EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

Zürich, Switzerland

19 November 2014

Host: Cordula Haas.
Chairman: Niels Morling.

A list of participants is attached.

Welcome
Head of the Institute of Forensic Medicine in Zürich, Prof. Michael Thali, welcomed members.

Updates from other groups

ENFSI guideline for the formulation of evaluative reports in forensic science – draft (attached) — Christophe Champod, Franco Taroni

Christophe Champod briefly mentioned the history and the purpose of the project. The second phase that included commenting/consultation from scientist has ended. The core group will meet and work on version 3 of the document the following days. The document is expected to be published in the middle of 2015.
In September 2014, some EDNAP members commented (attached).
Peter Gill commented on the possible difficulties caused by acceptance of subjective evidence and reporting on activity level. It was mentioned that, in some situations, interpretative reporting may be a necessary supplement to evaluative reporting that is performed in the great majority of the cases.
Christophe Champod recommended to use the values of the LRs and not the verbal translation of LRs that have been criticized by members of EDNAP.

Forensic Science Regulator, UK: Cognitive bias effects relevant to forensic science examinations — Niels Morling
Niels Morling briefly presented the draft (attached).

A probabilistic assessment of secondary transfer at the crime scene — Elida Fonnelop
Elida Fonnelop presented results on secondary transfer of DNA (attached).

LRmix, further developtments — Peter Gill
LRmix Studio will most likely be released on 15 December 2014 by the NFI.
A new software piece based on a continuous gamma model (R. Cowell et al.) has been implemented.
New training activities including LRmix: CEPOL in Madrid, Euroforgen Train the trainers course in April 2015 in Copenhagen, ISFG Precongress Workshop in September 2015 in Krakow.

Update on exercises

mRNA exercise no 6 and 7 Cordula Haas

A manuscript of the last exercise is under review. Cordula Haas will respond to the comments from the reviewers.

Cordula Haas reported on quantification of mRNA (attached). Manfred Kayser is interested in collaboration; however, the quantification method is not ready. Experiments performed in the laboratories in Zürich and Orlando showed that the correlation between concentrations of human RNA and body fluid specific expression of mRNA seems to be limited. It was decided to explore the possibility of performing a collaborative EDNAP exercise in order to explore if quantification of RNA is improving the mRNA results compared to those obtained without quantification.

The IrisPlex exercise on genetic prediction of eye colour Niels Morling

The manuscript was published in FSI Genetics.

EDNAP ancestry informative marker exercise Chris Phillips

Chris Phillips reported that two binary AIM sets of 34 SNPs and 46 Indels were circulated together with five controls and artificial 3:1 mixtures. The DNA reference 9947A and six DNA samples were supplied. A total of 19 labs returned results concerning mixtures and ancestry assignments using Snipper. Chris Phillips is preparing a manuscript.

Updates from other groups (continued)

EMPOP Walther Parson

Walther Parson reported on the development on mtDNA typing and the EMPOP database (attached). Seven mtDNA articles were published during the last year, including a population genetic study from Macedonia, updated ISFG guidelines on the analysis of mtDNA and studies on full mitochondrial genome analyses using conventional Sanger technology and massively parallel sequencing. A critical assessment of the sensitivity of mtDNA heteroplasmy detection was presented. WP reported on the DNA in Forensics 2014 meeting in Brussels and EMPOP training in Quito, Ecuador, organized by the ISFG-GHEP sub-working group.

Nomenclature of STR sequences Walther Parson

Walther Parson gave an update on the discussion of the nomenclature of STR sequences that are being produced in large numbers using massively parallel sequencing (attached). The ISFG is not yet ready to formulate recommendations on the issue. EDNAP will analyse the challenges and, if possible, formulate considerations that, hopefully, will help to guide the forensic genetic community into a direction with one or more reasonable nomenclature(s).

High quality DNA sequence database Walther Parson

Walther Parson proposed to update STR.base to accommodate tools for quality control on STR data in the framework of the reviewing process in FSI Genetics.

Interpol Richard Scheithaur

Richard Scheithaur gave a short summary of the DNA activities of Interpol, including implementation of Interpol's missing person database.

ENFSI Roman Hradil

General update

Roman Rhadil gave a short update on the activities of the DNA Working Group of ENFSI (attached).

Guidance on the conduct of proficiency tests and collaborative exercises within ENFSI Roman Hradil
The guidance is attached.

Euroforgen-NoE Peter Schneider
Peter Schneider gave an update concerning the project (attached).

EUROFORGEN - NGS and SNP analysis assessments Chris Phillips
Chris Phillips gave an update (attached).

EDNAP web site update (www.isfg.org/EDNAP) Peter Schneider
Members are encouraged to visit the website. Suggestions are welcome.

Future activities
A SNaPshot targeting common mtDNA mutations Arnoud Kal
Arnoud Kal suggested a collaborative EDNAP exercise concerning typing of a limited number of SNPs in mtDNA with the SNaPshot method in 2015. The members agreed to do the exercise. Titia Sitien and Arnoud Kal will send out invitations with e-mail.

Next meeting Niels Morling
The next EDNAP meeting will be held on 28 April 2015 in Copenhagen in conjunction with the ENFSI meeting.

Any other business Niels Morling
There was no other business.

Closing of the meeting
The meeting closed with sincere thanks to Cordula Haas and colleagues at the laboratory in Zürich.

Ammendment
Attached please find a document from the UK Forensic Science Regulator on DNA contamination detection - The management and use of staff elimination DNA databases.

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

- List of participants
- Presentations
  - ENFSI guideline for the formulation of evaluative reports in forensic science – draft
  - Comments to the draft from some EDNAP members
  - FSR, UK: Cognitive bias effects relevant to forensic science examinations – draft
  - Elida Fonnelop: A probabilistic assessment of secondary transfer at the crime scene
  - Cordula Haas: mRNA exercise
  - Chris Phillips: Ancestry marker exercise
  - Walther Parson: EMPOP report
  - Walther Parson: STR sequence nomenclature
  - Guidance on the conduct of proficiency tests and collaborative exercises within ENFSI
  - Roman Hradil: Three reports and plans from ENFSI
  - Peter Schneider: EUROFORGEN-NoE report
  - Titien/Kal: A SNaPshot targeting common mtDNA mutations

- Jack Ballantyne: Update from Orlando
- Chris Phillips: EUROFORGEN - NGS and SNP analysis assessments.

Dr. Ricky Ansell
National Laboratory of Forensic Science
S-58194 Linköping
Sweden
Tel: +46 1056 28119
Fax: +46 13 14 57 15
E-mail: ricky.ansell@skl.polisen.se

Dr. Natascha Arora
Institut für Rechtsmedizin Zurich
Winterthurerstr. 190
CH-8057 Zurich
Switzerland
Tel: +41 44 63 56 070
Fax:
E-mail: natasha.arora@irm.uzh.ch

Dr. Jack Ballantyne
Associate Director for Research
Department of Chemistry
National Center for Forensic Science
University of Central Florida
P.O.Box 162367
4000 Central Boulevard, Bldg#5
FL 32816-2366 Orlando
USA
Tel: +1 407 823 4440
Fax: +1 407 823 4042
E-mail: jack.ballantyne@ucf.edu

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, Dr.med. Walter Bär
Institut für Rechtsmedizin
Universität Zürich-Irchel
Winterthurerstrasse 190
CH-8057 Zürich
Switzerland
Tel: +41 1 635 5611
Fax: +41 1 635 6815
E-mail: baer@irm.uzh.ch

Dr. Christopher Champod
Ecole des sciences criminelles
Universite de Lausanne
Batochime
CH-1015 Lausanne-Doigny
Switzerland
Tel:
Fax:
E-mail: christophe.champod@unil.ch

Dr. Denise Syndercombe 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. Elida Fonnelod
Department of Forensic Biology
Norwegian Institute of Public Health
University of Oslo
P.O.Box 4404
Nydalen
N-0030 Oslo
Norway
Tel: +47 99 55 95 11
Fax:
E-mail: aneelida@gmail.com

Dr. Katherine Gettings
Biotechnology Division
National Institute of Standards and Technology
100 Bureau Drive, Mail Stop 8311
MD-20899-8314 Gaithersburg
USA

Tel: +01 301 975 6401
Fax:
E-mail: katherine.gettings@nist.gov

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. Erin Hanson
Department of Chemistry
National Center for Forensic Science
University of Central Florida
FL 32816-2366 Orlando
USA
Tel: +01 407 823 0647
Fax:
E-mail: erin.hanson@ucf.edu

Dr. Tom Heylen
Institut National de Criminalistique
98-100 Chaussée de Vilvorde
B-1120 Bruxelles
Belgium
Tel: +32 2243 4614
Fax: +32 2240 0501
E-mail: tom.heylen@just.fgov.be

Dr. Roman Hradil
Institute of Criminalistics Prague
P.O.Box KUP/62
Strojnicka 27
170 89 Prague 7
Czech Republic
Tel:
Fax:
E-mail: rhradil@gmail.com

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. Helen Johannessen
Department of Forensic Biology
Norwegian Institute of Public Health
University of Oslo
P.O.Box 4404
Nydalen
N-0030 Oslo
Norway
Tel: +47 2107 7610
Fax:
E-mail: hejo@fhi.no

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. Adi Kratzer
Institut für Rechtsmedizin
Universität Zürich-Irchel
Winterthurerstrasse 190
CH-8057 Zürich
Switzerland
Tel: +41 1 635 5647
Fax: +41 1 635 6852
E-mail: adelgunde.kratzer@irm.unizh.ch

Dr. Helle Smidt Mogensen
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 6212
Fax: +45 3532 6270
E-mail: helle.smidt@sund.ku.dk

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
Innsbruck Medical University
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. Elizabet Petkowski
Service Identification Génétique
Laboratoire National de Santé
42, rue du Laboratoire
L-1911 Luxembourg
Luxembourg
Tel: + 352 28 100-671
Fax:
E-mail: elizabet.petkovski@lns.etat.lu

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

Dr. Maria João Anjos Porto
Forensic Genetic Service
Instituto de Medicina Legal
University of Coimbra
Largo da Sé Nova
P-3000-213 Coimbra
Portugal
Tel: +351 239 854230
Fax: +351 239 826132
E-mail: mariajoao.porto@dcinml.mj.pt

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. Franco Taroni
Ecole des sciences criminelles
Universite de Lausanne
Batochime
CH-1015 Lausanne-Doigny
Switzerland
Tel: +41 21 692 46 08
Fax: +41 76 399 51 02
E-mail: franco.taroni@unil.ch

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

Notes on ENFSI guidelines for formulation of evaluative reports

from

Professor Angel Carracedo, Santiago de Compostela, Spain

Professor Peter Gill, Oslo, Norway

Professor Niels Morling, Copenhagen, Denmark

Professor Peter Schneider, Cologne, Germany

Forwarded 15 September 2014 on behalf of the group by Niels Morling

To the editing group

All contributors of the comments below support the majority of the statements in the draft of the document. However, we all have some comments and concerns that are mentioned below.

We hope to have the opportunity to discuss the draft at the next meeting of EDNAP 19 November 2014 in Zürich.

Best wishes,

Niels Morling, MD DMSc
Professor of Forensic Genetics
Director and chairman of the Department of Forensic Medicine
Faculty of Health and Medical Sciences, University of Copenhagen
11 Frederik V's Vej, DK-2100 Copenhagen, Denmark
E-mail: niels.morling@sund.ku.dk

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COMMENTS

Niels Morling, MD, DMSc, Director, Professor of Forensic Genetics, Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Denmark

1. We have concerns about the limited guidance concerning how to address the risk of errors throughout the document. An example could be the discrepancy between a DNA profile contamination rate of e.g. 1:10.000 and a reported LR of 10E+18 as positive weight of the evidence.

2. We have concerns about the concept of translating a likelihood ratio (LR) into a verbal expression for the following reasons:

1) If only the verbal expression is reported, it is not possible to evaluate the evidence in an objective manner. How is “moderately strong support” combined in a mathematically correct way with other pieces of information in the case?
2) Numbers are exact and are understood the same way by most people. Verbal expressions are not necessarily understood in the same way by various people. We are all happy when we ask a shop keeper for six bottles of beer and he gives us six beers and asks us to pay 4 Euros. Imagine a situation where we ask the shop keeper to sell us a “small amount” of beer, he gives us somewhere between 1 and 10 bottles and finally asks us to pay “a moderate amount” of money - will that be 4, 6, 10 Euro? No one would accept such a situation.
3) A discrete verbal scale of conclusion as described may mislead the court to think that a likelihood ratio of 9,999 is significantly different from a likelihood ratio of 10,001. In addition the court could get the impression that it makes no difference if the likelihood ratio is 10,001 or 999,999.

Therefore, we propose that – if a likelihood ratio can be calculated, it shall be reported. Verbal translations shall be avoided if possible. If a likelihood ratio has been calculated, the verbal equivalent shall not be reported without also reporting the value of the LR. If a verbal, equivalent statement is given because a likelihood ratio cannot be calculated, the premises on which the verbal statement is based shall be clearly stated.

3. We find the guidance very much influenced by the situation in the United Kingdom. Large parts of the guidance are not relevant for Denmark – and it would cause difficulties if we followed the proposed guidelines. Some kind of a disclaimer is needed.

Angel Carracedo, Institute of Legal Medicine, University of Santiago de Compostela, Spain

I agree with most of the aspects of the document but I strongly disagree with the idea of translating likelihood ratios into verbal predicates.

In addition to the arguments given by Copenhagen:

1. A verbal predicate is not based in any scientific evidence. Why a LR of 999 provides moderately strong support and one of 1001 strong support? We are degrading scientific evidence to a mere opinion of an expert or in this case of a group of experts. Some of the opinions, especially those dealing with low and very high LR ones can be easily misunderstood.
2. The role of the judge or the court and the expert are not clearly defined if we assess in this way the value of the evidence. This is the classical paternalist position of the expert thinking that the judge cannot understand a LR so it is better to help him with an opinion (without being asked to give it) and therefore assuming the role of a judge.
3. When many years ago when Hummel's verbal predicates were introduced to explain the probability obtained in paternity cases, it had a disastrous effect in many countries. In Spain for instance, even nowadays, a probability of more than 99.7% with a priori value of 0.5 is considered "proven" by a sentence of the supreme court. The only question that judges and lawyers ask in trials is if we have reached that value (without having the minor idea about what a priori value of 0.5 means.
4. The introduction of verbal equivalents is clearly against education of judges on the understanding

of LR and how to combine it with other type of evidence. It this approved and generalized, how the LR is calculated and specially communicated will have little interest, the only important conclusion being the verbal predicate. I think it would be difficult to avoid “transposed conditionals” when presenting the evidence if we report the value of the evidence using verbal statements.

Peter Schneider, Institute of Legal Medicine, University of Cologne, Germany

I support the statements made in this document.

Some additional comments:

General comment: The current reporting guidelines are influenced by the way evidence is being reported in an adversarial system such as in the UK. In the introduction, a statement should be included reflecting the different roles for experts or scientists in adversarial vs. inquisitorial justice systems.

Section 3.14, lines 184-5): "The choice of the reported verbal equivalent is based on the likelihood ratio and not the reverse." It is my impression that exactly this has happened in the example in the annex, DNA case 2, where the statement "provides strong support" is translated back into " By strong support, I mean that the finding is over a 1000 times more likely …"
… and, in the GSR case, it is stated " The strength of the findings is qualified as strong and by this term, we consider that it is 100 to 500 times more probable to obtain these findings if proposition 1 is true than the alternative." This also appears to be a back translation, although an incorrect one, as "strong" is correlated with a LR of 1000 to 10,000 according to section 3.14.

Section 3.14, lines 465-6): "Therefore, it is incorrect to use different scales for different types of evidence". I tend to agree but find this hard to achieve. The extent of statistical data for calculating DNA genotype probabilities are several orders of magnitude larger than those for glass and other types of forensic evidence. Population data can hardly be numerically compared with collections of commercial products such as glass panes.

Also, given the current sizes of national DNA databases, a LR of e.g. 1,000,000 for observing a genotype based on a partial profile could be poor evidence if the suspect was identified following a database search with a size of 2 million, as one would expect two random matches in a population of this size.

Peter Gill, University of Oslo, Norway

1. **Areas of agreement :**
    a. Use of the ‘framework of propositions’ is a useful way to think about evidence
    b. Section 2.3 states: “Evaluation of forensic science findings in court uses probability as a measure” – yes this is really the only way to numerically evaluate data – but my main concern is that there is conflict with this recommendation in relation to the case-examples that are described (Note I have only considered the biology cases)

c. Section 3.1 recommends agreement by discussion with the prosecution and defence teams in order to evaluate propositions. And section 3.3 recommends pre-assessment to assign probabilities.
d. Various caveats are listed in sections 3.4-3.10
e. The framework in section 3 appears to be reasonable
f. Section 4 has the aim "The reporting of the value of scientific findings shall conform to four requirements: Balance, Logic, Robustness and Transparency. These requirements are met by following the principles of forensic evaluation."
g. Section 4 is reasonable

2. Areas for discussion (please note I only evaluated biological examples) :

a. The examples provided conflict with the advice given namely the four requirements of "balance, logic, robustness and transparency
b. The dependency fallacy is committed: the use of a 'presumptive test' to assign a DNA profile to a body fluid (blood). This leads (inadvertently) to the 'prosecutor's fallacy'. In the example, the presence/absence of blood is conditioned on the evidence of the presumptive test. The correct question to ask is: "what is the probability of the evidence <u>if</u> the body fluid is blood?" This is a separate question to "what is the probability of the evidence <u>if</u> the DNA originates from Mr X".
c. As the body fluid test and the DNA test are entirely separate – the statistic of the DNA test cannot be transposed to the 'uncertain' observation of a body fluid, unless the probabilities of false negatives/positives are taken into account.
d. In the drugs wrap example, the propositions are formulated in a prosecution biased way that would inadvertently direct a court down a narrow path. The possibilities of contamination or secondary transfer are not properly considered in the prosecution and defence hypotheses.
e. The drugs wrap example is a good illustration of the point highlighted on line 673: namely the LR is strictly conditioned upon the propositions, "neither may be true". The problem is that the reports are presented in way that gives the court little opportunity to consider different alternative sets of propositions. The court is unlikely to appreciate this and needs much more guidance. The questions need to be related to data analysis, otherwise the 'evaluation' in a report is no better than 'speculation' (which we should avoid). This is illustrated by the miscarriage of justice in Adam Scott.

3. Background to the interpretation of DNA profiling evidence

Forensic Genetics has solid scientific foundation that is based on genetic theory. A large amount of research has been directed towards ensuring that strength of evidence calculation is based upon data-sets that are comprised of local populations. The ENFSI DNA working group has expended much effort to collate European databases. The assumption of '*independence'* between genetic markers is not implicit. In addition, the community has taken great effort to establish whether there is any *'dependence'* between linked markers. Therefore there is strong precedent to carry out strictly objective statistical analysis using datasets within the DNA field.

We argue that the same objectivity is required if DNA profiling statistics are used to infer 'source', or 'activity' within the agreed framework of propositions. This means that any conclusions made must be cross referenced to a coherent set of data. Although the recommendations appear to implicitly support this, the difficulty is that the examples do not

themselves refer to coherent datasets. Although there is mention of subjective inference, this is no different from 'speculation', and this is to be avoided in an objective framework.

There would be no need to rely upon subjective or speculative inference if the relevant datasets were available Unfortunately, there is surprisingly little research that can be used to make these inferences. This does not mean that the scientist is then given license to use speculation in the absence of datasets based on vague notions of 'expert opinion'. On the contrary, if there is no coherent set of data available, then no inferences can be made.

4. The association fallacy

a. The *dependency* assumption that a DNA profile has come from a body fluid is not implicit – this is problematic with 'trace-evidence' ie the body fluid and the DNA is not automatically associated (Guidance note 2). However, *it is implicit* that the statistical evidence of a DNA profile is a property of 'DNA' and not the body fluid from which it is purported to have originated. Therefore it is also implicit that all DNA profiling evidence statistics must originate at the *sub-source* level – the statistic is not a direct property of 'blood' or 'semen' *per se* as a separate test is needed to identify protein markers or RNA.

b. In order to 'associate' a DNA profile with a 'body fluid' or other tissue, then the assumption of *dependency* is required (this is the opposite of the *independence assumption*). This will lead to a serious statistical fallacy (*the association fallacy*) hence caution is required when attempting to apply strength of evidence from *sub-source* to *source* level – especially where traces are low-level. On line 257, it is stated *"a presumptive test and appearance allows the scientist to establish the 'stain is blood'…"* however neither test is definitive and there is always some uncertainty about the association, hence the statistic that is applied to the fact of the DNA profile cannot be directly applied to the body fluid 'source' - the statistics applied at the *sub-source* cannot simply be transposed to the *source* level.

c. Therefore we need to refer to bodies of data that clarify the *positive* and *negative* errors that occur when *presumptive* body fluid tests are applied. Remarkably, there has been no detailed study carried out (unless I am missing something). This has significant impact on the proposals to infer *source* level propositions.

   i. If we don't know error rates, then this does not mean that they can be ignored.

   ii. The possibility needs to be considered that the body fluid detected and the DNA profile have different origins. This will happen if there is admixture of two body fluids and one of these fails to give a DNA profile (insufficient quantity or degraded). See: Peel, C., and P. Gill. "Attribution of DNA profiles to body fluid stains. "*International Congress Series*. Vol. 1261. Elsevier, 2004.

5. Guidance note 2:

In general, it is problematic for the forensic scientist to report at the 'activity level' in the way that is proposed in the document. The advice provided in lines 332-346 appears to be on the right lines (it isn't particularly well written though and suggest a rewrite) – the crucial point here is that the authors explain an example:

a. there is a considerable amount of DNA recovered from the hands of the suspect

b. Digital penetration is alleged
c. Legitimate social contact is alleged
d. The authors correctly assert that source level propositions are inappropriate
e. For some reason the authors do not recommend the obvious course of action – which is to test for vaginal cells.
f. However, putting (point *e*) to one side, the authors accept that the "potentially large likelihood ratio could be misinterpreted" – which is my point made above in 3.
g. The introductory sections go to lengths to explain that the likelihood ratio framework informed by probabilities is optimal, and to inform the probabilities *data* are required.
h. However, this recommendation *contradicts* the advice provided in the *casework examples* that I examine below.

6. Guidance note 3:

a. There is some emphasis on the requirement to base findings on data in line 401. There needs to be a note that '*unpublished data shall be made available for scrutiny*'. There is a good recommendation (lines 422-4) that if a likelihood ratio cannot be assigned by the forensic scientist (due to a lack of knowledge for example), then no appropriate evaluative assessment of the findings can be made.
b. However, there is a contrary (controversial) recommendation in line 430 that recommends *'subjective probabilities'* using expert knowledge can be used. But if there are no data then there is no basis for an 'expert opinion' as the inference is clearly outside the experts experience and we get into the realms of speculation. The example given in line 433 is not appropriate since it refers to a *'relevant population'* (i.e. data will be available) hence there is no subjectivity involved with this example. Conversely, subjective probabilities are used without reference to datasets in the set of example cases examples (pages 1-6 at the end).

7. Analysis of the example cases (note I have only evaluated the DNA statements)

a. In the first case the association is made between the DNA profile and 'blood'. As previously stated above, this association is not implicit (two separate tests that are *not dependant*) and a note needs to be made to this effect
b. In the evaluation of the evidence, the propositions are best put forward:
  i. The DNA at the point of entry…
  ii. The DNA at the point of entry….
c. I am not sure why it is necessary to state the expectation of a full DNA profile is somehow associated with the body fluid 'blood'. This provides no proof of such a body fluid. I also expect to find a full DNA profile if John Smith touches the area of entry.
d. The propositions that provide 1 in 1 billion evidence should default to DNA sub-source, not blood source. Keep the fact of the DNA and the assessment of the blood evidence separate in the evaluation, unless there are data to inform proof of association with no uncertainty. See additional note 2.
e. Not all the explanations are included here – for example the possibility of contamination and secondary transfer are ignored – there should be a caveat that covers the possibilities

8. **Proposed changes to the first example (highlighted in red):**

Issue

The issue in this case is whether the DNA at the scene came from John Smith or another person. To help address this issue, the DNA profile generated from the point of entry will be compared with the profile of John Smith.

A concurrent test indicated that the area from which the DNA profile was obtained may be blood. However, the test used is subject to positive and negative errors. The DNA and the body fluid may originate from different people.

(Section 3.14 and Guidance Note 1) – The issues are here at Sub-source level because the DNA cannot be implicitly associated with the presumptive blood stain (Guidance Note 2).

Examination and Results

The DNA profile from the ~~blood staining at~~ the point of entry matched John Smith. I estimate the chance that a person unrelated to John Smith would have this profile is less than one in a thousand million.

Evaluation

To evaluate my finding I have addressed the following propositions:

(i) The DNA at the point of entry came from John Smith

(ii) The DNA at the point of entry came from an unknown person who is not a relative of John Smith

9. **Analysis of the second case (heroin bag)**
    - **a.** Here a mixed profile is obtained. Mr J is implicated and a number of propositions are listed. The profile is clearly within the low-template definition; hence the sub-source propositions are not disputed.
    - **b.** The list is incomplete. We have:
        i. Mr J handled the bag of heroin
        ii. Someone else handled the bag of heroin and Mr J's DNA transferred via Officer P

(note according to the authors in line 620, this is an *investigative set of propositions since they are not a coherent pair of competing hypotheses – i.e. should be mirror images. On line 13 the document states "It does not cover the requirements for intelligence, investigative or technical reporting therefore there is a contradiction here.)*

    - **c.** We also need in the list of alternative propositions:

i. An identified person transferred Mr J's DNA profile (it does not have to be a particular officer and the transfer could have occurred before the crime had been detected).

ii. A laboratory contamination event occurred – the lab should be able to provide details of negative controls and laboratory contamination records – but it cannot act as proxy for the possibility of contamination outside the laboratory.

d. The evaluation of the evidence listed in the document follows a narrow point of view since it specifically considers Officer P as the route. It demonstrates 'cognitive bias' where the *defence propositions* are weighted towards supporting the *prosecution hypothesis*. Latex gloves are excellent means of transferring DNA by secondary transfer. If officer P failed to change gloves after handling various items of evidence then there is a firm *expectation* of secondary transfer. Hence a fair statement that is not prosecution biased would be*:*

e. *"If Officer P had contact with Mr J or another item handled by him, then I have an expectation of observing Mr J's DNA profile on the drug bag – furthermore I do not expect to observe Officer P's profile since his transfer of his DNA is prevented by the latex barrier" (the authors missed this crucial point)*

f. The conclusion is not supported by any data, i.e. this is firmly at odds with previous recommendations in the document. It is not clear why a 'finding of over 1000 times more likely if Mr J handled the bag rather than someone else' is given here – the figure seems to be plucked out of thin air and arguably prosecution biased. Also it is based on a selected set of propositions that are not fully inclusive of all reasonable possible explanations of the evidence (i.e. the propositions are clearly *investigative* within the authors own working definitions).

g. I argue that the interpretation of this case should be at sub-source level and that *all reasonable possibilities* should be exposed to the court, rather than the specific selection of rather prosecution orientated explanations – scientists must think much more from the defence perspective.

h. The forensic scientist usually has no knowledge of what happened at the crime scene investigation and cannot exclude alternative possibilities of DNA transfer. This means that 'proxy' propositions that depend upon assumptions about officer P are not valid unless the scientist was actually present at the crime scene investigation and observed officer P. How can the scientist possibly know if officer P failed to change his gloves before handling evidence – this is a matter for officer P, not the scientist who was absent from the crime scene (note that as a matter of course I would suggest that the discarded gloves themselves are made available for testing)

10 Additional Notes

1) See Gill, Peter. "Application of low copy number DNA profiling." *Croatian Medical Journal* 42.3 (2001): 229-232 for an explanation of limitations of reporting evidence of low-template profiles
2) Note that the statement format proposed in the "case examples" have been used in at least one miscarriage of justice, For example in the case of Adam Scott, the reporting scientist made several errors including:

a. Wrongly assigning a body fluid to source (DNA from saliva cells was attributed to sperm)
b. Wrongly formulating the expectations
c. Note the 'activity' level is so close to the ultimate issue of guilt/innocence that this is itself problematic, since the forensic scientist encroaches upon the role of the court

10.1 The report in miscarriage of justice of Adam Scott (here follows the actual statement that was shown to be based on false logic)

> *Interpretation and conclusions*: The DNA detected in the sample recovered from (victim's name) vulval swab (GE2b) can be accounted for by a mixture of DNA from (victim's boyfriend) and Adam Scott. In my opinion these _findings are what I would expect if Adam Scott had some form of sexual activity with (victim's name).
>
> In order to assess the overall findings in this case I have therefore considered the following propositions:
> - Adam Scott had vaginal intercourse with (victim's name),
> - Adam Scott has never been to Manchester and does not know (victim's name).
>
> In my opinion, the scientific findings in relation to (victim's name) vulval swab *provide strong scientific support for the view that Adam Scott had sexual intercourse with (victim's name) rather than he did not.* However, given the position of the semen matching Adam Scott and an absence of semen on (victim's name) internal swabs, the findings do not specifically support vaginal penetration with ejaculation inside the vagina. They may also support vaginal-penile contact with external ejaculation or vaginal intercourse with no internal ejaculation.'
> *End of statement*

Note the italicised part of the statement above: "*provide strong scientific support for the view that Adam Scott had sexual intercourse with (victim's name) rather than he did not."*

An example where the strength of evidence at activity level is described as "strong" but this statement was not based on any data. To reiterate, the defendant was found to be innocent of the offence. The problem was that the alternative propositions that were chosen would effectively drive the jury down a narrow (speculative) path that has the superficial appearance of scientific rigor. To construct guidelines, it is an essential test to compare against the Adams case in order to prevent similar errors.

APPROVED BY THE MEMBERSHIP JULY 2014

ENFSI ACTION PLAN

PERIOD: 2014-2015	REF: BRD-FWK-009	ISSUE NO: 1	DATE: 12 JULY 2014

Preamble

The ENFSI Action Plan is regulated by the guidance document – FRAMEWORK FOR PLANNING & REPORTING – last amended 29-05-2013. Based on this framework, the ENFSI action plan

- shall present actions of the Board together with the Standing Committees covering the forthcoming year of the P&R-cycle;
- shall be based on the current Strategic Plan, affected by the topical developments and formulated as concrete targets;
- shall be sent to the Membership no later than six weeks after the Annual Meeting.

Introduction

The ENFSI Action Plan 2014-2015 is based on the **ENFSI Strategic Plan 2014-2017** as approved by the membership at the Annual Meeting 2014 in Bratislava containing the following strategic objectives:

1. Strengthening the empirical scientific basis of forensic science by
    a. developing Pan European Databases based on shared data models;
    b. conducting statistical/scientific research from these databases to solidify the scientific basis of forensic science.
2. Creating funding opportunities for the forensic community by
    a. providing information on funding topics and opportunities and creating funding possibilities through joint lobbying;
    b. building consortia and providing grant application support.
3. Formulating and improving forensic quality standards by
    a. creating standards for interpretation of scientific evidence;
    b. developing complete process standards.
4. Improving Forensic Governance by
    a. sharing best practices and experience on forensic management, service delivery, and stakeholder relations;
    b. facilitating education and training of managers of forensic service providers.

The ENFSI strategy and the derived actions are focused on those matters that are impossible or difficult to achieve by individual members themselves.

In the framework of the ENFSI strategy the European Council Conclusions on the Vision for European Forensic Science 2020 play an important role. This vision includes the creation of a **European Forensic Science Area** (EFSA2020) and the development of a forensic science infrastructure in Europe, accentuating the following 10 areas:

1. accreditation of forensic science institutes and laboratories;
2. respect for minimum competence criteria for forensic science personnel;

3. establishment of common best practice manuals and their application in daily work of forensic laboratories and institutes;
4. conduct of proficiency tests/collaborative exercises in forensic science activities at international level;
5. application of minimum quality standards for scene-of-crime investigations and evidencemanagement from crime scene to court room;
6. recognition of equivalence of law enforcement forensic activities with a view to avoiding duplication of effort through cancellation of evidence owing to technical and qualitative differences, and achieving significant reductions in the time taken to process crimes with a cross-border component;
7. identification of optimal and shared ways to create, update and use forensic databases;
8. use of advances in forensic science in the fight against terrorism, organised crime and other criminal activities;
9. forensic awareness, in particular through appropriate education and training of the law enforcement and justice community;
10. research and development projects to promote further development of the forensic science infrastructure.

The ENFSI strategy and the strategic actions derived are in line with the actions of the EFSA 2020 initiative (see Action list below)

1. ACTIONS RELATED TO ENFSI STRATEGY

I - Strengthening the empirical scientific basis of forensic science by:

- *Developing Pan European Databases based on shared data models;*
- *Conducting statistical/scientific research from these databases to solidify the scientific basis of forensic science.*

ACTION – 1.1		
Create an overview of databases currently maintained by ENFSI members and/or Expert Working Groups, incl. the criteria for availability for the ENFSI community for potential data exchange and usage.		
Remark: RDSC will create a survey on existing databases send out to the Membership and EWGs who shall provide relevant data		**EFSA:** 7
Owner: RDSC	**Deliverable:** Report	**End date:**31-03-2015

ACTION – 1.2		
Create an overview of databases that still need to be developed		
Remark: RDSC will create a survey on existing databases send out to the Membership and EWGs who shall provide relevant data		**EFSA:** 7, 10
Owner: RDSC	**Deliverable:** Development plan	**End date:** 31-03-2015

ACTION – 1.3		
Stimulate statistical Research using existing databases to achieve sound empirical data for interpreting forensic evidence		
Remark: The ENFSI Expert Working Groups should look into improving the empirical data for improving forensic evidence. Some guidance may be obtained from FORSTAT.		**EFSA:** 7, 10
Owner: RDSC	**Deliverable:** Progress Report	**End date:** 31-03-2015

ACTION – 1.4		
Manage the ENFSI Monopoly Projects related to R&D		
Remark: These are: - MP2011 – Improving Forensic Methodologies across Europe (IFMAE, HOME/2011/ISEC/MO/ENFSI/4000002384), and - MP2013 – Towards the Vision for European Forensic Science 2020 (TVEFS-2020)		**EFSA:** 7, 10
Owner: Board	**Deliverable:** Progress Report	**End date:** 31-03-2015

II - Creating funding opportunities for the forensic community by:

- *Providing information on funding topics and opportunities as well as creating funding possibilities through joint lobbying;*
- *Building consortia and providing grant application support.*

ACTION – 1.5		
Develop and maintain an active network of key persons within European or other national or international funding bodies for the purpose of information sharing and lobbying		
Remark: The action involves setting up regular meetings with potential funding organisations.		**EFSA:** 10
Owner: Board	**Deliverable:** Progress report	**End date:** 31-03-2015

ACTION – 1.6		
Establish an ENFSI brokerage platform that offers multidirectional communication on R&D related issues with respect to consortia, grants and funding possibilities.		
Remark: Main supplier of communication to the ENFSI members is the R&D Liaison Group		**EFSA:** 10
Owner: RDSC	**Deliverable:** Progress Report	**End date:** 31-03-2015

APPROVED BY THE MEMBERSHIP JULY 2014

III - Formulating and improving forensic quality standards by:

- *Creating standards for interpretation of scientific evidence;*
- *Developing complete process standards.*

ACTION – 1.7		
Management of current Monopoly Projects related to Quality and Competence		
Remark: These are: - MP2010 – Strengthening the Evaluation of Forensic Results across Europe (STEOFRAE; HOME/2010/ISEC/MO/ENFSI/4000001759) and, - MP2012 – Towards European Forensic Standardisation through Best Practice Manuals (TEFSBPM; HOME/2012/ISEC/MO/ENFSI/4000004278)		EFSA: 1, 2 ,3, 5, 6
Owner: Board	**Deliverable:** Progress Report	**End date:** 31-03-2015

ACTION – 1.8		
Dissemination of knowledge concerning all Monopoly Program activities on Quality		
Remark: Project deliverable from the Monopoly programme BPM, Interpretation, PT guidance, Validation, G19 successor & Knowledge Exam		**EFSA:** 1, 2, 3, 4, 5,6
Owner: QCC	**Deliverable:** Progress Report , OOS, workshop, conference	**End date:** 31-03-2015

ACTION – 1.9		
Participate in the CEN PC 419 and ISO PC 272 for developing European standards for the whole forensic process (from Crime Scene to Court room)		
Remark: Attend meetings as liaison and bring in ENFSI knowledge to CEN and ISO		**EFSA:** 1, 5, 6
Owner: QCC	**Deliverable:** Report	**End date:** 31-03-2015

ACTION – 1.10		
ENFSI will undertake a gap analysis of available Proficiency Tests and Collaborative Exercises against the needs of Expert Working Groups. The goal is to provide sufficient proficiency tests covering the major disciplines.		
Remark: - QCC gathers the information from EWG - Where gaps are established QCC to work with the EWG and commercial providers		**EFSA:** 1, 4
Owner: QCC	**Deliverable:** Report	**End date:** 31-03-2015

IV - Improving Forensic Governance by

- *Sharing best practices and experience on forensic management, service delivery, and stakeholder relations;*
- *Facilitating education and training of managers of forensic service providers.*

ACTION – 1.11		
Develop a plan in order to create an infrastructure for sharing best practices and experience with respect to forensic governance		
Remark: --		**EFSA:** --
Owner: Board	**Deliverable:** Development plan	**End date:** 31-12-2014

ACTION – 1.12		
Create and overview of demand with respect to Training and Education		
Remark: ENFSI will play the brokerage role between supply and demand with respect to education and training. The QCC and RDSC will do this in their respective fields.		**EFSA:** 9
Owner: QCC and RDSC	**Deliverable:** Report	**End date:** 31-03-2015

2. ACTIONS RELATED TO ENFSI's INTERNAL ORGANISATION

In order to professionalize the ENFSI internal structure and prepare it for the challenges in the nearby future a number of actions are necessary.

ACTION – 2.1		
Develop a roadmap towards the establishment of an ENFSI legal entity, taking into account the organisational integration of the ENFSI Secretariat		
Remark: --		**EFSA:** --
Owner: Board	**Deliverable:** Roadmap	**End date:** 31-10-2014

ACTION – 2.2		
Develop an internal website for ENFSI		
Remark: --		**EFSA:** --
Owner: Board	**Deliverable:** Internal website	**End date:** 31-10-2014

ANNUAL REPORT 2013

European Network of Forensic Science Institutes

ENFSI

CONTENT

ENFSI BOARD

The composition of the 17th ENFSI Board

Üllar Lanno,
Chairman (Estonia)

Tjark Tjin-A-Tsoi,
Chairman designate
(The Netherlands)

Maria Lourdes
Puigbarraca I Sol,
Member (Spain)

Hans Henrik Jensen,
Member (Denmark)

Thomas Andermann,
Member (Germany)

ENFSI SECRETARIAT

Ewa Klimuk,
ENFSI Secretary
(Poland)

Katarzyna Zwierzyk,
ENFSI Co-secretary
(Poland)

Dear Reader,

Today, we can say that the year of 2013 was successful for ENFSI and its partners all over the world. It was the time when, through several valuable events, forensic science got a lot of attention in Europe and worldwide. We can friendly call the last year the Year of Detection. The below examples will illustrate this statement.
My chairmanship started at the ENFSI Annual Meeting, held in Belgrade, in May 2013. My dream team was enlarged with two new and strong partners from The Netherlands and Germany. The work of the new Board started with improvements in communication by establishing a schedule of teleconferences to be organized between regular Board Meetings. Today, I am convinced that this step improved the quality of governance and optimised our costs.
On the 5th of July 2013, at Eurojust Headquarters located in the Hague, ENFSI met with its closest law-enforcement partners from Eurojust and Europol. All the three parties gathered together to openly discuss mutually important issues related to forensic science and focused on European Forensic Science Area 2020 (EFSA2020). The discussion was channelled towards creating a synergy through joint activities towards the European Financial Perspective 2014–2020. All the three organisations found this gathering necessary for regular activities to be continued in the forthcoming years.
Then, in September 2013, the first ever forensic governance training course was held at The Netherlands Forensic Institute (NFI) Academy in the Hague. This remarkable achievement was possible to carry out with the support of the European Commission (EC) and the contribution of enthusiastic Dutch colleagues. I am ready to declare, as one of the course participants, that ENFSI has now its own, forensic science highest level knowledge sharing platform, readily available on the top management level.
In October, the 17th International Forensic Sciences Symposium (IFSS) taking place at INTERPOL General Secretariat headquarters in Lyon was held in order to share global expertise of forensic science. The three-day (8–10 October) forum brought together more than 160 forensic scientists, investigators and researchers from 61 countries and three international organisations, in addition to the representatives from private companies. The forensic forum allowed to discuss the latest advances in the applications of forensic techniques and sharing best practices.
Two days prior to the IFSS, the International Forensic Strategic Alliance (IFSA) had its regular annual meeting. All the six global networks, constituting IFSA, together with Interpol and UNODC delegates, presented their latest developments and identified ways in which they can be applied in criminal investigations.
This ENFSI Annual Report highlights the activities that ENFSI has recently been involved in through its 64 Members and through its different task forces. Characterising all the ENFSI projects and achievements would need a space which is much bigger, than the next 24 pages.
Enjoy your reading and let us keep in touch.

Üllar Lanno
Chairman of the17th ENFSI Board

NEW PLACES NEW FACES

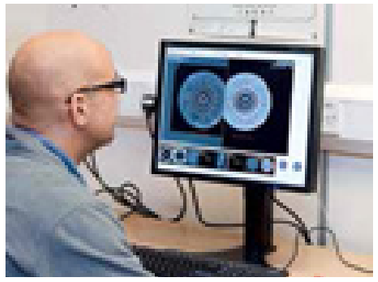
METROPOLITAN POLICE SERVICE – MESSAGE FROM MR. GARY PUGH

It is with great pleasure and pride that the Metropolitan Police Service (Scotland Yard) has become a full member of ENFSI. As a Director in the Forensic Science Service (FSS) before taking up my current post, I was the secretary of the ENSFI Crime Scene Working Group and attended many meetings with colleagues across Europe. In my current post of Director of Forensic Services in the MPS, I am responsible for all of the forensic services within the MPS including the crime scene examination and management, digital forensic science, fingerprint examination and forensic science. We continue to buy in from the UK commercial sector in DNA profiling and analytical services.

Some members of ENSFI may remember the "Met Lab" based at Lambeth. Lambeth which became the London Laboratory of the FSS and has now returned to the MPS and will be future base for the delivery of forensic science. We will undertake a £40m refurbishment of the Lambeth site in 2014–2015 and consolidate around 350 forensic science, digital, fingerprint and firearms staff (out of our 1,000 forensic staff) into the site from February 2015. This is an exciting development and we are looking forward to make fundamental changes to our delivery of digital forensic services and to establish scientific and technical partnerships with commercial organizations to take advantage of this latest advances in science and technology including the next generation DNA profiling techniques, AFIS systems, digital forensic data recovery and interpretation. We have also established a strategic alliance with King's College London, which is part of London University and has a long tradition in supporting academic teaching and research in forensic science.

Our motivation to join ENSFI is recognition of the influence and input of the increasing challenge we all face to contribute to the investigation of crime and the administration of justice across national boundaries and the common interest we have in ensuring that our citizens can have confidence and support the development of forensic science.

Gary Pugh, MPS Director



MPS Forensic Expert during his Daily Work

MPS Forensic Expert during his Daily Work

MPS Expert Presenting ENFSI Proficiency Test

BPA PG

Attendees of BPA PG Meeting held in Nantes, France on 28–30 January 2014

BLOOD PATTERN ANALYSIS PROJECT GROUP

The idea of establishing ENFSI Bloodstain Pattern Analysis Project Group (ENFSI BPA PG) was first presented by the 17th ENFSI Board at the ENFSI Annual Meeting 2013, held in Belgrade, where it was approved without any objection by the Membership.

The group discussed and agreed on its mission at the inaugural meeting held on 28–30 January 2014 in Nantes, France. The current scope of the ENFSI BPA PG covers development of a Best Practice Manual (BPM) for bloodstain pattern analysis, that is agreed upon in the ENFSI community. The BPM shall be used as a guidance when setting up standard operating procedures for blood pattern analysis in the member organizations. In addition, the group streams towards the creation of proficiency tests (PT), collaborative exercises (CE) for the BPA field of expertise, with the aim to deliver an annual test to the ENFSI Members.

The main challenge that the group has accepted is to bring together member organizations of ENFSI and other national agencies/organizations with professional interest in BPA methods for exchanging, disseminating and improving BPA knowledge.

The aims of BPA PG shall be achieved by:

- discussing, sharing and comparing BPA methods, protocols and research;
- establishing BPA quality assurance guidelines and quality controls for Europe (e.g. PT or CE);
- co-operating with other national and international organizations in developing European uniformity for BPA, including standardization of BPA terminology (in accordance with International Association of Bloodstain Pattern Analysts recommendations);
- producing an European BPM to be a reference guide, not only for bloodstain pattern analysts, but for all involved in the forensic process;
- disseminating to the European Forensic Bloodstain Pattern Analysis community, ENFSI guidelines, forensic research results, the provision of training and any other work of benefit.

COMPOSITION OF BPA PG STEERING COMMITTEE

Name	Function	Country
Philippe Esperança	Chairperson	IGNA, France
Lino Henriques	Deputy Chairperson	LPC, Portugal
Weine Drotz	Deputy Chairperson	SKL, Sweden
Jonny Irons	Secretary	FSNI, United Kingdom
Kamil Januszkiewicz	Secretary	CFLP, Poland
Mikle van der Scheer	Treasurer	NFI, The Netherlands

ENFSI FORENSIC ARCHAEOLOGY PROJECT GROUP

In December 2012, an application letter with a request to launch a Forensic Archaeology Project Group (PG) was sent to the 17th ENFSI Board. The Board agreed with the submitted proposal and the approval decision of all the ENFSI Member-representatives was taken at the Annual Meeting held in Belgrade in May 2013. The first meeting of the PG took place at The Netherlands Forensic Institute (NFI) on 29 August 2013, and was attended by representatives from three PG founding members and one associate member: the NFI, the German Crime Scene Unit of the Federal Criminal Police Office, the French Forensic Sciences Institute of the National Gendarmerie and the British Forensic Archaeology Expert Panel (IFA). Mr. Mike Groen (NFI) was appointed the Chair of the PG.

The group defined forensic archaeology as a `discipline that uses archaeological theory, methods and techniques in legal contexts and which combines archaeological, taphonomical and criminalistic knowledge to localise, document and interpret pedological, ecological and osteological patterns at a (possible) place of incident or a crime scene´. The Group itself has been established in order to understand how forensic archaeology is organized and practiced within the different European countries. Its founders wanted to meet and learn from foreign colleagues. The beginnings of the group can be traced back to 2012 when the NFI initiated a meeting on European Forensic Archaeology. This meeting, organized as a joint venture between the NFI and the IFA, was attended by approximately 50 delegates, covering 12 countries and representing ENFSI Member-institutes, police forces, NGOs, forensic science providers, universities and freelance professionals. One important outcome was the wish to establish a scientific forensic archaeological platform for European practitioners. It was felt that it would be most convenient for the forensic community and for forensic archaeologists, if this platform were not a stand-alone entity. The most logical option for such a platform was ENFSI, for the ENFSI Membership would not only underline the forensic nature of the forensic archaeological platform to be established, but would also provide a direct access to the European forensic community. The key aims of the established PG are to raise awareness of forensic archaeological possibilities at places of incident or scenes of crime within the ENFSI community and to explore the possibility of the establishment of Forensic Archaeology Working Group. The membership of the PG is not limited to forensic archaeologists only, but is open to all scientists who apply forensic archaeological theories, methods and techniques during their case work and who represent European forensic laboratories, NGOs, police forces and universities. The PG believes that such a mix of professionals is necessary to succeed in introducing and maintaining forensic archaeological theories, methods and techniques at places of incident and scenes of crime across Europe. Individuals interested in joining the PG are invited to contact the group at DI_ENFSI_PG_Forensic_Archaeology@nfi.minvenj.nl.

2nd European Meeting on Forensic Archaeology, 30–31 August, The Hague, The Netherlands

ENFSI IN CLOSE-UP

E&T STANDING COMMITTEE

In 2012, E&T SC took a number of initiatives to be carried out the following year. On one hand the Committee was keen to see results of a first survey on E&T among ENFSI members, on the other hand it wanted to improve the interaction with the ENFSI Working Groups in order to achieve better understanding of mutual expectations on shared E&T. The results of the survey were promising. Indeed, it contained information regarding E&T needs in different forensic disciplines. This input was discussed when the E&T SC met in Warsaw for the spring meeting 2013. A common agreement was made that more information will be necessary to get a better understanding on format and content of E&T modules which will have to be set up by the thematically concerned ENFSI Working Groups as well as other E&T providers. As far as the second objective is concerned – interaction with Working Groups, E&T SC planned to visit throughout the year a number of Working Group meetings for comments on the needs of E&T in ENFSI. However, the review of a first series of visits was less promising than expected. The visiting program was therefore put on hold and so were any further E&T SC activities. There was a strong feeling that it will be up to the ENFSI Board to examine the general terms of reference for the Committe and to provide guidance in order to be able to focus on the right strategic approach for E&T in ENFSI. A preliminary discussion on this subject was held during the ENFSI Joint Meeting 2013 in Barcelona. The inputs given by the Chairs of the ENFSI Working Groups made it clear that the ideas related to the added value of E&T in ENFSI are considerably varying and will need further consideration in 2014.

COMPOSITION OF E&T SC

Name	Function	Country
Peter Pfefferli	Chair	FSI, Switzerland
Jan Blok	Member	NFI, The Netherlands
Inge Buys	Member	INCC, Belgium
Gunnel Carlson	Member	SKL, Sweden
Gokhan Ersoy	Member	ATE, Turkey
Aleksandar Ivanovic	Member	FCPDM, Montenegro
Jozef Mlkvik	Member	IFS, Slovakia
Piotr Trojanowski	Member	CFLP, Poland
Hans Henrik Jensen	Board Representative	KTC, Denmark

QCC STANDING COMMITTEE

The main goal of Standing Committee of Quality and Competence (QCC SC) is to act as an advisor and coordinator to relevant ENFSI entities on matters of quality and competence in a broad sense. During 2013, QCC SC continued to work on the activities regarding continuous improvement of the quality and competence among ENFSI community. The Committee worked on plans to improve validation processes, PT/CE schemes and competency issues. 2013 brought also some changes in the composition of the QCC as such. Mrs. Carolina Sanchez de la Torre and Mr. Sebastien Nicholas left their positions, and three new members came on board – Mrs. Maria Kambosos from Bundeskriminalamt in Wiesbaden, Germany, Mrs. Chanda Lowther-Harris from Metropolitan Police Service in London, UK and Mrs. Merike Rump from Estonian Forensic Science Institute in Tallin.

Among other responsibilities, QCC was strongly involved in cooperation with the most relevant organizations regarding quality and accreditation (EA, ILAC, ISO, Eurachem, NIST, CEN/CENELEC, etc.). QCC worked actively on the development of new version of ILAC G19 Guideline for forensic laboratories which, including both ISO 17025 and ISO 17020, will be more comprehensive than the previous one. New Guideline is expected to be approved and published in 2014. Furthermore, QCC was involved in drafting of EA guidance document concerning opinions and interpretations, which constitute a highly important subject in the forensic process.

As every year, in 2013, QCC organized Quality and Competence Liaison Group (QCLG) meeting, gathering the QCLG representatives from ENFSI Member-laboratories and ENFSI Working Groups. This year's QCLG meeting was kindly hosted by the Forensic Science Laboratory in Dublin, Ireland on 5–8 November. It turned out to be one of the biggest events devoted to forensic quality organized in Europe in 2013. The conference was attended by over 70 individuals. The delegates included 27 Quality Managers from European forensic organizations, Australia as well as the United States. It was a successful conference which touched upon three other subjects from ENFSI Monopoly projects: M2 executed under MP2010 – General Forensic Knowledge Exam – project aiming at the development of an online examination enabling ENFSI laboratories to demonstrate the knowledge of their forensic practitioners in the field of general forensic science; P5 executed under MP2009 – Development of the Guidance on the Conduct of Proficiency Tests and Collaborative Exercises and last but not least, P4 executed under MP2009 – The development of guidelines for the validation of analytical and comparative methods in forensic science.

During the second half of the year, QCC put a lot of efforts in the preparation of one the largest ENFSI Monopoly Programme – MP2012 Towards European Forensic Standardisation through Best Practice Manuals (TEFSBPM). The key objective of TEFSBPM will be the harmonisation of the forensic Best Practice Manuals (BPMs) across Europe involving a wide range of forensic areas. QCC prepared the opening conference of MP2012 which was held in Amsterdam on 31 January 2013. The conference marked the formal start of the MP2012 activities. The conference was attended by the project team leaders together with the QCC team, responsible for introduction of BPM Field Specific Template and creating an overview of the MP2012 project activities. Through participation in some BPM team meetings, QCC will play a significant role in supporting and guiding all the BPM teams.

In the nearest future, QCC, in accordance to the ENFSI Policy on Standards for Accreditation will continue to monitor accreditation scopes and quality improvement among ENFSI Members.

COMPOSITION OF QCC

Name	Function	Country
Christina Bertler Edlund	Chairperson	SKL, Sweden
Saša Žugaj	Vice Chair	FSC Ivan Vucetic, Croatia
Ralph Kleuskens	Member	NFI, The Netherlands
Chanda Lowther Harris	Member	MPS, UK
Maria Kambosos	Member	BKA, Germany
Merike Rump	Member	EFSI, Estonia
Wim Neuteboom	Member, MP2012 leader	NFI, The Netherlands
Birgitta Roseen Pettersson	Secretary	SKL, Sweden
Lourdes Puigbarraca i Sol	Board representative	CME, Spain

R&D STANDING COMMITTEE

The Research and Development Standing Committee (R&D SC) has continued to work along the lines of the ENFSI Strategy, thus aiming to facilitate the increase of the amount of forensically relevant R&D in Europe.

In collaboration with the ENFSI Board and other Standing Committees, the R&D SC has contributed to the development of the view on the future role in ENFSI's strategic areas.

As in the previous years, in 2013 the Committee concentrated its activities on designated priority areas which included: creation of a R&D Liaison Group, Searching for R&D funding possibilities and, the mid and long term research needs within the ENFSI community.

COMPOSITION OF R&D SC

Name	Function	Country
Marcel van der Steen	Chair	NFI, The Netherlands
Tapani Reinikainen	Member	RTL, Finland
Yves Schuliar	Member	IRCGN, France
Sean McDermott	Member	FSL, Ireland
Thomas Biermann	Member	BKA, Germany
Christophe Champod	Member	ESC, Switzerland
Jim Fraser	Member	CFS, UK
Bart Nys	Member	INCC, Belgium
Laurence Dujourdy	Member	INPS, France

R&D LIAISON GROUP

The R&D Liaison Group has grown significantly and although not all the ENFSI members were able to appoint a member for the Liaison Group, the network now has a substantial size. Currently the liaison group has 55 members,17 of which were selected from ENFSI Working Groups.

MID AND LONG TERM RESEARCH NEEDS

A clear view from the European forensic community on its future research needs is important, as this can facilitate the discussions with potential collaboration and funding partners. The R&D SC, has continued to work together with the ENFSI Board and the ENFSI Working Groups to establish this view, among others by the creation of whitepapers on mid and long term research needs in all of the forensic areas covered by ENFSI. More than 50% of all the Working Groups have created draft white papers, some of which have already proved their value in discussions with the European Union on future funding programmes.

R&D SC

During the year 2013, a number of changes took place in the R&D SC membership. Mr. Michael Pütz, Mrs. Birgitta Rasmussen and Mr. Dariusz Zuba ended their terms as SC Members.

After a series of discussions with the ENFSI Board, it was decided to increase the size of the R&D SC with one extra member and all the vacant positions were filled during the course of 2013. The new members include: Mr. Tapani Reinikainen, Mr. Yves Schuliar, Mr. Sean McDermott and Mr. Thomas Biermann. The entire Committee is ready for the coming years to help to support the ENFSI Board with the implementation of the new ENFSI strategy.

16th Annual Meeting of Fire and Explosion Investigation Working Group, 1–4 October 2013, Wiesbaden, Germany

ENFSI EXPERT WORKING GROUPS – THE STRONGEST WORKFORCE OF ENFSI NETWORK

From fields such as Animal, Plant, and Soil Traces, Digital Imaging and DNA to the examination of documents, drugs, explosives, fingerprints, firearms/gunshot residues, fire and explosions, information technology, speech and audio, handwriting, marks, paint and glass, road accidents, crime scene or textile and hair – the diversity of scientific fields professionally covered by the 17 ENFSI Expert Working Groups is indeed exceptional. Undoubtedly, the Expert Working Groups constitute the backbone of the ENFSI network. In addition, ENFSI has currently established two project groups in order to verify a possible need for additional Expert Working Groups in the fields of Blood Pattern Analysis and Forensic Archaeology (see: New Places, New Faces).

The ENFSI Expert Working Groups provide an ongoing exchange of information on the established methods of examination as well as novel approaches in the respective scientific fields. They develop best practices, carry out workshops on specific topics as well as initiate and conduct joint research and development projects. Experts working for ENFSI Member-institutes across Europe and for associated institutes across the world, provide a suitable platform for an open and trustful exchange of knowledge. Co-operation built on mutual trust is facilitated by personal relationships and even friendship among the experts of forensic institutes, whose work is actively encouraged and promoted by ENFSI. Personal contacts have positive impact on the mutual assistance in everyday casework, e.g. exchange of comparison materials, sharing information on forensic providers or simply giving professional advice. Through active participation in the Annual Meetings, membership in Steering Committees, active contribution to projects related to the establishment of best practice or the design and conducting of proficiency tests and collaborative exercises, the experts generate an enormous benefit for the ENFSI community as a whole and their respective home institutes in particular. In order to maintain good relations and connections within ENFSI, it is important that Directors of ENFSI Member – laboratories support the Expert Working Groups by encouraging experts from their institutes to actively participate in the Annual Meetings of Working Group, Steering Committees and Project Groups.

ENFSI WORKING GROUPS

1 **Monika Hilgert**
GERMANY
Fingerprint

2 **Udo Zerell**
GERMANY
Drugs

3 **Stephen Maxwell**
NORTHERN IRELAND
Handwriting

4 **Niamh Nic Deaid**
UK
Fire and Explosion

5 **Jürgen Bügler**
GERMANY
Documents

6 **Urlich Simmross**
GERMANY
Paint and Glass

7 **Keith Fryer**
UK
Scene of Crime

8 **Mattijs Koeberg**
THE NETHERLANDS
Explosives

9 **Roman Hradil**
CZECH REPUBLIC
DNA

10 **Andreas Hellmann**
GERMANY
Animal, Plant and Soil

11 **Kornelia Nehse**
GERMANY
Textile and Hair

12 **Christophe Lambert**
FRANCE
Digital Imaging

13 **Florin Rusitoru**
ROMANIA
Road Accident Analysis

14 **Chris Moynehan**
UK
Firearms/GSR

15 **Andrzej Drygajło**
SWITZERLAND
Forensic Speech and Audio Analysis

16 **Zeno Geradts**
THE NETHERLANDS
Forensic Information Technology

17 **Michael Braune**
GERMANY
Marks

ENFSI SECRETARIAT

At the 24th ENFSI Annual Meeting, held at the historical castle in Dublin, Ireland, from 23 till 26 of May 2012, ENFSI Member-representatives, present at the meeting, arrived at the decision to transfer ENFSI Secretariat from the Hague, The Netherlands, to Warsaw, Poland, as of 1 January 2013.

According to the transfer plan, the new Secretariat is running based on tripartite agreement between ENFSI, Central Forensic Laboratory of the Police (CFLP) in Poland and European Forensic Initiatives Centre Foundation (EFIC). It is located at Aleja Wyzwolenia 3-5/29, 00-572 Warsaw, Poland.

The post of the Secretary is held by Ms. Ewa Klimuk (CFLP). The Polish team involves also Ms. Katarzyna Zwierzyk (Co-secretary) as well as Mrs. Beata Stefańska and Mr. Grzegorz Gutkowski, both responsible for ENFSI finances.

Apart from the regular ENFSI business, the Polish team is tasked with financial management and financial administration of ENFSI Monopoly Programmes, currently running within the network.

Taking the opportunity, ENFSI Secretariat would like to thank the entire ENFSI Community for the ongoing support and trust placed in the Polish team.

Ms. Ewa Klimuk,
ENFSI Secretary

Ms. Katarzyna Zwierzyk,
ENFSI Co-secretary

ENFSI SECRETARIAT IN POLAND STARTED ITS ACTIVITIES AS OF 1ST JANUARY 2013

ENFSI MEETINGS

ENFSI Annual Meeting 2013, Belgrade, Serbia

ENFSI ANNUAL MEETING

The 25th ENFSI Annual Meeting was held in Belgrade, Republic of Serbia, on 22–24 May 2013. The meeting was attended by 59, out of 64, ENFSI Member-representatives who gathered in Serbia in order to discuss and share their views on the issues of importance to the entire ENFSI Community. The meeting participants included also the Chairs of ENFSI SCs as well as representatives from ENFSI Working Groups.

The Belgrade Meeting was hosted by National Crime-Technical Center (NCTC) of Serbia and its Director, Mr. Lazar Nesic. It took place in the famous Hotel Moscow, located in the very heart of Belgrade city.

Following ENFSI tradition, the Annual Meeting consisted of two parts, thematic and business one.

Thematic Day started with a welcome speech by Mr. Dejan Radenković from Criminal Investigation Department in Serbia. He warmly welcomed the participants and continued his speech with mentioning current initiatives running within ENFSI, pointing to their importance. In addition, Mr. Lazar Nesic, Director of NCTC, gave Serbian gifts to the ENFSI Board Members and the Secretariat. He wished all the participants a nice stay in Belgrade and a fruitful meeting. Then, the next speaker, Mrs. Jelica Nedeljković described the results of psychological tests carried out on the employees of the NCTC with the aim to check mental state of the employees, identifying problematic workers, showing a role model of successful superior and worker as well as answering a question on how to improve working conditions for the individuals employed in the Serbian Police Forensic Centre. The thematic part was concluded with the presentation on "Consequences of disregarding contemporary forensic standards – case Racak and Ljuboten" by Mr. Aleksandar Ivanović from Forensic Center Montenegro.

The business part started shortly after the thematic one. The Member-representatives discussed the issues concerning the future of ENFSI, ENFSI Working Groups, ENFSI finances and, last but not least, the development of new internal web-based platform for ENFSI Members.

In addition to the above, during the meeting, ENFSI chose the new Chairman designate, replacing Mr. Üllar Lanno, the new Chairman of the 17th ENFSI Board, replacing Mr. Paweł Rybicki, and the new Board Member, taking place of Mr. Torsten Ahlhorn, stepping down

ENFSI Annual Meeting 2013, Belgrade, Serbia

ENFSI Annual Meeting 2013, Belgrade, Serbia

Gala Dinner, held during ENFSI Annual Meeting 2013

Thematic Part of ENFSI Annual Meeting 2013

from his function. The raised topics included also the reports on recent activities of ENFSI SCs and the Board as such. The Chairman designate, Mr. Üllar Lanno took a lead on the voting procedure. The candidates for Board Member position, Dr. Thomas Andermann and Mrs. Dominique Saint-Dizier as well as for Chairman designate, Dr. Tjark Tjin-A-Tsoi and Mr. Pavel Kolar had a chance to present themselves before the voting.

The Membership elected Dr. Thomas Andermann for the position of a new Board Member. Dr. Tjark Tjin-A-Tsoi was elected as Chairman designate by acclamation, as Mr. Pavel Kolar decided to withdraw himself due to internal obligations in his own country.

At the end of the meeting, the new Chairman of the 17th ENFSI Board Mr. Üllar Lanno presented his plans, aims and objectives for the forthcoming two-year period. He proposed the ENFSI community to focus on transparency of ENFSI finances, accreditation of ENFSI Members, increasing the effectiveness of internal communication, including the development of a web-based platform, as well as other PR activities of ENFSI. He also pointed to the importance of International Forensic Strategic Alliance (IFSA) and the significance of the EU support. The new Chairman's speech was concluded with kind words directed towards the stepping down Board members. He handed over memory gifts to the Chairman of 16th ENFSI Board Mr. Paweł Rybicki and the Board Member Mr. Torsten Ahlhorn.

Thanks to the local organizers and the social program offered by them, the Belgrade Annual Meeting was an excellent opportunity to mix business with pleasure. Definitely, due to the pleasant atmosphere, the 25th Annual Meeting will be remembered for a long time.

The next, 26th Annual Meeting will take place in Bratislava, Slovakia, on 21–24 May 2014.

THE 25TH ENFSI ANNUAL MEETING WAS HELD IN BELGRADE, REPUBLIC OF SERBIA, ON 22–24 MAY 2013.

ENFSI JOINT MEETING 2013

ENFSI Joint Meeting (JM) 2013 took place in Barcelona, Spain, and was held on 3–4 December 2013. The attendees constituted the 17th ENFSI Board, ENFSI Working Group Chairs, ENFSI Project Group Chairs, Standing Committees Leaders, ENFSI Secretariat and other invited guests. The meeting was organized by Ms. Lourdes Puigbarraca i Sol, one of the ENFSI Board members, and hosted in the state-of-the-art building of Departament d'Interior – Generalitat de Catalunya, Sabadell, Spain.

Despite the winter time, Barcelona welcomed the participants with beautiful, warm weather. The rays of the sun suffused the conference room bringing pleasant atmosphere and inspiration for fruitful discussions. Joint Meeting 2013, similarly to the previous gatherings of this kind, consisted of several thematic parts and included separate plenary sessions of the Board, Working Groups and Standing Committees representatives.

The discussed topics covered reports and plans of Working Groups, Project Groups and Standing Committees, ENFSI standard for the formulation of evaluative reports in forensic science and ENFSI Strategic Plan for 2015–2018. Furthermore, during the QA session the attendees touched upon the matters of WG finances, WG Associate Members, ENFSI website, publication of ENFSI documents, ENFSI Monopoly Programmes and Minimum Requirement Standards developed by International Forensic Strategic Alliance (IFSA).

At the very beginning, the ENFSI Chairman welcomed and introduced the new faces among the Working Group Chairs, Project Groups and other invited guests. The substantial part of the JM 2013 was devoted to the 2013 reports and 2014 plans of the ENFSI statutory bodies. Each of 17 Working Group, 2 new Project Groups and 3 Standing Committees presented their composition, structure, activities performed in 2013 and goals for the year 2014. These presentations allowed the participants to get familiar with the groups' achievements. During the QA session, ENFSI Board explained and discussed the issues of the utmost importance to WGs.

Another important subject of the meeting was the project entitled "The development and implementation of an ENFSI standard for reporting evaluative forensic evidence" executed under Monopoly Programme 2010. The leader and member of this project team, Mrs, Sheila Willis and Mr. Christophe Champod respectively, presented its aims and the progress in preparation of standard's draft, which provoked a lively discussion among the attendees of Barcelona meeting.

During the ending session, ENFSI Board handed out the Best Working Group Award. The winner was DNA Working Group, recognized for being well organized, active and progressive in the forensic domain it represents. The award was handed to Mr. Roman Hradil, the Chair of DNA WG. In addition, the ENFSI Chairman expressed his gratitude to Ms. Lourdes Puigbarraca i Sol for the efforts related to the organization of JM 2013. Thanks to the host, the participants not only spent their time on productive discussions but also had an opportunity to get to know each other better while sightseeing Barcelona's old town or Parliament of Catalonia.

Attendees of ENFSI Joint Meeting 2013

Attendees of ENFSI Joint Meeting During their Visit to the Parliament of Catalonia

ENFSI Joint Meeting 2013

17th ENFSI Board and ENFSI Secretary

BOARD MEETINGS

No.	Date	Venue
1	24–25 January 2013	Prague (Czech Republic)
2	18–19 April 2013	Copenhagen (Denmark)
3	20–21 May 2013	Belgrade (Serbia)
4	17 June 2013	Barcelona (Spain)
5	29–30 August 2013	Warsaw (Poland)
6	17–18 October 2013	Rome (Italy)
7	2 and 4 December 2013	Barcelona (Spain)

ENFSI BOARD MEETINGS

In the year of 2013, the ENFSI Board gathered seven times. The meetings were held in Prague (16th ENFSI Board), in Copenhagen (16th ENFSI Board), in Belgrade (16th ENFSI Board), in Barcelona (17th ENFSI Board), in Warsaw (17th ENFSI Board), in Rome (17th ENFSI Board) and last but not least, the last Board Meeting in 2013 was organized in Barcelona (17th ENFSI Board). The detailed dates of the respective meetings are presented in a table on the left.

In 2013, the Board's priorities covered the improvement of ENFSI internal communication, optimization of ENFSI expenditures as well as facilitating daily activities of ENFSI Working Groups. The meetings served their purposes and allowed for effective management of all the routine ENFSI business.

ENFSI Board Meeting in Warsaw, Poland

KEY PROJECTS

EMFA-2

The accreditation of all ENFSI-members in compliance with ISO17025 standard has been a top priority for ENFSI. In line with this priority, ENFSI developed a special programme to support non-accredited members in achieving accreditation called "The European Mentorship for Forensic Accreditation" (EMFA), also referred to as the 'flying mentors'. According to the programme's rules, accredited ENFSI laboratories act as mentors for non-accredited ones. In this way non-accredited laboratories (trainees) are taught in the area of building up their quality assurance systems. By making twin combinations between accredited and non-accredited laboratories, those without accreditation can achieve it much faster. The programme ends with a real, broad scale pre-audit. After this audit the mentor draws the conclusion whether the trainee is 'ready for accreditation'.

ENFSI gave financial support for the trainees as well as the mentors by covering the costs related to travel and accommodation.

Following the first EMFA programme (2007–2010), the EMFA-2 programme started in 2011 and ended,as scheduled, in November 2013.

Unfortunately, the trainee laboratory from Sarajevo, Bosnia and Herzegovina, had to leave EMFA-2 due to a number of internal problems after only one orientation visit. Thus, the programme had to be continued with 3 instead of 4 twinning pairs.

Mrs. Christina Bertler Edlund (SKL – Sweden) and Mr. Wim Neuteboom (NFI – The Netherlands) were responsible for the programme management.

Three plenary meetings were held within the framework of EMFA-2.These were the Opening Conference (Belgrade, Serbia, March 2011), the Mid Term Conference (Danilovgrad, Montenegro, March 2012) and the Closing Conference (Zagreb, Croatia, November 2013). Starting from the orientation visits of the mentors to the trainee laboratories in the summer of 2011, a series of 21 visits followed in the course of the programme. Some visits involved only Quality Assurance Managers, others included also experts in the selected fields.

EMFA-2 was scheduled to be executed from the summer of 2011 till the end of 2013 (2.5 years). Due to the fact that the ENFSI budget year runs from April 01 till March 31, the total programme budget was spread over 4 ENFSI financial years. The overall expenses have been considerably within the budget.

At the Closing Conference in Zagreb, Croatia, the mentor laboratories gave their opinion about their 'own' trainee laboratories. The opinions were all positive and the trainee laboratories were qualified as 'ready for accreditation'.

EMFA-2 TWINNING PAIRS

Trainee laboratory	Mentor laboratory
The National Crime-Technical Center of Serbia	Forensic Science Centre "Ivan Vučetić", Croatia
Forensic Center Montenegro	Estonian Forensic Science Institute
North-Western Forensic Science Center Russia	State Forensic Science Bureau, Latvia
Forensic Expertise Department, Bosnia and Herzegovina	Forensic Science Laboratory, Slovenia

OVERVIEW OF EMFA-2 BUDGET AND EXPENSES

Budget period	Budget (original project plan)	Budget (approved by ENFSI Members at Annual Meetings)	Actual expenses
2010–2011	8.800 Euro	5.000 Euro	1.690 Euro
2011–2012	35.600 Euro	25.000 Euro	14.250 Euro
2012–2013	40.500 Euro	30.000 Euro	18.386 Euro
2013–2014	41.400 Euro	Not explicitly fixed	13.396 Euro
Total	126.300 Euro	60.000 Euro (+ unfixed budget 2013–2014)	47.722 Euro

EMFA-2 Opening Conference

EMFA-2 Team

MONOPOLY PROGRAMME

The year 2013 has seen important milestones for ENFSI Monopoly Programmes. The EU monopoly grants to support forensic co-operation across Europe, are made available under the General Programme on Security & Safeguarding Liberties Specific Programme on Prevention of and Fight against Crime (ISEC) managed by the EC Directorate-General Home (DG Home). The grants are awarded to ENFSI as an organization recognized by the EU as having a monopoly status in the area of forensic science. The year 2013 was the final year of the ISEC programme funding and ENFSI submitted its last grant application to the EC in December 2013. In addition, the implementation of the very first Monopoly Programme (funded by the 2009 grant) was completed on 15th December 2013 and the final report has been delivered to the EC. Although no new ISEC grants will be announced beyond 2013, the implementation of the current ENFSI monopoly work will continue for several years to come.
The completed 2009 project "Sustainable Quality within European Forensic Science" (SQWEFS) was designed to promote the exchange and dissemination of best practice in the critical area of quality standards within European forensic science. The work has made significant contributions in several key areas:

1. Progress towards Europe-wide consistency in the process of accrediting forensic laboratories and ensuring that national accreditation bodies have appropriate knowledge and support:
   - Work with the International Laboratory Accreditation Cooperation organisation (ILAC) has produced a new guideline for the implementation of ISO-standards (17020 and 17025) throughout the whole forensic process. This document (G19) will be published in 2014.
   - Joint work with European Cooperation for Accreditation (EA) has delivered training for 31 representatives from national accreditation bodies across Europe to raise their general awareness of different forensic fields.

ENFSI MONOPOLY PROGRAMME 2010

Programme Theme: "Strengthening the Evaluation of Forensic Results across Europe (STEOFRAE)".
Current Status: Grant Agreement signed / Work commenced on 1st January 2012

M1	The development and implementation of an ENFSI standard for reporting evaluative forensic evidence.	Sheila Willis (FSL - Ireland)
M2	The development of a knowledge examination for competence assessment.	Didier Meuwly (NFI - The Netherlands)
M3	Upgrading the ENFSI STR BASE.	Ingo Bastisch (BKA - Germany)
M4	An international training seminar on the use of sub class characteristics in firearm investigations.	Ruprecht Nennstiel (BKA - Germany)
M5	Workshops on the application of the Bayesian approach in gunshot residue investigation.	Ludwig Niewoehner (BKA - Germany)
M6	Guidelines for the representative sampling of drugs for quantitative analysis.	Laurence Dujourdy (INPS - France)
M7	The evaluation of computer proficiency tests within computer forensics.	Hakan Bergstedt (SKL - Sweden)

ENFSI MONOPOLY PROGRAMME 2011

Monopoly Projects (2011) – 3 year programme (grant awarded € 646,931)
Programme Theme: "Improving Forensic Methodologies across Europe (IFMAE)".
Current Status: Grant Agreement signed / Work commenced on 1st January 2013

Z1	Dating of questioned documents by resins and binders in ballpoint ink entries.	Fritz Koehler (BKA - Germany)
Z2	The development of an internet accessible database on textile fibres.	Kornelia Nehse (LKA Berlin - Germany)
Z3	Methodological guidelines for semi-automatic and automatic speaker recognition for case assessment and interpretation.	Andrzej Drygajlo (ESC - Switzerland)
Z4	International cooperation for testing, validation and application of ink dating methods.	Juergen Buegler (BLKA Munich - Germany)
Z5	Standardization of forensic image and video enhancement (S-Five).	Patrick De Smet (INCC - Belgium)

ENFSI MONOPOLY PROGRAMME 2012

Monopoly Projects (2012) – 2 year programme (grant awarded € 537,982)
Programme Theme: "Towards European Standardisation through Best Practice Manuals (TEFSBPM)".
Current Status: Grant Agreement signed / Work commenced on 1st January 2014.

B1	Guidelines for best practice in the forensic examination of digital technology.	Gregory Webb (MPS - UK)
B2	Pan-European best practice in forensic handwriting examinations.	Jonathan Morris (SPSAFS - UK)
B3	Best practice manual for colouring methods in gunshot residue analysis.	Amalia Brouwer-Stamouli (NFI - The Netherlands)
B4	Best practice manual for road accident reconstruction examination.	Lina Lazarenko (FSCL - Lithuania)
B5	Microscopic identification and comparison of human and animal hair best practice manual.	Chris Gannicliffe (SPSAFS - UK)
B6	Best practice manual for fingerprint examination.	Slobodan Oklevski (MoI- Republic of Macedonia)
B7	Specification for DNA pattern recognition and comparison.	Ulrich Neuhaus-Steinmetz (LKA Berlin - Germany)
B8	Best practice manual for the application of molecular methods for the forensic examination of non-human biological traces.	Andreas Hellmann (BKA - Germany)
B9	Best practice manual for the forensic recovery, identification and analysis of explosives traces.	Matthew Beardah (Dstl - UK)
B10	Best practice manual for the forensic investigation of fire scenes which have resulted in fatalities.	Niamh Nic Daeid (CFS - UK)
B11	Best practice manual for the forensic investigation of fire scenes which involve the clandestine manufacture of improvised or home-made explosive devices.	Niamh Nic Daeid (CFS - UK)
B12	Best practice manual for the forensic investigation of fire scenes which involve the clandestine manufacture of illicit synthetic drugs.	Niamh Nic Daeid (CFS - UK)

- Further cooperation with EA, represented by United Kingdom Accreditation Service (UKAS), has delivered training for 44 senior forensic scientists as 'technical experts' to work alongside national accreditation bodies when assessing forensic laboratories. The training involved different forensic disciplines (digital evidence, pattern recognition, fingerprints, and scene of crime & fire scene investigation). Prior to the SQWEFS project there has been a shortage of such trained people across Europe.

2. Two new guideline documents have been produced:
   - "Guidelines for the Single Laboratory Validation of Instrumental and Human Based Methods in Forensic Science".
   - "Guidance on the Conduct of Proficiency Tests and Collaborative Exercises within ENFSI".

A report has been produced providing a broad overview of forensic education and training (E&T) across Europe and contact points have been established for E&T matters.
During 2013, ENFSI has signed a further monopoly grant agreement, arising from the ISEC 2012 Annual Work Programme (AWP) with the implementation starting on 1 January 2014 and an opening conference being held in Amsterdam on 31st January 2014. Thus, in January 2014 three simultaneous work programmes were being implemented (2010, 2011 and 2012).
The new ENFSI application to the EC for the 2013 monopoly grant was made in December 2013 with a proposal for a 2 year work programme entitled "Towards the Vision for European Forensic Science 2020 (TVEFS-2020)". The theme and the activities continue ENFSI's work towards realising the EU "Council conclusions on the vision for European Forensic Science 2020 including the creation of a European Forensic Science Area and the development of forensic science infrastructure in Europe" approved by the Justice and Home Affairs Council (December 2011). In this way, the monopoly funding will continue to play an important role in helping to achieve the aims of forensic science co-operation and standardisation.
The full details of the ENFSI Monopoly Programmes (2010 to 2013) are provided in the tables.

ENFSI MONOPOLY PROGRAMME 2013

Monopoly Projects (2013) – 2 year programme (application for 645,649)
Programme Theme: "Towards the Vision for European Forensic Science 2020 (TVEFS-2020)".
Current Status: Formal application submitted to the EC in December 2013 (expected start date 1st January 2015)

T1	Creation and shared use of an international database of ignitable liquids and substances.	Jeanet Hendrikse (NFI - The Netherlands)
T2	Development and implementation of new analytical methods and databases for the detection of additives in fuels and fire debris.	Frank Schäfer (BKA - Germany)
T3	Proficiency tests for the fingerprint domain.	Aldo Mattei (RaCIS - Italy)
T4	SmartRank: Likelihood ratio software for searching national DNA databases with complex DNA profiles.	Hinda Haned (NFI - The Netherlands)
T5	SmartRank: Likelihood ratio software for searching national DNA databases with complex DNA profiles.	Ate Kloosterman (NFI - The Netherlands)
T6	The development of a statistical software package for likelihood ratio calculations.	Annabel Bolck (NFI - The Netherlands)

IFSA MEETING IN LYON

In 2013, International Forensic Strategic Alliance (IFSA) held its meeting in Lyon (France) on 6–7 October 2013. The event was combined with the 17th International Forensic Science Symposium (IFSS) which took place at INTERPOL premises in Lyon on 8–10 October 2013. The meeting was attended by Chairpersons from continental forensic networks constituting part of IFSA as well as representatives from UNODC and INTERPOL. ENFSI was represented by the Chairman of the 17th ENFSI Board, Mr. Üllar Lanno, and the Secretary, Ms. Ewa Klimuk.

The complete list of participants included:

1. Mr. Jose Antonio Lorente, Academia Iberoamericana de Criminalística y Estudios Forenses (AICEF)
2. Mr. Alastair Ross, Senior Managers of Australian and New Zealand Forensic Laboratories (SMANZFL)
3. Mr. Üllar Lanno (ENFSI)
4. Ms. Ewa Klimuk (ENFSI)
5. Mrs. Soraya McClung American Society of Crime Laboratory Directors (ASCLD)
6. Mr. Kermit Channell, American Society of Crime Laboratory Directors (ASCLD)
7. Mr. Paul Ludik South African Regional Forensic Science Network (SARFS)
8. Mr. Kong Boon LIM, Asian Forensic Science Network (AFSN)
9. Mrs. Angeline YAP Tiong Whei, Asian Forensic Science Network (AFSN)
10. Mr. Justice Tettey (UNODC)
11. Mrs. Susan Hitchin (INTERPOL)

The gathering was chaired by Mr. Jose Antonio Lorente, IFSA President representing AICEF.
The main topics raised at the meeting included, but were not limited to, issues related to minimum requirements, standard documents elaborated by IFSA with the aim to raise the level of forensic science in underdeveloped countries, IFSA website and IFSA Secretariat. In addition to this, the meeting gave an opportunity for every single network to present itself, speak of its priorities and name current directions of forensic science development, observed on given continents.
The IFSA attendees took a decision that IFSA website shall be further developed in the form of a subpage published on the open ENFSI website www.enfsi.eu and maintained by the ENFSI Secretariat. Also, it was decided that, from now on, IFSA Secretariats will be run by continental networks of given IFSA Presidents.
At the meeting, Mr. Jose Antonio Lorente completed the two year period of carrying out the function of President of IFSA. He was congratulated and warmly thanked by the meeting participants for the excellent fulfillment of this mission. The next IFSA Chairman will be Mr. Lam Kian Ming from the Health Sciences Authority, Singapore. He will play his role, representing Asian Forensic Science Network (AFSN), for the two forthcoming years (2014–2015).

Participants of IFSA Meeting in Lyon

IFSA Discussions Held in Lyon

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Tripartite Meeting of ENFSI, Eurojust and Europol, The Hague, The Netherlands

TRIPARTITE MEETING OF ENFSI, EUROJUST AND EUROPOL IN THE HAGUE

On the 5th of July 2013, at Eurojust Headquarters located in the Hague, two delegates from ENFSI met with Eurojust and Europol representatives. ENFSI was represented by the Chairman of the 17th Board, Mr. Üllar Lanno and the Chair of Research & Development Standing Committee (R&D SC), Mr. Marcel van der Steen. The Hague meeting was chaired by Mr. Klaus Rackwittz, Administrative Director at Eurojust. The second representative from Eurojust was Mr. Jerzy Iwanicki, Assistant to the National Member of Poland. Europol was represented by Mr. Olivier Burgersdijk, Head of Strategy, European Cybercrime Centre by Mr. Pierre van Renterghem, Senior specialist and forensic expert. Mr. Renterghem is known to the ENFSI network as Deputy Chair of DNA Working Group and a member of Scene of Crime Working Group (SoC WG), who runs the Europol Platform of Experts (web portal) used by SoC WG as a tool for internal communication. The three parties gathered together in order to openly discuss mutually important issues related to forensic science and European Forensic Science Area 2020 (EFSA2020). They considered the way of creating the synergy between their activities and work towards the European Financial Prospective 2014–2020. In connection to this topics, ENFSI presented an initiative to arrange a high level meeting in Brussels with the participation of ENFSI, Eurojust, Europol and European Commission to talk about EFSA2020, Horizon2020 and other security related common interest areas. The joint meeting organized by the three organizations proceeded in a pleasant atmosphere, which resulted in a fruitful discussion.

ON THE 5TH OF JULY 2013, AT EUROJUST HEADQUARTERS LOCATED IN THE HAGUE, TWO DELEGATES FROM ENFSI MET WITH EUROJUST AND EUROPOL REPRESENTATIVES

STANDARDIZING FORENSIC SCIENCE

STANDARDIZING FORENSIC SCIENCE

Throughout the 2013, CEN Technical Committee 419 – "Forensic science processes" continued its work on the standard concerning the whole forensic science delivery process covering all aspects from the scene of crime to Court room.

Appointed by CEN on 24 May 2012, the Committee focuses of four areas which include:

1. Crime scene, exhibit handling and control;
2. Delivery of results through the processes of forensic science examinations and analysis of various types of physical material;
3. Evaluation and interpretation of the results of forensic science examinations and analysis in the context of the case;
4. Reporting results and conclusions from the forensic science examination and analysis, data exchange and the standardization of the documentation used for forensic purposes.

