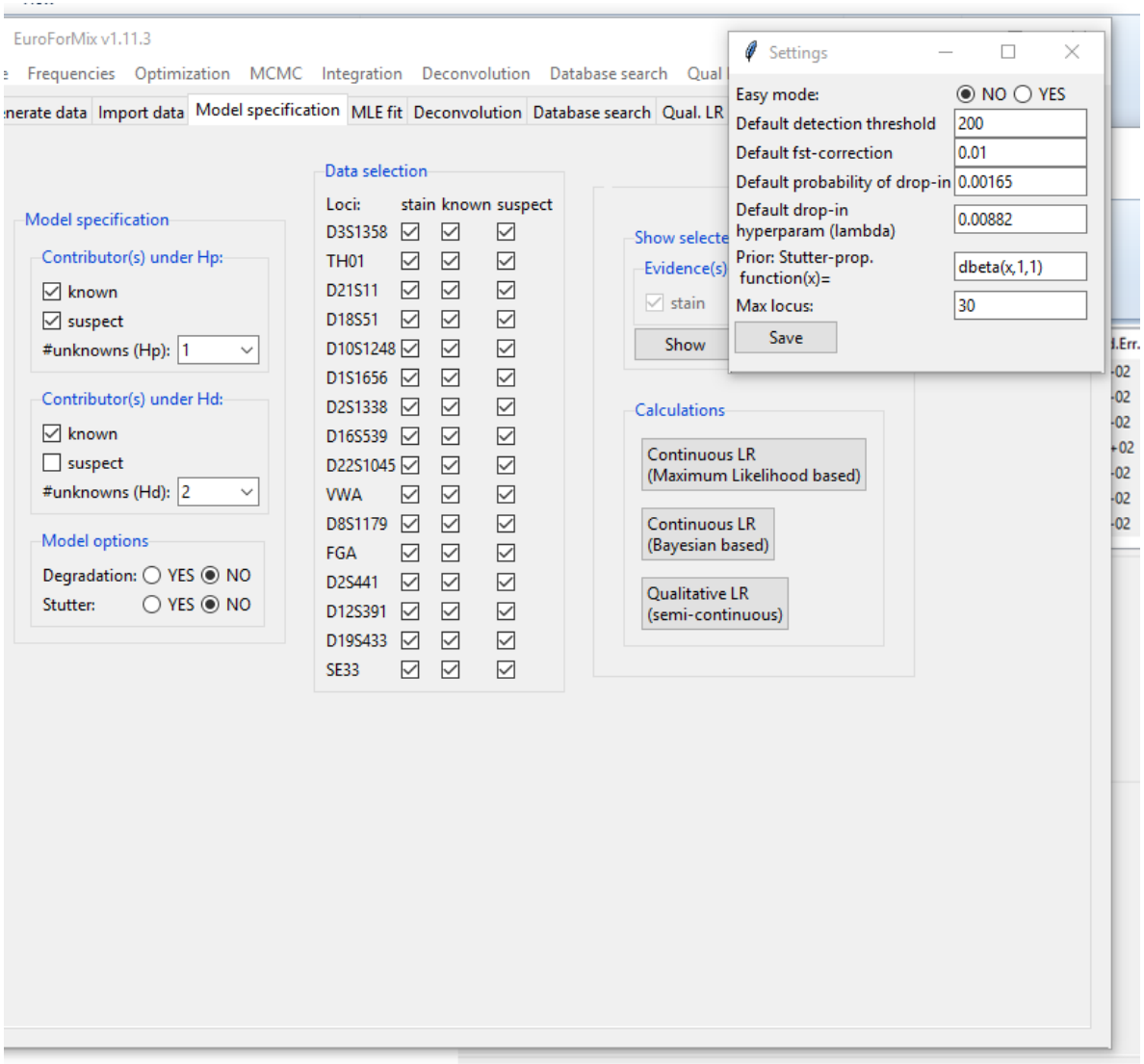
LRmix studio is a commonly used *qualitative* program that utilises the peak designation only http://lrmixstudio.org/.

EuroForMix is a more complicated *quantitative* program. Its development was supported by a EU-FP7 inititative (EuroForGen-NOE). Peak height, stutter and degradation are incorportated into the model. This package is 'state of the art', and is also used for analysis of massive parallel sequencing data http://www.euroformix.com/

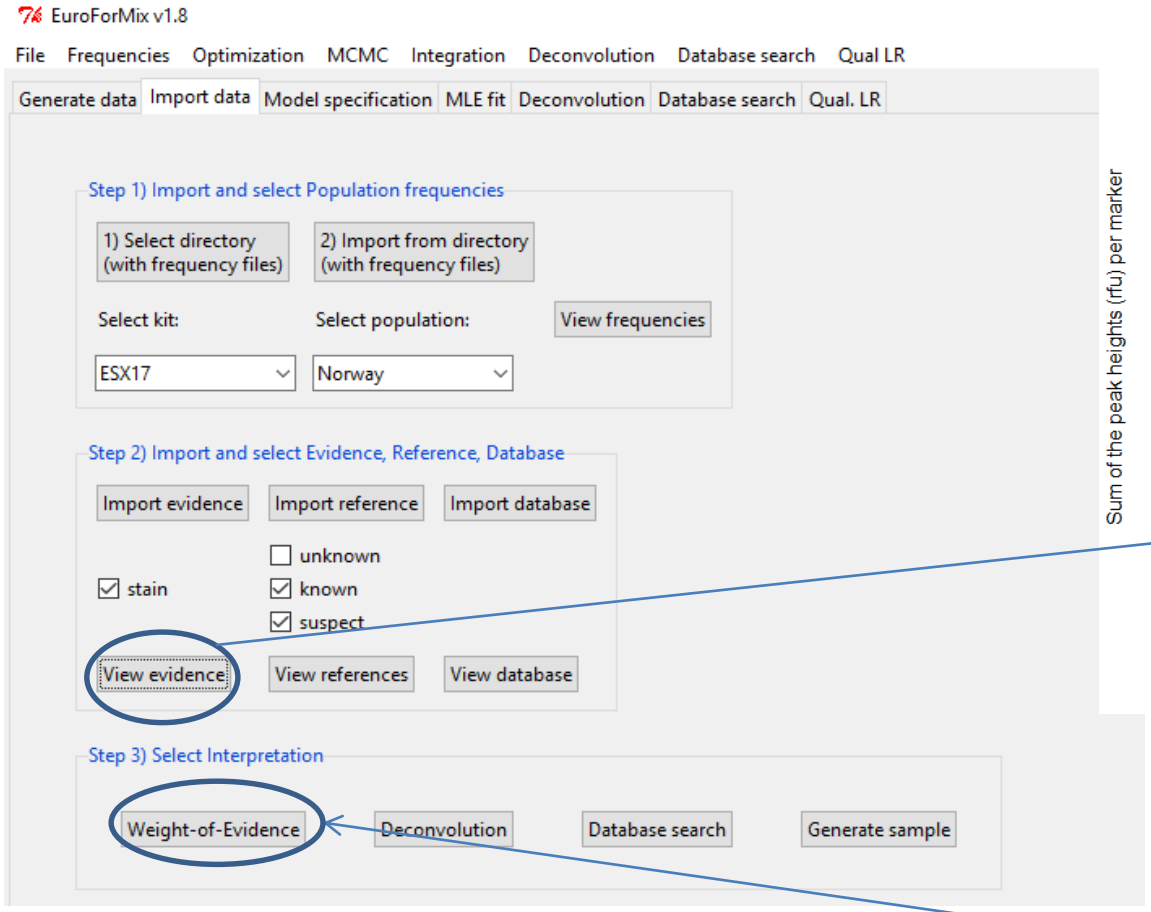
Case Solver is a program that is linked with EuroForMix. It is used to automate the analysis of complex cases where there may be multiple suspects and crime-stains http://www.euroformix.com/.

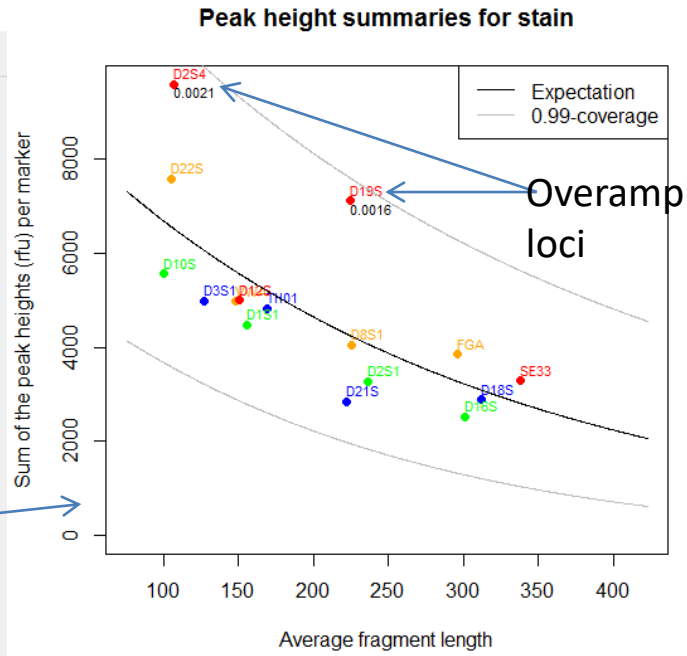
DNAxs is a new program under development by the NFI. EuroForMix will be integrated into this package, along with a host of features designed to assist the reporting officer to interpret complex DNA profiles. Corina Benschop will provide feedback and demonstrate program features.

Numbers of contributors (robber case) step 1: load settings



Euroformix Analysis
Step 1b: load the samples





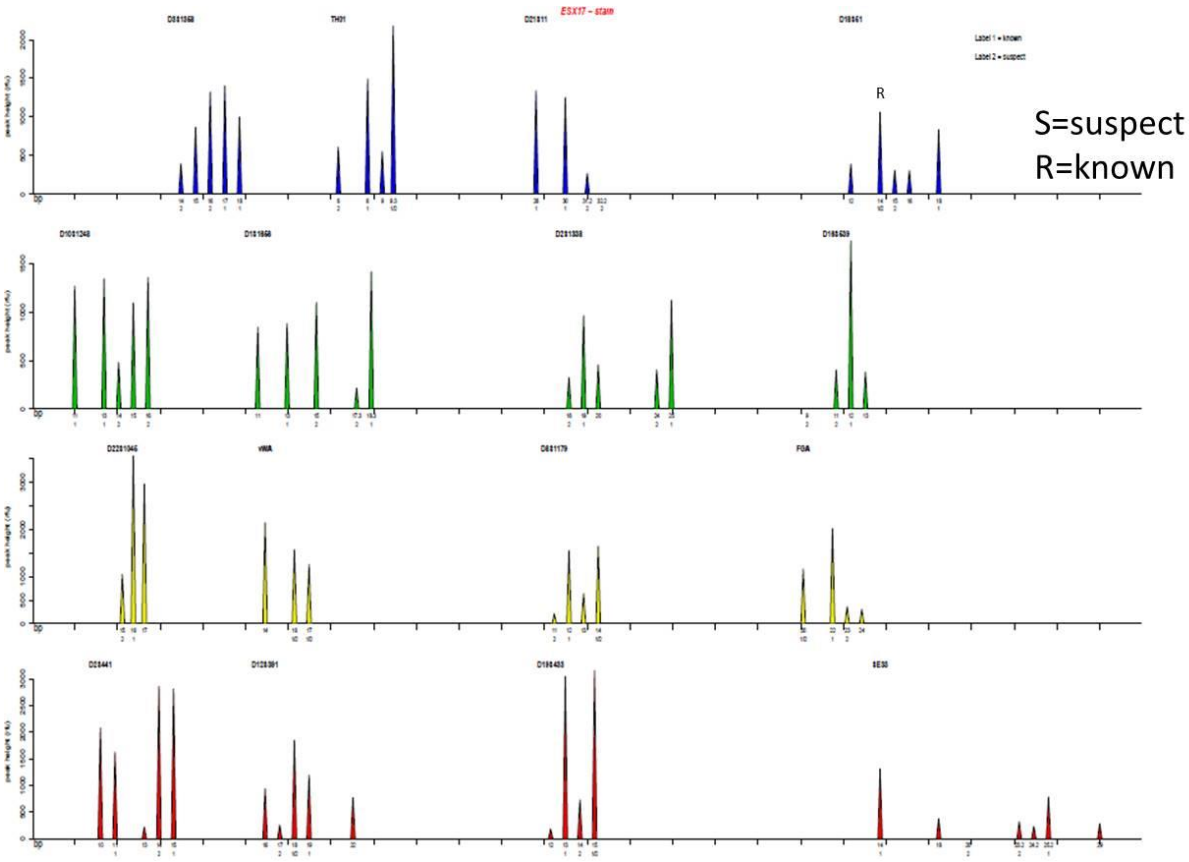
Overamplified loci

Note that it is easiest to go to the File tab and load the project from ‘robberSample’