The first meeting of CEN/TC 419 took place on 23 October 2013 in Warsaw, Poland. At the meeting, the participants agreed on the title and scope of the Project Committee and discussed the work programme. In 2013, the Committee members gathered also in Stockholm, Sweden, to held the 2nd Plenary Meeting. ENFSI was represented there by Mr. Tore Olsson and Mrs. Christina Bertler-Edlund.

Last year, ENFSI obtained liaison status with the Committee. This means that the network is allowed to send its representatives to CEN/TC 419 meetings and contribute to the work process by the committee by providing its input to draft standard document elaborated by the committee members. This allows ENFSI to play an important role in the European standardization process and therefore, execute strategic goals mentioned in ENFSI Strategic Plan 2011–2014.

1st Meeting of CEN/TC419 in Warsaw, Poland

1st Meeting of CEN/TC419 in Warsaw, Poland

MAP OF ENFSI MEMBERS

ENFSI MEMBERS

1 National Bureau of Expertises, NDE-SNPO YEREVAN, Armenia; 2 FORENSIC SCIENCES CENTRE FSC MJ BAKU, Azerbaijan; 3 Criminal Intelligence Service Austria, Forensic Science Service, DFS VIENNA, Austria; 4 National Institute of Criminalistics and Criminology, INCC BRUSSELS, Belgium; 5 Forensic Expertise Department, OKT SARAJEVO, Bosnia and Herzegovina; 6 Research Institute of Forensic Science and Criminology, RIFSC SOFIA, Bulgaria; 7 Forensic Science Centre "Ivan Vucetic", FSC ZAGREB, Croatia; 8 Criminalistic Services Cyprus Police CSCP NICOSIA, Cyprus; 9 Institute of Criminalistics Prague, ICP PRAGUE, Czech Republic; 10 National Forensic Services, KTC VANLÖSE, Denmark; 11 Estonian Forensic Science Institute, EFSI TALLINN, Estonia; 12 National Bureau of Investigation Forensic Laboratory, RTL VANTAA, Finland; 13 Forensic Sciences Institute of the French Gendamerie, IRCGN ROSNY-SOUS-BOIS, France; 14 National Forensic Science Institute, INPS ECULLY, France; 15 Legal Entity under Public Law Levan Samkharauli National Forensics Bureau, NFB TBILISI, Georgia; 16 Forensic Science Institute BKA, KTI-BKA WIESBADEN, Germany; 17 Forensic Science Institute, LKA KT BERLIN, Germany; 18 Forensic Science Institute, LKA / KTI DÜSSELDORF, Germany; 19 Forensic Science Institute, LKA/KTI STUTTGART, Germany; 20 Forensic Science Institute, LKA/KTI HESSEN, Germany; 21 Forensic Science Institute, BLKA – KTI MUNICH, Germany; 22 Forensic Science Institute, LKA HAMBURG, Germany; 23 Forensic Science Institute, LKA/KTI HANNOVER, Germany; 24 Hungarian Institute for Forensic Sciences, HIFS BUDAPEST, Hungary; 25 Forensic Science Laboratory, FSL DUBLIN, Ireland; 26 Central Anticrime Directorate of Italian National Police, Forensic Science Police Service, DAC-SPS ROMA, Italy; 27 Forensic science laboratories of Carabinieri Force, RaCIS ROMA, Italy; 28 State Forensic Science Bureau, SFSB RIGA, Latvia; 29 State Police Forensic Service Department of the Ministry of Interior of Latvia, FSD RIGA, Latvia; 30 Forensic Science Center of Lithuania, FSC VILNIUS, Lithuania; 31 Lithuanian Police Forensic Science Centre, FSC-LT VILNIUS, Lithuania; 32 Forensic Center of Police Directorate of Montenegro, FCPDM DANILOV-GRAD, Montenegro; 33 The Netherlands Forensic Institute, NFI THE HAGUE, The Netherlands; 34 National Criminal Investigation Forensic Science Division, NCIS OSLO, Norway; 35 Institute of Forensic Research, IFR KRAKOW, Poland; 36 Central Forensic Laboratory of the Police, CFLP WARSAW, Poland; 37 The Internal Security Agency, Forensic Laboratory, ABW WARSAW, Poland; 38 Laboratory of Scientific Police, LPC LISBON, Portugal; 39 National Institute of Forensic Expertise, NIFE BUCHAREST, Romania; 40 National Forensic Science Institute NFSI BUCHAREST, Romania; 41 The Russian Federal Center of Forensic Science of the Ministry of Justice, RFCFS MOSCOW, Russia; 42 North-Western Forensic Science Centre, NWFSC ST. PETERSBURG, Russia; 43 National Criminalistic-Technical Centre NCTC BELGRADE, Serbia; 44 Institute of Forensic Science IFS BRATISLAVA, Slovakia; 45 Ministry of Interior Forensic Science Laboratory, FSL LJUBLJANA, Slovenia; 46 General Commissary of Scientific Police GCSP MADRID, Spain; 47 Criminalistic Service of the Civil Guard, SECRIM MADRID, Spain; 48 National Institute of Toxicology and Forensic Science, INTCF MADRID, Spain; 49 Scientific Police Division CME, SABADELL, Spain; 50 Forensic Science Unit, Basque Country Police, FSU, ERANDIO, Spain; 51 Swedish National Laboratory of Forensic Science, SKL LINKÖPING, Sweden; 52 School of Forensic Science, ESC LAUSANNE, Switzerland; 53 Forensic Science Institute Zurich, FSI ZÜRICH, Switzerland; 54 Institute of Forensic Medicine Istanbul University, ATE ISTANBUL, Turkey; 55 Gendarmerie Forensic Department GFD ANKARA, Turkey; 56 Headquarters of Forensic Department KDB KPL ANKARA, Turkey; 57 The Council of Forensic Medicine, ATK ISTANBUL, Turkey; 58 State Scientific Research Expertise and Forensic Centre, SREFC KYIV, Ukraine; 59 Forensic Science Northern Ireland, FSNI BELFAST, United Kingdom; 60 University of Strathclyde, Centre for Forensic Science, CFS GLASGOW, United Kingdom; 61 Scottish Police Services Authority-Forensic Services, SPSAFS GLASGOW, United Kingdom; 62 LGC Forensics, LGCF OXFORDSHIRE, United Kingdom; 63 Key Forensics Services Ltd, KFS COVENTRY, United Kingdom; 64 Metropolitan Police Service, MPS LONDON, United Kingdom.

COLOPHON

EDITOR
Ewa Klimuk

CONTRIBUTORS
Thomas Andermann
Marcel van Der Steen
Philippe Esperança
Richard Gill
Mike Groen
Üllar Lanno
Wim Neuteboom
Peter Pfefferli
Gary Pugh
Sasa Zugaj
Katarzyna Zwierzyk

LOGISTIC COORDINATOR
Ewa Klimuk

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secretariat@enfsi.eu

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European Forensic Initiatives Centre Foundation
Aleja Wyzwolenia 3-5/29
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Tel/Fax: +48 22 622 29 67
e-mail: secretariat@enfsi.eu
website: www.enfsi.eu

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ENFSI Secretariat
Aleja Wyzwolenia 3-5/29
00-572 Warsaw, Poland
Tel/Fax: +48 22 622 29 67
e-mail: secretariat@enfsi.eu
website: www.enfsi.eu

folkehelseinstituttet

A probabilistic assessment of secondary transfer at the crime scene

Elida Fonneløp

Department of Forensic Biology

Norwegian Institute of Public Health

Background

- Transfer of DNA
  - active
  - Passive
- Tendency to associate profiles to the crime

Background

- Increasing sensitivity in analysis
  - Mini-tapes
  - New amplification multiplexes
  - New Instrumentation

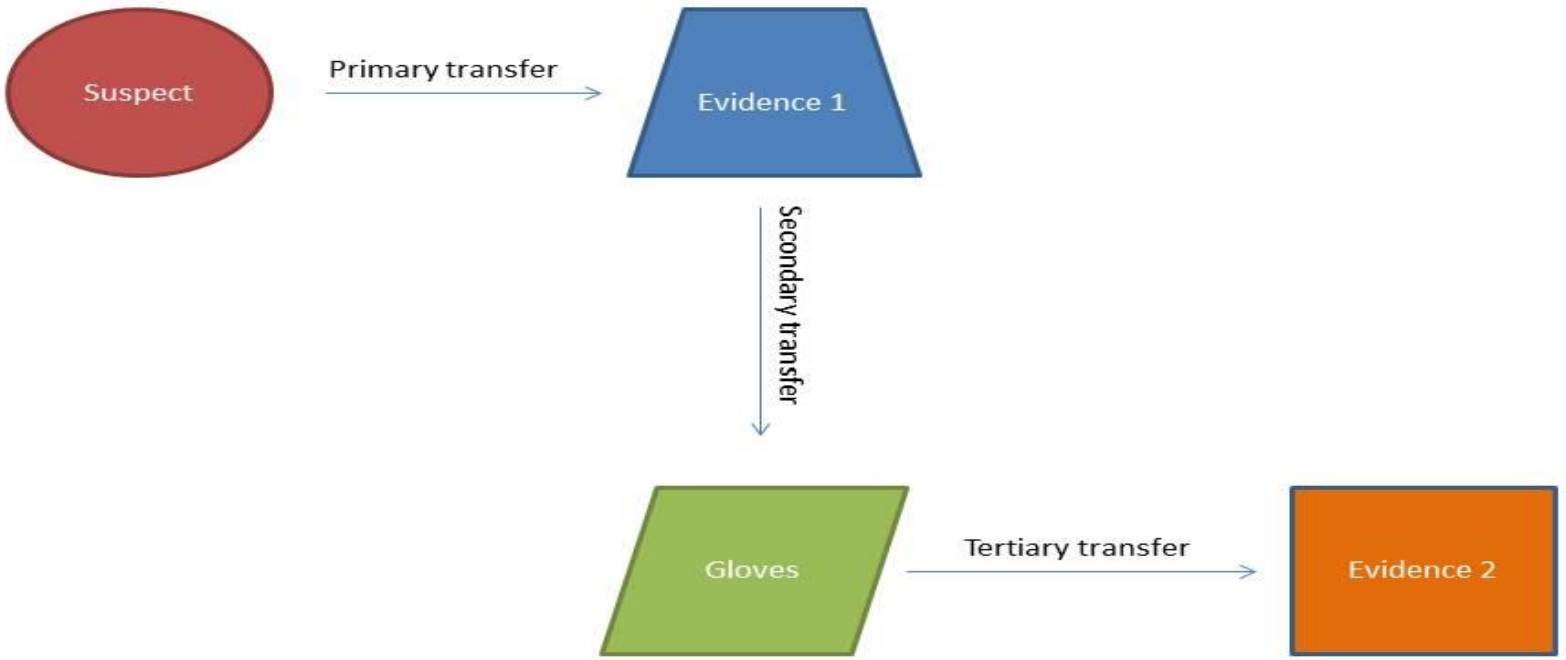
+ Higher success rates

÷ Potential of innocent DNA transfer is not properly understood

Background

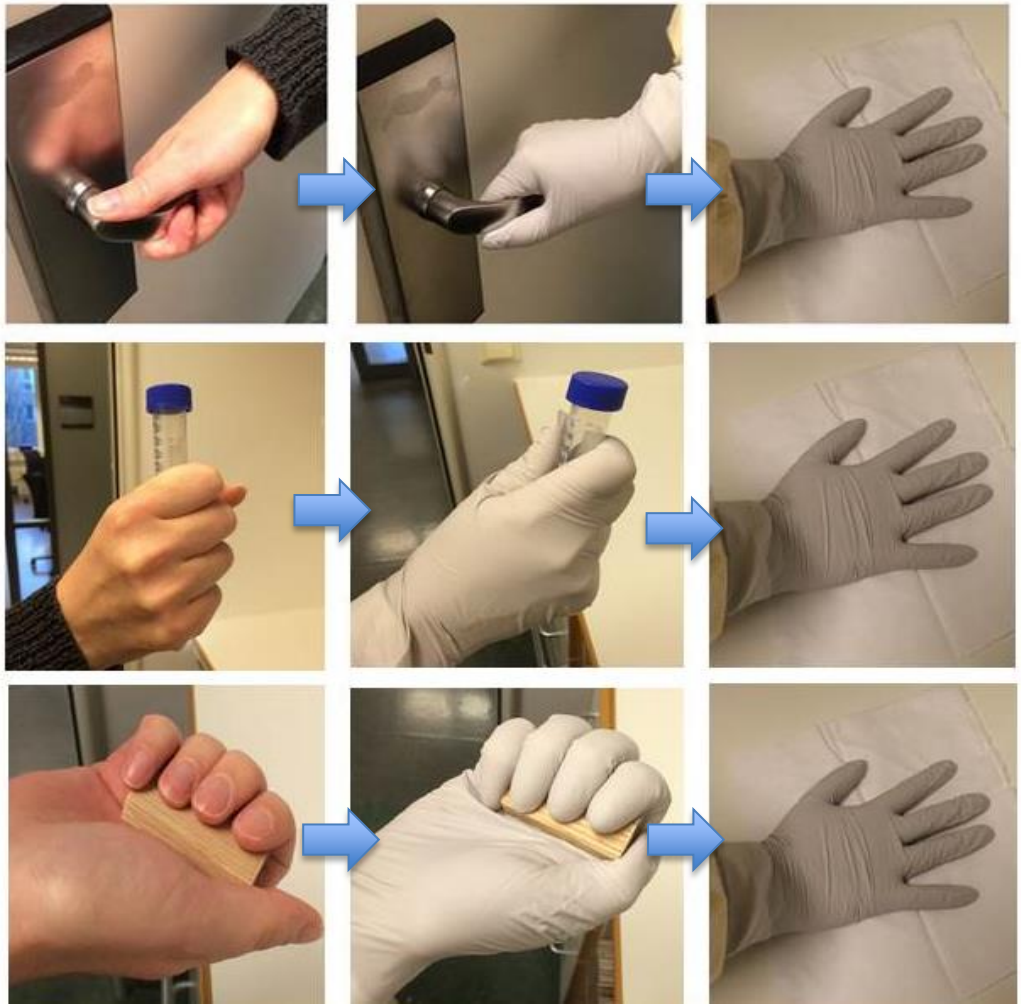
- Investigators always use protective clothing to protect integrity of crime scene
- Is there potential for the investigator to act as a vector of DNA-transfer between items within the crime-scene itself?

Design


Suspect
Primary transfer
Evidence 1
Secondary transfer
Gloves
Tertiary transfer
Evidence 2

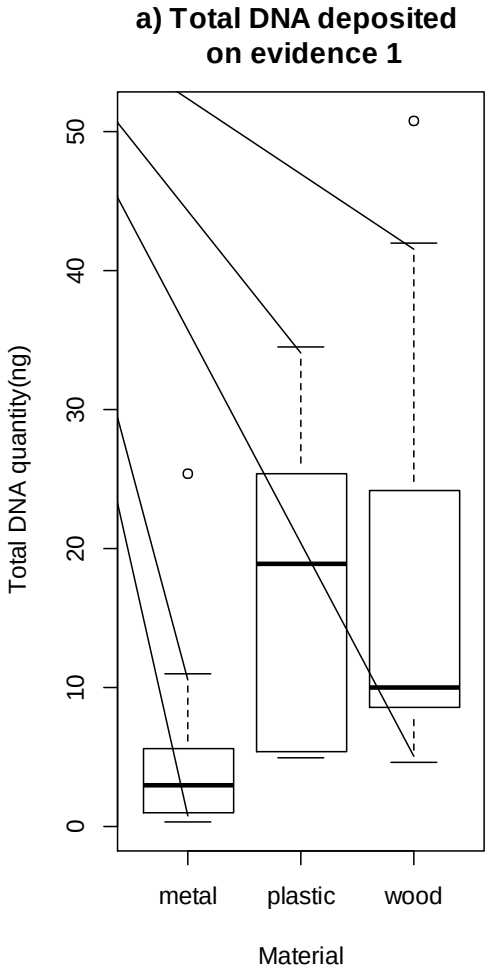
Experiments

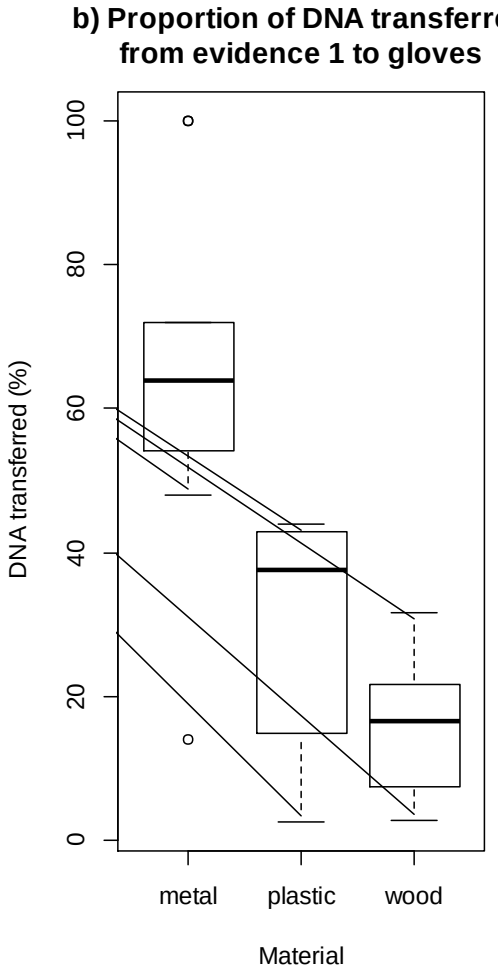
Evidence 1 Gloves Evidence 2



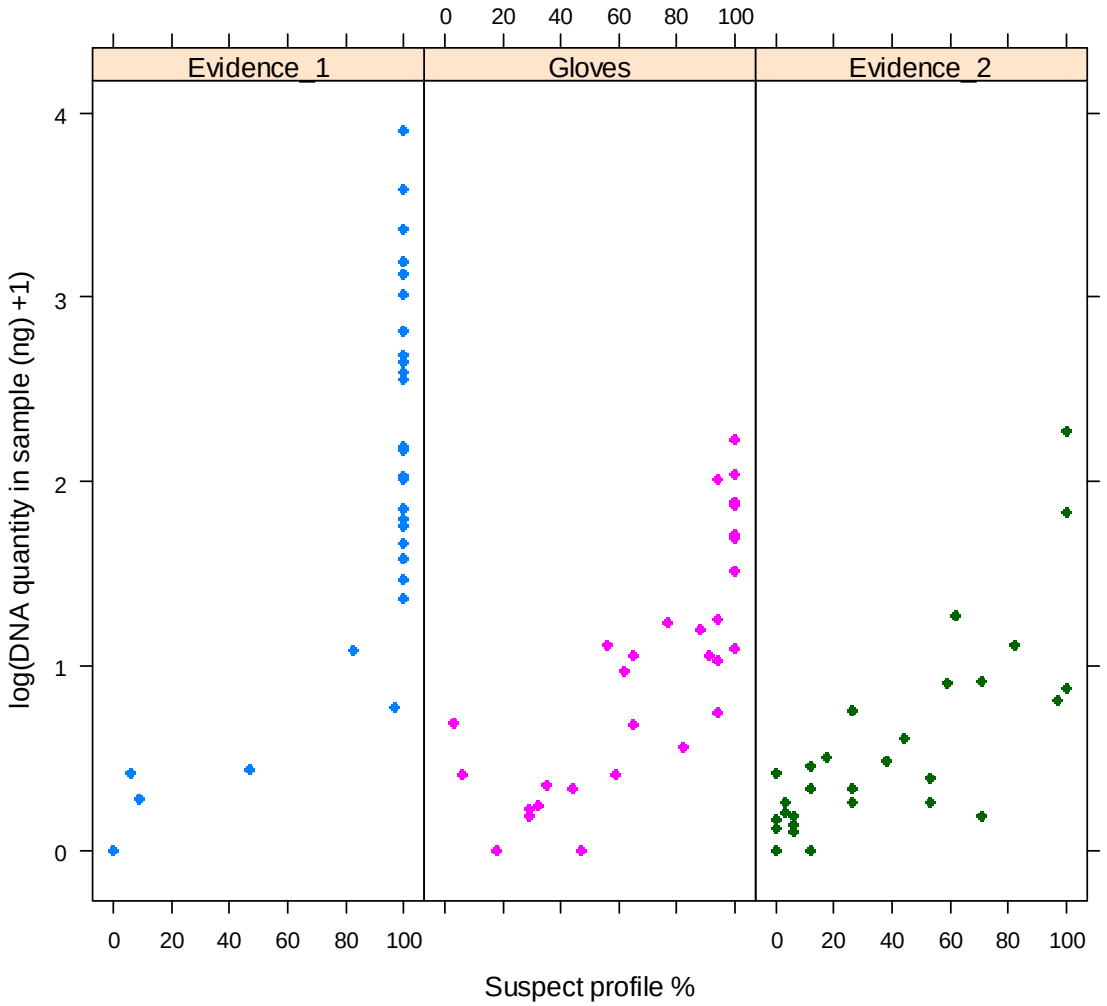
- Evidence 1: Metal, plastic or wood
- Nitrile disposable gloves
- Evidence 2: Fabric or paper
- 30 experiments
- 3 good shedder donors
- DNA-free surfaces
- Mini-tapes
- Standard analysis

Results – transfer rates

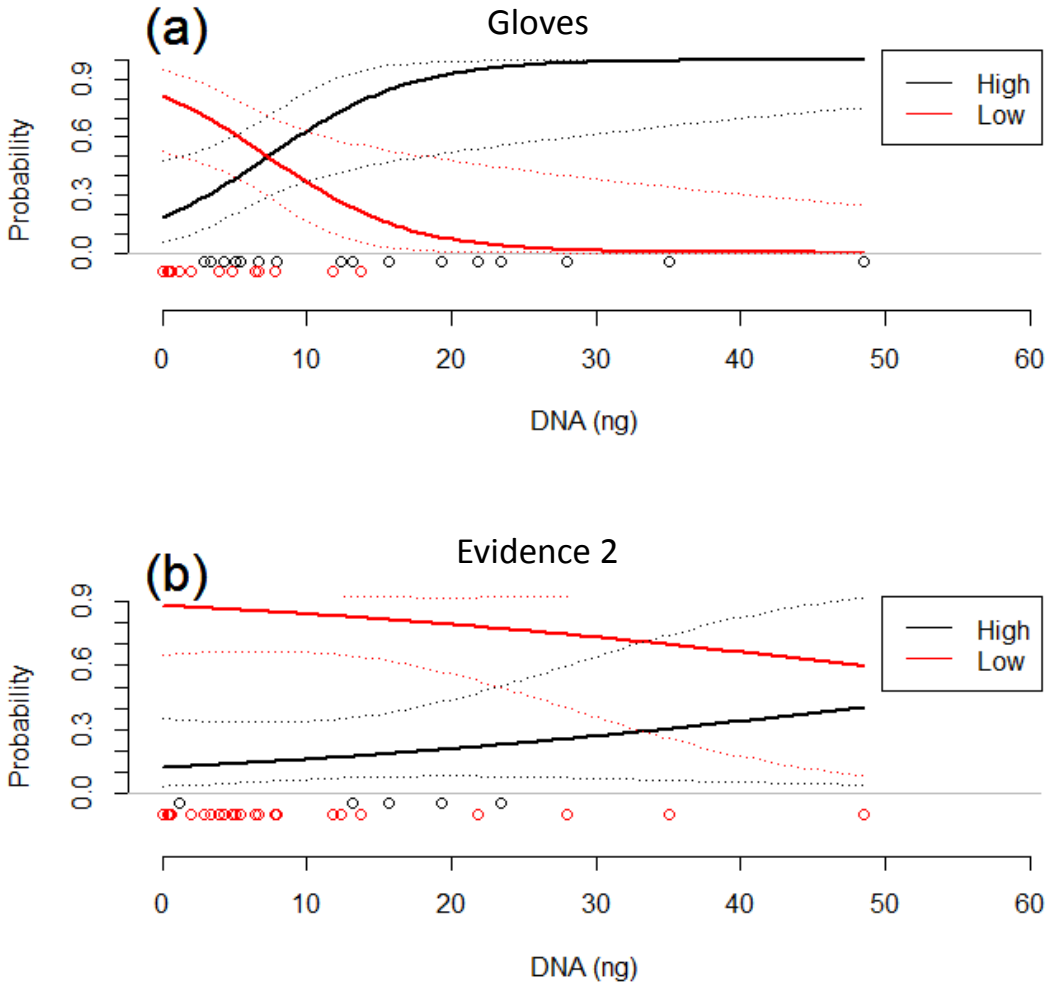




Results - DNA profiles



Prediction model



- Probability of secondary and tertiary transfer given the amount of DNA deposited on evidence 1 by the suspect

- High quality profile ≥ 10 loci
- Low quality profile ≤ 9 loci

Evidence of quaternary transfer events

- Mixture in all elements of a transfer chain
- Match with reference sample
- 27 of 32 alleles (18 exclusive)
- Proposed chain of transfer

  gf→suspect→evidence1→gloves→evidence2

Conclusions

- Our findings show that “touch-DNA” can be transferred between multiple objects, and that disposable gloves can act as an efficient transfer vector.

Continous work

- Collaboration with police (SOCO)

NIST Update
Applied Genetics Group

EDNAP
November 19, 2014

Katherine Butler Gettings, Ph.D.
Applied Genetics Group
Biomolecular Measurement Division

National Institute of
Standards and Technology
U.S. Department of Commerce

NIST Applied Genetics Group Update

Group Leader – peter.vallone@nist.gov

Standard Reference Materials

PCR-based DNA Profiling Standard 2391c

- 6 Components: 4 liquid extracts, 2 paper, 1 mixture
- Tested with new kits: GlobalFiler, YFiler Plus, PowerPlex Fusion, PPY23, etc.
- Certifying for new loci: D6S1043, 12 additional Y-STRs, 12 X-STRs, 30 InDels
- Certifying for sequences: to assist with transition to NGS

Human DNA Quant Standard 2372a

- Migrating away from UV based measurements
- Certifying for "copy/target number" using digital PCR
- BioRad QX100 (droplet) and Fluidigm BioMark (chamber)

mtDNA Sequencing Standard 2392/2392-I

- 3 Components
  - 9947A, HL60, CHR
- Certifying whole genome using NGS
  - PGM, MiSeq, HiSeq, SOLiD
- Concordance across platforms
- Heteroplasmies below Sanger LOD

Rapid DNA

ANDE – NetBio

RapidHit 200 – IntegenX

- Positive and Negative Control Study
- Fall 2014 Maturity Assessment
- Feedback to SWGDAM R-DNA subcommittee

STR Mixture Interpretation

- LR mix Studio: Haned and Gill
  - LR mix in a user-friendly GUI
- DNA-View Mixture Solution: Charles Brenner
  - windows version under development
- STRmix: ESR and S. Australia collaboration
- LikeLTD: Balding
- Lab Retriever: Lohmueller, Rudin and Inman
- TrueAllele: Cybergenetics

Next-Generation Sequencing

PGM SNP Panels

- Degraded DNA & Sensitivity Study
  - IISNP, GlobalFiler, IdentiFiler Plus, MiniFiler, DIPplex
- Ancestry SNPs
  - Analysis Software / Interpretation Models / Reference Databases

Illumina FGx

- ForenSeq (STR + SNP)

Promega PowerSeq Auto - STR

2391c – becky.hill@nist.gov · 2372a – margaret.kline@nist.gov · 2392 – michael.coble@nist.gov

Rapid DNA – erica.butts@nist.gov · NGS – katherine.gettings@nist.gov · kevin.kiesler@nist.gov

Forensic STR Sequence Diversity

NIST Population Samples

- N=183
  - Caucasian (70)
  - Hispanic (45)
  - African American (68)

Amplification & Library Prep

- 2 x 0.5 ng input DNA
- PowerSeq Auto System
- Illumina TruSeq HS PCR-Free

Sequencing

- MiSeq

Bioinformatics

- STRait Razor
- ExactID (Battelle)
- CE concordance

Pop Gen

- Prob of Identity
- Heterozygosity

Forensic STR Sequence Diversity

Plate 1 Samples with Lowest and Highest Coverage

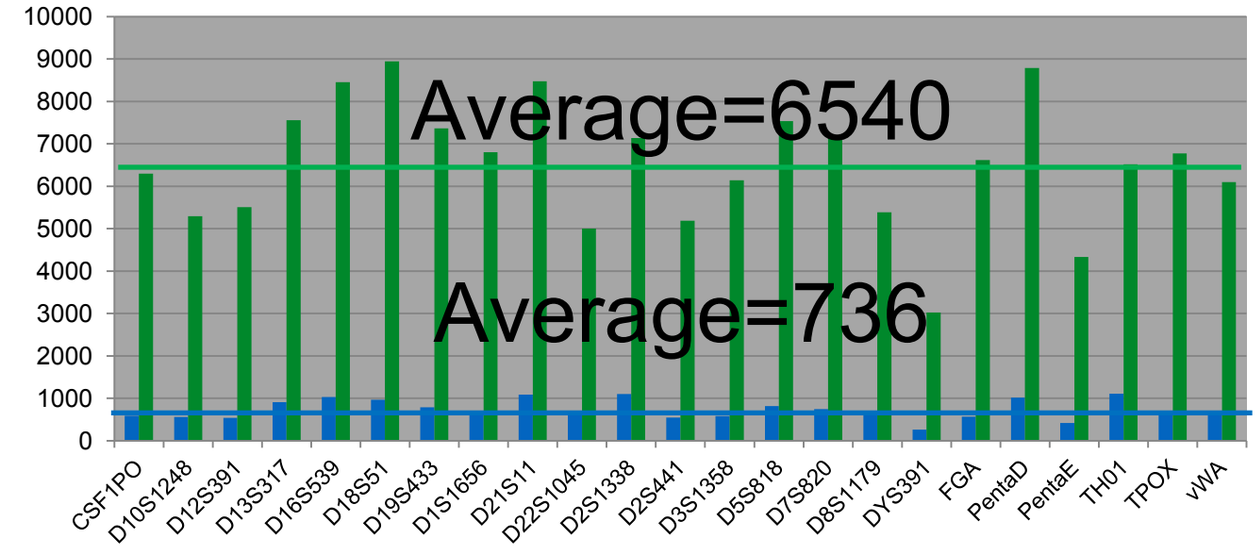
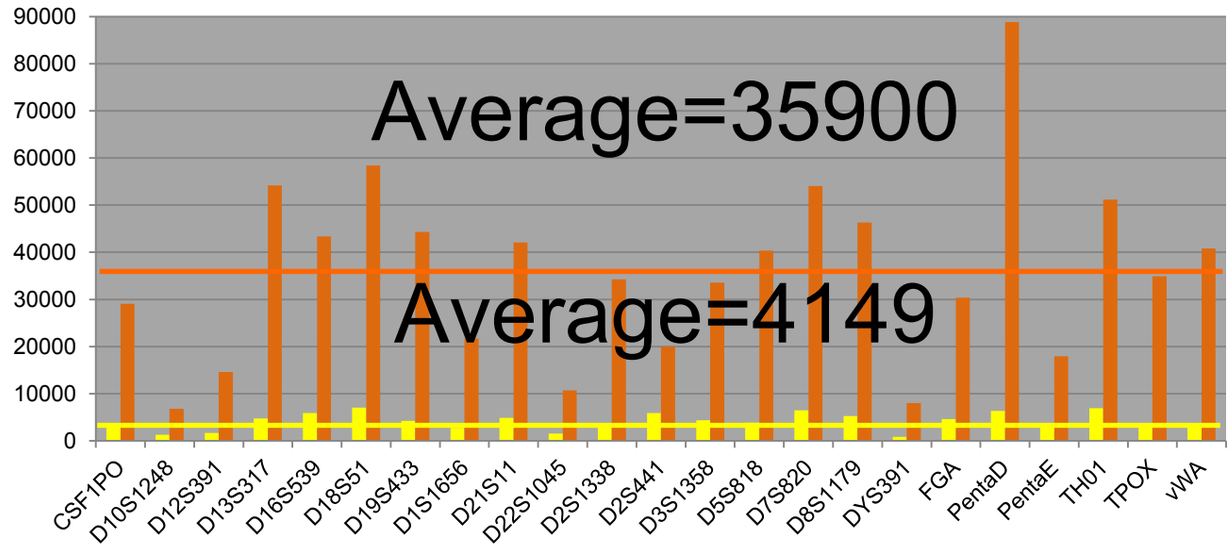
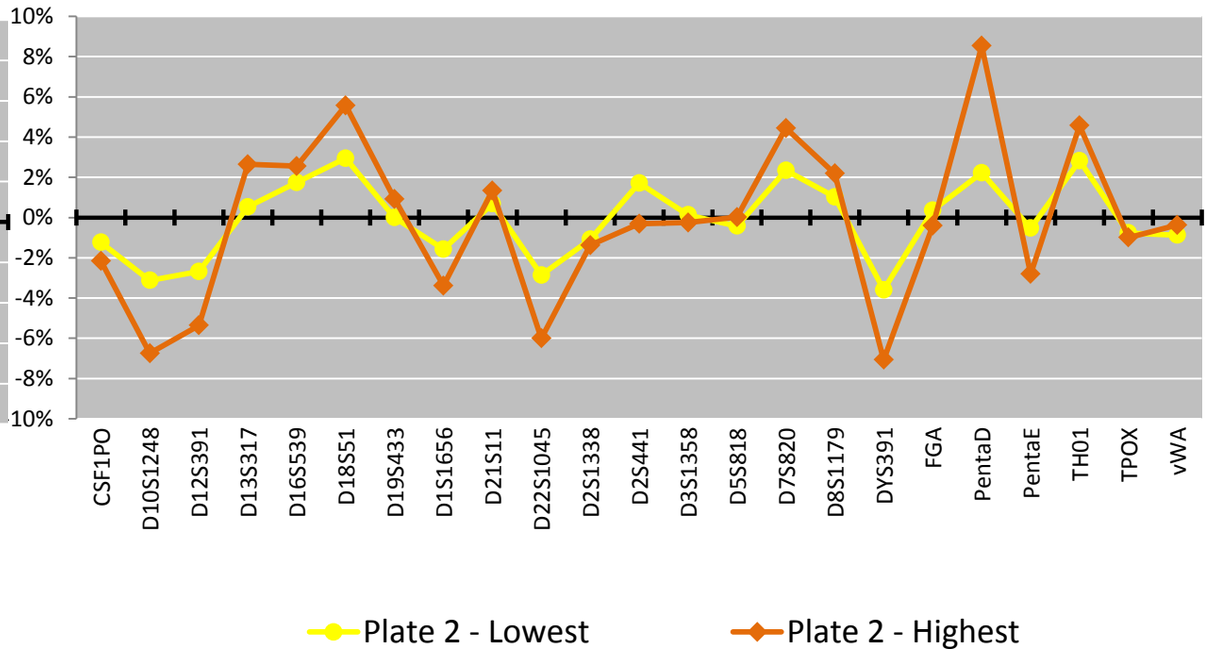


Plate 2 Samples with Lowest and Highest Coverage



10 fold difference in coverage shows similar inter-locus balance



Forensic STR Sequence Diversity

CE Concordance Check Results:

24 loci x 183 samples = 4392 loci evaluated

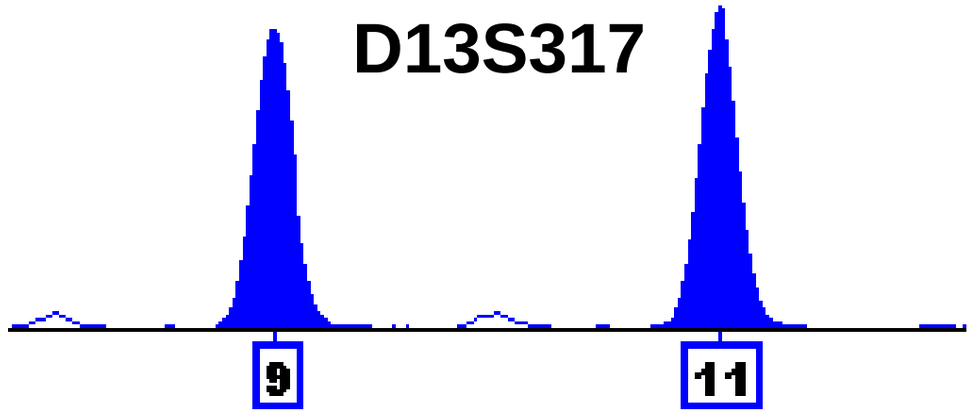
ExactID and STRait Razor

> 99% concordance with CE data

	Discordant Loci in CE compare		
	Exact ID Only	ExactID+ STRait Razor	STRait Razor Only
D13S317		5	
D7S820		1	
Penta D	15		
D18S51	3		
D19S433		1	2
D12S391			3

Forensic STR Sequence Diversity

	Discordant Loci in CE compare		
	Exact ID Only	ExactID+ STRait Razor	STRait Razor Only
D13S317		5	
D7S820		1	
Penta D	15		
D18S51	3		
D19S433		1	2
D12S391			3



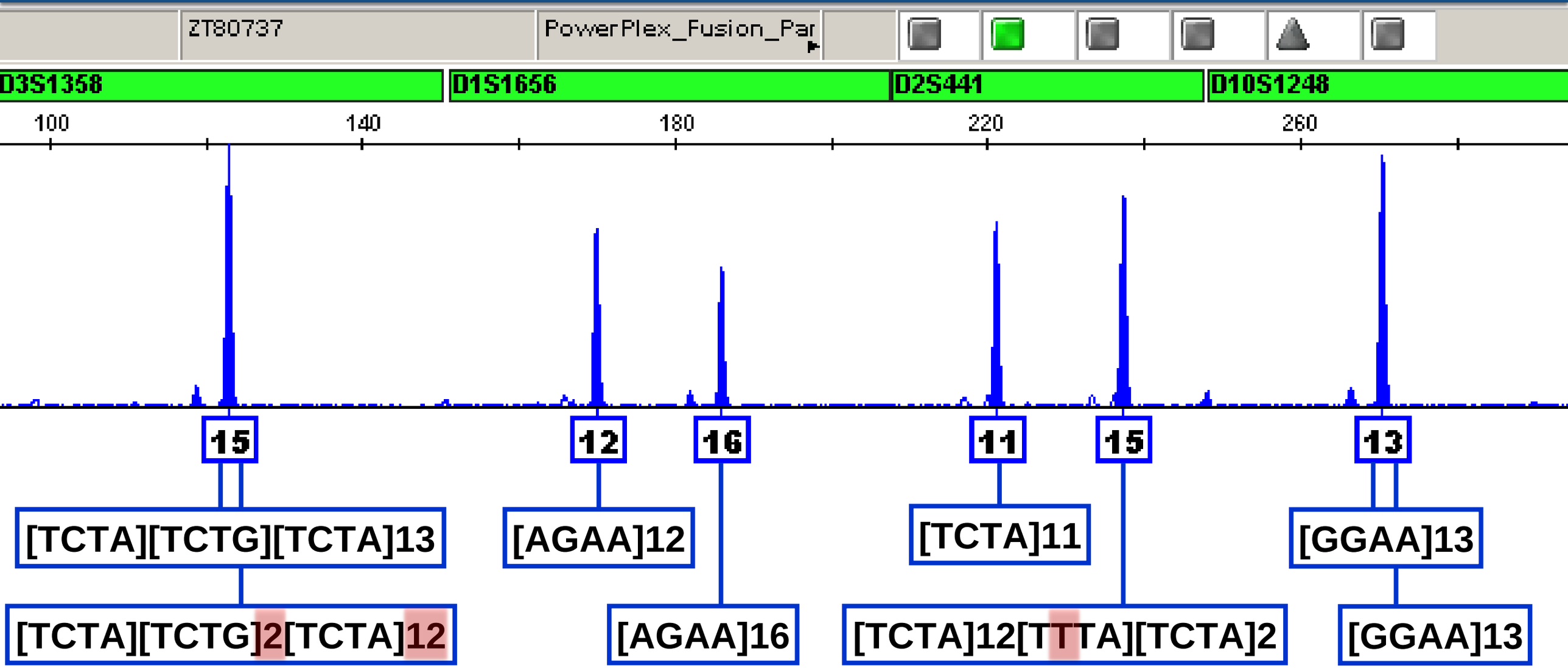
NGS Sequence	$[TATC]_{10}$ $[TATC]_{11}$

← Repeat Region NGS Recognition Region 4 bp Deletion CE Primer Binding Site→

```
TATC TATC TATC AATCAATCATCTATCTATCTTTCTGTC----TTTTTGGGCTGCCTATGGCTCAA
TATC TATC TATC AATCAATCATCTATCTATCTTTCTGTCTGTCTTTTTGGGCTGCCTATGGCTCAA
```

Flanking region InDel: Bioinformatic pipelines may reduce the region used for genotyping, resulting in deletions not being “counted” as they would via CE

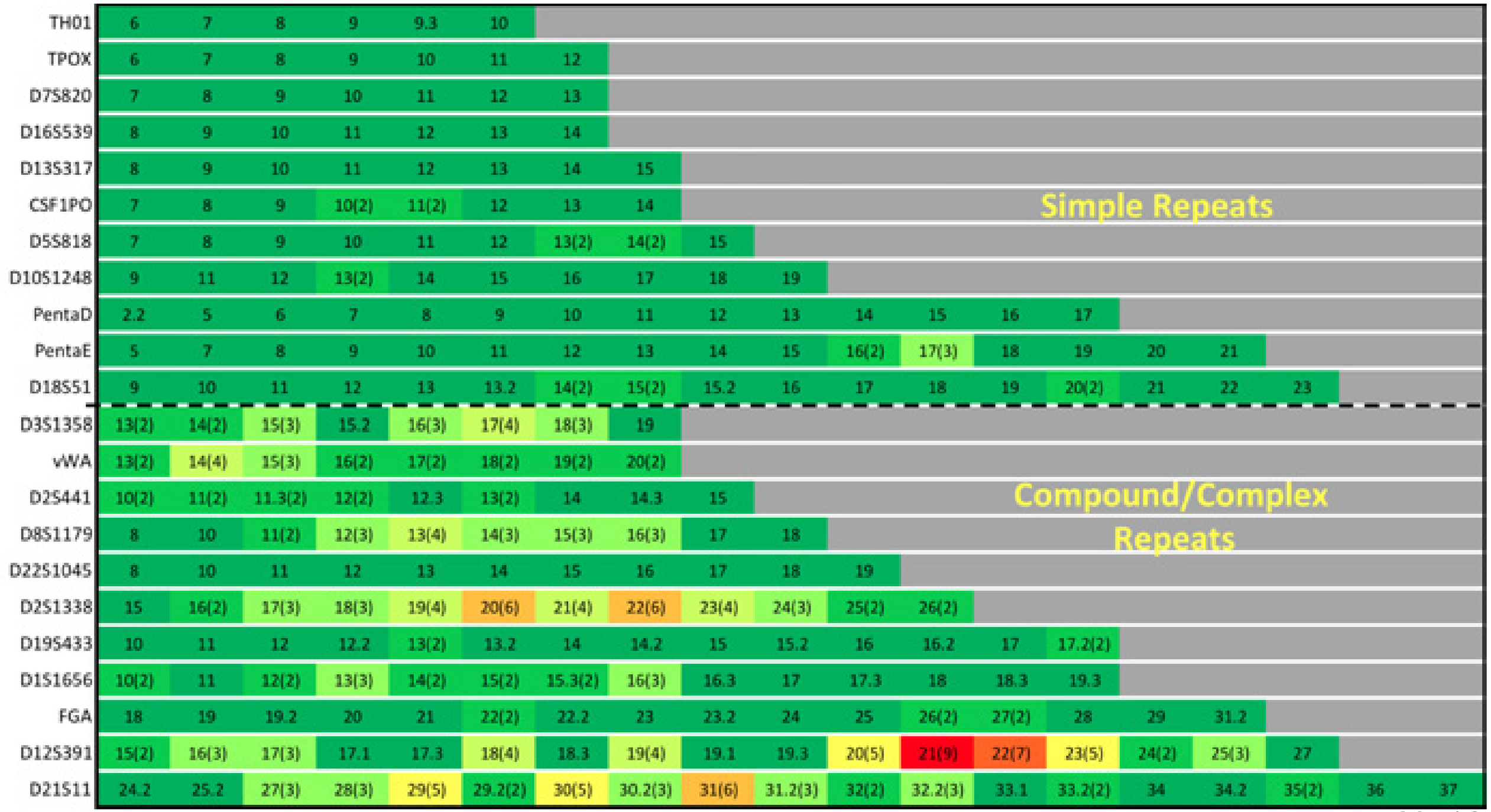
Forensic STR Sequence Diversity



Sequence-Based Heterozygote: A locus that appears homozygous in length-based measurements (such as CE), but is heterozygous by sequence

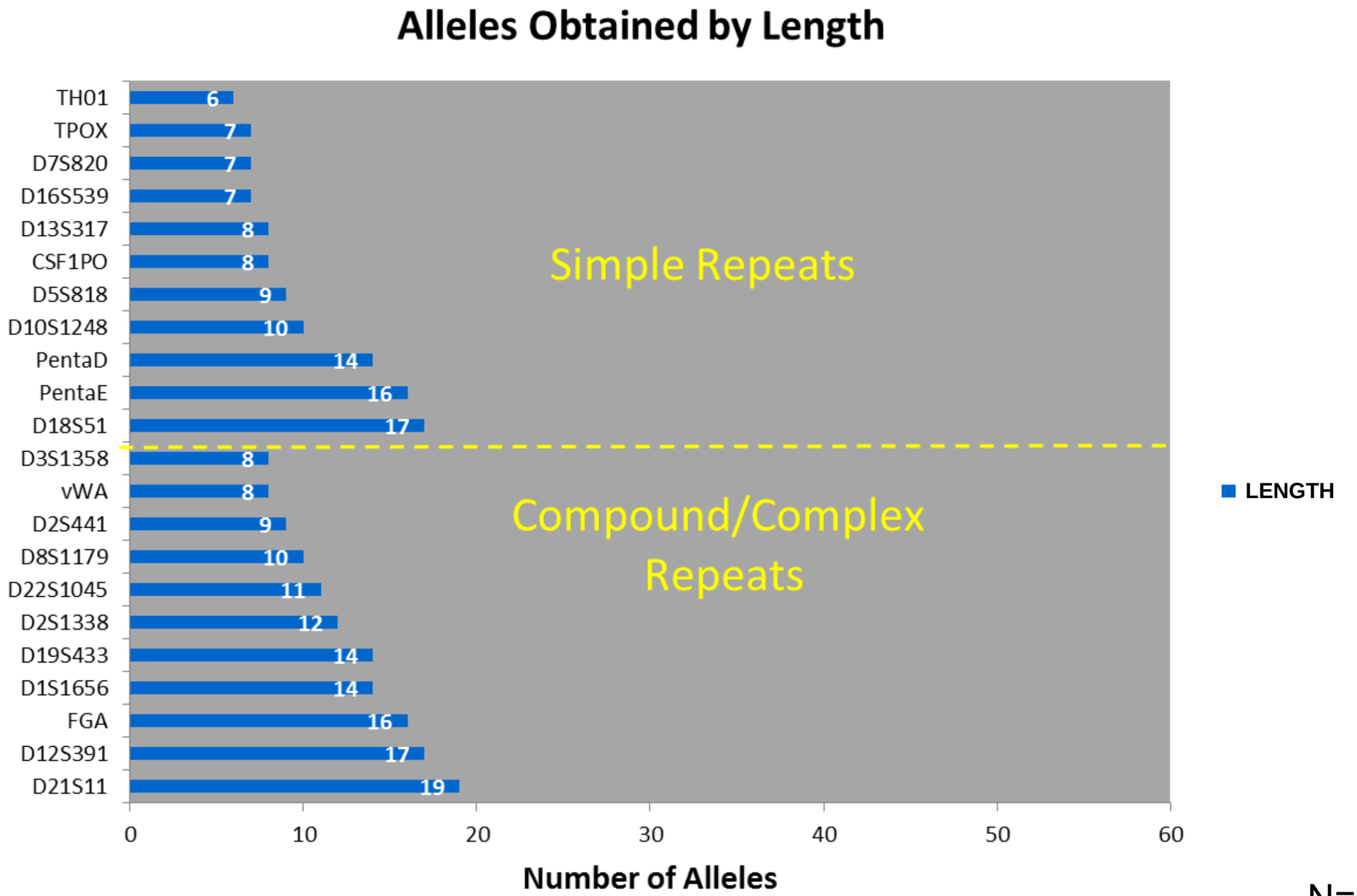
Forensic STR Sequence Diversity

Additional Alleles Obtained by Sequencing



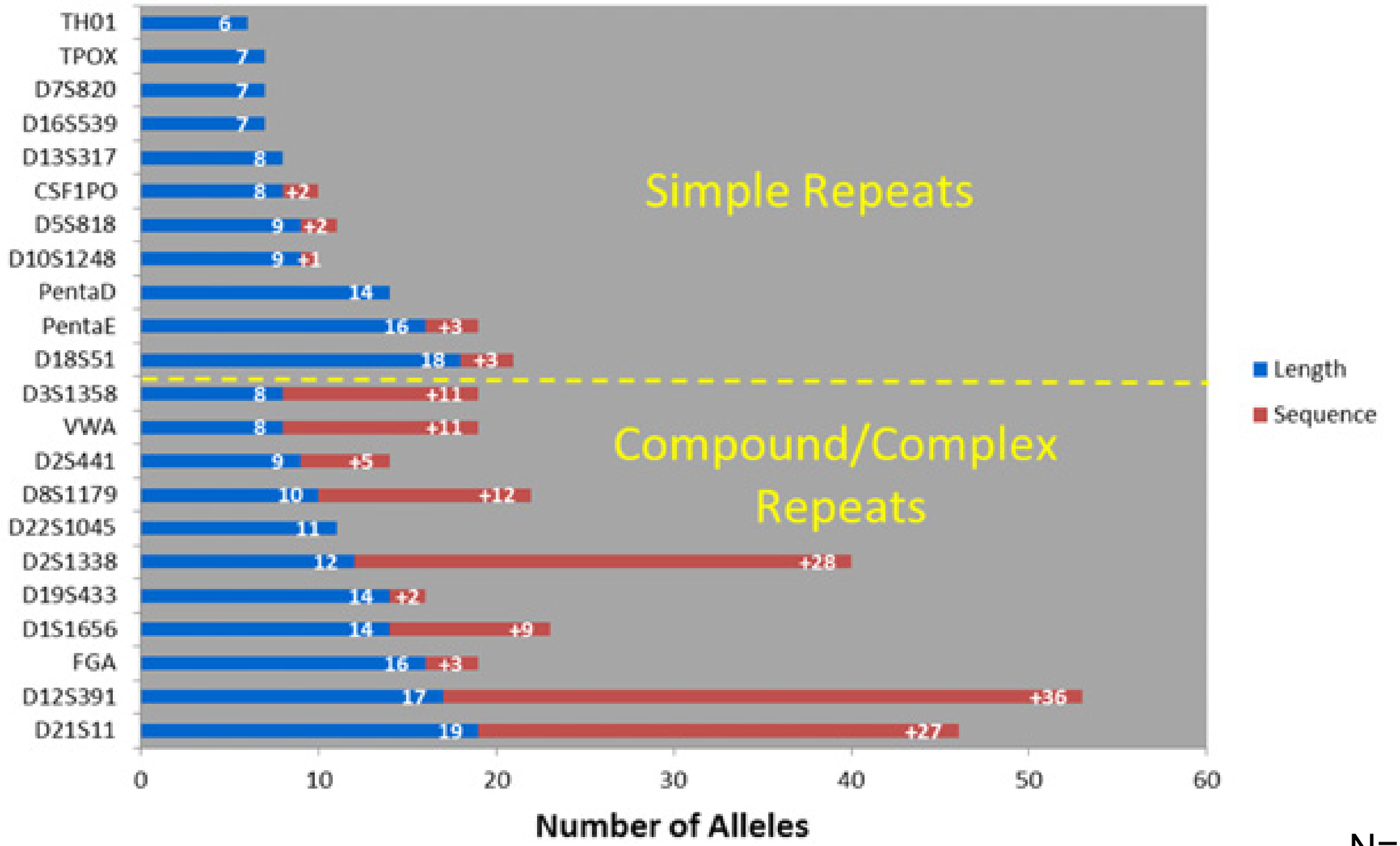
N=183

Forensic STR Sequence Diversity



N=183

Forensic STR Sequence Diversity



N=183

Forensic STR Sequence Diversity

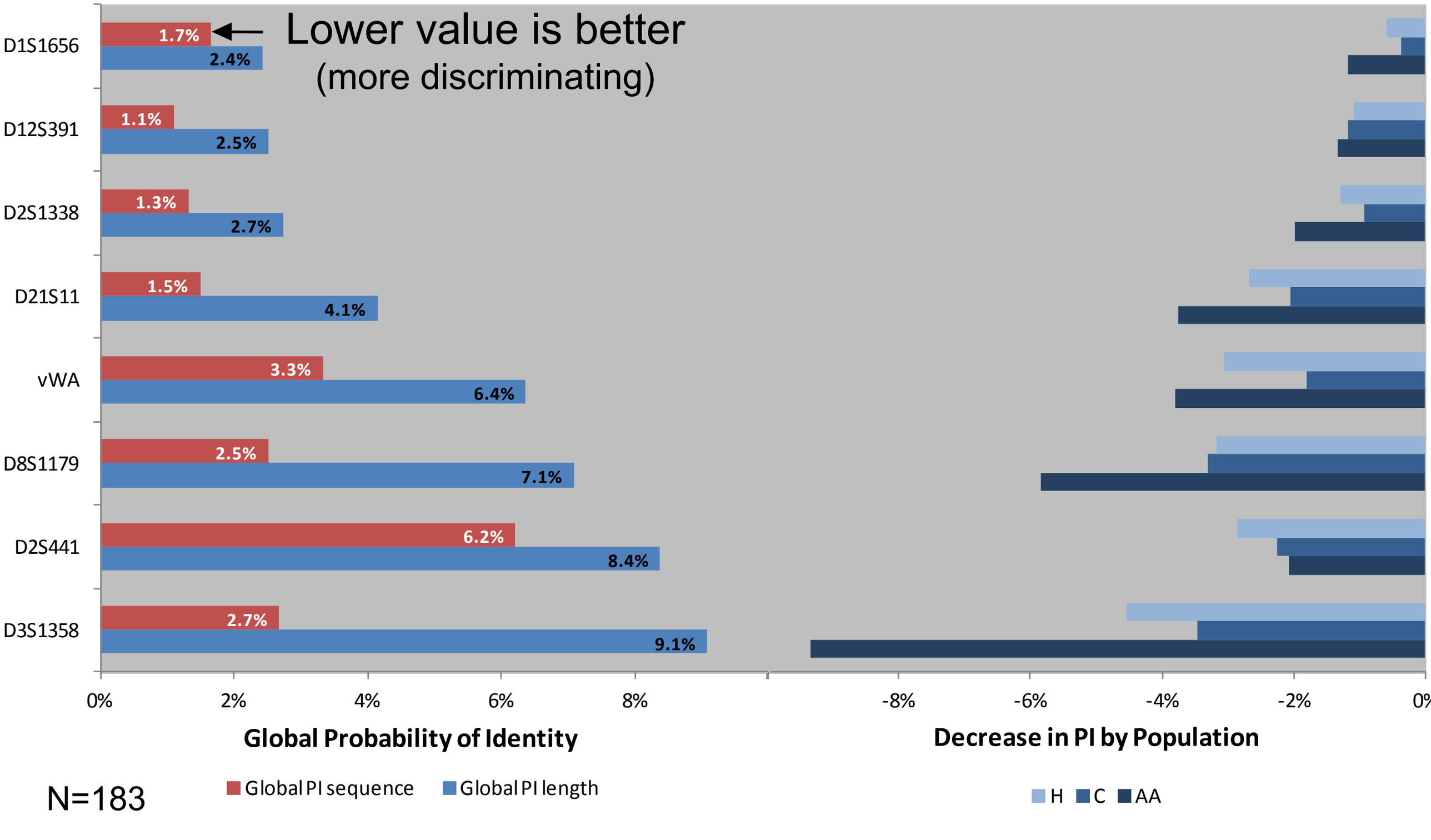
Probability of Identity

Sum of each genotype frequency^2 at each locus

$$= \sum_{i=1}^{n} x_i^2$$

Probability that two unrelated individuals selected at random will have the same genotype at a locus

Forensic STR Sequence Diversity



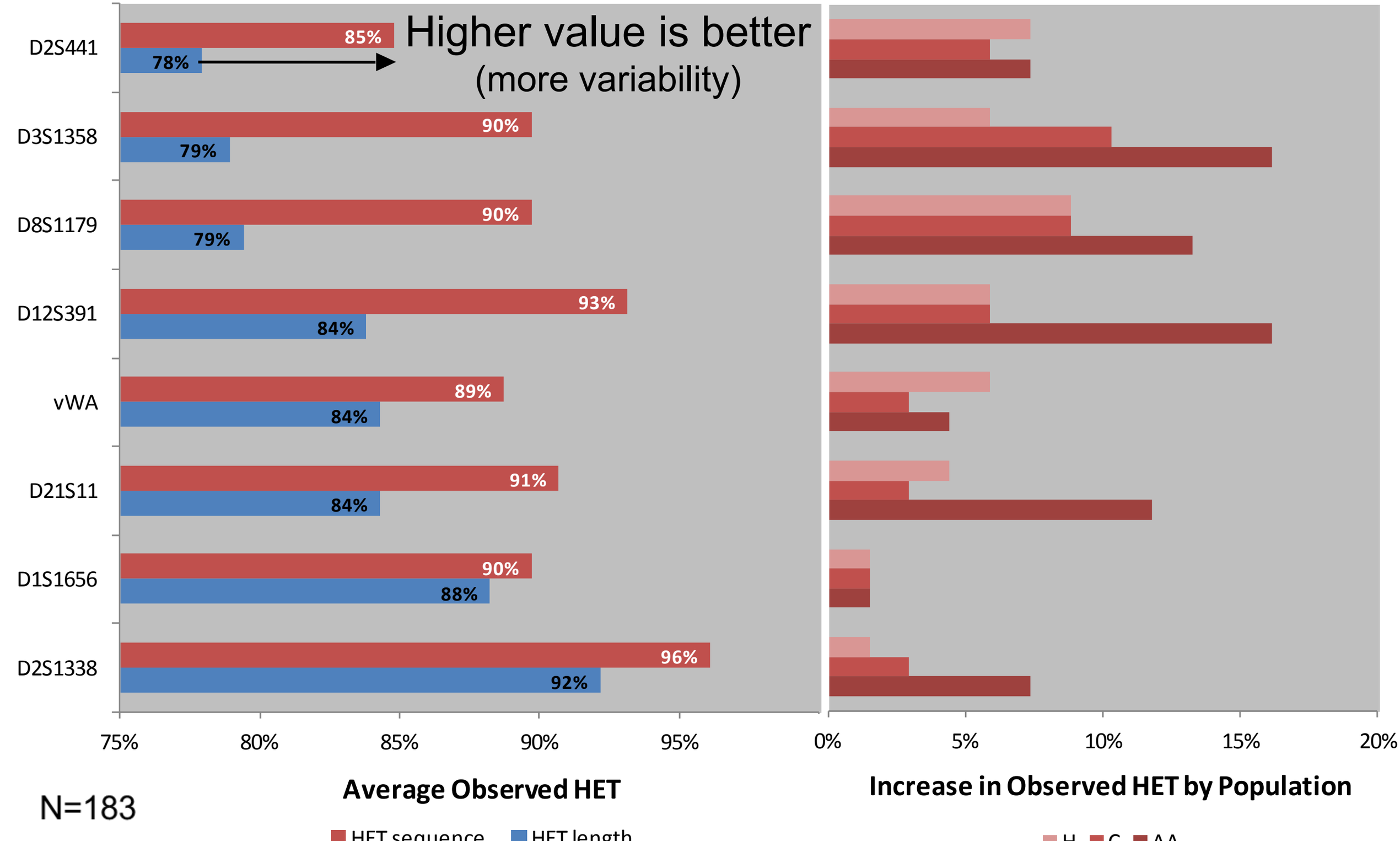
Forensic STR Sequence Diversity

Heterozygosity

$$\frac{\text{\# heterozygotes observed}}{\text{\# of loci tested}}$$

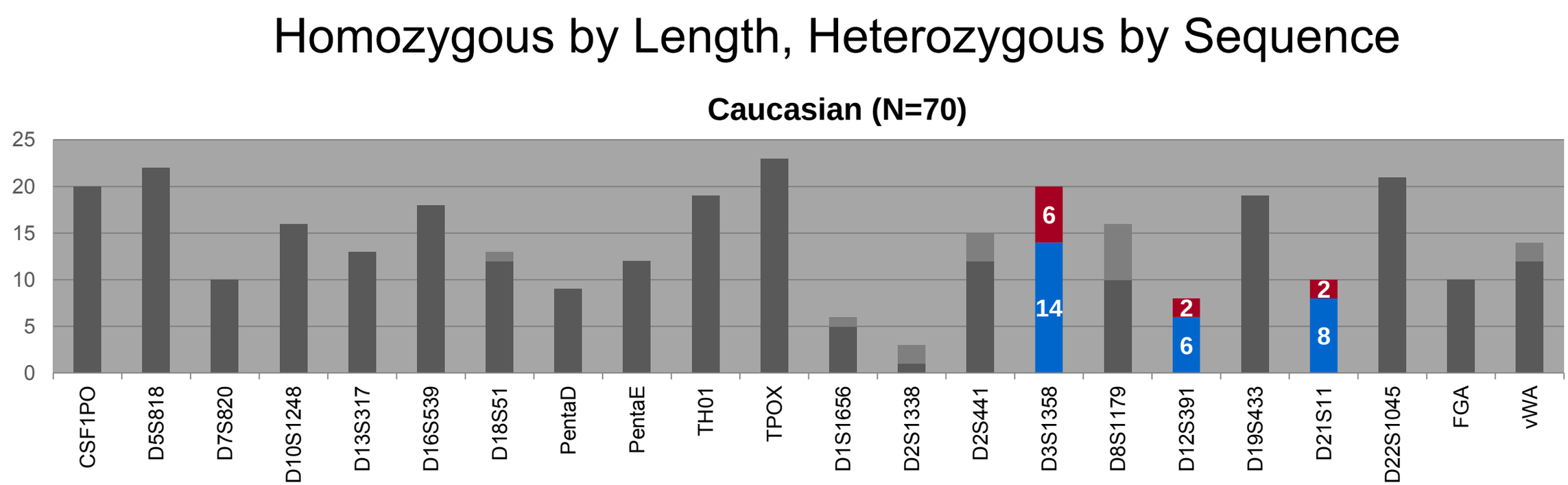
Indicates genetic variability at a locus

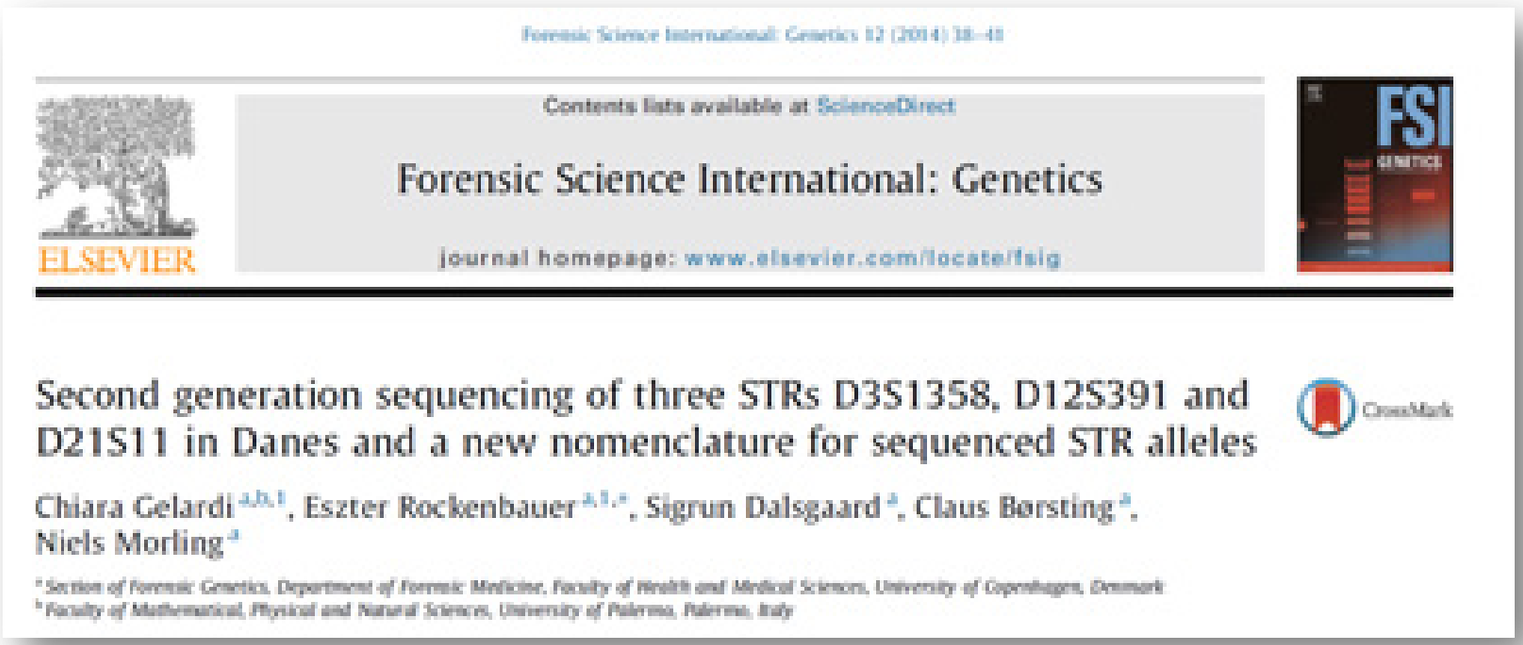
Forensic STR Sequence Diversity



N=183

Forensic STR Sequence Diversity





Forensic Science International: Genetics 12 (2014) 38–43

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Second generation sequencing of three STRs D3S1358, D12S391 and D21S11 in Danes and a new nomenclature for sequenced STR alleles

Chiara Gelardi[a,b,1], Eszter Rockenbauer[a,1,*], Sigrun Dalsgaard[a], Claus Børsting[a], Niels Morling[a]

[a] Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Denmark
[b] Faculty of Mathematical, Physical and Natural Sciences, University of Palermo, Palermo, Italy

Frequency of Sequence-Based Heterozygotes		
Locus	Gelardi (N=197)	NIST (N=70)
D3S1358	23%	30%
D12S391	33%	25%
D21S11	41%	20%

Forensic STR Sequence Diversity

Forensic Science International: Genetics 8 (2014) 20–23

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Characterising the STR locus D6S1043 and examination of its effect on stutter rates

Jo-Anne Bright [a,b,*], Kate E. Stevenson [a], Michael D. Coble [c], Carolyn R. Hill [c], James M. Curran [b], John S. Buckleton [a]

Allele	Repeat Structure
9	[AGAT]9
10	[AGAT]10
11	[AGAT]11
12	[AGAT]12
13	[AGAT]13
14	[AGAT]14
15	[AGAT]15
16	[AGAT]10[ACAT][AGAT]5
17	[AGAT]11[ACAT][AGAT]5
18	[AGAT]12[ACAT][AGAT]5
19	[AGAT]13[ACAT][AGAT]5
20	[AGAT]14[ACAT][AGAT]5
21	[AGAT]15[ACAT][AGAT]5

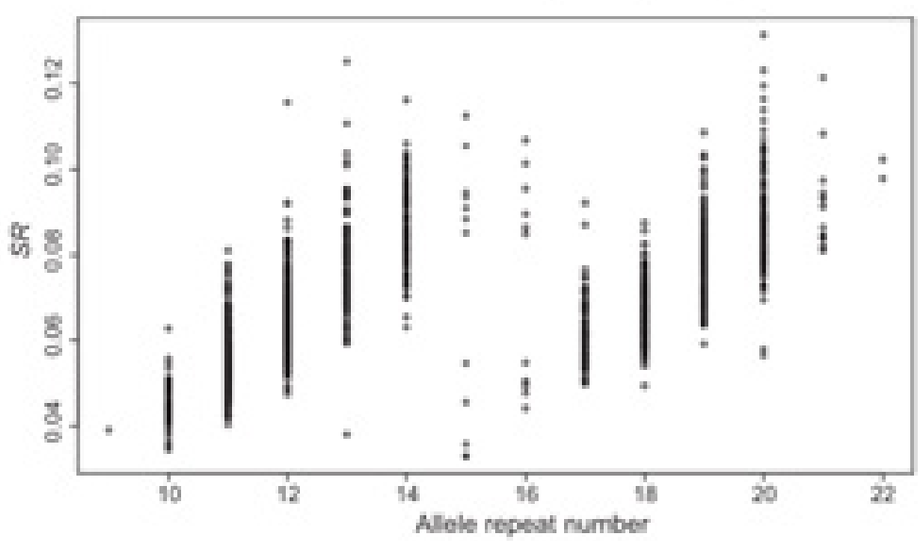


Fig. 1. A plot of *SR* versus allele repeat number.

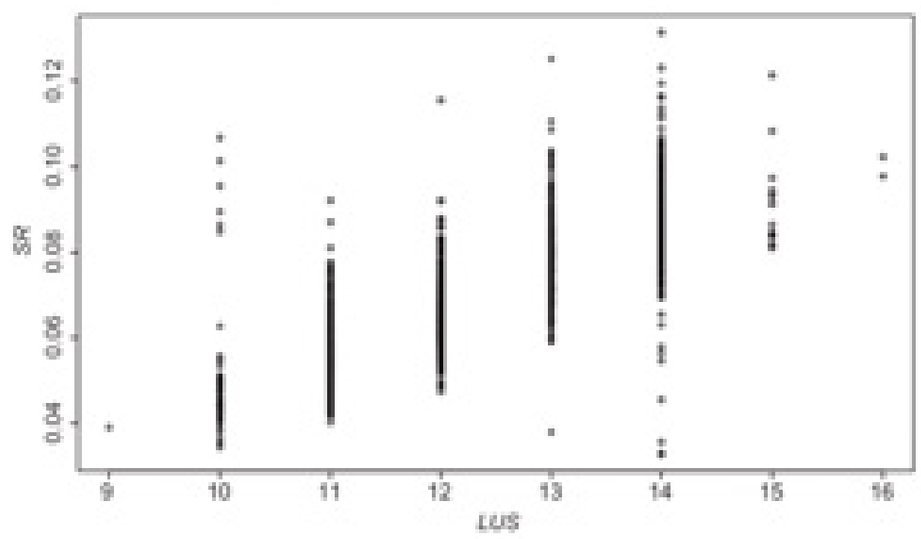


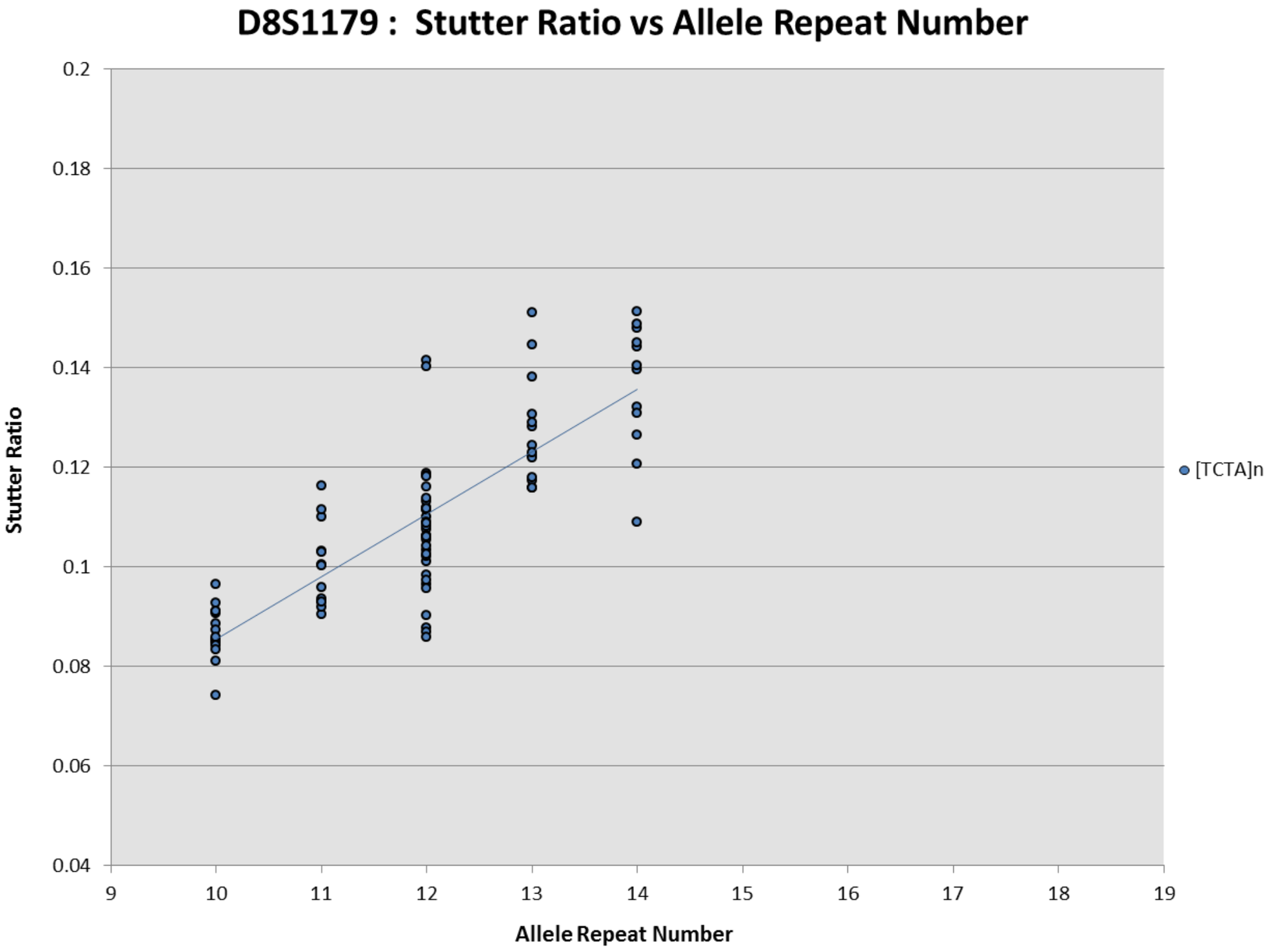
Fig. 2. A plot of *SR* versus *LUS*.

Forensic STR Sequence Diversity

D8S1179

Allele	Repeat Structure
	[TCTA]10-14
10	[TCTA]10
11	[TCTA]11
12	[TCTA]12
13	[TCTA]13
14	[TCTA]14

Forensic STR Sequence Diversity

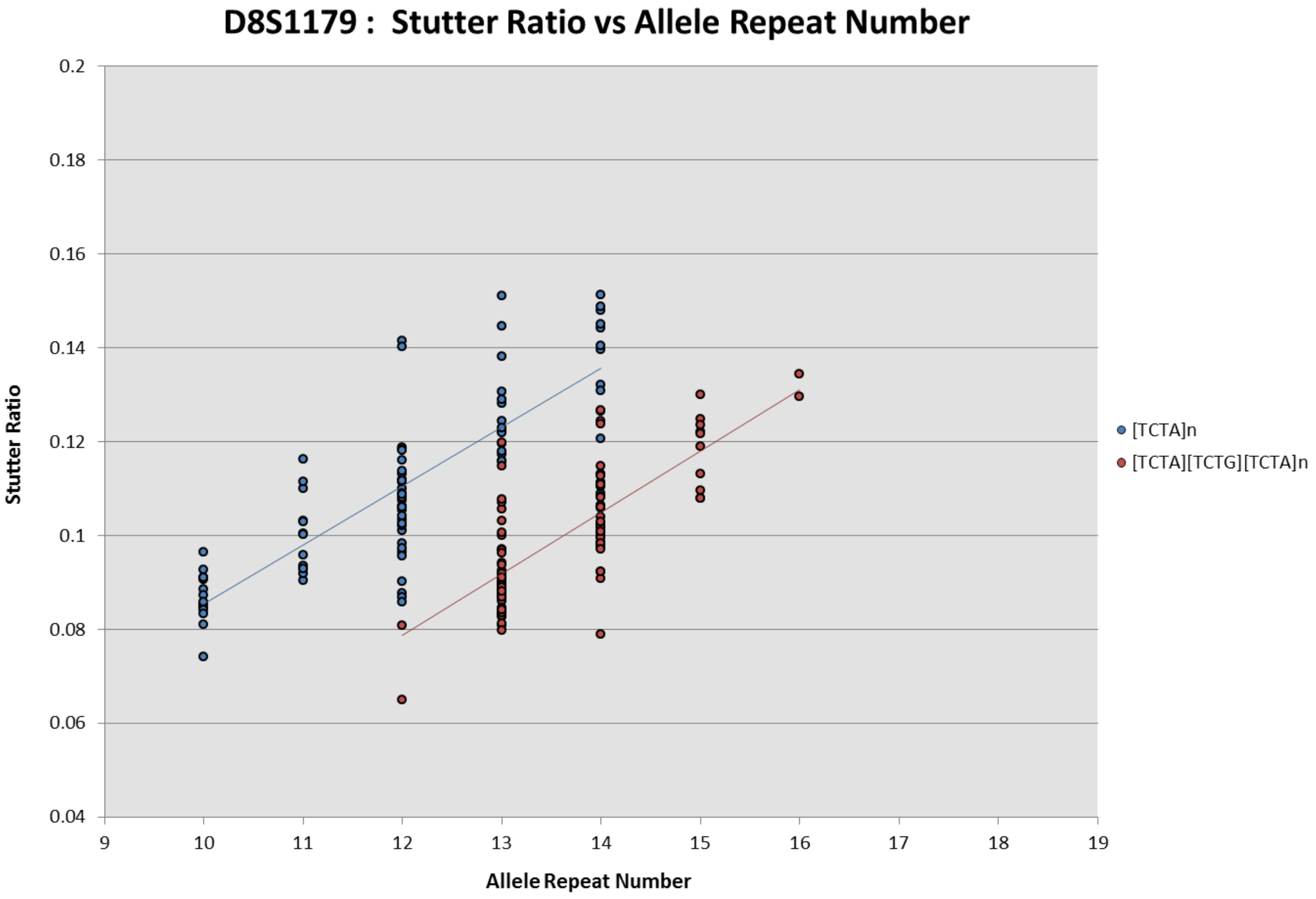


Forensic STR Sequence Diversity

D8S1179

Allele	Repeat Structure
	[TCTA]10-14
10	[TCTA]10
11	[TCTA]11
12	[TCTA]12
13	[TCTA]13
14	[TCTA]14
	[TCTA][TCTG][TCTA]10-14
12	[TCTA][TCTG][TCTA]10
13	[TCTA][TCTG][TCTA]11
14	[TCTA][TCTG][TCTA]12
16	[TCTA][TCTG][TCTA]14

Forensic STR Sequence Diversity

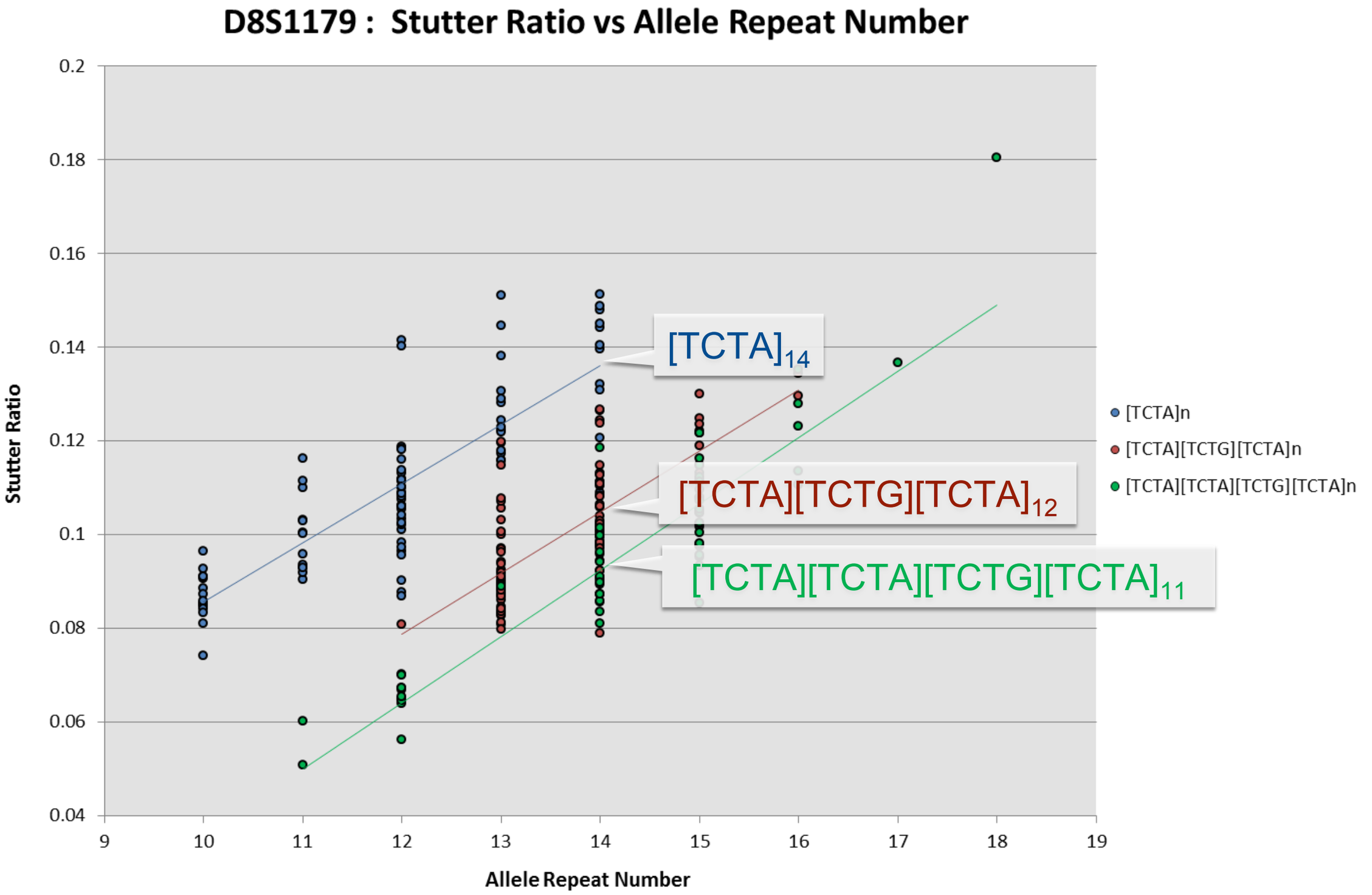


Forensic STR Sequence Diversity

D8S1179

Allele	Repeat Structure
	[TCTA]10-14
10	[TCTA]10
11	[TCTA]11
12	[TCTA]12
13	[TCTA]13
14	[TCTA]14
	[TCTA][TCTG][TCTA]10-14
12	[TCTA][TCTG][TCTA]10
13	[TCTA][TCTG][TCTA]11
14	[TCTA][TCTG][TCTA]12
16	[TCTA][TCTG][TCTA]14
	[TCTA][TCTA][TCTG][TCTA]8-15
11	[TCTA][TCTA][TCTG][TCTA]8
12	[TCTA][TCTA][TCTG][TCTA]9
13	[TCTA][TCTA][TCTG][TCTA]10
14	[TCTA][TCTA][TCTG][TCTA]11
15	[TCTA][TCTA][TCTG][TCTA]12
16	[TCTA][TCTA][TCTG][TCTA]13
17	[TCTA][TCTA][TCTG][TCTA]14
18	[TCTA][TCTA][TCTG][TCTA]15

Forensic STR Sequence Diversity

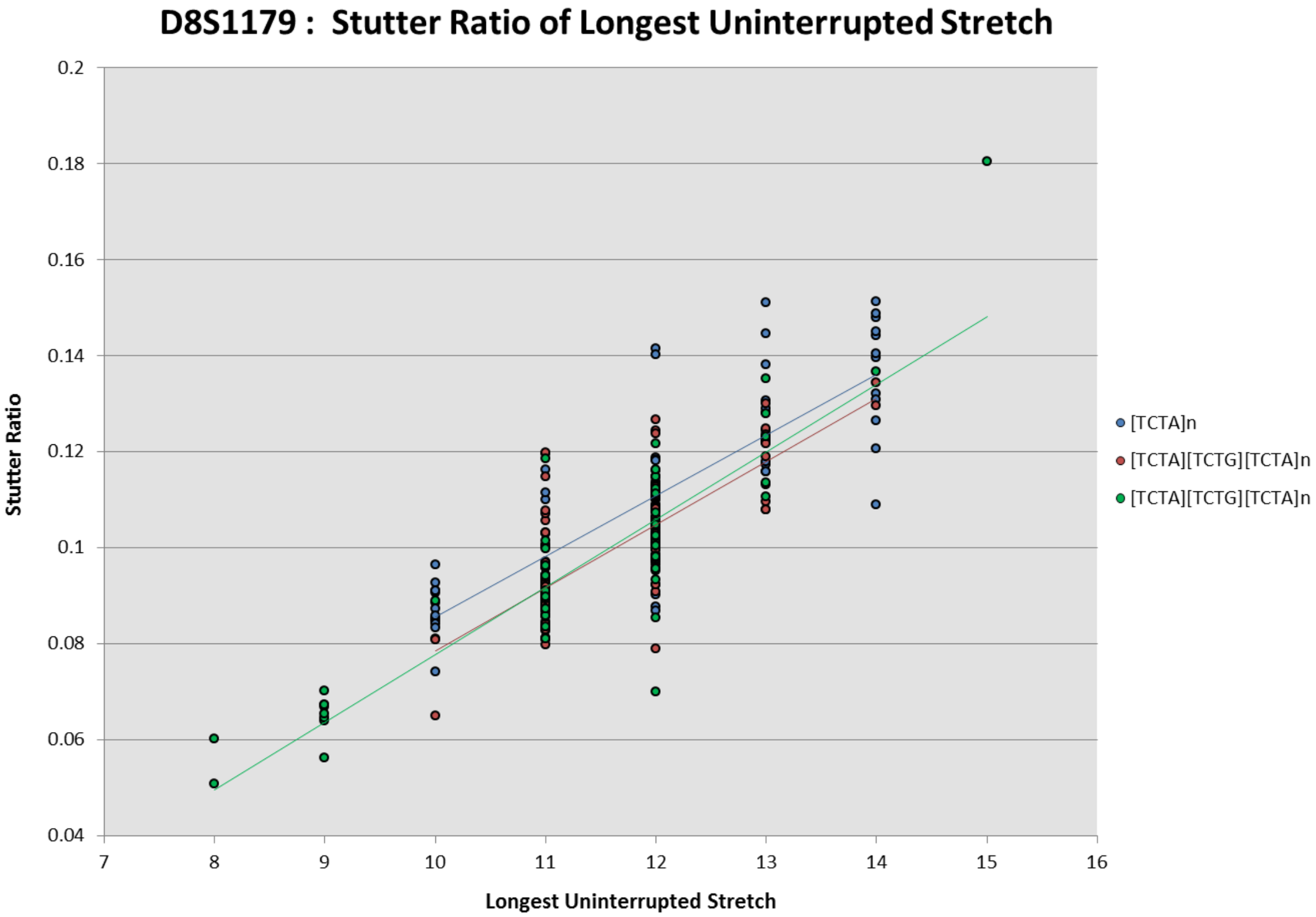


Forensic STR Sequence Diversity

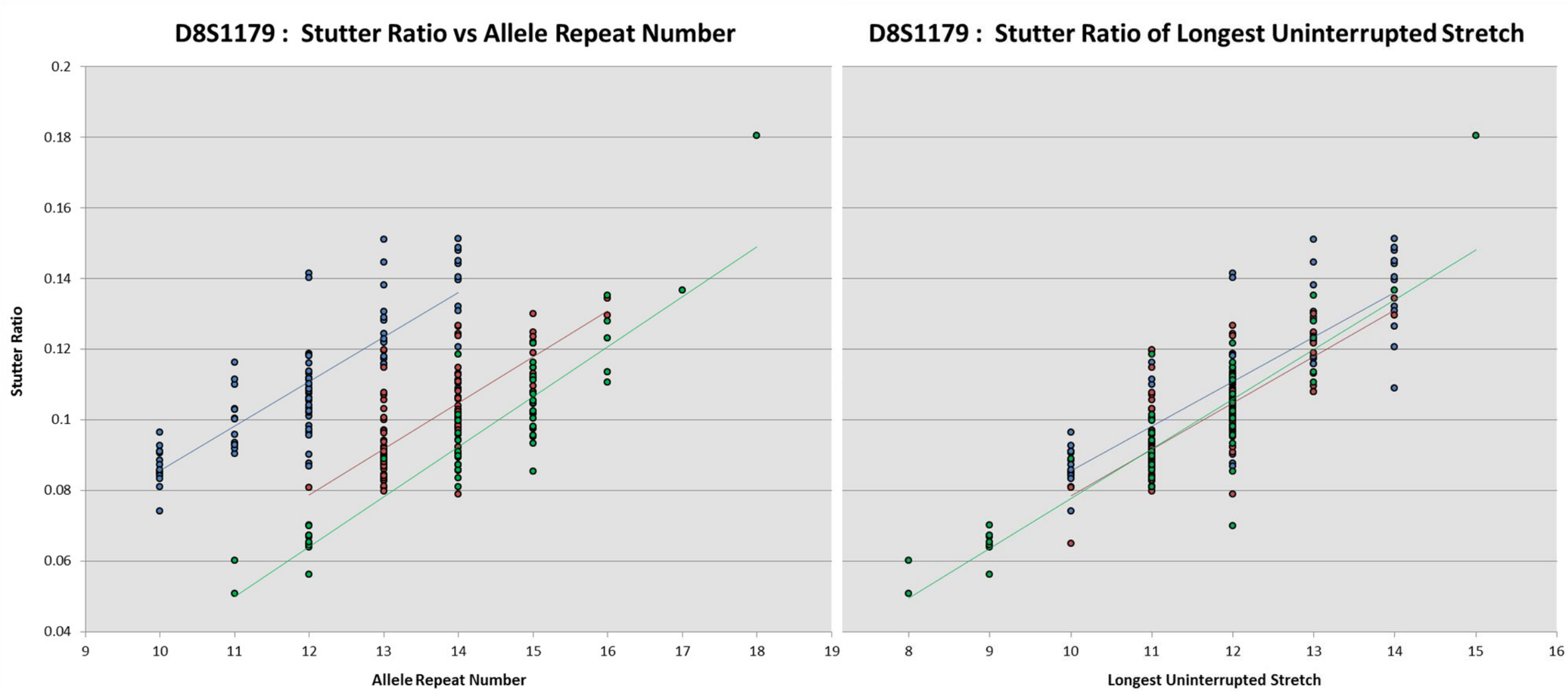
LONGEST UNINTERRUPTED STRETCH

Allele	Repeat Structure
	[TCTA]10-14
10	[TCTA]10
11	[TCTA]11
12	[TCTA]12
13	[TCTA]13
14	[TCTA]14
	[TCTA][TCTG][TCTA]10-14
12	[TCTA][TCTG][TCTA]10
13	[TCTA][TCTG][TCTA]11
14	[TCTA][TCTG][TCTA]12
16	[TCTA][TCTG][TCTA]14
	[TCTA][TCTA][TCTG][TCTA]8-15
11	[TCTA][TCTA][TCTG][TCTA]8
12	[TCTA][TCTA][TCTG][TCTA]9
13	[TCTA][TCTA][TCTG][TCTA]10
14	[TCTA][TCTA][TCTG][TCTA]11
15	[TCTA][TCTA][TCTG][TCTA]12
16	[TCTA][TCTA][TCTG][TCTA]13
17	[TCTA][TCTA][TCTG][TCTA]14
18	[TCTA][TCTA][TCTG][TCTA]15

Forensic STR Sequence Diversity



Forensic STR Sequence Diversity



Forensic STR Sequence Diversity

Conclusions

Sequencing forensic STR loci in a HTP manner is possible
(automation is needed)

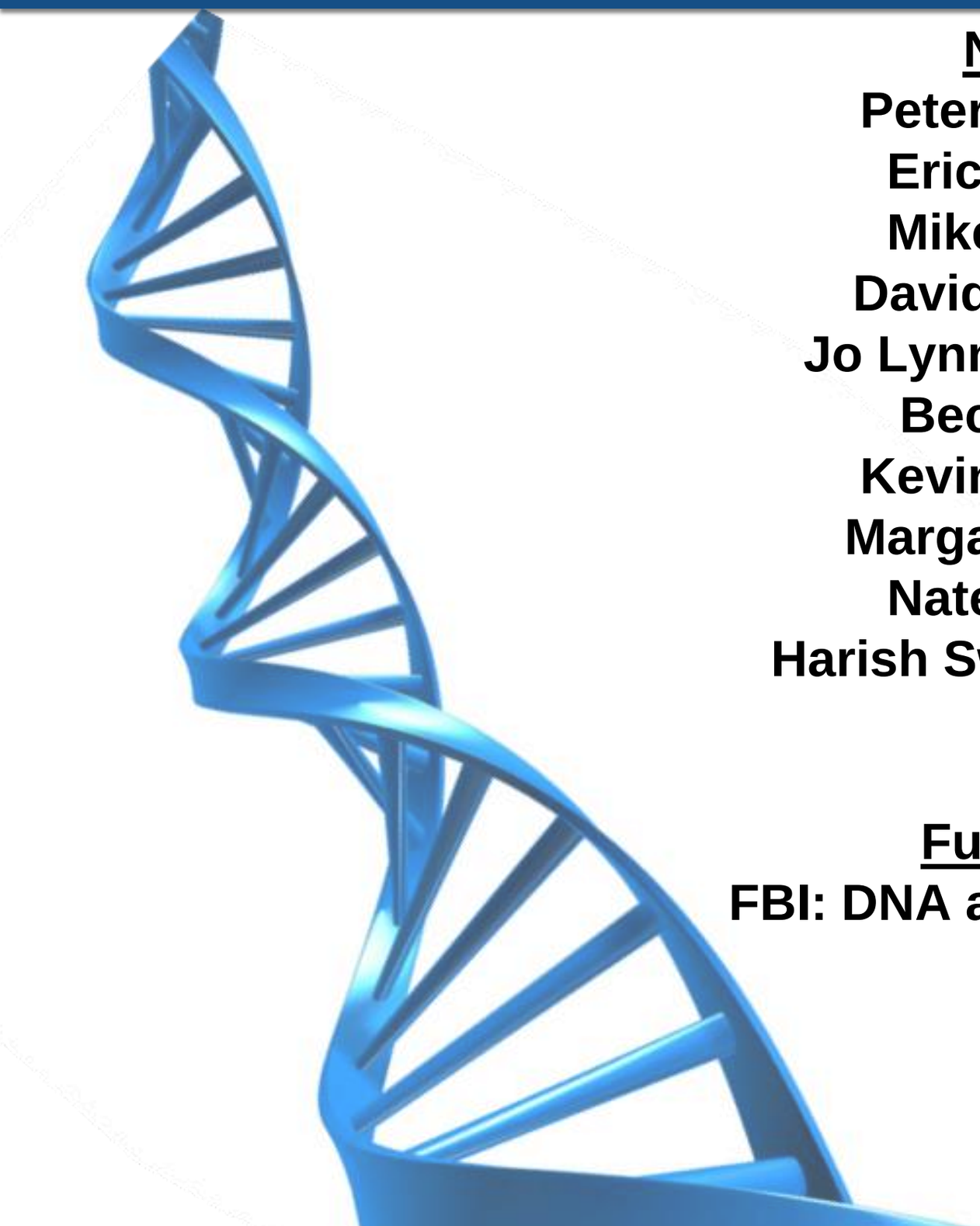
Bioinformatic tools are in their infancy,
testing across platforms and pipelines is important

At some loci, sequencing will offer significant gains
("core set" for mixture analysis)

Extending analysis to the flanking regions
will increase effective number of alleles

Allele frequency databases are needed prior to implementation
(150-200 per population → 5/2n)

Acknowledgements



NIST

Peter Vallone
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Margaret Kline
Nate Olson
Harish Swaminathan

Promega

Doug Storts
Jay Patel

Battelle

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FBI: DNA as a Biometric

Contact Information
katherine.gettings@nist.gov

University of Zurich[UZH]

Institute of Legal Medicine

EDNAP mRNA exercise 6 (skin)

Human specific RNA quantification

Cordula Haas, Erin Hanson, Jack Ballantyne

EDNAP meeting, 19. November 2014, Zurich

EDNAP mRNA exercise 6

Ms. Ref. No.: FSIGEN-D-14-00304
Title: RNA/DNA co-analysis from human skin and contact traces - results of a sixth collaborative EDNAP exercise
Forensic Science International: Genetics

Dear Cordula,

The reviewers have commented on your above paper and both have suggested a few minor modifications.
Please consider the comments below and address each one as usual.

I look forward to receiving your revised manuscript.

Yours sincerely,

Professor Adrian Linacre, Ph.D.
Associate Editor
Forensic Science International: Genetics

Few minor modifications: 4 A4 pages…

Last EDNAP meeting

→ Human specific mRNA quant assay (Ballantyne)

Other mRNA quant assay (Zubakov, Kayser)?

→ Suggestion for a collaborative exercise on mRNA quantification (EDNAP mRNA exercise 7) at next EDNAP meeting, Nov. 2014 in Zurich

mRNA profiling workflow

- RNA extraction
- DNase treatment (TURBO DNA-free kit)
- *Optional: total mRNA quantification*
- Reverse transcription (RT)
- body fluid specific PCR-multiplex
- Capillary electrophoresis

→ too little RNA into RT: no result
too much RNA into RT: cross contamination

Total mRNA quantification

- RiboGreen & Qubit (Fluorescence)
- RiboGreen & ELISA-Reader (Fluorescence)
- Bioanalyzer (Chip-Gelelectrophoresis)
- NanoDrop (Absorption A_{260})

Human specific mRNA quant assay - UCF

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

- ➢ human specific
- ➢ abundant in body fluids
- ➢ sensitive

Human specific mRNA quant assay – Zurich approach

RNA-Extraction:

Manual (Phenol-Chloroform) or Kit (Qiagen RNeasy Mini Kit)

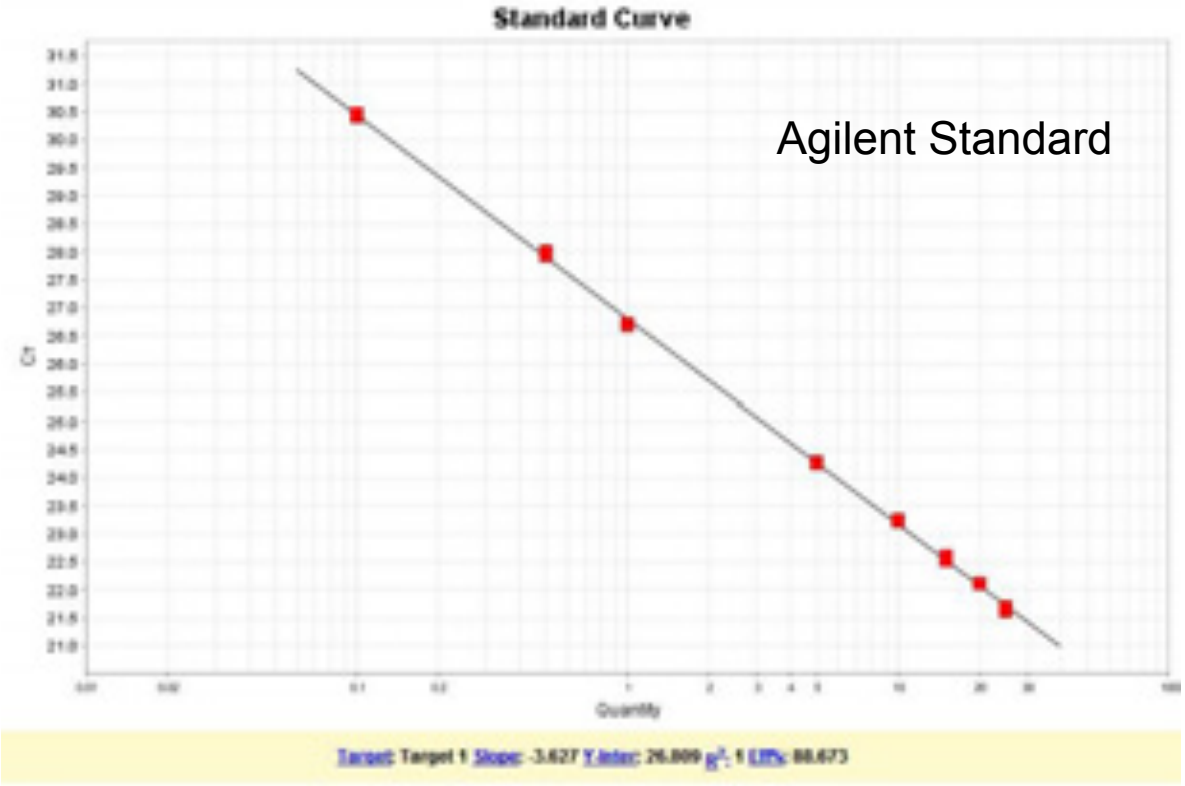
- **Manual:** Difference between RT+ und RT- (ΔC_t) is small
  → DNA-Contamination
- ✓ 2 x DNase digestion, different DNase-Buffer

- **Kit:** no issues with RT-

Human specific mRNA quant assay – Zurich approach

2 RNA/cDNA Standards (Agilent, Qiagen)

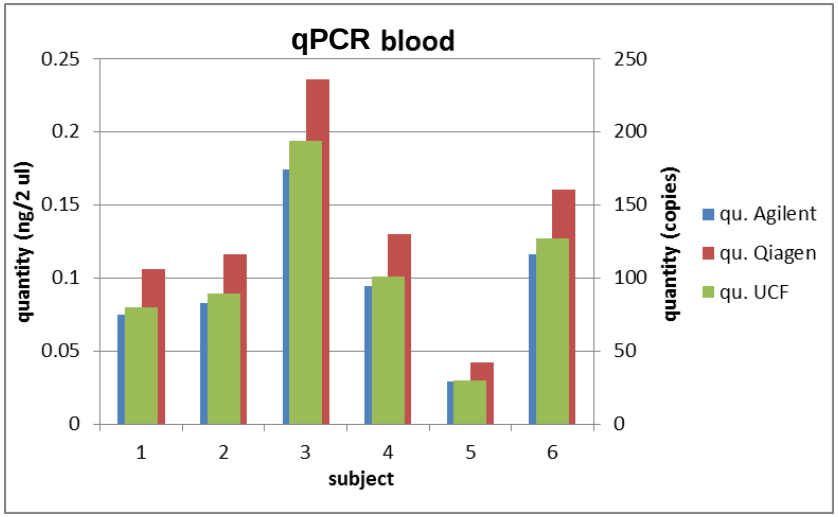
- Reference RNA (Cell lines (Agilent), Tissues (Qiagen))
  25 / 20 / 15 / 10 / 5 / 1 / 0.5 / 0.1 ng RNA into RT

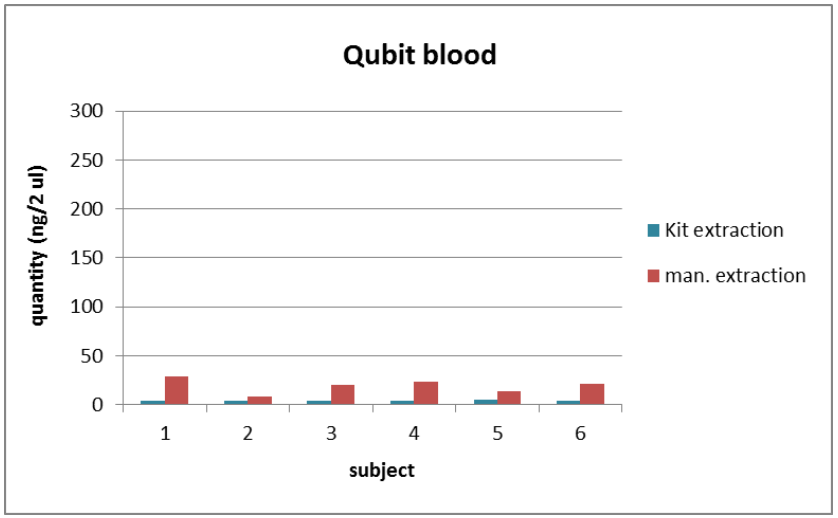


Human specific mRNA quant assay – Zurich approach

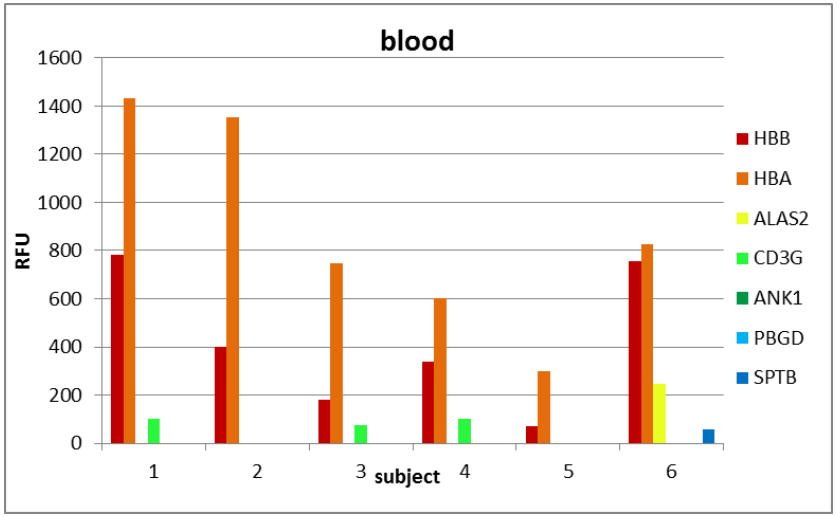
- 5 body fluids
- 6 donors for each body fluid
- RNA-Extraction Qiagen RNeasy Mini Kit and manual
- DNase digestion
- Total RNA quantification (Qubit)
- Reverse Transkription (2 µl and 25 ng)
- qPCR quantification
- End point-PCRs (Blood: 2plex & 5plex, Saliva: 3plex, Semen: 5plex, Vaginal secretion: 3plex, Menstrual blood: 3plex, HKG: 3plex)
- CE

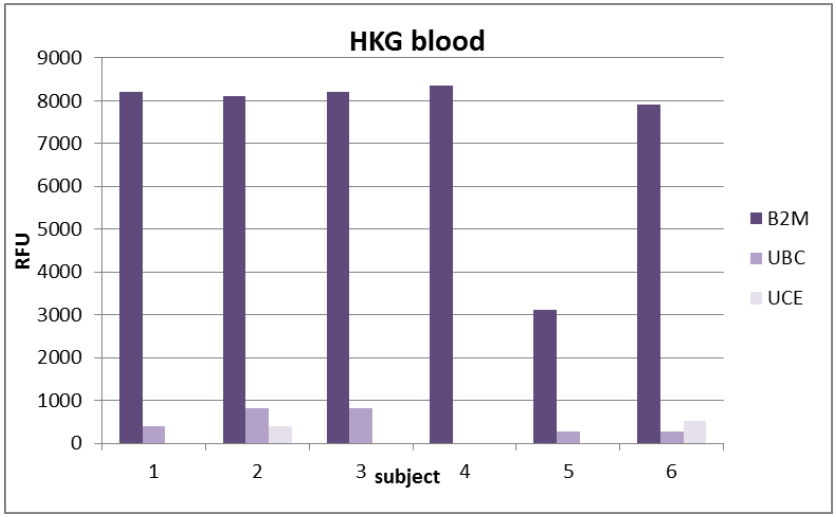
Quantification





Blood-specific marker expression





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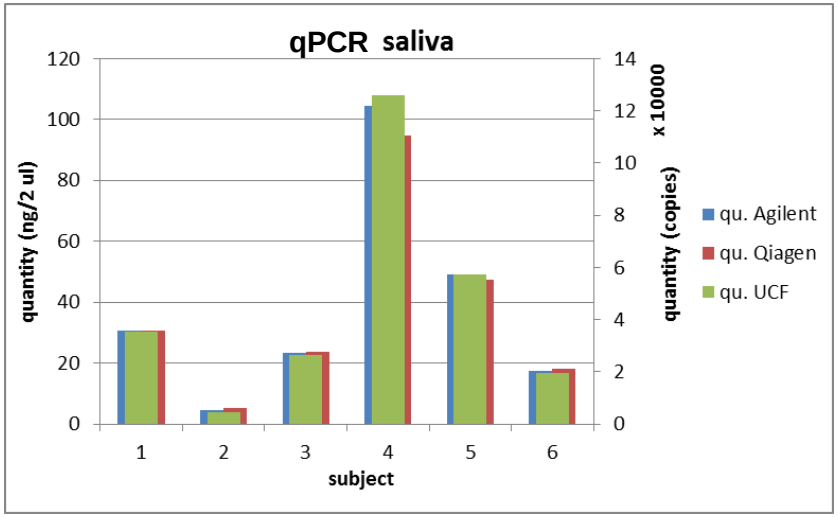
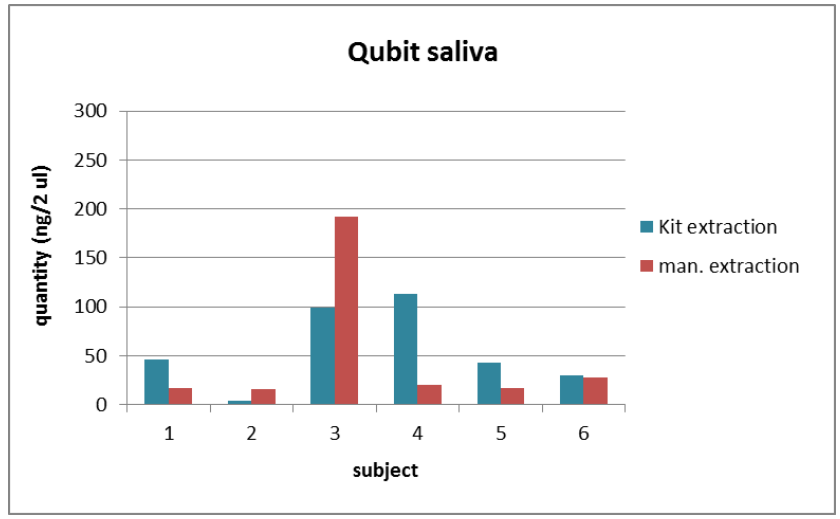
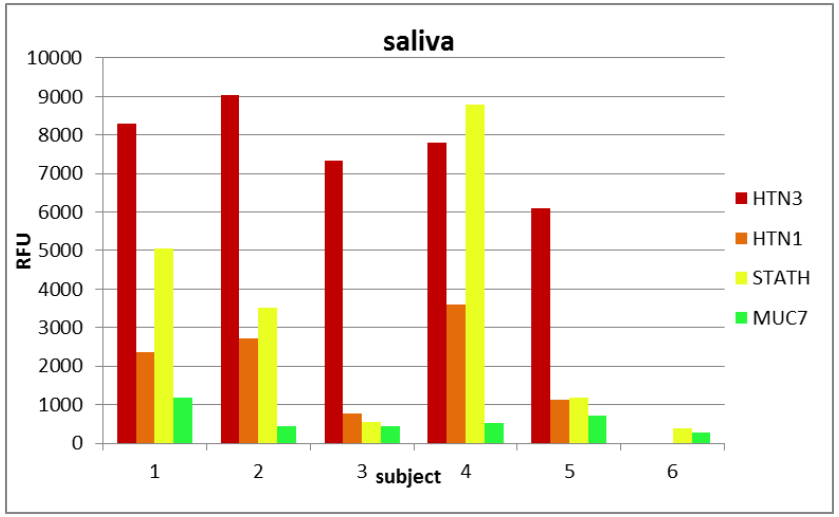
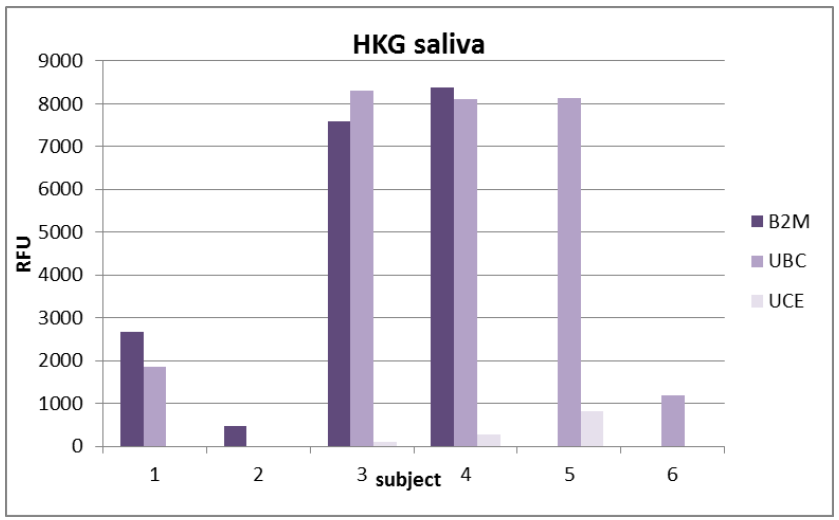




Saliva-specific marker expression





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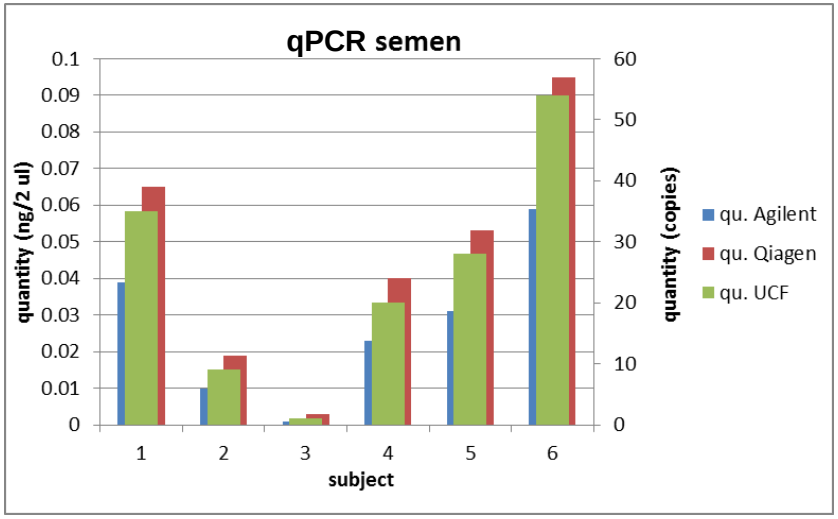
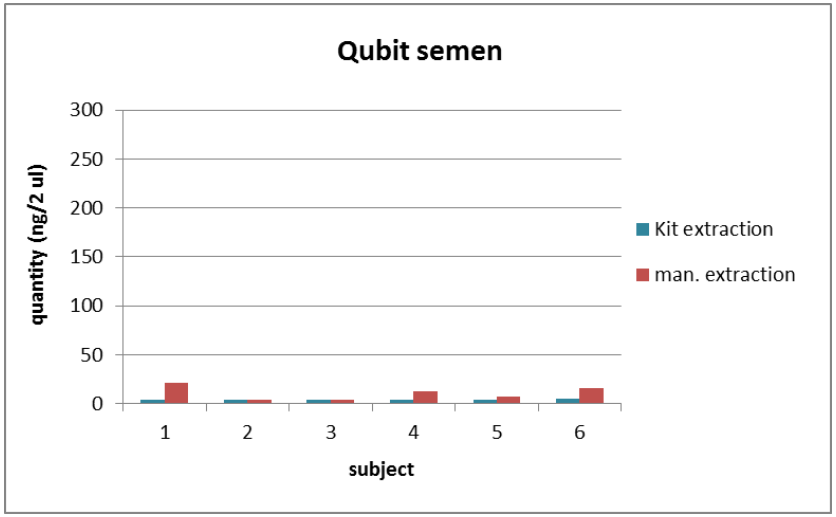
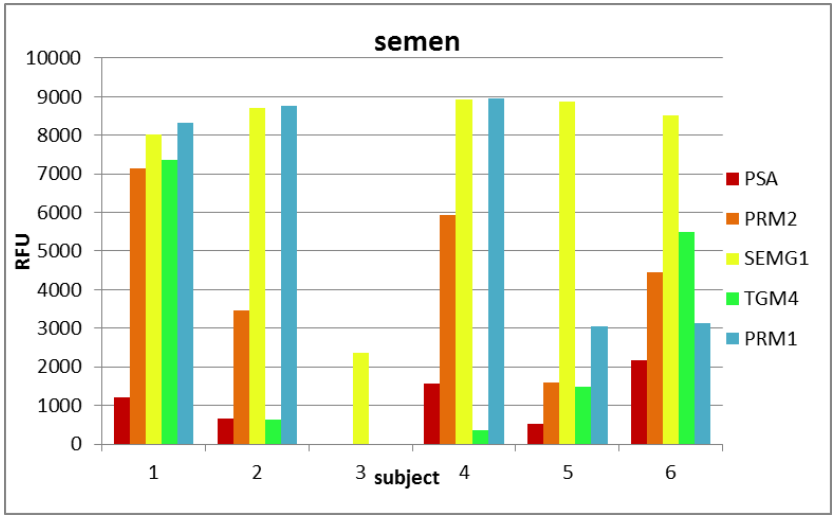
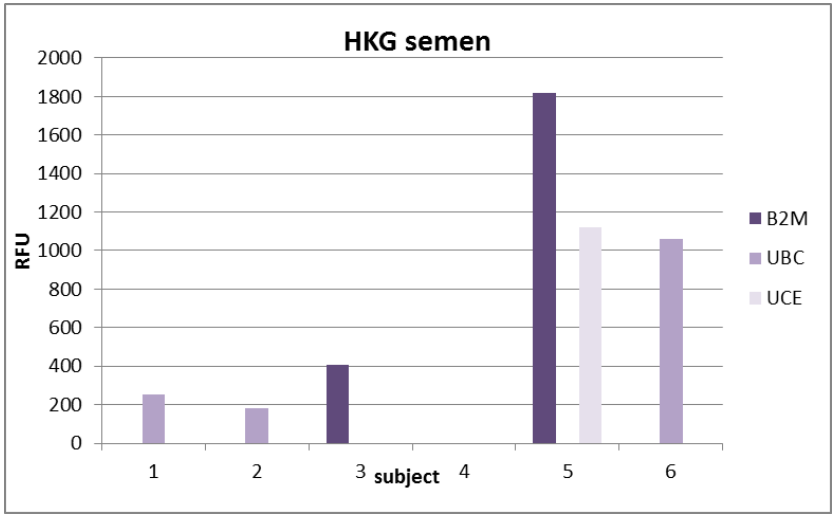




Semen-specific marker expression





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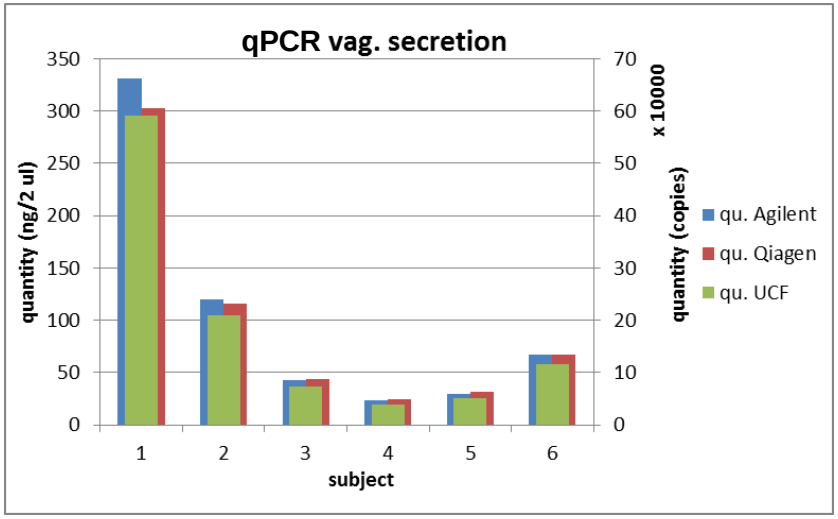
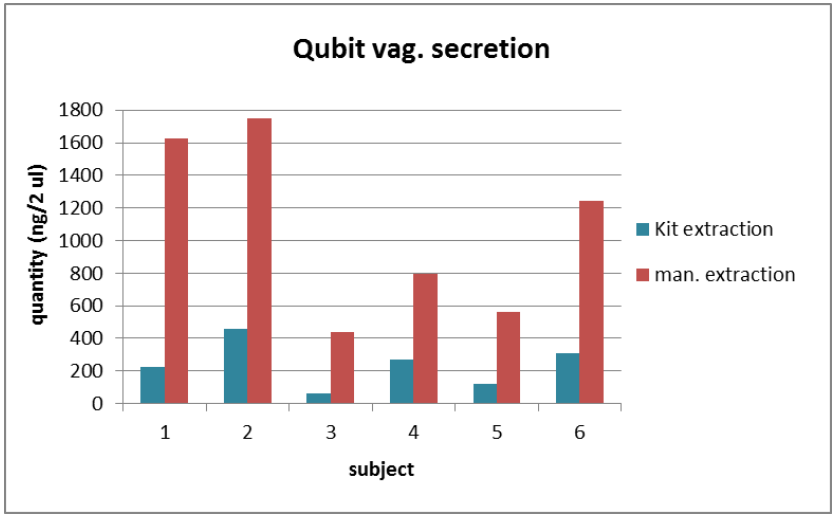
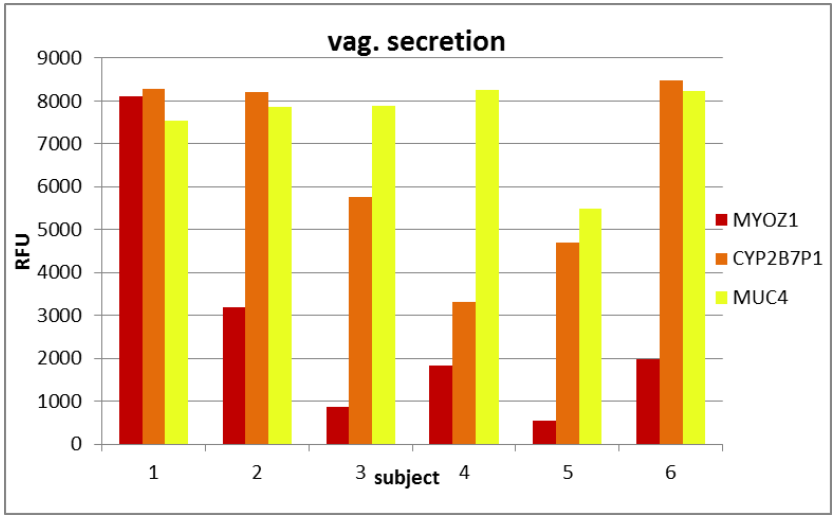
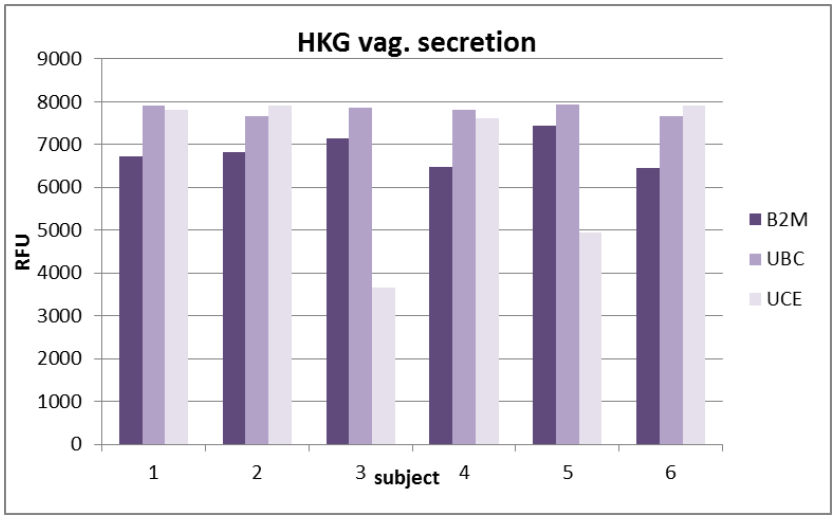




Vag-specific marker expression





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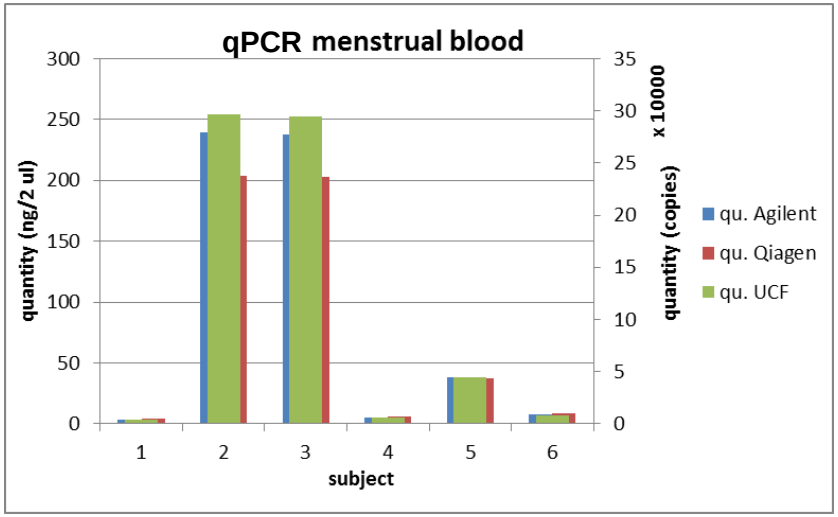
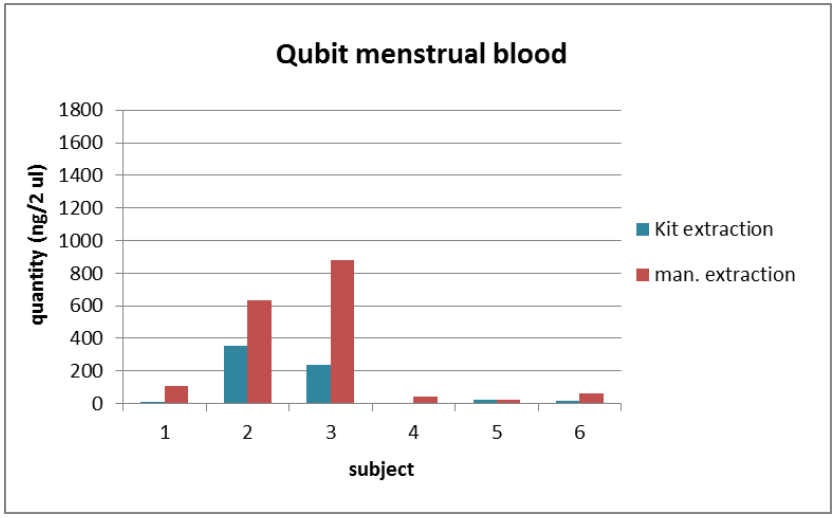
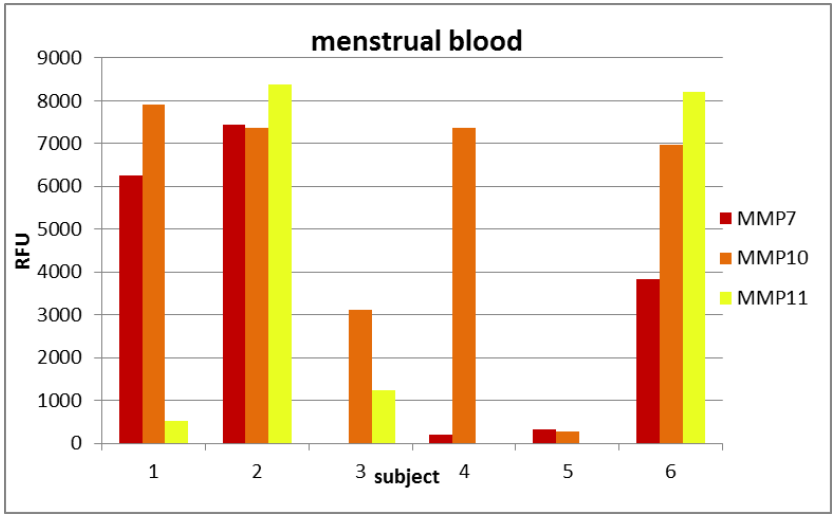
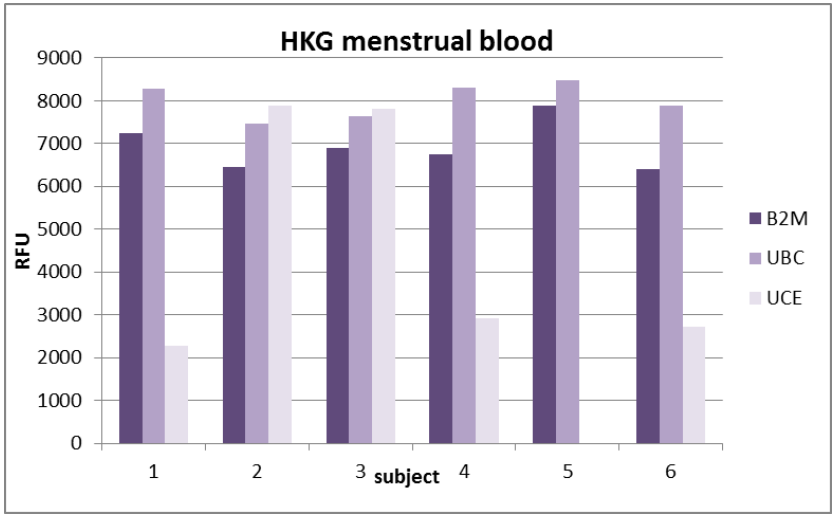




MB-specific marker expression





Human specific mRNA quant assay – Zurich approach

- 5 body fluids
- 6 donors for each body fluid
- RNA-Extraction Qiagen RNeasy Mini Kit and manual
- DNase digestion
- Reverse Transkription (2 µl and 25 ng)
- qPCR quantification → ng RNA
- optimal RNA input amount into RT
- End point-PCRs (Blood: 2plex & 5plex, Saliva: 3plex, Semen: 5plex, Vaginal secretion: 3plex, Menstrual blood: 3plex, HKG: 3plex)
- CE

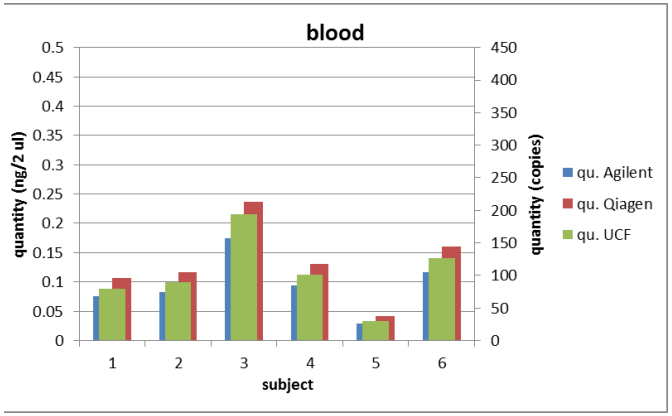
2 ul RNA into RT

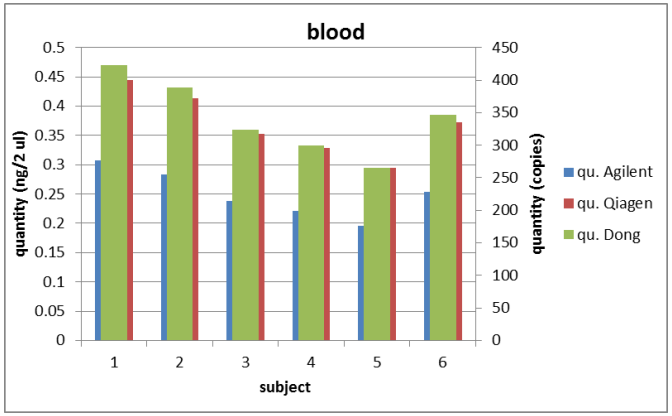
0.25 ng into RT

0.25 ng corresponds to

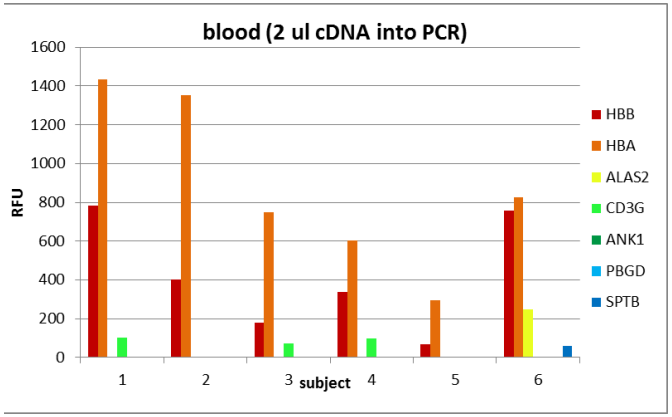
6.06 ul (1)	4.33 ul (4)
5.36 ul (2)	8.00 ul (5)
2.59 ul (3)	3.72 ul (6)

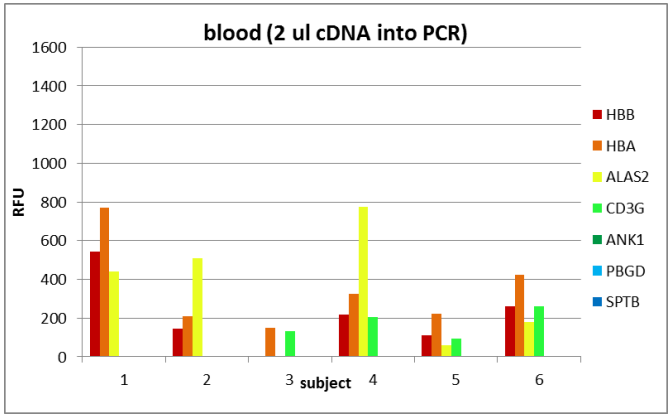
human-specific quantification





blood-specific marker expression





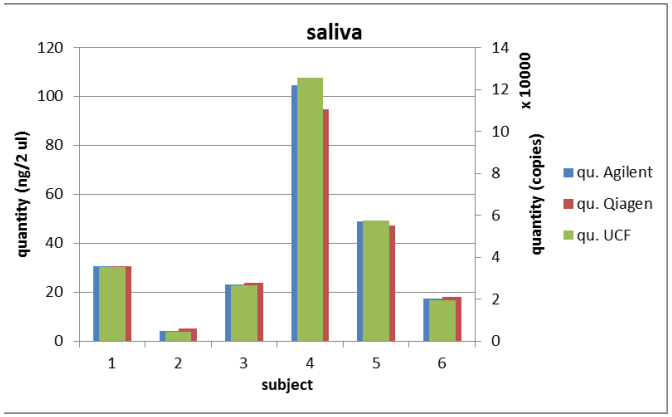
2 ul RNA into RT

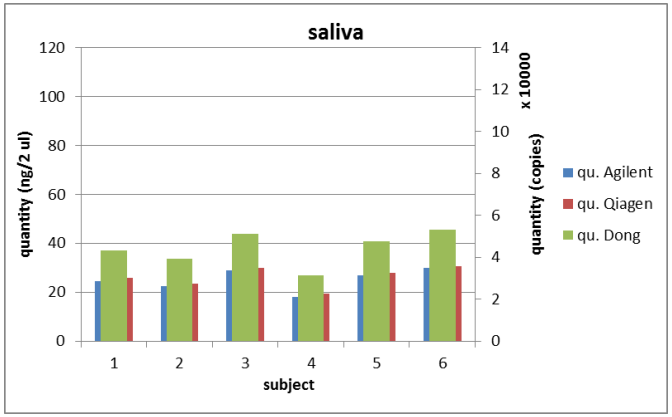
25 ng into RT

25 ng corresponds to

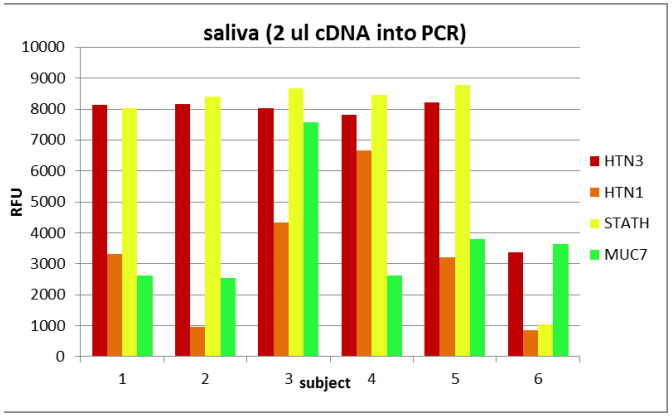
1.59 ul (1)	0.55 ul (4)
8.00 ul (2)	1.05 ul (5)
2.09 ul (3)	2.89 ul (6)

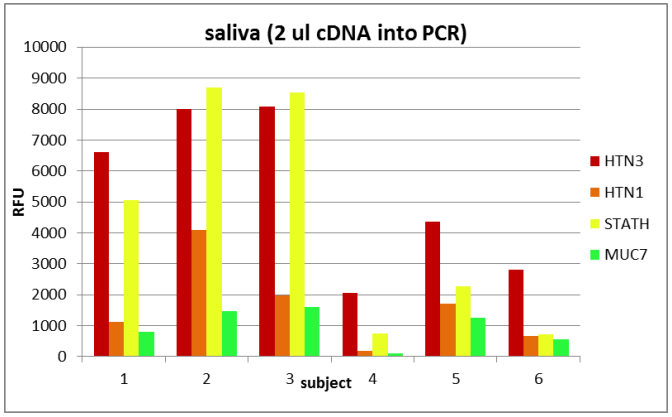
human-specific quantification





saliva-specific marker expression





2 ul RNA into RT

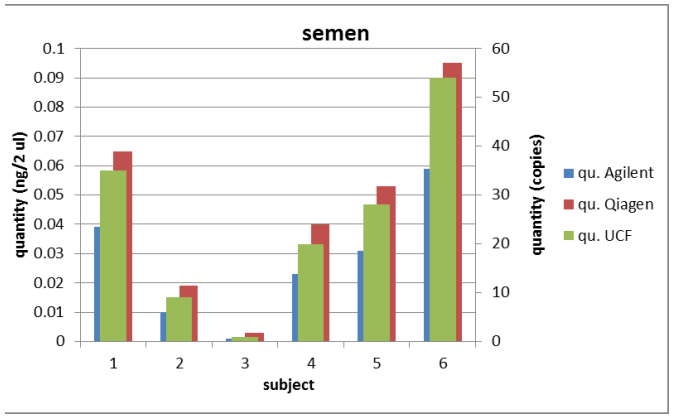
0.025 ng into RT

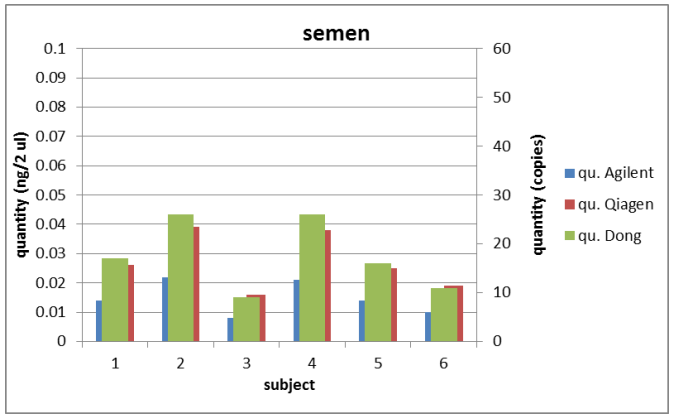
0.025 ng corresponds to

1.00 ul (1)	1.61 ul (4)
3.57 ul (2)	1.16 ul (5)
8.00 ul (3*)	0.68 ul (6)

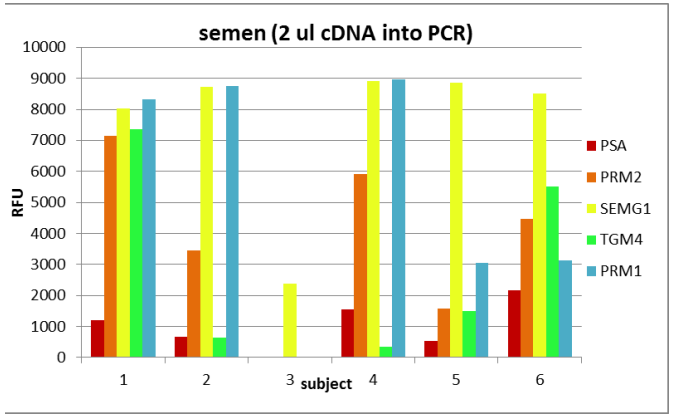
* azoospermic

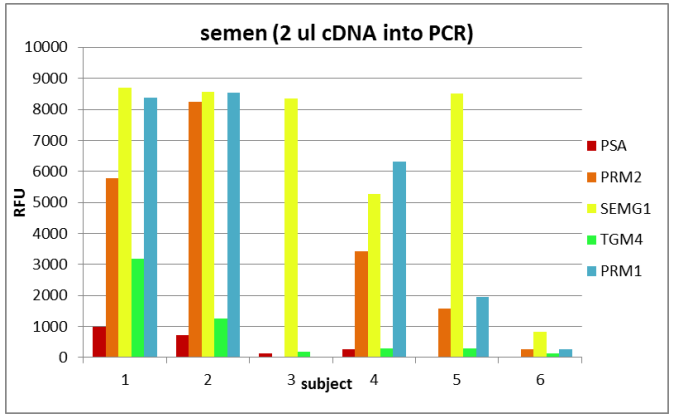
human-specific quantification





semen-specific marker expression





2 ul RNA into RT

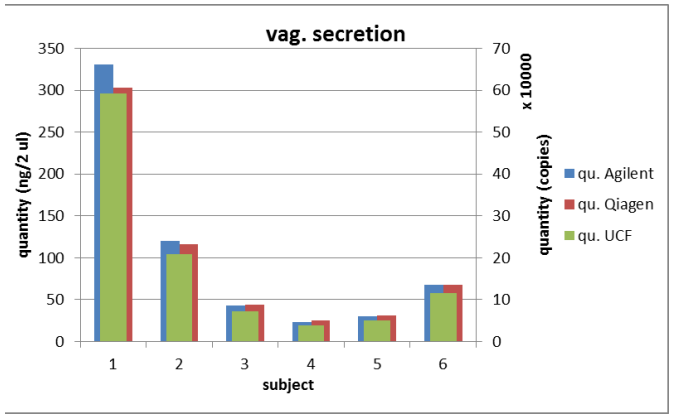
25 ng into RT

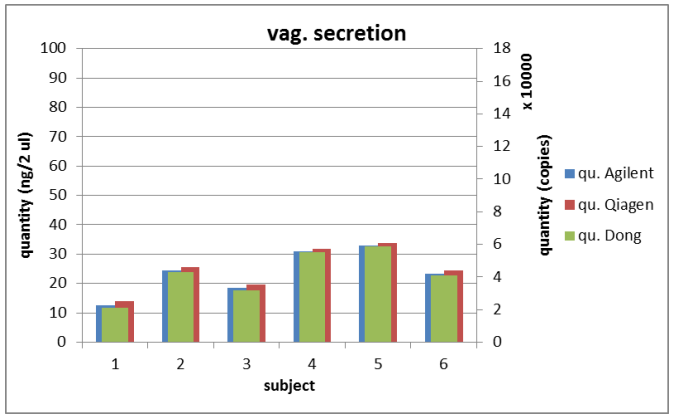
25 ng corresponds to

1.52* ul (1)	2.16 ul (4)
3.56* ul (2)	1.58 ul (5)
1.12 ul (3)	8.02* ul (6)

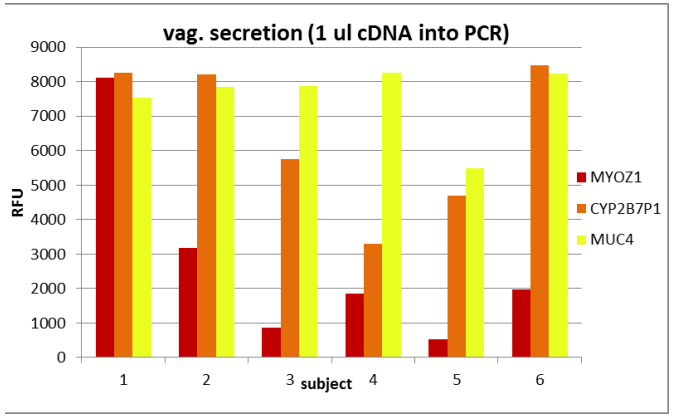
* 1:10 dilution

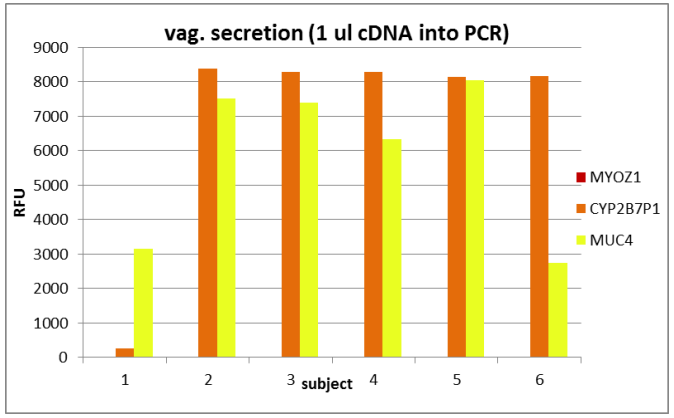
human-specific quantification





vag-specific marker expression





2 ul RNA into RT

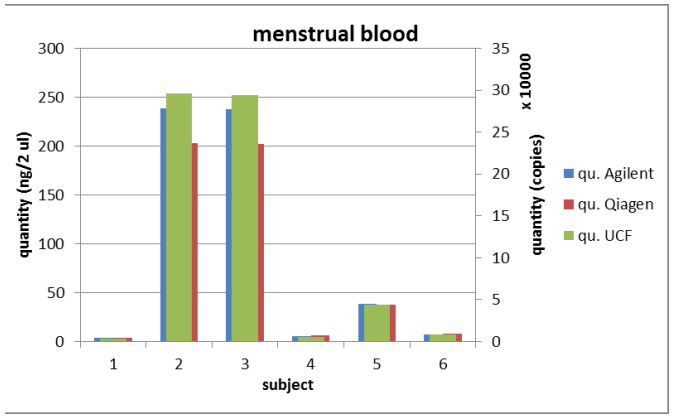
25 ng into RT

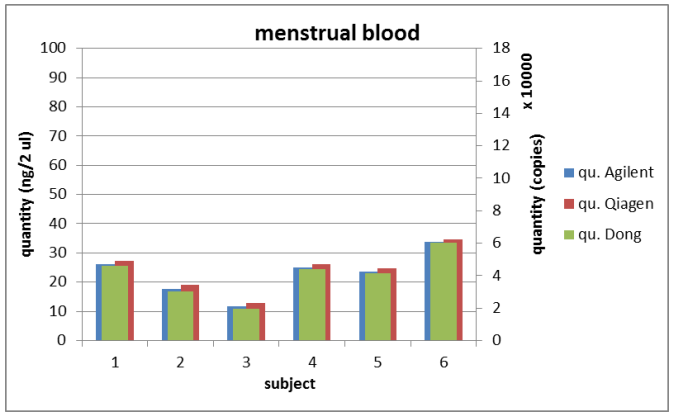
25 ng corresponds to

8.00 ul (1)	8.00 ul (4)
2.24* ul (2)	1.36 ul (5)
2.44* ul (3)	6.38 ul (6)

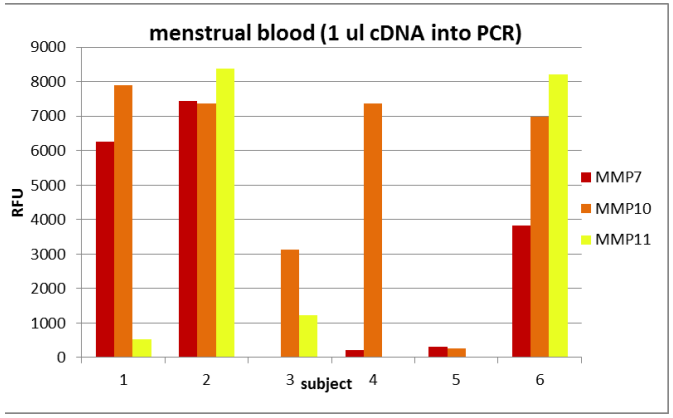
* 1:10 dilution

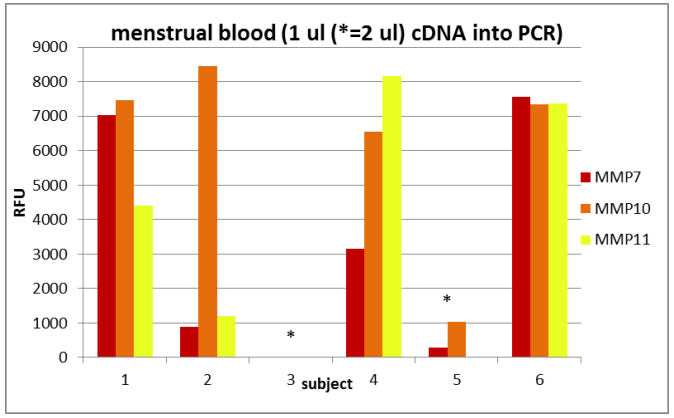
human-specific quantification





MB-specific marker expression





mRNA quantification – way forward?

- Correlation between RNA-concentration (copy numbers) and body fluid specific expression (peak height in RFU) only marginal
- Collaborative exercise?
- Test ‘no quant’ compared to ‘some sort of quant’ (however imperfect with respect to human specificity)?

Thank you for your attention!

Jack Ballantyne, Erin Hanson, Dong Zhao
Cordula Haas, Sabrina Ingold

EDNAP Meeting, Zürich, Switzerland, November 19, 2014

EMPOP Update

Walther Parson
Institute of Legal Medicine
Innsbruck Medical University
Austria

EMPOP update

1. New EMPOP related publications
2. Past meetings
3. EMPOP Trainings

1. New publications - 1

Forensic Science International: Genetics 13 (2014) 1–2

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Short Communication

Mitochondrial DNA control region analysis of three ethnic groups in the Republic of Macedonia

Renata Jankova-Ajanovska [a], Bettina Zimmermann [b], Gabriela Huber [b], Alexander W. Röck [b], Martin Bodner [b], Zlatko Jakovski [a], Biljana Janeska [a], Aleksej Duma [a], Walther Parson [b,c,*]

[a] Institute of Forensic Medicine, Criminalistic and Medical Deontology, Medical Faculty, University "Ss. Cyril and Methodius", Skopje, Macedonia
[b] Institute of Legal Medicine, Innsbruck Medical University, Innsbruck, Austria
[c] Penn State Eberly College of Science, University Park, PA, USA

1. New publications - 2

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]

[a] Institute of Legal Medicine, Innsbruck Medical University, Innsbruck, Austria
[b] Penn State Eberly College of Science, University Park, PA, USA
[c] DNA Diagnostic Laboratory (LDD), State University of Rio de Janeiro (UERJ), Brazil
[d] IPATIMUP, Institute of Molecular Pathology and Immunology of the University of Porto, Portugal
[e] FBI Laboratory, Quantico, VA, USA
[f] Division of Blood Group Serology, Medical University of Vienna, Austria
[g] Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark
[h] Department of Sciences, John Jay College for Criminal Justice, New York, NY, USA
[i] Unidade de Xenética, Departamento de AnatomíaPatolóxica e CienciasForenses, and Instituto de CienciasForenses, Grupo de MedicinaXenómica (GMX), Facultade de Medicina, Universidade de Santiago de Compostela, 15872 Galicia, Spain
[j] Institute of Legal Medicine, Medical Faculty, University of Cologne, Cologne, Germany
[k] International Commission on Missing Persons, Alipasina 45a, 71000 Sarajevo, Bosnia and Herzegovina

1. New publications - 3

Forensic Science International: Genetics 10 (2014) 73–79

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Short Communication

Development of forensic-quality full mtGenome haplotypes: Success rates with low template specimens☆

Rebecca S. Just [a,b,c,*], Melissa K. Scheible [a,b], Spence A. Fast [a,b], Kimberly Sturk-Andreaggi [a,b], Jennifer L. Higginbotham [a,b], Elizabeth A. Lyons [a,b,1], Jocelyn M. Bush [a,b], Michelle A. Peck [a,b], Joseph D. Ring [a,b], Toni M. Diegoli [a,b], Alexander W. Röck [d], Gabriela E. Huber [d], Simone Nagl [d], Christina Strobl [d], Bettina Zimmermann [d], Walther Parson [d,e], Jodi A. Irwin [a,b,2]

[a] Armed Forces DNA Identification Laboratory, 115 Purple Heart Dr., Dover AFB, DE 19902, United States
[b] American Registry of Pathology, 120A Old Camden Rd., Camden, DE 19934, United States
[c] University of Maryland, College Park, 8082 Baltimore Ave., College Park, MD 20740, United States
[d] Institute of Legal Medicine, Innsbruck Medical University, Müllerstrasse 44, Innsbruck, Austria
[e] Penn State Eberly College of Science, 517 Thomas Building, University Park, PA 16802, United States

1. New publications - 4

Forensic Science International: Genetics 14 (2015) 141–155

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

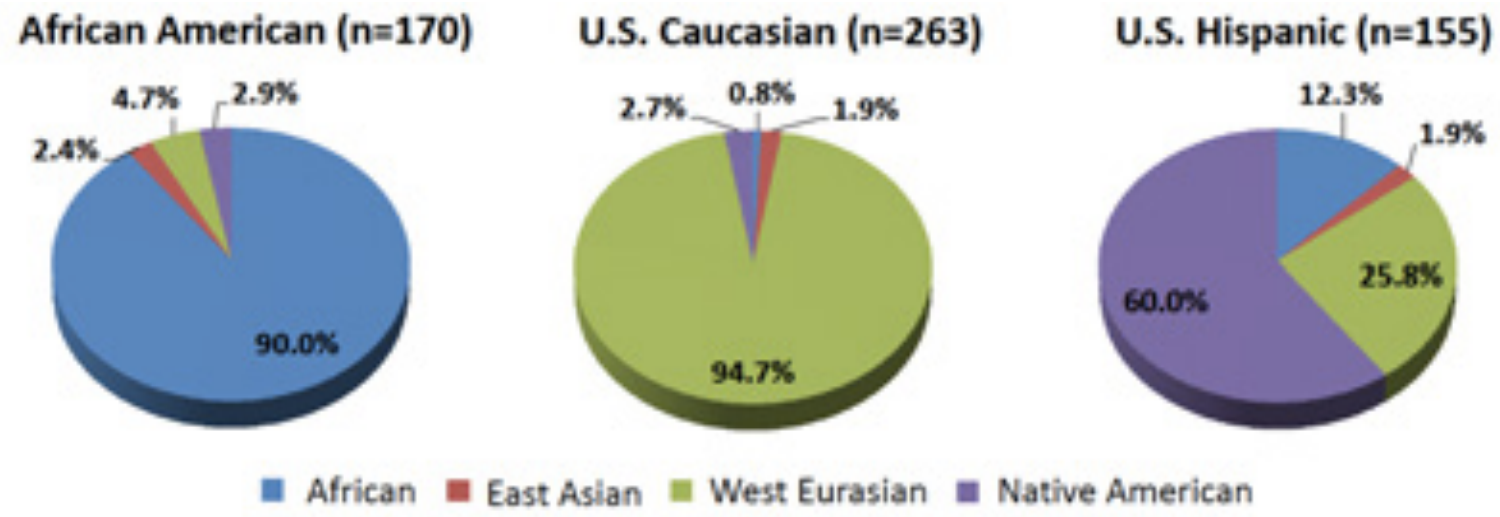
Forensic Population Genetics – Original Research

Full mtGenome reference data: Development and characterization of 588 forensic-quality haplotypes representing three U.S. populations

Rebecca S. Just [a,b,c,*], Melissa K. Scheible [a,b], Spence A. Fast [a,b], Kimberly Sturk-Andreaggi [a,b], Alexander W. Röck [d], Jocelyn M. Bush [a,b], Jennifer L. Higginbotham [a,b], Michelle A. Peck [a,b], Joseph D. Ring [a,b], Gabriela E. Huber [d], Catarina Xavier [d], Christina Strobl [d], Elizabeth A. Lyons [a,b,1], Toni M. Diegoli [a,b], Martin Bodner [d], Liane Fendt [d,2], Petra Kralj [d], Simone Nagl [d], Daniela Niederwieser [d], Bettina Zimmermann [d], Walther Parson [d,e], Jodi A. Irwin [a,b,3]

[a] Armed Forces DNA Identification Laboratory, 115 Purple Heart Dr., Dover AFB, DE 19902, United States
[b] American Registry of Pathology, 15245 Shady Grove Rd, Suite 335, Rockville, MD 20850, United States
[c] University of Maryland, College Park, 8082 Baltimore Ave., College Park, MD 20740, United States
[d] Institute of Legal Medicine, Innsbruck Medical University, Müllerstrasse 44, Innsbruck, Austria
[e] Penn State Eberly College of Science, 517 Thomas Building, University Park, PA 16802, United States

1. New publications - 4



African American (n=170)					Percentage Increase		
	HV1	HV1/HV2	CR	mtG	HV1 to HV1/HV2	HV1/HV2 to CR	CR to mtG
# Haplotypes	124	140	148	169	12,9%	5,7%	14,2%
# Unique Haplotypes	106	120	130	168	13,2%	8,3%	29,2%
Power of Discrimination	99,20%	99,67%	99,81%	99,99%			
U.S. Caucasian (n=263)					Percentage Increase		
	HV1	HV1/HV2	CR	mtG	HV1 to HV1/HV2	HV1/HV2 to CR	CR to mtG
# Haplotypes	151	200	229	259	32,5%	14,5%	13,1%
# Unique Haplotypes	122	170	211	255	39,3%	24,1%	20,9%
Power of Discrimination	97,62%	99,42%	99,78%	99,99%			
U.S. Hispanic (n=155)					Percentage Increase		
	HV1	HV1/HV2	CR	mtG	HV1 to HV1/HV2	HV1/HV2 to CR	CR to mtG
# Haplotypes	119	134	141	147	12,6%	5,2%	4,3%
# Unique Haplotypes	102	121	130	140	18,6%	7,4%	7,7%
Power of Discrimination	99,37%	99,74%	99,86%	99,92%			

Just et al 2014

1. New publications - 5

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Helena, the hidden beauty: Resolving the most common West Eurasian mtDNA control region haplotype by massively parallel sequencing an Italian population sample

Martin Bodner [a], Alessandra Iuvaro [a,b], Christina Strobl [a], Simone Nagl [a], Gabriela Huber [a], Susi Pelotti [b], Davide Pettener [c], Donata Luiselli [c,*], Walther Parson [a,d,**]

[a] Institute of Legal Medicine, Innsbruck Medical University, Innsbruck, Austria
[b] Department of Medical and Surgical Sciences, Institute of Legal Medicine, University of Bologna, Bologna, Italy
[c] Department of Biological, Geological and Environmental Science, Laboratory of Molecular Anthropology, University of Bologna, Bologna, Italy
[d] Penn State Eberly College of Science, University Park, PA, USA

1. New publications - 5

Table 1
Comparison of the diversity parameters in the 29 Italian samples using different sequence ranges.

	mtDNA range		
	CR	CR+39 codR SNPs[a]	Complete mtGenome
Haplotypes	1	6	28
Unique haplotypes	0	2	27
Haplogroups[b]	1	6	20
Unique haplogroups[b]	0	2	18
RMP[c]	1.000	0.296	0.037
Haplotype diversity	0.0%	72.9%	99.8%

[a] Thereof 17 specific for haplogroup H clades [44].
[b] According to Ref. [15], build 16. H* is considered a haplogroup.
[c] Random match probability.

1. New publications - 6

Forensic Science International: Genetics 12 (2014) 128–135

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

High-quality and high-throughput massively parallel sequencing of the human mitochondrial genome using the Illumina MiSeq

Jonathan L. King [a,1,*], Bobby L. LaRue [a,1], Nicole M. Novroski [a], Monika Stoljarova [a], Seung Bum Seo [a], Xiangpei Zeng [a], David H. Warshauer [a], Carey P. Davis [a], Walther Parson [b,c], Antti Sajantila [a,d], Bruce Budowle [a,e]

[a] Institute of Applied Genetics, Department of Molecular and Medical Genetics, University of North Texas Health Science Center, 3500 Camp Bowie Blvd., Fort Worth, TX 76107, USA
[b] Institute of Legal Medicine, Innsbruck Medical University, Innsbruck, Austria
[c] Penn State Eberly College of Science, University Park, PA, USA
[d] Department of Forensic Medicine, Hjelt Institute, P.O. Box 40, 00014 University of Helsinki, Helsinki, Finland
[e] Center of Excellence in Genomic Medicine Research (CEGMR), King Abdulaziz University, Jeddah, Saudi Arabia

1. New publications - 6

Three major U.S. populations (n=283)

	HV1/HV2			mtG		
	AFA	CAU	HIS	AFA	CAU	HIS
# Individuals	87	83	113	87	83	113
# Unique haplotypes	76	77	96	85	83	111
				+11.8%	+7.8%	+15.6%

		HVI/HVII		mtGenome	
Populations	*n*	RMP	GD	RMP	GD
AFA	*87*	2.42%	98.72%	1.31%	99.84%
CAU	*83*	3.12%	98.06%	1.20%	100.00%
HIS	*113*	3.33%	97.53%	0.98%	99.91%
Mean ±SD		2.96 ±0.48%	98.10 ±0.59%	1.16[c] ±0.17%	99.91[d] ±0.08%

King et al (2014)

1. New publications - 7

LETTER

Questioning the prevalence and reliability of human mitochondrial DNA heteroplasmy from massively parallel sequencing data

In their analysis of massively parallel sequencing data (MPS) from the 1000 Genomes Project, Ye et al. (1) report a very high rate of human mitochondrial DNA (mtDNA) heteroplasmy (89.68% of individuals), including up to 71 point heteroplasmies within a single individual, when using an ~1% minor allele frequency (MAF) threshold. Inspection of the heteroplasmy data detailed in dataset S1 of ref. 1 revealed that contamination, diagnostic positions remained for sample HG00740, but a 25% threshold would be required to achieve the same result for sample HG01108. Regardless, it is evident that the conclusions drawn by the authors should be revisited if the data themselves are flawed. For example, in contrast to the positive correlation between substitution rates and heteroplasmy rates reported by Ye et al. (1), no correlation was observed ($R^2 = 0.003979$, $P =$

ACKNOWLEDGMENTS. We thank Melissa Scheible (American Registry of Pathology and Armed Forces DNA Identification Laboratory) for data review and Eric Pokorak (Federal Bureau of Investigation) for valuable feedback on the manuscript.

Rebecca S. Just[a,b,c,1], Jodi A. Irwin[d], and Walther Parson[e,f]

[a]Emerging Technologies Section, Armed Forces DNA Identification Laboratory, Armed Forces Medical Examiner System,

PHP detected by Sanger:

Control region - 6% (Irwin et al 2009) = evol. hotspots

mtGenome - 24% (Just et al 2014) = random positions

max. 3 PHPs per haplotype

Extensive pathogenicity of mitochondrial heteroplasmy in healthy human individuals

Bullet points:

Analyzed mtDNA sequence data from 1000 Genomes Project

Mean coverage of ~2,000x

Use a combination of stringent thresholds and a maximum-likelihood method to define heteroplasmy

~90% of the individuals carry at least one heteroplasmy (1% minor allele frequency (MAF) threshold)

Positive correlation between substitution rates and heteroplasmy rates (!)

Extensive pathogenicity of mitochondrial heteroplasmy in healthy human individuals

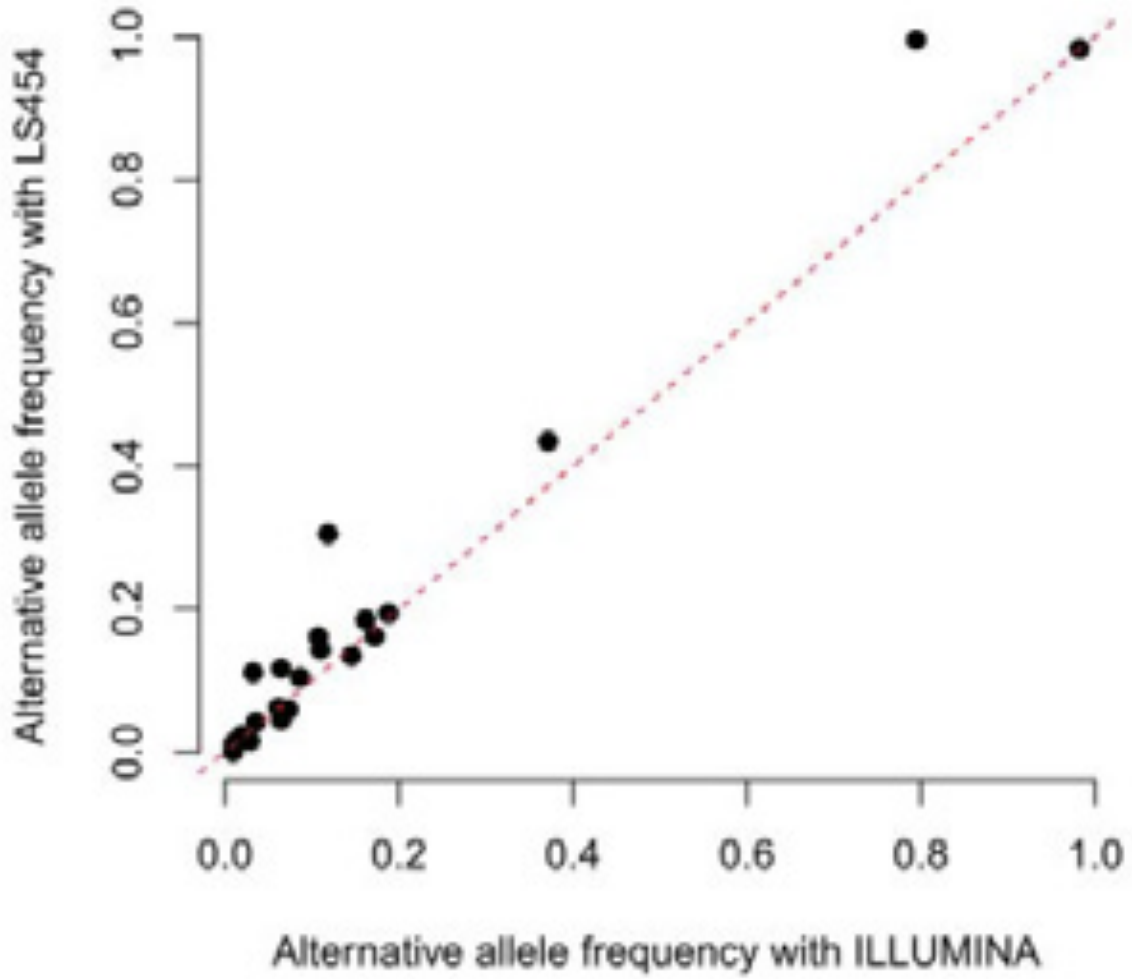


Fig. S2. The comparison of alternative allele frequencies for heteroplasmies identified in 9 individuals sequenced by both ILLUMINA and LS454. The allele frequencies were estimated by ML method.

Extensive pathogenicity of mitochondrial heteroplasmy in healthy human individuals

Inspection of Dataset (Dataset S1):

15 samples with 20 or more PHPs (**up to 71!**)

80.7% of the 584 PHPs occurred at hg-specific sites

HG00740: 90% of the **71 PHPs** can be ascribed to either hg L1b1a7a and B2b3a

HG01108: 50 and 12 of the **69 PHPs** are diagnostic for hgs L0a1a2 and M7c1b

Heteroplasmy???

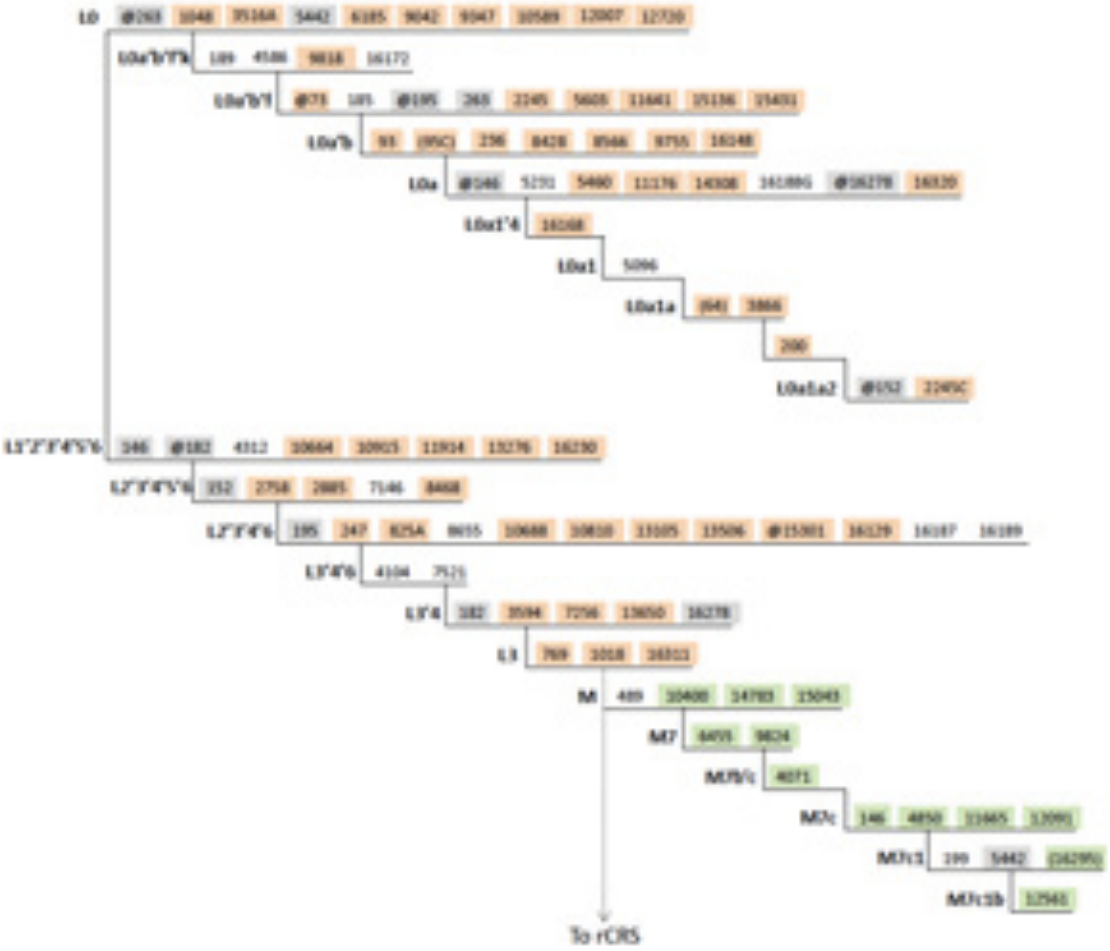
Just et al (2014)

No hg-specific PHPs remain when

a 15% MAF is applied for HG00740 and a 25% MAF for HG01108

No correlation between substitution rates and PHP rates was observed (R2=0.003979, p=0.23) when only the coding region PHPs with a MAF greater than 15% were analyzed using the same substitution rate data employed by the authors

1. New publications



2. Meetings

NICC INCC

DNA IN FORENSICS 2014

- Home
- Book of Abstracts
- Programme
- Illumina workshop

BRUSSELS 2014

On 14, 15 and 16 May

9th International **Y-chromosome workshop**
6th International **EMPOP meeting**

YHRD.ORG 3.0

EMPOP

Invitation

DNA in Forensics 2014 is the place to be for students, academics, and forensic experts who want to learn more about the latest evolutions in Y-chromosome and mitochondrial DNA research.