Click on this button next

Numbers of contributors (robber case)

- Some advice



Estimation of numbers of contributors is not just about the maximum number of alleles in the profile (SE33 example)

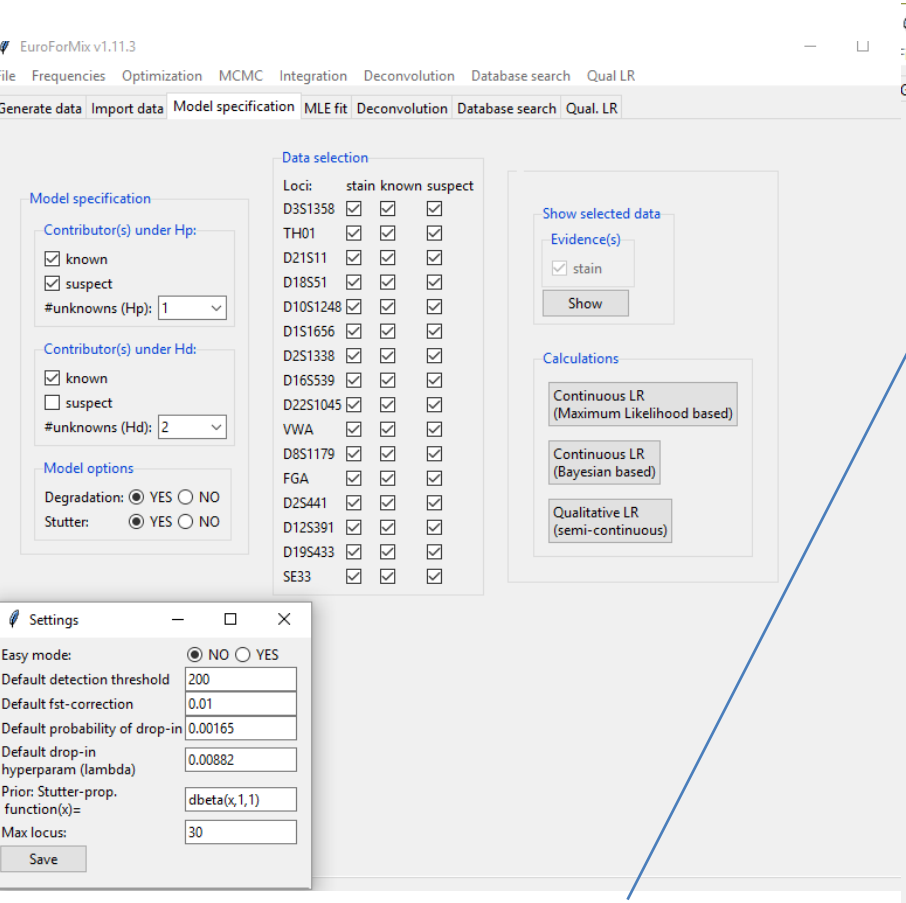
	Alleles in the crime-stain						Alleles in references			
Conditioning hypothesis	**1**	**2**	**3**	**4**	**5**	**6**	**Known**	**Suspect**	**List of unique alleles**	**Minimum no. of contributors**
Hp	14	18	23.2	24.2	25.2	29	14, 25.2	23.2, 20	14,18,20,23.2,24.2,25.2,29	4
Hd	14	18	23.2	24.2	25.2	29	14, 25.2		14,18,23.2,24.2,25.2,29	3
Peak heights	1312	373	318	229	779	279				

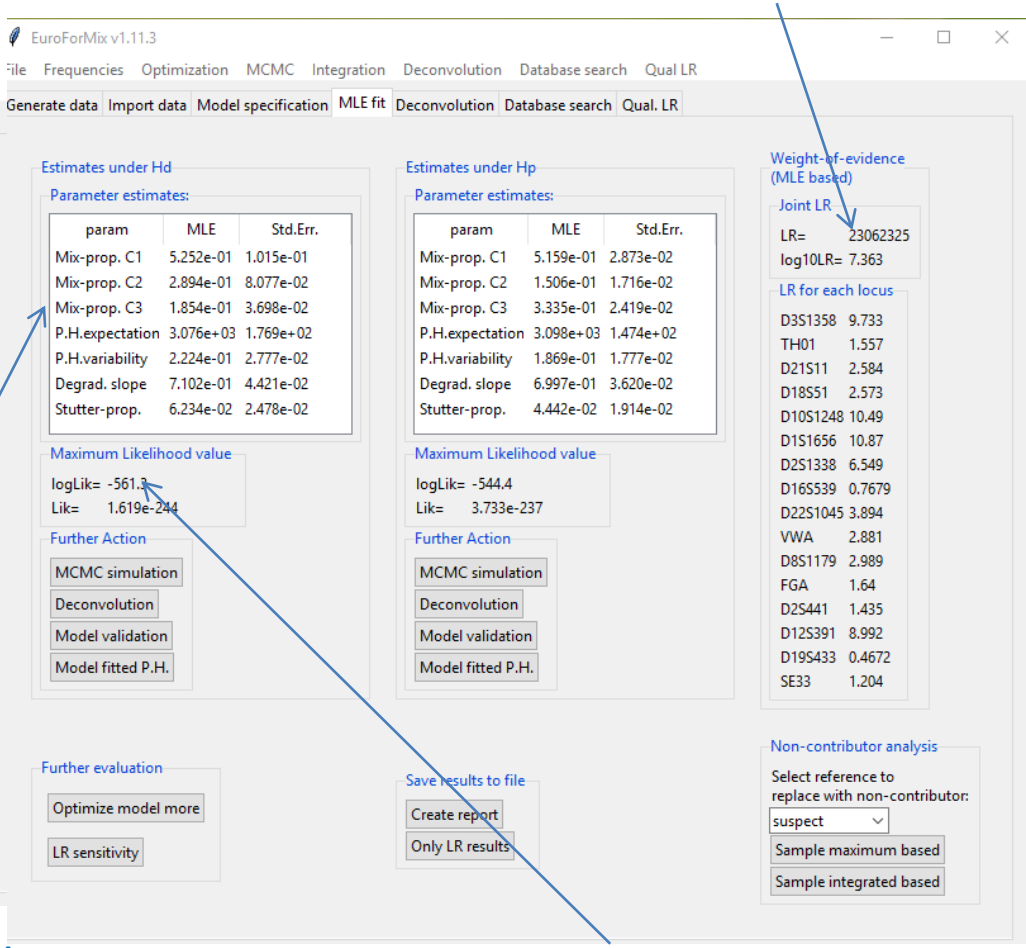
- First find the minimum number of contributors needed to explain the profile
- It is necessary to add together the unique alleles in the profile plus the conditioned genotypes
- These may be different under Hp and Hd.

Why doesn’t it make any difference if we postulate too many contributors?

Determine the best model. 2 minutes to compute 3 person mixture with stutter/deg

(unadjusted) LR

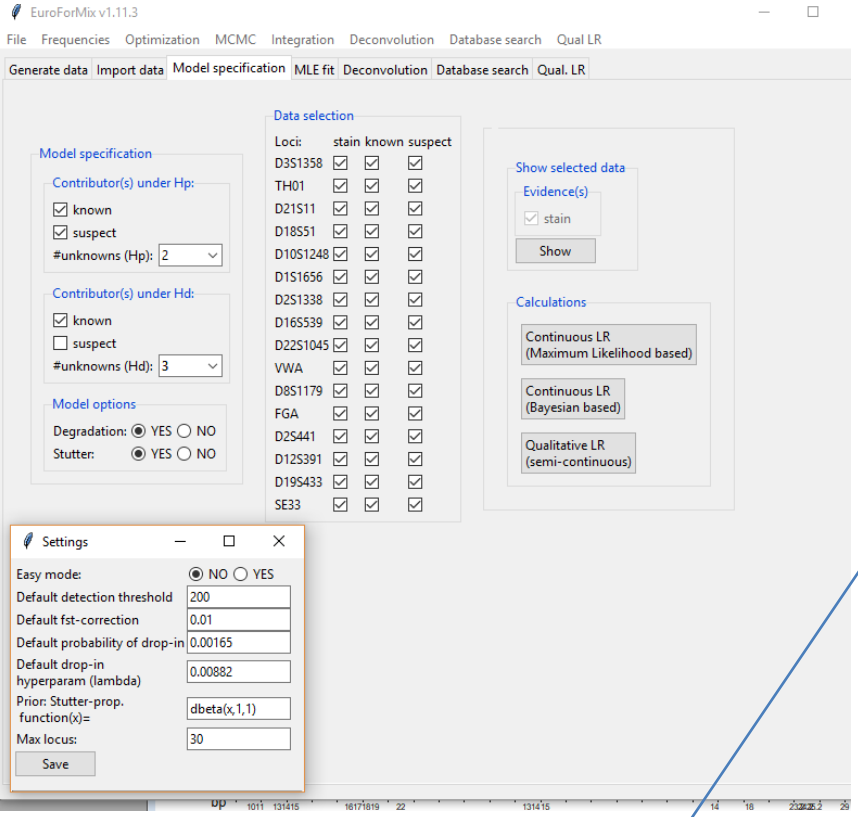


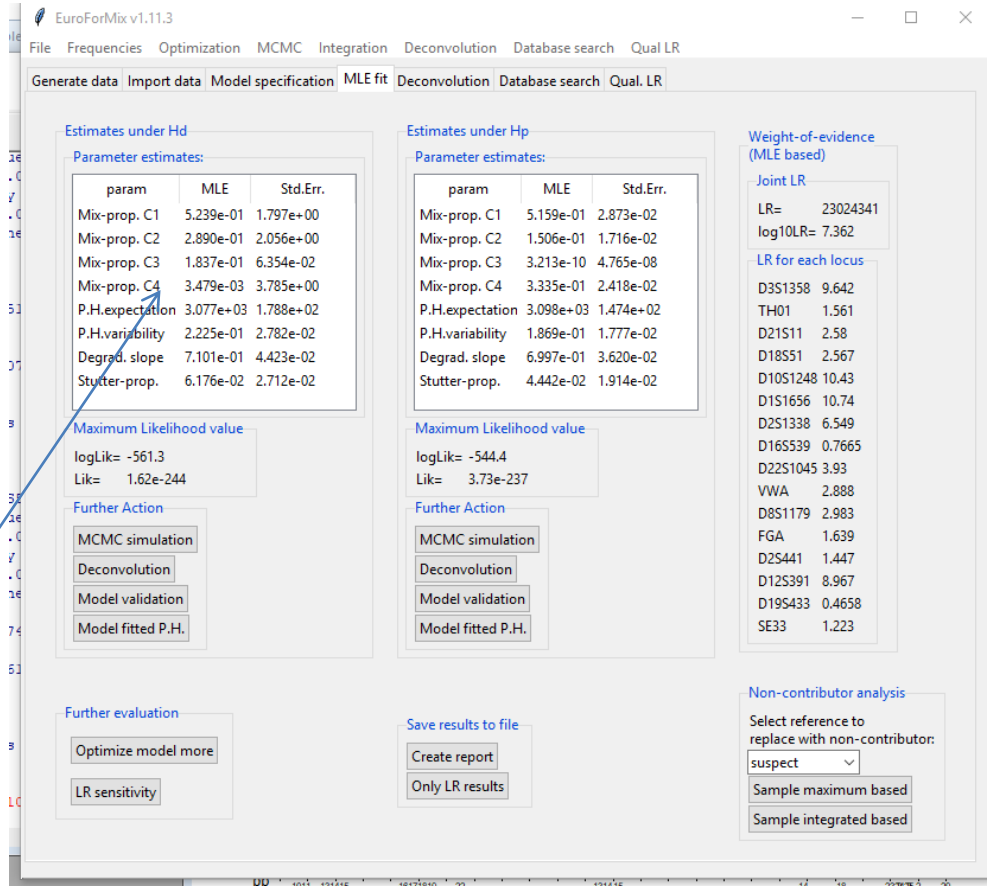


Check the mixture proportions: 0.525;0.289;0.185

Record the LogLik

Step 3: Determine the best model 120mins to compute 4 persons with stutter/deg





Mx =0.003 is very low

We can carry out multiple analyses for various contributors

- LRs are different but the best fit model is denoted by the Log Likelihood value.