The conferences on haploid markers have become a tradition in the forensic scientific meetings with four of them in Berlin (1996, 2000, 2004 and 2010), two in Innsbruck (2006, 2012), one in Porto (2002) and one in Ancona (2008). In the spirit of the previous events, the organization of DNA in Forensics 2014 was inspired by an actual demand of experts and scientists in the forensic field. Therefore, the upcoming meeting will deal with the question: Is NGS NowGS in forensics?

EMPOP

GMI DNA

3. MtDNA/EMPOP Trainings

XIX JORNADAS DEL GHEP-ISFG

Quito (Ecuador), 9-12 septiembre 2014

ghep-isfg

Cruz Roja Ecuatoriana

CRUZ VITAL

Massively Parallel Sequencing of STRs
Considerations on nomenclature and databasing

Dr. Walther Parson with ideas from colleagues
assoc. Prof. Institute of Legal Medicine, Innsbruck, Austria
adj. Prof. Penn State Eberly College of Sciences, PA, USA
walther.parson@i-med.ac.at

Known nucleotide variation

Barber et al (1995) ***Int J Legal Med*** Structural variation of novel alleles at the Hum vWA and Hum FES/FPS short tandem repeat loci

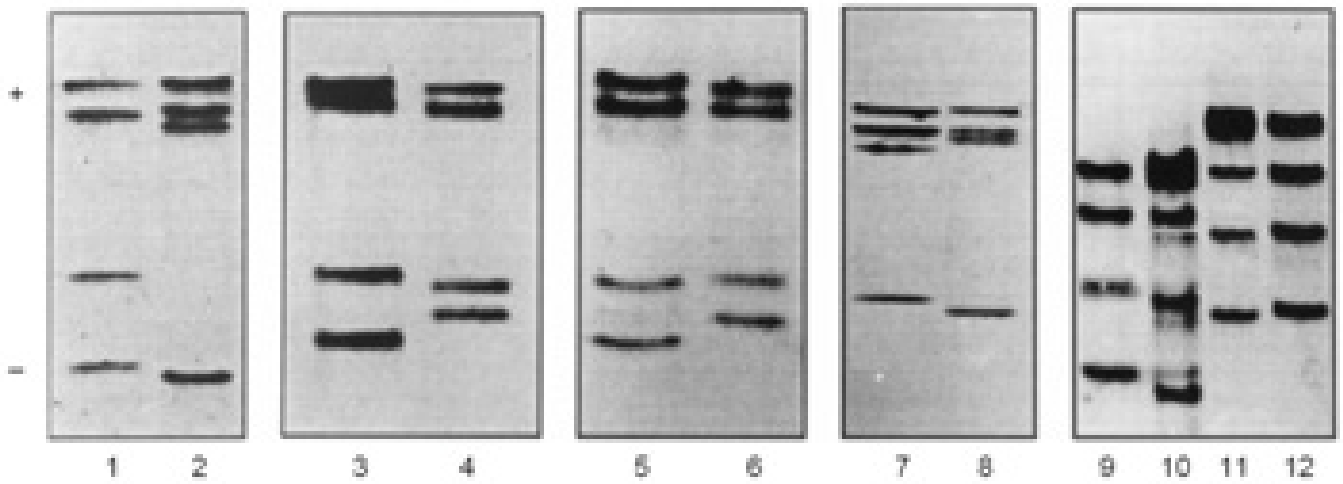


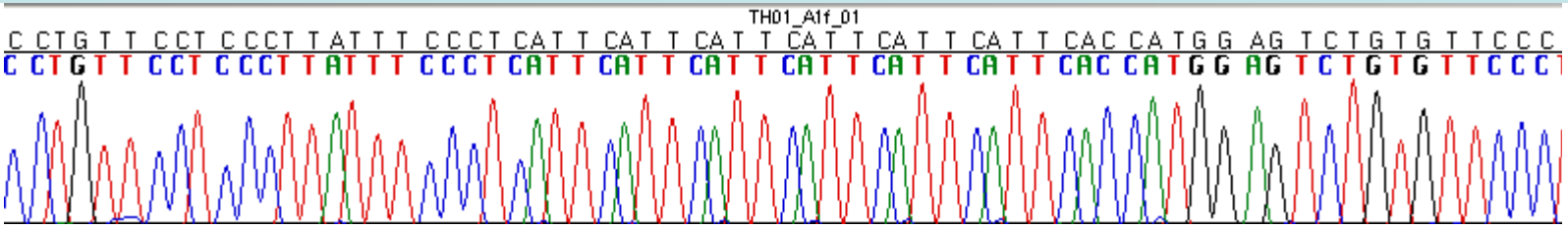
Fig. 1 Different types of heteroduplex patterns observed in some genotypes of FES/FPS. Lanes 1: 8–10 T(177), 2: 8–10 C(177), 3: 9–10 T(177), 4: 9–10 C(177), 5: 10 T(177)-11, 6: 10 C(177)-11, 7: 10 C(177)-12, 8: 10 T(177)-12, 9:10 T(177)-13, 10: 10 C(177)-13, 11: 7–10 T(177), 12: 7–10 C(177)

Pereira et al (1999) ***Int J Legal Med***

Analysis of STRs

forward primer (21 bp) 1 bp Repeat 24 bp reverse primer (21 bp)

CCTGTTCCTCCCTTATTTCCCTCATTCATTCATTCATTCATTCATTCACCATGGAGTCTGTGTTCCC



example TH01 allele 6 – 67 bp

~ 67 bp fragment (size marker)
no information on base composition

p_D depends frequency of size categories

A	313
C	289
G	329
T	304

~ 20263 Da molecule
Estimate size (67 bp)
Estimate base composition (Ref)

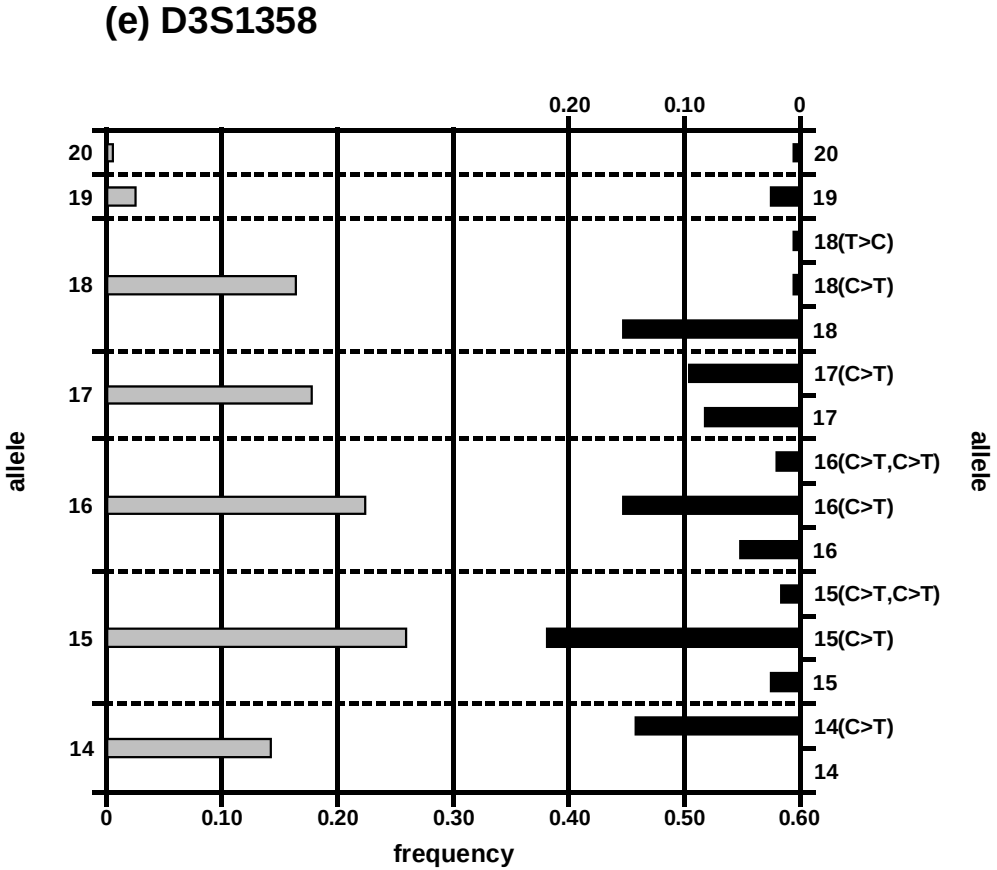
p_D depends frequency of size categories **and** nucleotide variability

Oberacher et al (2001) ***Anal Chem***

Increasing the discrimination power

Austrian population study (N=98)

7 allele categories



14 allele categories

Oberacher et al (2008) ***Human Mutation***

Increasing the discrimination power

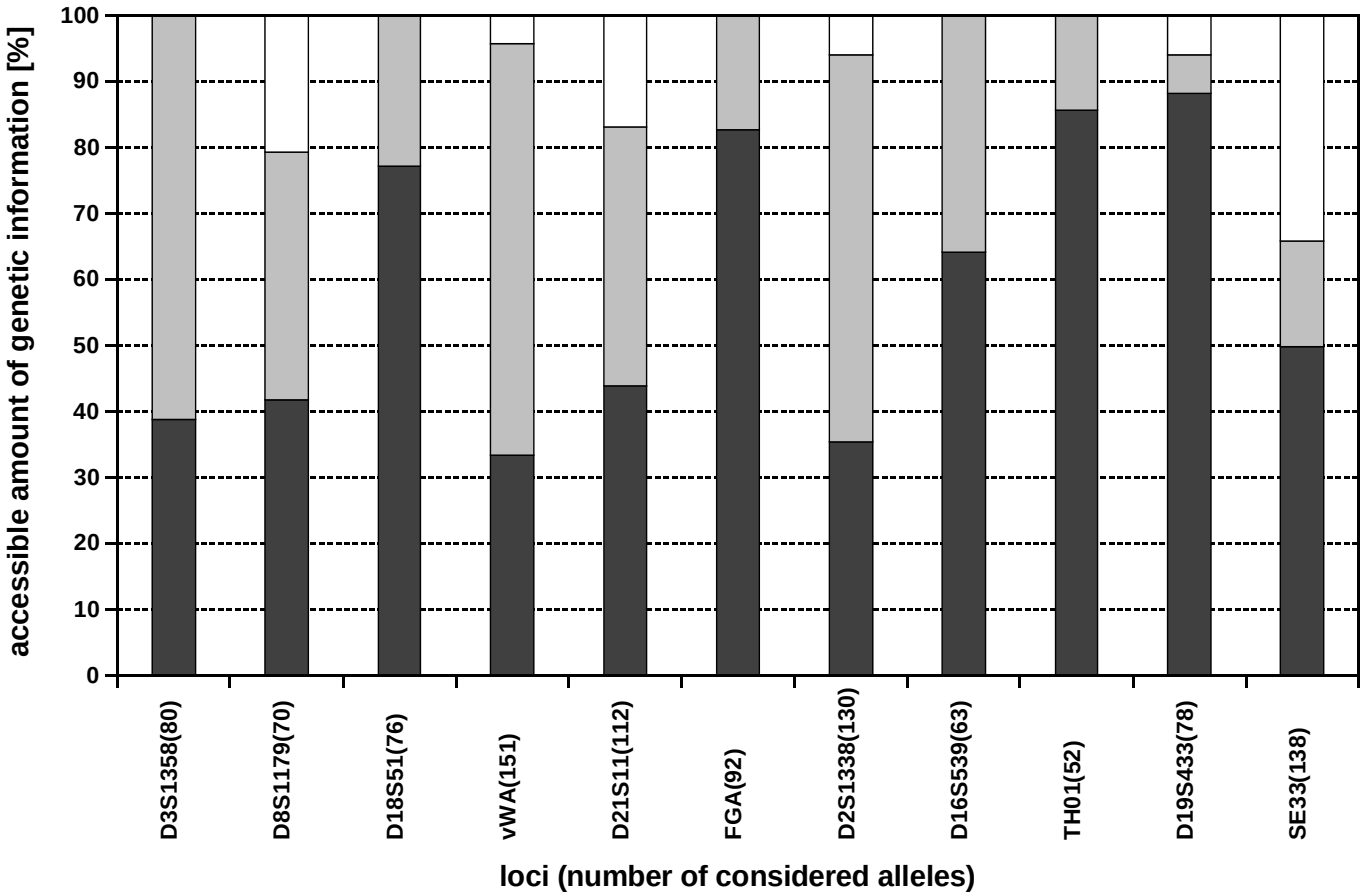
D3S1358: Sequenced alleles 15 and 16

15	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGACAGATAGA	TACATG
15	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGACAGATAGA	TACATG
15(C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGATAGA	TACATG
15(C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGATAGA	TACATG
15(C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGATAGA	TACATG
15(C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGATAGA	TACATG
15(C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGATAGA	TACATG
15(C>T,C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGATAGA	TACATG
15(C>T,C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGATAGA	TACATG
15(C>T,C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGATAGA	TACATG
16	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGACAGATAGA	TACATG
16	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGACAGATAGA	TACATG
16(C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGATAGA	TACATG
16(C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGATAGA	TACATG
16(C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGATAGA	TACATG
16(C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGATAGA	TACATG
16(C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGACAGATAGA	TACATG
16(C>T,C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGATAGA	TACATG
16(C>T,C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGATAGA	TACATG
16(C>T,C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGATAGA	TACATG
16(C>T,C>T)	GACAGAGCAAGACCCTGTCTCA	TAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGATAGACAGATAGA	TACATG

Relative discrimination

Population studies: Austria (N=100), South Africa (N=108), Sakha Republic (N=94)

total number of sequences alleles = 1042



Massively Parallel Sequencing of STRs

ANALYSIS REPORTING STORAGE QUERY

Scheible et al (**2011**) FSIG SS, Fordyce et al (**2011**) BioTechniques
Bornmann et al (**2012**) BioTechniques, van Neste et al (**2012**) FSIG
Dalsgaard et al (**2013**) FSIGSS, Rockenbauer et al (**2013**) FSIGSS, Warschauer et al (**2013**) FSIG
Gelardi et al (**2014**) FSIG, Gettings et al (**2014**) Poster at AAFS

Reporting

General:

Report coding region strand whenever possible (coding genes, pseudogenes, introns). Seems straight forward for cases where this is known. In other cases it has been proposed to use the nomenclature described in the literature, which may be in conflict with the rule.

Rather than trying to chase down all individual exceptions databases should be able to work with coding region strand AND complement reverse of it.

Reporting is then de-coupled of matching (similar experience in EMPOP).

Reporting

General:

Determine repeat motif according to established guidelines from Sanger data using 5' end of first repeat motif; revisit earlier guidelines and outline synopsis - not always unambiguous

Keep repeat structure in mind (simple, compound, complex). Consensus method is preferred for complex STRs with (large) deletions. This process needs to be software-based.

Will benefit from examples - therefore data generation will be elemental

Reporting

Specific:

1) Report unaligned nucleotide strings

cannot be understandably reported as such, but could serve as basis for database searches (see below)

2) Unique identifiers using sequence-specific designators

(e.g. 13F; Planz 2012)
stand alone solution, difficult to use between groups

3) Bracketed sequence (suggested by Gelardi 2014)

e.g. $[STR1]_m[STR2]_n$non-repetetive_sequence$[STR3]_o$rs123456[STR3]
non-repetetive_sequences need to be considered to maintain compatibility with old data

Storage

Databases need to be capable to turn allele designations into an unaligned string of nucleotides (easy) and back into one or more accepted allele designation nomenclature (more difficult)

Searching needs to be performed on unaligned nucleotide strings (easy but computationally intensive - strong server architecture required)

Searching

Unaligned string-based search is the only solution, because there are many ways to name one and the same nucleotide string; done in EMPOP

Event-based string search: capture combined indels

Discussion

We should only allow for one searching system (string-based)

We may need to allow for multiple reporting systems; if we do not need it - fine

Most difficult part is reporting:
What are we suggesting to report?
Only the STR region?

- Excludes discriminatory information in flanking region (e.g. AIMs, indels, …)
- Likely makes the new data incompatible to existing ones

STR and FR?

- add sequence as it is?

EDNAP AIMs Exercise 2014

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- Two binary AIM sets of 34 SNPs and 46 Indels. Five controls from each corner of the globe and a 3:1 artificial mixture.

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- Genotyping concordance and ease-of-use of each system
- Use of statistical tools for ancestry inference: Bayes analysis / PCA
- Mixture detection - albeit with a single sample.

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- 21 Packages sent out, 19 Labs successfully completed: 1 US; 3 ANZ; 15 EUR. 16/19 labs participated in 2013 *Irisplex* exercise, but 34-plex SNaPshot profiles are more challenging to assess: 8>34.

Forensic ancestry analysis with two simple capillary electrophoresis AIM panels: Results of a collaborative EDNAP exercise

C. Santos[a1], M. Fondevila[a1], D. Ballard[b1], R. Banemann[c], A.M. Bento[d], C. Børsting[e1], W. Branicki[f2], F. Brisighelli[g], M. Burrington[h], T. Capal[i], L. Chaitanya[j], R. Daniel[k], R. England[l], K.B. Gettings[m], T.E. Gross[n1], C. Haas[o], J. Harteveld[p], P. Hoff-Olsen[q], A. Hoffmann[c], M. Kayser[j], A. Linacre[r], P. Kohler[q2], M. Mayr-Eduardoff[s1], C. McGovern[l], N. Morling[e1], F. Noël[t], G. O'Donnell[h], W. Parson[s1], V.L. Pascali[g], M.J. Porto[d], A. Roseth[q], P.M. Schneider[n1], T. Sijen[p], V. Stenzl[i], D. Syndercombe Court[b1], J. Templeton[r], M. Turanska[u], D. Valérie[t], P.M. Vallone[m], R.A.H. van Oorschot[k], L. Zatkalikova[u], The EUROFORGEN-NoE Consortium; Á. Carracedo[a1], C. Phillips[a1*]

[a] Forensic Genetics Unit, Institute of Legal Medicine, University of Santiago de Compostela, Santiago de Compostela, Spain

[b] Department of Forensic and Analytical Science, School of Biomedical Sciences, King's College, London, UK

[c] Kriminaltechnik, Bundeskriminalat, Wiesbaden, Germany

[d] Serviço de Genética e Biologia Forenses, Delegação do Centro, Instituto Nacional de Medicina Legal e Ciências Forenses, Coimbra, Portugal

[e] Section of Forensic Genetics, Department of Forensic Medicine, Faculty of Health and Medical Sciences, University of Copenhagen, Frederik V's Vej 11, DK-2100 Copenhagen, Denmark

[f] Section of Forensic Genetics, Institute of Forensic Research, Kraków, Poland

[g] Forensic Genetics Laboratory, Institute of Legal Medicine, Università Cattolica del Sacro Cuore, Rome, Italy

[h] Forensic Science Laboratory, Dublin, Ireland

[i] Department of Forensic Genetics, Institute of Criminalistics, Prague, Czech Republic

[j] Department of Forensic Molecular Biology, Erasmus MC University Medical Centre Rotterdam, Rotterdam, The Netherlands

[k] Office of the Chief Forensic Scientist, Forensic Services Department, Victoria Police, Australia

[l] ESR, Private Bag 92021, Auckland, New Zealand

[m] Material Measurement Laboratory, National Institute of Standards and Technology, Gaithersburg, MD, USA

[n] Institute of Legal Medicine, Faculty of Medicine, University of Cologne, Cologne, Germany

[o] Institute of Legal Medicine, University of Zurich, Zurich, Switzerland

[p] Department of Human Biological Traces, Netherlands Forensic Institute, The Hague, The Netherlands

[q] Department of Forensic Biology, Norwegian Institute of Public Health, Oslo, Norway

[r] School of Biological Sciences, Flinders University, Adelaide, South Australia 5042, Australia

[s] Institute of Legal Medicine, Innsbruck Medical University, Innsbruck, Austria

[t] National Institute of Criminalistics and Criminology, Chaussée de Vilvoorde 100, 1120 Brussels, Belgium

[u] Institute of Forensic Science, Ministry of the Interior, Department of Biology and DNA Analysis, Slovenská Lupca, Slovakia

[1] Also participated in the EUROFORGEN ancestry analysis inter-laboratory exercise pilot

[2] Participated in the EUROFORGEN exercise only

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[1] Also participated in the EUROFORGEN ancestry analysis inter-laboratory exercise pilot
[2] Participated in the EUROFORGEN exercise only

EUROFORGEN piloted the exercise. Purchased Indel primer stocks for any lab to assess and tested their stability in transit. Feasibility of mixture analysis with NIST SRMs.

Variation in CE systems used - 5 labs chose Indels only

	CE			
	ABI Sequencer	Polymer	Dilution factor	
			34-plex	AIM-indels
1	3130xl	POP-4	None	None
2	3130xl	POP-4	None	1:10 (samples A-E) 1:5 (F & NTC)
4	3100	POP-6	None	None
5	3130xl	POP-4		None
6	3130xl	POP-7	None	1:10
7	3130	POP-4	None	None
8	3130xl	POP-4	None	1:20
9	3130xl	POP-7	None	None
11	3130xl	POP-4	None	1:10 (samples E & F)
12	3500	POP-4		None
13	3130	POP-4	1:10	1:10
14	3500xl	POP-4		1:5
15	3130xl	POP-4		(not reported)
16	3130xl	POP-4		None
17	3130xl	POP-4	None	1:20
18	3130	POP-4	None	None
19	3500xl	POP-4	None	1:20
20	3130xl	POP-4	None	1:10
21	3130	POP-4	None	None

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	ABI Sequencer	Polymer	Dilution factor	
			34-plex	AIM-indels
1	3130xl	POP-4	None	None
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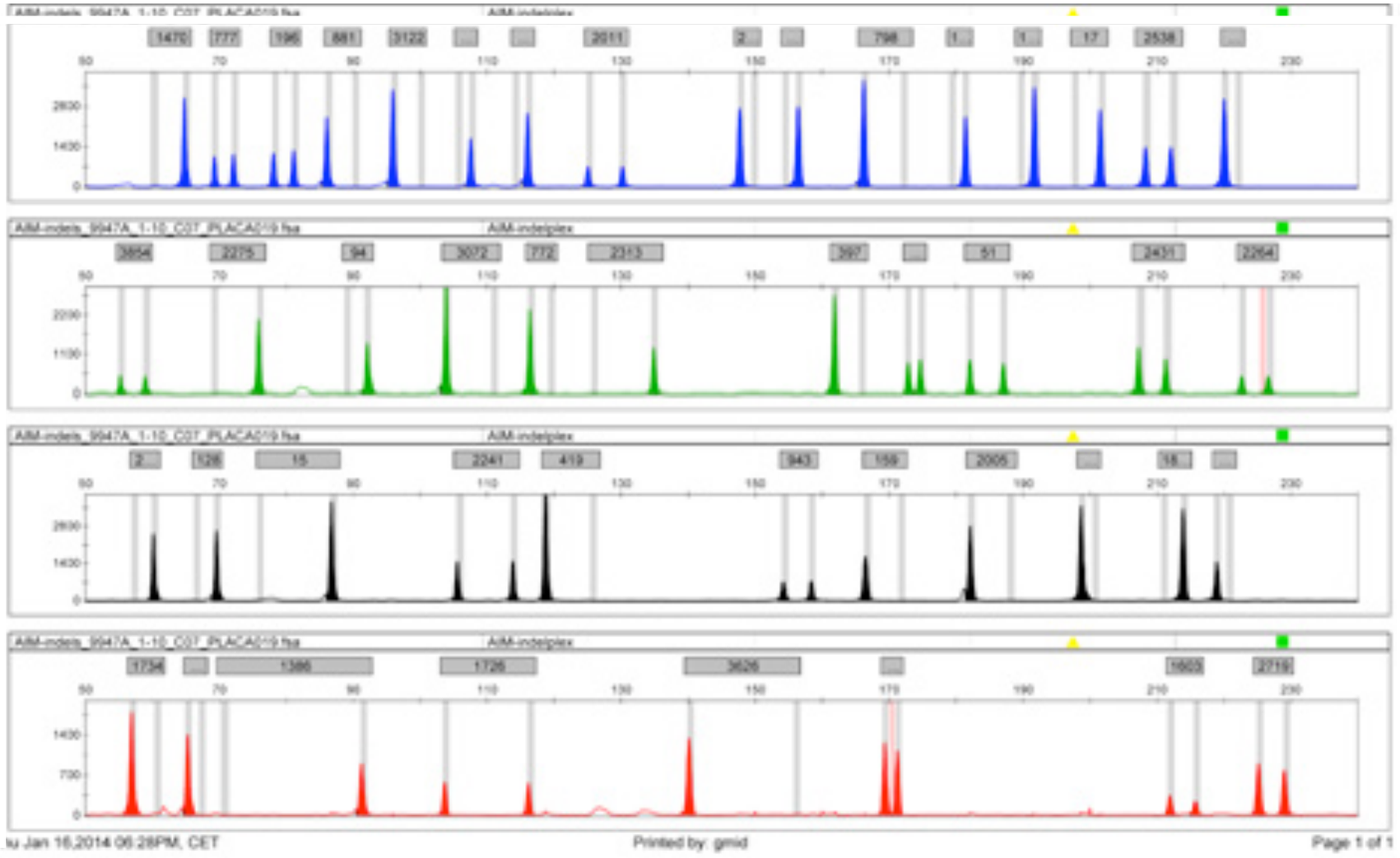
6	3130xl	POP-7	None	1:10
7	3130	POP-4	None	None
8	3130xl	POP-4	None	1:20
9	3130xl	POP-7	None	None
11	3130xl	POP-4	None	1:10 (samples E & F)
12	3500	POP-4		None
13	3130	POP-4	1:10	1:10
14	3500xl	POP-4		1:5
15	3130xl	POP-4		(not reported)
16	3130xl	POP-4		None
17	3130xl	POP-4	None	1:20
18	3130	POP-4	None	None
19	3500xl	POP-4	None	1:20
20	3130xl	POP-4	None	1:10
21	3130	POP-4	None	None

13 labs: 3130 / POP-4

3 labs: 3500 / POP-4

2 labs: 3130 / POP-7

1 lab: 3100 / POP-6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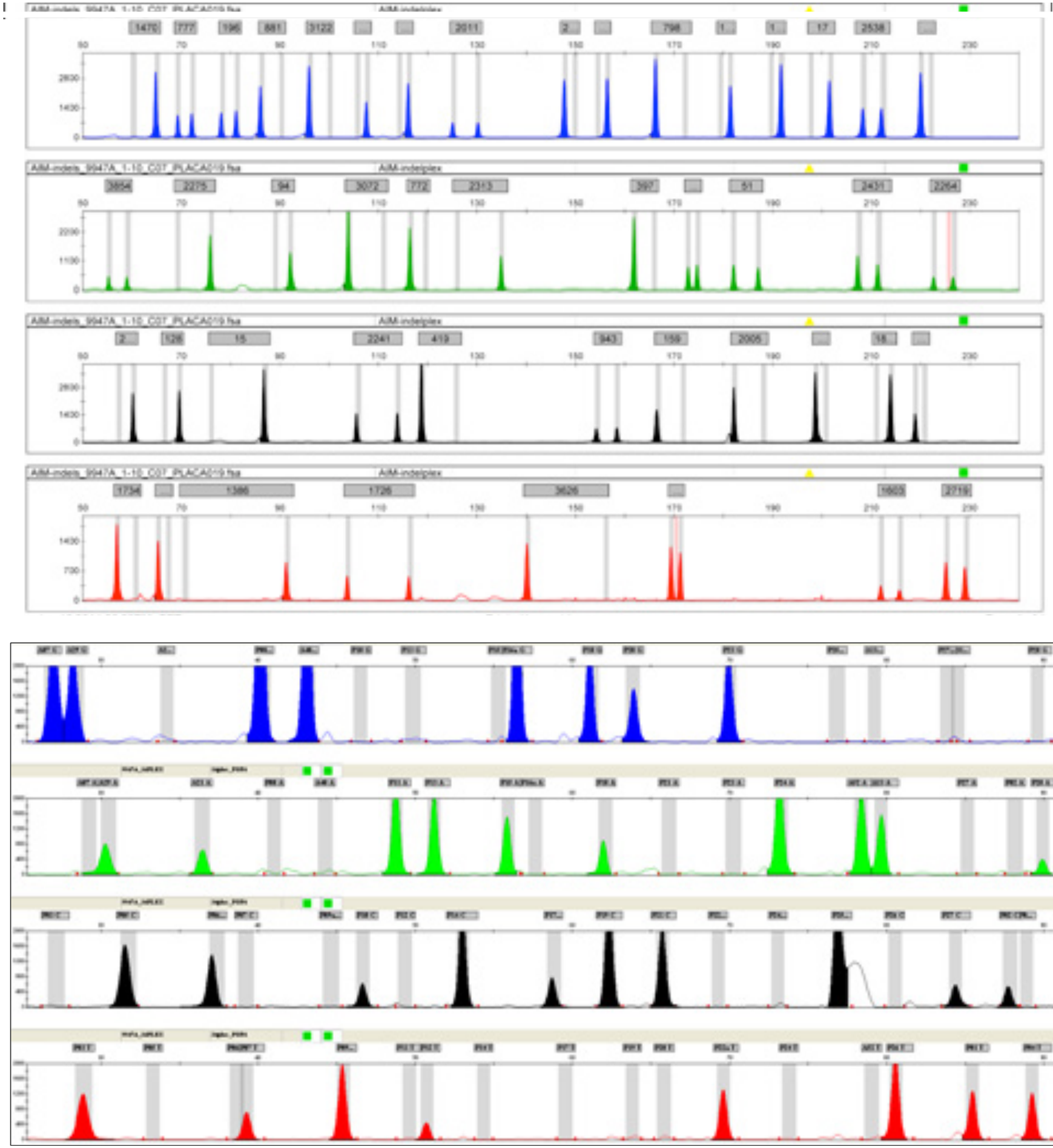
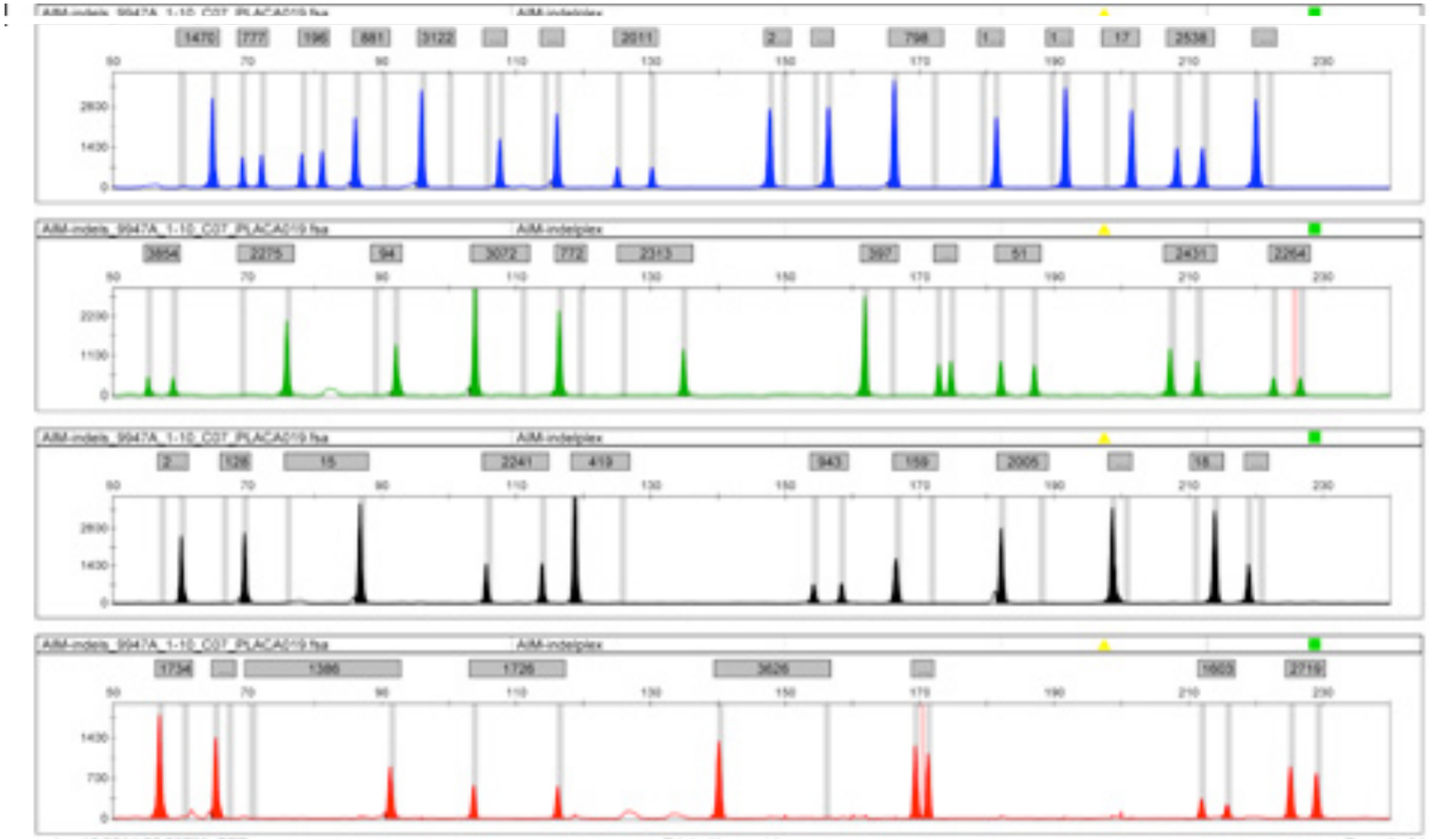
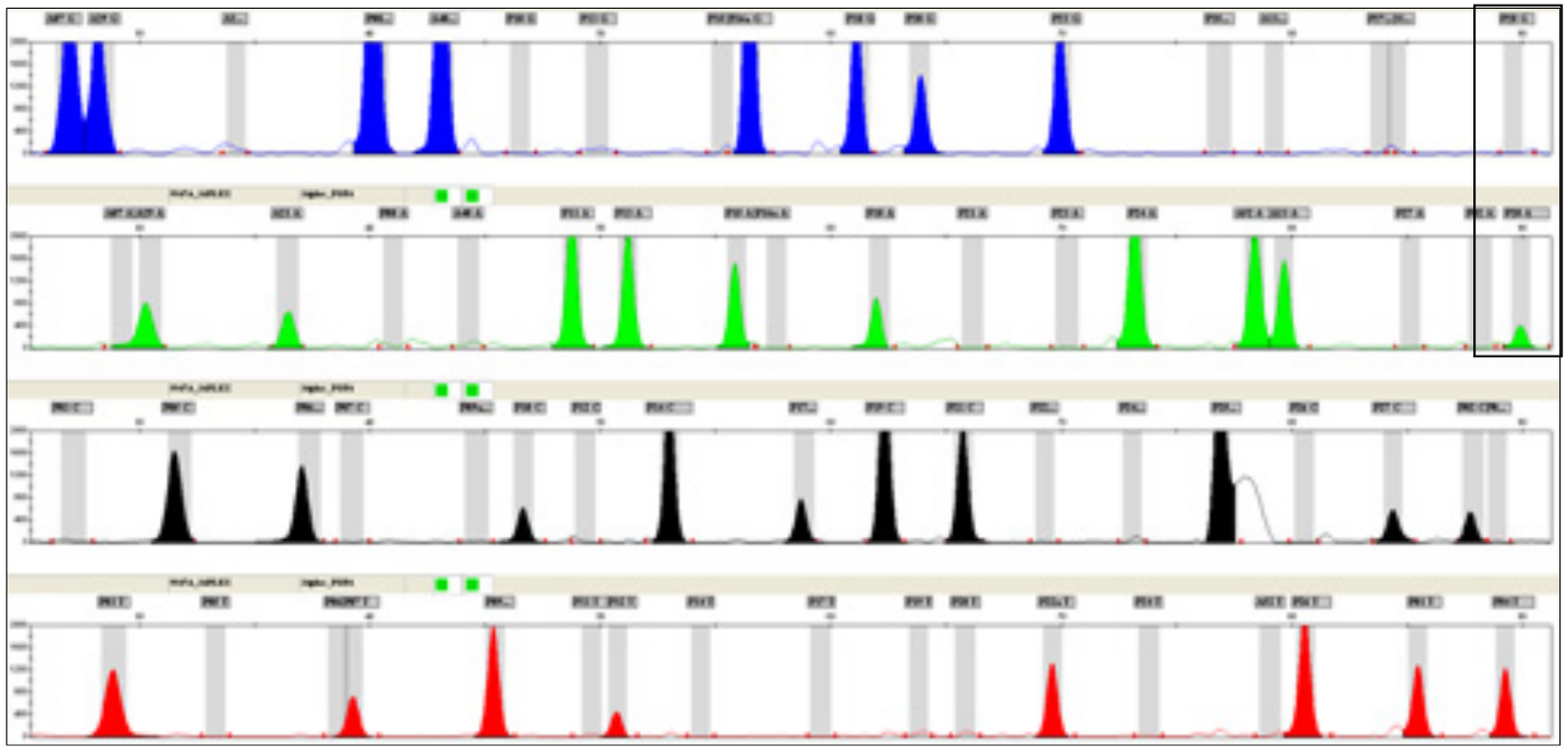
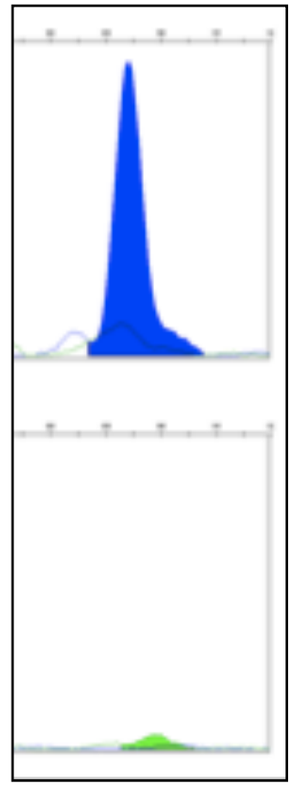
AIM-indelsplex
1470 777 196 881 3122 2011 2 798 1 1 17 2538
3854 2275 94 3072 772 2313 397 51 2431 2264
2 126 15 2241 419 943 159 2005 18
1734 1386 1726 3626 1603 2719



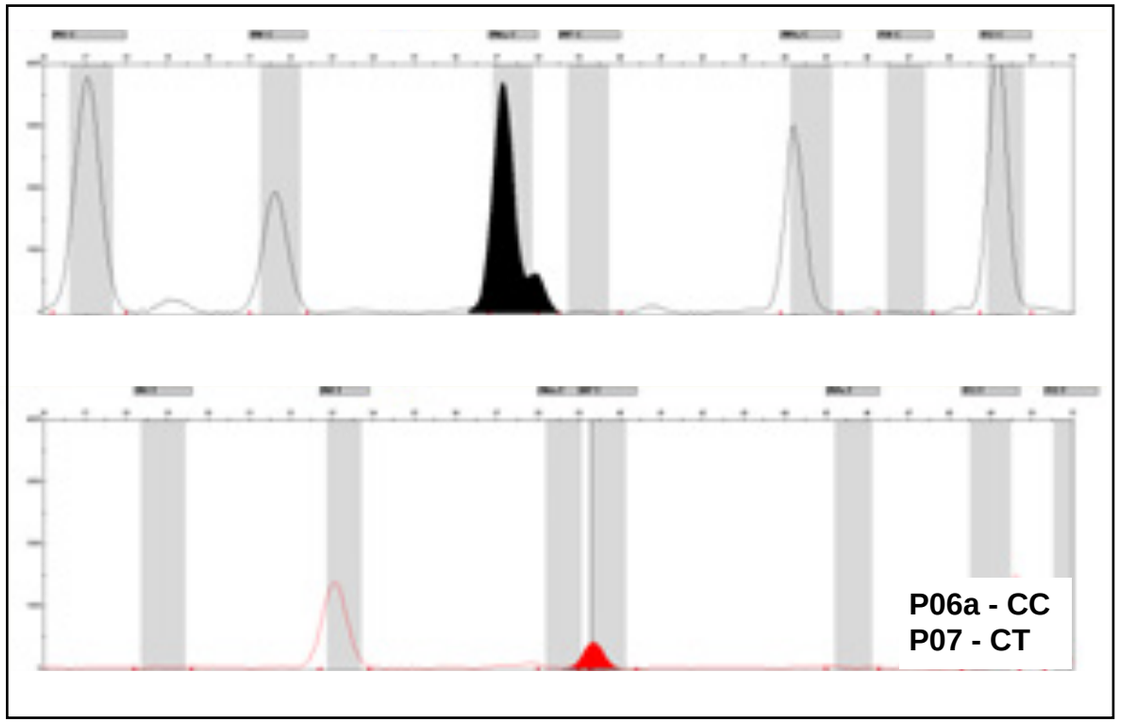


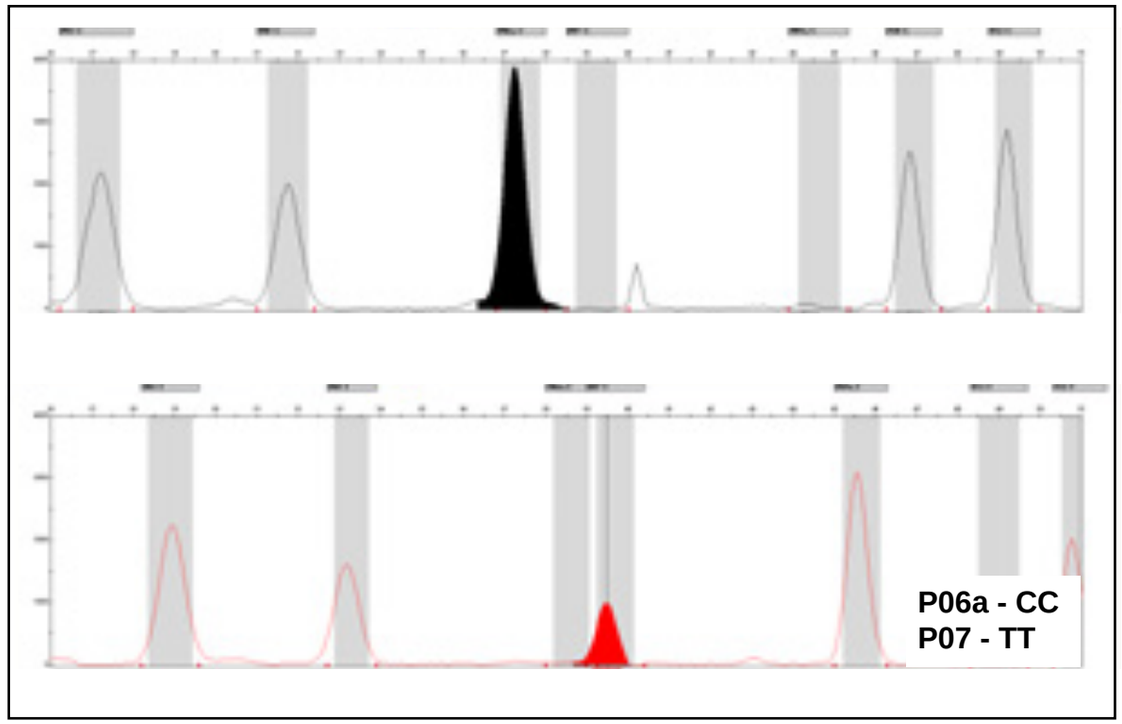


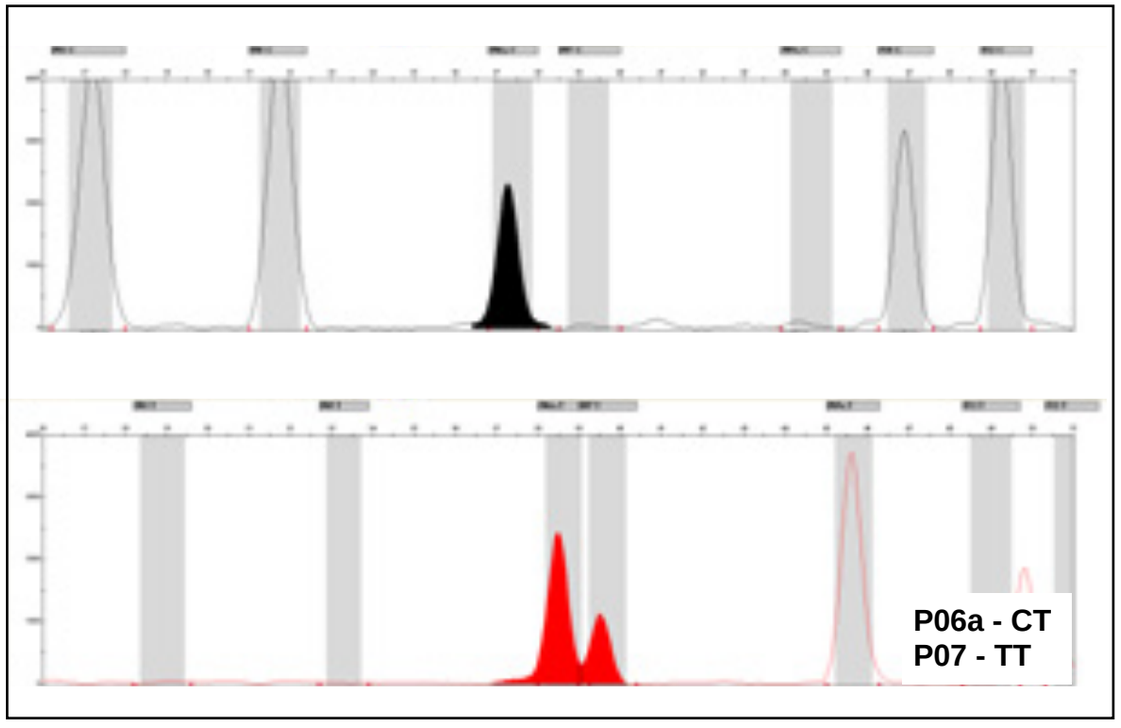


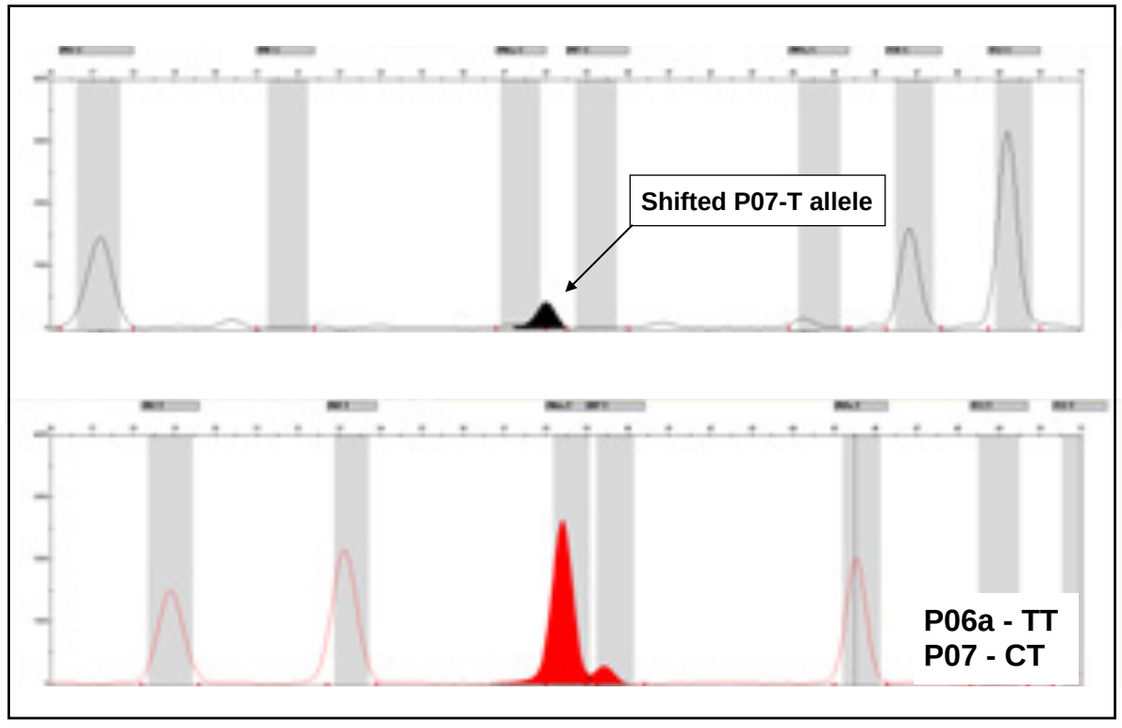
34-plex: known SNaPshot profile issues

A. P06a-P07 peak pair very close together



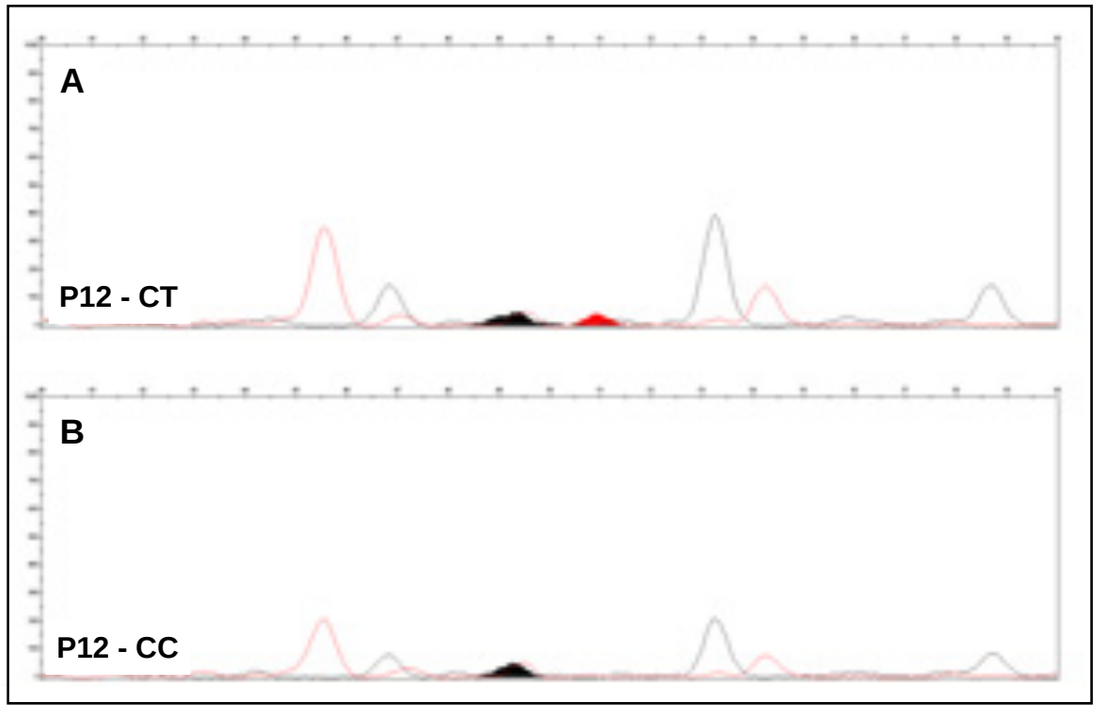


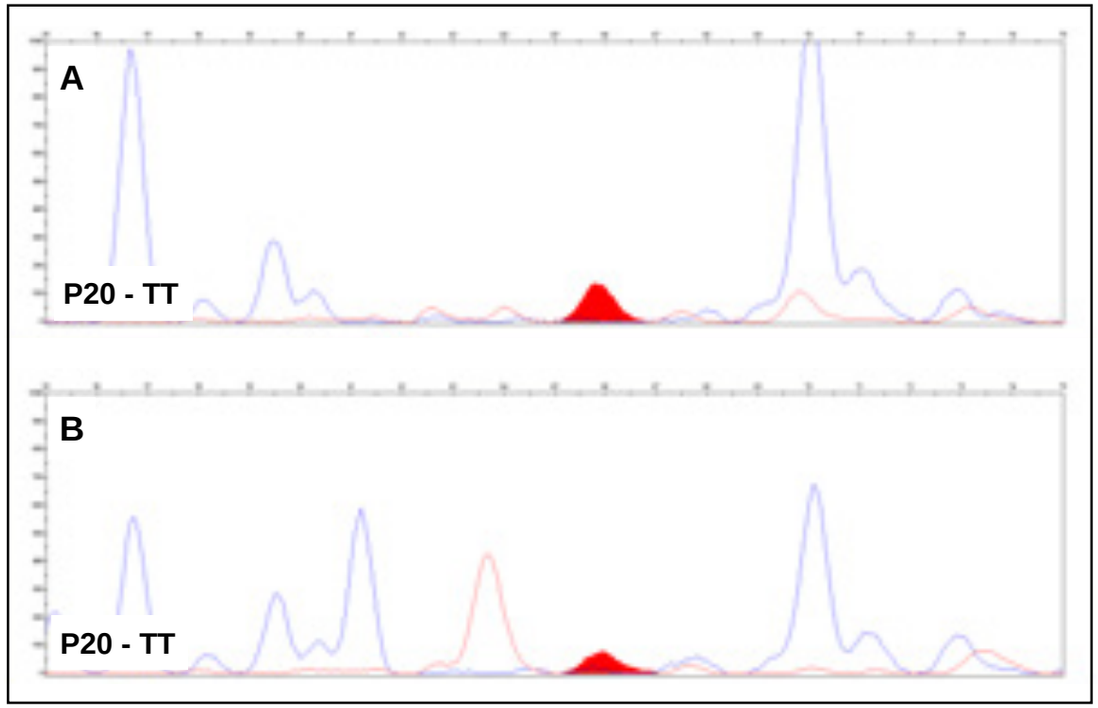


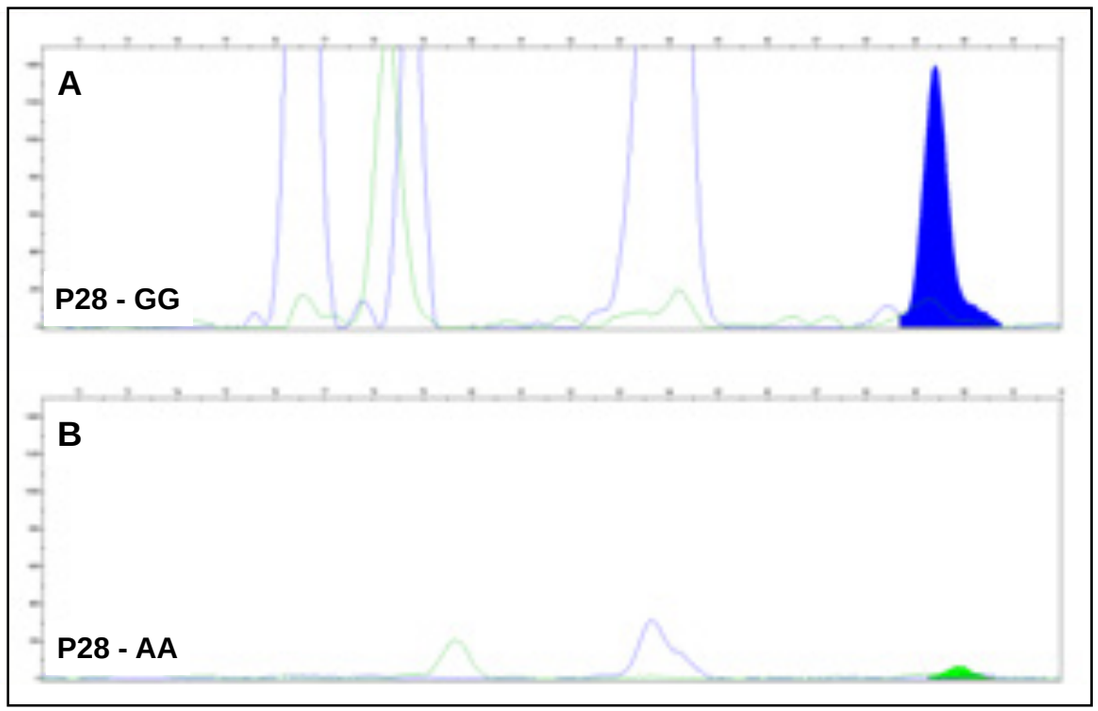


34-plex: known SNaPshot profile issues

B. P12, P20 and P28 have low peak heights for one or both alleles

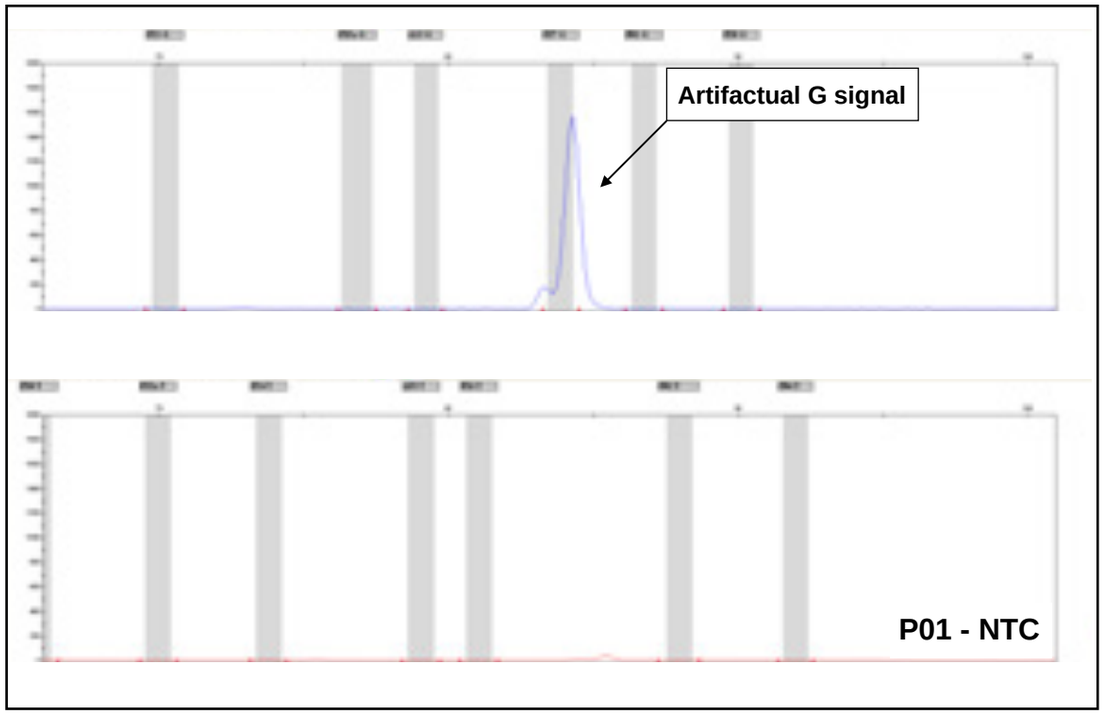


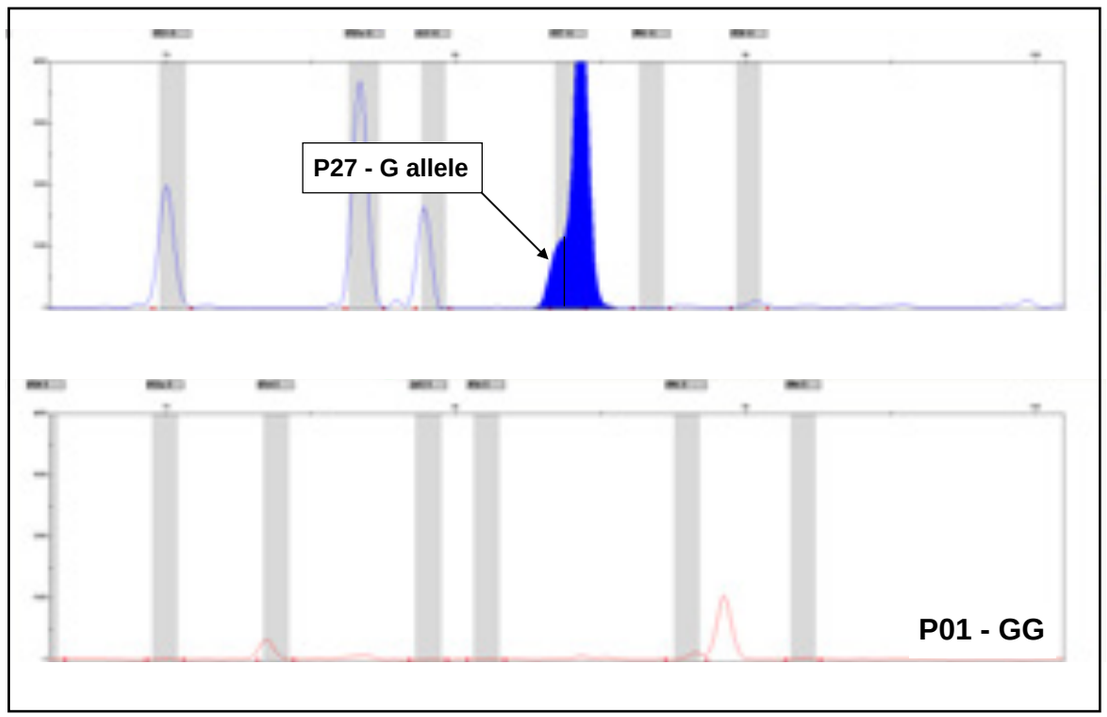


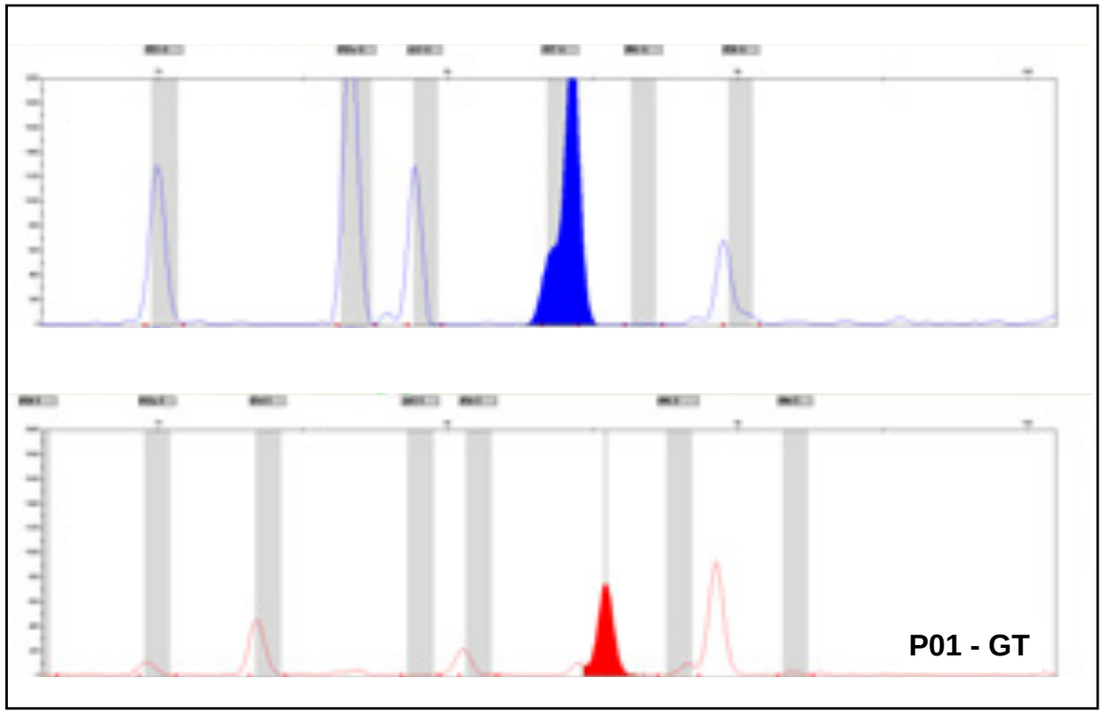


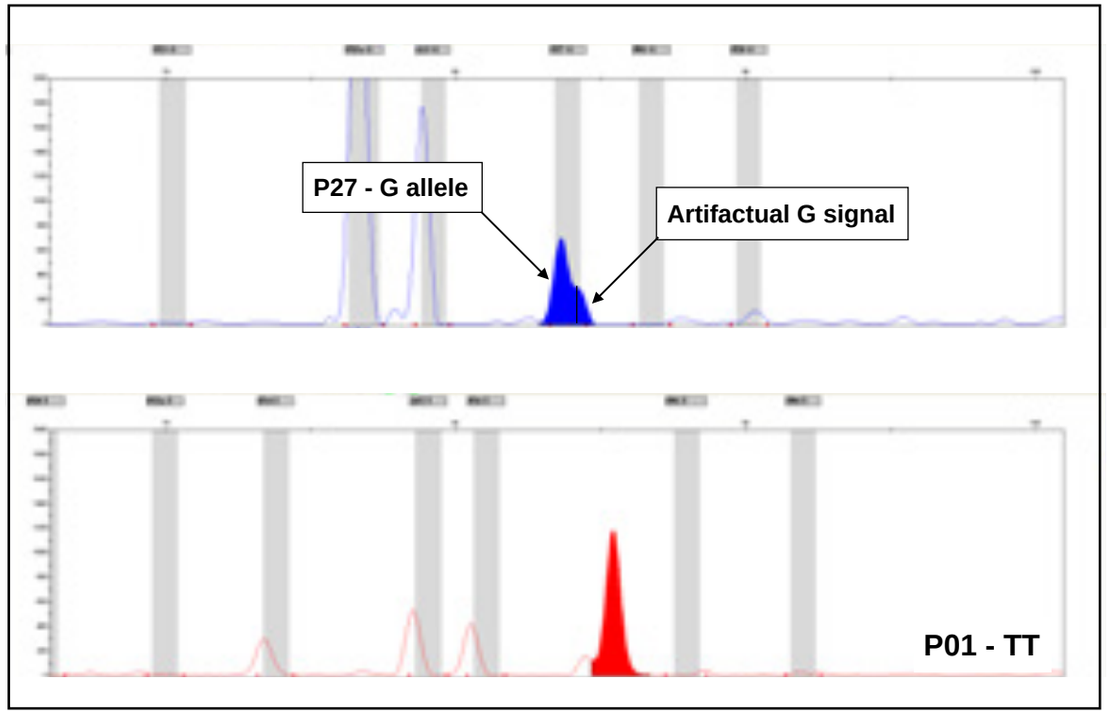
34-plex: known SNaPshot profile issues

C. P01 has a G-like artifactual peak in NTC + mobility shift into P27 position









Genotyping concordance: 34-plex, samples A-E

14 labs

34-plex SNPs ordered by decreasing genotyping reliability for participants

	1	2	4	5	6	7	8	9	11	12	13	14	15	16	17	18	19	20	21	Genotype completeness	Genotype concordance
rs2065160	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs3785181	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs896788	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs1573020	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				0 | 0	0 | 0	0 | 0	0 | 0	0 | 5	92.86	98.57
rs1426654	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs2572307	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs2814778	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs730570	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs1886510	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 3	0 | 0	0 | 5	0 | 0	0 | 0	88.57	97.14
rs3827760	0 | 0	0 | 1	0 | 0		0 | 0	0 | 2	0 | 4	0 | 1	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	0 | 0	87.14	97.14
rs2040411	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	1 | 0	100	95.71
rs2065982	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	95.71
rs7897550	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	95.71
rs1978806	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	1 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	95.71
rs2026721	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 1	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	1 | 0	97.14	95.71
rs16891982	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	94.29
rs12913832	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	94.29
rs1321333	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	94.29
rs773658	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 1	0 | 0	0 | 0	98.57	94.29
rs2303798	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	1 | 1	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	98.57	92.86
rs1335873	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	1 | 2	97.14	92.86
rs1498444	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		1 | 0				2 | 0	0 | 0	0 | 0	0 | 2	2 | 0	97.14	92.86
rs4540055	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	1 | 2	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 1	0 | 0	0 | 0	95.71	92.86
rs5997008	0 | 0	1 | 1	0 | 0		0 | 0	1 | 0	0 | 1	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	1 | 0	95.71	92.86
rs881929	0 | 0	0 | 1	0 | 0		0 | 0	0 | 0	1 | 1	0 | 0	0 | 0		0 | 2				4 | 0	0 | 0	0 | 1	0 | 5	0 | 0	85.71	92.86
rs1024116	0 | 0	0 | 0	0 | 0		2 | 0	0 | 0	0 | 0	0 | 0	0 | 1		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	1 | 0	98.57	91.43
rs10843344	1 | 0	0 | 0	0 | 0		1 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 1	0 | 4	0 | 0	92.86	90.00
rs182549	0 | 0	0 | 0	0 | 0		4 | 0	0 | 0	0 | 0	0 | 1	0 | 0		0 | 4				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	92.86	90.00
rs10141763	0 | 0	0 | 0	0 | 0		3 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	88.57
rs722098	1 | 1	0 | 0	0 | 0		0 | 0	0 | 0	2 | 0	0 | 0	0 | 0		0 | 0				5 | 0	0 | 0	0 | 0	0 | 1	0 | 0	97.14	88.57
rs5030240	0 | 0	2 | 0	0 | 0		1 | 0	0 | 0	0 | 2	0 | 0	0 | 1		1 | 0				5 | 0	0 | 1	0 | 0	0 | 1	0 | 0	92.86	87.14
rs2304925	0 | 0	1 | 0	1 | 0		0 | 0	0 | 0	2 | 0	0 | 0	0 | 1		1 | 0				2 | 0	0 | 0	0 | 5	1 | 1	1 | 0	90.00	87.14
rs917118	1 | 0	0 | 0	0 | 0		3 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				5 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	85.71
rs239031	2 | 1	1 | 0	0 | 0		2 | 0	1 | 0	0 | 4	0 | 1	0 | 0		0 | 0				2 | 0	2 | 0	1 | 1	2 | 2	0 | 5	80.00	81.43
Profile completeness	98.82	98.24	100		100	98.82	90.59		98.24		96.47				98.24	99.41	91.18	86.47	92.94	***96.3***	
Profile concordance	95.29	97.06	99.41		90.59	98.82	94.12		99.41		98.24				44.71	98.82	99.41	98.24	95.29		***93.5*** *(14 labs)*
																					97.5 *(13 labs)*

miscalls | no-calls

Genotyping concordance: 34-plex, samples A-E

14 labs

34-plex SNPs ordered by decreasing genotyping reliability for participants

	1	2	4	5	6	7	8	9	11	12	13	14	15	16	17	18	19	20	21	Genotype completeness	Genotype concordance
rs2065160	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs3785181	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs896788	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs1573020	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				0 | 0	0 | 0	0 | 0	0 | 0	0 | 5	92.86	98.57
rs1426654	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs2572307	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs2814778	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs730570	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs1886510	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 3	0 | 0	0 | 5	0 | 0	0 | 0	88.57	97.14
rs3827760	0 | 0	0 | 1	0 | 0		0 | 0	0 | 2	0 | 4	0 | 1	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	0 | 0	87.14	97.14
rs2040411	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	1 | 0	100	95.71
rs2065982	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	95.71
rs7897550	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	95.71
rs1978806	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	1 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	95.71
rs2026721	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 1	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	1 | 0	97.14	95.71
rs16891982	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	94.29
rs12913832	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	94.29
rs1321333	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	94.29
rs773658	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 1	0 | 0	0 | 0	98.57	94.29
rs2303798	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	1 | 1	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	98.57	92.86
rs1335873	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	1 | 2	97.14	92.86
rs1498444	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		1 | 0				2 | 0	0 | 0	0 | 0	0 | 2	2 | 0	97.14	92.86
rs4540055	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	1 | 2	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 1	0 | 0	0 | 0	95.71	92.86
rs5997008	0 | 0	1 | 1	0 | 0		0 | 0	1 | 0	0 | 1	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	1 | 0	95.71	92.86
rs881929	0 | 0	0 | 1	0 | 0		0 | 0	0 | 0	1 | 1	0 | 0	0 | 0		0 | 2				4 | 0	0 | 0	0 | 1	0 | 5	0 | 0	85.71	92.86
rs1024116	0 | 0	0 | 0	0 | 0		2 | 0	0 | 0	0 | 0	0 | 0	0 | 1		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	1 | 0	98.57	91.43
rs10843344	1 | 0	0 | 0	0 | 0		1 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 1	0 | 4	0 | 0	92.86	90.00
rs182549	0 | 0	0 | 0	0 | 0		4 | 0	0 | 0	0 | 0	0 | 1	0 | 0		0 | 4				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	92.86	90.00
rs10141763	0 | 0	0 | 0	0 | 0		3 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	88.57
rs722098	1 | 1	0 | 0	0 | 0		0 | 0	0 | 0	2 | 0	0 | 0	0 | 0		0 | 0				5 | 0	0 | 0	0 | 0	0 | 1	0 | 0	97.14	88.57
rs5030240	0 | 0	2 | 0	0 | 0		1 | 0	0 | 0	0 | 2	0 | 0	0 | 1		1 | 0				5 | 0	0 | 1	0 | 0	0 | 1	0 | 0	92.86	87.14
rs2304925	0 | 0	1 | 0	1 | 0		0 | 0	0 | 0	2 | 0	0 | 0	0 | 1		1 | 0				2 | 0	0 | 0	0 | 5	1 | 1	1 | 0	90.00	87.14
rs917118	1 | 0	0 | 0	0 | 0		3 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				5 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	85.71
rs239031	2 | 1	1 | 0	0 | 0		2 | 0	1 | 0	0 | 4	0 | 1	0 | 0		0 | 0				2 | 0	2 | 0	1 | 1	2 | 2	0 | 5	80.00	81.43
Profile completeness	98.82	98.24	100		100	98.82	90.59		98.24		96.47				98.24	99.41	91.18	86.47	92.94	***96.3***	
Profile concordance	95.29	97.06	99.41		90.59	98.82	94.12		99.41		98.24				44.71	98.82	99.41	98.24	95.29		***93.5*** *(14 labs)*
																					97.5 *(13 labs)*

miscalls | no-calls 6.5% | 3.7%

Genotyping concordance: 34-plex, samples A-E

14 labs

34-plex SNPs ordered by decreasing genotyping reliability for participants

	1	2	4	5	6	7	8	9	11	12	13	14	15	16	17	18	19	20	21	Genotype completeness	Genotype concordance
rs2065160	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs3785181	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs896788	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs1573020	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				0 | 0	0 | 0	0 | 0	0 | 0	0 | 5	92.86	98.57
rs1426654	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs2572307	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs2814778	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs730570	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs1886510	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 3	0 | 0	0 | 5	0 | 0	0 | 0	88.57	97.14
rs3827760	0 | 0	0 | 1	0 | 0		0 | 0	0 | 2	0 | 4	0 | 1	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	0 | 0	87.14	97.14
rs2040411	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	1 | 0	100	95.71
rs2065982	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	95.71
rs7897550	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	95.71
rs1978806	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	1 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	95.71
rs2026721	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 1	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	1 | 0	97.14	95.71
rs16891982	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	94.29
rs12913832	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	94.29
rs1321333	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	94.29
rs773658	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 1	0 | 0	0 | 0	98.57	94.29
rs2303798	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	1 | 1	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	98.57	92.86
rs1335873	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	1 | 2	97.14	92.86
rs1498444	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		1 | 0				2 | 0	0 | 0	0 | 0	0 | 2	2 | 0	97.14	92.86
rs4540055	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	1 | 2	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 1	0 | 0	0 | 0	95.71	92.86
rs5997008	0 | 0	1 | 1	0 | 0		0 | 0	1 | 0	0 | 1	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	1 | 0	95.71	92.86
rs881929	0 | 0	0 | 1	0 | 0		0 | 0	0 | 0	1 | 1	0 | 0	0 | 0		0 | 2				4 | 0	0 | 0	0 | 1	0 | 5	0 | 0	85.71	92.86
rs1024116	0 | 0	0 | 0	0 | 0		2 | 0	0 | 0	0 | 0	0 | 0	0 | 1		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	1 | 0	98.57	91.43
rs10843344	1 | 0	0 | 0	0 | 0		1 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 1	0 | 4	0 | 0	92.86	90.00
rs182549	0 | 0	0 | 0	0 | 0		4 | 0	0 | 0	0 | 0	0 | 1	0 | 0		0 | 4				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	92.86	90.00
rs10141763	0 | 0	0 | 0	0 | 0		3 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	88.57
rs722098	1 | 1	0 | 0	0 | 0		0 | 0	0 | 0	2 | 0	0 | 0	0 | 0		0 | 0				5 | 0	0 | 0	0 | 0	0 | 1	0 | 0	97.14	88.57
rs5030240	0 | 0	2 | 0	0 | 0		1 | 0	0 | 0	0 | 2	0 | 0	0 | 1		1 | 0				5 | 0	0 | 1	0 | 0	0 | 1	0 | 0	92.86	87.14
rs2304925	0 | 0	1 | 0	1 | 0		0 | 0	0 | 0	2 | 0	0 | 0	0 | 1		1 | 0				2 | 0	0 | 0	0 | 5	1 | 1	1 | 0	90.00	87.14
rs917118	1 | 0	0 | 0	0 | 0		3 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				5 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	85.71
rs239031	2 | 1	1 | 0	0 | 0		2 | 0	1 | 0	0 | 4	0 | 1	0 | 0		0 | 0				2 | 0	2 | 0	1 | 1	2 | 2	0 | 5	80.00	81.43
Profile completeness	98.82	98.24	100		100	98.82	90.59		98.24		96.47				98.24	99.41	91.18	86.47	92.94	***96.3***	
Profile concordance	95.29	97.06	99.41		90.59	98.82	94.12		99.41		98.24				44.71	98.82	99.41	98.24	95.29		***93.5*** *(14 labs)*
																					97.5 *(13 labs)*

miscalls | no-calls 6.5% | 3.7%

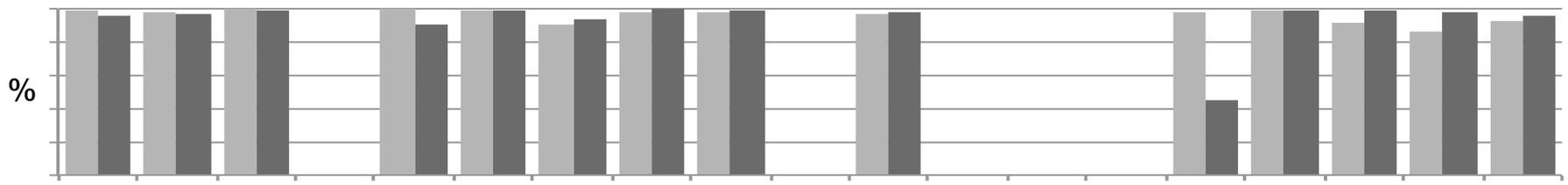
2.5% | 3.7%

Genotyping concordance: 34-plex, samples A-E

14 labs

34-plex SNPs ordered by decreasing genotyping reliability for participants

	1	2	4	5	6	7	8	9	11	12	13	14	15	16	17	18	19	20	21	Genotype completeness	Genotype concordance
rs2065160	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs3785181	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs896788	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				1 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.57
rs1573020	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				0 | 0	0 | 0	0 | 0	0 | 0	0 | 5	92.86	98.57
rs1426654	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs2572307	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs2814778	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs730570	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.14
rs1886510	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 3	0 | 0	0 | 5	0 | 0	0 | 0	88.57	97.14
rs3827760	0 | 0	0 | 1	0 | 0		0 | 0	0 | 2	0 | 4	0 | 1	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	0 | 0	87.14	97.14
rs2040411	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 0	1 | 0	100	95.71
rs2065982	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	95.71
rs7897550	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	95.71
rs1978806	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	1 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	95.71
rs2026721	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 1	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	1 | 0	97.14	95.71
rs16891982	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	94.29
rs12913832	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	94.29
rs1321333	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	94.29
rs773658	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 1	0 | 0	0 | 0	98.57	94.29
rs2303798	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	1 | 1	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	98.57	92.86
rs1335873	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	1 | 2	97.14	92.86
rs1498444	0 | 0	0 | 0	0 | 0		0 | 0	0 | 0	0 | 0	0 | 0	0 | 0		1 | 0				2 | 0	0 | 0	0 | 0	0 | 2	2 | 0	97.14	92.86
rs4540055	1 | 0	0 | 0	0 | 0		0 | 0	0 | 0	1 | 2	0 | 0	0 | 0		0 | 0				3 | 0	0 | 0	0 | 1	0 | 0	0 | 0	95.71	92.86
rs5997008	0 | 0	1 | 1	0 | 0		0 | 0	1 | 0	0 | 1	0 | 0	0 | 0		0 | 0				2 | 0	0 | 0	0 | 0	0 | 1	1 | 0	95.71	92.86
rs881929	0 | 0	0 | 1	0 | 0		0 | 0	0 | 0	1 | 1	0 | 0	0 | 0		0 | 2				4 | 0	0 | 0	0 | 1	0 | 5	0 | 0	85.71	92.86
rs1024116	0 | 0	0 | 0	0 | 0		2 | 0	0 | 0	0 | 0	0 | 0	0 | 1		0 | 0				3 | 0	0 | 0	0 | 0	0 | 0	1 | 0	98.57	91.43
rs10843344	1 | 0	0 | 0	0 | 0		1 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 1	0 | 4	0 | 0	92.86	90.00
rs182549	0 | 0	0 | 0	0 | 0		4 | 0	0 | 0	0 | 0	0 | 1	0 | 0		0 | 4				3 | 0	0 | 0	0 | 0	0 | 0	0 | 0	92.86	90.00
rs10141763	0 | 0	0 | 0	0 | 0		3 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				4 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	88.57
rs722098	1 | 1	0 | 0	0 | 0		0 | 0	0 | 0	2 | 0	0 | 0	0 | 0		0 | 0				5 | 0	0 | 0	0 | 0	0 | 1	0 | 0	97.14	88.57
rs5030240	0 | 0	2 | 0	0 | 0		1 | 0	0 | 0	0 | 2	0 | 0	0 | 1		1 | 0				5 | 0	0 | 1	0 | 0	0 | 1	0 | 0	92.86	87.14
rs2304925	0 | 0	1 | 0	1 | 0		0 | 0	0 | 0	2 | 0	0 | 0	0 | 1		1 | 0				2 | 0	0 | 0	0 | 5	1 | 1	1 | 0	90.00	87.14
rs917118	1 | 0	0 | 0	0 | 0		3 | 0	0 | 0	1 | 0	0 | 0	0 | 0		0 | 0				5 | 0	0 | 0	0 | 0	0 | 1	0 | 0	98.57	85.71
rs239031	2 | 1	1 | 0	0 | 0		2 | 0	1 | 0	0 | 4	0 | 1	0 | 0		0 | 0				2 | 0	2 | 0	1 | 1	2 | 2	0 | 5	80.00	81.43
Profile completeness	98.82	98.24	100		100	98.82	90.59		98.24		96.47				98.24	99.41	91.18	86.47	92.94	***96.3***	
Profile concordance	95.29	97.06	99.41		90.59	98.82	94.12		99.41		98.24				44.71	98.82	99.41	98.24	95.29		



93.5 *(14 labs)*

97.5 *(13 labs)*

% Profile/genotype completeness

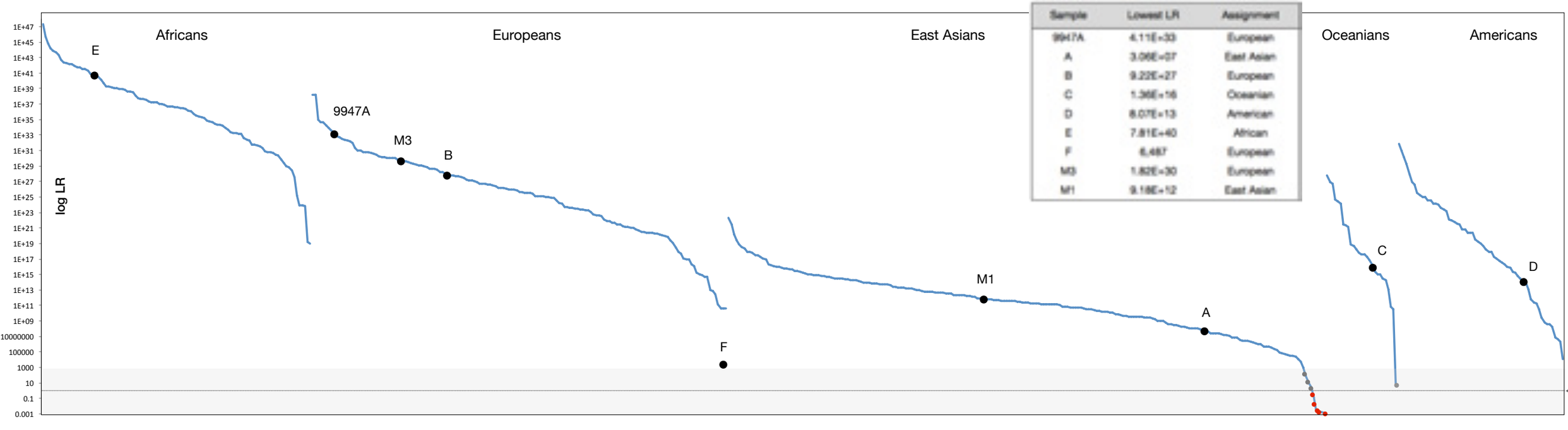
% Profile/genotype concordance

46 Indels Test DNAs A-E: **19 labs**

miscalls | no-calls	1	2	4	5	6	7	8	9	11	12	13	14	15	16	17	18	19	20	21	Genotype completeness	Genotype concordance
rs2307666	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs1610863	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs16635	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs1610965	**1 | 0**	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.95
rs35451359	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs140837	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs1160893	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs2308203	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs33974167	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs1160852	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs1610884	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	**0 | 1**	0 | 0	0 | 0	98.95	100
rs2067280	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs2308067	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	**0 | 1**	0 | 0	0 | 0	98.95	100
rs4183	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs3054057	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs2307840	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs60612424	**1 | 0**	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.95
rs3033053	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs16384	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs34611875	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs1610859	**0 | 1**	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	98.95	100
rs3045215	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs25621	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs2307832	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs16343	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs3031979	**0 | 1**	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	98.95	100
rs34122827	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	**2 | 0**	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.89
rs133052	**1 | 0**	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	**1 | 0**	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	97.89
rs6490	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs4181	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs3030826	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs140708	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs1611026	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	**1 | 0**	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	98.95
rs16438	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs2308161	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs16687	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs2307998	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs2307803	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs2307930	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs25630	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs2307582	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs2307922	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	**0 | 3**	0 | 0	96.84	100
rs11267926	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs25584	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs2307799	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
rs34541393	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	0 | 0	100	100
Profile completeness	99.1	100	100	100	100	100	100	100	100	100	100	100	100	100	100	100	99.1	98.5	100	***99.8***	
Profile concordance	98.7	100	100	100	100	98.7	100	100	100	100	99.6	100	100	100	100	100	100	100	100		***99.8***

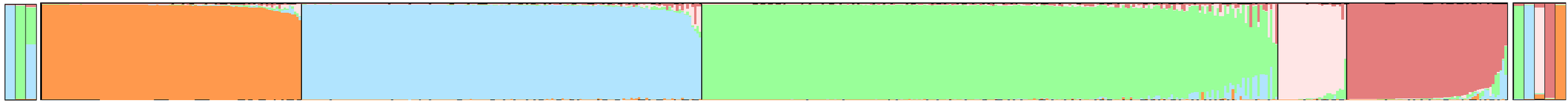
Statistical analysis of ancestry: three best approaches

Bayes analysis

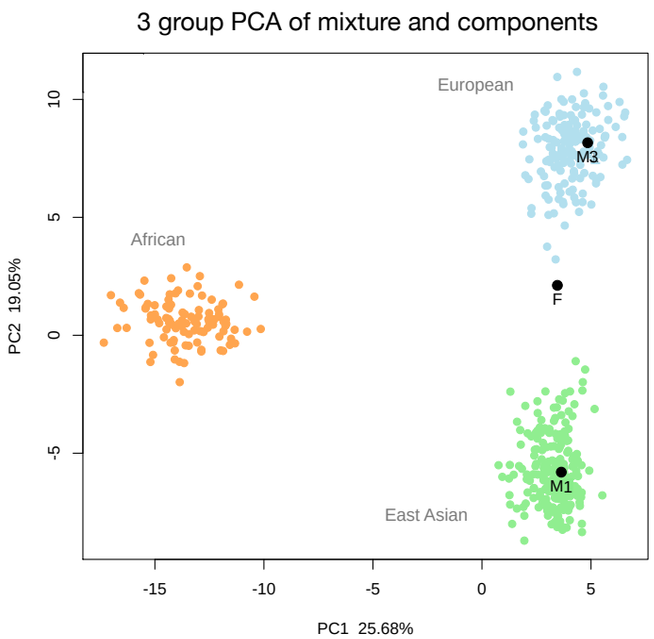


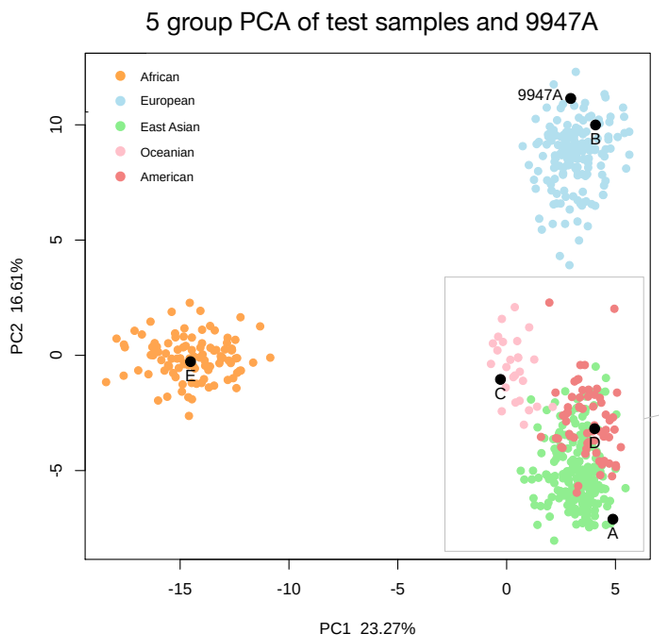
Sample	Lowest LR	Assignment
9947A	4.11E+33	European
A	3.06E+27	East Asian
B	9.22E+27	European
C	1.36E+16	Oceanian
D	8.07E+13	American
E	7.81E+40	African
F	6.487	European
M3	1.82E+30	European
M1	9.18E+12	East Asian

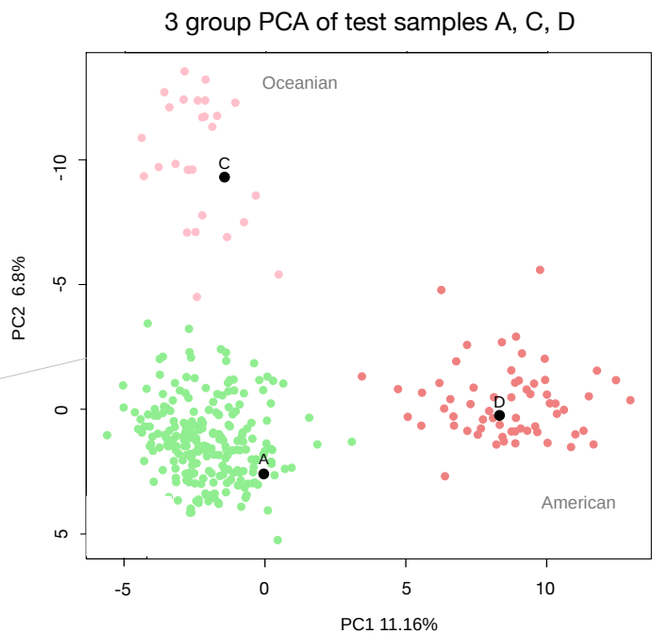
STRUCTURE



PCA







Statistical analysis of ancestry: Bayes and PCA

Binary AIM classification of multiple individuals
with an Excel file of populations (.xlsx format)

Step 1: Data input (population)

Select your local Excel file of populations in the required format (hybrid profile and training set file). You can download a valid example file if necessary clarify ideas, or use the input file generator wizard.

Datei auswählen | Keine Datei ausgewählt

Step 2: Choose classifier

- (•) Naive Bayes (Hardy-Weinberg principle applies)
- ( ) Naive Bayes (Hardy-Weinberg principle need not apply)
- ( ) Genetic distance algorithm

Get PCA graph and classify

Binary AIM classification of individuals as
Europe-East Asia-Africa-America-Oceania (34 SNPs or 46 Indels or both)

Step 1: Choose marker set

- (•) 34-plex (previous) with rs727811.
- ( ) 34-plex (revised) with rs3827760.
- ( ) 46-plex AIM-Indels.
- ( ) 80-plex AIM-Indels (all of the above).

Step 2: Choose populations

- (•) 3 populations (Europe, East Asia, Africa).
- ( ) 4 populations (Europe, East Asia, Africa, America).
- ( ) 5 populations (Europe, East Asia, Africa, America, Oceania).

Step 3: Choose classifier

- (•) Naive Bayes (Hardy-Weinberg principle applies)
- ( ) Naive Bayes (Hardy-Weinberg principle need not apply)
- ( ) Multinomial logistic regression
- ( ) Genetic distance algorithm

Step 4: Data input

Type either **34 SNPs or/and 46 Indels of the individual to classify** as appropriate. For the required SNP/Indel (rs) numbers, see graphics on left. For instance, the following 34 SNP profile would be valid in the case of 34-plex (previous) with rs727811, and 3 populations:

AAAACTGGCCAATTCCAACCCCAGTTGGAAAAAGCCTTTTGGTTCCCTCGCCAGACTTCCCTGTCCAG

Statistical analysis of ancestry: Bayes and PCA

Binary AIM classification of multiple individuals
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Datei auswählen | Keine Datei ausgewählt

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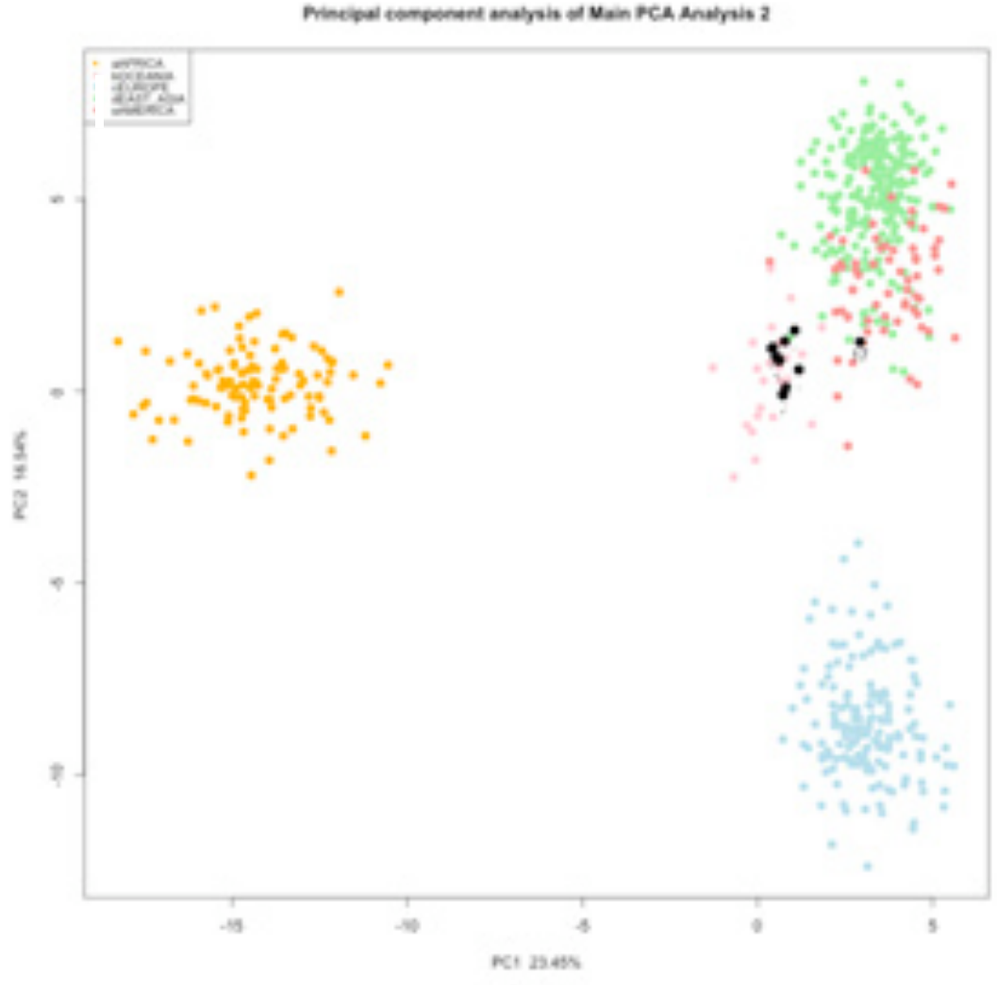
Get PCA graph and classify



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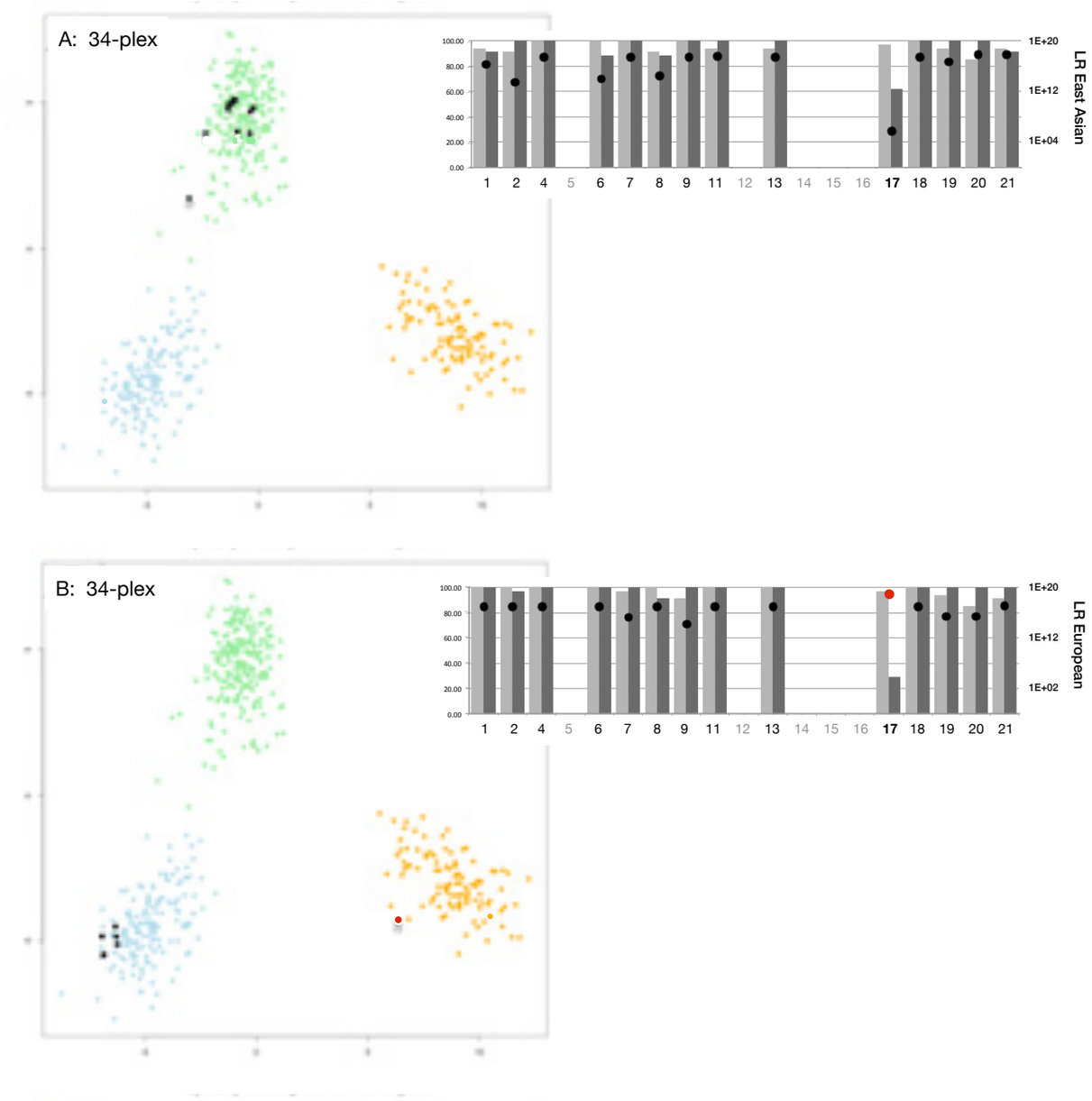
- ( ) Multinomial logistic regression
- ( ) Genetic distance algorithm

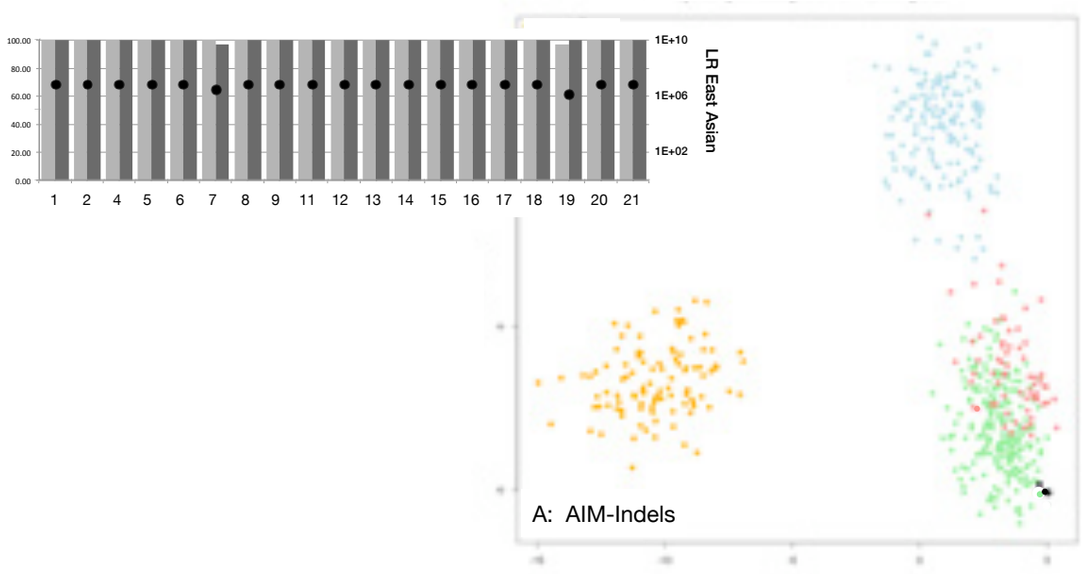
Step 4: Data input

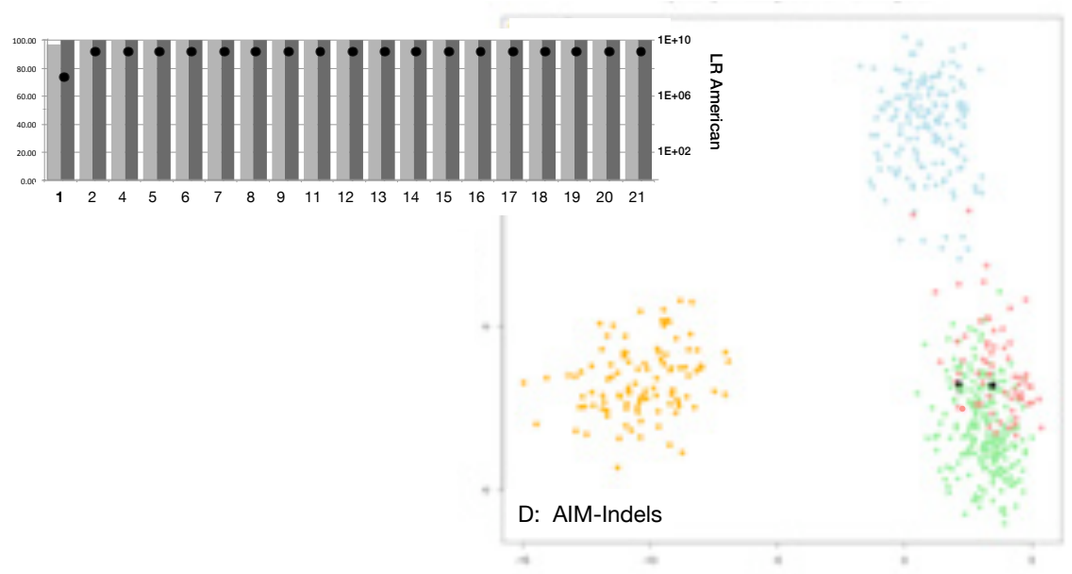
Type either **34 SNPs or/and 46 Indels of the individual to classify** as appropriate. For the required SNP/Indel (rs) numbers, see graphics on left. For instance, the following 34 SNP profile would be valid in the case of 34-plex (previous) with rs727811, and 3 populations:

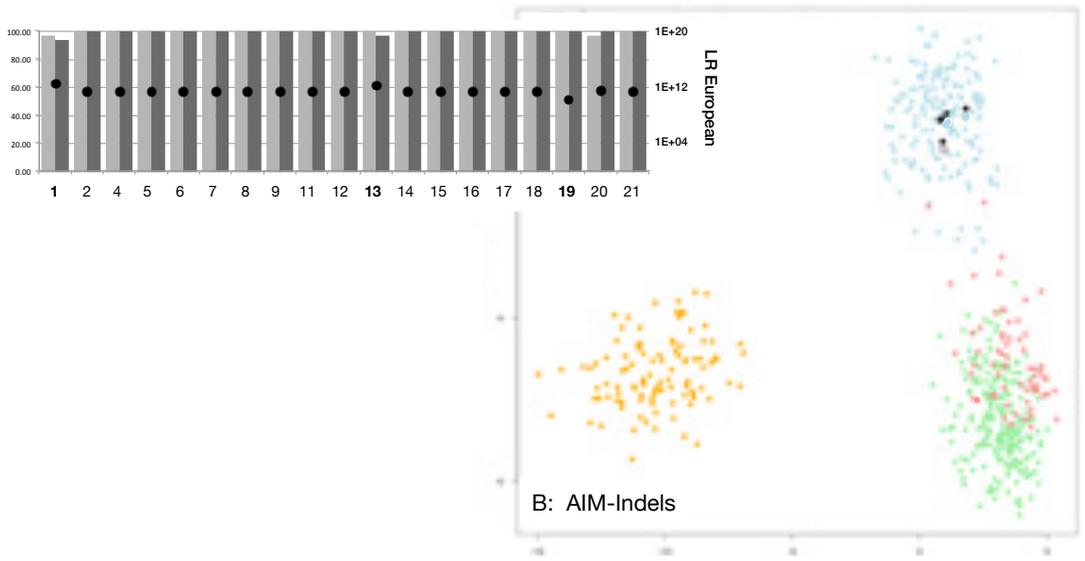
AAAACTGGCCAATTCCAACCCCAGTTGGAAAAAGCCTTTTGGTTCCCTCGCCAGACTTCCCTGTCCAG

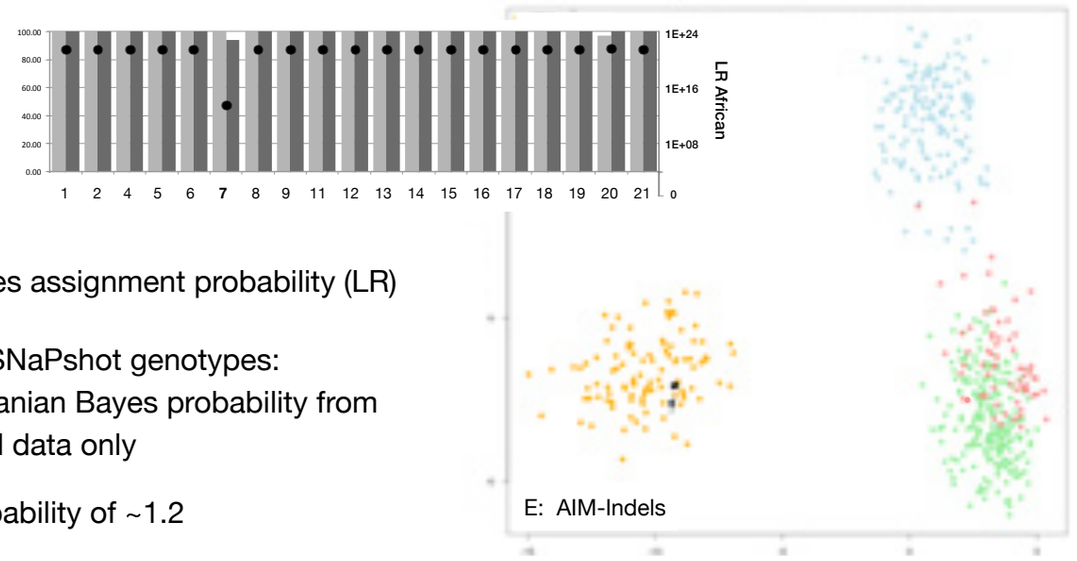


34-plex PCAs, Bayes LRs and quality







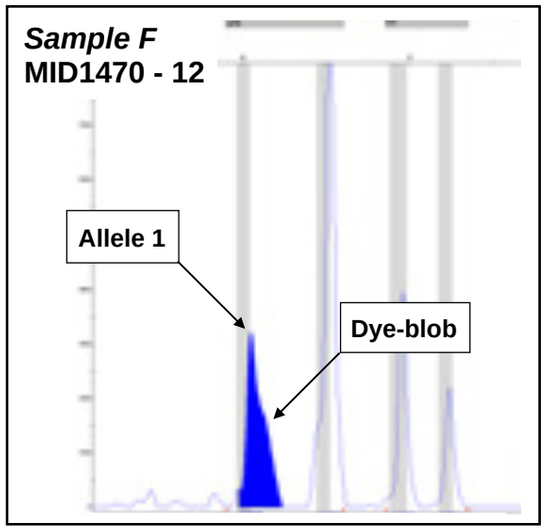


- Bayes assignment probability (LR)
- No SNaPshot genotypes: Oceanian Bayes probability from Indel data only
- Probability of ~1.2

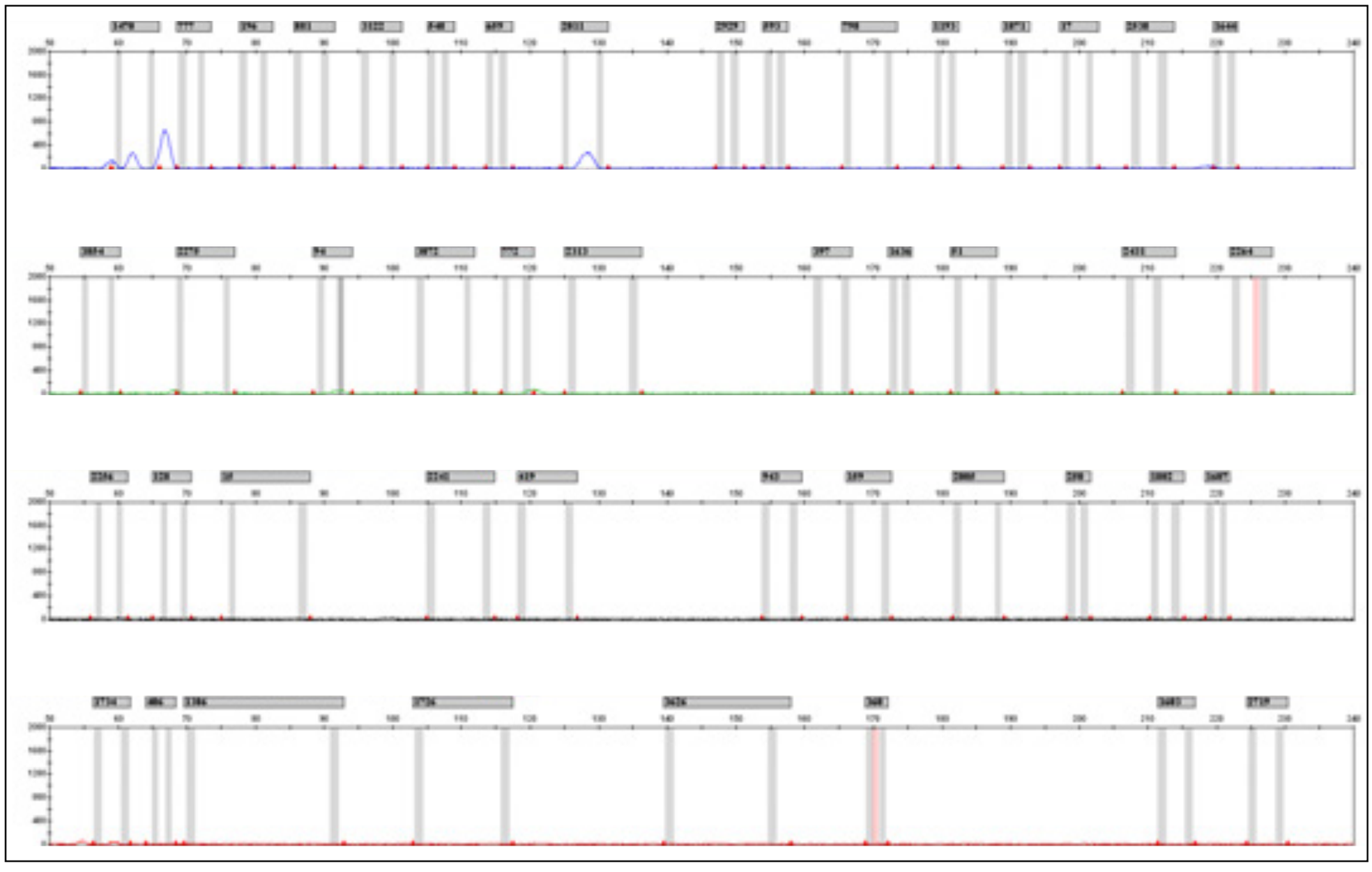
Indels

Indels: known PCR-to-CE profile issues

A. Some dye-blobs are present

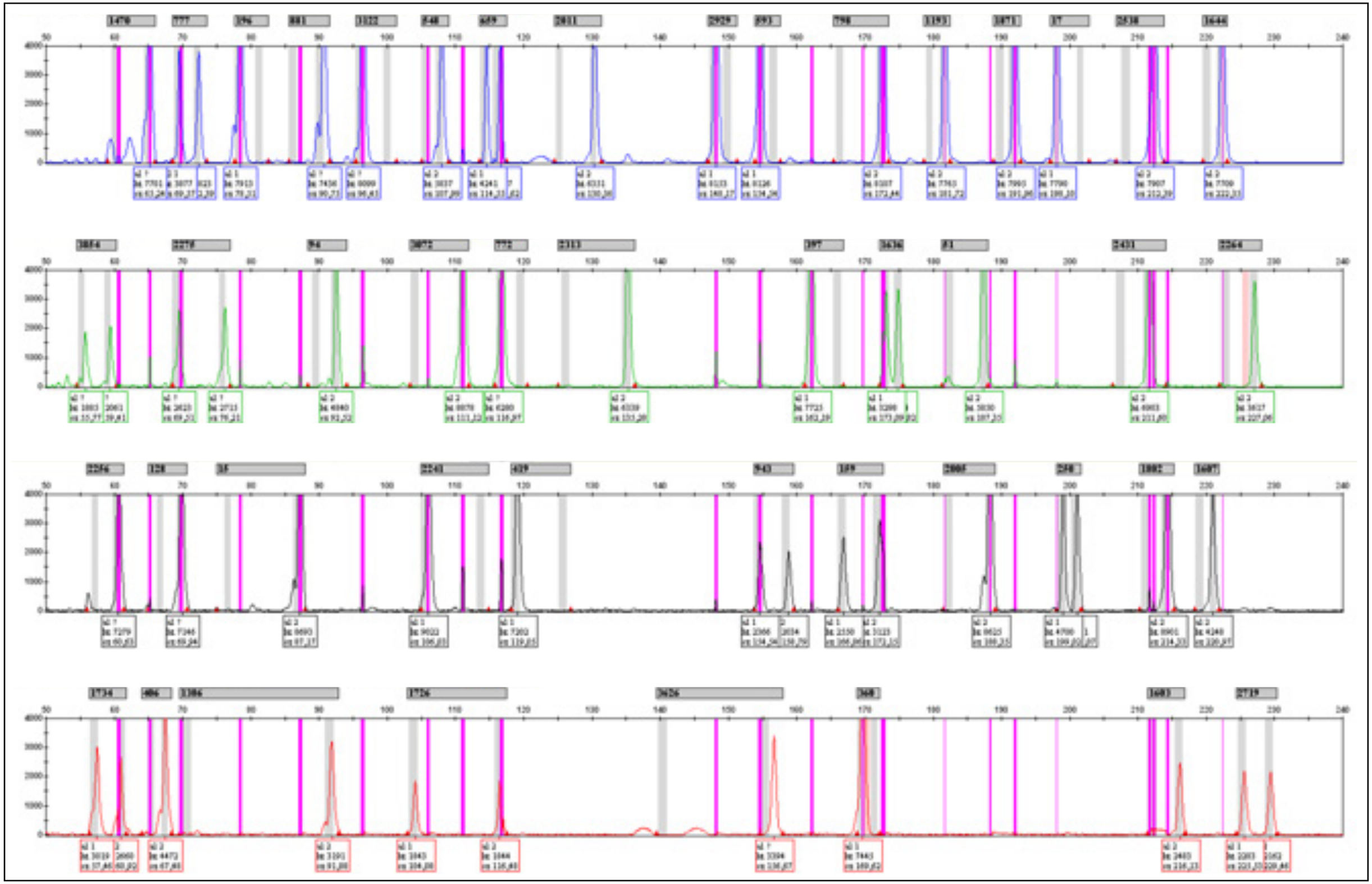


Dye-blobs in NTC



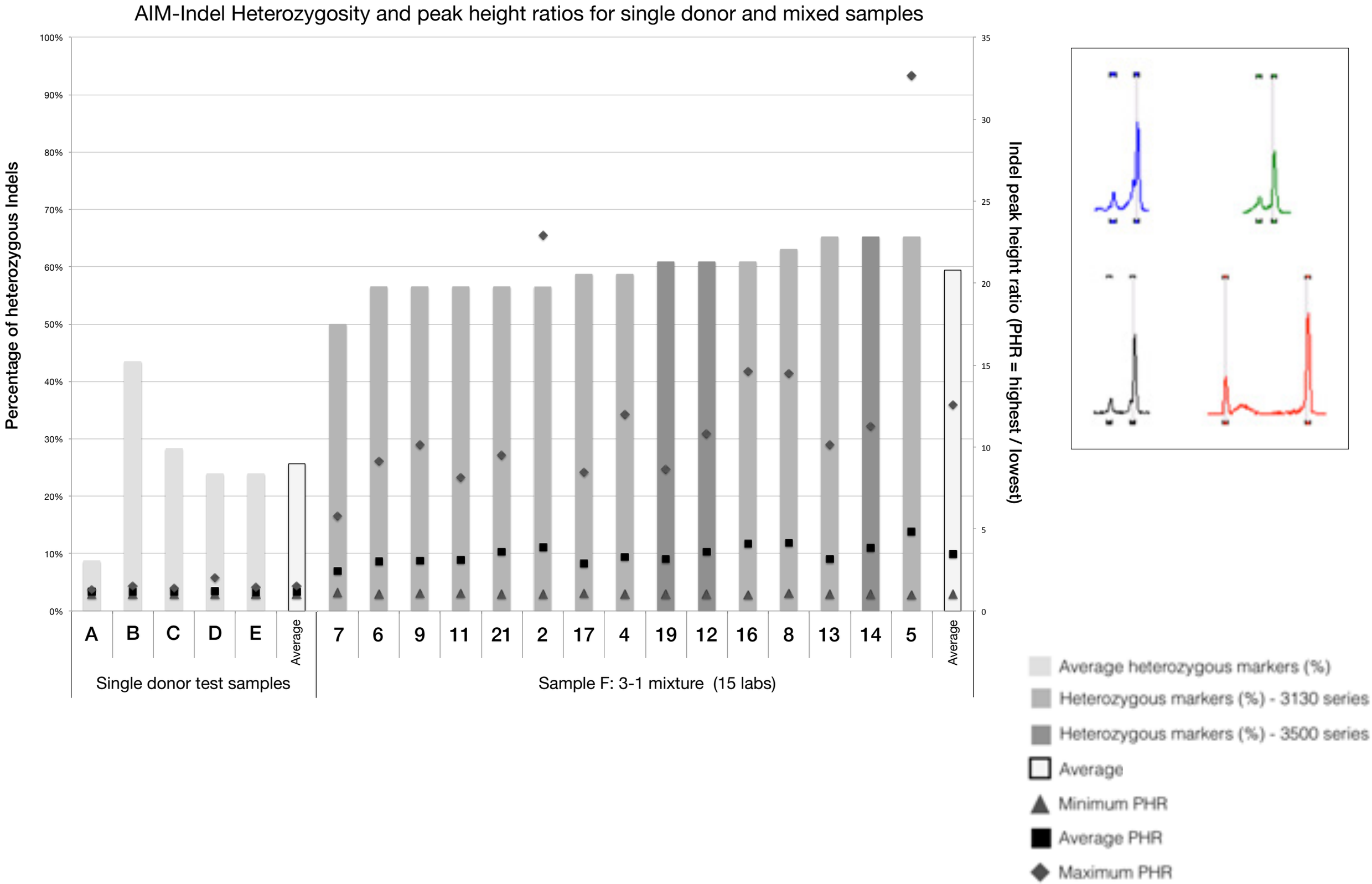
Indels: known PCR-to-CE profile issues

B. Pull-ups are frequent



Mixtures: 15 labs provided PHR data from Indels

a)



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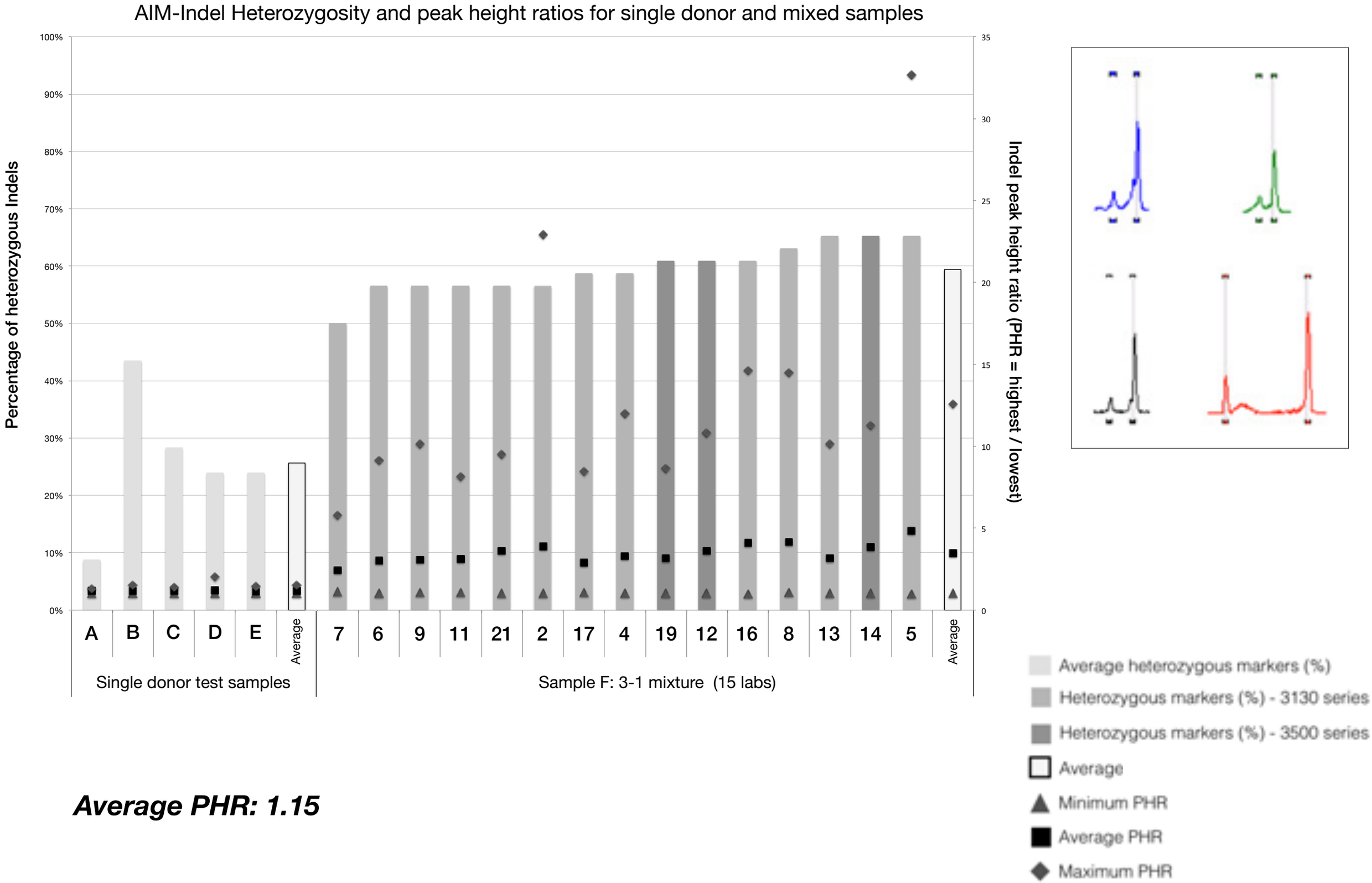

Average PHR: 1.15

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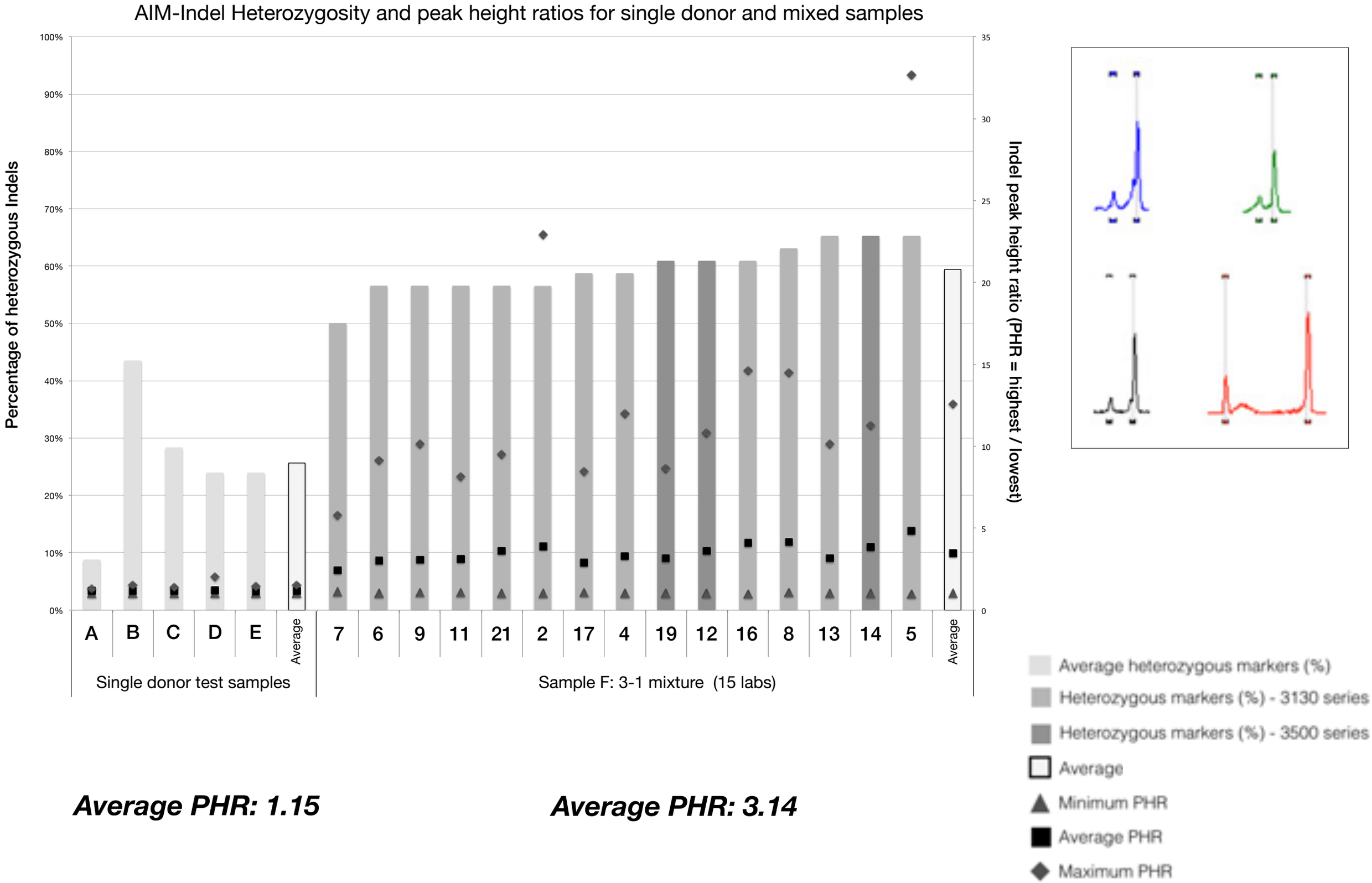
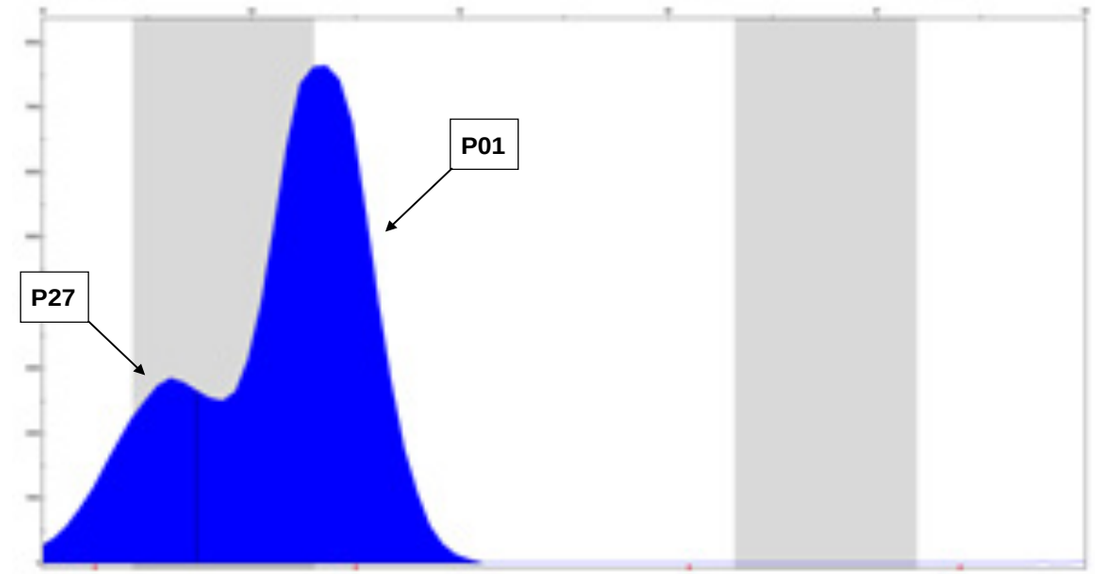


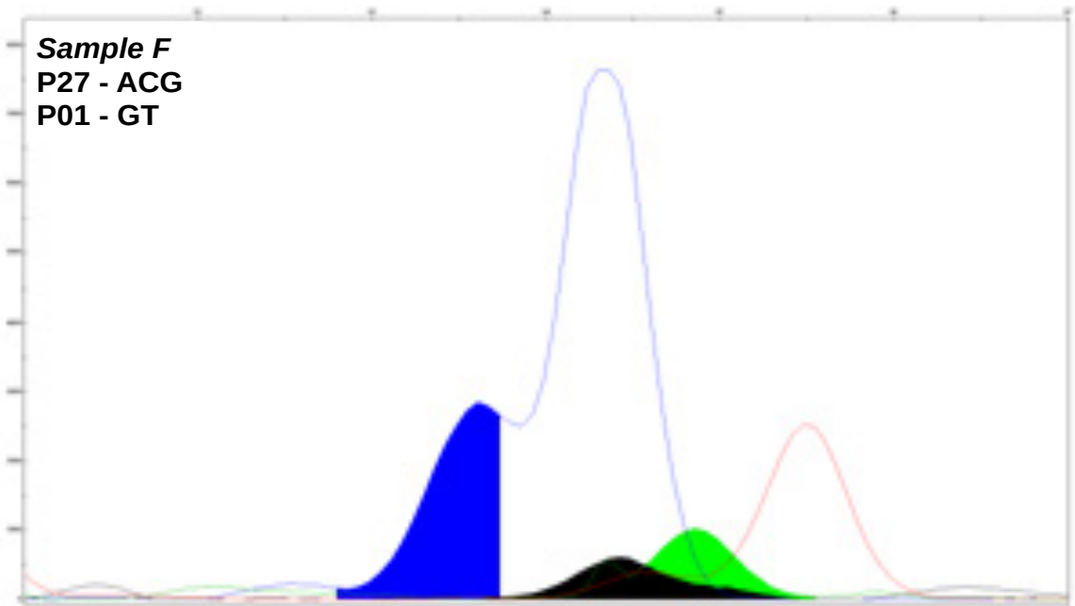
Average PHR: 1.15

Average PHR: 3.14

Mixtures: One 34-plex triallelic SNP has three peaks

Tri-allelic P27 (rs5030240) has 3 alleles present in sample F





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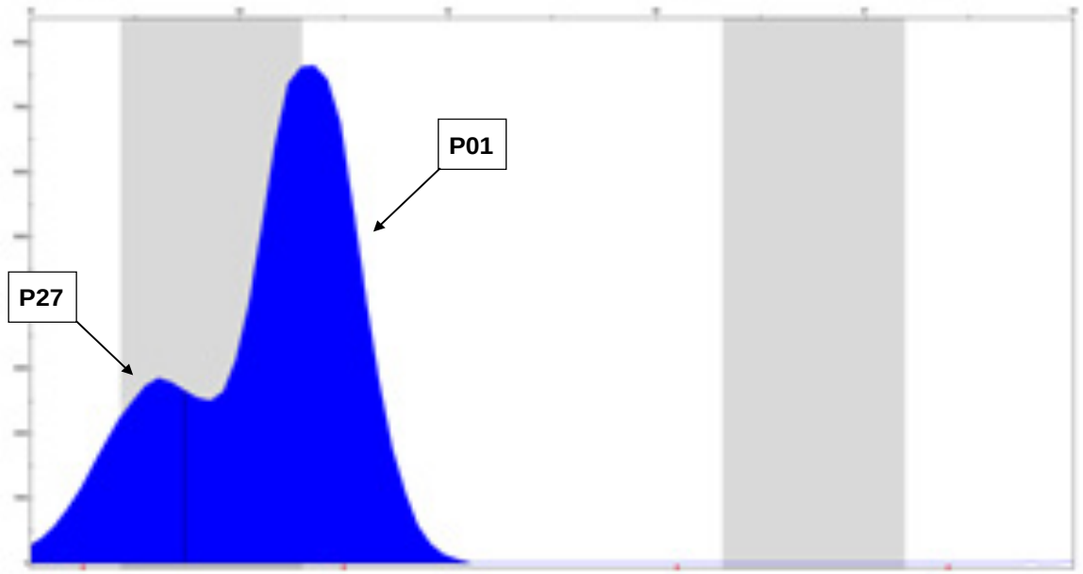

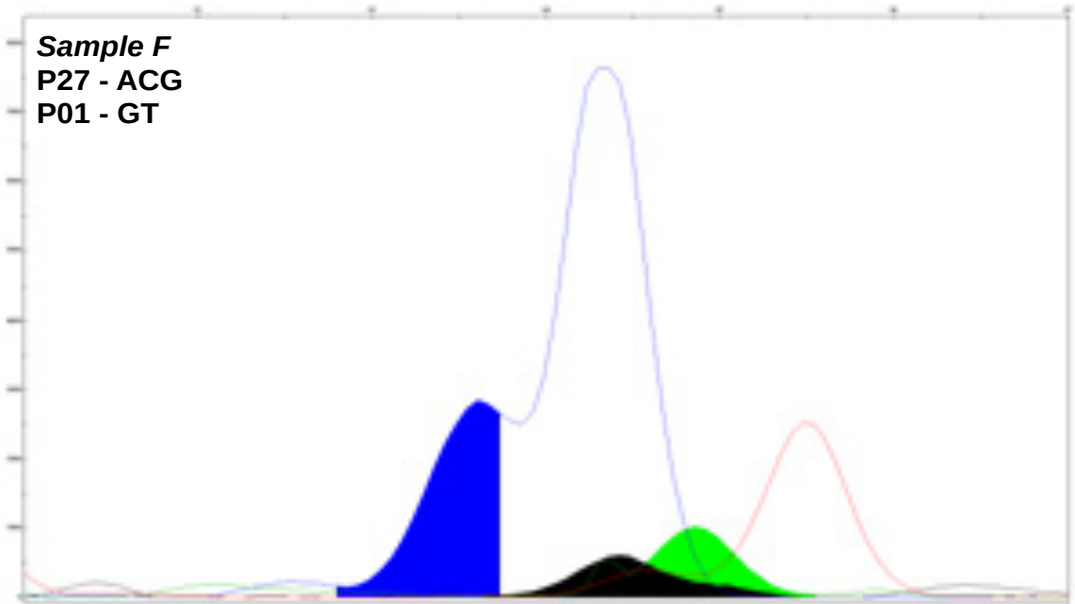

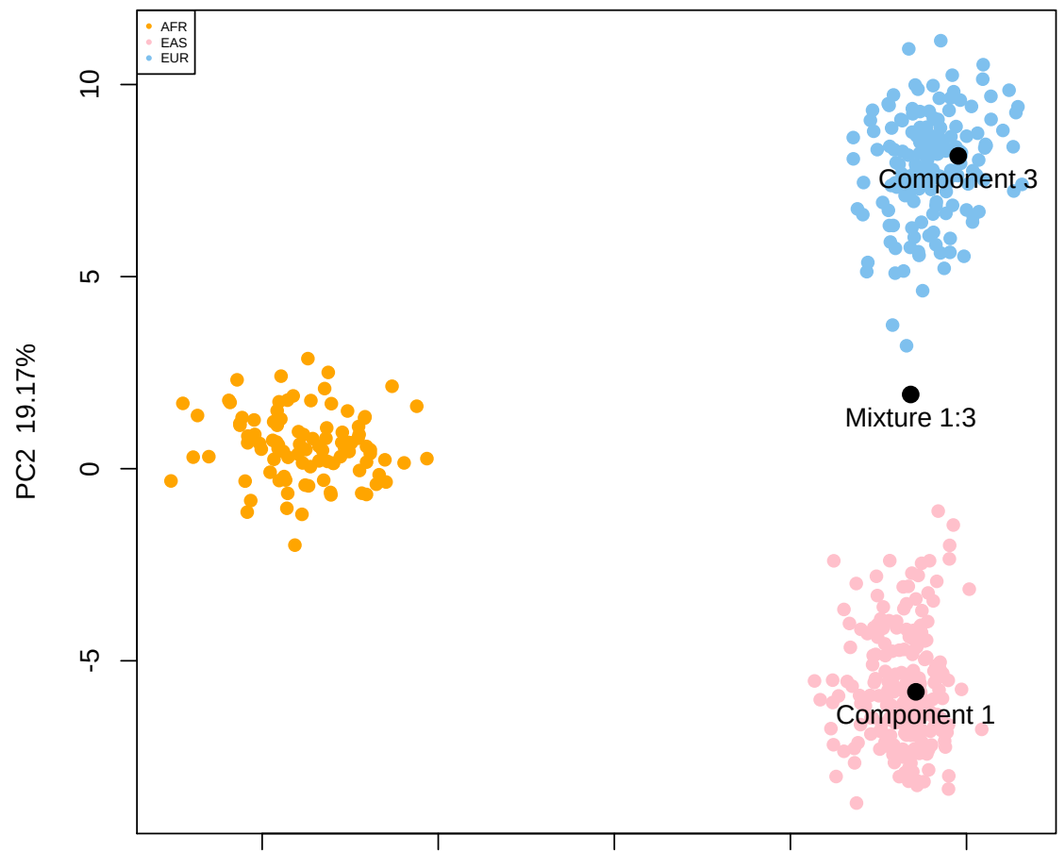

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- 18/19 labs achieved correct ancestry assignments for samples A-E from Bayes and PCA statistical analysis.

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- Applied a standardized validation program to the Ion PGM 169-SNP forensic identification set: ‘HID SNP’
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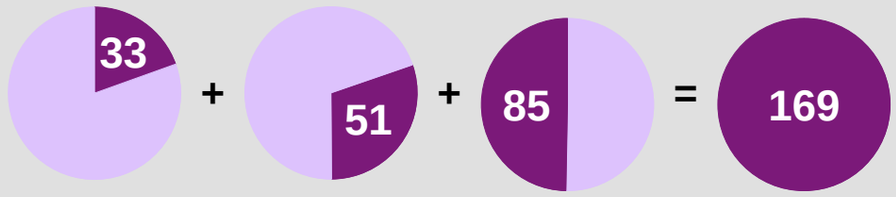
- Scrutinized context sequence for previously uncharted features such as flanking SNPs or indels that can disrupt alignments.

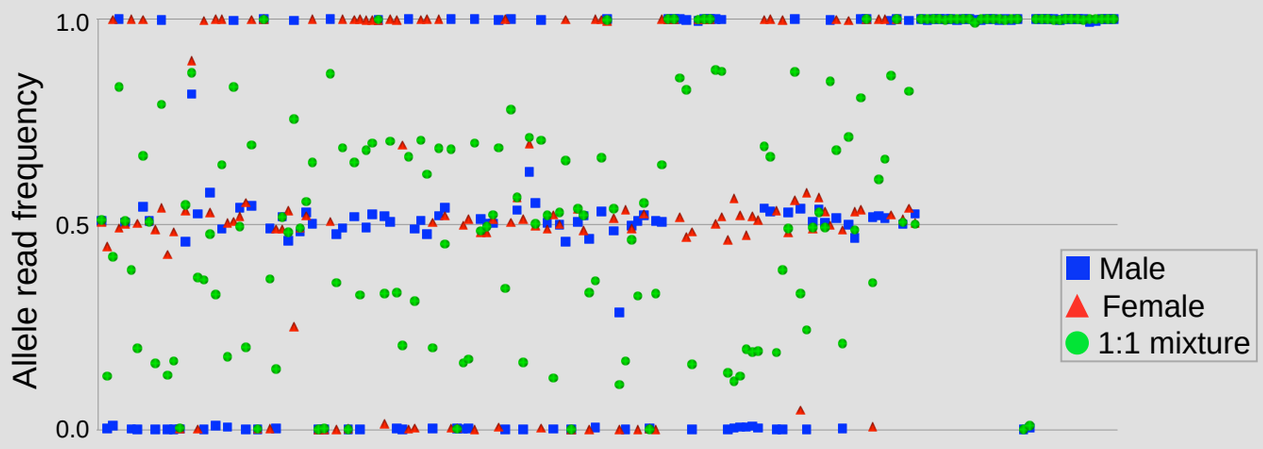
EDNAP AIMs Exercise 2014 - Conclusions

Graphical Abstract (for review)

HID-Ion AmpliSeq™ Identity Panel v2.2



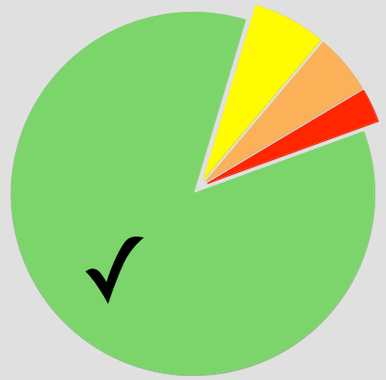
Mixture detection



Inter-run | Inter-lab | With databases

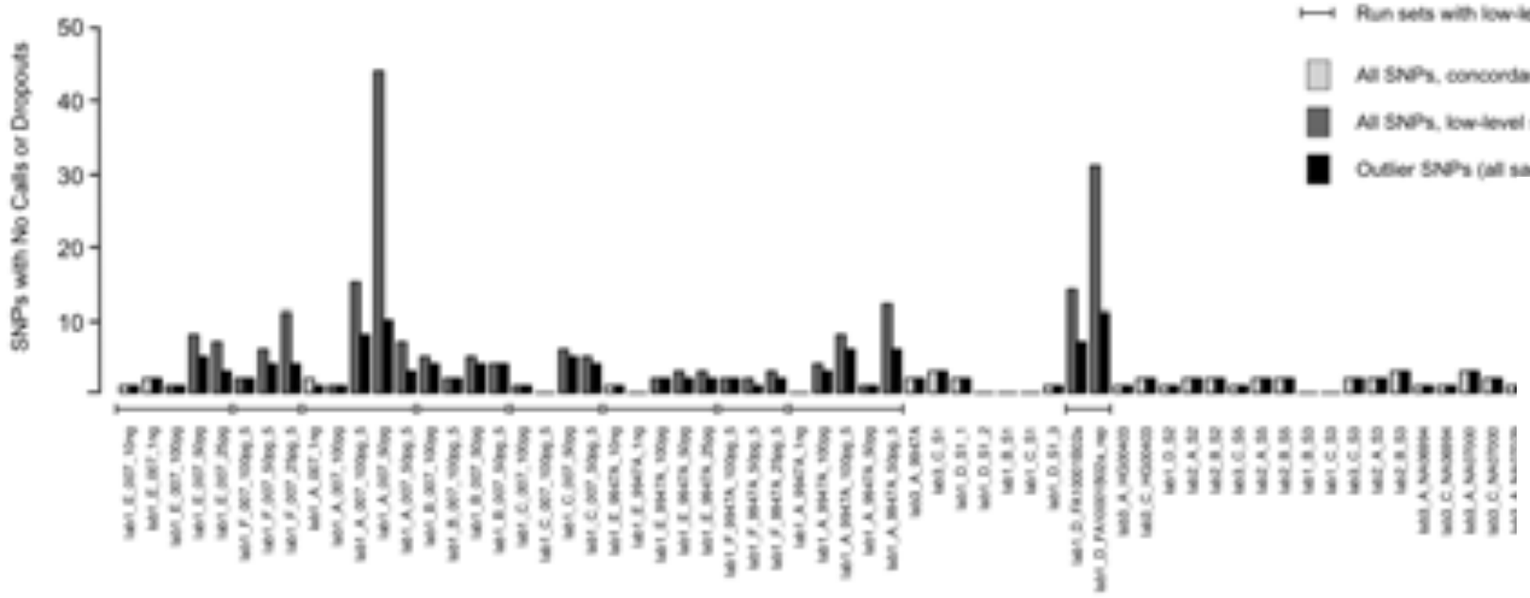
Overall concordance 99.8%

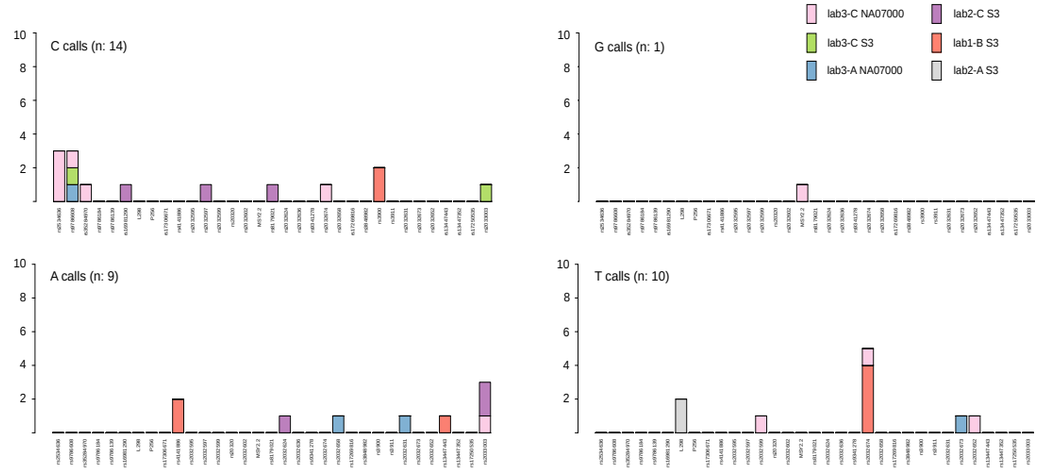
- Concordant genotypes
- No-call
- Discordant genotypes



Outlier SNPs

Atypical	No-calls	Discordant
rs1979255	rs1004357	rs938283
rs2032597	rs2399332	





1000 Genomes final public SNP data release

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NGS AIMs Panels

- Illumina MiSeq *FGx* panel: 55 AIMs (2/55 FDP)
- Ion PGM: 165 AIMs panel
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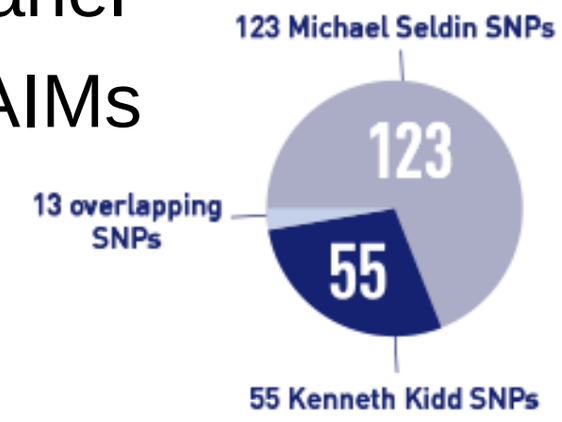

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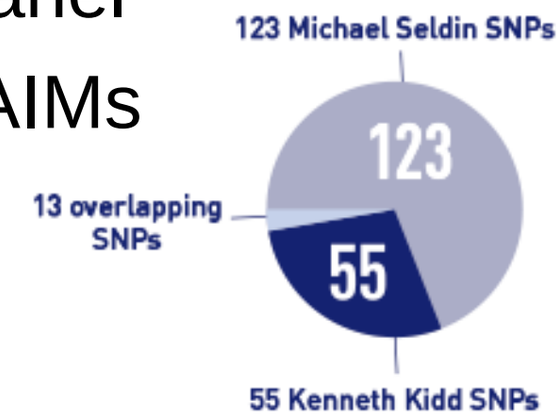

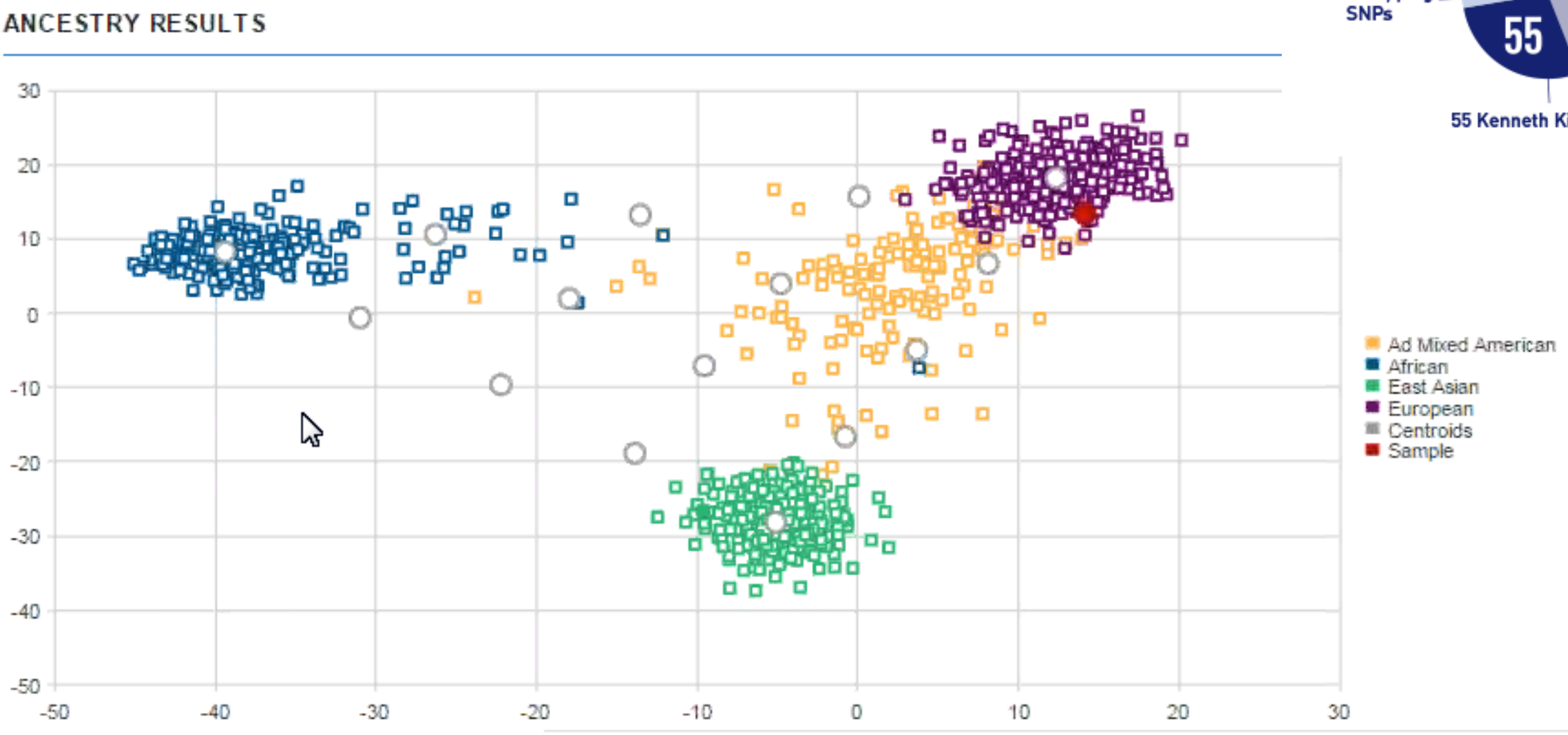

DISTANCE TO NEAREST CENTROID

5.10

SNP PANEL USED

› 1000genomes populations with samples in centroid with sample

› Reference samples in centroid with sample

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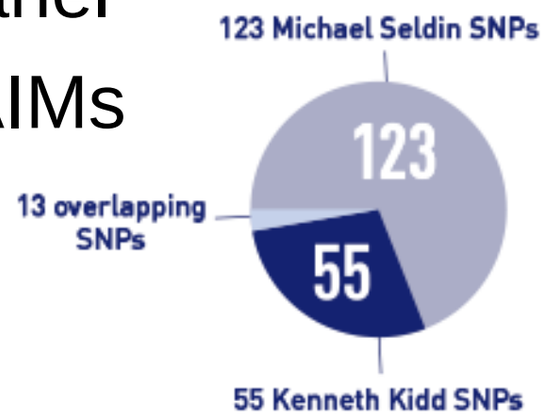

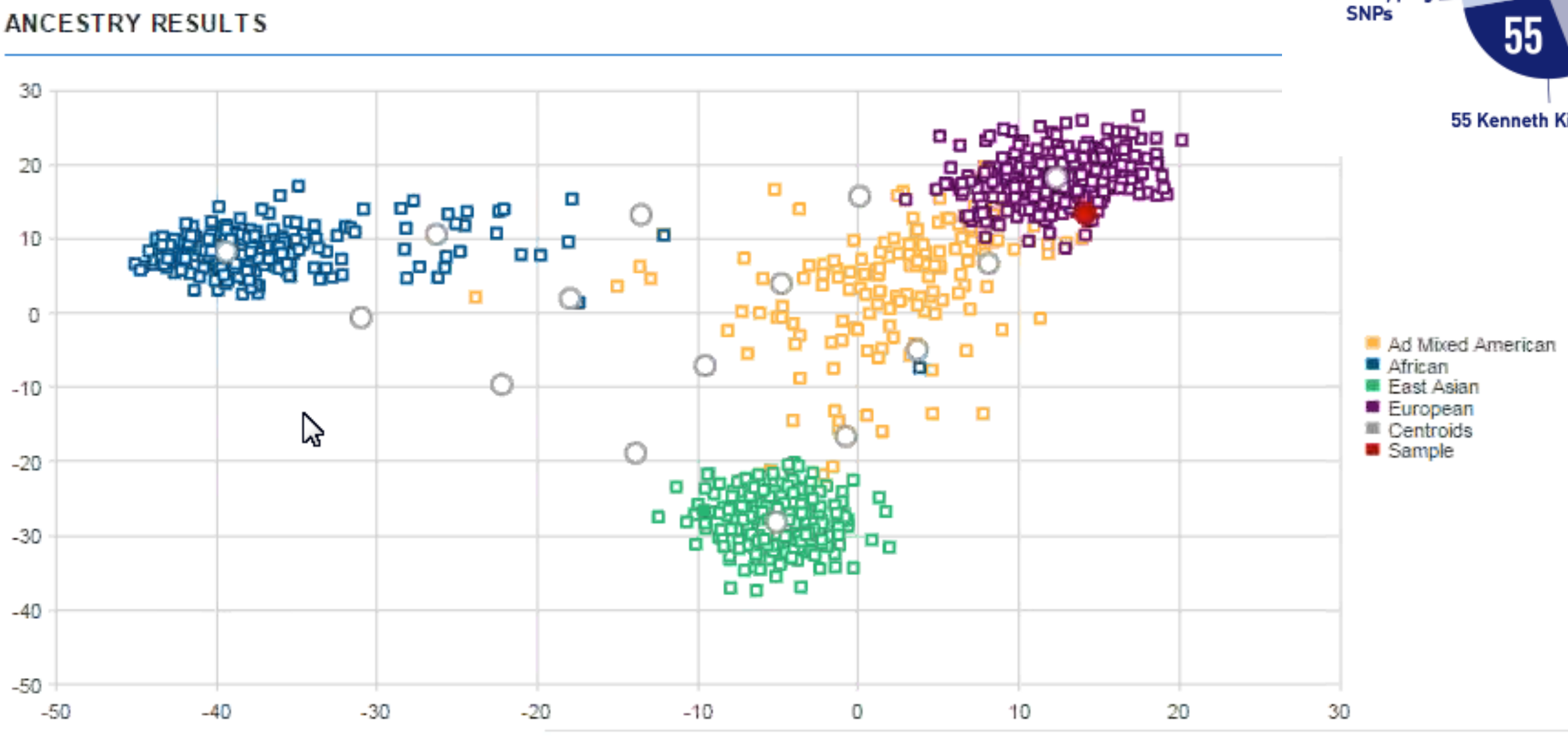

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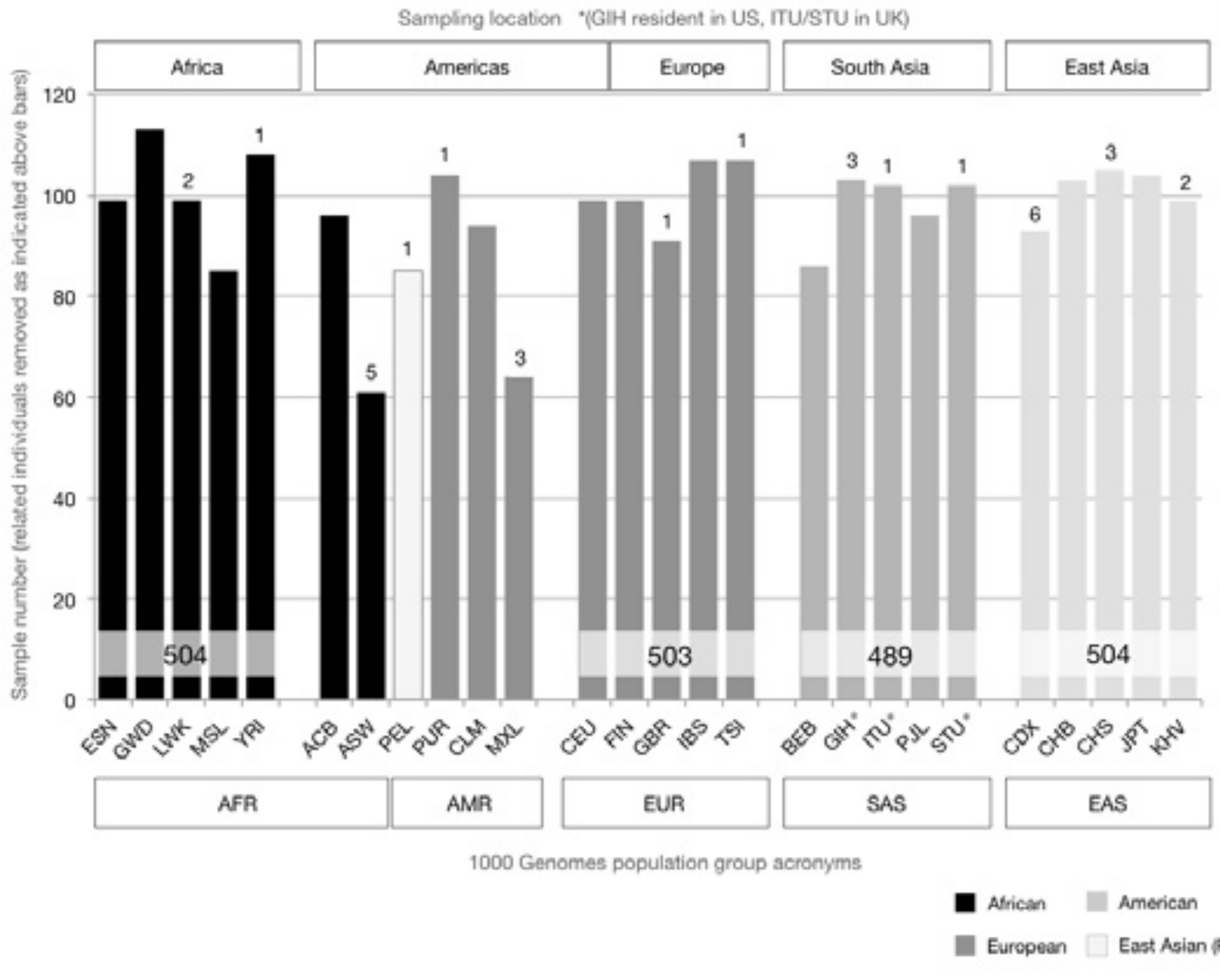
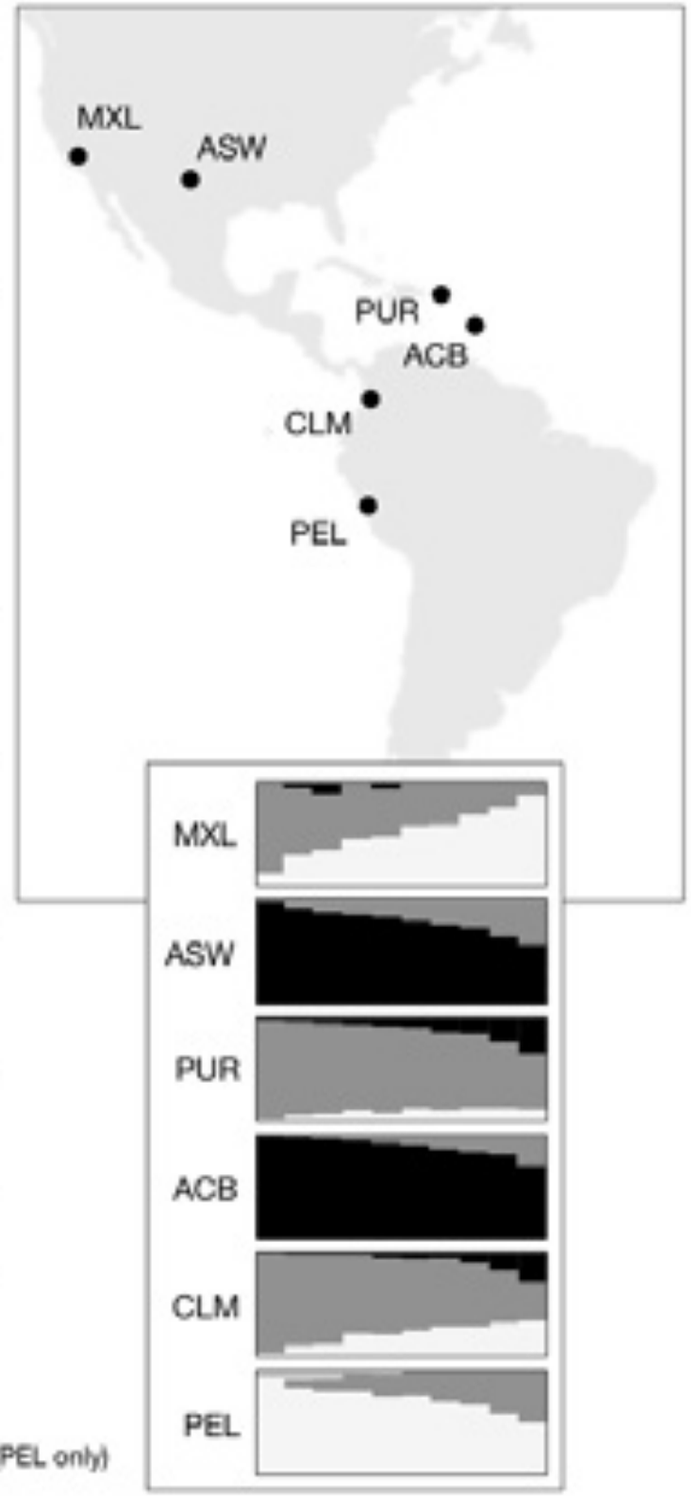
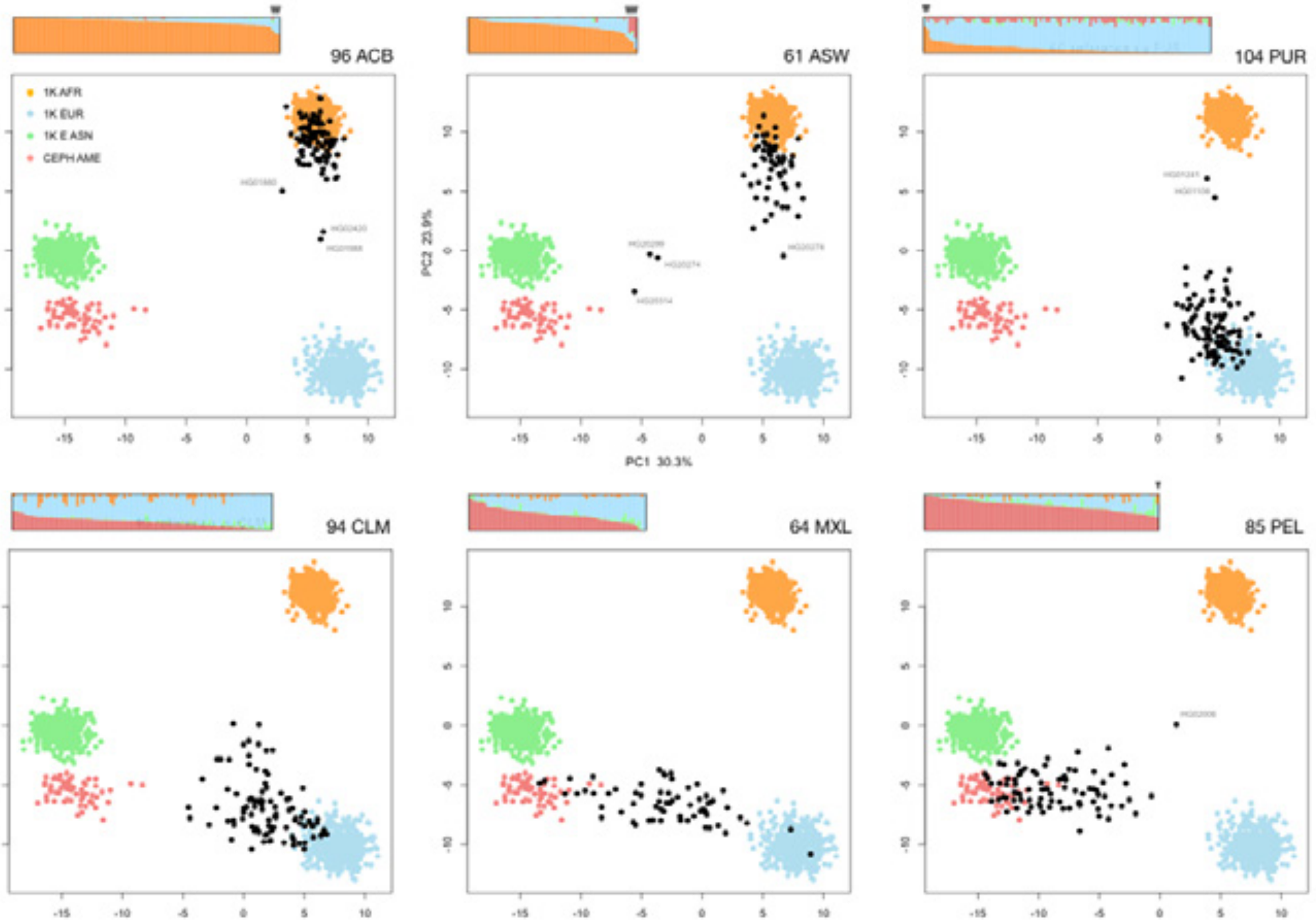





1000 Genomes Africans
1000 Genomes Europeans
1000 Genomes East Asians
Ame.
ACB
ASW
PUR
CLM
MXL
PEL
K:4


96 ACB
61 ASW
104 PUR
94 CLM
64 MXL
85 PEL
1K.AFR
1K.EUR
1K.E.ASN
CEPH.AME
PC2 23.9%
PC1 30.3%

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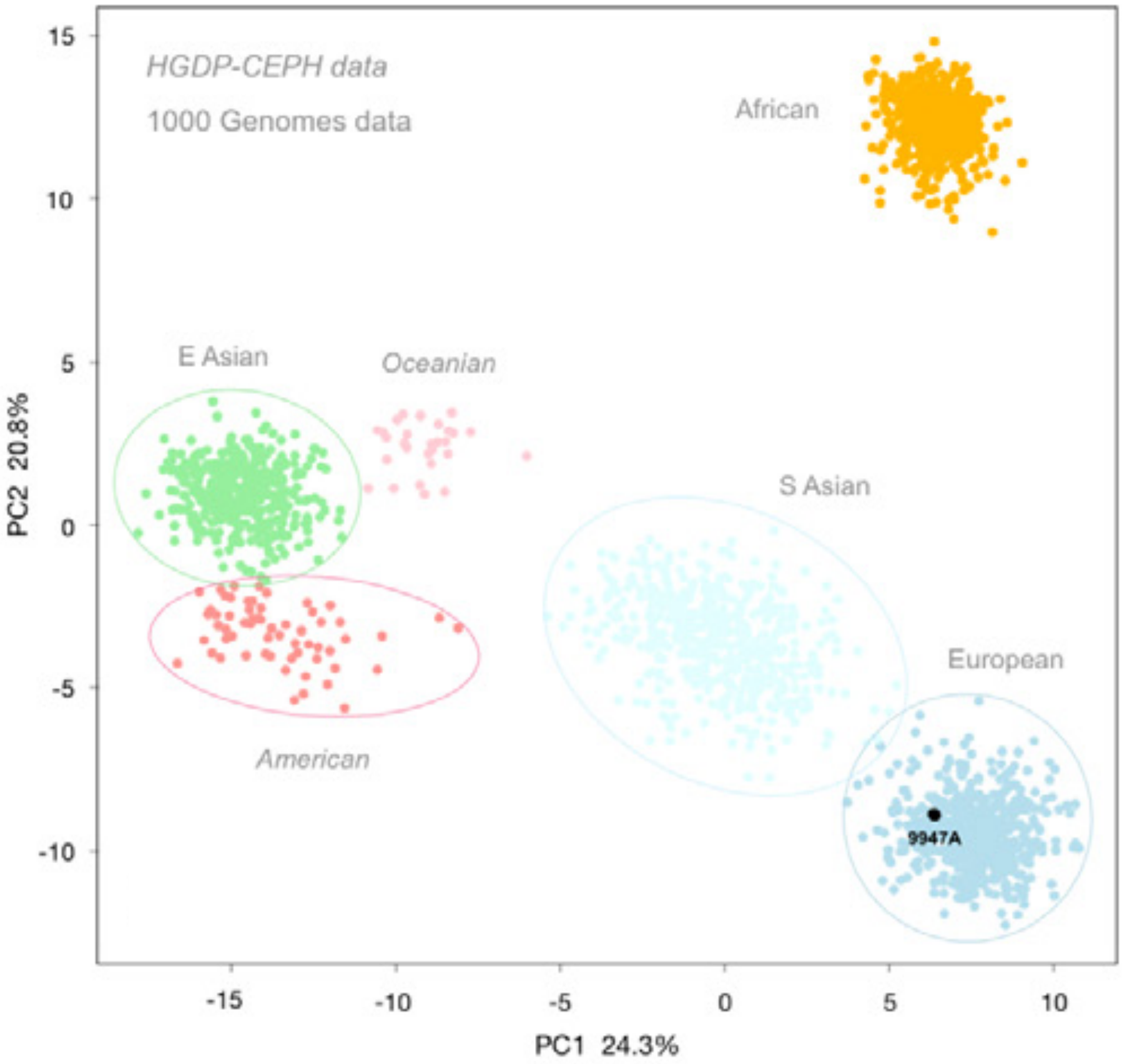

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Coding : non-coding status of 80 AIMs

	ID	Chr	Position	Gene	Functional Class
1	rs2307666	11	64729920	C11orf85	
2	rs1610863	16	6551830	RBFOX1	
3	rs16635	6	99789775	FAXC	
4	rs1610965	5	79746093	ZFYVE16	
5	rs35451359	18	45110983	-	
6	rs140837	6	3708907	-	
7	rs1160893	2	2.25E+08	WDFY1	
8	rs2308203	2	1.09E+08	RANBP2	
9	rs33974167	8	87813725	-	
10	rs1160852	6	1.37E+08	IL20RA	
11	rs1610884	5	56122323	MAP3K1	
12	rs2067280	5	89818959	LYSMD3	
13	rs2308067	7	1.27E+08	SND1	
14	rs4183	3	3192524	CRBN	
15	rs3054057	15	86010538	AKAP13	
16	rs2307840	1	36099090	PSMB2	
17	rs60612424	6	84017510	ME1	
18	rs3033053	14	42554496	-	
19	rs16384	22	42045009	XRCC6	
20	rs34611875	18	67623917	CD226	
21	rs1610859	5	1.28E+08	SLC27A6	
22	rs3045215	1	2.35E+08	IRF2BP2	
23	rs25621	6	1.4E+08	-	
24	rs2307832	1	55590788	USP24	
25	rs16343	4	17635560	FAM184B	
26	rs3031979	8	73501951	KCNB2	
27	rs34122827	13	63778778	-	
28	rs133052	22	41042364	-	
29	rs6490	12	1.08E+08	PRDM4	
30	rs4181	2	42577803	COX7A2L	
31	rs3030826	6	67176774	-	
32	rs140708	6	1.71E+08	-	
33	rs1611026	5	82545545	XRCC4	
34	rs16438	20	25278470	PYGB	
35	rs2308161	10	69800909	HERC4	
36	rs16687	7	83887878	-	
37	rs2307998	5	7814345	ADCY2	
38	rs2307803	3	1.09E+08	-	
39	rs2307930	6	84476378	-	
40	rs25630	6	14734341	-	
41	rs2307582	1	2.48E+08	OR2G3	
42	rs2307922	1	39896964	MACF1	
43	rs11267926	15	45526069	-	
44	rs25584	12	1.12E+08	ACAD10	
45	rs2307799	5	70828419	BDP1	
46	rs34541393	20	30701405	TM9SF4	

?