EuroForMix analysis							The reported log10LR	
K	**Stutter**	**Degrad.**	**#param.**	**LogLik**	**adjusted LogLik**	**log10LR**	**EFM**	**LRmix Studio**
3	no	no	0	-577.7	-577.7	2.6	1.15	-2.85
3	no	yes	-1	-567.3	-568.3	4.5		
3	yes	no	-1	-572.7	-573.6	4.7		
3	yes	yes	-2	-561.3	**-563.3**	**7.4**	**5.44**	
4	no	no	-1	-574.9	-575.9	4	**1.94**	**1.62**
4	no	yes	-2	-564.3	-566.3	6.5		
4	yes	no	-2	-572.7	-574.7	4.6		
4	yes	yes	-3	-561.3	-564.3	7.4	6.1	
Hp=4;Hd=3	yes	yes	-2.5	-561.3	-563.8	7.4	6.3	-0.22

Summary of guidance

- To determine the number of contributors try out different numbers using exploratory analysis.
- Check whether an additional contributor is significant. If it is very low (Mx=0.001) in the example then it doesn’t make any difference.
- Decide the best model to use with LogLik analysis.

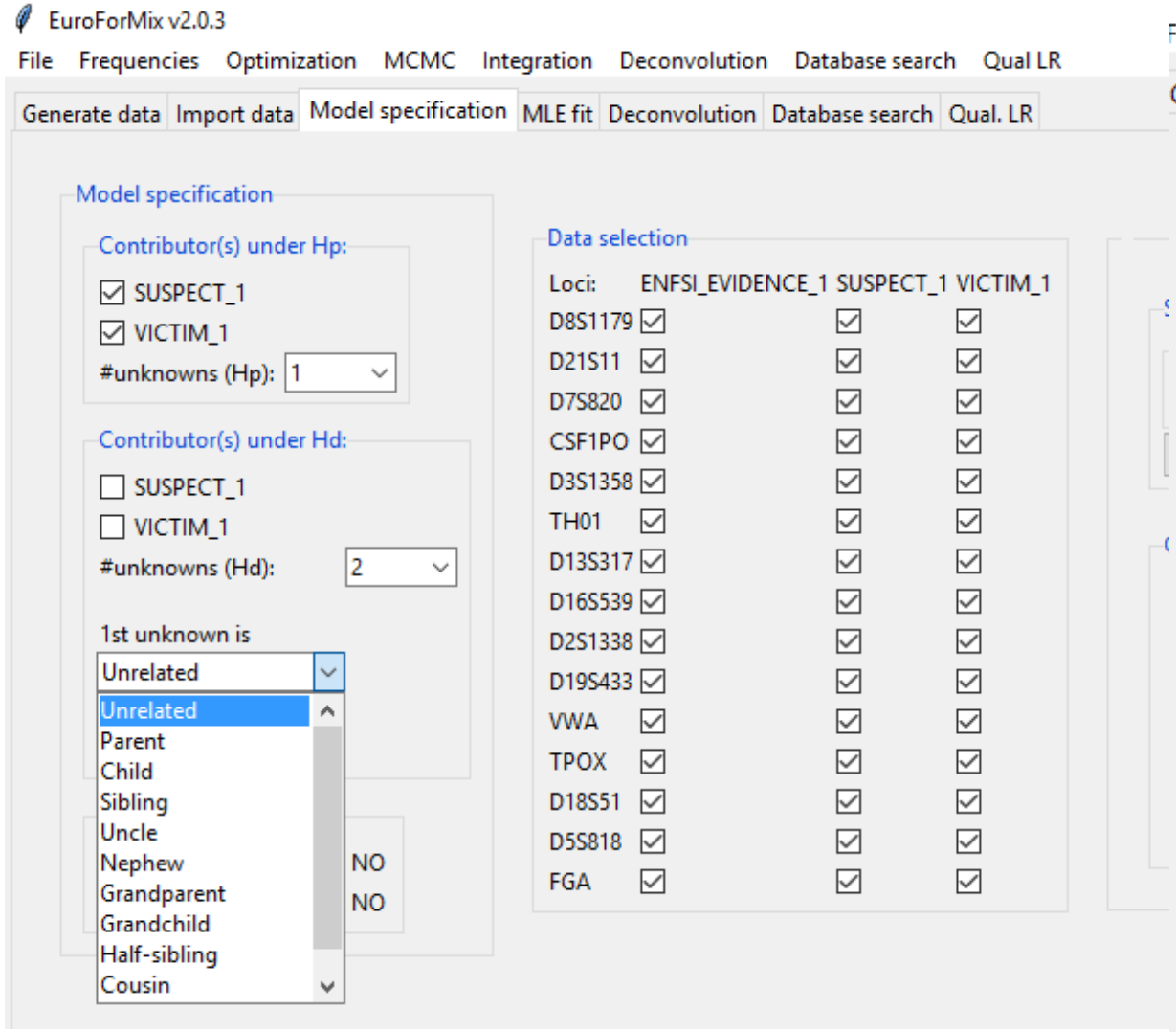
Conclusion

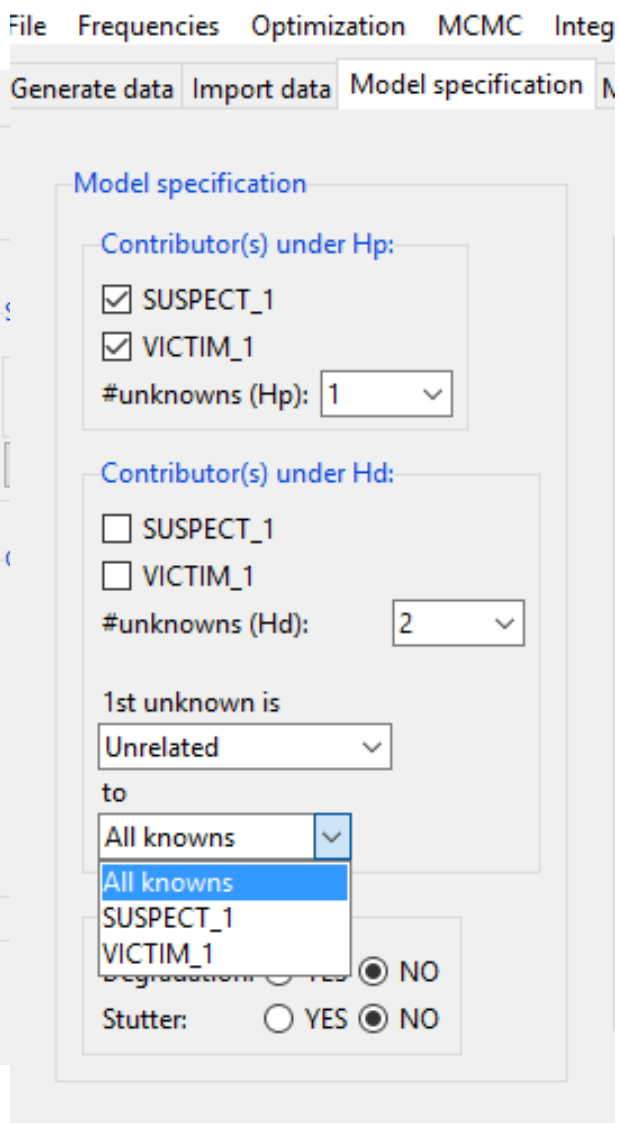
- We never know the number of contributors
- It really doesn’t matter, so long as we choose sufficient to explain the profile.
- If too many contributors are chosen, a good model will assign excess contributors a very low Mx

Euroformix version 2 release

- New update
- Supports NGS SNPs and STRs
- Forenseq multiplex is coded into the latest package