	ID	Chr	Position	Gene	Functional Class
1	rs10141763	14	36170607	RALGAPA1	intronic
2	rs1024116	18	75432386	-	
3	rs10843344	12	29369871	-	
4	rs12913832	15	28365618	HERC2	promotor for OCA2
5	rs1321333	20	38849642	-	
6	rs1335873	13	20901724	-	
7	rs1426654	15	48426484	SLC24A5	coding THR 111 ALA
8	rs1498444	3	1.69E+08	-	
9	rs1573020	1	36768200	-	
10	rs16891982	5	33951693	SLC45A2	coding PHE 374 LEU
11	rs182549	2	1.37E+08	MCM6	promotor for LCT
12	rs1886510	13	22374700	-	
13	rs1978806	10	34755348	PARD3	intronic
14	rs2026721	4	1.59E+08	-	
15	rs2040411	22	47836412	-	
16	rs2065160	1	2.05E+08	-	
17	rs2065982	13	34864240	-	
18	rs2303798	19	42410331	ARHGEF1	intronic
19	rs2304925	17	75551667	-	
20	rs239031	21	17710424	-	
21	rs2572307	21	25672460	-	
22	rs2814778	1	1.59E+08	DARC	5' UTR (creates null)
23	rs3785181	16	90105333	GAS8	intronic
24	rs3827760	2	1.1E+08	EDAR	coding VAL 370 ALA
25	rs4540055	4	38803255	TLR1	intronic
26	rs5030240	11	32424389	WT1	intronic
27	rs5997008	22	26350103	MYO18B	intronic
28	rs722098	21	16685598	-	
29	rs730570	14	1.01E+08	-	
30	rs773658	12	56603834	RNF41	intronic
31	rs7897550	10	17064992	CUBN	intronic
32	rs881929	16	31079371	ZNF668	intronic
33	rs896788	2	7149155	RNF144A	intronic
34	rs917118	7	4457003	-	

3 of 34 non-synonymous coding SNPs and 3 of 34 ‘control element’ SNPs involved in gene expression (one for a non-EVC)

32 of 46 Indels are in coding regions but their function or effect on these gene’s behaviour is not known yet

Coding : non-coding status of 80 AIMs

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1	rs2307666	11	64729920	C11orf85	
2	rs1610863	16	6551830	RBFOX1	
3	rs16635	6	99789775	FAXC	
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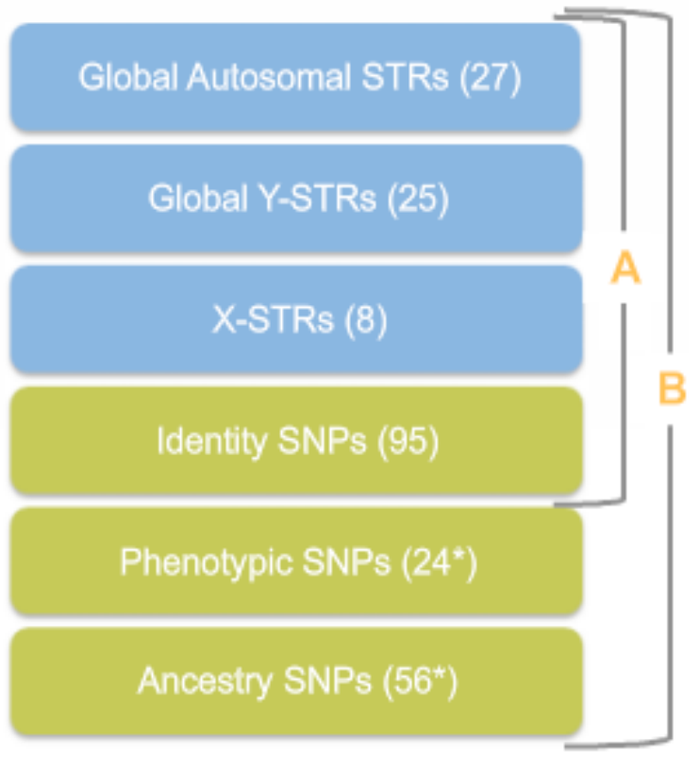

- 233 markers
- 2 primer sets
- 60-460 bp

A 155

B 78

EUROFORGEN - NGS and SNP analysis assessments

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- Applied a standardized validation program to the Ion PGM 169-SNP forensic identification set: ‘HID SNP’
  - Shared/Universal control DNAs 9947A - staff - Coriell CLDs
  - Sensitivity dilution series and non-probative v degraded DNA
  - Artificial mixtures of 1:1, 3:1, 9:1

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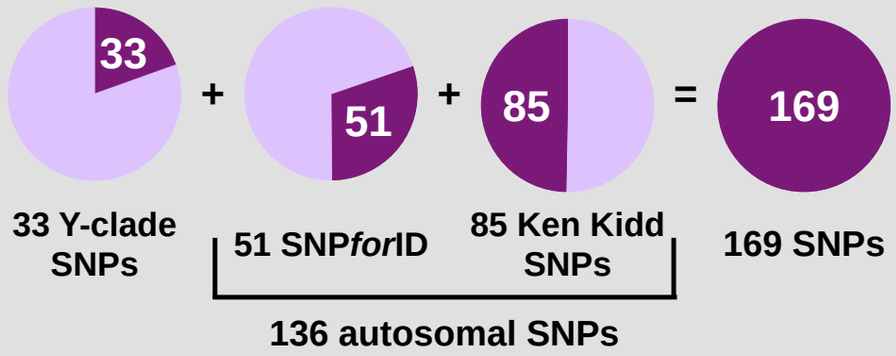
- Scrutinized context sequence for previously uncharted features such as flanking SNPs or indels that can disrupt alignments.

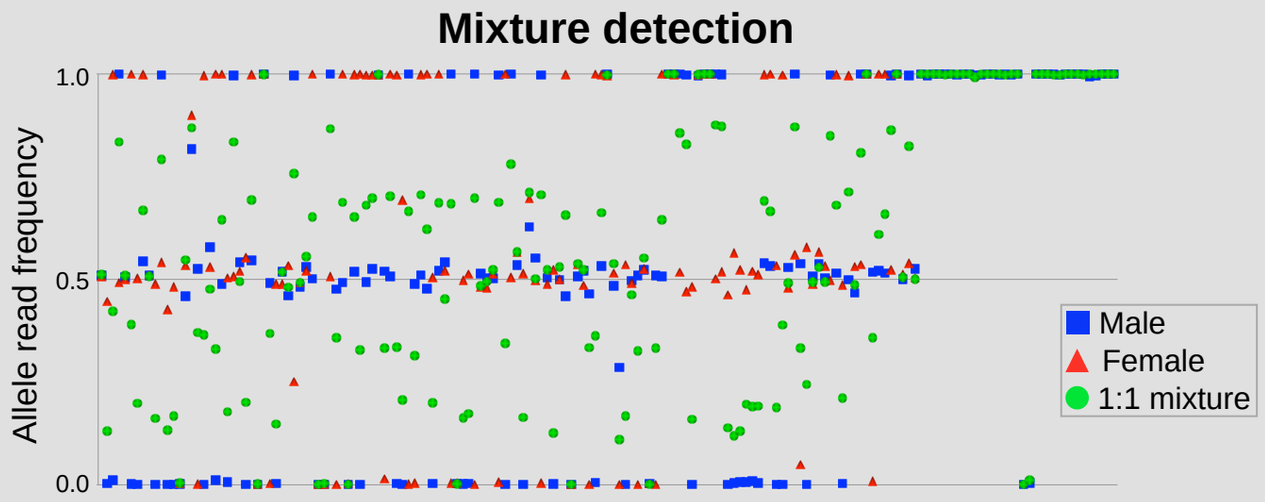
EUROFORGEN - HID SNP findings in a nutshell

EUROFORGEN Network of Excellence

Graphical Abstract (for review)

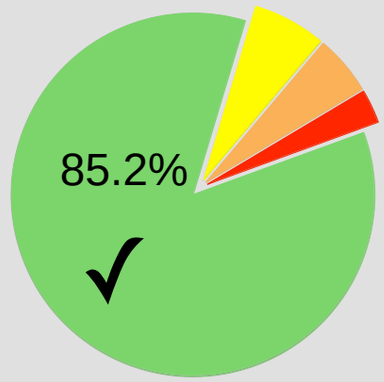
HID-Ion AmpliSeq™ Identity Panel v2.2





Overall concordance
99.8%

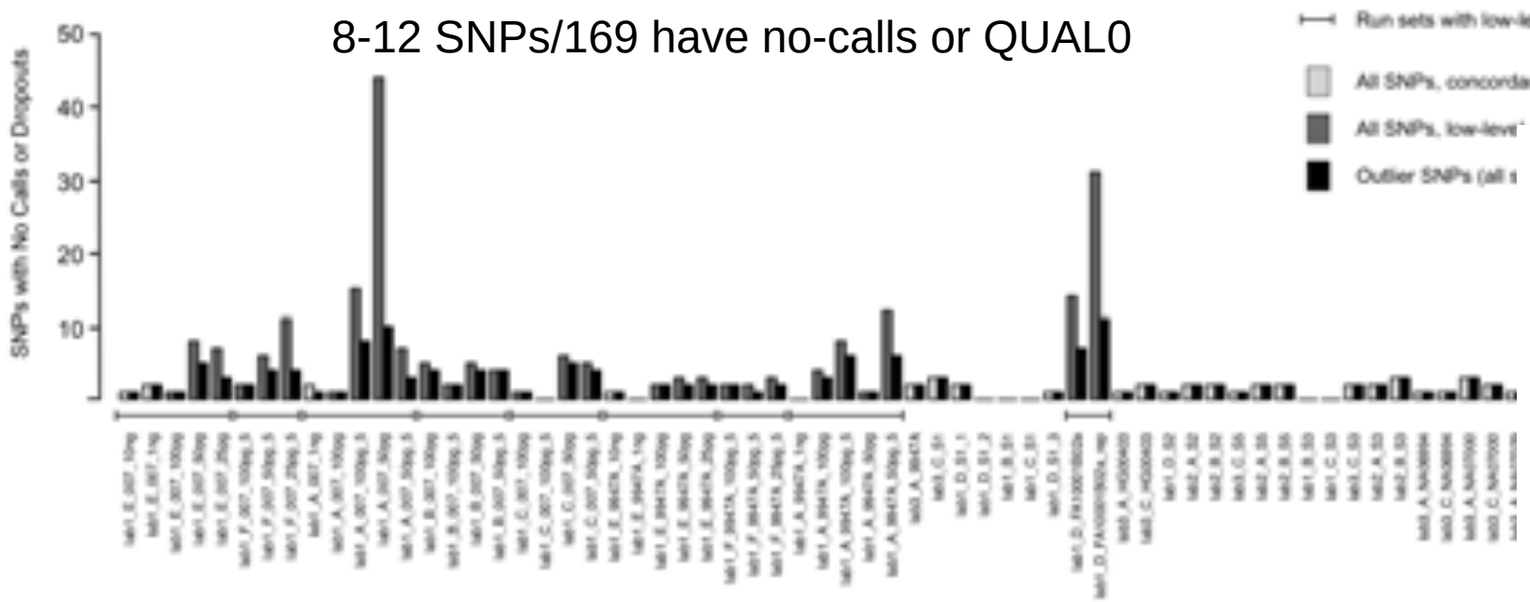
- Concordant genotypes
- No-call
- Discordant genotypes

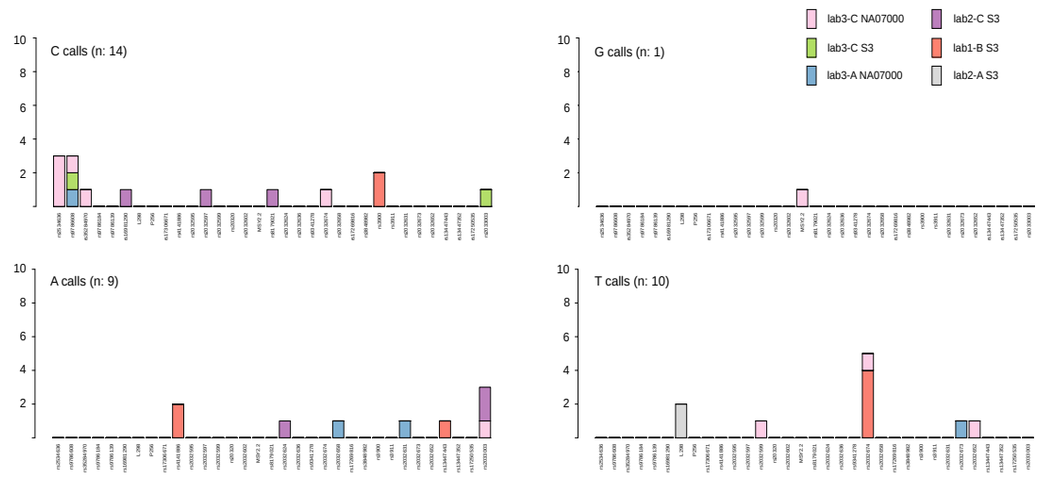


Outlier SNPs

Atypical
No-calls
Discordant
rs1979255
rs1004357
rs938283
rs2032597
rs2399332

8-12 SNPs/169 have no-calls or QUAL0





34 male sequences in >2 million

IGV rs13182883

IGV rs1029047

C T A G Indel

misalignment point Direction

TT homozygote

AA homozygote with 19% T-base calls

Total count: 408
A : 328 (80%, 196+, 132-)
C : 1 (0%, 0+, 1-)
G : 0
T : 79 (19%, 41+, 38-)
N : 0

DEL: 1
INS: 203

The SNP target base sequence counts indicate 19% spurious T-base calls were made due to misalignment of 3-T tract in the forward strand or misalignment of both 3-T and 8-A tracts in the reverse strand

EUROFORGEN - Goals for SNPs/NGS/Ancestry-FDP

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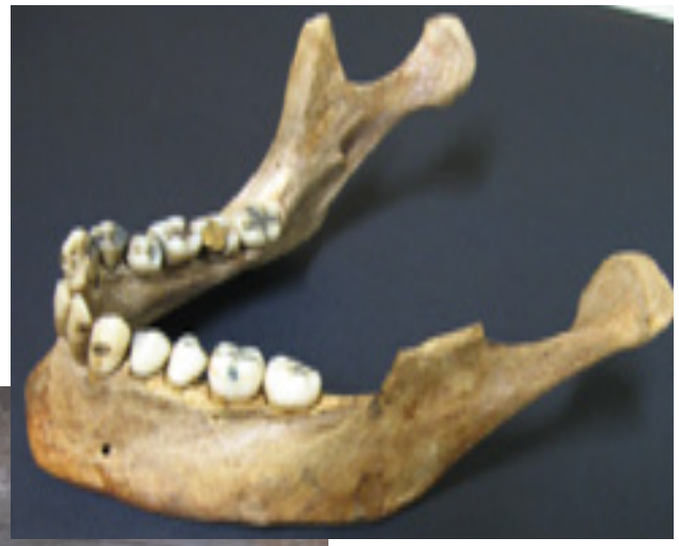
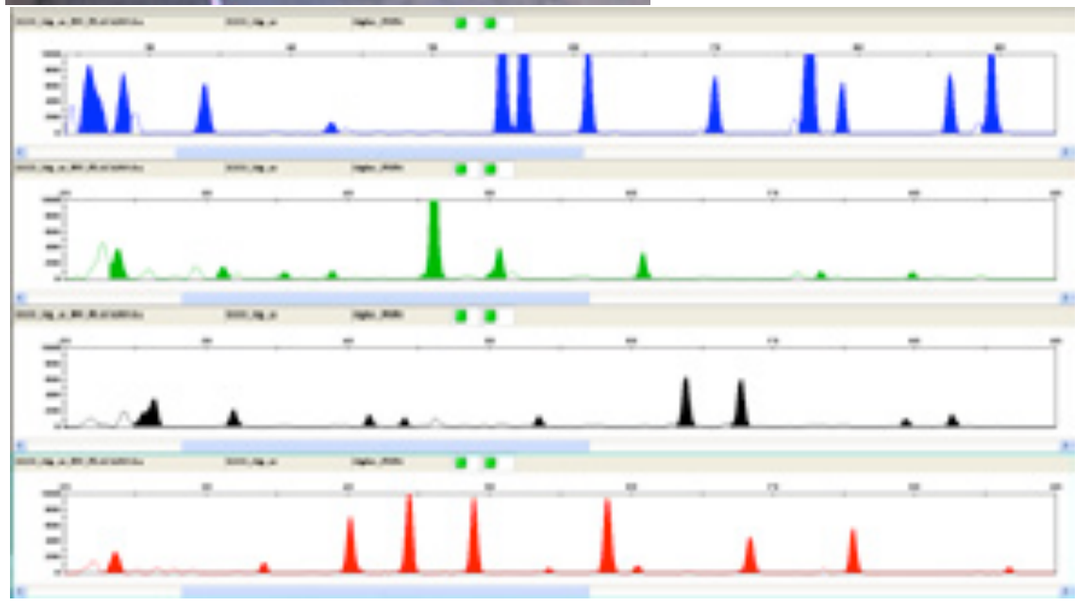
- Developed the *Global* 128-AIM panel and now validating these SNPs with a similar program to the commercial HID SNP multiplex

Adelaide U. WWII soldier identification in PNG

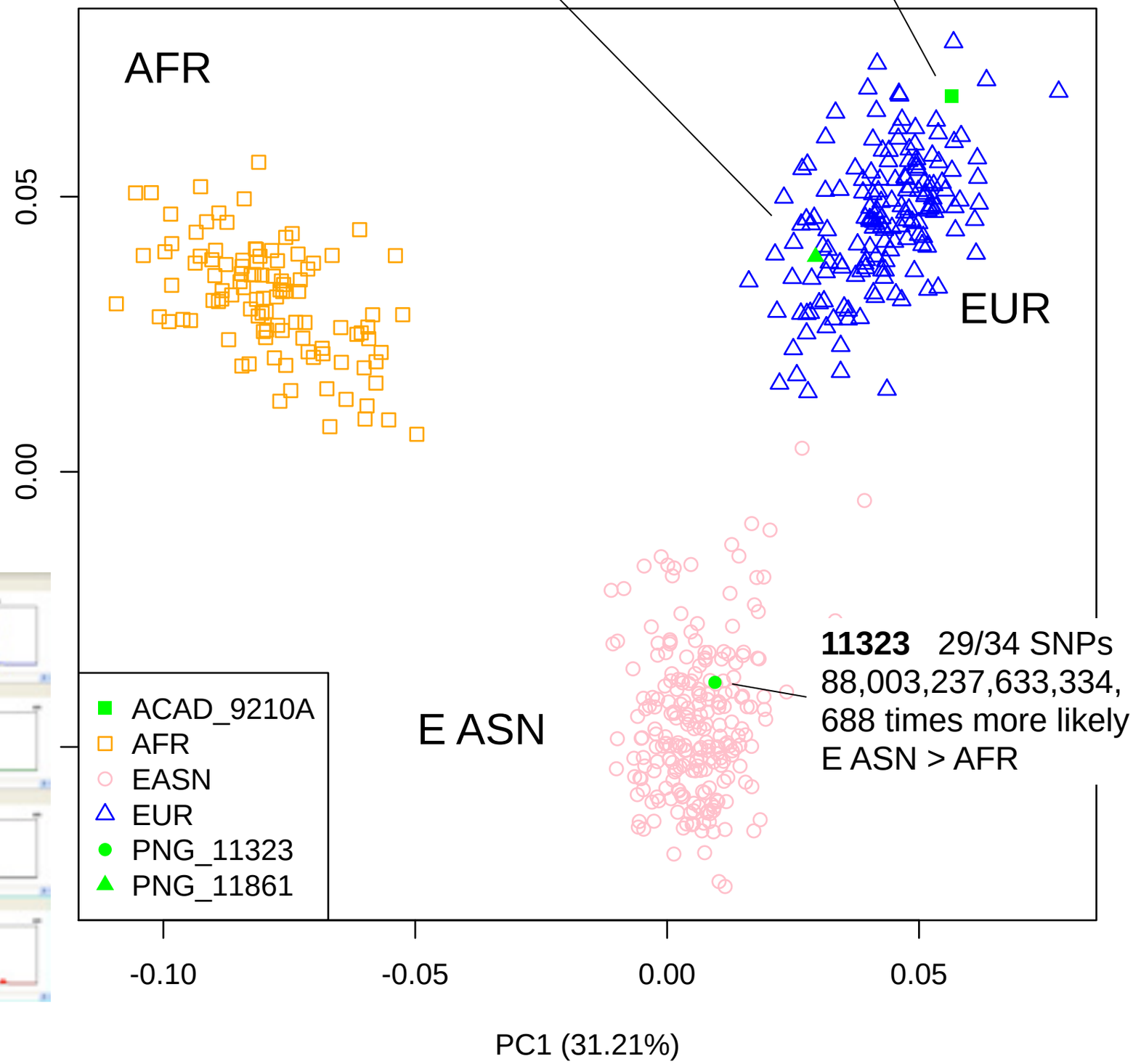




11323

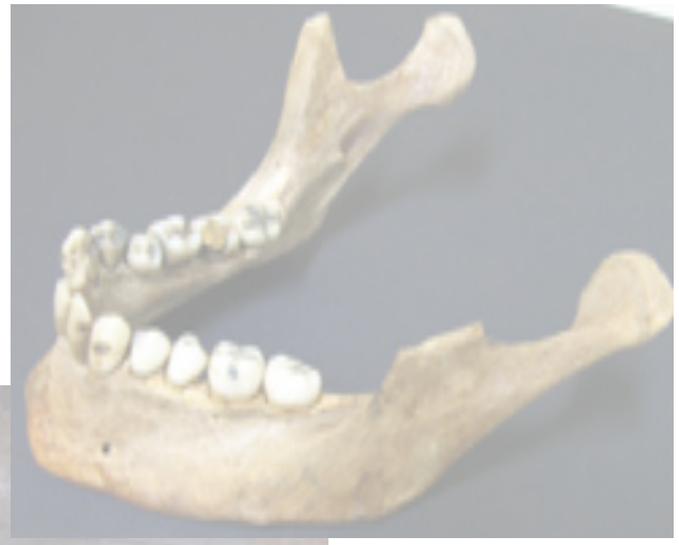
9210A 31/34 SNPs
335,954,698,878,504,337,408
times more likely EUR > E ASN

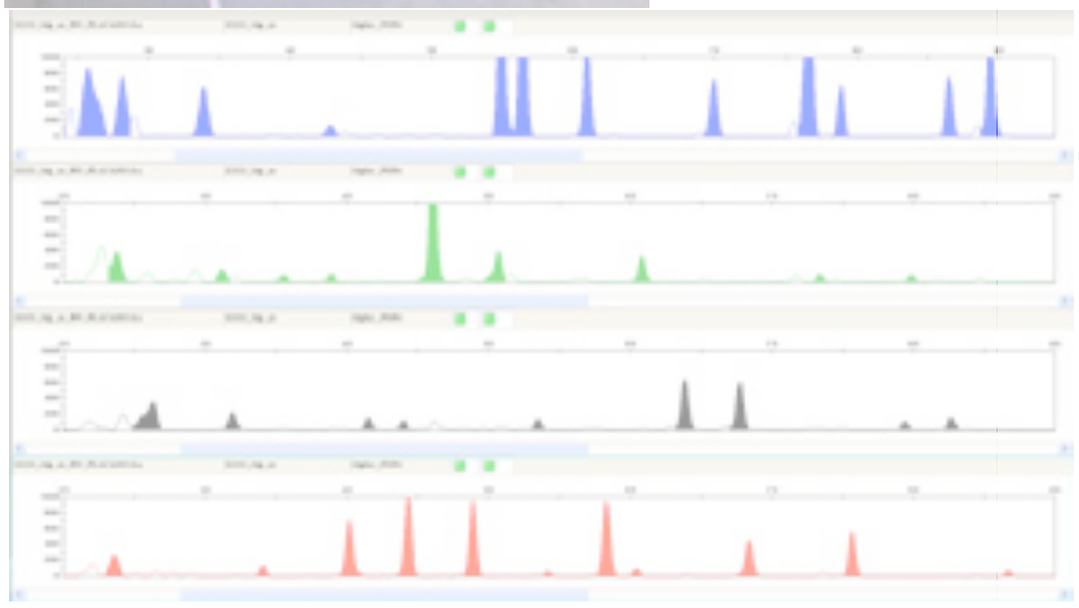
11861 18/34 SNPs
8,594,617,739 times more
likely EUR > E ASN



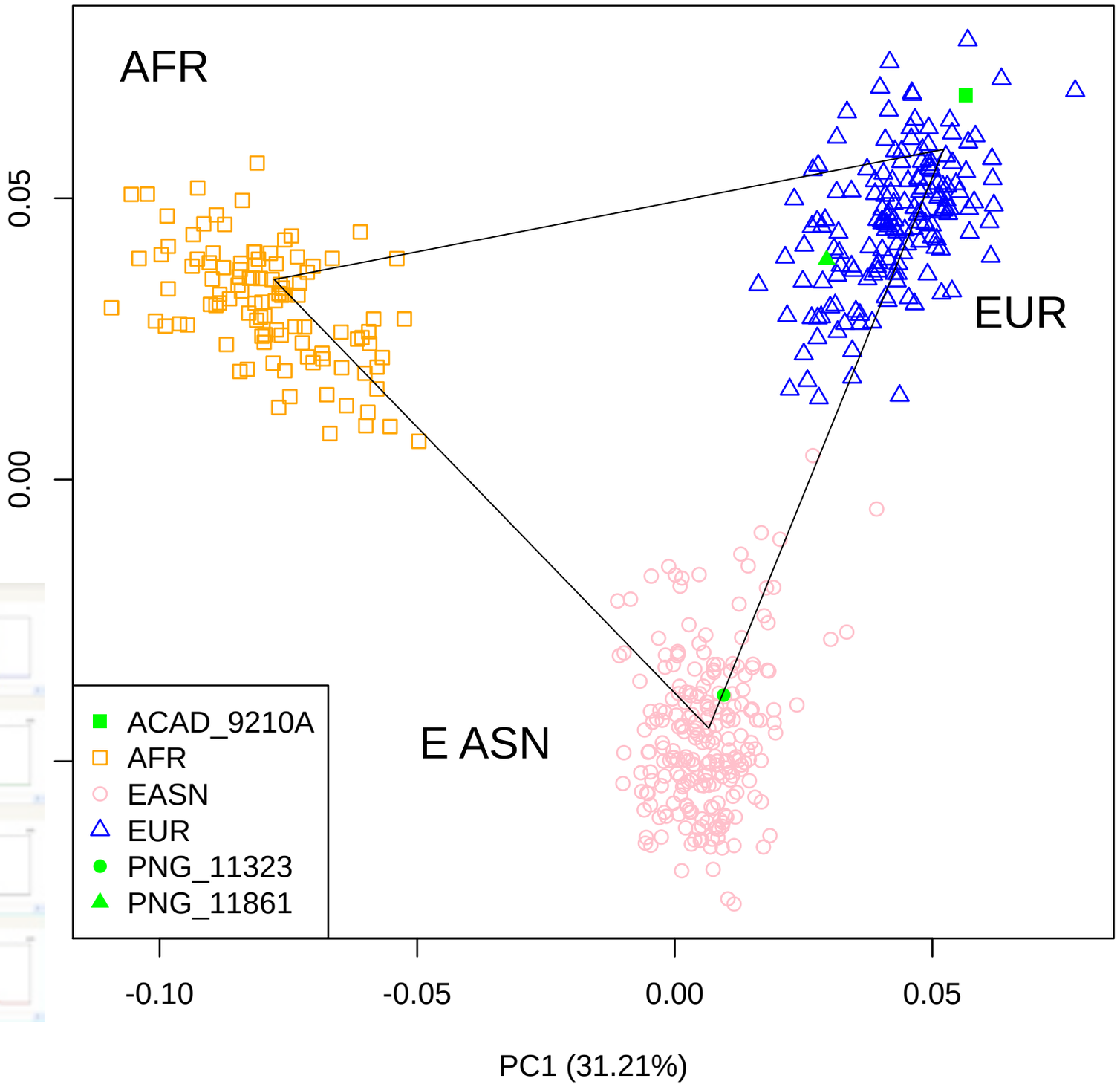
11323 29/34 SNPs
88,003,237,633,334,
688 times more likely
E ASN > AFR

Population group differentiation balance





11323



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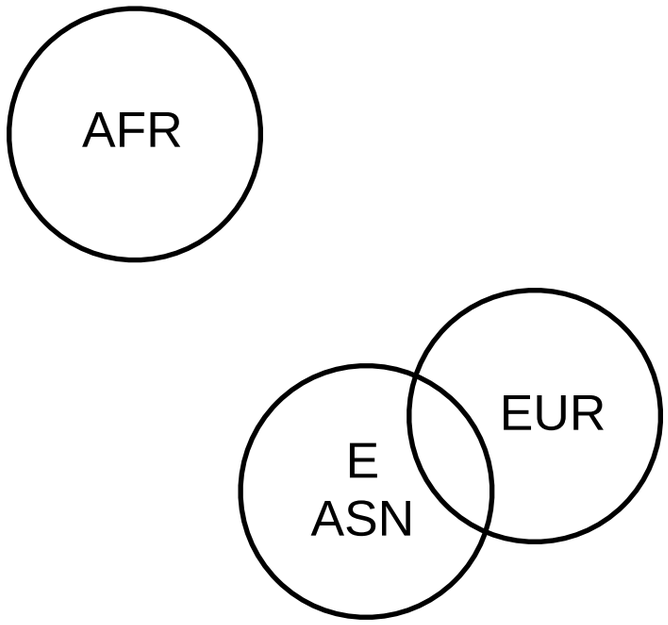

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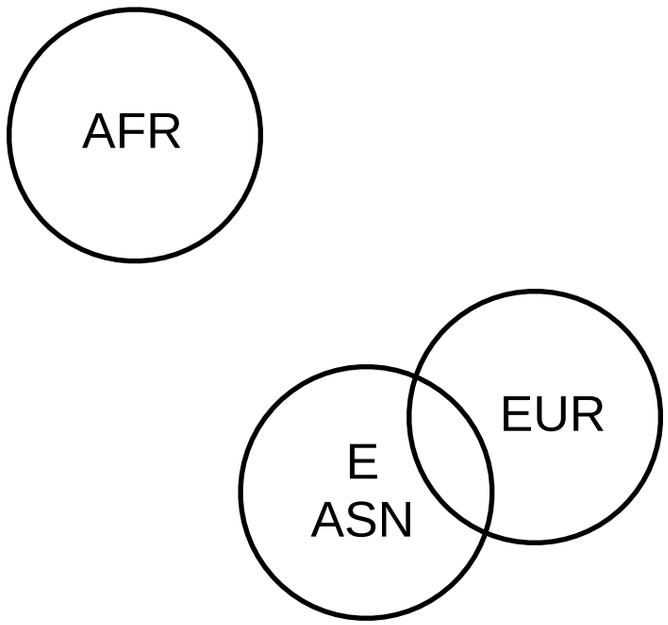
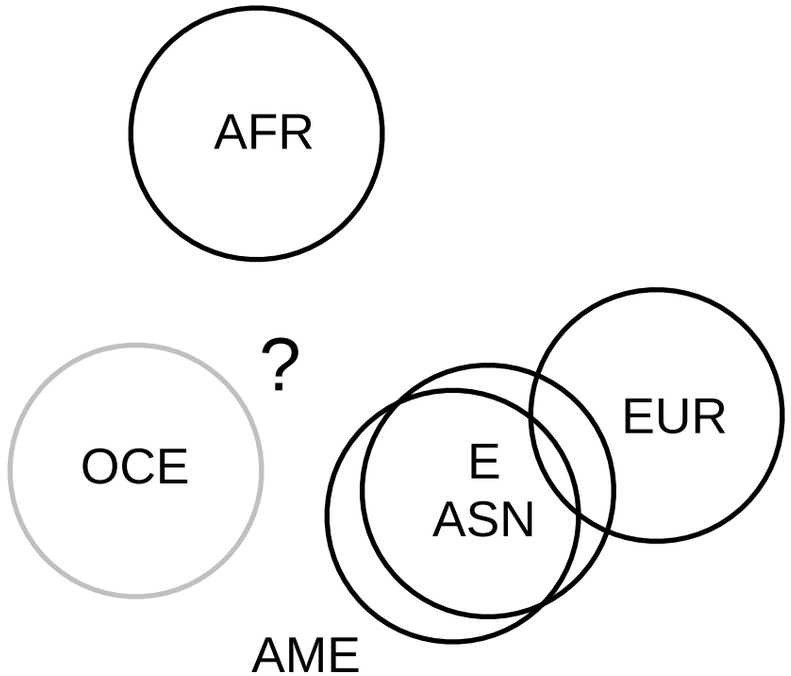


changing to five populations complicates the degree of divergence



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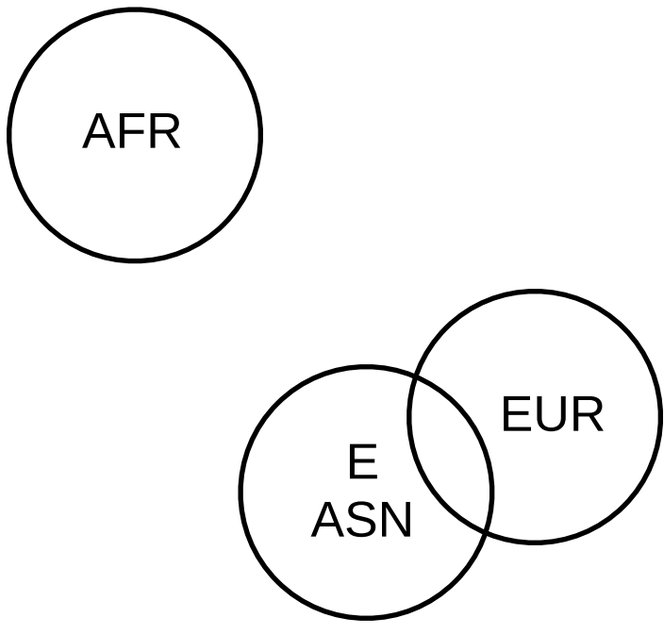

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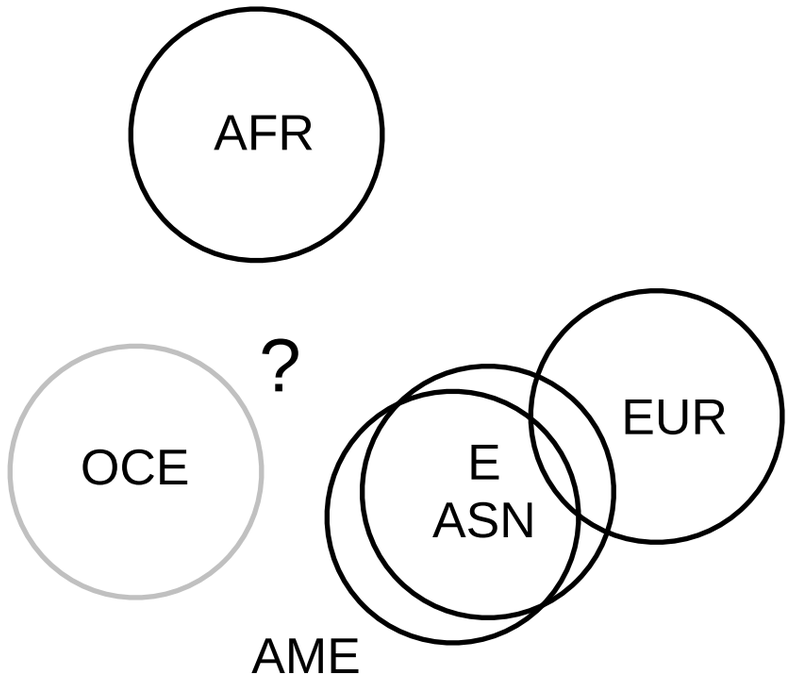

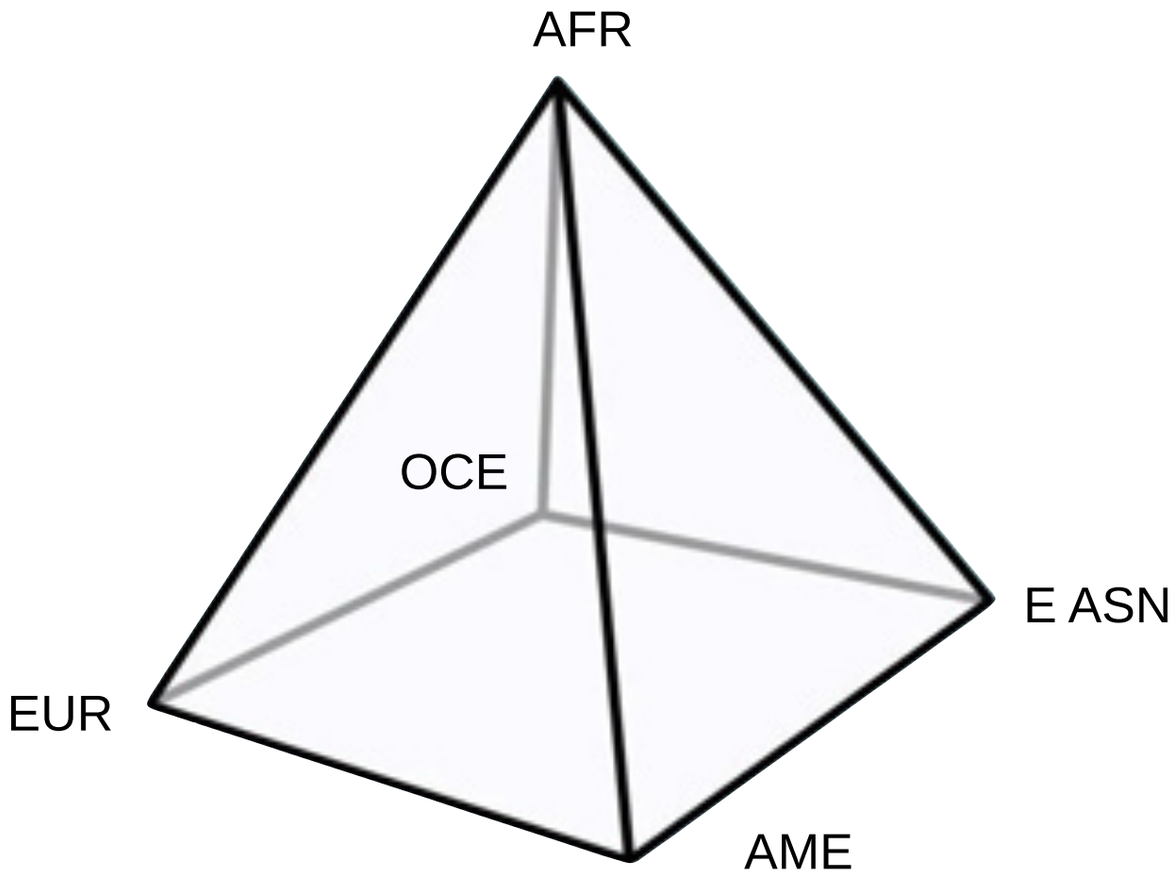

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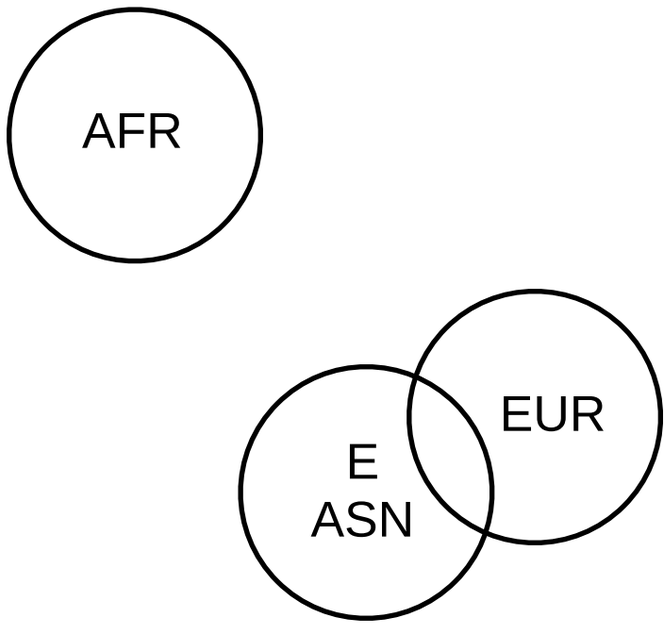

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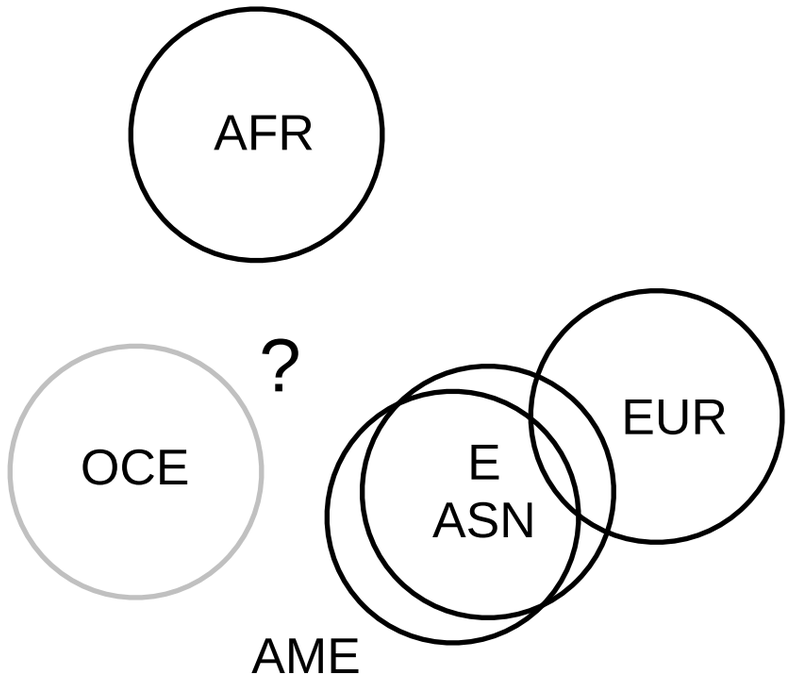

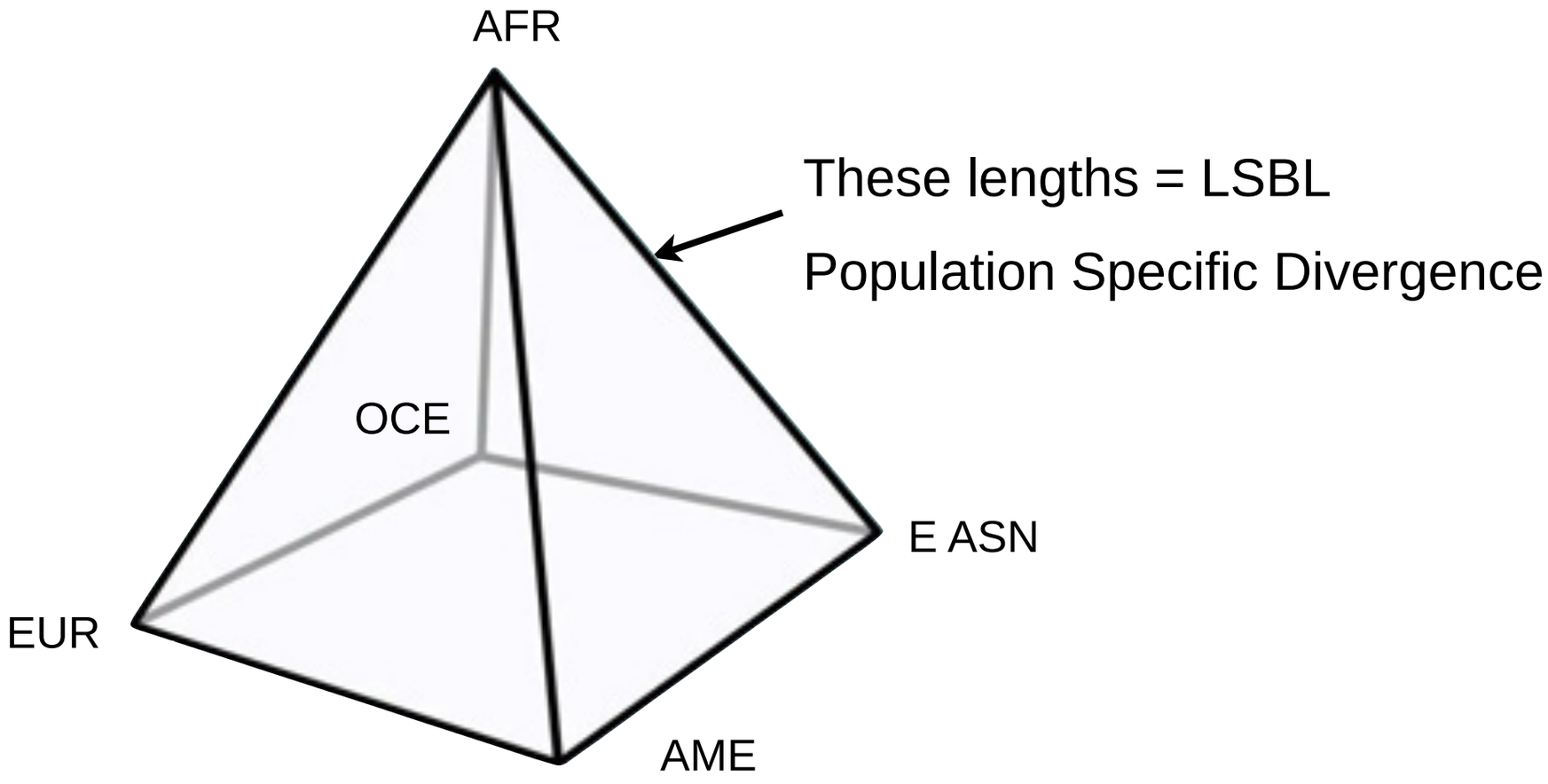

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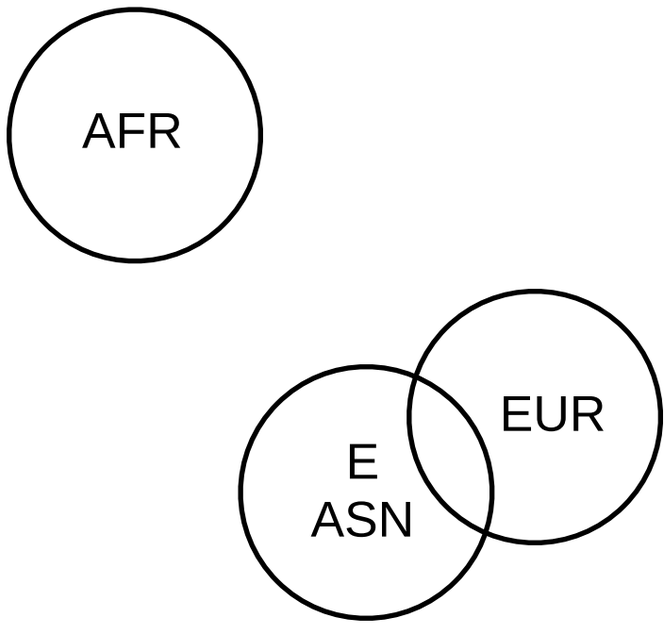

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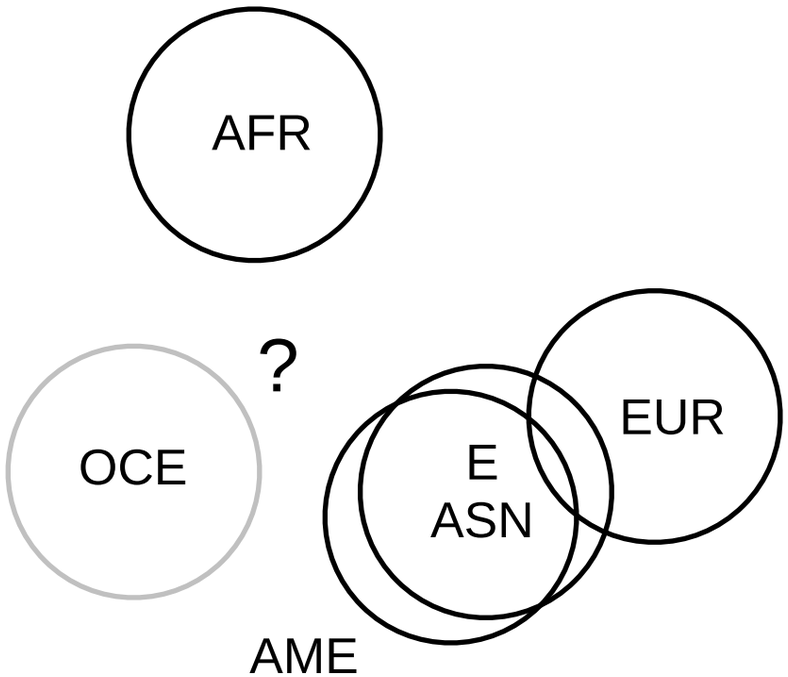

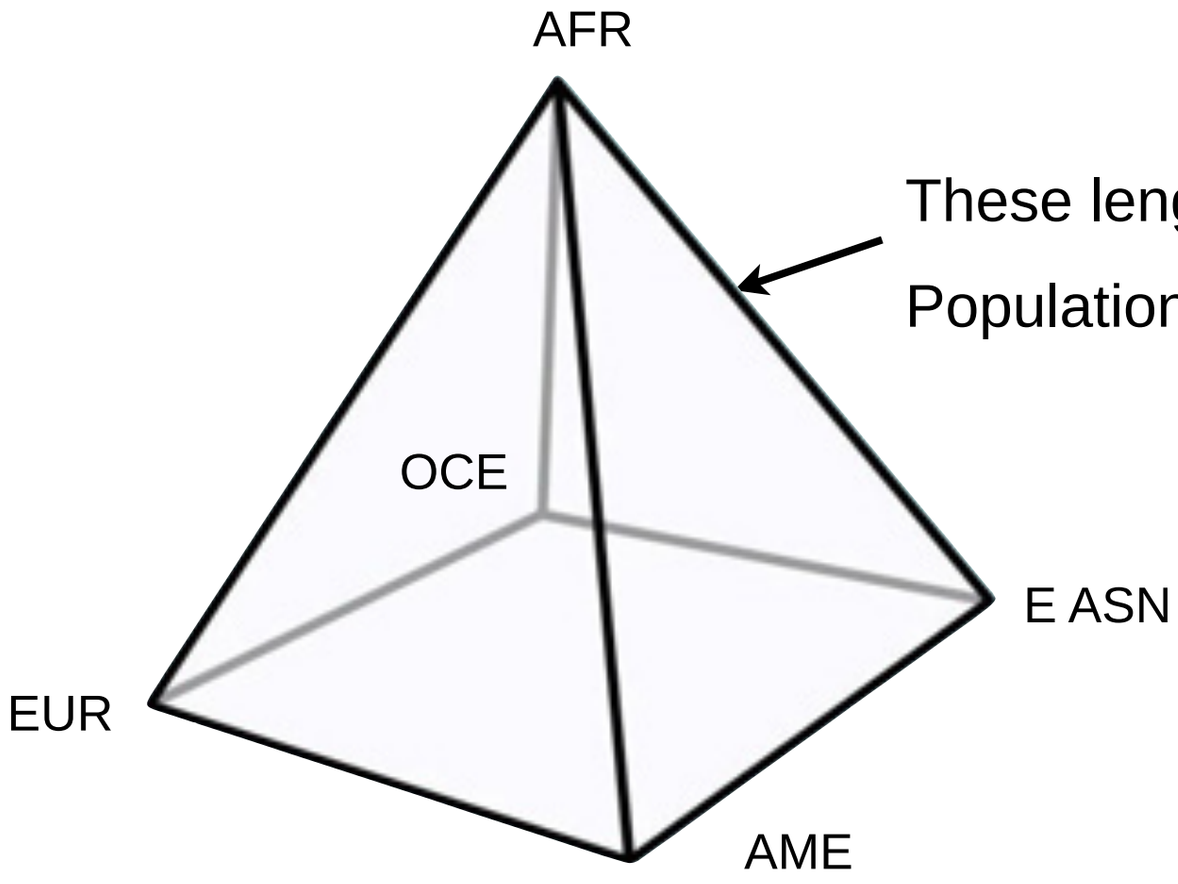

I_n

Carefully tracked PSD balance to a point of convergence at 122 SNPs

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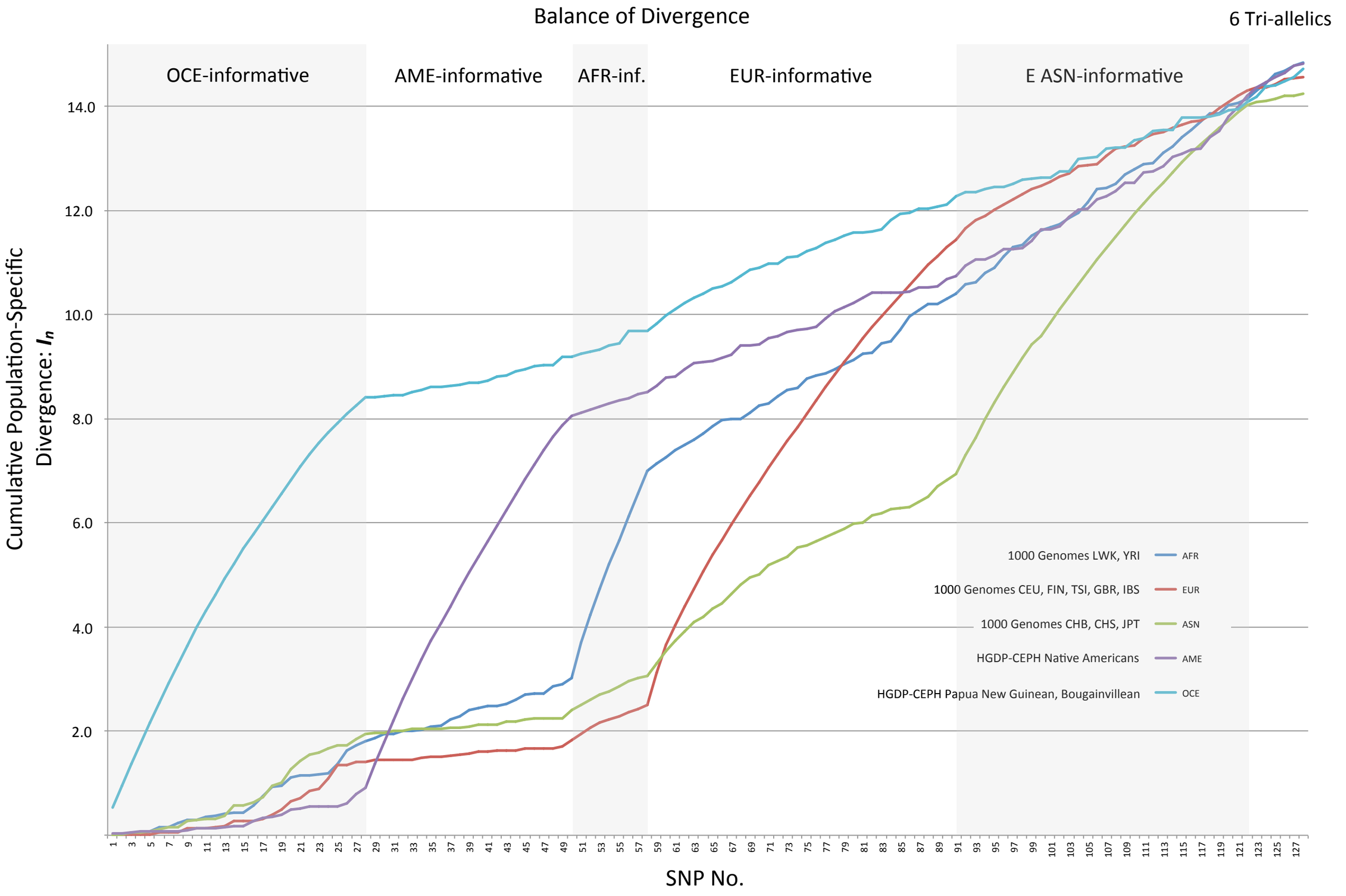

EUROFORGEN - Goals for SNPs/NGS/Ancestry-FDP

- Developed the *Global* 128-AIM panel and now validating these SNPs with a similar program to the commercial HID SNP multiplex

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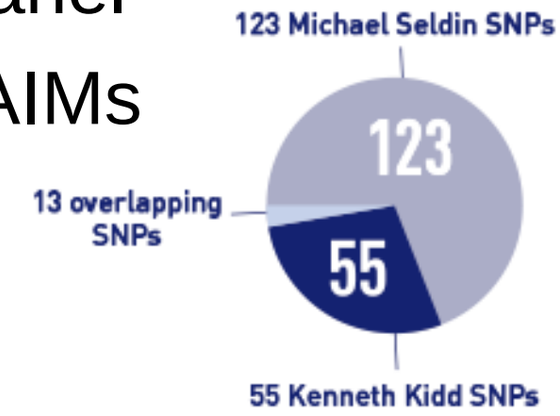
- USC: ancestry inference toolbox enhanced in concert with SPS

NGS AIMs Panels

- Illumina MiSeq *FGx* *ForenSeq* panel: 55 AIMs (2/55 FDP)
- Ion PGM: 165 AIMs panel
- Euroforgen: 128 Global AIMs



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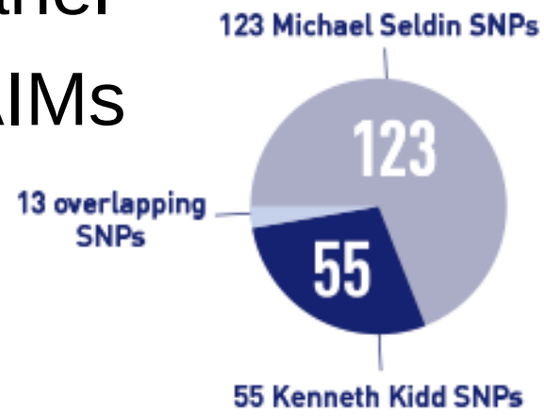
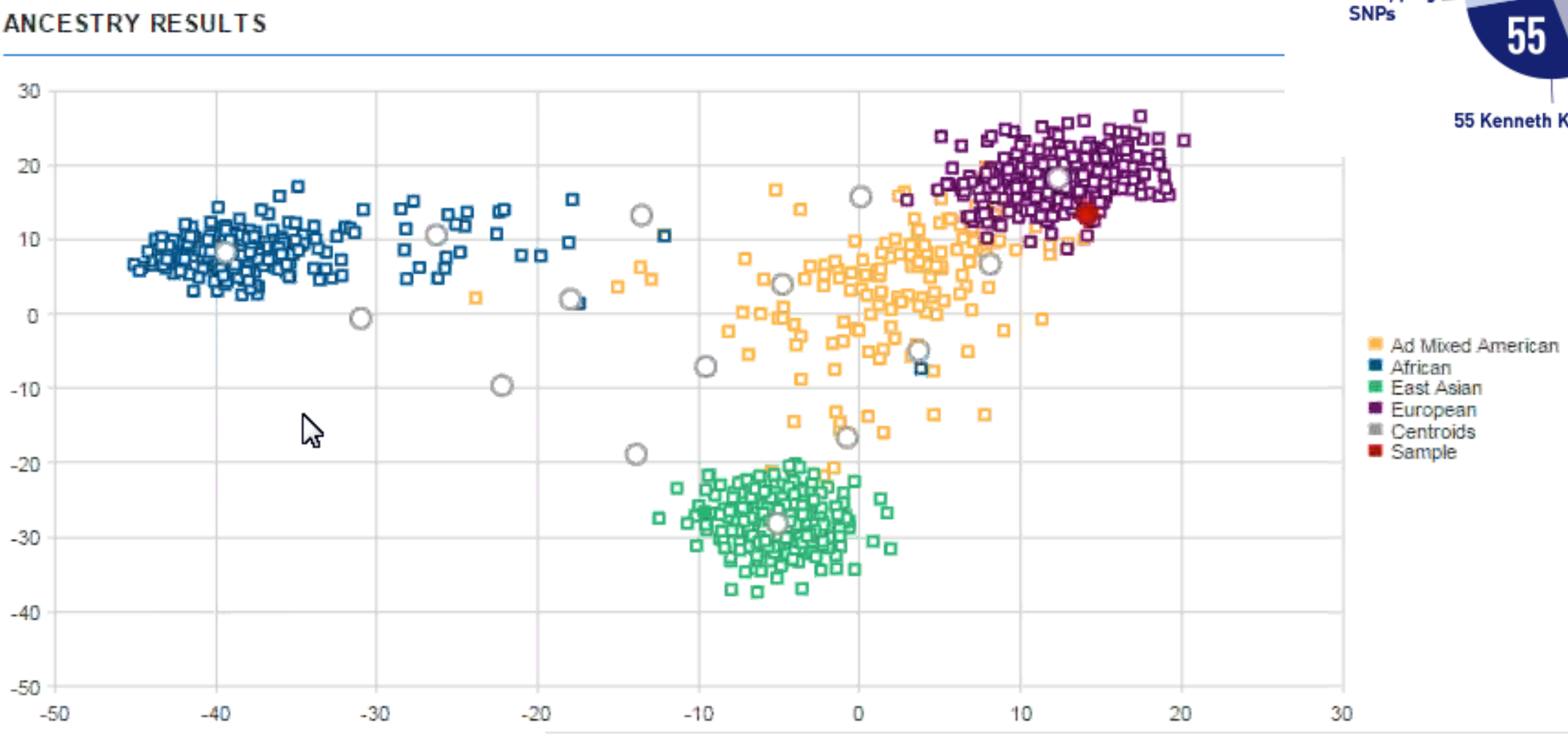




DISTANCE TO NEAREST CENTROID

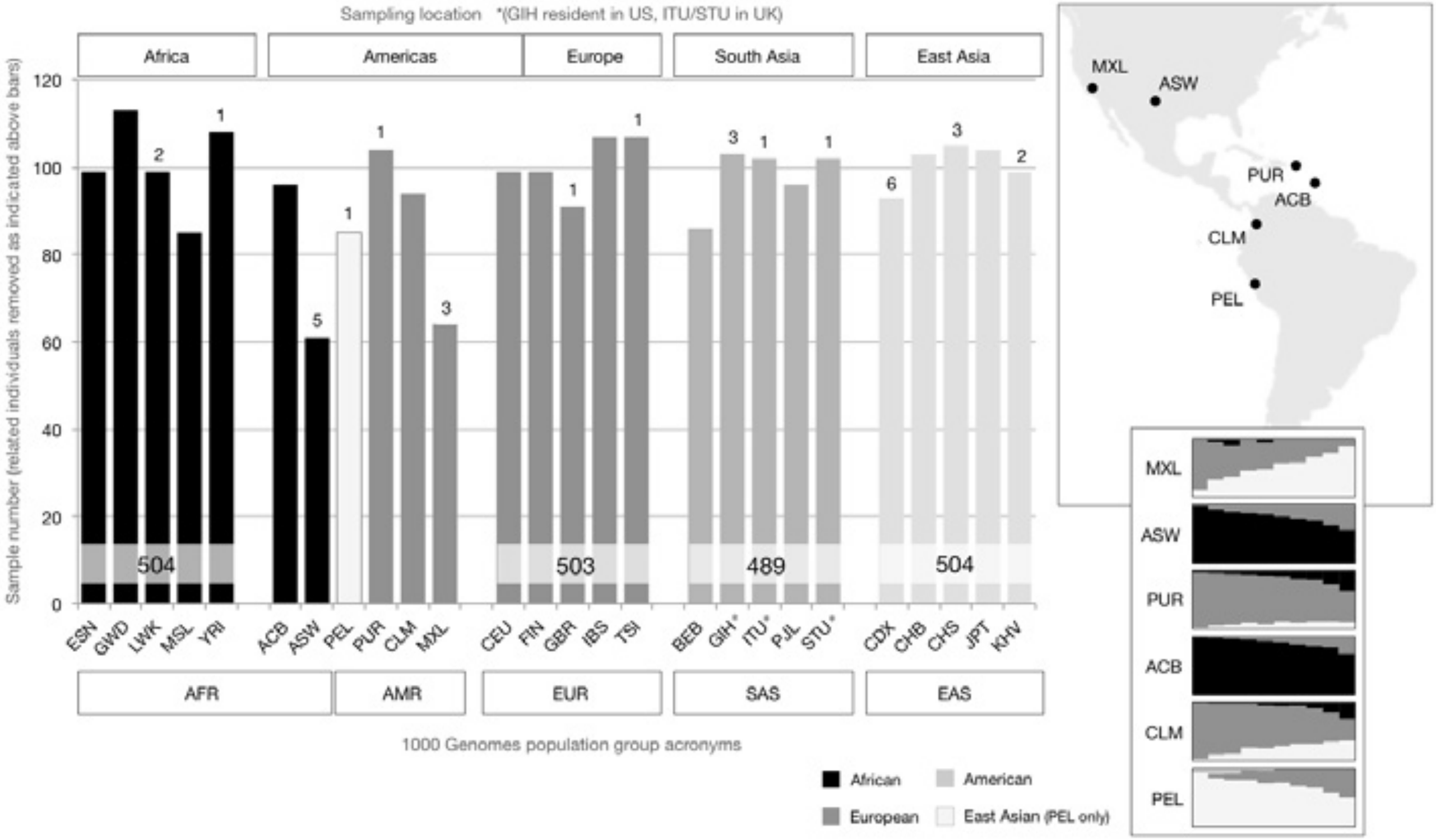
5.10

SNP PANEL USED

› 1000genomes populations with samples in centroid with sample

› Reference samples in centroid with sample

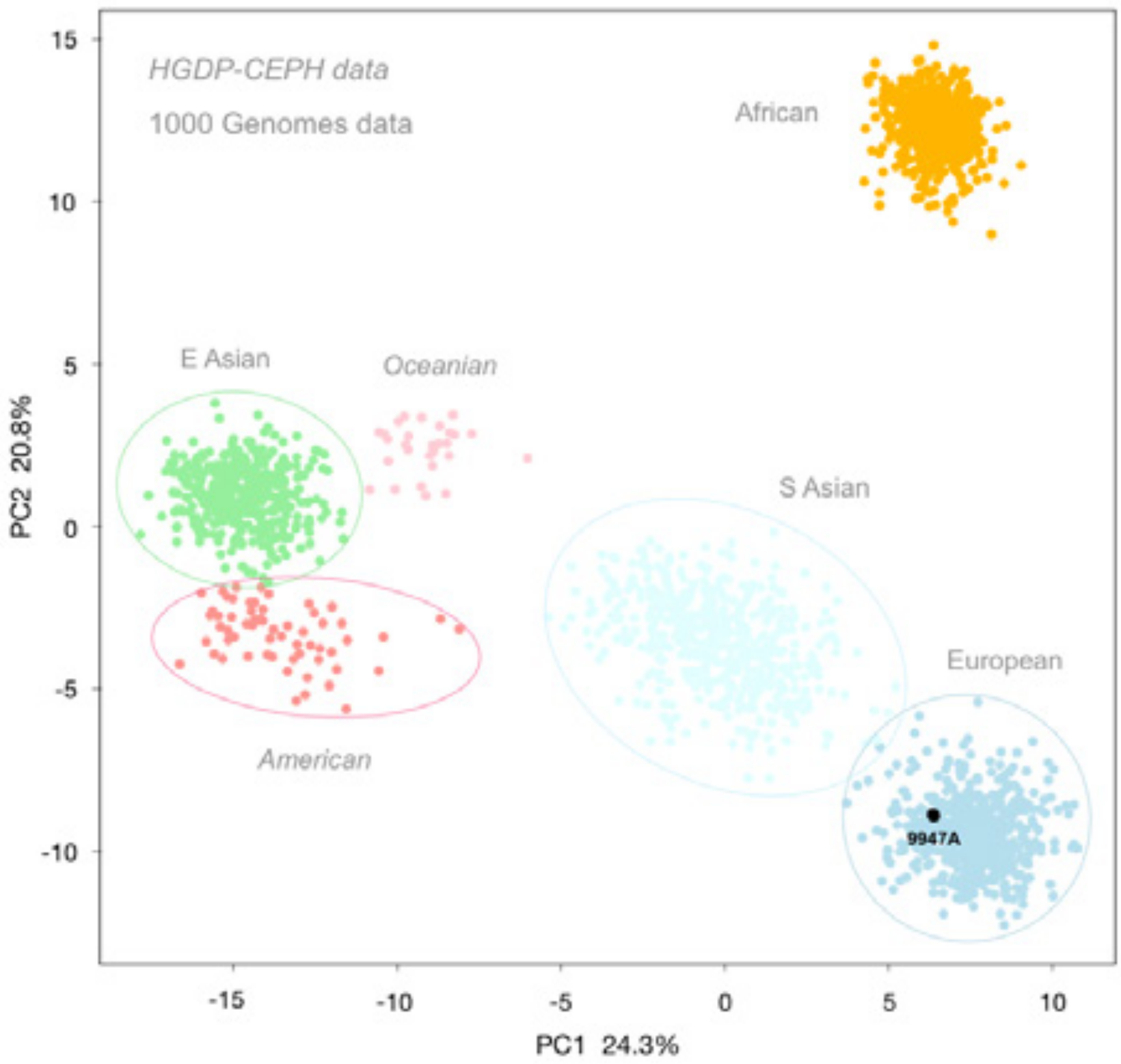
1000 Genomes final public SNP data release



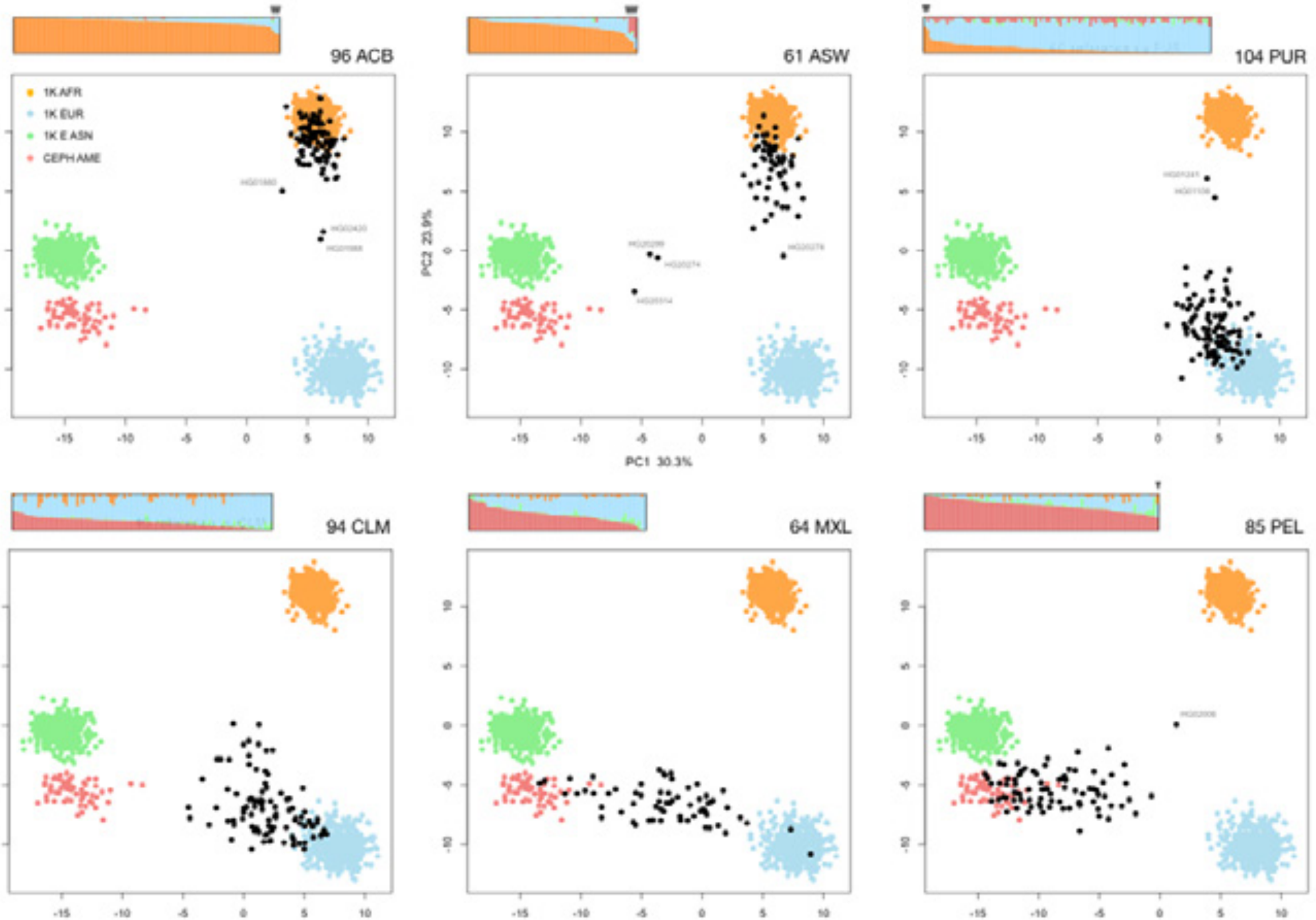
Phase I: ~28 million variants in 629 individuals from 12 populations
Phase III: ~79 million variants (77,520,219 are single nucleotide SNPs of A/C/G/T substitution) in 2,535 individuals from 26 populations.

1000 Genomes final data brings S Asians to ancestry analyses

122/128 Global AIMs




1000 Genomes Africans
1000 Genomes Europeans
1000 Genomes East Asians
Ame.
ACB
ASW
PUR
CLM
MXL
PEL
K:4


96 ACB
61 ASW
104 PUR
94 CLM
64 MXL
85 PEL
1K.AFR
1K.EUR
1K.E.ASN
CEPH.AME
PC2 23.9%
PC1 30.3%

Coding : non-coding status of 80 AIMs

	ID	Chr	Position	Gene	Functional Class
1	rs2307666	11	64729920	C11orf85	
2	rs1610863	16	6551830	RBFOX1	
3	rs16635	6	99789775	FAXC	
4	rs1610965	5	79746093	ZFYVE16	
5	rs35451359	18	45110983	-	
6	rs140837	6	3708907	-	
7	rs1160893	2	2.25E+08	WDFY1	
8	rs2308203	2	1.09E+08	RANBP2	
9	rs33974167	8	87813725	-	
10	rs1160852	6	1.37E+08	IL20RA	
11	rs1610884	5	56122323	MAP3K1	
12	rs2067280	5	89818959	LYSMD3	
13	rs2308067	7	1.27E+08	SND1	
14	rs4183	3	3192524	CRBN	
15	rs3054057	15	86010538	AKAP13	
16	rs2307840	1	36099090	PSMB2	
17	rs60612424	6	84017510	ME1	
18	rs3033053	14	42554496	-	
19	rs16384	22	42045009	XRCC6	
20	rs34611875	18	67623917	CD226	
21	rs1610859	5	1.28E+08	SLC27A6	
22	rs3045215	1	2.35E+08	IRF2BP2	
23	rs25621	6	1.4E+08	-	
24	rs2307832	1	55590788	USP24	
25	rs16343	4	17635560	FAM184B	
26	rs3031979	8	73501951	KCNB2	
27	rs34122827	13	63778778	-	
28	rs133052	22	41042364	-	
29	rs6490	12	1.08E+08	PRDM4	
30	rs4181	2	42577803	COX7A2L	
31	rs3030826	6	67176774	-	
32	rs140708	6	1.71E+08	-	
33	rs1611026	5	82545545	XRCC4	
34	rs16438	20	25278470	PYGB	
35	rs2308161	10	69800909	HERC4	
36	rs16687	7	83887878	-	
37	rs2307998	5	7814345	ADCY2	
38	rs2307803	3	1.09E+08	-	
39	rs2307930	6	84476378	-	
40	rs25630	6	14734341	-	
41	rs2307582	1	2.48E+08	OR2G3	
42	rs2307922	1	39896964	MACF1	
43	rs11267926	15	45526069	-	
44	rs25584	12	1.12E+08	ACAD10	
45	rs2307799	5	70828419	BDP1	
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	ID	Chr	Position	Gene	Functional Class
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2	rs1024116	18	75432386	-	
3	rs10843344	12	29369871	-	
4	rs12913832	15	28365618	HERC2	promotor for OCA2
5	rs1321333	20	38849642	-	
6	rs1335873	13	20901724	-	
7	rs1426654	15	48426484	SLC24A5	coding THR 111 ALA
8	rs1498444	3	1.69E+08	-	
9	rs1573020	1	36768200	-	
10	rs16891982	5	33951693	SLC45A2	coding PHE 374 LEU
11	rs182549	2	1.37E+08	MCM6	promotor for LCT
12	rs1886510	13	22374700	-	
13	rs1978806	10	34755348	PARD3	intronic
14	rs2026721	4	1.59E+08	-	
15	rs2040411	22	47836412	-	
16	rs2065160	1	2.05E+08	-	
17	rs2065982	13	34864240	-	
18	rs2303798	19	42410331	ARHGEF1	intronic
19	rs2304925	17	75551667	-	
20	rs239031	21	17710424	-	
21	rs2572307	21	25672460	-	
22	rs2814778	1	1.59E+08	DARC	5' UTR (creates null)
23	rs3785181	16	90105333	GAS8	intronic
24	rs3827760	2	1.1E+08	EDAR	coding VAL 370 ALA
25	rs4540055	4	38803255	TLR1	intronic
26	rs5030240	11	32424389	WT1	intronic
27	rs5997008	22	26350103	MYO18B	intronic
28	rs722098	21	16685598	-	
29	rs730570	14	1.01E+08	-	
30	rs773658	12	56603834	RNF41	intronic
31	rs7897550	10	17064992	CUBN	intronic
32	rs881929	16	31079371	ZNF668	intronic
33	rs896788	2	7149155	RNF144A	intronic
34	rs917118	7	4457003	-	

3 of 34 non-synonymous coding SNPs and 3 of 34 ‘control element’ SNPs involved in gene expression (one for a non-EVC)

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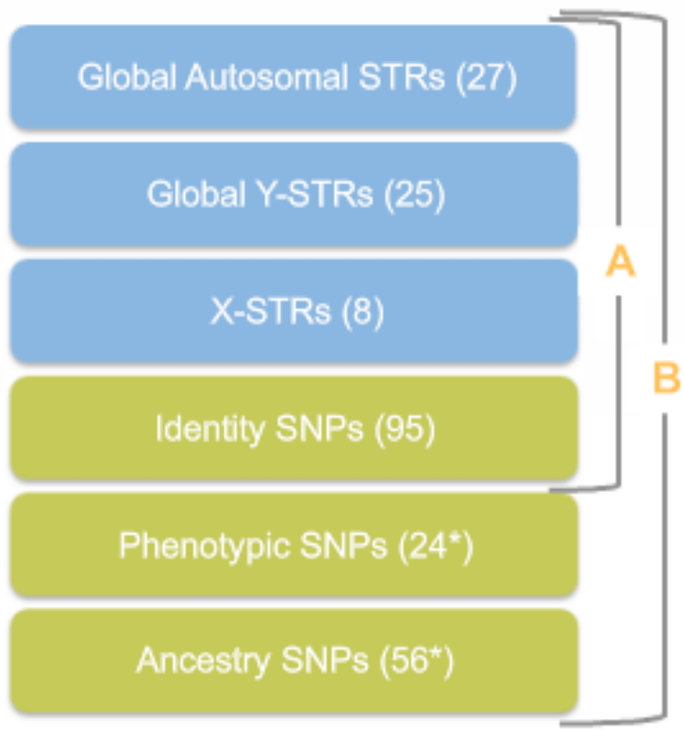


- 233 markers
- 2 primer sets
- 60-460 bp

A 155

B 78

EUROFORGEN-NoE update: EDNAP Meeting Zurich 2014

Peter M. Schneider
Institute of Legal Medicine
University of Cologne (Germany)

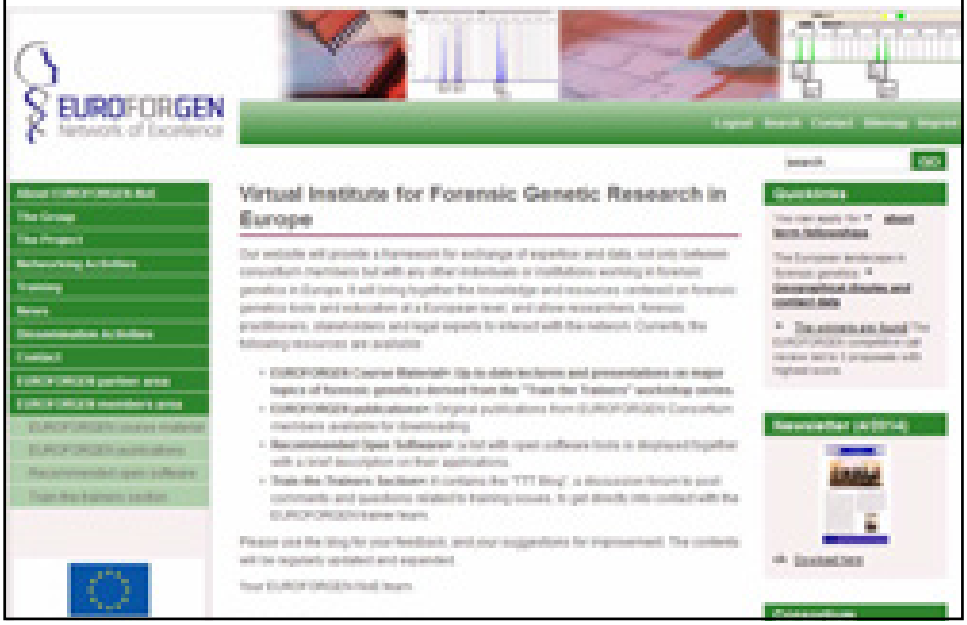
EUROFORGEN-NoE is funded by the European Commission within the 7th Framework Programme

Recent activities

- **Expansion of the EUROFORGEN website**
  - The Virtual Institute of Research for Forensic Genetics
- **Call for Proposals**
  - 3 new projects selected
- **Public Relations Conference**
  - Lobbying for more funding for research
- **Training news**
  - and other sources of support

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

The Virtual Institute of Research for Forensic Genetics



The Virtual Institute of Research for Forensic Genetics

- **Dedicated "for members only" area of website**
  - Can only accessed after individual registration, and obtaining a user name and password
  - All colleagues working in institutions that have submitted their contact data by submitting a questionnaire in the initial inquiry will be admitted
  - Please do not hesitate to inquire if you are not sure about the participation of your lab!

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

The Virtual Institute of Research for Forensic Genetics

The Virtual Institute of Research for Forensic Genetics

- **Privileged access to new content:**
  - **Course Material:** Up-to-date lectures and presentations on major topics of forensic genetics derived from the "Train the Trainers" workshop series.
  - **Publications:** Original publications (PDF) from Consortium members available for downloading.
  - **Open Software:** a list with open source / accessible software tools is displayed together with a brief description on their applications.
  - **Train-the-Trainers Section:** a discussion forum to post comments and questions related to training issues, to get directly into contact with the EUROFORGEN trainer team.

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

Competitive Call for Proposals 2014

Announcing the three winning proposals

- **Dr. Cordula Haas, Zürich**
  "Association of a Body Fluid with a DNA Profile by Targeted RNA and DNA Deep Sequencing"
- **Prof. Manfred Kayser, Rotterdam**
  "Forensic DNA phenotyping of hair structure for investigative purposes"
- **Dr. Marielle Vennemann, Münster (with Lynn Dennany, Strathclyde)**
  "Development of innovative electrochemical biosensor technologies for the detection of tissue specific DNA methylation"

Competitive Call for Proposals 2014

- The new partners will be integrated into the consortium agreement as full partners according to the FP7 rules
- Projects will begin in January 2015
- New collaborations will be established
- New deliverables have been defined based on the research objectives


Public relation conference on
Millions of genetic traces and no suspects
– what can be done?
30 September 2014, 17:30 – 19:00
Representation of the State of North Rhine Westphalia, Germany, Rue Montoyer 47, 1000 Brussels
in the center of the EU district
Agenda
About the speakers


PR Conference in Brussels (30.09.2014)
EUROFORGEN-NoE is funded by the E
within the 7th Framework Programme

PR Conference in Brussels (30.09.2014)

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

PR Conference in Brussels (30.09.2014)

Programme details

- Opportunities for funding by the ERC
  José Labastida, Head of the Scientific Management Department of the European Research Council (ERC)
- Forensic sciences in the 21st century
  Üllar Lanno, Chairman of the European Network of Forensic Science Institutes (ENFSI)
- The European Forensic Genetics Network of Excellence - meeting the challenges of biological evidence
  Peter Schneider, Institute of Legal Medicine, University of Cologne
- Recent developments in Forensic Genetics research and the funding situation in Europe
  Angel Carracedo, Institute of Legal Medicine, University of Santiago de Compostela
- Innovations in Forensic Genetics: For and With Society
  Robin Williams, Centre for Forensic Science, Northumbria University at Newcastle

http://www.euroforgen.eu/news/public-relations-conference/

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

PR Conference in Brussels (30.09.2014)

- **The Consortium members are urging the legislators and policy makers**
  - to provide a suitable funding framework ensuring that relevant topics will be selected for the upcoming calls for proposals in HORIZON 2020,
  - and that a significant amount of funding earmarked for forensic sciences will be available for research projects
    - with a direct impact on providing more investigative leads on unsolved crime cases,
    - as well as for a sound and reliable presentation of forensic evidence in the courtroom.

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

Train the Trainers Workshops

- **The 4th TTT workshop will be organized in April 2015**
  - Increasing the number of trainers from participating countries
- **EUROFORGEN instructors are available to support satellite trainings at national level**

  Colleagues from
  - Italy
  - Spain
  - Belgium

  … have already organized such local training events!
- **Pre-Congress Workshops at the next ISFG Congress in Kraków 2015 will be supported by EUROFORGEN**

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

The Short Term Fellowship Program

- **First Call 2013**
  - 14 fellowships awarded to 13 colleagues from 9 countries
  - Details on website

- **Second Call 2014-2015**
  - 20 new fellowships open
    - Laboratory visits for 3-5 days
    - Active participation in workshops related to EFG aims
    - Other research/training activities related to scope of WPs 2-5
  - Application details on the website
  - Travel support up to EUR 500

Last but not least …

find us on Facebook

Forensic Science Regulator
Overseeing Quality

Draft guidance: Cognitive bias effects relevant to forensic science examinations

August 2014

This is a consultation draft and therefore should not be regarded or used as a standard. This draft is issued to allow comments from interested parties; all comments will be given consideration prior to publication. Comments should be sent to FSRConsultation2@homeoffice.gsi.gov.uk and should be submitted by 31st October 2014. This mailbox is not for general correspondence and is not routinely monitored so no acknowledgement will normally be sent.

THIS DRAFT IS NOT CURRENT BEYOND 31st OCTOBER 2014.

CONTENTS

1. INTRODUCTION

1.1.1 A key requirement of the Forensic Science Regulator's Codes of Practice and Conduct for forensic science providers and practitioners (the Codes) is that they "Act with honesty, integrity, objectivity and impartiality..." (p9 bullet point 2).

1.1.2 However many fields of forensic science include subjective assessment and comparison stages that are potentially susceptible to unconscious personal bias (cognitive contamination), which in turn could undermine the objectivity and impartiality of the forensic process. The focus of this appendix to the Codes is on providing general guidance on cognitive bias relevant to forensic examinations with the aim of alerting readers on how to recognise it and therefore help safeguard against biasing effects, through adherence to good practice. This document also provides examples of good practice for specific subject areas listed in sections 7 to 12. This document sets out the policy to ensure the format and content of all annexes issued by the Regulator are consistent.

2. EFFECTIVE DATE

2.1.1 This is a draft issue of this document for consultation.

3. SCOPE

3.1.1 These guidelines are limited to the consideration of cognitive bias within processes associated with forensic science examinations at scenes and within the laboratory only and therefore do not cover the wider aspects of the criminal justice system (CJS) such as court processes including activities of the judiciary/legal profession.

4. MODIFICATIONS

4.1.1 This is a draft issue of this document.

5. TERMS AND DEFINITIONS

5.1.1 **Anchoring or focalism:** The tendency to rely too heavily on one piece of information when making decisions.

5.1.2 **Blinding:** Shielding the forensic examiner from information about the case that is not required in order to conduct the examination.

5.1.3 **Cognitive bias:** a pattern of deviation in judgement whereby inferences about other people and situations may be drawn in an illogical fashion.

5.1.4 **Confirmation bias:** The tendency to test hypotheses by looking for confirming evidence rather than potentially conflicting evidence.

5.1.5 **Contextual bias:** The tendency for a consideration to be influenced by background information.

5.1.6 **Debias:** The reduction or elimination of the impact of bias in decision making and problem solving.

5.1.7 **Expectation bias:** also known as experimenter's bias, is where the expectation of what you will find affects what you do actually find.

5.1.8 **Photogrammetry:** The art science and technology of obtaining reliable information about physical objects through the processes of recording measuring and interpreting photographic images.

5.1.9 **Psychological contamination:** Exposure to other information which is irrelevant to their assessment but introduces unconscious bias into their findings.

5.1.10 **Reconstructive effects:** The tendency when people rely on memory, to fill in gaps on recall with what they believe should have happened.

5.1.11 **Role effects:** The tendency for individuals to identify themselves as part of a team with common goals which may introduce subconscious bias.

6. AN EXPLANATION AND BRIEF OVERVIEW OF COGNITIVE BIAS

6.1 Overview

6.1.1 Cognition is the mental process of knowing, including awareness, perception, reasoning and judgement[1], and is distinct from emotion and volition[2]. Cognitive bias may be defined as a pattern of deviation in judgement whereby inferences about other people and situations may be drawn in an illogical fashion[3]. We all tend to display bias in judgements that we make in everyday life, indeed this is a natural element of the human psyche: Jumping to a conclusion, tunnel vision, only seeing what we want to see, being influenced by the views of others, are all behaviours we recognise in ourselves and others. However whilst such biases may be commonplace and part of human nature, it is essential to guard against these in forensic science, where many processes require subjective evaluations and interpretations. The consequences of cognitive bias may be far-reaching: decisions by the investigator to follow a particular line of enquiry, the CPS to prosecute or not, and decisions in the CJS as to guilt or innocence of an individual upon which may rest their liberty or even their life in some jurisdictions, frequently depends on the reliability of the forensic evidence and the conclusions drawn from its interpretation.

6.1.2 Cognitive bias has been identified as a potential issue within criminal justice systems since the 1970s[4,5,6], and in more recent years some high profile cases

[1] The American Heritage® Science Dictionary Copyright © 2005

[2] The Concise Oxford Dictionary, 18th edition

[3] Haselton, M. G., Nettle, D., & Andrews, P. W. (2005). *The evolution of cognitive bias*. In D. M. Buss (Ed.), The Handbook of Evolutionary Psychology: Hoboken, NJ, US: John Wiley & Sons Inc. pp. 724–746

[4] Tversky, A., & Kahneman, D. (1974). Judgment under uncertainty: Heuristics and biases. Science, 185, 1124–1131. http://dx.doi.org/10.1126/science.185.4157.1124

[5] Charlton, D., Fraser-Mackenzie, P.A.F. & Dror I.E. (2010). Emotional experiences and motivating factors associated with fingerprint analysis. Journal of Forensic Sciences, 55, p385-393

including false positive fingerprint identifications[7,8] have brought the issue into sharp relief. This has been reinforced by an assessment of forensic science published in 2009 by the US National Academy of Sciences in which a diverse range of forensic disciplines within the USA were identified to have wide-ranging issues including lack of validation, standardisation, reliability, accuracy and potential for bias[9].

6.2 Categories of cognitive bias

6.2.1 There are a number of categories of cognitive bias, including those described briefly below; some are very similar and can sometimes apply in combination in real life situations. Further information on different sources of bias in forensic science is provided in a paper by Dror[10].

6.2.1.1 **Expectation bias**, also known as experimenter's bias, is where the expectation of what you will find affects what you do actually find i.e. where there is scope for ambiguity, people only see what they expect to see. For example, an experimenter may disbelieve or downgrade the significance of findings that conflict with their original expectations, whilst believing and certifying material that supports preexisting expectations. This is also closely related to observer expectancy effects in which a researcher unconsciously manipulates an experiment or data interpretation in order to find a result consistent with expectations.

6.2.1.2 **Confirmation bias** is closely related to expectation bias, whereby people test hypotheses by looking for confirming evidence rather than potentially conflicting evidence[11,12]. For example, in the evaluation of DNA mixtures, if the reference sample is compared before the crime profile has been interpreted, confirmation bias would result if the analyst then looked only for features supporting the inclusion of the reference profile within the mixture. Some verification processes have potential for confirmation bias if the verifier has knowledge of the original examiner's findings before reaching their own conclusions. They may also be influenced by the experience or status of the previous examiner where these are known to them (so-called conformity effects, and institutional bias).

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[6] Dror, I.E., Peron, A.E., Hind, S.-L. & Charlton, D. (2005), When emotions get the better of us: The effect of contextual top-down processing on matching fingerprints. Applied Cognitive Psychology, 19, p799-809.

[7] Office of the Inspector General (2006). A review of the FBI's handling of the Brandon Mayfield case. Office of the Inspector General, Oversight & Review Division, US Department of Justice.

[8] Campbell, A. (2011). The fingerprint inquiry report. Available at: http://www.thefingerprintinquiryscotland.org.uk/inquiry/3127-2.html

[9] NAS. (2009). Strengthening forensic science in the United States: A path forward. Washington, DC: National Academy of Sciences, National Academies Press.

[10] Dror, I.E. (2009) How can Francis Bacon help forensic science? The four idols of human biases. Jurimetrics, 50, p93-110

[11] Balcetis, E., Dunning, D. (2006) See What You Want to See: Motivational Influences on Visual Perception, Journal of Personality & Social Psychology, Vol.91, No.4, p612-625

[12] Sanitioso, R., Kunda, Z., Fong, G.T., 1990. Motivated Recruitment of Autobiographical Memories, Journal of Personality & Social Psychology, 59 p229-241

6.2.1.3 Examples such as a request to "Quickly check this match" demonstrate the potential for confirmation bias in verification processes.

6.2.1.4 **Anchoring effects** or focalism is closely related to both the above and occurs when an individual relies too heavily on an initial piece of information when making subsequent judgements, which are then interpreted based around the anchor. For example investigators may fix too readily on a specific subject early on in an investigation and look to explain the circumstances around that person, whilst subsequently ignoring simpler alternative explanations of what may have happened, or who else may have committed the crime.

6.2.1.5 **Contextual bias** is where someone has other information aside from that being considered which influences (either consciously or unconsciously) the outcome of the consideration. Psychological research has demonstrated that perception is responsive to both the individual's psychological and cognitive state along with the environment in which they are operating. For example, a scientist working within a police laboratory could be influenced by knowing that detectives believe they have a strong suspect, or that the suspect has already confessed to having committed the crime. Provision of information not required by the scientist to undertake their evaluation and that potentially influences this type of biasing has been termed 'psychological contamination' or 'cognitive contamination'[13], as opposed to the more widely understood issue within forensic science of 'physical contamination'[14].

6.2.1.6 **Role effects** are where scientists identify themselves within adversarial judicial systems as part of either the prosecution or defence teams, and this may introduce subconscious bias which can influence decisions especially where some ambiguity exists. In fibre examinations when potential contact between two textile items is under consideration but no matching fibres are found, cognitive bias may be seen from a scientist acting on behalf of the prosecution, and interpreting the findings as neutral rather than considering whether the absence of matching fibres might support the view that the contact had not occurred. Role effects are differentiated from a similar effect called motivational bias, which is often considered separately to cognitive biases. Motivational bias occurs where, for example, motivational influence on decision making results in information consistent with a favoured conclusion tending to be subject to a lower level of scrutiny than information which may support a less favoured outcome[15,16]. An extreme example of this is where an individual wants one side

[13] Dror, I.E. (2013) Practical solutions to cognitive and human factor challenges in forensic science. Forensic Science Policy & management 4 p1-9.

[14] Kassin, S.M. et al (2013). The forensic confirmation bias: Problems, perspectives, and proposed solutions. Journal of Applied Research in Memory and Cognition. 2, p42-52

[15] Pyszczynski, T., Greenberg, J., 1987 Toward an Integration of Cognitive & Motivational Perspectives on Social Inference: A biased Hypothesis-testing Model, Advances in Experimental Social Psychology, vol 20 p297-340.

[16] Dawson, E., Gilovich, T., Regan, D. T., 2002 Motivated Reasoning and Performance on the Wason Selection Task, *Personality & Social Psychology Bulletin*, 28 p1379-1387

to win and when in doubt will always make a conscious decision in one direction i.e. to routinely inculpate (or conversely exculpate) suspects; examples of such misconduct have been well documented[17].

6.2.1.7 **Reconstructive effects[18]** can occur when people rely on memory rather than taking contemporaneous notes: people tend to subsequently fill in gaps with what they believe should have happened and so may be influenced by protocol requirements when recalling events some time later from memory.

6.3 Academic research into cognitive bias in forensic science

6.3.1 Academic research into cognitive bias in forensic science, conducted through both experimentation and identification of examples from past cases, has indicated that effectively any technique or process which includes subjective assessment and comparison is potentially susceptible to bias. A particularly useful overview of this topic has been published recently by Kassin et al[19]. Other research papers have describe studies on bias in DNA mixture interpretation[20], fingerprint comparison[21,22], handwriting comparison[23], fire investigation[24,] forensic odontology[25], bullet comparisons[26], hair comparison[27], and forensic anthropology[28]. The extent of the issue in real life has yet to be fully evaluated, however it is likely to be highly variable depending on the type of forensic analysis being conducted and the extent of safeguards built into the

[17] Giannelli P.C. (2010) Independent crime laboratories: the problem of motivational and cognitive bias: Utah Law Review 2, p247-256

[18] Risinger, D.M. et al (2002) The Daubert/Kumho Implications of Observer Effects in Forensic Science: Hidden Problems of Expectation and Suggestion Author(s): California Law Review, Vol. 90, No. 1, pp. 1-56

[19] Kassin, S.M. et al (2013). The forensic confirmation bias: Problems, perspectives, and proposed solutions. Journal of Applied Research in Memory and Cognition. 2, p42-52

[20] Dror, I. & Hampikian, G. (2011). Subjectivity and bias in forensic DNA mixture interpretation. Sci. Justice 51 p204-208

[21] Dror, I. et al (2006 check) Contextual Information Renders Experts Vulnerable to Making Erroneous Identifications: Forensic Science International 156 74-78

[22] Dror, I.E & Charlton, D. (2006) Why experts make errors, J. Forensic Identif. 56 600–616.

[23] Found, B. & Ganas, F. (2013) The management of domain irrelevant context information in forensic handwriting examination casework, Sci. Justice 53 p154–158.

[24] Bieber, P. (2012) Measuring the impact of cognitive bias in fire investigation. International symposium on fire investigation. Sci. Technol. (2012) p3–15.

[25] Page, M. et al (2012), Context effects and observer bias—implications for forensic odontology, J. Forensic Sci. 57 p108–112.

[26] Kerstholt, J., Eikelboom, A., Dijkman, T., Stoel, R., Hermsen, R., van Leuven, B., Does suggestive information cause a confirmation bias in bullet comparisons? (2010) *Forensic Science International* **198** 138–142

[27] Miller, L. (1987) Procedural Bias in Forensic Science Examinations of Human Hair, Law and Human Behaviour 11(2) p157-163

[28] S. Nakhaeizadeh, et al., Cognitive bias in forensic anthropology: Visual assessment of skeletal remains is susceptible to confirmation bias, Sci. Justice (2013), http://dx.doi.org/10.1016/j.scijus.2013.11.003

processes within which organisations or individuals are working. From a global perspective, it will also depend on the overarching quality requirements and expectations of the particular justice system within which the outcomes are delivered.

6.4 Bias countermeasures (also known as "Debiasing techniques")

6.4.1 Blinding precautions

6.4.1.1 Providing the forensic examiner only with information about the case that is required in order to conduct an effective examination is the most powerful means of safeguarding against the introduction of contextual bias. Such information could be for example a statement from the victim, and for this reason direct contact with the investigating officer should be avoided prior to assessment. That said, it should be borne in mind that the information required may vary from case to case, and it is hard to perform case assessment and interpretation effectively without having access to background information. For example, targeting effectively for "touch" DNA may require information from witness statements.

6.4.1.2 Most forensic science providers would be able to control the flow of information to analysts, however some forensic science practitioners are in sole practice and the instructing agency needs to have role and therefore a working knowledge. In such situations, the practitioner may need to ensure the officer in the case is well aware of appropriate information, images and disclosure through the investigation.

6.4.1.3 Good practice in forensic science requires that independent checking of critical findings is undertaken (Codes 15.3.2). Independent checking that minimizes the risk of cognitive bias would entail assessment without knowing the outcome of the initial analysis, or even where practicable the identity of the original examiner in order to avoid confirmation bias.

6.4.2 Structured approach

6.4.2.1 Application of a structured approach to performing a comparison and arriving at a decision using an essentially "linear" process can effectively reduce or eliminate the influence of the target (i.e. information pertaining to suspect) from the conclusions drawn. A good example of a general methodology for undertaking comparisons is "Analysis, Comparison Evaluation and Verification" (ACE-V). It is the most commonly accepted approach to fingerprint comparison in the UK and USA. The sequence of working is: i) an examiner analyses a mark: ii) the examiner then compares the mark to a known print: iii) having compared the images, the examiner evaluates what they have seen and reaches a decision iv) the results are then subject to verification by one additional examiner or more. Although most literature sets out the ACE-V process as a sequential process it is in fact not linear in application to fingerprint comparisons – the Analysis phase can be revisited in a well-structured way

during the comparison phase. However the evaluation is a separate stage as described.

6.4.2.2 Another framework that has been applied to give structure to the evaluation of scientific findings is the Case Assessment and Interpretation (CAI) model[29,30]: this helps scientists design effective, efficient, and robust case-examination strategies. The CAI model is founded on Bayesian[31] thinking and provides clarity on the role of forensic scientists within the criminal justice process. It also encourages consistency of approach, and helps direct research effort. In common with ACE-V it describes an approach in which examination and analysis of scene-related material is undertaken prior to assessment. However whilst ACE-V often entails some re-iteration of the assessment process, CAI is essentially a linear approach and both provide a practical means of safeguarding against confirmation bias. Further information on the CAI-type approach is given in section 7.

6.4.3 Method development

6.4.3.1 As the potential for cognitive bias arises at different stages in the examination process, method development ought to look at risks or perceived risks in the method and apply the most practicable control strategy. It ought to be borne in mind that simply because there is a risk of an event, it doesn't mean it automatically manifests itself affecting critical judgment.

6.4.3.2 Having a complete picture is often vital for constructing and testing relevant hypothesis and propositions. However if knowing about certain aspects are assessed to work against the objective process in a particular method (i.e. assessment recommends a blinding method is used), then the methodology right down to design and content of paperwork as well as interaction with the officers in the case might be considered. If the whole case file is handed over to an analyst with all the extraneous detail, then even if there is no perceptible bias there is the perception that it could have occurred and may be open to challenge in court.

6.4.4 Awareness, training and competence assessment

6.4. It is not sufficient to simply have well defined evaluation procedures in place as outlined above: practitioners need to be aware of the risks and issues arising from cognitive bias, and to receive substantial training in how to overcome these in their respective roles. Similarly those involved in method development require training regarding the risks and issues so that they are best equipped to design out cognitive bias from processes as far as is practicable.

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[29] Cook, R. et al (1998a) A model for case assessment and interpretation. Science and Justice 38: 151-156.

[30] Association of Forensic Science Providers. (2009). Standards for the formulation of evaluative forensic science expert opinion. Sci. Justice 49, 161–164.10.1016/j.scijus.2009.11.004

[31] The use or application of Bayes' Theorem, a mathematical formula that can be applied to update probabilities of issues in the light of new evidence.

6.4.4.2 Given that susceptibility to psychological and cognitive influences varies between individuals, there may be merit in assessing these susceptibilities as part of the recruitment or selection procedures for new staff, such as the recruitment testing procedure for fingerprint examiners developed by Dror et al[32]. Competence in applying evaluative processes should be formally assessed prior to commencing casework and thereafter on a regular basis. This may be achieved through a proficiency testing programme, utilizing mocked up casework samples for which the expected outcomes of testing and evaluation are known. Whilst blind trials are effectively the gold standard in providing the most reliable indicator of real-life performance, in reality they can be very time-consuming and challenging to set up, especially in avoiding alerting the person being assessed that it is a trial rather than another piece of casework. Good practice adopted by many laboratories is to undertake a mixed programme of both declared and undeclared trials, with the proficiency of all individuals tested on a regular basis.

6.4.5 Avoidance of reconstructive effects

6.4.5.1 The taking of contemporaneous notes or technical records is another stipulation in the Codes (section 15.2.3) Adherence with this requirement wherever it is practicable to do so at and at all stages in the collection and processing of forensic evidence provides the best safeguard against potential reconstructive effects.

6.4.6 Avoidance of role effects

6.4.6.1 Role effects whereby scientists are subconsciously influenced by acting on behalf of the defence or prosecution are difficult to demonstrably eliminate given the adversarial nature of the CJS within the UK, and which are potentially compounded by the pressures of a commercial market in which a supplier/customer relationship for the delivery of forensic science is the norm. These pressures apply whether an FSP is providing contracted services to the prosecuting side or to the defence, or in the case of police laboratories in providing services to an internal customer.

6.4.6 However a wider customer is being served here i.e. the CJS, not just the defence or prosecution sides paying for the services: the Regulator's Codes of Conduct for forensic science stipulate that practitioners shall:

a. Have an overriding duty to the court and to the administration of justice, and,
b. Act with honesty integrity and impartiality.

6.4.6.3 This is reinforced in section 7.2 of the Regulator's Codes of practice, in which conflicts of interest, perceived or otherwise, and threats to impartiality of a practitioner are identified, including the following:

a. Being the sole reviewer of their critical findings.

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[32] Charlton, D., Fraser-Mackenzie, P.A.F. & Dror I.E. (2010). Emotional experiences and motivating factors associated with fingerprint analysis. Journal of Forensic Sciences, 55, p385-393

b. Being over-familiar with or trusting another person instead of relying on objective evidence.
c. Having organisational and management structures that could be perceived to reward, encourage or support bias, where for example a culture of performance measurement and time pressures could potentially pressurize examiners into biasing decisions.

6.4.6.4 Whilst point c) may be erring towards misconduct rather than being a cognitive phenomenon, the overriding issue with all these points is the effect of subconscious influences on impartiality. Furthermore, compliance with the ISO 17025 quality standard which is an integral requirement of the Codes stipulates that personnel undertaking the analyses shall be free from any undue commercial, financial and other pressures which might influence their technical judgement. In other words, organisational systems and safeguards are required to ensure scientists are insulated from potential biasing pressures.

6.4.6.5 The Criminal Procedure Rules state in part 33.2 that (1) An expert must help the court to achieve the overriding objective by giving objective, unbiased opinion on matters within his expertise; (2) This duty overrides any obligation to the person from whom he receives instructions or by whom he is paid; (3) This duty includes an obligation to inform all parties and the court if the expert's opinion changes from that contained in a report served as evidence or given in a statement. Every expert report must contain a statement that the expert understands his duty to the court, and has complied and will continue to comply with that duty.

6.4.6.6 Adoption of a structured approach such as the CAI principles as described in 4.3.1.2 and expanded further in section 6 below, in which consideration of both prosecution and defence hypotheses, can help ensure evidence is evaluated and presented in a more balanced manner, regardless of defence or prosecution role. This requires that:

a. Experience is brought to bear by a person who has all the information regarding the case in formulating a coherent strategy that underpins the rationale for analytical submissions;
b. Analysis is undertaken only with relevant facts disclosed to the analyst; and,
c. The results of the analysis are reviewed and interpreted from the perspective of the whole case, and should accept the conclusions drawn by the analyst.

7. A GENERIC PROCESS TO MANAGE COGNITIVE BIAS FOR A RANGE OF FORENSIC EVIDENCE TYPES

7.1 The role of the investigating officer or instructing authority

7.1.1 Appropriate flow of information is very important in all cases, one limiting factor in the assistance forensic science can give to the investigation is pertinent information not being passed on. Contextual or case information can be made available for the leading examiner for case building purpose, the lead can then ensure analysts receive information appropriate for that stage, while still

ensuring proper case assessment can be made and the most appropriate techniques are used.

7.1.2 However, when instructing experts in sole practice, a greater onus is placed on the investigating officer (or instructing authority) to manage the flow of information. The expert is still likely to need the contextual or case information, but this may be required to be held back until certain analytical stages are complete.

7.1.3 However, anybody instructing experts should always think hard about including comments such as the 'suspect admitted to the crime', 'we already have a DNA match', or even in the question asked '…can you identify whether suspect A (the stabber) is carrying anything and, if he is, what that item is…' Being exposed to such information doesn't automatically result in a biased decision, but it can influence and should be guarded against.[33]

7.1.4 The investigating officers or instructing authority should deals with the following in their forensic strategy:

a. information flow based upon the nature of the evidence type, the phase of the analysis and the capability of the forensic science provider.

i. Is the provider able to apply any debiasing techniques themselves i.e. a larger provider will probably control the flow of information to the analyst?

ii. Is this a smaller provider or niche specialism where the lead examiner is the sole examiner? If this is the case then agree with them beforehand how the initial, and sometimes follow up, communications might be best handled.

7.2 The role of the scientist in the analysis or initial evaluation stage

7.2.1 The analyst should know through their training that they must stay separate from the rest of the investigation and accept the fact that they should undertake the analysis "blind", and not to seek other information beyond what is required, in order to protect their impartiality. If potentially biasing information is inadvertently disclosed to them, for example that someone is in custody or has confessed, the lead scientist should be informed that this has happened.

7.3 The role of a forensic expert

7.3.1 The role of the forensic science expert is to evaluate scientific findings and the results of analytical tests in the context of the relevant case circumstances. An expert opinion should meet the criteria that it is balanced, robust, logical and transparent[34]:

[33] In R v Rogers [2013] EWCA Crim 2406 the Court of Appeal (Criminal Division) rejected the argument the admission of a police officer's identification of the accused from photographs after being informed that there was a DNA match rendered the trial unfair or conviction unsafe.

[34] Cook, R. et al (1998a) A model for case assessment and interpretation. Science and Justice 38: 151-156.

a. Balanced – the expert has considered both the prosecution and defence views in their evaluation
b. Robust – it is based on data that are available for inspection and discussion
c. Logical – in the approach taken to the evaluation
d. Transparent - another suitably qualified scientist could follow all the steps and decisions taken[35].

7.3.2 If all of the above criteria are met, then any difference of opinion between experts could be limited to a well-defined part of the opinion rather than being a general disagreement, as well as identifying the reasons for each of the opinions. This is most helpful to the court in identifying the areas of dispute between scientists.

7.4 Process Outline

7.4.1 A very brief outline of forensic process within the laboratory is as follows:

a. Define requirement
b. Develop examination strategy
c. Agree examination strategy with client
d. Carry out forensic examinations and analyses
e. Review quality and content of examination results
f. Compare the results with the reference samples and marks
g. Evaluate and interpret the scientific findings and analytical tests
h. Verification by second expert
i. Communicate the scientific findings and analytical tests

7.4.2 During this process it is the responsibility of the expert to record, retain and reveal their work. This requires that they:

a. Record all information received
b. Record details of interpretation

7.4.3 Risks of cognitive bias

7.4.4 If it is not practical to mitigate or control the main forms of cognitive bias then the following may occur:

a. An incorrect conclusion may be made.
b. A critical check might be inadvertently administrative or cursory

7.4.5 The evidence may be challenged.

7.4.6 The risks associated with relying on the scientific findings and analytical results as a way of assigning a weight of evidence are that:

7.4.7 It can be difficult to consider alternative hypotheses since knowledge of the actual outcome provides a source of confirmation bias.

[35] Association of Forensic Science Providers. (2009). Standards for the formulation of evaluative forensic science expert opinion. Sci. Justice 49, 161–164.10.1016/j.scijus.2009.11.004

7.4.8 The limitations of the examination and tests performed can be overlooked when evaluating the findings.

7.4.9 Risk management in all disciplines usually starts with an assessment, and a process map detailing the critical control points as required in the Codes (19.4.2.) for building in contamination controls during method is development may be useful for this purpose. This practice should identify the stages where individuals being knowledge rich is not ideal and stages where being knowledge poor is damaging. This approach can inform the examination strategy as well as communication strategy. As the officer in the case may have a role, such a visual tool might be included in officer awareness training or supplied as service information.

7.5 Mitigation strategies to reduce the risk of cognitive bias:

7.5.1 The expert goes through a formal process of pre-assessing the expected probabilities for an exhaustive range of possible outcomes, in as many or as few categories as is sensible for the examination, recording their opinions.

7.5.2 Each category in the exhaustive list of outcomes is considered firstly under the assumption that the prosecution hypothesis is true, and secondly under the assumption that the defence hypothesis is true.

7.5.3 These are used to provide an expected outcome which may be either qualitative or quantitative with the latter expressed as a Likelihood Ratio (LR).

7.5.4 The background data and experience used for assessing the expected outcomes are documented and any gaps identified.

7.5.5 A second expert carries out the same process independently, without viewing the decisions made by first examiner and the experts jointly agree the expected outcomes.

7.5.6 Posterior probabilities are not provided for evaluation of findings[36].

7.6 Recommended good practice

7.6.1 Define requirement[37]:

a. Identify whether the scientist's role in the case is investigative (intelligence) or evaluative (judicial).
b. Seek clarity on which tests are required, the purpose and how this fits into the hierarchy of sub-source (e.g. touch DNA), source, activity and offence level propositions[38,39].

[36] The posterior probability is the conditional probability assigned after the scientific evidence has been taken into account; so considers the probability of the hypothesis *given* the evidence. This is an example of the prosecutors fallacy or transposed conditional. The scientist should provide the probability of the evidence *given* the hypothesis.

[37] Cook, R. et al (1998b). A hierarchy of propositions: Deciding which level to address in casework. Science and Justice 38:231-239.

7.6.2 Develop examination strategy:

a. Formulate relevant prosecution and defence alternatives based on the case circumstances and information provided.
b. Consider any agreed assumptions that are used in formulating these alternatives.
c. Use assessment of possible outcomes to determine which tests are most informative and discriminating.
d. Use this pre-assessment to assign a weight to an exhaustive list of possible outcomes, giving the expected outcome for each, expressed as a Likelihood Ratio (LR) where these are quantitative.

7.6.3 This approach provides clarity on the alternatives being considered, and the pre-assessment of weight for all outcomes avoids the potential bias of using the observed results to assign weight of evidence.

7.6.4 **Carry out forensic examinations and analyses**

7.6.5 Review quality and content of examination results: decisions on the suitability of the results and marks for later comparison are made at this stage, to avoid post-comparison rationalisation of opinion on quality.

7.6.6 Compare the results with the reference samples and marks: quality and suitability of the questioned result has already been assessed so this is not influenced by the reference result.

7.6.7 **Evaluate and interpret the scientific findings and analytical tests**

7.6.8 Verification by second expert: independent review at this stage in advance of communicating the result to the client.

7.6.9 Communicate the scientific findings and analytical tests.

7.6.10 Interpret the scientific findings and analytical tests:

a. Confirmation bias is mitigated by using the LR or qualitative expectation which has already been assigned to each outcome, before the examinations and tests have been performed.
b. Pre-assessment enables the scientist to explain how the weight of evidence has been assigned.
c. Provide details of the assumptions that have been made.
d. Give the basis of the expert opinion and specify the propositions considered, with reasoning for these, based on the case context.
e. Include any limitation of the opinion.
f. Describe the range of other opinions.

[38] Jackson, G. et al (2006) The nature of forensic science opinion--a possible framework to guide thinking and practice in investigations and in court proceedings. Science & justice : Journal of the Forensic Science Society 46, 33–44.

[39] RSS Practitioner Guide No 4: Case Assessment and Interpretation of Expert Evidence, Graham Jackson, Colin Aitken, Paul Roberts.

8. GOOD PRACTICE GUIDELINES - SCENES OF CRIME

8.1.1 The police response to a reported crime requires many factors to be taken into consideration and for priorities to be balanced accordingly. Preserving the scene, securing evidence, speed of response including making most effective use of the "Golden Hour", proportionate use of resources based on the seriousness of the crime: all are potentially conflicting in their requirements, and all are overridden by the most pressing priority of all, the preservation of life.

8.1.2 Within this context and from the outset of the investigation, the investigative team seeks to answer many questions that will assist in making sense of the incident under investigation. Frequently the answers to these questions can be provided by material which is obvious and readily to hand, but there will also be gaps. The latter may be filled by gathering of further information or material, identified during the course of the investigative decision-making process, and which may be present at the scene of crime, at other related sites or from other sources[40].

8.2 Scene of crime process

8.2.1 Serious crime

8.2.1.1 In major or serious crime investigations, forensic science resources are called upon by the Crime Scene Manager to attend the scene based on the specific needs of a case, especially where other evidence to detect the case is not readily available, and these resources are in proportion to the seriousness of the crime. Prior to entering the secured and controlled scene the examiners (e.g. Crime Scene Examiners, forensic scientists) are briefed regarding the scenario being evaluated and the questions that need to be answered. However, the emphasis here is on ensuring that relevant expertise is deployed with the capacity to look at the case and the inquiry to determine what value may be added and what inferences may be drawn from the collection and analysis of physical evidence[41].

8.2.2 Volume crime

8.2.2 The process for volume crime is markedly different to serious crime, due primarily to significant financial constraints impacting on time, personnel and other resources available. Therefore these processes deployed are about maximizing the benefits from these limited resources as a whole rather than for each crime that is reported. The process constitutes the following steps:

8.2.2.2 On notification of a crime, the police call handler has to make a decision based on information received, and guided by force policy regarding response to volume crime incidents, on whether or not to dispatch a police officer to attend.

[40] National Centre for Policing Excellence (2006) Murder investigation manual

[41] Tilley, N. & Townsley, M. (2009) Forensic science in UK policing: strategies, tactics and effectiveness. Published in Handbook of Forensic Science eds J. Fraser & R. Williams p359-379

8.2.2.3 If a police officer is dispatched to attend the scene they may collect physical evidence themselves or will determine whether a crime scene examiner is to be called to examine the scene for any physical evidence.

8.2.2.4 If an examiner attends the scene, they may be briefed regarding the offence and what might be most usefully looked for, in advance of their searching for and recovering physical evidence from the scene.

8.2.2.5 Recovered evidence is packaged labelled and transported back to police facilities, after which a decision is made on what if any evidence is subsequently processed[35].

8.2.3 **Crime scene activities and risk of bias**

8.2.3.1 Whilst some crime scene studies have been published by criminology specialists[42,43], cognitive bias at scenes of crime has been less comprehensively evaluated than other areas of forensic activity. Nevertheless its potential impact may be significant: for example, it could result in failure to secure the required evidence if a crime scene investigation is closed prematurely resulting in crucial evidence being lost; it could mislead an investigation by investigators focusing too early and incorrectly on a false lead, so that other evidence is potentially overlooked; or if undertaken incorrectly activities could result in "psychological contamination" of evidence downstream in the forensic analysis and interpretation processes.

8.2.4 Both volume and serious crime scene activities may be prone to errors and bias. For volume crime, given the severe time constraints, there is little scope to undertake anything more than a basic examination and recovery of evidence: focus is likely to be concentrated on the aspects of the case which are known from past experience to be most likely to yield fruitful results, e.g. fingerprints and DNA collection at the point of entry in a house burglary or vehicle theft, and on items which may have been handled or discarded at the scene, which the victim may be able to assist in identifying. Conversely, in major crime, context may be more of an issue with a risk that forensic strategies are written with a pre-conceived 'story' in mind.

8.2.5 Opportunities for cognitive bias can be usefully considered within the context of activities related to the crime scene, which can be categorised are as follows, as applied to serious crimes unless otherwise stated and is adapted from a conference presentation[44]:

[42] Lingwood, J., Smith, L.L., & Bond, J.W. (in press) 'Amateur vs professional: Does the recovery of forensic evidence differ depending on who assesses the crime scene?' International Journal of Police Science and Management

[43] Adderley, R., Smith, L.L., Bond, J.W., & Smith, M. (2012) 'Physiological measurement of crime scene investigator stress' International Journal of Police Science and Management 14 (2): 166-176.

[44] Fraser, J. (2013) Crime scene examination –final frontier or forgotten function? Paper presented at Forensic Horizons 2013: supporting research and development & delivering best practice for the justice system

8.2.6 **Gathering of information prior to scene attendance**

8.2.6.1 Prior to scene attendance information is gathered from any available source regarding the incident to be investigated. This may include witness or victim accounts as to what is alleged to have happened and by their nature these may be consciously or unconsciously biased. With volume crime, decisions on whether or not to attend the scene may be based on this potentially biased information and could therefore affect whether the crime is even investigated at all.

8.2.7 **Controlling the forensic process at scenes**

8.2.7.1 This entails creating inner and outer cordons to secure the scene, and establishing a common approach pathway. The cognitive processes entail determining locations and boundaries of the scene and the entry/exit points of the offender, based on observations, information received and inferences. Whilst there may be scope for bias to affect these decisions for example the past experiences of an individual on which they may base their decisions are subjective may not be reflective of typical scenes. However other factors may be more relevant, and have more impact in real life such as convenience: for example establishing the boundary by taping from lamppost to lamppost is commonplace simply because they are already there.

8.2.8 **Creating a record of the scene**

8.2.8.1 This includes image capture and writing notes and statements. The cognitive processes include selection of equipment, plus decisions on which images to capture, and entails assessment of the current case needs plus some anticipation of future needs. Depending on Force requirements, these may allow wide variation in how findings are documented and are therefore open to subjectivity. Depending on how the written record is crafted, there is a risk that contextual or confirmation bias may be introduced downstream in the investigative process. A gross example is “item X was recovered from suspect Y, a known repeat offender”.

8.2.9 **Undertaking forensic examinations at scenes**

8.2.9 This requires an understanding of the investigative needs of the case, plus to observe, discover and recover evidence to meet both these present needs and those anticipated for the future. If guidance for these decision-making processes is not explicitly documented then actions taken at this stage are largely reliant on the examiners intuition and tacit knowledge, which in turn are susceptible to bias.

8.2.10 **Packaging, storing, labelling and transporting recovered items**

8.2.10.1 These actions are largely procedural rather than cognitive. However there is still scope for introduction of psychological contamination if inappropriate information is included on the labelling of recovered items, as described in section 6.2.1.3.

8.3 Bias Countermeasures and good practice

8.3.1 It is impossible to undertake certain tasks effectively without being provided with context within which to operate, and this is certainly true with scenes of crime

investigations, where some briefing regarding the alleged crime and circumstances are an essential starting point for the examiner's activities. Examiners must be safeguarded against the risks of contextual and other biases through their training and through adherence to formal documented evidence-based guidance. Of necessity such guidance may be more prescriptive in volume crime where scenarios under investigation are relatively consistent scene to scene and are amenable to application of highly directive, standardised and efficient approaches. For example an examiner is better able to make a balanced and informed decision on which parts of a scene to sample for touch DNA analysis if they are armed with knowledge of Force-wide success rates from the substrates available, rather than relying on their own subjective experience of outcomes from just a few of their own cases. However it is also essential that volume crime investigators are trained not to "switch off": given their extensive experience of volume crime scenes, they are better placed than anyone else to identify anything slightly out of the ordinary and therefore potentially indicative of an alternative explanation to that posited by the victim which may be biased or even completely false, e.g. identify evidence that a "burglary" has been staged in order to make a false claim on insurance.

8.3.2 Serious crime investigations of necessity require much more latitude in terms of approach by examiners, although fact-based guidance regarding approaches at their disposal is just as important as in volume crime. Regardless of this latitude of approach it must be demonstrably systematic and it is essential that examiners fully and contemporaneously document information regarding their examination. The latter provides transparency to the process, and is of particular value in:

a. subsequently reviewing the case internally to identify whether issues may have been introduced due to bias, and
b. facilitating review by the defence[45].

8.3.3 Communication of the examiners findings to others through written reports rather than verbal updates, whilst slower, is preferable as the former provides less risk of introducing bias into the transfer of information.

8.3. The activities of examiners are guided at the outset by briefing regarding the scenario being evaluated and the questions that need to be answered (6.1.1). Some may be readily answered by material that is easily available but there will also be gaps that cannot be filled[46]. Under these circumstances good practice has been identified of building hypotheses which can help bridge the knowledge gap and indicate where further material may be gathered[47].

8.3.5 The key points when building hypotheses have been identified in this guidance as follows:

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[45] Butt, L. (2013) The forensic confirmation bias: Problems, perspectives, and proposed solutions – Commentary by a forensic examiner. Journal of Applied Research in Memory and Cognition 2 p59–60

[46] National Centre for Policing Excellence (2006) Murder investigation manual

[47] ACPO (2005) Practice Advice on Core Investigative Doctrine

a. Ensuring a thorough understanding of the relevance and reliability of all material gathered;
b. Ensuring that the investigative and evidential test has been applied to all the material gathered in the investigation;
c. Ensuring there is sufficient knowledge of the subject matter to interpret the material correctly;
d. Defining a clear objective for the hypothesis;
e. Developing hypotheses that 'best fit' with the known material;
f. Consulting colleagues and experts to formulate hypotheses;
g. Ensuring sufficient resources are available to develop or test the hypotheses;
h. Ensuring that hypotheses-building is proportionate to the seriousness of the offence.

8.3.6 This guidance emphasises that these assumptions must be developed objectively and that investigators should be aware of the dangers of making assumptions or believing that assumptions made by others are fact. It further states that where assumptions are used to develop hypotheses this should be made explicit.

8.3.7 In some circumstances where collection and analysis of physical evidence is complex spanning several different evidence types, a co-ordination and integration role is required to be undertaken by experienced forensic practitioners, termed crime scene coordinators, or 'Byford Scientists'. These liaise with senior investigating officers in overseeing the collection of physical evidence and ensuring that the disparate strands of forensic analysis are brought together and appropriate inferences are drawn[48]. This role was introduced after an HMIC inquiry into failings in the Yorkshire Ripper Inquiry[49] due to important leads not being followed up, and false ones being persisted with i.e. classic anchoring effects. It is also important that those undertaking this integration role are also aware of, and thereby safeguard against the fact that these activities are also fraught with potential bias and it may be appropriate under certain circumstances for the coordinators to act as gatekeepers for contextual information and only impart to practitioners information required to fulfill their tasks[50].

[48] Tilley, N. & Townsley, M. (2009) Forensic science in UK policing: strategies, tactics and effectiveness. Published in Handbook of Forensic Science eds J. Fraser & R. Williams p359-379

[49] Byford, L. (1982) Report by Sir Lawrence Byford into the police handling of the Yorkshire Ripper case. London: Home Office (Released in June 2006, under the Freedom of Information Act)

[50] Charman, S. (2013) The forensic confirmation bias: A problem of evidence integration, not just evidence evaluation. Journal of Applied Research in Memory and Cognition 2 (2013) 56–58

9. DNA MIXTURES GOOD PRACTICE GUIDANCE

9.1 Outline of the Forensic Process Involving DNA Mixture Interpretation

9.1.1 The generic forensic process that encompasses the interpretation and reporting of DNA profiling results, including complex DNA results, can be briefly described as follows and in figure 1:

a. Items are received along with case information and questions to be addressed by the scientific work.
b. The case information, supplied by the law enforcement customer, is used to direct the DNA recovery and analysis strategy, ideally within a framework of appropriate propositions.
c. If non-complex DNA results are obtained that match a suspect, an appropriate random match probability or Likelihood Ratio (LR) estimate is assigned.
d. If complex mixed DNA results are obtained that can be numerically evaluated the probability of the mixed result is calculated under appropriate prosecution and defence hypotheses and a LR is assigned.
e. If complex DNA results are obtained that do not lend themselves to statistical evaluation, in some circumstances, a qualitative assessment is made and an opinion about the significance of the DNA results can be put forward.
f. Findings are checked by a competent colleague/peer.
g. A statement or report is issued.
h. The scientist may be called to court to give oral testimony.

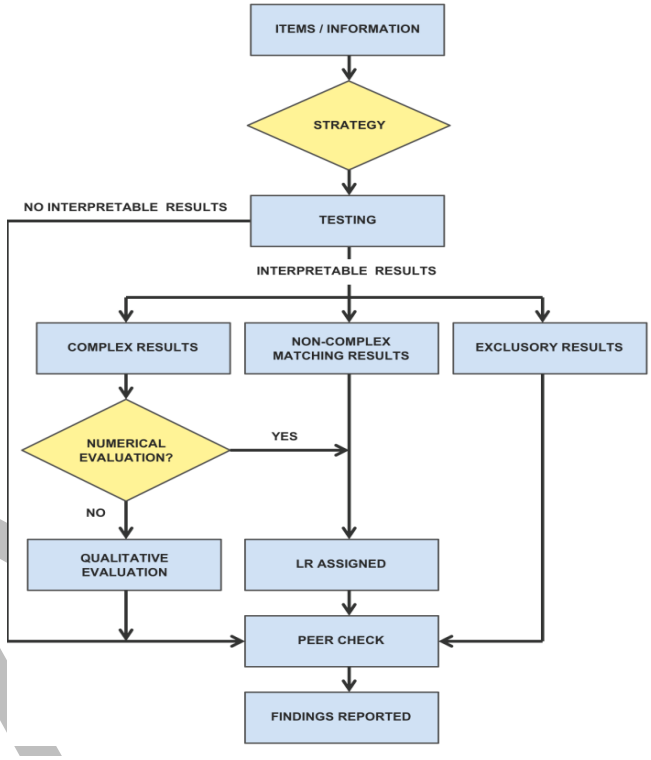

Figure 1: Outline of the Forensic Process Involving DNA Mixture Interpretation

9.2 The Risk of Cognitive Bias in DNA Mixture Interpretation

9.2.1 General Considerations

9.2.1.1 Just like other areas of science, the interpretation of DNA profiles can potentially be affected by some form of unconscious and unintended bias[51]. This can occur at points in the interpretation process where scientists are free to make decisions or put forward opinions that are formed outside of the mechanical application of a set of rules. Such opinions and decisions can be described as being subjective, since they arise from the individual's mental capabilities, relevant experiences, depth of knowledge and skill as well as any cognitive influences impacting on them at the time both manifest and unapprehended. Usually decisions are made and opinions are formed in the context of the information the scientist has been given about the case.

9.2.1.2 The interpretation of complex DNA mixtures requires care and skill and often includes a degree of qualitative and subjective decision-making. Indeed, regardless of any case-specific contextual information, practitioners may have a higher expectation of observing DNA profile matches simply because samples were submitted for analysis by police investigators.

9.2.2 General Conditions Impacting on the Level of Cognitive Bias Risk

9.2.3 Within DNA mixture interpretation there is a spectrum of bias risk that is shaped by multiple factors including the following:

a. Risks are low when results are clear and unambiguous and greater when results are complex, of poor quality and there is an increased reliance on subjective opinion.
b. Risks are lower when there is a methodical approach with defined standards built on principles that have been tested and validated, and greater when the approach is un-researched, ad hoc and personal to the operator.
c. Risks are lower when operators and checkers are well trained, experienced and continuously meet acceptable standards of competence; they are greater when operators and checkers are inexperienced, unmonitored and left to adopt their own approach.
d. Risks are lower when interpretation is checked by a competent peer who conducts a separate interpretation fully independent and without influence from the reporting scientist. Risks are higher when checking is less rigorous and/or conducted collaboratively.

[51] Dror, I. & Hampikian, G. (2011). Subjectivity and bias in forensic DNA mixture interpretation. Sci. Justice 51 p204-208.

Risk Source	Low risk	High risk
Result Quality	Results are clear and unambiguous	Results are complex, of poor quality and there is an increased reliance on subjective opinion.
Interpretation Approach	There is a methodical approach with defined standards built on principles that have been tested and validated	The approach is un-researched, ad hoc and personal to the operator.
Operator Competence	Operators are well trained, experienced and continuously meet acceptable standards of competence	Operators are inexperienced, unmonitored and left to adopt their own approach.
Checking	Full independent reinterpretation	Checking is conducted collaboratively

Table 1. Summary of Conditions Impacting on the Risk of Cognitive Bias

9.2.4 **Advancing Technology**

9.2.5 DNA testing technology continues to develop apace. In addition to the routine application of enhanced sensitivity techniques, today's new multiplexes frequently achieve results from low quantities of DNA (low template samples). The incidence of complex mixtures and of low template profiles exhibiting stochastic effects is increasing and so the conditions in which subjective opinion tends to be relied upon are more commonly encountered. As a consequence, there is an increasing risk of cognitive contamination affecting DNA evidence.

9.2.6 **Contemporaneous Case and Reference Sample Interpretation**

9.2.6 A substantial part of the risk relating to DNA mixture interpretation arises if the case sample is interpreted alongside the reference sample, or if the case sample interpretation is revised after examination of the reference sample. For example, during the interpretation of a two-person mixture (when the interpretation is not conditioned on the presence of an undisputed DNA source) knowledge of the reference sample may result in confirmation bias in the genotype combinations that are included or excluded as being possible, based on allele quantities.

9.2.7 **Use of Qualitative and/or Subjective Approaches**

9.2.7.1 Significant risk is also associated with the use of qualitative and subjective evaluation approaches that have increased considerably since the recent publication of the judgment in R v Dlugosz *et al* (R v Dlugosz, R v Pickering and R v MDS [2013] EWCA Crim 2). The Dlugosz judgment has been taken as a broad license to allow the qualitative evaluation of complex results and subjective expressions of evidential weight when a statistical approach is either difficult or considered inappropriate. Such non-statistical assessments can only

be conducted by comparing a reference sample directly with the complex result from the case sample and drawing conclusions based on the presence of alleles in common between case sample and reference sample, the absence of particular alleles and inferences from allele quantities. The Dlugosz judgment does specify safeguards that relate to whether or not such an evaluation can be considered admissible as evidence and how the evidence should be presented. The safeguards require that the expert is experienced, that the extent of their experience is explained for the consideration of the jury and that caveats relating to the limitations of the findings are clearly explained. Whilst the safeguards might seem reasonable they are dependent on the following underlying assumptions that might be considered dubitable in some circumstances:

a. That general familiarity with complex DNA mixtures and numerical evaluation methods is wholly relevant to the use of what is essentially a new and un-researched evaluative practice; and
b. Such experience enables the practitioner to form safe, reliable opinions relating to sources of DNA within complex mixtures.

9.2.7.2 To provide assurance in the use of methods that rely on the accuracy of such assumptions, it would assist if clear standards were developed relating to the circumstances in which such an approach is valid and when it is not. Also testing the performance of individual practitioners against developed standards would reduce the risk of inaccurate estimates of evidential strength having an impact in criminal trials. Current application of qualitative methods appears to be largely *ad hoc* without specifically designed controls. If effective quality, training and competency measures are in place, the impacts of cognitive contamination can be minimized.

9.2.8 Potential Oversights in DNA Interpretation Induced By Cognitive Bias

9.2.8.1 Unconscious cognitive bias has the potential to manifest itself as a skewed evaluation, partly because its influence can increase the likelihood of oversights during the DNA interpretation process. Some possible oversights are described below; most are applicable regardless of whether a numerical or qualitative approach is applied and, with most, the risk is either reduced or eliminated if an assessment is made without knowledge of the reference sample result. Examples include:

a. Restricted assumptions about numbers of contributors.
b. Automatic assumptions that a part of a mixture has originated from one individual.
c. Underestimating the significance of non-matching peaks when they can be considered sub-threshold or designated as artifacts.
d. Underestimating the uncertainty introduced by stochastic effects.
e. Overestimating the significance of unconfirmed matching peaks.
f. Underestimating the significance of unconfirmed non-matching peaks.
g. Taking account of matching alleles where their presence is uncertain due to masking by other components of the mixture.
h. Double counting peaks as homozygous that do not clearly represent a double contribution when the subject is homozygous.

i. Over emphasizing the absence of non-matching alleles when it is not clear if contributors are fully represented.

9.2.9 **Further Flaws Potentially Induced by Cognitive Bias**

9.2.9.1 The following points describe some further flaws that may be induced or exacerbated by cognitive bias. Most of these are afforded some latitude by the way in which disclosure tends to be approached by defendants and their representatives. The rules of disclosure within the legal system of England and Wales require no prior disclosure of the defendant's account. This often means that the DNA scientist is required to make their own, uninformed suppositions about appropriate defence hypotheses when deciding on analysis strategy and conducting their evaluation:

a. Greater focus on strategies for DNA recovery and testing that are likely prove a case rather than disprove a case.
b. Choice of propositions that maximize the strength of evidence against the suspect.
c. Observations that support the defence case are less rigorously considered or evaluated and are not given their true weight, particularly relating to the absence of evidence.
d. Failure to express alternative explanations.
e. Reluctance to express doubt particularly during oral evidence at court.

9.3 Case Examples Where Cognitive Bias May Have Contributed to Error

9.3.1 In this section, the identity of specific cases or the practitioners involved are not disclosed; rather, anonymised issues are described in several real cases that may have been caused or exacerbated by unintended cognitive bias. The examples are from cases in which the authors of this guidance had direct experience; all were reported in 2013. They stem from inaccurate evaluations or misleading descriptions of complex DNA mixtures, all biased in favour of the prosecution's case. It is, of course, not possible to be certain to what extent the issues were influenced by cognitive bias or some other source of inaccuracy but they illustrate the difficulties that relate to non-numerical evaluation of complex DNA results. As such, they are helpful in identifying procedural steps and controls that are likely to be effective to both limit cognitive bias and/or demonstrate that it has not occurred.

9.3.2 **Qualitative evaluation shown to be at odds with numerical evaluation**

9.3.2.1 A complex mixed DNA result from a case sample contained alleles in common with profiles in all four reference samples that were compared in the case. Most of the alleles in the case sample profile matched Subject X. No statistical analysis was conducted initially but, based on the reporting scientist's experience, s/he gave the opinion the result provided "at least moderate support" for the assertion that some of the DNA on the swabs came from Subject X. The results were later interpreted with the aid of LikeLTD[52], recently

[52] There are several relatively recently developed software programs that are available to providers and are designed to aid the numerical evaluation of some types of complex DNA profiles including complex mixtures.

developed software that is capable of numerical evaluation of some types of complex DNA mixture. The use of this software produced a LR of 4 indicating that, based on commonly accepted verbal descriptors, the strength of support should more fairly have been described as "weak".

9.3.3 **Implying the absence of alleles is due to masking by a major component**

9.3.3.1 One case relates to a duplicated, standard sensitivity test on vaginal swabs containing a trace of semen. A full, major component profile was obtained matching the complainant, together with a number of low-level minor component bands that were all present in the defendant's profile. Six duplicated bands in the minor component all matched the defendant and a further five unduplicated bands also matched the defendant. The unduplicated bands were described as unconfirmed. No other, non-matching, minor component bands were visible in either duplicate test and the ratio of the major component to the minor would not have allowed the identification of minor component alleles that were masked by the major component. Comparison of one duplicate result with the other showed that significant stochastic variation, including allelic drop-out, was a reality within these samples. It was not possible to tell whether or not there was full representation of the DNA source(s) within the minor component across the duplicates or to use peak quantities to determine whether there was more than a singular contribution from a specific minor component allele. In the presence of the jury, the scientist was invited to add up the number of alleles in the mixed profile that matched with the suspect's profile. The response was that there were six confirmed bands, five unconfirmed bands, seven that were shared with the major component profile and one further because the suspect was homozygous at one position. The scientist concluded that there were nineteen out of a possible twenty alleles matching the suspect within the mixed profile. There was no attempt to explain that the possible presence of minor component alleles in positions where the minor component would have been invisible was completely neutral to prosecution and defence hypotheses. There was a significant risk that this description of the evidence would be misleading to the jury in favour of the prosecution's case. There may be issues here relating to the approach to quality at the parent laboratory, in particular with the monitoring of competence and/or the support and training provided to reporting officers in the specialist field of low template mixture interpretation. Where there is a lack of understanding of evidence the potential for cognitive contamination is increased.

9.3.4 **Ignoring the possibility that a sub-threshold peak is an intrinsic allele**

9.3.4.1 This example relates to a major/minor mixed result from a standard sensitivity test in which a statistical evaluation of eight low level alleles in the minor component was reported. The low level alleles could only have been from the suspect if several of his alleles were not visible due to allelic drop-out. A sub-

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The following have been used in criminal trials in the UK: *LikeLTD*, developed by David Balding, Professor of Statistical Genetics at University College London. *STRmix*, developed by forensic experts at ESR Ltd in New Zealand (J. Bright and J. Buckleton) and at Forensic Science South Australia (D. Taylor). *TrueAllele*®, developed by Mark Perlin of Cybergenetics in the USA.

threshold peak, distinct from background and with acceptable allelic morphology was present in one of two duplicates and did not match an allele in the suspect's profile. The presence of this peak was presumably considered a spurious occurrence (drop-in or artefact) and was not taken into consideration for the purpose of the statistical evaluation; its presence was not otherwise mentioned in the scientist's report. Although this peak did not satisfy the criteria to be included as a confirmed component of the profile, further testing may have clarified the presence of the peak and if not, a more appropriate statistical approach could have been taken. Failing to take account of the peak or to attempt to replicate it through further work may have been a consequence of cognitive bias.

9.3.5 **Assuming all DNA bands in a low level profile are from the same person**

9.3.5.1 This assumption is often made but not always explicitly stated and, based on the quality of the profile and nature of the mixture, there are varying extents to which it can be justified. In low-level profiles it is important for the scientist to consider whether or not it is appropriate to use the result for comparison purposes and to consider the possible number of contributors prior to comparing to any reference sample. When mixed DNA profiles are interpreted alongside reference sample(s) without any prior assessment of their suitability for comparison, the risk of cognitive bias increases substantially.

9.3.6 **Only addressing the prosecution's case when a suspect cannot be excluded**

9.3.6.1 This relates to cases in which the complexity of the DNA result is such that it cannot provide evidence of inclusion but is only suitable to exclude individuals as a possible contributing source. The assertion that an individual cannot be excluded as a possible contributor to such a mixture is often reported without the qualification that there are many other individuals with different profiles who similarly could not be excluded. Only expressing an inability to exclude the presence of the defendants DNA from a case sample invites an interpretation by jurors that favours the prosecution's case more than is justified.

9.4 Mitigation strategies currently deployed in the UK and overseas

9.4.1 Below are examples of mitigation strategies that are variously used in current practice. All are experience-based examples of good practice in appropriate circumstances and should be applied as described:

9.4.2 Prior-interpretation of case sample result before reference result is revealed. Formally noting the following from the DNA result, prior to comparison with the reference profile:

a. suitability to include or exclude;
b. assessment of number of contributors;
c. level of representation of contributors;
d. potential for stochastic effects;
e. identification of likely/unlikely genotype combinations that might explain the mixture.

9.4.3 This is a critical step and is recommended for DNA profile interpretation in all circumstances.

9.4.4 Full checking via repeat interpretation by an experienced and competent colleague including prior-interpretation of case sample result before reference result is known. The check should be conducted independent of, and uninfluenced by, the reporting scientist, and should use original unmodified hard copy or electronic results that are free from annotation. This is a critical step and is recommended for DNA profile interpretation in all circumstances.

9.4.5 Case Assessment and Interpretation. Comparison of expected, pre-assessed outcomes with actual results under appropriate hypotheses. Some documented indication of expected outcome is recommended in all cases.

9.4.6 Careful selection of case stains/samples for testing to minimise the occurrence of mixtures and low template issues. Selection should be informed by case information and is good practice whenever case circumstances present a choice of DNA case stain targets.

9.4.7 Duplicate (or multiple) analyses to assess stochastic effects in low template samples. Replication is often used in conjunction with interpretation in a consensus framework, but can also be used prior to probabilistic evaluation of the results separately. Replication should be applied whenever a poor quality profile is to be relied upon to progress an investigation or provide evidence against a suspect. It assists in evaluating reproducibility, identifying spurious peaks and informing conclusions relating to the likelihood of allelic drop-out and the number of contributors. Replication allows a fuller understanding of the nature of the sample and reduces scope for conjecture and the risk of misinterpretation; it improves the scientist's ability to accurately gauge whether or not the sample is suitable for any form of comparison or statistical evaluation.

9.4.8 Analysis and interpretation is carried out blind, in the complete absence of any information about the case. This approach is practiced in some jurisdictions and eliminates the risk of some types of bias. It does present the practical challenge of separating case strategy, hypotheses testing, stain selection etc. from result interpretation and reporting in the context of the case. The risk of missing identification of realistic alternative explanations for the evidence given the case circumstances may be greater using this approach.

9.4.9 Use of recently developed interpretation software for complex mixtures[53] such as LikeLTD[54], STRmix™ (Institute of Environmental Science and Research (ESR) or TrueAllele® (Cybergenetics). Ideally should be used with all suitable results whenever other objective numerical methods are not appropriate. Efforts should be made to ensure practitioners are able to use them reliably whenever required.

[53] Suitable validation of all such methods would be expected prior to introduction in casework.

[54] A software package developed by David Balding, Adrian Timpson, Christopher Steele, Mayeul d'Avezac and James Hetherington. Further details available from: http://cran.r-project.org/web/packages/likeLTD/likeLTD.pdf [Accessed 27/08/2014]

9.4.10 Appropriate training of practitioners in the method employed, who can demonstrate initial and ongoing competency. This is a critical step and is recommended for DNA profile interpretation in all circumstances.

9.4.11 Transparency and disclosure of appropriate experimental data used to support conclusions and opinions. Research work should ideally be published in a peer reviewed scientific journal.

9.5 Further recommendations for good practice

9.5.1 In addition to the good practice described in 7.4 we also recommend the following:

9.5.1.1 When a numerical evaluation is not possible, it remains of crucial importance that qualitative and subjective judgments of pertinent profile features and their combined likelihood are assessed under the hypotheses framed by both the prosecution hypothesis (Hp) and defence hypothesis (Hd) separately. The final opinion of evidential weight must be based on how much, if any, comparison of separate assessments favours one hypothesis over the other, as with a likelihood ratio. For example, consider a complex mixture that cannot be conditioned on the presence of a known profile: If it is not possible to form a properly reasoned and reliable view about the probability that the mixture could arise if it came from a combination of unknown individuals (Hd), then the result can be of little, if any, probative value because half of the LR is unknown. If this approach is always adopted, it helps practitioners to identify when an observation favours neither prosecution nor the defence and is likely to prevent issues like those described in case examples 7.3.2 and 7.3.5.

9.5.1.2 Use a completely “blind” checker who repeats the full interpretation described in 7.4.2 but in the absence of any contextual information relating to the case. This may present practical challenges, particularly within smaller organisations. However, it will assist in a continuous learning and improvement cycle, where Reporting Officers can identify instances where they may have been affected by bias. Further, it provides assurance for the courts that the interpretation is free from contextual bias.

9.5. If there is no suitable option for objective evaluation, only employ qualitative and subjective based approaches that have been validated and therefore have demonstrated the robustness of resultant conclusions and opinions. Such procedures should include system performance data indicating when the approach breaks down and is no longer valid. The approach should be quality managed with defined standards and safeguards using trained staff who demonstrate initial and ongoing competence. It is also recognised that some scientists perform better than others under cognitive pressures and if a suitable measure can be adopted by providers this would help to mitigate the risks through improved staff selection, training and self-awareness.

9.5.1.4 Training and education in relation to the risks of cognitive bias generally and specifically in relation to complex DNA interpretation.

9.6 Further Research

9.6.1 The wider use of software packages (see note 50) capable of numerical evaluation of complex DNA results is likely to reduce the frequency with which

issues relating to subjectivity are encountered. However, such software does not yet offer a complete solution and there will continue to be a gap filled by non-numeric interpretation. Whilst best practice will minimise the inherent issues it is likely that there will continue to be a risk of cognitive bias and general disagreement between experts. We recommend continued research into objective methodology that will increase the power of DNA technology and improve the reliability and robustness of the evaluative processes for the benefit of criminal justice.

10. FINGERPRINTS GUIDANCE

10.1 Brief Outline of the Forensic Process

10.1.1 Every finger, palm or sole of foot comprises an intricate system of ridges and furrows, known as friction ridge skin. The arrangement and appearance of features within friction ridge skin are unique to each individual, persist throughout life and are accepted as a reliable means of human identification. Fingerprint Examiners are trained to interpret arrangements of ridge features and to report their opinion as to the common origin or otherwise of any two areas of friction ridge.

10.1.2 The fingerprint examination process consists of stages frequently referred to as Analysis, Comparison, Evaluation and Verification (ACE-V), terms which provide useful descriptors of the cognitive process undertaken by the examiner in arriving at their final opinion.

10.1.3 Each mark is analysed to establish the quality of detail visible within the mark and to determine its suitability for further examination taking account of variables such as:

a. The surface on which the impression was left
b. Any distortion arising from pressure applied when the impression was deposited
c. The clarity, quality and quantity of detail visible in the print.

10.1.4 During the comparison stage the examiner will systematically compare the ridge pattern and sequence of ridge characteristics in an impression from an unknown source with that of a known source impression. They will establish their opinion of the level of agreement or disagreement between the unique sequence of ridge characteristics visible in both impressions.

10.1.5 During the evaluation stage of the process the examiner will review all of their previous observations and come to their final opinion and conclusions about the outcome of the examination process. The ACE-V process is iterative in application with the analysis and comparison stages overlapping on occasion. The examination of a latent print against a known reference print may allow examiners to observe further features within the mark by directing their attention to areas, which require particular attention and further processing. This comparison activity may cause the examiner to reconsider their initial analysis of the mark and which could require further documentation by way of technical notes. The evaluation stage however remains a separate and distinct phase of the ACE-V process.

10.1.6 If the quality and/or quantity of detail visible within either or both impression is lacking, the examiner will record the impression(s) as **insufficient** and generally no further examination will occur. If the examiner is satisfied that the level of agreement between both impressions is sufficient to determine that they were made by a common donor, then they will consider the unknown impression **identified** to a particular individual. If the examiner feels that the level of disagreement between the two impressions is so significant that they are able to determine that both impressions could not have been made by the known donor, then they will consider that particular individual **excluded** as a potential donor of the unknown print. The examiner may conclude that, although there may be some agreement evident, the extent of disagreement and/or the quality and quantity of detail visible in both or either impression is such that it is not possible to come to a definitive conclusion at this time. In such a circumstance the examiner would consider the outcome of that examination to be **inconclusive**[55].

10.1.7 Although the process is often described sequentially, it is important to note that fingerprint examination is iterative in practice and each stage is not mutually exclusive throughout the process.

10.1.8 It is common practice across the fingerprint discipline globally that identifications are subject to verification by further examiner(s) who will conduct a personal analysis, comparison and evaluation of the impressions under examination.

10.1.9 Due to the subjective nature of the interpretative cognitive process undertaken by the examiner in arriving at their final opinion, it is accepted that the information used to come to conclusions may vary between examiners. For example, individual examiners may approach their examination from different starting points or consider the visible features in differing sequences; however, the original conclusions are shown to be reliable through demonstrating consistent end results from all subsequent examiners.

10.2 Risks of Cognitive Bias

10.2.1 The subjective, iterative and interpretative elements inherent within the fingerprint examination process expose the fingerprint examiner to a range of cognitive influences which, if not properly managed, could impact on the reliability of examination outcomes and examiner opinion.

10.2.2 Significant research has already been undertaken across the fingerprint discipline to explore the impact of cognitive influence and human factors on the examination process and the examiners personal decision-making behaviours. Studies undertaken to date have established that fingerprint examiners will, on occasion, alter their original opinions and conclusions in circumstances when

[55] Not every UK bureau use the same toolbox terminology at this time and 'inconclusive' may not be an option for some to use. This places a cognitive burden on the examiner to side with decisions that may lead to stronger biasing implication. To this extent 'inconclusive' could be a valuable tool to the decision-making armoury.

the original material is presented in a different context[56]. Further research has indicated that this influence is more prevalent when the impressions under examination are of poorer quality[57].

10.2.3 The risks of cognitive bias inherent in the fingerprint examination process can be categorised as contextual, confirmation and cultural.

10.2.4 **Contextual bias**

10.2.4.1 Fingerprint examiners are exposed to a wealth of contextual information which will impact on their decision making process such as;

a. Nature and details of the crime including background information
b. Association with or personal knowledge of the victim or their circumstances
c. Status of suspects or person(s) already in custody for the crime
d. Previous criminal activity of suspects or persons of interest
e. Location of the crime (an area close to their home)
f. Media or public interest associated with the crime
g. Personal moral codes or behaviours
h. Time pressure from investigating officers or office managers

10.2.4.2 For many organisations, contextual influence relating to crime type is in fact imbedded within their standard operating procedures. Crimes of a serious nature such as murder, rape and sexual assault are often given priority over other case work, have additional quality assurance measures in place or have specialist teams dedicated to this type of case work.

10.2.4.3 Prior knowledge of contextual information can influence the decision making process of a fingerprint examiner. For example, during an analysis an examiner may be more likely to retain an impression of borderline quality submitted as part of a serious crime than if the same impression was submitted as part of a low level volume crime. Prior knowledge of the status of an arrested person can lead to particular focus or emphasis on that individual to the exclusion of others.

10.2.5 **Confirmation Bias**

10.2 Within operational fingerprint bureaus, the majority of examination requests are received from police officers or prosecution services, with both hoping that the examination outcomes will help “solve the case” or “secure a conviction”. Contributing to the detection of crime is considered a fundamental aspect of fingerprint bureau service delivery. Also, personal identification or “hit” rates are used as key performance indicators at both organisational and individual level.

[56] Dror, I. et al (2006 check) Contextual Information Renders Experts Vulnerable to Making Erroneous Identifications: Forensic Science International 156 74-78

[57] Dror, I. et al (2005) When Emotions Get the Better of Us: The effect of Contextual Top-down Processing On Matching Fingerprints, Applied Cognitive Psychology, Wiley InterScience DOI:10.1002/acp 1130

10.2.5.2 Combined with a personal moral code to "do the right thing," this emphasis on "identification" as the most favoured hypothesis will exert powerful cognitive influence on examiner decision making.

10.2.5.3 Having prior knowledge of the previous examiner's findings and conclusions may also expose fingerprint examiners to the risk of confirmation bias and this will have a particular importance during the verification process.

10.2.5.4 At a technical level, examiners can be unduly influenced by confirmation bias when, having found a number of features from an unknown impression to agree with features in an impression from a known source, the examiner will then begin to reason backward, finding features in the unknown impression which are suggested by those in the known print rather than being visible without reference to the known source material.

10.2.5.5 Dror's paper "Practical Solutions to Cognitive and Human Factor Challenges in Forensic Science"[58] discusses the issue of base rate regularities and the impact of new technology into the fingerprint examination process. Within the context of automated fingerprint identification systems (AFIS) examiners become accustomed to having positive hits positioned at or near the respondent list. AFIS systems are designed to return those candidates most similar to the mark under search. The combination of heightened expectation of an identification being at top of the list along with the most similar candidates being returned at the top of the list carries with it an increased risk of cognitive influence on the decision making of fingerprint examiners.

10.2.6 **Cultural Bias**

10.2.6.1 Individual perception is influenced by the environment in which they are operating. Prior to the publication of The Fingerprint Inquiry Report in 2011, there was a tendency to represent the findings of fingerprint examiners as statements of objective fact rather than expressions of informed technical yet subjective opinion, albeit an opinion based on sound training and experience.

10.2.6.2 Historically, investigating officers and courts have accepted fingerprint evidence without challenge, which further contributed to the perception that fingerprint examination enjoyed "practical infallibility".

10.2.6.3 Operating in environments where differences of opinions are perceived as disputes with a "right" or "wrong" answer can also exert a powerful cognitive influence on examiners, leaving them reluctant to challenge their own or the findings of others.

10.2.6.4 Further examples of cultural influence which can impact on the decision making process include;

a. Strict hierarchical structures based on time served rather than competence.
b. Over confidence in individual or organisational competence.

[58] Dror, I.E. (2013) Practical solutions to cognitive and human factor challenges in forensic science. Forensic Science Policy & management 4 p1-9.

c. Lack of interaction with peers or exposure to alternative methods of working.
d. Lack of acceptance of the potential for errors or effective root cause analysis of errors.

10.2.6.5 The Fingerprint Inquiry report called for the profession to move away from any presentation of fingerprint evidence with 100% certainty, to fully explore the cogency of explanations offered for any evident differences between impressions and most importantly to recognise that fingerprint evidence is opinion evidence and as such is inherently subjective.

10.2.6.6 Any process which relies on the subjective personal interpretation of data as part of the decision making process is at risk from the influence of cognitive bias. This influence is typically exerted at an unconscious level and examiners often believe that their personal strategies are sufficient to mitigate any associated risk of cognitive bias. However experience has shown this not to be the case.

10.2.6.7 The challenge for the fingerprint profession is to adopt effective risk management strategies at individual and organisational level but without impacting on service delivery.

10.3 Examples where cognitive risks have become an issue

10.3.1 Brandon Mayfield Case 2006

10.3.1.1 In May 2004 Brandon Mayfield, an Oregon attorney, was arrested by the Federal Bureau of Investigation (FBI) as a material witness in an investigation of terrorist attacks on commuter trains in Madrid, Spain. In March 2004, the FBI fingerprint department had conducted a computer database search of an impression found on a bag of detonators and identified the impression to Brandon Mayfield. Two weeks after Mayfield's arrest, the Spanish National Police (SNP) informed the FBI that they had in fact identified the print to an Algerian national called Daoud.

10.3.1.2 The FBI compared Daoud's prints with the impression on the bag of detonators and agreed the findings of the SNP. They subsequently withdrew their previous identification of Brandon Mayfield.

10.3.1.3 The U.S. Department of Justice, Office of the Inspector General (OIG) launched a review into the FBI's handling of the case and provided an assessment of the causes of the misidentification. FBI examiners initially found 10 features they believed to be in agreement with Mayfield's prints. The OIG report [E] concludes; "…the unusual similarity in position and ridge counts was a critical factor that misled four examiners and contributed to their overlooking other important differences between LFP 17 and Mayfield's fingerprint" (Executive Summary). This conclusion implies that due to the unusual level of similarity, examiners were less focused on information which would negate the hypothesis of identification. The report further states; "There were also other subtle but important differences between the prints in the positioning of the features. But the unusual similarity in position and ridge counts was a critical factor that…..contributed to their overlooking other important differences" (Executive

Summary). It would appear that the examiners applied a lower level of scrutiny to the information which supported their favoured hypothesis of identification.

10.3.1.4 The OIG found that the examiner's interpretation was also influenced by circular reasoning, working backward from the known source material; "Having found as many as 10 points of unusual similarity, the FBI examiners began to 'find' additional features that were not really there, but rather were suggested to the examiners in the Mayfield prints" (Executive Summary). Again the examiners would seem to be unconsciously seeking out information to confirm their favoured hypothesis of identification and this is a consistent theme throughout the assessment of the causes of the errors, particularly with regard to the explanation offered by the examiners for observed differences between the prints. "This explanation required the examiners to accept an extraordinary set of coincidences. The OIG found that the support for this explanation was, at best, contradictory" (Executive Summary).

10.3.2 Shirley McKie Case 1999

10.3.2.1 During the 1997 trial of Mr. David Asbury for the murder of Miss Marion Ross, Ms. McKie, one of the investigating officers, did not accept that an impression from the crime scene, identified to her by experts from the then Scottish Criminal Records Office (SCRO) could have been made by her.

10.3.2.2 Ms. McKie was subsequently charged with perjury in 1999 and at her trial the SCRO identification was challenged and refuted by American Fingerprint Experts, Mr. Pat Wertheim and Mr. David Grieve. These experts also challenged the identification of an impression which had been presented as part of the prosecution case against Mr. Asbury.

10.3.2.3 The jury unanimously found Ms. McKie not guilty; however the fingerprint evidence remained a matter of dispute and controversy across the national and international fingerprint community for the next decade and was subject to a Scottish Government Justice Committee Inquiry in 2006. In March 2008 Sir Anthony Campbell was appointed to hold a public inquiry into the identification and verification of the fingerprints associated with HM Advocate v McKie 1999. The Fingerprint Inquiry Report was published in December 2011 stating that two misidentifications had occurred and also presented an in-depth scrutiny of fingerprint examination methodology and associated issues.

10.3.2.4 On discussing the causes of the errors Sir Anthony Campbell stated; "The method of work described by the four SCRO officers displays a number of recognised risks factors and in the case of Y7 and QI2 Ross it is likely that these risks crystallised into the misidentification"[59].

10.3.2.5 Amongst risk factors identified in the SCRO methodology listed below are those which are relevant to the cognitive bias issues under discussion in this paper:

[59]Campbell, A. (2011). The fingerprint inquiry report. Available at: http://www.thefingerprintinquiryscotland.org.uk/inquiry/3127-2.html

10.3.2.6 Practitioners being taught 100% certainty which could be attained prematurely in the examination process on the basis of relatively few characteristics.

10.3.3 Establishes an inner conviction which can lead to a circular argument discounting differences which must be capable of explanation even if the examiner is not sure what that explanation is.

10.3.4 Diminishes the independence of the verification process because a verifying examiner might tend towards confirming the view of the first examiner particularly if the examiner is senior in experience or rank.

10.3.5 Diminishes the usefulness of asking an examiner to reconsider their findings – if they have already reached a conclusion with 100% certainty then unsurprising that a re-examination would typically lead to a confirmation of the initial findings

10.3.6 The ethos in the SCRO fingerprint bureau where pride was taken in an ability, particularly on the part of more experienced officers, to identify marks that other bureaus might not consider sufficient for identification[60].

10.3.7 **An inappropriate hierarchical philosophy**

10.3.8 Examiners could be influenced to make identifications or confirm identifications of senior officers, where the quality and volume of information did not properly support identification.

10.3.9 The application of inappropriate tolerances in the observation and interpretation of detail in marks and prints, reverse reasoning and the influence of repeated viewing of known prints.

10.3.10 Contextual information from the police, which may subconsciously influence the conclusions of fingerprint examiners.

10.4 Examples of mitigation strategies.

10.4.1 IPOL Unit, Netherlands Police Service, Zotermeer

10.4.1 The IPOL unit has introduced a structure and workflow process specifically designed to mitigate the risks associated with cognitive bias.

10.4.1.2 The fingerprint unit is established around regional centres and a central hub. Latent images are input by staff at the regional centres, sent for search on the automated fingerprint recognition system and then processed by examiners at the central hub. These examiners receive only the on-screen image, with all lifts and case information retained at the regional centres.

10.4.1.3 This structure effectively removes any risk of contextual influence affecting the examiner's technical decision making.

[60] This topic is discussed in some detail in: Charlton, D., Fraser-Mackenzie, P.A.F. & Dror I.E. (2010). Emotional experiences and motivating factors associated with fingerprint analysis. Journal of Forensic Sciences, 55, p385-393

10.4.1.4 Prior to processing the search, the examiner must conduct an onscreen analysis without reference to any comparison print. They are required to demonstrate a minimum of 12 unique features in the print before proceeding with the features graded for suitability for use in the initial findings. Any further features identified at comparison phase are highlighted as such and appropriate tolerances applied. This type of workflow mitigates the risks of cognitive influence associated with the application of inappropriate tolerances in the observation and interpretation of detail in impressions.

10.4.2 **Federal Bureau of Investigation (FBI) Latent Print Unit**

10.4.2.1 Following the procedure review instigated as a result of the Brandon Mayfield Case, the FBI introduced a system of blind verification. They have defined blind verification as "the independent application of Analysis, Comparison, and Evaluation (ACE) to a friction ridge print by another qualified examiner who does not know the conclusions of the primary examiner"[61]. The FBI further state that blind verification should; "eliminate confirmation bias and limit contextual bias in the examination process".