Latest developments relatedness tests

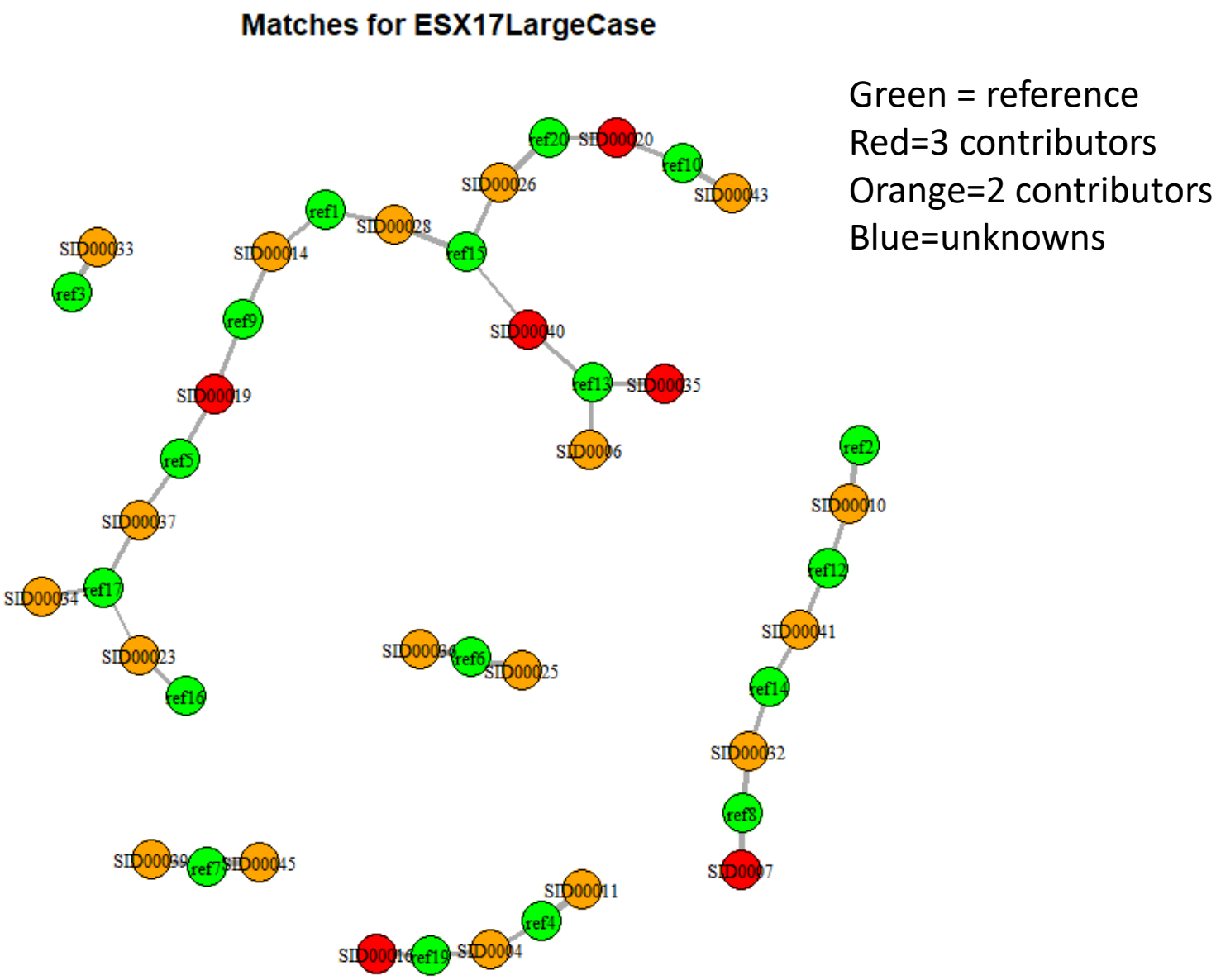


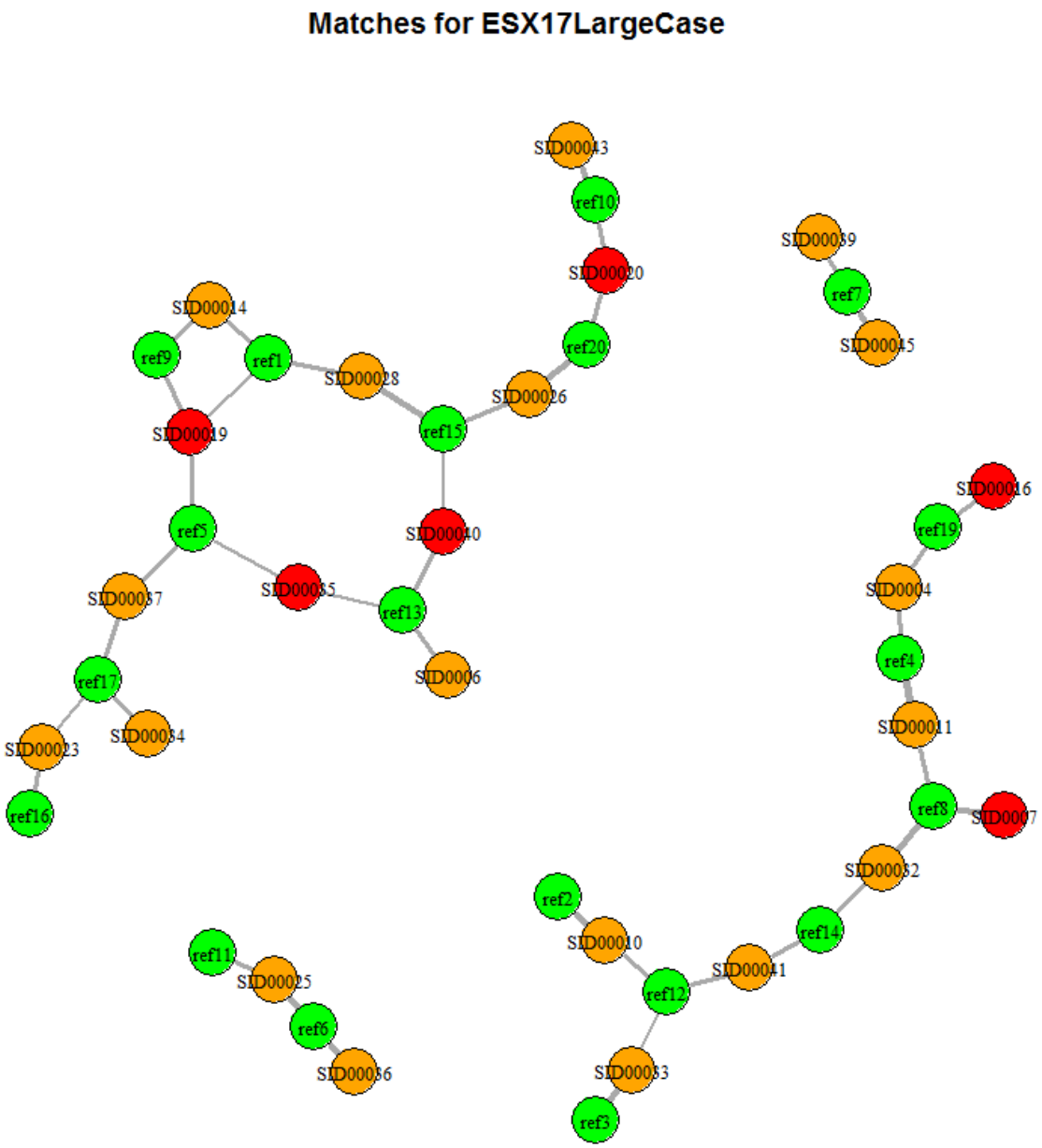


Speed of analysis is greatly improved

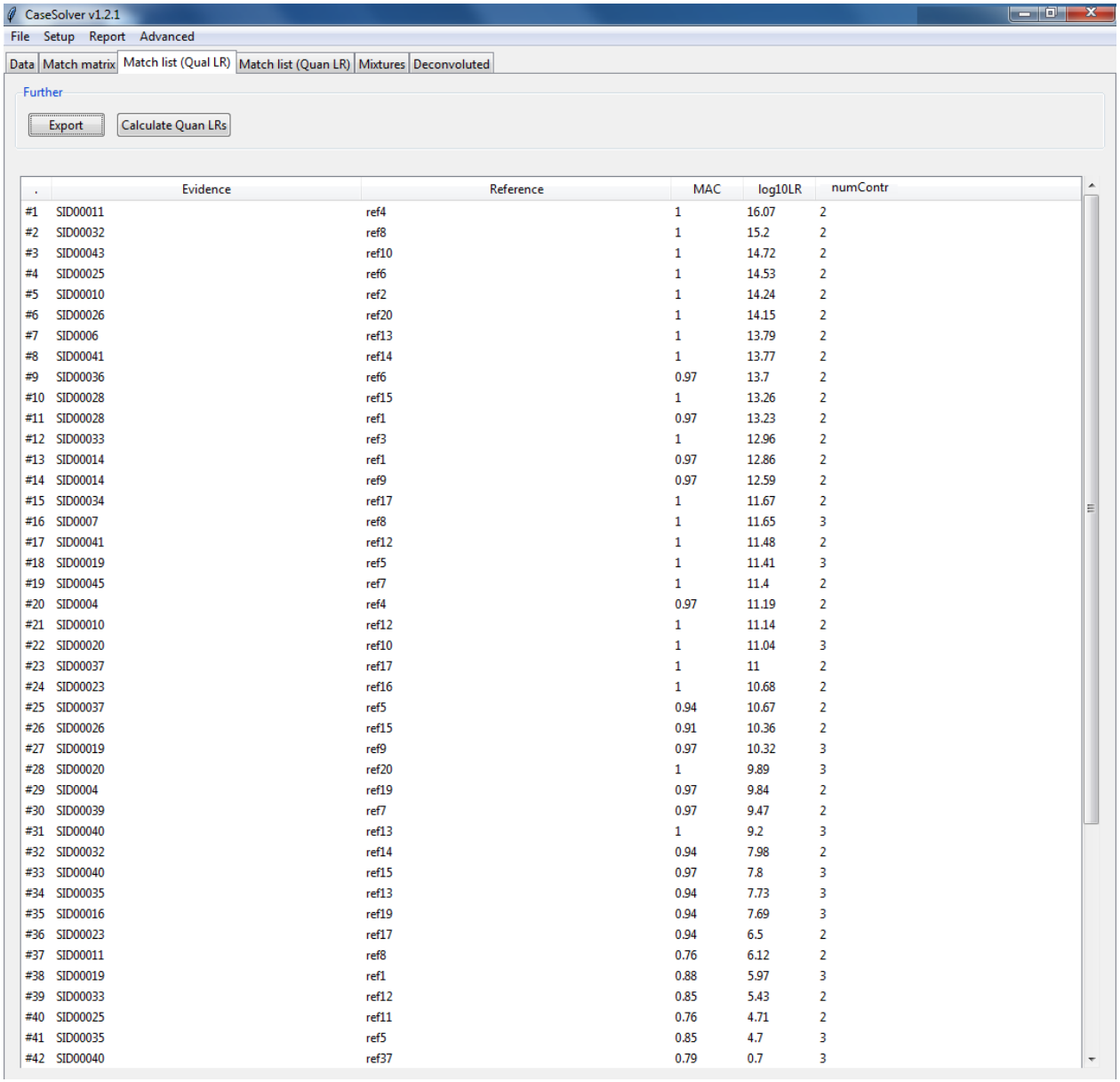
- 5 person mixture (with conditioning one person under Hd/Hp) can now be achieved in approximately 3 hours (instead of 18 hours with previous version).

Case solver

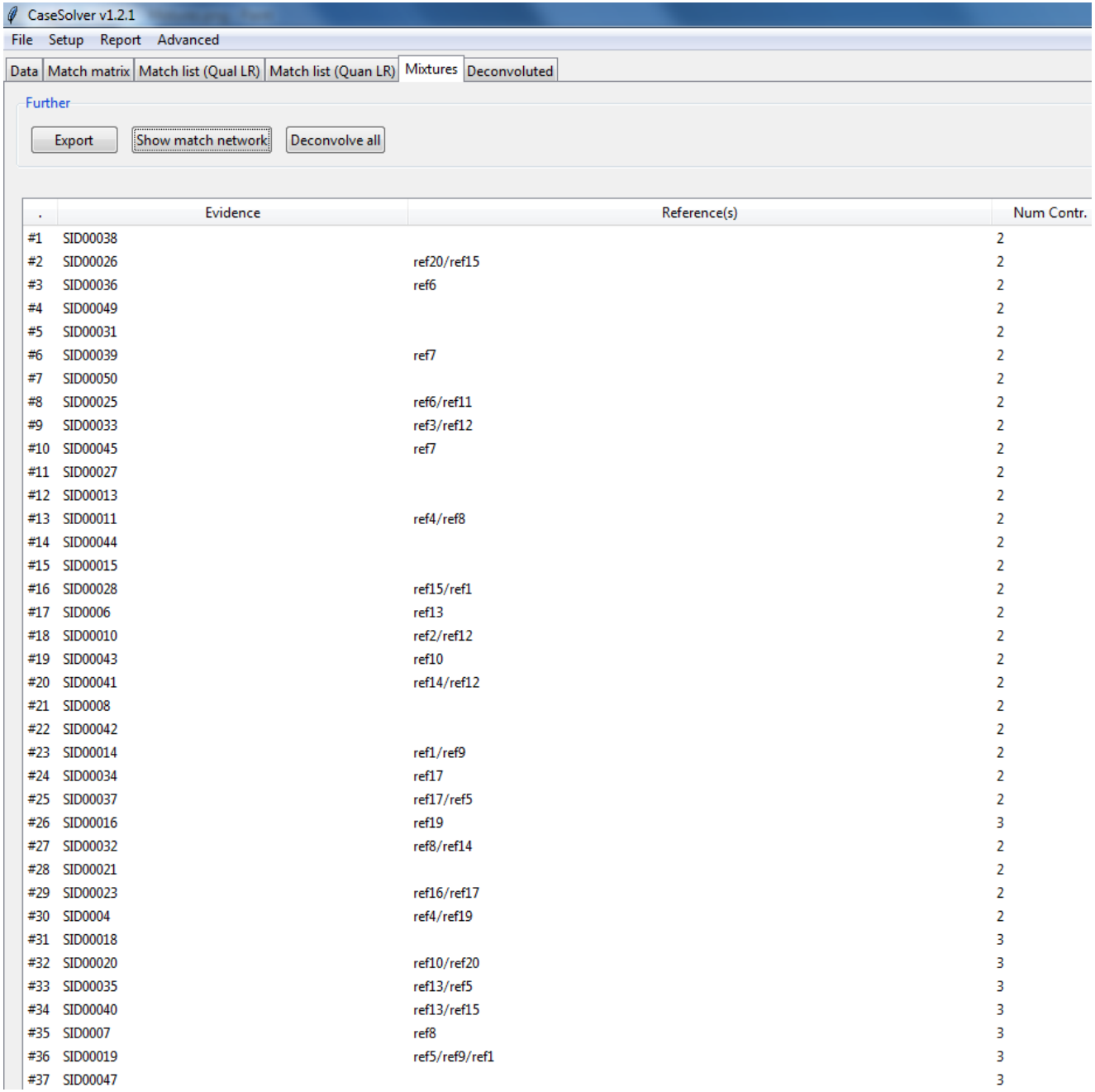

Matches for ESX17LargeCase
Green = reference
Red=3 contributors
Orange=2 contributors
Blue=unknowns
ref20
SID00020
ref10
SID00043
SID00026
ref1
SID00028
SID00033
SID00014
ref15
ref3
ref9
SID00040
SID00019
ref13
SID00035
SID00006
ref5
ref2
SID00037
SID00010
ref12
SID00034
ref17
SID00041
SID00038
ref6
SID00025
SID00023
ref14
ref16
SID00032
ref8
SID00039
ref7
SID00045
SID00007
SID00011
ref4
SID00016
ref19
SID00004



Green = reference
Red=3 contributors
Orange=2 contributors
Blue=unknowns



.	Evidence	Reference	MAC	log10LR	numContr
#1	SID00011	ref4	1	16.07	2
#2	SID00032	ref8	1	15.2	2
#3	SID00043	ref10	1	14.72	2
#4	SID00025	ref6	1	14.53	2
#5	SID00010	ref2	1	14.24	2
#6	SID00026	ref20	1	14.15	2
#7	SID0006	ref13	1	13.79	2
#8	SID00041	ref14	1	13.77	2
#9	SID00036	ref6	0.97	13.7	2
#10	SID00028	ref15	1	13.26	2
#11	SID00028	ref1	0.97	13.23	2
#12	SID00033	ref3	1	12.96	2
#13	SID00014	ref1	0.97	12.86	2
#14	SID00014	ref9	0.97	12.59	2
#15	SID00034	ref17	1	11.67	2
#16	SID0007	ref8	1	11.65	3
#17	SID00041	ref12	1	11.48	2
#18	SID00019	ref5	1	11.41	3
#19	SID00045	ref7	1	11.4	2
#20	SID0004	ref4	0.97	11.19	2
#21	SID00010	ref12	1	11.14	2
#22	SID00020	ref10	1	11.04	3
#23	SID00037	ref17	1	11	2
#24	SID00023	ref16	1	10.68	2
#25	SID00037	ref5	0.94	10.67	2
#26	SID00026	ref15	0.91	10.36	2
#27	SID00019	ref9	0.97	10.32	3
#28	SID00020	ref20	1	9.89	3
#29	SID0004	ref19	0.97	9.84	2
#30	SID00039	ref7	0.97	9.47	2
#31	SID00040	ref13	1	9.2	3
#32	SID00032	ref14	0.94	7.98	2
#33	SID00040	ref15	0.97	7.8	3
#34	SID00035	ref13	0.94	7.73	3
#35	SID00016	ref19	0.94	7.69	3
#36	SID00023	ref17	0.94	6.5	2
#37	SID00011	ref8	0.76	6.12	2
#38	SID00019	ref1	0.88	5.97	3
#39	SID00033	ref12	0.85	5.43	2
#40	SID00025	ref11	0.76	4.71	2
#41	SID00035	ref5	0.85	4.7	3
#42	SID00040	ref37	0.79	0.7	3

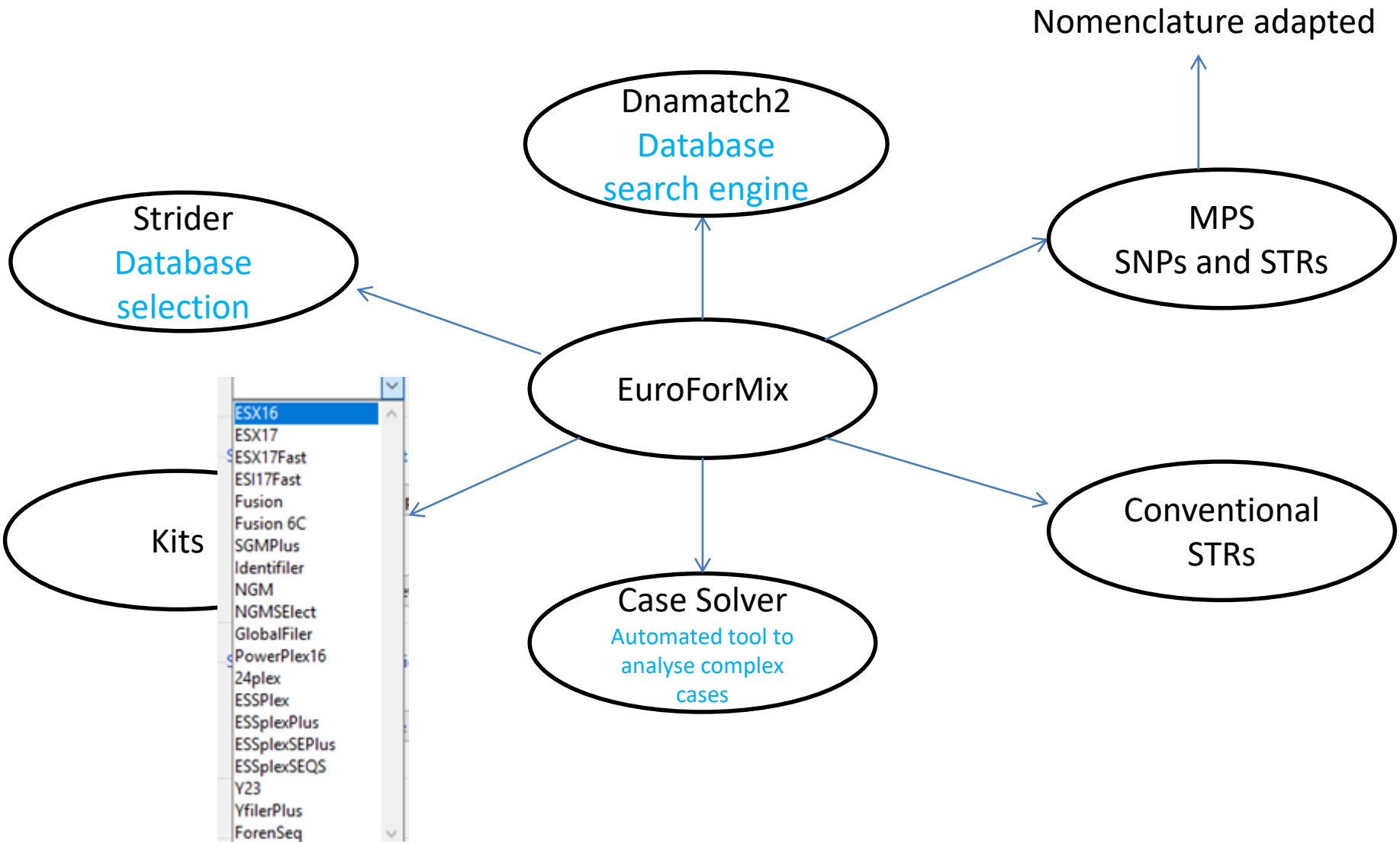


.	Evidence	Reference(s)	Num Contr.
#1	SID00038		2
#2	SID00026	ref20/ref15	2
#3	SID00036	ref6	2
#4	SID00049		2
#5	SID00031		2
#6	SID00039	ref7	2
#7	SID00050		2
#8	SID00025	ref6/ref11	2
#9	SID00033	ref3/ref12	2
#10	SID00045	ref7	2
#11	SID00027		2
#12	SID00013		2
#13	SID00011	ref4/ref8	2
#14	SID00044		2
#15	SID00015		2
#16	SID00028	ref15/ref1	2
#17	SID0006	ref13	2
#18	SID00010	ref2/ref12	2
#19	SID00043	ref10	2
#20	SID00041	ref14/ref12	2
#21	SID0008		2
#22	SID00042		2
#23	SID00014	ref1/ref9	2
#24	SID00034	ref17	2
#25	SID00037	ref17/ref5	2
#26	SID00016	ref19	3
#27	SID00032	ref8/ref14	2
#28	SID00021		2
#29	SID00023	ref16/ref17	2
#30	SID0004	ref4/ref19	2
#31	SID00018		3
#32	SID00020	ref10/ref20	3
#33	SID00035	ref13/ref5	3
#34	SID00040	ref13/ref15	3
#35	SID0007	ref8	3
#36	SID00019	ref5/ref9/ref1	3
#37	SID00047		3

Case Solver

- Continued development
- Now possible to use conditioning profiles
- Very fast software used to investigate unlimited numbers of stains and reference samples – in fact entire databases can be used.
- Practical limitation of number of contributors=3
- Investigative tool

Scope


Nomenclature adapted
Dnamatch2
Database
search engine
Strider
Database
selection
MPS
SNPs and STRs
EuroForMix
Kits
ESX16
ESX17
ESX17Fast
ESI17Fast
Fusion
Fusion 6C
SGMPlus
Identifiler
NGM
NGMSElect
GlobalFiler
PowerPlex16
24plex
ESSPlex
ESSplexPlus
ESSplexSEPlus
ESSplexSEQS
Y23
YfilerPlus
ForenSeq
Conventional
STRs
Case Solver
Automated tool to
analyse complex
cases

Education

- It is important that biologists can understand the software mechanism
- There is a barrier between statisticians and biologists which inhibits understanding.
- Use of complicated formulae
- Solution is to prepare simple explanations that are spreadsheet based so that users can explore the formular behind the spreadsheet.

Educational initiative
Explaining how does it work?

- Mastermix excel spreadsheet has been expanded to include the determination of probabilistic weights

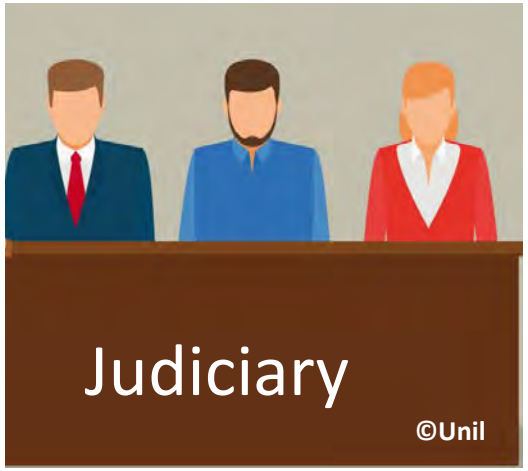
	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O
	D8S1179						**Expected peak height (shape*scale)**				**weightings**				**weights**
		Height	Proportions	Allele	Frequency	Genotype	**A**	**B**	**C**	**Q**	**A**	**B**	**C**	**Q**	
	A	**288**	**0.098**	**9.000**	0.0158	**AB,CQ**	301.810	301.810	783.281	783.281	2.90E-03	7.90E-09	4.91E-04	5.11E-11	**5.751E-25**
	B	**1525**	**0.518**	**13.000**	0.2852	**AC,BQ**	301.810	783.281	301.810	783.281	2.90E-03	3.18E-05	1.10E-06	1.14E-11	**1.160E-24**
	C	**1129**	**0.384**	**14.000**	0.2482	**AQ,BC**	301.810	783.281	783.281	301.810	2.90E-03	3.18E-05	4.91E-04	2.55E-03	**1.155E-13**
	SUM	2942		Q	0.4508	**BC,AQ**	783.281	301.810	301.810	783.281	5.94E-05	7.90E-09	1.10E-06	5.11E-11	**2.640E-29**
						BQ,AC	783.281	301.810	783.281	301.810	5.94E-05	7.90E-09	4.91E-04	2.55E-03	**5.884E-19**
						CQ,AB	783.281	783.281	301.810	301.810	5.94E-05	3.18E-05	1.10E-06	2.55E-03	**5.301E-18**
	Mx	**1-Mx**				**AA,BC**	603.6207592	783.2807	783.2807		5.68E-04	3.18E-05	4.91E-04		8.87E-12
0	0.278143	0.72				**BB,AC**	783.2806998	603.620759	783.2807		5.94E-05	3.22E-06	4.91E-04		9.40E-14
1						**CC,AB**	783.2806998	783.2807	603.620759		5.94E-05	3.18E-05	1.13E-04		2.13E-13
2	**Parameters (global)**					**AB,AC**	1085.091079	301.81038	783.2807		3.25E-07	7.90E-09	4.91E-04		1.26E-18
3	shape	16.47842	(alpha)			**BC,AC**	783.2806998	301.81038	1085.09108		5.94E-05	7.90E-09	1.41E-03		6.61E-16
4	scale	6.58E+01	(beta)			**AB,BC**	301.8103796	1085.09108	783.2807		2.90E-03	3.62E-04	4.91E-04		5.15E-10
5						**BC,AA**	1566.5614	301.81038	301.81038		5.78E-12	7.90E-09	1.10E-06		5.02E-26
6	**Parameters per contributor**					**AC,BB**	301.8103796	1566.5614	301.81038		2.90E-03	1.26E-03	1.10E-06		4.02E-12
7	shape A=	4.583357	in contributor 1			**AB,CC**	301.8103796	301.81038	1566.5614		2.90E-03	7.90E-09	5.45E-04		1.25E-14
8	shape A=	11.89507	in contributor 2			**AC,AB**	1085.091079	783.2807	301.81038		3.25E-07	3.18E-05	1.10E-06		1.14E-17
9	scale=	65.8492	universal			**AC,BC**	301.8103796	783.2807	1085.09108		2.90E-03	3.18E-05	1.41E-03		1.30E-10
0	mu=	301.8104	in contributor 1			**BC,AB**	783.2806998	1085.09108	301.81038		5.94E-05	3.62E-04	1.10E-06		2.36E-14
1	mu=	783.2807	in contributor 2												
2														**St dev**	0.246
3														mu	1.085E+03
4															

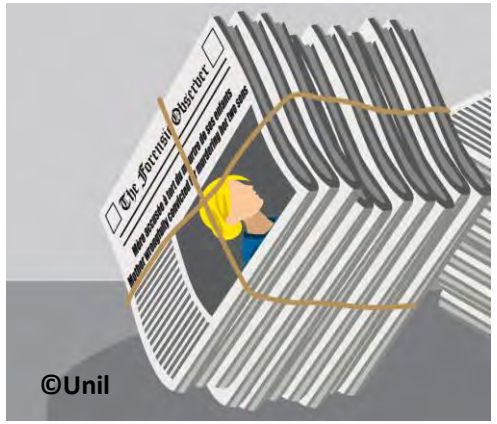
O	P	Q	R	S	T	U
weights						
	Pr(g|Hd)	Product Hd	Pr g|Hp	product Hp	LR with peak heights	LR without weighting
5.751E-25	0.002016751	1.15979E-27	0.22377712	7.288E-11	90.62173307	4.876806695
1.160E-24	0.002016751	2.3403E-27	0.25713632	1.870E-11	23.24943852	18.51396408
1.155E-13	0.002016751	2.3285E-16	0.14157328	1.63458E-14	0.020325113	7.270832097
2.640E-29	0.002016751	5.32513E-32	0.01424528	2.182E-16	0.00027132	0.482792597
5.884E-19	0.002016751	1.18657E-21	0.00784312	4.615E-21	5.73798E-09	0.402802059
5.301E-18	0.002016751	1.06912E-20	0.00901232	4.77763E-20	5.94073E-08	0.462849102
8.87E-12	3.53424E-05	3.13584E-16	0.14157328			
9.40E-14	0.000637952	5.99587E-17	0.00784312			
2.13E-13	0.000555188	1.18512E-16	0.00901232			
1.26E-18	0.0001	8.92021E-23	0.00784312			
6.61E-16	0.001110376	7.3423E-19	0.00784312			
5.15E-10	0.001275904	6.56806E-13	0.14157328			
5.02E-26	3.53424E-05	1.77453E-30	0.00024964			
4.02E-12	0.000637952	2.56406E-15	0.08133904			
1.25E-14	0.000555188	6.92849E-18	0.06160324			
1.14E-17	7.06847E-05	8.03727E-22	0.00901232			
1.30E-10	0.001110376	1.44083E-13	0.14157328			
2.36E-14	0.001275904	3.0157E-17	0.00901232			
	0.0194714	**8.04217E-13**				
0.246		hp	7.288E-11	85.47118229		
1.085E+03		hd	8.52679E-13			

MOOC Challenging forensic science: how Science should speak to Court

Christophe Champod, Franco Taroni
Alex Biedermann, Tacha Hicks

Assessing and conveying the value of forensic science results is a challenge for all stakeholders:





Media

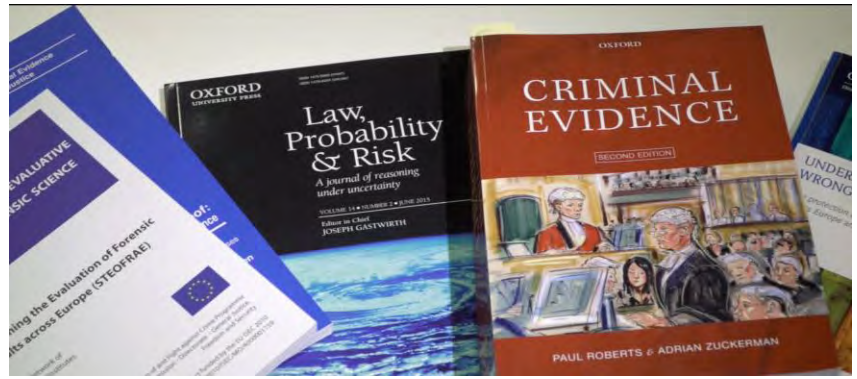
General public

100% scientific certainty…

We must go beyond conventional image that is promoted through TV series.

Alert on the limits of the techniques for sound administration of forensic science

Education is key!



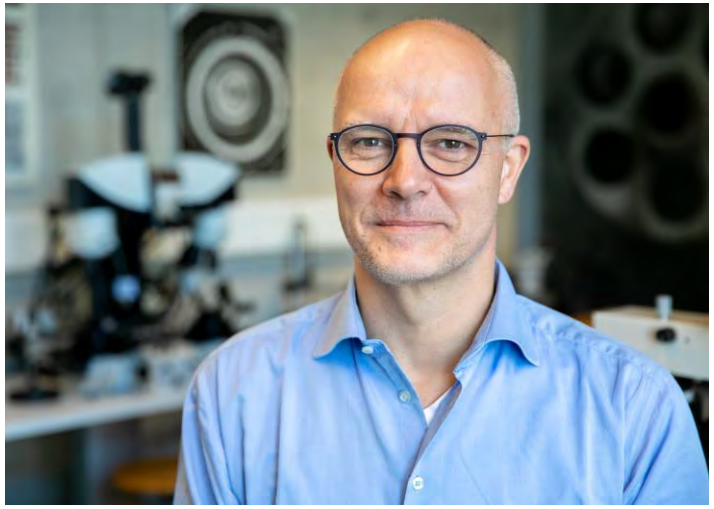
University curricula, on-going education, conferences, articles, books…

MOOC to promote critical thinking with regard to forensic science

Causes célèbres supplemented with interviews

Aim of the course : realise that uncertainty is an inevitable part of forensic science and probabilistic reasoning is essential to avoid misleading the court.

The MOOC team

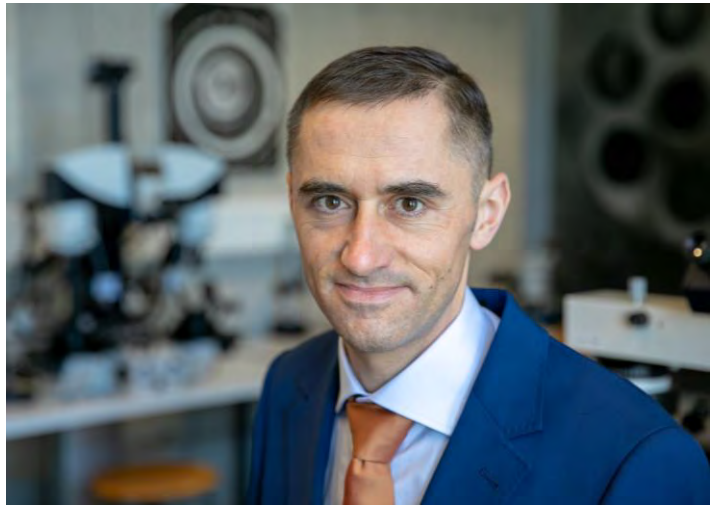


Franco Taroni

Tacha Hicks

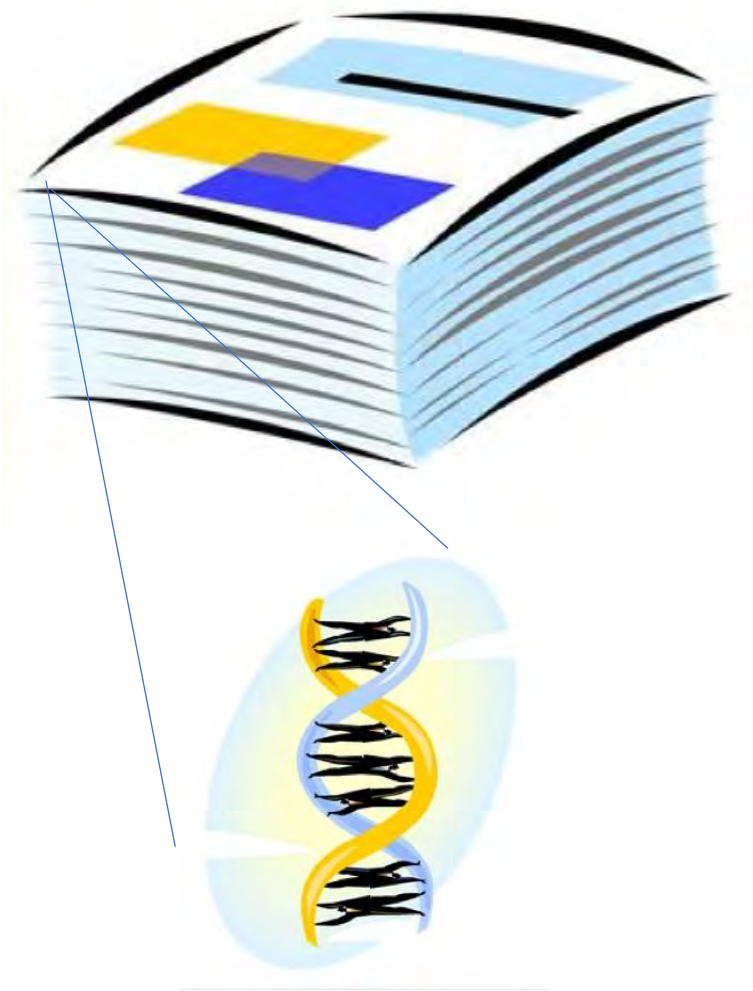
Christophe Champod

Alex Biedermann



Batochime: one week, one room

What is the DNA of a good forensic report?



Forensic science and evaluative reporting

Uncertainty in the criminal trial

Principles of forensic reporting

ENFSI guideline for evaluative reporting

Elementary: source is not activity!

DNA recovered on a suspect: Weller case

Gunshot residues on a suspect: George case

DNA recovered on a victim: Butler and Nealon cases

DNA is not the magic bullet -

DNA in the laboratory

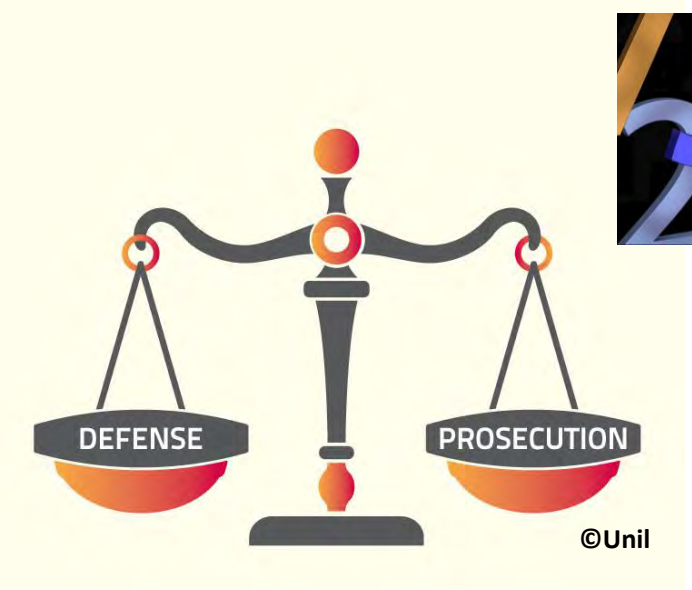
Small quantities of DNA: the Knox and Sollecito case

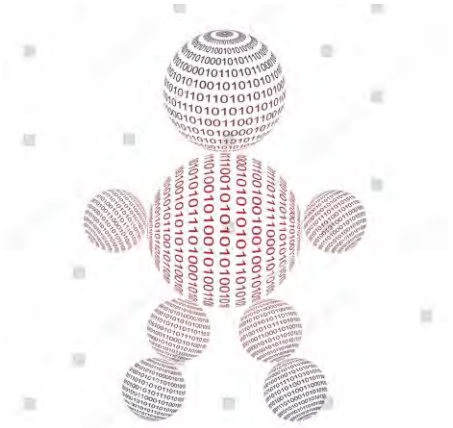
Transfer and pollution: the Jama case

Transfer and pollution: Anderson and Scott cases

Interviews

Trials by numbers ≠ numbers on trial





The defendant :Mr. Number

Beware of the transposed conditional
Adams and Dreyfus cases

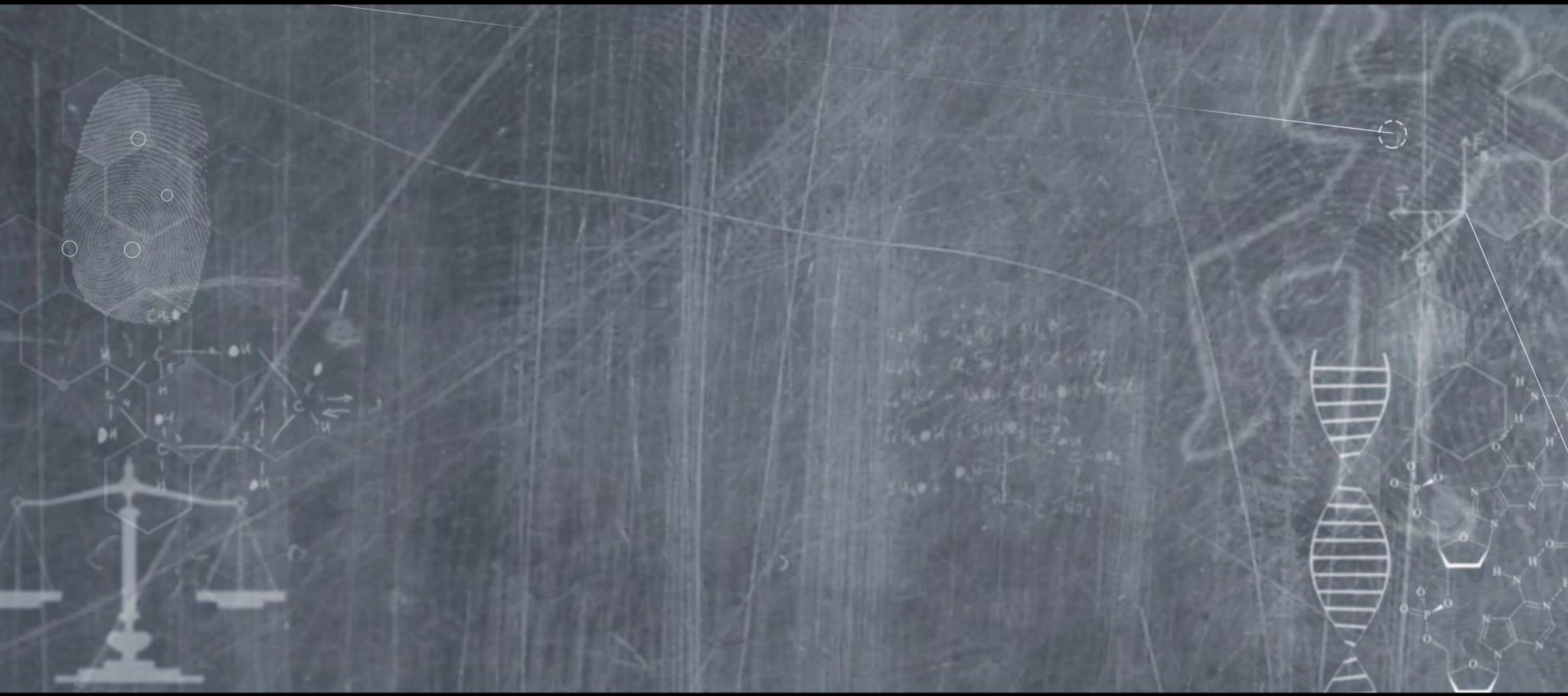
Statistics in Court: Clark and Collins cases

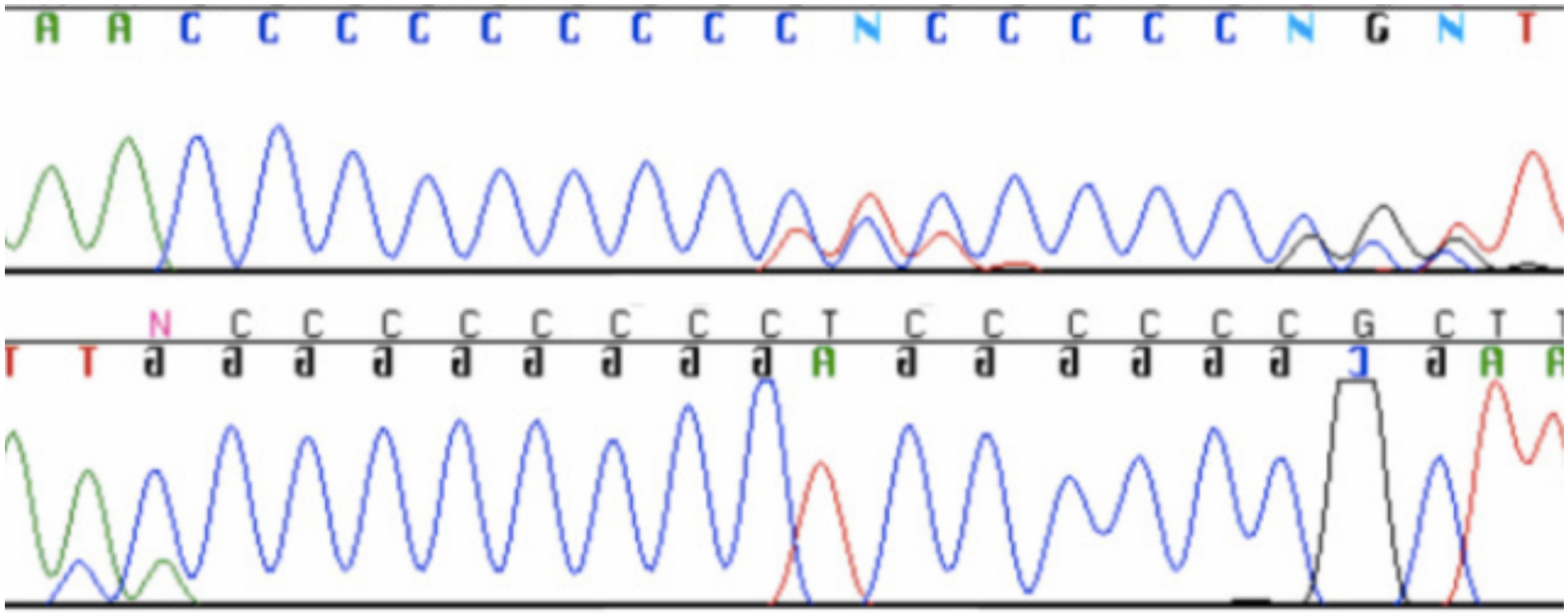
The wonderland of certainty

Identification with earmarks:
The Dallagher case

Identification with fingermarks:
The Mayfield case

Identification with fingermarks:
The McKie case





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
walther.parson@i-med.ac.at

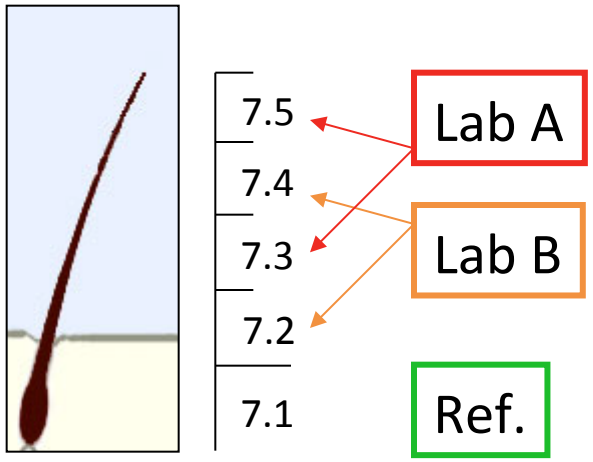
EMPOP

Point (sequence) heteroplasmy - PHP

Length heteroplasmy - LHP

Point heteroplasmy EDNAP study

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

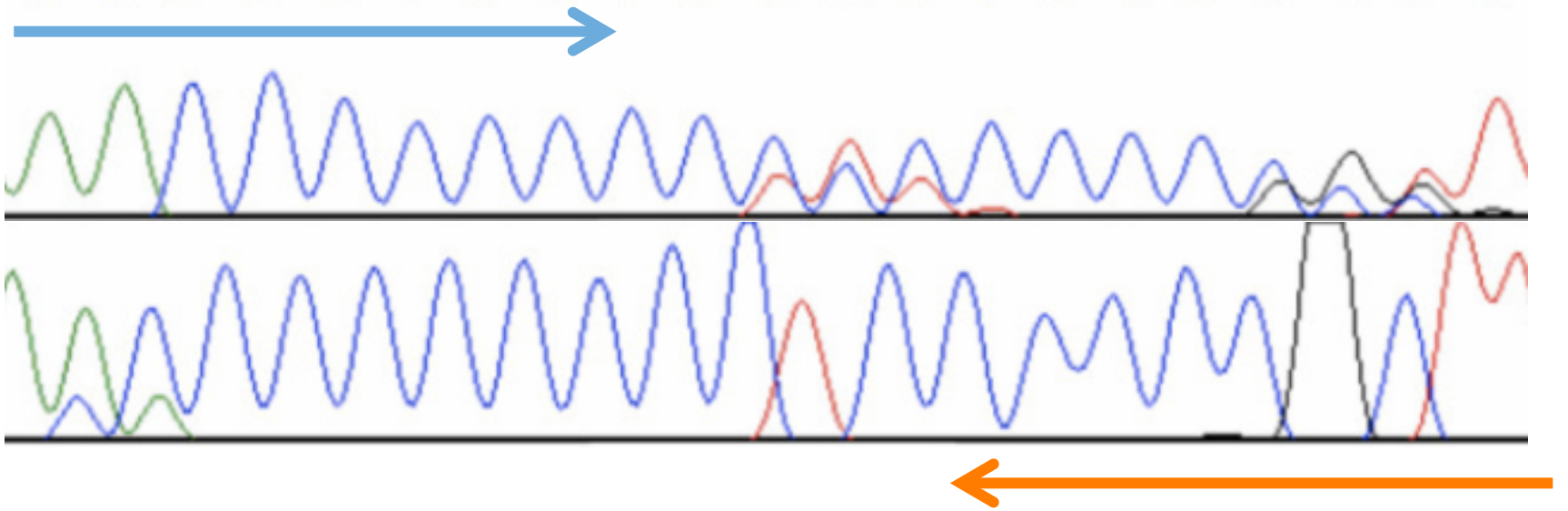
Donor‘s haplotype (blood)

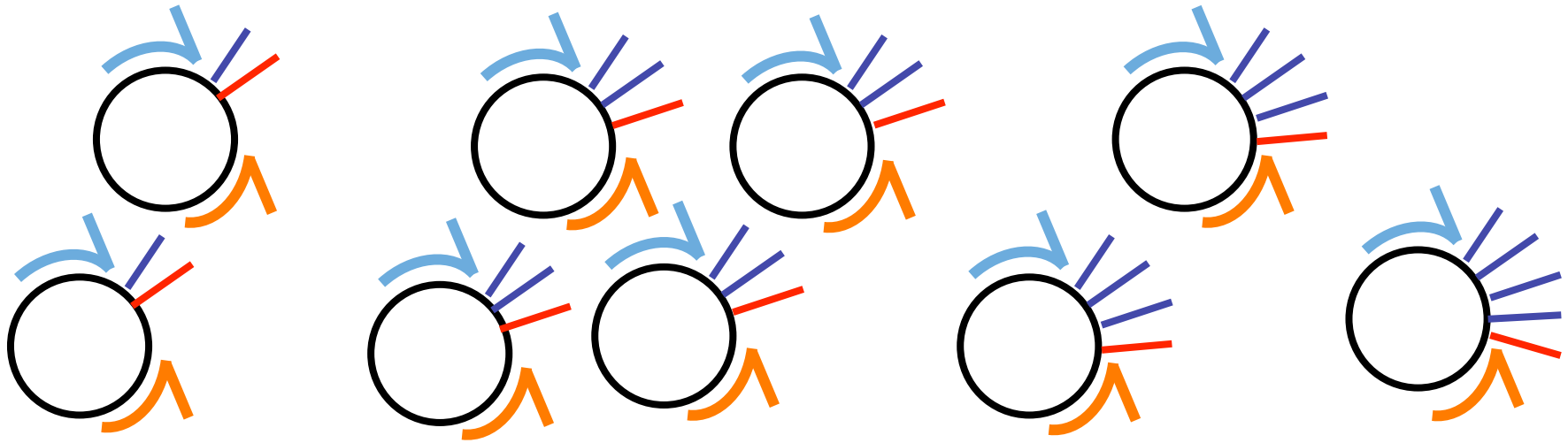
16093C 16129A 16162G 16172C 16234Y 16304C
73G 249DEL 263G 309.1C 315.1C (hg F1a1)

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

Tully et al (2004)

Length heteroplasmy





Length heteroplasmy

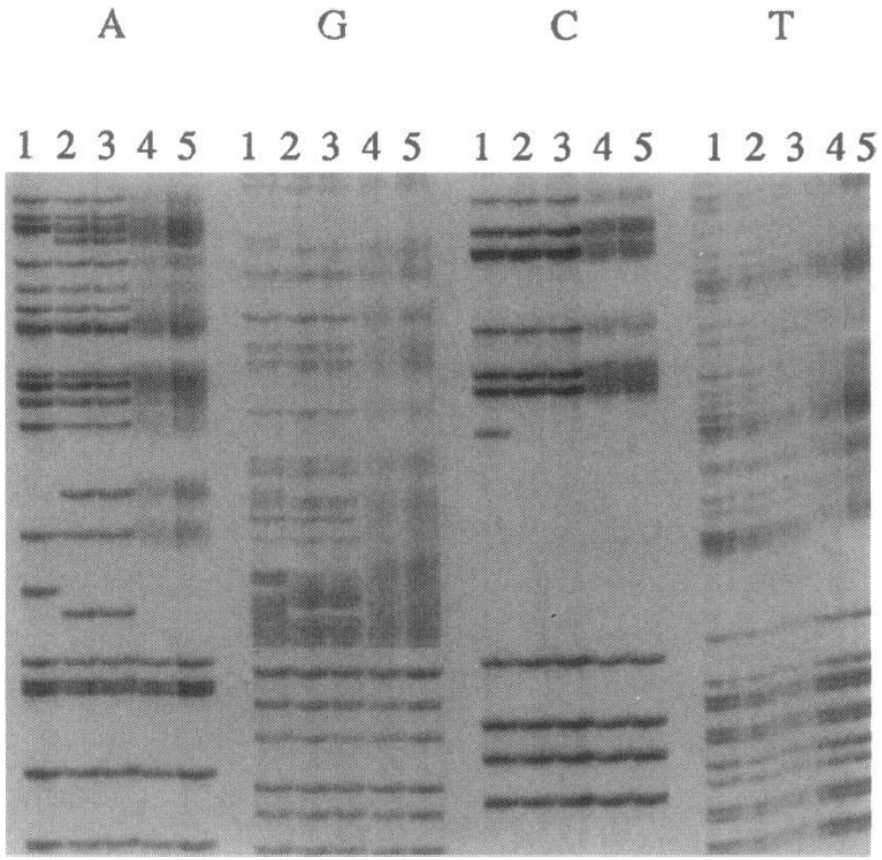
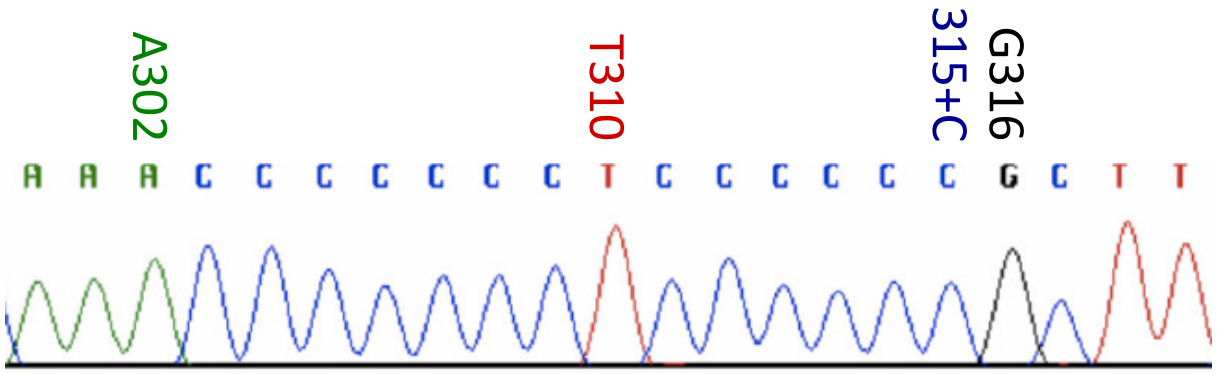


Figure 2 Heavy-strand sequences of individuals with and without variants in the homopolymeric tract. Lane 1, Variants at nt 16186 and 16189. Lanes 2 and 3, No variants. Lanes 4 and 5, Variant at nt 16189. A, G, C, and T lanes, Short run.

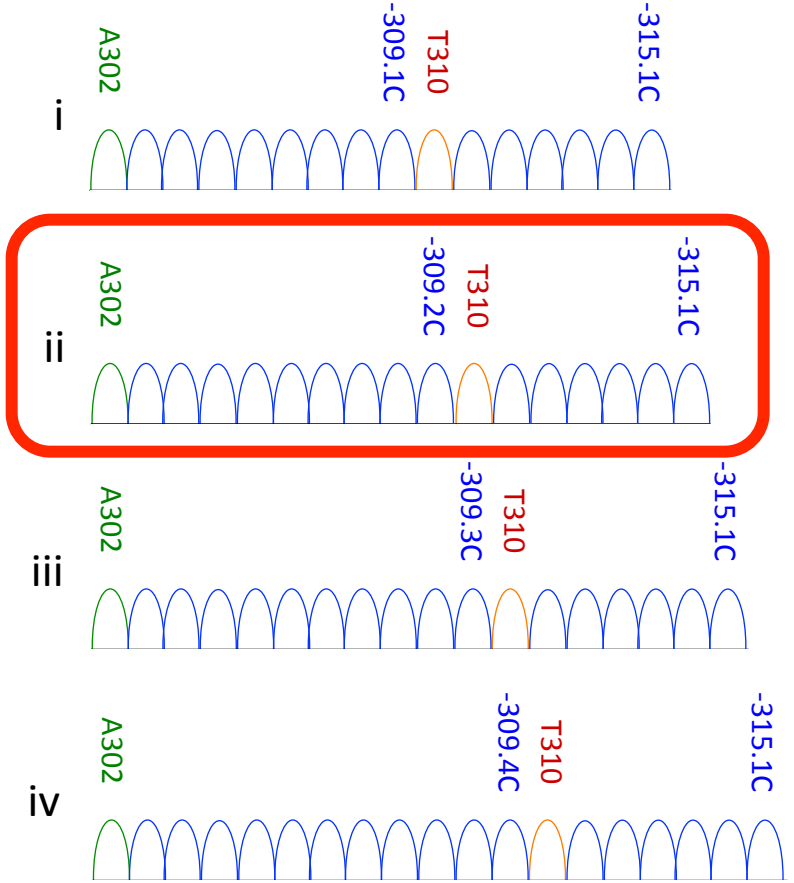
Bendall and Sykes (1995)

- LHP in HVS-I C-tract 16189C
  (Bendall and Sykes 1995)
- Excessive LHP in C-tracts > 8Cs
  (Parson et al 1998)
- Quantitative profiling of LHP
  (Lee et al 2004)
- Different inheritance of HVS-II LHP
  (Asari et al 2008)
- Segregation in MZ twins
  (Lutz-Bonengel et al 2008)
- 6% generational mutation rate
  (Forster et al 2010)
- Dominant variant interpretation
  (Berger et al 2011)

Length Heteroplasmy Dominant Variant

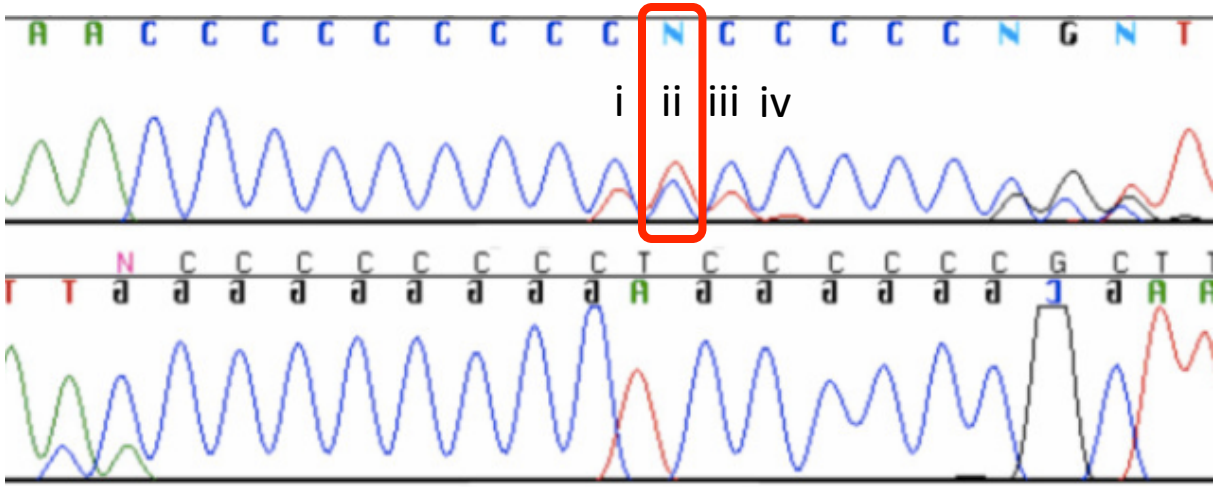


“dominant type/major molecule”



LHP example

forward strand



reverse strand

EMPOP LHP interpretation guide

EMPOP mtDNA database

EMPOP guidelines for standardized analysis and interpretation of length and point heteroplasmic positions in the human mtDNA control region

Version date: 10.03.2017

Suggestion for an EDNAP LHP exercise

Compare results of LHP between technologies and laboratories

- Sanger sequencing
- Illumina/Ion Torrent MPS

Labs to use their amplification/sequencing protocols

Apply individual analysis tools and compare to custom software (NFI offered LDS tools)

Analyse data in Innsbruck

Select samples that display LHP and can be shared between laboratories

- e.g. Control DNA, GEDNAP, …

confirming participation

Europe

Coimbra - FDUC
Cologne - UoC
Copenhagen - Forensic Medicine
Freiburg - Legal Medicine
Innsbruck - GMI
Linkoping - NFC
London - Kings College
Lyon - INPS
Madrid - IUICP
Madrid - NITFS
Oslo - UiO
Rome - Catholic University
Santiago - USC
The Hague - NFI
Wiesbaden - BKA
Zürich - Forensic Medicine

International

Auckland, NZ - ESR
Dover, USA - AFDIL
Fort Worth, USA - UNT
Gaithersburg, USA - NIST
Orlando, USA - NCFS
Singapore, SG - HSA
State College - PennState U