10.4.2.2 Blind verifications take place in cases with a single mark conclusion, circumstances where there are conflicts between examiners and also on decisions of "value" or "no value". The FBI are clear that blind verifications cannot be performed by any examiner who has previously been consulted by the primary examiner, who has knowledge of the previous examiner's conclusions, any knowledge of the information used by the primary examiner or and specific background case details.

10.4.2.3 The FBI accepts that some consultation is necessary for the sharing of expertise and that not every consultation between examiners is indicative of a complex analysis. However an analysis is considered complex when dissimilarities or factors influencing the quality of the print could interfere with the proper interpretation of the impression. When a complex analysis or conclusion results in an identification, examiners are required to document any explanation for differences caused by apparent distortion and identify the supporting data for their explanation in the case record.

10.4.3 **Scottish Police Authority Forensic Services (SPA FS), Fingerprint Units**

10.4.3.1 In anticipation of the publication of The Fingerprint Inquiry Report 2011 SPA FS established a series of work streams to consider good practice in relation to the cognitive influence issues raised as a result of the McKie case.

10.4.3.2 It was accepted that a certain amount of case context is required to allow the initial examiner to develop an effective case assessment strategy, however SPA FS recognised that it was not essential for subsequent examiners to have access to this information on every occasion.

[61] Dror, I.E., & Cole, S.A., (2010). The vision in "blind" justice: Expert perception, judgment, and visual cognition in forensic pattern recognition. *Psychonomic Bulletin & Review* **17(2)**, 161-167

10.4.3.3 A proportionate risk management approach was adopted to mitigate risks of cognitive influence without impacting on service delivery. A range of measures was developed;

a. Improved note taking, including demonstration of features used in lead identifications.
b. A complex marks process to manage variance in opinion between examiners. This process includes a blind technical review process, where examiners are required to prepare technical reports and supporting visuals following a completely independent review of the relevant impressions. Those involved in the technical review process have no prior knowledge or access to case-related information or the technical findings of any other examiners.
c. A blind verification process for lead identifications in which verifying examiners have no knowledge of the technical findings of any previous examiners.
d. The removal of any case context information or related communication documentation from the verification process in any circumstance.
e. Regular dip-sampling of all completed case work.
f. Training programmes for examiners exploring cognitive bias and its impact on the human decision making process.

10.4.4 **Surrey and Sussex Forensic Identification Services Unit (FISU)**

10.4.4.1 Surrey and Sussex Forensic Identification Services Unit have followed similar processes to SPA, and have also introduced cognitive profiling recruitment tests which have proven very effective at predicting cognitive skills of new staff, thus improving effectiveness and efficiency in managing cognitive influence.

10.4.4.2 Other parameters under consideration by FISU are longitudinal studies to underpin cognitive issues with overall accuracy and performance, and embedding cognitive processes to mitigate risks in using new technologies (remote transmission and on screen annotation tools).

10.5 Recommended good practice

10.5 The Codes (section 20.4) states that once a method has been designed or determined, there should be an assessment to identify any risks including; "identifying areas where the operation of the method, or interpretation of the results, requires specialist skills or knowledge to prevent ambiguous or misleading outputs or outcomes". An organisation should therefore adopt a risk management approach to the fingerprint methodology as applied within their organisation to identify, assess and evaluate the threats and consequences posed by the issue of cognitive bias. Practical solutions could include the introduction of a blind element to the verification process or randomising the respondent lists delivered through AFIS searches[62].

[62] Dror, I.E. (2013) Practical solutions to cognitive and human factor challenges in forensic science. Forensic Science Policy & management 4 p1-9.

10.5.2 Further generic guidance from The Institute of Risk Management states that; "Risk Identification should be approached in a methodical way to ensure that all activities within the organisation (or method) have been identified and all the risks flowing from these activities defined"[63]. Once identified, the risks should be displayed in a structured format, which can then be used to evaluate the consequences of the risk including the probability of occurrence. Risk assessment in this manner allows the organisation to break down each stage of the process and consider how best the impact can be mitigated. Areas to be considered can include:

a. Name of Risk
b. Scope of Risk
c. Nature of Risk
d. Stakeholders
e. Quantification of Risk
f. Risk Tolerance
g. Risk Treatment & Control Mechanisms
h. Potential Action for Improvement.

10.5.3 Suitable Risk Treatment and Control Mechanisms for consideration with regard to fingerprint examination are listed below:

a. Survey and breakdown extent of current contextual information available to examiners & assess added value each piece of information brings to the examination process.
b. Remove or limit contextual information which adds no tangible value to the fingerprint examination process.
c. Remove or limit contextual information made available to verifying or subsequent examiners.
d. Introduce a blind verification process for identified case work assessed as at greatest risk from contextual, confirmation and/or cultural bias.
e. Introduce a blind element to a technical review process for analyses, comparisons and/or evaluations which are considered complex or cause a variance in opinion between examiners.
f. As part of a technical review process for complex marks or circumstances where examiners have a variance in opinion, introduce an appropriate and proportionate note-taking strategy which requires examiners to provide written and visual accounts of their reasoning and findings.
g. Develop bespoke training programmes to raise awareness of the cognitive issues involved in human perception, judgement and decision making.
h. As part of an established quality management system, instigate an effective review and monitoring process to provide assurance that the risk treatment and control measures continue to provide effective risk management.

[63] Institute of Risk Management (2002) "A Risk Management Standard" IRM

11. FOOTWEAR, TOOL MARK AND FIREARMS COMPARISON AND FIREARMS CLASSIFICATION GUIDANCE

11.1 The generic marks comparison process

11.1.1 Introduction

11.1.1.1 The generic forensic process that is outlined below encompasses the interpretation and reporting of 'marks' comparison cases. It is applicable to a wide range of evidence types such as firearms, footwear, and tool marks and outlines a practical strategy that can be used to counter potential cognitive bias when carrying out 'marks' comparison cases:

11.1.1.2 With regards to tool mark comparison this section should be read in conjunction with Regulator Codes of Practice and Conduct – Draft Appendices Toolmarks – HOS/12/027

11.1.1.3 With regards to footwear marks related comparisons this section should be read in conjunction with Regulator Codes of Practice and Conduct – Draft Appendices Footwear – (HOS/11/059)

11.1.1.4 With regards to firearms related comparisons this section should be read in conjunction with the Regulator Codes of Practice and Conduct – Draft Appendices Firearms – HOS/12/026, Microscopy and Firing Marks.

11.1.1.5 The strategy also addresses the possible low expectation of a 'hit" when screening through a firearms Open Case File (OCF)[64]

11.1.1.6 Confirmation bias in firearms classification examinations is also addressed. In this context this section should be read in conjunction with Forensic Science Regulator Codes of Practice and Conduct – Draft Appendices Firearms – HOS/12/026, Classification of Firearms and Ammunition.

11.1.2 Process outline

11.1.2.1 Items are recovered from the crime scene and may consist of the original item or a 'true' copy of the mark generated by other methods.

11.1.2.2 Items are received along with case information and questions to be addressed by the scientific work.

11.1.2.3 The case information, supplied by the customer, is used to direct the item examination recovery and analysis strategy, ideally within a framework of appropriate propositions.

a. Examination of the item/mark recovered from the crime scene.
b. Use of recovery and enhancement techniques as required.
c. Generation/Examination of the 'control' item
d. Make test marks if required in the appropriate manner.
e. Undertake a comparison using appropriate methods and equipment

[64] An OCF is defined as an organised collection of ammunition components derived from crime scenes that is intended to be compared against test fired and crime scene ammunition samples in order to establish whether or not a single gun has been used at one or more scenes.

f. Interpret and evaluate findings
g. Verification of result
h. Findings are described in a statement or report.
i. The scientist may be called to court to give oral testimony.

11.2 Risks of cognitive bias

11.2.1 A marks comparison seeks to establish if a 'mark' (the unknown) has been made by the submitted exhibit (the known) or has been made by the same item e.g. a revolver which has not been recovered could be responsible for discharging multiple bullets recovered from multiple scenes. It is based on the comparison of detail and is therefore observational. The scientist is looking to determine if the detail present in the mark matches characteristic detail on the item or in a test mark or is significantly different. An assessment of what the detail is and how it has been produced must consider general characteristics common to a set of items (CLASS), unintentional manufacturing marks present on a sub-set of items (SUB-CLASS) through to random damage/wear and tool mark characteristics (INDIVIDUAL). Any examination is therefore dependent upon the visual quality and clarity of the detail that is observed by the examiner. The process is one of pattern recognition aided by the use of equipment such as photographic/imaging, low power microscopy and comparison microscopes. The final assessor of the level of significance of any agreement between the marks is the human operator; there is no significant instrumental analysis [W]. In footwear mark comparisons, the methods employed by footwear practitioners are normally side-by-side comparisons or overlay. In this way the footwear expert assesses the level of agreement in terms of the pattern, pattern configuration, mould/moulding detail, wear and damage. The assessment is subjective, although reference material and data can be used to support the evaluation of the findings. In tool mark/firearms comparisons there are currently two methods; traditional pattern recognition where the examiner's opinion is based on the relative extent of detailed agreement with a best known-non-match and Consecutive Matching Straie (CMS) where the examiner applies a conservative criteria of runs of aligned straie to establish a possible match. Both techniques use subjectivity.

11.2.2 The interpretation and evaluation of a 'marks comparison' may potentially be affected by some form of unintended bias. In the interpretation process there are no results produced by a 'black box'; opinions and decisions are based on the individual's, relevant experience, depth of knowledge and skill as well as their disposition at the time. Every effort must be made to make it logical, transparent, balanced and robust. Usually the opinions are formed in the context of supplied case information, introducing the possibility of contextual bias.

11.2.3 Within marks interpretation it is considered that there is a spectrum of bias risk (table 2).

Risk factor	Low risk	High risk
Detail	The detail in the mark(s) is clear, well defined and unambiguous	Marks are confused and complex, of poor quality and the detail present is poorly defined.
Equipment	Optimum visualisation of the detail in a mark using appropriate equipment/imaging and enhancement techniques.	Poor or inappropriate equipment/imaging and enhancement techniques.
Approach/Examiner	There is a methodical approach with defined standards built on principles that have been tested and validated. Possible confirmation bias may reduce as a consequence of the comparison reviewer having less contextual information[65]	When the approach is un-researched, ad hoc and personal to the operator. When the expectation of an OCF hit is very low.
Scientist/Examiner	Scientist/examiners are well trained, experienced and continuously meet acceptable standards of competence	Scientist/examiners are inexperienced, unmonitored and left to adopt their own approach.

Table 2: Spectrum of bias risk in marks interpretation

a. Risks are low when results are clear and unambiguous and greater when results are complex, of poor quality and there is an increased reliance on subjective opinion.
b. Risks are lower when there is a methodical approach with defined standards built on principles that have been tested and validated and greater when the approach is un-researched, ad hoc and personal to the operator.
c. Risks are lower when equipment is well maintained and functioning to the required standard.
d. Risks are lower when operators are well-trained, experienced and continuously meet acceptable standards of competence and results are peer reviewed, and greater when operators are inexperienced, unmonitored and left to adopt their own approach.
e. Contextual and confirmation bias risk is lower when the contextual information is minimised, particularly at the comparison review stage and the reviewer is unaware of the examiner's opinion, or other evidence that relates to the 'marks' examination.

[65] Kerstholt, J., Eikelboom, A., Dijkman, T., Stoel, R., Hermsen, R., van Leuven, B., Does suggestive information cause a confirmation bias in bullet comparisons? (2010) *Forensic Science International* **198** 138–142

f. Expectation bias manifesting in the missing of an OCF hit is lower when there is an expectation of success[66].

11.2.4 Other more general bias risks within "Marks" and firearms examination and classifications:

a. Observations that support the defence case are less rigorously considered or evaluated and are not given their true weight.
b. Interpreting the Firearms Act 1968 when classifying potential component parts or antiques. Confirmation bias on the status of firearms should be avoided; this is particularly pertinent where the prosecution expert relies upon Home Office Guidance, which is not explicitly reflected in the legislation.
c. Reluctance to express doubt particularly during oral evidence at court.
d. Reluctance to clearly understand and express the limitations of a comparison after a time delay between the offence and the recovery of a suspect item.
    i. The comparison of footwear a footwear mark recovered at a crime scene to footwear recovered months later.
    ii. The assessment of the significance when there is matching and non-matching characteristic detail in the mark.
e. Failure to express alternative explanations, such as possible sub-class origins and arguments for alternative firearms legal classifications.
f. A failure to assess detail correctly due to a lack of knowledge and the inability to investigate due to location of manufacturing plant or time and cost considerations.

11.3 Examples where risks of bias have become an issue

a. The identification of a tool being responsible for cutting a wire fence, where detail was clearly visible that excluded the suspect tool.
b. Situation where critical findings checks were being undertaken on a basis of 'I will check yours if you check mine'. An independent approach was not maintained.
c. The association of two crime scenes in the same geographic area, involving crimes of similar *modus operandi*, calibre, make and model of gun. Possibly due to confirmation and contextual bias compounded by lack of awareness of differences between sub-class and individual characteristics.
d. The automatic classification of vintage firearms as not being subject to the section 58(2) exemption provided for antique firearms, due to the prosecution expert relying on "official" guidance as opposed to statute, possibly as a result of confirmation bias.

[66] Nennstiel R., (2010). The Human Factor in Detecting Cold Hits, Association of Firearms and Toolmarks Examiners Annual Training Seminar. Henderson, Nevada, USA, 2nd – 7th May 2010.

e. Classification of possible component parts of a firearm as being subject to the 1968 Act without consideration of any alternative hypothesis most probably due to confirmation bias.

11.4 Mitigation strategies currently deployed in the UK and overseas

11.4.1 Examples of mitigation strategies that are variously in current practice are listed below. These are considered to be good practice in appropriate circumstances:

a. Case Assessment and Interpretation. Comparison of expected, pre-assessed outcomes with actual results under appropriate hypotheses.
b. Full disclosure of all data used in the evaluation.
c. In all firearms classification cases, the reviewer should clearly set out what is official guidance and what is statute, ensuring that alternative classification hypotheses are addressed to counter any confirmation bias.
d. Use a completely "blind" checker who repeats the full interpretation, but in the absence of any contextual information relating to the case. Initially, the checker should not be aware of the opinion of the reporting scientist.
e. An acceptable alternative is that result will be subject to a critical findings check by a second authorised examiner. The initial practitioner completes the comparison and records what items they have examined, their findings together with their conclusion. The checker then undertakes a detailed independent review wherever possible without knowledge of the previous practitioner's conclusion. The aim of the check is as follows:
    i. The examiner has followed the appropriate documented examination process and applied the appropriate relevant scientific methodology and techniques.
    ii. The work and findings of the examination are reflected in the conclusion of the report. The results must support the conclusion and clearly there should be an understanding or statement of the findings.
    iii. The maximum evidence has been obtained, that nothing has been overlooked and there are no other marks that may change the outcome.
    iv. The submitting authority's question has been fully addressed.

11.4.2 In addition to the good practice described above the following are also recommended:

a. Validation testing of qualitative and subjective based approaches to demonstrate the robustness of conclusions and opinions.
b. Development of standards and quality managed procedures for qualitative and subjective based methods, including system performance data indicating when the approach breaks down and is no longer valid.
c. Practitioner training in the specific method used, together with initial and on-going competency assessment.
d. Training and education in relation to the risks of cognitive bias in firearms classification and marks comparison generally.
e. An approach to quality that includes the assessment and monitor of on-going competence of practitioners including the use of proficiency tests, declared and undeclared trials.

f. Providers should ensure that a validated form of Context Management is applied.
g. The use of blind trials should be introduced to increase the "success" rate of cold OCF hits.

12. TRACE EVIDENCE (INCLUDING HAIR AND FIBRE) GUIDANCE

12.1 Outline of the Forensic Process for Trace Evidence analysis

12.1.1 The examination of trace evidence covers a wide range of materials including particulate material such as glass, paint, hairs and fibres. However whilst the range of trace materials is wide, the analysis of such material essentially follows the same process which involves comparison of crime (unknown/recovered) material with one or more known/reference samples. This process can briefly be described as follows:

12.1.2 Item receipt: items are received along with case information and questions to be addressed by the scientific work. When dealing with contact traces, taking and submitting the right reference samples (from the crime scene or individuals) is critical as it can have a fundamental impact on the subsequent comparison.

12.1.3 Case assessment: case information is used to direct the strategy for item examination and trace evidence recovery and analysis. Ideally case assessment should be carried out with in a framework of appropriate propositions. By its nature trace evidence examination is time consuming, so practicality and cost have to be considered. Case assessment can assist with targeting the exhibits most likely to yield probative evidence.

12.1.4 **Recovery of trace materials using appropriate techniques**

12.1.5 Identification of target material and comparison with reference sample(s):

a. Whichever recovery technique is used, the examiner is often presented with a large amount of debris which may potentially contain some of the target material. Where there is a limited amount of target material of interest which can be immediately identified, e.g. glass fragments, paint fragments, this material can be recovered in its entirety or a sample taken. The material can then be compared with the relevant reference sample(s) using the appropriate microscopy and instrumental/analytical techniques.
b. With other evidence types, for example fibres and hairs, there will often be a large amount of material collected which is of no relevance to the case. For this reason it is necessary to review the reference sample(s) and use features to enable an initial search of the recovered material to locate that which is of potential interest. For example, for hairs and fibres a search of tapings under a low power microscope would be conducted to locate hairs/fibres with similar macroscopic features (colour, length etc.) to the recovered hairs/fibres. This material can then be recovered for more detailed comparison with the reference samples using the appropriate microscopy and instrumental/analytical techniques.

c. Evaluation of the scientific findings and interpretation within the context of the case specific information available (may be at source or activity level as appropriate).
d. Provision of report or statement describing the findings and providing opinion on their significance.
e. Oral testimony - the scientist may be called to court to give evidence.

12.2 The Risk of Cognitive Bias in Trace Evidence analysis

12.2.1 As in other areas of forensic science, trace evidence analysis can potentially be affected by some form of subconscious and unintended bias and will be a particular risk where subjective interpretations are required. Trace evidence examinations can broadly be divided into two groups:

12.2.2 Those that are entirely subjective and based on mainly observational skills, for example, the microscopic comparison of hairs or the comparison of the layers of paints in a microscopic fragment, which relies exclusively on a subjective assessment of whether the crime and reference samples match.

12.2.3 Those that may include an initial subjective element, followed by the use of objective instrumental techniques to confirm or eliminate matches. For example, analysis of paint after a visual comparison and fibre comparisons where the subjective microscopic examinations can usually be followed by the use of a range of instrumental/analytical techniques including Microspectrophotometry, Fourier Transform Infrared, Raman spectroscopy and Thin Layer Chromatography. Hair comparisons have no similar follow up tests (unless dyed), other than DNA analysis (nuclear or mitochondrial DNA) which, because of the cost and the destructive nature of the testing, is often not an option.

12.2.4 Additionally, opinions are formed in the context of the information supplied about the case and the samples submitted e.g., where and how the glass was broken, how close the person was to the breaking glass, how long after the incident/alleged contact clothing was recovered etc. This may introduce contextual bias[67]. Regardless of contextual case information, practitioners may have a higher expectation of observing matching hairs, fibres, glass etc., simply because the samples have been submitted by the police investigators.

12.2.5 Due to the nature of trace evidence, the recovery and comparison is time consuming and requires a high level of skill, knowledge and often patience. In all cases involving contact traces, there is a requirement for relevant case information to be available to the practitioner to allow effective case assessment. Where fibre evidence is being considered, without information it would be impossible in all but the simplest cases to effectively target those fibre transfers which are viable and would be most probative, thus keeping the time expenditure at a level commensurate with the requirements of the case. This will also apply to hair examinations, where the population of hairs potentially of interest is large.

[67] Miller, L. (1987) Procedural Bias in Forensic Science Examinations of Human Hair, Law and Human Behaviour 11(2) p157-163

12.2.6 Within trace evidence examinations, there is a spectrum of bias risk:

Risk Source	Low risk	High risk
Case Assessment	Full case assessment considering potential outcomes, preferably considering at least two competing hypotheses	No case assessment; only one hypothesis considered.
Examination process	Empirical analysis using instrumental techniques	Subjective microscopic analysis only
Result Quality	Results are clear and unambiguous	Results show wide intra-sample variation, are of poor quality and there is an increased reliance on subjective opinion.
Interpretation Approach	There is a methodical approach with defined standards built on principles that have been tested and validated	The approach is un-researched, ad hoc and personal to the operator.
Operator Competence	Operators are well trained, experienced and continuously meet acceptable standards of competence	Operators are inexperienced, unmonitored and left to adopt their own approach.
Checking	Independent confirmation of critical observations. Full independent reinterpretation	No checking or checking is conducted collaboratively

Table 3: Spectrum of bias risk within trace evidence examinations

a. Risks are high where no case assessment is carried out with respect to the potential outcomes of the examinations and the expectations of the examiner, preferably considering at least two competing hypotheses. Risks are reduced significantly where a documented assessment is carried out, the potential outcomes of the examinations are considered in the light of the relevant contextual information available, and the expectations of the examiner are recorded.
b. Risks are low when empirical analysis forms part of the examination processes, and greater where there is an increased reliance on subjective observational analysis.
c. Risks are low where results are clear and unambiguous (for example with a strongly coloured manmade fibre sample which shows little intra-sample variation) and is higher where there is wide intra-sample variation

(for example with a shoddy mix of fibres where it may not be possible to use instrumental techniques to confirm microscopic matches).

d. Risks are low if there are sufficient reference samples showing all possible variations for example within a painted surface, hair from different parts of the head, all broken windows have been sampled etc. Risks are higher if only a limited reference sample is available and may result in the practitioner making a subjective assessment of the match.
e. Risks are lower when there is a methodical approach with defined standards built on principles that have been tested and validated and greater when the approach is un-researched, ad hoc and personal to the operator.
f. Risks are lower when operators/checkers are well trained, experienced and continuously meet acceptable standards of competence; they are greater when operators/checkers are inexperienced, unmonitored and left to adopt their own approach.
g. Risks are lower when critical observations, such as paint layer colours and sequence, are checked independently by another competent practitioner and higher where no critical observation checks are carried out.
h. Risks are lower when interpretation is checked by a competent peer who conducts a separate interpretation, fully independent and without influence from the reporting scientist. Risks are higher when checking is less rigorous and/or conducted collaboratively.

12.2.7 For some trace evidence there are data to support the practitioner. Studies of glass have been undertaken over many years and provide a great deal of data regarding background population, persistence on clothing, breaking windows and the transfer of glass fragments; refractive index information and analytical data for different types of glass are also available. For fibres, there is considerable empirical data to support interpretations, such as population studies and target fibre studies but there is currently no fibre database which provides any guidance with respect to how common a particular fibre might be in the general fibre population. Previous databases (Forensic Science Service) went some way to providing this, but constantly changing fashions and fibre technology changes mean that any database is almost impossible to keep up to date. Therefore, any assessment regarding how common (or otherwise) a fibre might be is essentially subjective and based on the scientist's experience, unless specific industrial enquiries can be made for a particular case.

12.2.8 Fibre, hair and trace evidence analysis generally are becoming less used, and therefore the risk that the examinations are not carried out by practitioners who are dealing with the evidence on a routine basis is increasing. The lack of work in this field has serious implications for the maintenance of scientists' experience and competence and a reduction in the number of practising scientists may ultimately result in there being no one suitable to undertake peer-review.

12.2.9 It is not operationally practical to carry out a full independent check of microscopic fibre matches where large numbers of fibres have been recovered from tapings and individually examined; but where a range of instrumental and analytical techniques are employed which back-up the subjective microscopic

matches this is not necessary. However, where subjective observational methods are the only option, for example in hair comparisons, a full independent check is vital.

12.2.10 With budgetary constraints a certain amount of 'pre-assessment' is often carried out by police forces before selected items are submitted to a forensic provider for examination. There is a bias risk inherent in this process, particularly where the practitioner is not fully informed. For example, other items seized but not submitted for examination may be potentially be an alternative, legitimate source of matching fibres.

12.3 Case Examples where Cognitive Bias May Contribute to Error

12.3.1 The analytical processes for trace evidence have largely remained the same for several decades. As a result methods have been validated and well-tested in forensic casework. The authors are unaware of any specific examples where the results of the microscopic comparison of trace evidence, or subsequent analytical testing of the material has been an issue in case work in the UK. The area of high risk with respect to bias in trace evidence analysis is that of the case evaluation and interpretation where contextual bias might be introduced. Whilst no specific casework examples can be provided where cognitive bias may have contributed to interpretational error, the following hypothetical examples involving glass and fibre examinations are offered where bias might be observed:

12.3.2 Absence of matching glass fragments concluded as being inconclusive

12.3.2.1 Clothing is submitted from a suspect who is believed to have been seen breaking a glass window and who was arrested shortly after the incident. The practitioner would have a high expectation of finding glass fragments on the persons clothing (choice of clothing to examine would depend on the height of the window). If the relevant clothing was examined and no glass is found then what should the practitioner conclude? As a simple observation then it could be said that no glass was recovered, however this provides no evaluation of the significance of the evidence. Often it is concluded that the findings are inconclusive as it is not possible to comment as no glass was found. If the practitioner evaluates the evidence using a structure of alternative propositions, one reflecting the prosecution view and one the defence view (or a hypothetical defence view if appropriate) the lack of any glass fragments may well support the view that the suspect was not involved in breaking the window as alleged. Therefore reporting the findings as inconclusive might be considered biased.

12.3.3 Absence of matching fibres concluded as being neutral

12.3.3.1 The examination of car seat tapings for a transfer of fibres from the clothing of an individual who is alleged to have stolen and driven the car for some hours results in no matching fibres being found. The defendant has made no comment. In this situation, it is tempting to conclude that the absence of matching fibres is neutral and does not assist in addressing whether or not the individual had been in the car. However, if the information available provides no explanation for the absence of matching fibres (for e.g., the defendant might have had had time to change clothing before arrest) and the scientist had a high expectation of finding matching fibres if the contact had occurred as alleged, the

absence of matching fibres may well support the view that the defendant had not been in the car. Even where a 'no comment' interview has been offered by the defendant, a good case assessment at the outset requiring consideration of the full range of outcomes and potential defence scenarios, including the absence of any matching fibres, would be likely to result in this type of bias being eliminated.

12.3.4 **Difference in treatment of crime and reference material post transfer**

12.3.4.1 A fibre examiner faces considerable difficulty in dealing with cases where clothing has been altered at a chemical level in the period between the offence and seizure of the clothing, for example where the body of a victim has been submerged in a river or at sea for some time, causing the dye in the clothing to fade. In this situation, the challenge for a fibre examiner is firstly searching for fibres without a reference sample that is representative of the fabric at the type of the offence, and then having to interpret a population of fibres on a suspect's garment which does not match the control, but perhaps did at the time of the offence.

12.3.4.2 A European Textile and Hair Group (ETHG) collaborative exercise in 2004 involved a hypothetical scenario involving blue pigmented viscose fibres found on the victim's clothing, which appeared the same as those from the putative source when compared under transmitted light, but differed markedly under UV light. Clearly these fibres did not match. Subsequent experimentation to test a theory that when the T-shirt had become wet, the fibres had 'taken up' washing detergent residues on T-shirt which contain optical brighteners causing them to fluoresce, demonstrated that this was possible. But the issue that the experiment does not address is how we tell whether the fibres on the T-shirt fluoresced the same as those from the mattress prior to the absorption of detergent. It is entirely possible that the fluorescent behaviour observed under the microscope is exactly what the fibres were like at the point of transfer. Whilst it is fair to explore the possibility that fibres have been changed at a chemical level and pursuing experiments to assess that, it would be biased for a laboratory to state that on the basis of such experiments more support is provided for the view that the fibres recovered from the T-shirt came from the mattress rather than from another source.

12.4 Mitigation strategies deployed both within the UK and overseas

12.4.1 The following are examples of mitigation strategies that are variously used in current practice. All are examples of good practice in appropriate circumstances and should be applied as described.

12.4.2 Independent checking – where only subjective observational assessments of a match are possible (for example hair comparisons, paint layer colours and sequences), full independent checking should be carried out and clearly documented. The check should be carried out independently of the original examiner.

12.4.3 Independent checking of analytical results – where instrumental techniques are used, either alone or to back up subjective microscopic matches, and the results are subject to interpretation by the operator (e.g., Microspectrophotometry result for analysis of colour of fibres, refractive index

measurements for glass, chemical analysis of glass fragments and paint layers), the interpretation of the results should, where possible, be carried out by two competent and experienced scientists, (operator plus one other) independently of each other.

12.4.4 Use of statistical approach to evaluation – to assess whether the refractive index of suspect glass fragments match that of reference glass sample(s) a statistical approach can be applied rather than relying on the experience of the practitioner.

12.4.5 Case Assessment and Interpretation – a robust and documented comparison of expected, pre-assessed outcomes with actual results under appropriate competing hypotheses. Some documented indication of expected outcome is recommended in all cases. Where results are at the least likely end of the expected outcomes, for example the absence of matching fibres where the most likely outcome was to find lots of matches, an independent review of the tapings would be advisable.

12.4.6 Training – appropriate training of practitioners in the methods employed who can demonstrate initial and ongoing competence.

12.4.7 Quality assurance trials – participation in internal and external quality assurance trials. Members of the ENFSI European Textile and Hair Group (ETHG) participate in an annual collaborative exercise which seeks to test various parts of the process of fibre examination. Membership of the ETHG is limited, and participation is only available to members. Forensic Science Providers (FSP) in the UK also participate in CTS (Collaborative Testing Services Inc.) trials which are available by subscription and cover fibre, paint and glass analysis. These trials are considered to be fairly basic and test the microscopic and analytical procedures employed, but do not assess the approach to evaluating the significance of the findings. At least one of the UK FSPs carrying out fibre work also carries out internal quality assurance testing with each of their scientists undertaking a mock case every 2 years to test their competency. Only some of these trials will be relevant with respect to assurance that bias is being avoided, however all provide some level of assurance of the ongoing competence of the scientists involved. There is a gap in the current system with respect to ‘blind’ trials – small organisations do not have the resources to conduct such testing.

12.4.8 **Further recommendations for good practice**

12.4.9 In addition to the good practice described in 11.4, also following may be considered:

a. Use of a completely independent (“blind”) checker who repeats the examination/interpretations described in 11.4.1 and .2 but in the absence of any contextual information relating to the case. This may present practical challenges, particularly within smaller organisations. However, it will assist in a continuous learning and improvement cycle, where reporting scientists can identify instances where they may have been affected by bias. Further, it provides assurance for the courts that the interpretation is free from contextual bias.
b. Documented case assessment and interpretation in all cases involving trace evidence analysis, preferably carried out independently by a

second scientist, but at the very least to be peer reviewed. Elements of the interpretation should also be included in the scientist's statement to explain to the court how their conclusion has been reached.

c. With a reduction in the use of trace evidence analysis in casework in the UK, maintaining competency and having sufficient trained and competent staff to allow independent checks and peer reviews will be a challenge, particularly for smaller organisations. Clear documentation of case assessment, interpretation and a report/statement which clearly states the limits of the examinations used (i.e. where appropriate their subjective nature, limitations of small amounts of reference material (hairs) and whether findings and interpretation have been reviewed) should be a requirement. Such transparency and disclosure provides the opportunity for scrutiny and the identification of potential bias.
d. Where items submitted to a forensic provider for examination have been the subject of 'pre-assessment' by the submitting force, ideally a list of other items seized should be made available to the scientist on request to allow consideration of potential alternative sources of transferred material.
e. Training and education in relation to the risks of cognitive bias in trace evidence examination generally and specifically in relation to highly subjective examinations.
f. A program of 'blind' or undeclared quality assurance trials in the UK submitted to all FSPs could address the issue of bias thus providing assurance to the courts that procedures are robust and areas of potential bias are identified and managed.

13. VIDEO AND AUDIO

13.1 Introduction

13.1.1 A video or audio comparison often seeks to establish if the image or signal associated with a suspected crime (the "item") is of a specific article or person (the "target"). This may be for example a person's face captured on CCTV, an item of clothing being worn by the perpetrator, a vehicle or indeed any other object that may be relevant to the crime scene. This is undertaken by comparison against a reference image or signal from the target, ideally which has been generated under identical conditions to the original item. The comparison may be subjective and may utilise either purely visual side by side comparisons, or may include use of tools to aid comparison, such as overlaying of the images and switching between the two to highlight any potential differences. Alternatively comparison may be aided by objective measurements of the images (photogrammetry) for example in facial comparison in which spatial proportions of facial features are compared using measurements of distances and angles between facial landmarks in order to quantify any differences or similarities observed. Elimination should be the fundamental aim in any comparison and presence of a single difference for which there is no viable explanation should be sufficient for an exclusion. Conversely where a number of features are seen to be in common and no differences are observed, then this can provide corroboration to other evidence of inclusion.

13.1.2 Any examination is therefore dependent upon the visual quality and clarity of the detail that is observed by the examiner plus how inherently discriminable the object is from other objects of the same type. In combination these ultimately impact on the strength of the conclusions that may be drawn. For example with a good quality image of a motor vehicle it may be possible to identify the make and model with confidence by observing a combination of class characteristic features such as the shape of the windows, lights, bumpers, doors, overall shape etc. However, narrowing the identification to a single specific car would require much more detail in the images in order to observe individual characteristics or features that differentiate one individual car of the same make/model from another e.g. registration number, intentional alteration such as cosmetic modifications, wear and tear such as scratches or other damage features[68].

13.1.3 The basis for opinions and conclusions reached lies in the detection of correspondence or discordance of features determined to be reliable. These in turn rely on the individual's, relevant experience, depth of knowledge and skill as well as their disposition at the time. Every effort must be made to ensure that opinions and conclusions are logical, transparent, balanced and robust. In some cases a statistical model may be applied to provide a formal probabilistic basis for a conclusion. In other cases a statistical model may not be feasible but this does not necessarily preclude reaching a sound conclusion where for example a CAI approach is adopted.

13.2 Generic video and audio process outline

13.2.1 The generic forensic process that is outlined below encompasses the interpretation and reporting of video and audio comparison cases. It is applicable to a wide range of evidence types including photographic evidence with motion and still images, plus audio recordings associated with a suspected criminal act under investigation:

a. Recovery of video, photo or audio material related to the crime scene consisting
b. Items are received by the analyst along with relevant case information and questions to be addressed by the scientific work.
c. Generation of an exact copy of the original then use of techniques as required to clarify or clean up the copy of the image or audio signal
d. Examination of the copied material recovered from the crime scene and notation of features determined to be reliable
e. Examination of the 'control' item
f. Undertake a comparison using appropriate methods and equipment
g. Interpret and evaluate findings
h. Verification of result
i. Findings are described in a statement or report.
j. The scientist may be called to court to give oral testimony.

[68] Scientific Working Group Imaging Technology (SWGIT) (2013) Best practices for forensic photographic comparison V1.1 Section 16

13.3 Risks of cognitive bias

13.3.1 Within video and audio comparison, there is a spectrum of bias risk:

Risk factor	Low risk	High risk
Detail & Presentation	The images/signals are clear detailed and unambiguous with item and reference images generated under identical conditions	The images are of poor quality and the detail present is poorly defined, and the images being compared have been generated under very different conditions
Equipment	Optimum visualisation of the detail in an image using appropriate equipment/imaging and enhancement techniques.	Poor or inappropriate equipment/imaging and enhancement techniques.
Approach	There is a methodical approach with defined standards built on principles that have been tested and validated. Item is characterized prior to exposure to reference image	When the approach is un-researched, ad hoc and personal to the operator. Item is characterized after exposure to reference image
Scientist/Examiner	Scientist/examiners are well trained, experienced and continuously meet acceptable standards of competence	Scientist/examiners are inexperienced, unmonitored and left to adopt their own approach.
Verification of results	Independent review of critical findings	There is no independent review, or reviewer knows findings and conclusions drawn from original assessment

Table 4: Spectrum of bias risk in video and audio comparison

13.4 Mitigation strategies and good practice guidance

13.4.1 Avoiding psychological contamination in the processing of material

13.4.2 One of the greatest risks of introducing cognitive bias is in the way the material is provided for assessment. Examiners should only be provided with the information relevant to the examination of the item image, and in the first instance and they should only be asked to describe what they see. The latter guards against confirmation bias, which is almost inevitable if the question asked is along the lines of “do you agree that this is item/individual x?”, or the examiner asks to be told what the item is so that they can consider whether or not they agree. Not being provided with the case notes and other extraneous information prior to the examination and comparison task at hand helps safeguard against contextual bias. For the same reason it is better for the

analyst to receive written briefing regarding the comparison to be made rather than being in direct verbal contact with the investigator, so that opportunity for transfer of non-relevant and potentially biasing information (both contextual and confirmatory) can be avoided.

13.4.3 Wherever possible, the item should be assessed prior to observing the reference image or signal, again so that confirmation bias can be guarded against. If a series of images are submitted of what is believed to be the same item, these should be assessed in sequence starting with the worst image first, so that the potential for confirmation bias between these images is avoided. Where a discriminatory feature is identified in the item only after comparison with the reference, this should be fully explained in the examination records, so that transparency of the assessment is maintained at all times.

13.4.4 Independent assessment of critical findings is also crucial. Independent checking that minimizes the risk of cognitive bias entails assessment without knowing the outcome of the initial analysis, or even where possible the identity of the original examiner in order to avoid confirmation bias.

13.4.5 **Use of validated processes**

13.4.5.1 All forensic processes should be validated prior to use in casework. Section 20 of the FSR Codes provides guidance on validation with more detailed explanations given in validation appendix currently due for publication by the FSR in September 2014 plus guidance on how to approach validation of digital forensic techniques in an currently being drafted for consultation by the FSR. Scientific validation is the process by which a new method or technique is assessed to ensure that it is fit for purpose and that once implemented will continue to function as such. This principle applies whether a system provides objective highly automated analysis and comparison of materials, or at the other extreme where the process relies almost entirely on subjective comparison and assessment by an analyst.

13.4.5.2 Bias is less likely when images are clear and well defined, whilst the risk of bias increases as images become less defined and ambiguity regarding interpretation increases. Therefore use of appropriate and validated methods to clarify images/signals may help reduce risk of bias. However certain techniques for image manipulation are “lossy” and can result in the loss of potentially discriminable detail (increasing the risk of false inclusion) whilst other enhancement techniques can create artefacts, thereby increasing the risk of false exclusion. It is crucial therefore that any manipulation processes are validated. This should include full characterization of the processes applied including determination of the limits within which the application can be reliably used and demonstration through experimentation not to increase the risk of false inclusion or exclusion. Likewise during application to casework, and especially in the enhancement of audio signals the analyst should frequently check back during processing against the original to ensure that the signal has

not become over-processed[69]. Likewise, when using colour as a comparator, the limitations of the approach should be fully evaluated and understood: under certain lighting conditions (e.g. sodium lamp), 2 items that are different in colour under natural illumination may appear to be the same, whilst the same item under different lighting conditions may appear to be markedly different in colour.

13.4.5.3 Techniques deployed to aid in the side by side comparison of images must be validated to ensure they do not introduce bias. For example overlaying techniques for comparison can highlight differences between images by rapid flicking between images. However a gradual transition between two overlaid images may cognitively mask any differences from the observer. Wherever possible the same context should be used to generate reference images for comparison against the original crime scene image by for example re-constructing the scene and capturing the reference image using the same equipment, lighting conditions, camera angles, environmental conditions etc. Where this is not possible, the resultant limitations in making a comparison should be declared in any statement.

13.4.6 **Proficiency testing/ QC measures**

13.4.6.1 The fact that the police have asked for a comparison to be made between two images or an image and an item can in itself create a bias towards confirmation. The use of appropriate procedures, plus the training, experience and competence of the examiner should in combination ensure that in this is being safeguarded against in practice, but these measures should be both strengthened by and demonstrated to be effective through the use of effective QA/QC measures. These measures include the following:

13.4.6.2 Initial competency assessment of an individual prior to commencing forensic casework: the individual is subjected to proficiency testing using characterized test material of known provenance to demonstrate that they, in combination with validated working practices, generate reliable unbiased outcomes.

13.4.6.3 Ongoing competency assessment through use of declared and undeclared trials. Undeclared or blind trials are of particular value as these are more likely to give a truer indication of typical performance and behaviours, unlike a declared trial where the individual knows that they are being observed, and may consequently behave differently to normal by for example being more cautious in their evaluation.

13.4.6.4 Provision of an image line up using “fillers”. This is akin to an identity parade in which for example the analyst may be presented with a number of images comprising that of the target plus a number of other broadly similar “innocent” items, and asked to determine which if any constitutes a match to the image corresponding to the crime scene[70]. A further refinement is to split this

[69] Manchester, P. (2010) An introduction to forensic audio. Sound on Sound. January 2010 http://soundonsound.com/sos/jan10/articles/forensics.html

[70] Kassin, et al (2013). The forensic confirmation bias: Problems, perspectives, and proposed solutions. Journal of Applied Research in Memory and Cognition. 2, p42-52

comparison into two sets so that the examiner does not know whether an individual set contains the target image.

14. ABBREVIATIONS

ACE-V	Analysis, Comparison, Evaluation and Verification
FBI	Federal Bureau of Investigation
ENFSI	European Network of forensic Science Providers
ETHG	European Textile and Hair Group
FSP	Forensic science provider
Hd	Defence hypothesis
Hp	Prosecution hypothesis
LR	Likelihood Ratio
OCF	Open Case File

15. ACKNOWLEDGEMENTS

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Forensic Science Regulator

Overseeing Quality

Codes of Practice and Conduct

Protocol: DNA contamination detection -The management and use of staff elimination DNA databases

FSR-P-302

ISSUE 1

1. INTRODUCTION

1.1.1 The purpose of this protocol is to preserve the integrity of forensic DNA evidence and databases by identifying and preventing the addition of DNA profiles derived as a result of contamination from individuals involved in the DNA process chain. Policies and procedures implemented to achieve this aim demonstrate respect for the privacy of individuals and compliance with the Data Protection Act 1998 with respect to holding relevant and accurate data.

1.1.2 Contamination events from individuals involved in the DNA process chain that have not been detected have:

a. misled high-profile police investigations;
b. wasted resources associated with significant costs; and
c. delayed cases reaching a judicial conclusion through the courts.

1.1.3 For the purposes of this protocol, contamination is defined as *"the introduction of DNA, or biological material containing DNA, to an exhibit or sample during or after its recovery from the scene of crime, or from a person"*. This is distinct from the adventitious transfer of biological material to an exhibit or sample that can also occur, usually prior to the exhibit or sample being recovered and before investigative agencies have intervened.

1.1.4 This protocol is intended to assist in the assessment of forensic science providers, police force scientific units and any other functions as appropriate against BS EN ISO/IEC 17025:2005 for which the operation of an effective staff elimination database is considered to be a prerequisite in order to achieve accreditation and demonstrate compliance with the *Codes of Practice and Conduct for Forensic Science Providers and Practitioners in the Criminal Justice System* (the Codes) (Forensic Science Regulator, 2014).

1.1.5 This protocol should be used in conjunction with other anti-contamination guidelines concerned with the prevention of contamination, being developed by the Forensic Science Regulator:

a. FSR-G-206 Guidance and standards on the control and avoidance of DNA contamination – crime scene examination;

b. FSR-G-207 Guidance and standards on the medical examination of adult and child sexual assault victims;

c. FSR-G-208 Guidance and standards on the control and avoidance of DNA contamination – laboratory examination and published standards;

d. PAS 377:2012 Specification for consumables used in the collection, preservation and processing of material for forensic analysis, and

e. ISO 18385:[1] Minimizing the risk of human DNA contamination in products used to collect and analyze biological material for forensic purposes. The interaction of these guidelines and standards is shown in Figure 1.

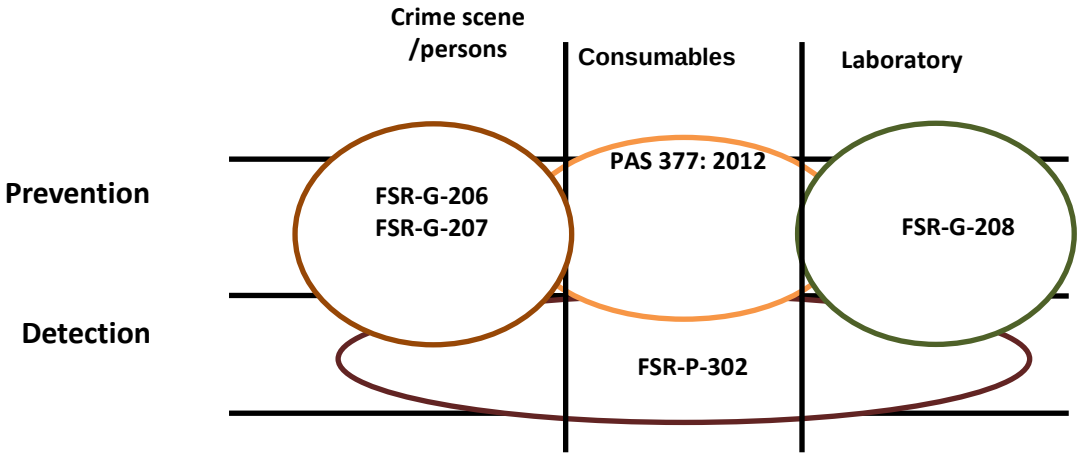

Figure 1: Interaction of anti-contamination guidelines.

1.1.6 From a forensic science perspective, crime investigation activities can be considered as two distinct phases:

a. the pre-submission phase (scene/victim/suspect), during which investigative agencies are involved in locating, recovering, packaging, storing and transporting exhibits; and

b. the analytical phase (laboratory) in which the recovered exhibit is processed within a laboratory.

1.1.7 Contamination can occur at any point in these investigation phases. The principal sources of DNA contamination are:

[1] ISO 18385 is currently under development and may ultimately replace Annex A in PAS 377:2012

a. from personnel to exhibit/DNA sample;
b. from contaminated consumables (for example, swabs, tubes) to exhibit/DNA sample; and
c. from exhibit to exhibit or sample to sample.

1.1.8 Anti-contamination measures fall into two core areas of activity.

a. Prevention of contamination as far as is practicable. Preventative measures entail:

    i. minimising the chance of contamination occurring by, for example, staff using barrier clothing;
    ii. restricting access to areas containing exhibits;
    iii. cleaning laboratory surfaces;
    iv. rendering consumables human DNA-free; and
    v. ensuring that equipment used at scenes of crime is adequately decontaminated between scenes.

b. Detection of contamination primarily entails:

    i. comparison of DNA profiles generated from items against a database of reference DNA profiles from personnel from whom there is a significant risk of contamination;
    ii. cross-checking of profiles within the same batch of samples and from different batches of samples processed within the same laboratory; and
    iii. investigation of unexpected results.

2. SCOPE

2.1.1 This protocol provides the requirements and recommendations on the management and use of elimination databases as a primary means of detecting contamination.

2.1.2 This protocol builds on section 19.4.5 of the *Codes of Practice and Conduct for Forensic Science Providers and Practitioners in the Criminal Justice System* (Forensic Science Regulator, 2014), which stipulates that policies and

procedures are required for elimination databases of laboratory staff, internal/external visitors, equipment suppliers and consumables manufacturers.

2.1.3 This protocol applies to England and Wales. Scotland and Northern Ireland should also institute parallel arrangements for their jurisdictions and databases.

3. IMPLEMENTATION

3.1.1 This protocol is available for incorporation into a forensic science provider's quality management system from the date of publication. This protocol comes into effect from April 2015.

4. MODIFICATION

4.1.1 This is the first issue of this document. The document will form part of the review cycle as determined by the Forensic Science Regulator.

5. TERMS AND DEFINITIONS

5.1.1 The terms and definitions set out in the *Codes of Practice and Conduct for Forensic Science Providers and Practitioners in the Criminal Justice System 2014* apply to this protocol. For the purposes of this protocol, abbreviations are spelled out in section [29] – Abbreviations. The definitions of terms are given in section [30] – Glossary.

5.1.2 The word 'shall' has been used in this document where there is a corresponding requirement in ISO/IEC 17025 or the Forensic Science Regulator's *Codes of Practice and Conduct for Forensic Science Providers and Practitioners in the Criminal Justice*; the word 'should' has been used to indicate generally accepted practice and the word 'may' has been used as recommendations.

6. ACCOMMODATION AND ENVIRONMENTAL CONDITIONS (ISO/IEC 17025 REF 5.3)

6.1.1 It is recognised that DNA contamination incidents cannot be eliminated completely, given the prevalence of human DNA within the environment in which we both live and work. The issue is exacerbated by the increasing sensitivity of DNA analytical techniques. Therefore, an effective DNA anti-

contamination process requires a combination of approaches to both minimise the risk of occurrence (ISO/IEC 17025 ref 4.12) and to maximise the ability to detect contamination when it does occur (ISO/IEC 17025 ref 4.9).

6.1.2 Following batch profile integrity checks and prior to the submission of DNA reference or casework profiles to the National DNA Database® (NDNAD)[2] or prior to communicating the casework results to customers and stakeholders in the criminal justice system (CJS),[3] the DNA profiles shall be compared against:

a. the Laboratory Elimination Database (LED) profiles;
b. the relevant subset of profiles pertaining to the investigating police force, including medical personnel; and
c. profiles of manufacturing staff relevant to the consumables used by the police force and laboratory/forensic science provider (FSP) (ISO/IEC 17025 ref 4.9 and 5.8.1).

6.1.3 Searches against the relevant elimination profile data sets shall also be conducted for profiles that do not meet the aforementioned criteria (for example, mixed profiles or a partial profile derived from a mixture) and those being used for investigative purposes (ISO/IEC 17025 ref 4.9 and 5.8.1).

6.1.4 Exceptionally, under urgent circumstances results may be communicated prior to the elimination databases check, but the fact that contamination checks have yet to be completed shall be made known to the customer and stakeholders (ISO/IEC 17025 ref 4.4).

6.1.5 Where relevant checks against appropriate staff elimination profiles is not possible for whatever reason, then this shall be made known to the customer and stakeholders, for example, by the use of an appropriately worded caveat.

6.1.6 All instances where a match against an elimination profile is observed shall be investigated (ISO/IEC 17025 ref 4.9 and 4.11). The approach shall be on the basis that there is an innocent explanation for the match (see section [18]).

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2 The National DNA Database is a registered trademark owned by the Secretary of State for the Home Department.

3 This applies to prosecution, defence and criminal case review authorities.

7. MANAGEMENT OF PERSONNEL WHO POSE RISK OF CONTAMINATION

7.1 Police personnel

7.1.1 The risk of DNA contamination from police personnel has long been recognised and a Police Elimination Database (PED) has been in existence since 2000. From April 1, 2003 there has been a requirement for all new recruits to provide a DNA sample for inclusion on the PED and since October 2012, for new recruit profiles to be compared against the National DNA Database® (NDNAD) on a one-off basis as part of the vetting procedure.

7.1.2 Unfortunately, since its introduction 'searching' the PED for potential contamination events has been ineffective. This is because authorisation has to be given by a senior police officer, and the check is restricted to a manual comparison of a specific PED profile against a particular result from a specific case in which contamination is suspected. Inevitably police personnel profiles have been inadvertently entered on the NDNAD due to this lack of screening for potential contamination events. The revised approach detailed in this protocol directly addresses these issues.

7.2 Management of profiles from high risk police personnel

7.2.1 High risk individuals shall be screened automatically and routinely against all DNA crime profiles and reference profiles generated from material collected by their own police force.[4] Roles and organisational structures can vary significantly between forces, so each force should conduct its own risk assessment of roles, but in general the following are considered to be high risk.

a. All scene-going staff: crime scene investigators; crime scene examiners; scenes of crime officers; etc. All such roles are considered high risk even if, for example, the individual in question is solely a footwear-mark examiner or fingerprint officer.

[4] For certain roles there will also be a requirement to screen profiles generated by bordering forces with which an individual may undertake overlapping operational activities.

b. All personnel involved in seizure of exhibits in planned operations, including drugs officers who may handle exhibits either at the scene or within a laboratory prior to submission for DNA analysis.

c. Evidence-related property officers, including those handling or opening exhibit bags and splitting those incorrectly containing more than one exhibit.

d. Custody officers and others recovering evidence and handling exhibits in custody suites, including those involved in taking buccal scrapes from detainees for submission to the NDNAD.

e. All personnel involved in handling unpackaged exhibits, i.e. police laboratory staff, including those searching for trace evidence material and screening exhibits. The provision for these staff shall meet the same requirements as those for laboratory staff working for forensic science providers (FSPs) as set out in section [7.7].

7.2.2 Given that it has only been a condition of police service since 2003, provision of a sample by staff recruited prior to this date is on a voluntary basis. Police forces must satisfy themselves that all high risk personnel (defined above) have provided a sample. Those not yet on the PED and who do not volunteer a sample shall either be moved to a low risk role, or their terms and conditions of employment shall be changed, through appropriate consultation, to make inclusion on the PED a requirement.

7.3 Management of profiles from low risk police personnel

7.3.1 Police roles other than those specifically identified as high risk shall be considered as low risk, for example, members of community policing teams. Profiles from low risk individuals shall not be screened unless this is required in particular circumstances, for example, where an officer attends the scene of a serious crime when this is not part of their regular role. Under these circumstances, a record is kept of all attendees entering the scene, as per police policy nationally. As part of the investigation, the Senior Investigating Officer (SIO) or another senior police officer shall typically authorise comparison of profiles from all individuals attending the scene against any recovered crime scene profiles.

7.3.2 It is national policing policy that all new police recruits consent to being entered on the PED and are searched against the NDNAD as part of the vetting process. Existing police staff who consent to be included on the PED as a new requirement for their existing role should also be screened against the NDNAD as a one-off exercise.

7.4 Additional non-police personnel

7.4.1 It is recognised that there are additional groups of non-police personnel, who through their roles may also pose a risk of contamination. These include:

a. vehicle recovery officers;
b. paramedics, doctors, ambulance staff;
c. partner agency staff, for example, social services, those involved in securing premises (boarding up doors and windows, etc); and
d. personnel working for FSPs who do not undertake DNA analysis but do nevertheless examine items (such as mobile phones) that could subsequently be the subject of DNA analysis.

7.4.2 Whilst it may not be proportionate to have elimination profiles for all such groups routinely, it is recommended that provision be made for any instance where this could become an issue.

7.5 Medical personnel

7.5.1 All individuals who routinely enter medical examination rooms, post-mortem facilities or any other rooms used for the examination and recovery of evidential material from either living or deceased victims of crime, shall provide DNA samples for elimination purposes. These include the following groups of individuals.

a. All staff working within Sexual Assault Referral Centres (SARCs) i.e. medical practitioners, crisis workers, cleaning staff, individuals, such as family members or friends, who may be present during a medical examination at the request of the victim.
b. All staff working within post-mortem facilities, including pathologists.

7.5.2 The sampling process, including the wording of consent forms, retention criteria and destruction of unused material are as per laboratory/FSP staff and visitors procedures (see sections [7.9], [7.10], [10] and [11]). Data recorded are the same as for police personnel records, including the details of the police force(s) for which the medical examinations are undertaken and of the particular facility in question.

7.6 Manufacturing staff

7.6.1 All parts of the criminal justice system (CJS) involved in the processing and analysis of DNA evidential material should utilise consumables, where these are available, that are free of detectable human DNA and comply with PAS 377:2012: *Specification for consumables used in the collection, preservation and processing of material for forensic analysis* and when published ISO 18385: *Minimizing the risk of human DNA contamination in products used to collect and analyze biological material for forensic purposes.*

7.6.2 Manufacturers and assemblers of consumables and kits shall establish and maintain an up-to-date collection of DNA profiles from all personnel with access to the manufacturing/assembly work environment and who pose a risk of contaminating the consumables with their own DNA. These can be held in an anonymised form, but ideally a master list should be maintained as per section [13] for FSP staff. This potentially enables the source of contamination to be pinpointed to a specific individual, which facilitates the adoption of effective improvement and corrective actions.

7.6.3 A risk assessment process shall be used to establish the scope of the DNA profile collection, as stipulated in PAS 377:2012. For example, personnel who are involved in physically handling the consumables should be included, as opposed to others who are involved in distribution of materials and are only handling boxes of packaged items. The risk assessment should also consider, where appropriate, the personnel involved in the supply of raw materials used in manufacture.

7.6.4 The anonymised profiles from manufacturing staff shall be provided to create a collection of profiles for contamination detection purposes, i.e. the

Manufacturers Elimination Database (MED). The data format shall meet the requirements for international DNA databases, including the country in which the individual is working as this may impact on the investigation process. Manufacturers may elect to provide this information directly to a centrally held and maintained MED or may provide DNA samples from the relevant personnel to an accredited DNA profiling provider to undertake profiling and submit the profiles on their behalf, for inclusion on the MED.

7.7 Laboratory staff/forensic science providers

7.7.1 Police laboratory staff is included in this category and shall meet the same requirements as laboratory staff in FSPs.

7.7.2 Each DNA profiling provider/FSP shall establish and maintain a Laboratory Elimination Database (LED) against which DNA profiles from casework and reference samples shall be compared for elimination purposes only (ISO/IEC 17025 ref 4.9 and 4.12).

7.7.3 The DNA elimination data shall contain profiles from laboratory trace evidence recovery staff, DNA processing staff, staff involved in sample reception, plus contractors and visitors who enter DNA-sensitive areas.

7.7.4 It shall be a condition of employment for new members of laboratory/FSP staff to give written consent to provide a DNA sample for profiling, and for this profile to be held on the LED. Where existing members of staff do not have this requirement in their original employment contract, they shall either give written consent to provide a DNA sample for the LED, or their terms and conditions of employment shall be modified, through appropriate consultation, to include this as a requirement.

7.7.5 Reference DNA samples shall be taken as part of the induction process for new staff and before they enter a DNA-sensitive area.

7.7.6 All contractors and visitors who require entry to a DNA-sensitive area shall give written consent for their DNA profile to be entered on the LED and provide

reference DNA samples prior to entry. Where possible, they should also be given advance notice of this requirement before arriving on site.

7.7.7 The LED shall also contain unsourced contaminant profiles. These are primarily profiles observed in negative controls and consumable batch tests, i.e. laboratory-owned quality data (ISO/IEC 17025 ref 4.9).

7.8 Unsourced contaminants

7.8.1 DNA profiling providers/FSPs shall search profiles that are categorised as unsourced (these include negative controls and consumable batch test results) against an appropriate MED. Any non-matched profiles shall be submitted and held as a subset of the centrally maintained MED, so that a current pooled collection of these profiles is used to search against profiles from all DNA profiling providers/FSPs. Profiles should meet the minimum load criteria for partial profiles to the MED. This effectively constitutes an unconfirmed supplement to the MED, the value of which is maximised by having the widest possible usage and contribution. This shall be regularly checked to remove duplicate profiles and those for which a source has been identified.[5]

7.9 Sampling

7.9.1 There is no breach of article 8 of the European Convention for the Protection of Human Rights and Fundamental Freedoms if samples are taken with informed consent as a condition of employment.

7.9.2 The sampling and analysis process is as per the requirements of the DNA profiling provider, who shall use a validated[6] DNA profiling method. Once a full designated DNA profile has been generated and quality checks completed, it shall be submitted to the appropriate elimination profile data set.

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5 Consideration should be given to the accepted match criteria for determining a match to an individual and the minimum load criteria. Any profile containing less than 12 alleles could be adventitious.

6 The method used must be proven to perform as required and may be covered by accreditation.

7.10 Destruction of unused DNA material

7.10.1 Following all quality control checks and confirmation that a full profile has been obtained from a donor, any unused sample, including DNA extract, shall be destroyed. Unless there are demonstrable proportionate reasons to retain[7] any unused sample, including DNA extract, it shall not be retained for longer than six calendar months after the sample has been taken. This time period takes due regard of practice set out in legislation for use of DNA samples elsewhere.

7.11 Business continuity

7.11.1 Business continuity plans are required for the operation of elimination databases for staff within the CJS in England and Wales, and where possible for staff of consumables manufacturers supplying to the CJS, in order to meet the provision to provide checks against ongoing cases, appeals and judicial reviews. In the event of closure or ceasing to provide the elimination database screening service, the organisation shall have in place a process to archive and transfer the data to an agreed authorised provider or archive (*Codes of Practice and Conduct for Forensic Science Providers and Practitioners in the Criminal Justice System,* section 6).

7.11.2 The requirement to transfer the data shall be built into the consent form (10.1.4).

8. THE USE AND MANAGEMENT OF DNA ELIMINATION DATABASES

8.1 Organisation of elimination databases

8.1.1 A unified approach to the organisation and use of DNA elimination databases comprising locally, nationally (centrally) and internationally managed DNA elimination databases should be agreed nationally by key policy and standards stakeholders. The sole purpose of this is to detect potential contamination from personnel involved in the manufacture of consumables (swabs, tubes, etc.), and the collection and processing of the DNA samples.

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[7] Consent from the donor to retain their sample for an extended period, or for additional profiling, is a legitimate reason for longer sample retention periods.

8.1.2 Elimination profile data sets shall be established and maintained within which the profiles of the following groups shall be held and compared against crime stain and reference DNA profiles purely for the purposes of identifying potential DNA contamination events (ISO/IEC 17025 ref 4.9 and 4.12).

a. Laboratory/forensic science provider (FSP) staff undertaking processing of evidential samples, and any visitors to the facility who pose a risk of contaminating the DNA samples processed within the organisation, for example, laboratory staff elimination data set – Laboratory Elimination Database (LED).
b. Police personnel, both officers and civilian staff, for example, national (central) staff elimination data sets – Police Elimination Database (PED).
c. All medical staff including forensic medical examiners, Sexual Assault Referral Centre (SARC) personnel, pathologists, and doctors directly or indirectly involved in recovery of evidence from victims of crime, both living and dead, or from arrested suspects, for example, national (central) staff elimination data sets – Medical Examiners Elimination Database (MedExD).
d. Personnel directly involved in the manufacture and assembly of consumables used in the collection, preservation and processing of material in order to generate DNA profiles, for example, national (central) or international staff elimination data sets – Manufacturers Elimination Database (MED).

8.1.3 Each of these groups shall be held within separate sub-databases, which shall be maintained completely separately from the National DNA Database® (NDNAD) and should comply with ISO 27001 Information Security Management.

8.1.4 The rationale for having laboratory elimination databases for DNA profiling providers/FSPs is that these contain profiles of individuals who pose a risk of contamination at a single site or by a single organisation only.

8.1.5 Conversely the PED, MedExD and MED contain profiles from police and other staff that over time may require checking against different FSP submissions depending on changes in service provider contracts, plus some staff may work

for two or more forces that may also use different/multiple FSPs to process their samples. FSPs and police forces may also over time change their consumable suppliers. Hence elimination databases for these groups should be managed nationally as a Central Elimination Database (CED) or internationally with authorised access for searching either by multiple individual forensic DNA profiling providers/FSPs or by central/national database operators on behalf of their criminal justice jurisdiction.

8.2 Subject access

8.2.1 Donors have the right to a copy of their DNA profile where it is associated to them as a named individual. On written request, the organisation shall provide them with a certified copy of their personal information stored on the elimination database. This certified copy can be used as a 'biometric passport', removing the need to be re-profiled if, for example, the person moves jobs to a different FSP or requires access to DNA-sensitive areas in a different organisation. It will be for the elimination database operator to determine whether a copy of the profile is acceptable and meets the profile requirements for inclusion on their elimination database.

8.3 Retention periods on elimination database

8.3.1 Consideration shall be given to retention periods that are relevant to their role once staff have left and the expected period of time that relevant material handled by them will be in the criminal justice system (CJS) before DNA profiles are generated.

8.3.2 The shelf life of manufactured consumables should be considered for determining the retention period of manufacturing staff data. Laboratory contamination with an 18-month interval has been observed; therefore unless contamination incidence data provide evidence to the contrary, then as a minimum profiles shall be retained for searching for 18 months after staff have left the organisation. In the case of contractors/visitors six to twelve months after last entering a DNA-sensitive area and for police officers in attendance (excluding crime scene recovery staff and subject to exhibit submission periods) six months may be more appropriate.

8.4 Archive

8.4.1 The requirement for the archive and the retention period should be determined for each elimination database or staff role, be relevant, proportionate and shall form part of the consent required from staff working within the CJS in England and Wales.

8.4.2 Once the period for retaining a profile on the live elimination database has elapsed for staff exiting the CJS [10.1.3] then the data may be deleted or stepped down by either annotating the record, removing the individual's name or transferring the record to an archive, providing consent has been given.

8.4.3 The record could be retained for up to 30 years in order to be available for:

a. checks against cold cases; and
b. appeals and judicial reviews.

Retention periods are set out in the code of practice issued under the Criminal Procedure and Investigations Act 1996 (CPIA) for England and Wales. As cases have been tested for DNA after 20 years, then the minimum retention period could be set at 20 years, if deemed appropriate (ISO/IEC 17025 ref 4.1.2).

8.4.4 Access and searching against any archived profiles shall be restricted [9.1.19] only for the purposes stated above [8.4.3].

8.5 Interface with international Manufacturers Elimination Databases

8.5.1 Consumables used in the processes of sampling and DNA profile production are widely used by the police, laboratories/FSPs globally; many examples of profiles from manufacturing staff having been observed in multiple countries have been documented (Sullivan *et al*., 2004). With increasing sharing of biometric data including DNA across borders, particularly in Europe as a result of the Prüm Treaty decisions, sharing of information regarding contamination is becoming ever more important if the integrity of the DNA comparisons is to be assured.

8.5.2 The DNA Working Group of the European Network of Forensic Science Institutes (ENFSI) is continuing to work towards shared manufacturers and unsourced contaminants databases. The forensic DNA community and FSPs should collaborate with such international initiatives, particularly in sharing unsourced contaminant profiles, including:

a. collaborations to have reciprocal agreements for facilitating searching of local, central or internationally held MEDs and unsourced contaminant profile records by both UK and international forensic DNA profiling laboratories/FSPs; or

b. where contaminations checks are carried out after loading to NDNADs, by central/national database operators on behalf of their forensic DNA profiling laboratories as appropriate for the purposes of their criminal justice system.

9. RESPONSIBILITIES AND CODE OF CONDUCT

9.1.1 All parties within the criminal justice system (CJS) involved either directly, for example, the police, forensic science providers (FSPs), medical examiners or indirectly, for example, consumables manufacturers, in the processing of DNA samples should recognise that contamination of samples and potential inclusion on the National DNA Database® (NDNAD) is an occupational hazard for workers within the CJS, and that employers have a duty of care to employees to minimise the risk of this happening.

9.1.2 Whilst the occurrence of contamination from personnel within the CJS can be minimised through the adoption of appropriate anti-contamination measures, this risk cannot be completely eliminated. Hence effective management of contamination requires a combination of actions both to minimise the frequency of occurrence and maximise the chances of its detection through the use of effective elimination databases (ISO/IEC 17025 ref 4.9 and 4.12).

9.1.3 All personnel should be given the option, if they wish, for their profile to be subject to a one-off search against the NDNAD (ISO/IEC 17025 ref 4.9). This is in response to a legacy issue where people working within the CJS may have been at high risk of contaminating evidential material prior to the implementation

of comprehensive and effective DNA elimination checks. For police personnel see section [7.1], but should also be extended to other groups, including medical personnel and consumables manufacturing staff.

9.1.4 All matches against DNA elimination databases shall be investigated (ISO/IEC 17025 ref 4.11) and all investigations shall be undertaken from a standpoint that the match has arisen due to an inadvertent contamination or other innocent circumstances; past experience has demonstrated this to almost always be the case.[8] Further use of the matching reference or crime stain profile shall be put on hold until the investigation has been completed. Responsibility for investigating an identified match lays with the organisation within which it has been observed, for example, the police, FSP, or consumables manufacturer. Outcomes of the investigation shall be fed back to the end user. See also section [17.1.2].

9.1.5 All investigations shall be undertaken sensitively and discreetly by nominated individuals. The individual being investigated (where known) shall be kept informed of the progress of the investigation, and the exercise should be undertaken as a means to identify potential improvement actions rather than as a route to disciplining staff, unless it transpires that the individual has repeatedly failed to follow written procedures.

9.1.6 Even when anti-contamination procedures have been correctly followed, contamination events are known to occur through no fault of the individual concerned. For example, some people are more prone to shed DNA than others and therefore more at risk than others of contaminating. In extreme cases this can result in an individual repeatedly contaminating with their own DNA despite wearing appropriate protective clothing and correctly following procedures. If all

[8] The Forensic Science Service maintained a comprehensive elimination database comprising both staff and personnel from consumables manufacturers. Over a period of more than a decade several million crime and reference DNA samples were processed and routinely compared against this elimination database. Virtually all observed matches were attributable to contamination occurring either within the laboratory or manufacture of consumables. Only two instances could not be attributed to contamination. On investigation, these were both found to be due to items that were attributable to members of staff being associated with a crime scene by innocent means.

preventative measures fail then consideration shall be given to moving the individual to a different role.

9.1.7 Where the FSP has conducted an investigation and the investigator is satisfied that the observed match is explicable through contamination, the police customer shall accept this outcome and the identity of the individual shall not be disclosed; only the alleles in the person's profile that match the crime stain shall be included in any contamination report.

9.1.8 Only in the rare event that the investigation concludes that the match is not explicable through contamination or other innocent means, it may be necessary, depending on circumstances, for the name of the individual concerned (where known), or the name of the organisation if individuals are anonymised, to be divulged to the police via a single point of contact, in order to facilitate further investigation for elimination purposes. For example, knowing whether the manufacturer is UK-based may have a bearing on police considerations regarding the need for follow-up investigations.

9.1.9 DNA profiling providers/FSPs and manufacturers shall work together collaboratively to address the issue of contamination of consumables. The fact that contamination cannot be completely eliminated should be the guiding principal. Detection of contamination should be used by FSPs as an opportunity to provide regular feedback to manufacturers to enable continuous review and improvement of their quality procedures, rather than as a reason to undertake legal action against the manufacturer for provision of a non-conforming product.

9.1.10 It is the responsibility of police forces to provide up-to-date data to the Police Elimination Database (PED), including changes to records and search parameters to ensure that, as far as is practicable, screens continue to be restricted to all relevant individuals and no others. These changes include the following:

a. PED records, reflecting movement of police personnel from one force to another or exiting the CJS;
b. medical staff records, reflecting any changes of the medical staff utilised by a force.

9.1.11 Search/trace evidence recovery laboratories shall provide their DNA profiling providers/FSPs with up-to-date information on the consumables that they use so that the relevant manufacturing staff can be searched against these.

9.1.12 It is the responsibility of DNA profiling provider/FSP and police laboratories to maintain up-to-date staff elimination profile data sets (ISO/IEC 17025 ref 4.1.2).

9.1.13 It is the responsibility of manufacturers to maintain a current collection of DNA profiles for contamination detection, and where appropriate to provide up-to-date data to the centrally held Manufacturers Elimination Database (MED), i.e. new profiles, removal of old profiles, update of details, etc.

9.1.14 For the MED it is the responsibility of manufacturers, as the data owners, to determine the user communities in addition to forensic DNA profiling laboratories that are authorised to check against their elimination profile records; these may, for example, include organisations that provide testing for them, produce reference DNA materials or generate proficiency test samples.

9.1.15 It is the responsibility of the MED operator(s) to establish the user communities in addition to forensic DNA profiling laboratories that are authorised by the manufacturers to check against the elimination profile records being held and processed on their behalf.

9.1.16 It is the responsibility of the elimination database operator(s) to ensure that they are registered with the Information Commissioner's Office (ICO), unless they are exempt, as failure to do so is a criminal offence.

9.1.17 It is the responsibility of the elimination database operator(s) to establish ownership of the data, whether they are the data owner (for example, data from their own staff), the data processor (for example, holding and processing data from other organisations) or both. It is important to clarify who the data controller is, and ensure compliance with the Data Protection Act 1998 for organisations based in the UK (ISO/IEC 17025 ref 4.1.2 and the *Codes of Practice and Conduct for Forensic Science Providers and Practitioners in the Criminal Justice System* [the Codes], 20.18.3). For manufacturers based

overseas, the data protection laws relevant to their own country shall be observed.

9.1.18 It is the responsibility of the elimination database operator(s) to demonstrate that the software and algorithms used are appropriate and fit for purpose (the Codes 20.18.4 and 21.1 and ISO/IEC 17025 ref 5.4.5 and 5.5).

9.1.19 Security of the elimination database records shall be maintained by enforcing restricted access to nominated authorised individuals, and through working practices that ensure compliance with the Data Protection Act 1998. The data shall be backed up and transmitted in accordance with the Government's *Security Policy Framework* (ISO/IEC 17025 ref 4.1.2 and 5.7 and the Codes 20.18, 20.18.2 and 20.18.3).

9.1.20 Procurement functions shall ensure that consumables purchased for the collection, retention and processing of DNA samples comply with PAS 377:2012, where these exist and are appropriate (ISO/IEC 17025 ref 4.6).

9.1.21 It is the responsibility of laboratories/FSPs and police forces to inform their consumable suppliers of the importance for the manufacturing staff to provide elimination DNA profiles to the MED as appropriate.

9.1.22 DNA profiling providers/FSPs shall provide the MED with regular updates of contamination profiles that are categorised as unsourced, so that a current pooled collection of these profiles is used to search against profiles from all FSPs. This effectively constitutes an unconfirmed supplement to the MED, the value of which is maximised by having the widest possible usage and contribution (ISO/IEC 17025 ref 4.9).

10. CONSENT FORM (see also section 25)

10.1.1 All individuals entered on to an elimination database shall sign and date a consent form that provides consent for providing the sample and confirms the basis on which a sample is provided, which should include but is not limited to the following.

a. The organisation is authorised to collect a DNA sample and generate a DNA profile from it.

b. The organisation shall provide a written explanation with the consent form explaining the management of the elimination database, including how investigations are conducted in the event of a match and arrangements for retention and removal of profiles, both on a routine basis and on request.

c. The results will be used solely for comparison with profiles generated from casework or reference samples in order to detect contamination incidents. Where contamination is observed, investigations are targeted towards identifying improvements rather than disciplining staff.

d. The organisation will retain a copy of the results of the tests performed on the sample, ideally with the metadata and profile stored separately and not accessible except by a restricted number of staff to conduct investigations. The organisation will not disclose the information in any way other than as authorised in the consent form, or as may be required by law.

    i. Specific authorisation may be sought on the form for limited disclosure of the profile to other accredited forensic providers where necessary, and following agreement between the respective Human Resources (HR) departments (see section [21]).

    ii. In the event of the operator of the elimination database ceasing to operate the data are transferred to another authorised operator or archive that meets the existing security and legal requirements of the organisation that owns the data to be transferred.

10.1.2 The individual agrees to provide a DNA sample on a voluntary basis if it is not part of their terms and conditions of employment, and the profile will not be uploaded to the National DNA Database® (NDNAD) nor compared against it except with their explicit permission, although the latter is strongly recommended for individuals exiting employment within the criminal justice system (CJS).

10.1.3 Profiles will be retained until the individual no longer poses a risk of contamination once they have left the DNA process chain.

10.1.4 After the specified time the individual's profile shall be stepped down in the live elimination database, deleted or transferred to a restricted archive database for cold case review appeals and judicial reviews use only. If this is not part of their terms and conditions then consent shall be required from staff as appropriate to work within the CJS in England and Wales.

10.1.5 If there are any specific proposals to vary the basis on which the data are held or processed, a further specific written consent would be required from the individual who originally provided the profile.

11. INFORMATION RECORDED AND RETAINED ON ELIMINATION DATABASES

11.1 Data format

11.1.1 As a minimum, entries of information shall use a data format and other configuration parameters that closely align to those defined in the 'DNA chapter 1' of the annex to the EU Council Decision 2008/616/JHA used for the Prüm DNA data exchange and applications. This allows for interoperability between different elimination databases.

11.1.2 Only unsourced profiles and by agreement with the manufacturer shall the Manufacturers Elimination Database (MED) data be shared with other countries.

11.2 Data fields

11.2.1 Each entry shall as a minimum include the following information.

a. A reference number unique to the individual.[9]
b. A country code[10].
c. The organisation for which the individual works/data owner/controller.
d. The multiplex kit(s) used.

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9 Personal information, for example, name shall not be held on any of the elimination databases.

10 Relevant to, manufacturing staff elimination data and laboratory location for unsourced contaminants.

e. The profiling organisation (if this is different to the organisation that the individual works for and is authorised by the data owner/controller to load, amend, delete that profile and for follow-up profile queries).

f. The sample category (for example, manufacturer, police, medic, contractor, visitor, or an unsourced profile).

g. A full designated short tandem repeat (STR) DNA profile provided using a validated profiling system.

11.2.2 For local and national (central) elimination database profiles – utilising the current standard multiplex in use and that meets the allele reporting requirements determined from the validation of the method used or the load criteria set for the National DNA Database® (NDNAD).

11.2.3 For MED profiles – utilising a multiplex that provides comprehensive coverage of the European Standard Set of loci (ESS) and the United States Combined DNA Index System (CODIS) loci in use globally, but as a minimum it should include all SGMPlus® loci. This is to account for the fact that consumables will be used for processing samples using any number of STR multiplex kits and will allow any users of these consumables to carry out meaningful searches for manufacturer contamination regardless of which STR profiling system is used by them.

11.2.4 The exception is for unsourced contaminants – as a minimum this shall be a partial profile utilising the current standard multiplex in use that meets the criteria set by the DNA profiling provider/forensic science provider (FSP) for searching their Laboratory Elimination Database (LED) for matches, and may be lower than the criteria for loading to the NDNAD. For those profiles to be added to the MED, the minimum load criteria set for the MED shall be met. It is recommended when appropriate for inclusion on the MED that re-profiling is undertaken, utilising a multiplex to obtain a more discriminating profile to minimise occurrence of adventitious matches.

11.2.5 For all elimination databases, including the MED, consideration should be given to data fields that indicate archive and deletion dates and a flag for records

where the profile is anonymised that any match against that record will not necessarily be traceable to the individual.

12. LEGACY PROFILES

12.1.1 For existing elimination databases the profiles will have been generated using short tandem repeat (STR) multiplexes that have been superseded by more sensitive discriminating STR profiling technology, therefore consideration should be given to re-sampling staff and upgrading profiles if at all possible. The discriminating power of the legacy profiles will have a bearing on the searching and matching regime used against these profiles.

13. ADDITIONAL RETAINED INFORMATION

13.1.1 Ideally Human Resources (HR) or an equivalent function or authorised individual, such as the data protection officer, should maintain a master list in which the names of individuals are linked to the unique reference number held within a secure system. Data shall be maintained in compliance with the Data Protection Act 1998 for UK operators. Overseas manufacturers should give due regard to the legislation of their own country. Access to this list shall be protected and available to only a few nominated authorised individuals permitted to search the data when a specific contamination incident is being investigated.

13.1.2 For manufacturers outside the UK, where national legislation would prevent the name of the individual being held, then information as to the parts of the manufacture process that they are involved in can be recorded to aid identifying possible areas for quality improvements should there be a match against an anonymised DNA profile record.

14. SEARCHES AGAINST ELIMINATION DNA PROFILE RECORDS

All profiles, either single source or component(s) of interest in interpreted mixtures, casework or reference, shall be compared against the relevant laboratory staff elimination profiles held on the Laboratory Elimination Database (LED)/Central Elimination Database (CED), plus the relevant subset of profiles

pertaining to the investigating police force, including medical personnel, unsourced and manufacturing staff profiles pertaining to the consumables used by the police force and laboratory (ISO/IEC 17025 ref 4.9 and 4.12) (also see section [6]). An example of a schematic for checking against elimination databases is shown in Figure 2.

14.1.1 Wherever possible data input should be automated to avoid DNA profile data errors. Where manual input of the data cannot be avoided then processes such as double entry or a second independent check shall be implemented and documented.

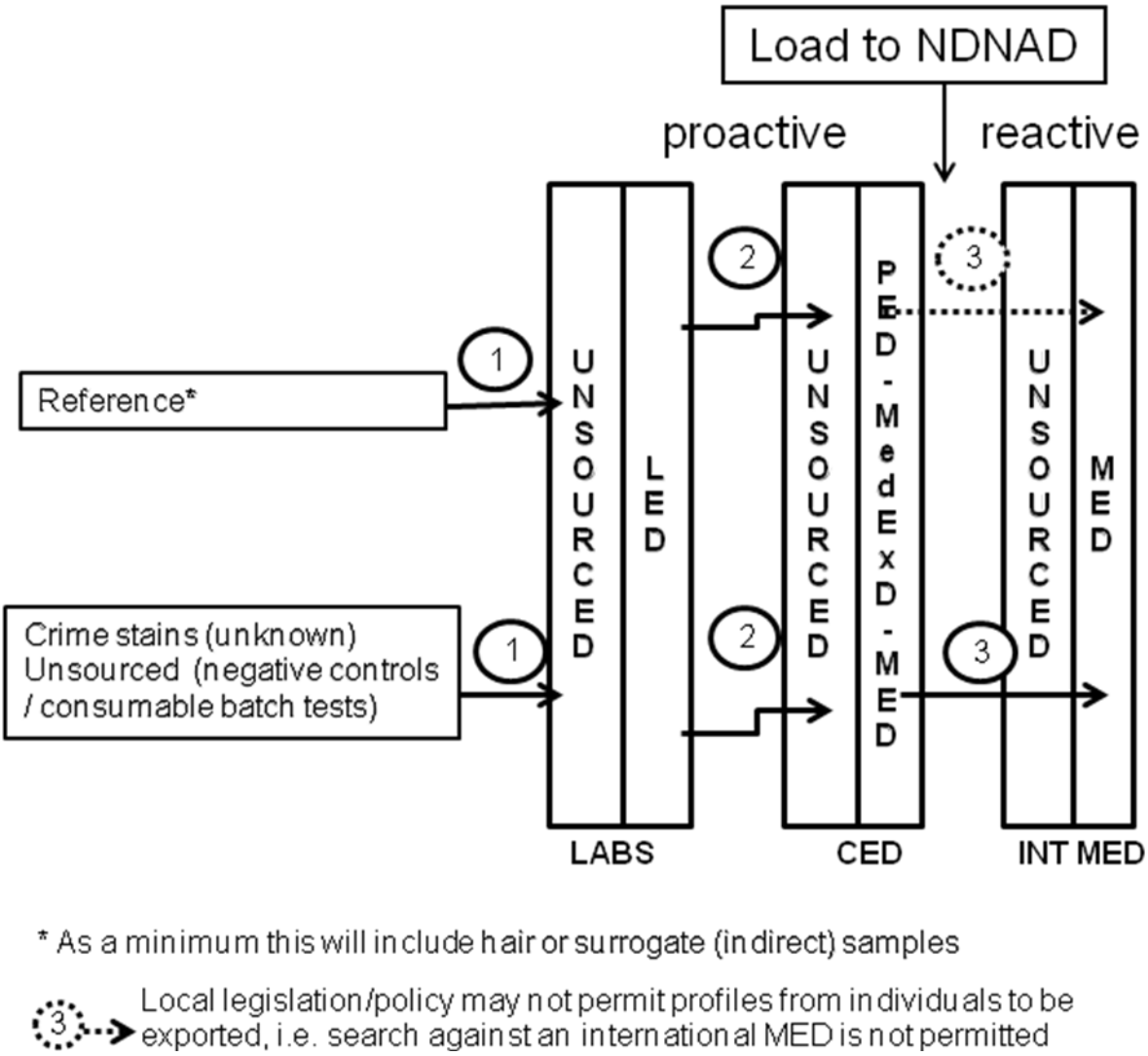

Figure 2: Example schematic for elimination database screening.

15. SEARCHING

15.1 Match regime

15.1.1 The searching and matching regime shall optimise the identification of contaminating profiles but minimise the number of adventitious matches. The regime shall take into account the number of alleles that a forensic science provider (FSP) will use to report:

a. a statistical match probability to the court;

b. the minimum load criteria for the local,[11] national and international databases;

c. the number of elimination records held in the elimination database;

d. the discriminating power of the elimination DNA profiles held; and

e. in particular, any legacy profiles and the short tandem repeat (STR) multiplex kit(s) used to generate the profiles being compared.

15.2 Same short tandem repeat polymerase chain reaction chemistry/multiplex

15.2.1 For searches against profiles generated using the same polymerase chain reaction (PCR) chemistry/multiplex, demonstrable consideration shall be given to high stringency searching and searching to accommodate for profile anomalies, such as allele mis-designation or omission and the rarer event of a somatic mutation.

15.2.2 Demonstrable consideration should be given to ensuring that the searching of profiles is conducted on numbers of alleles that maximise the chances of detecting contamination, yet also minimise the numbers of false positives generated (*Codes of Practice and Conduct for Forensic Science Providers and Practitioners in the Criminal Justice System* [the Codes], 20.18.4 and ISO/IEC 17025 ref 5.4.5).

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[11] The load and search criteria for the Laboratory Elimination Database (LED) can be less than that permitted for loading and searching against the Central Elimination Database (CED) and the Manufacturers Elimination Database (MED) as the LED will contain fewer staff profiles to search against, thus will have a higher tolerance to adventitious matches for partial profiles. This will allow for the identification of profiles that are more prevalent but difficult to identify due to the partial nature of the profiles.

15.2.3 For a highly discriminating search profile then high stringency matching with an N-1 routine shall be undertaken. N-1 means that the search profile can contain a single designated allele difference at one locus and is not position specific (i.e. can be in either the high or low molecular weight position).

15.2.4 The partiality of the profile where N-1 searching is unsuitable should be determined using the considerations listed in [15.1.1] (for example, for search profiles with less than 8 alleles present against the LED and less than 10 alleles present against the CED and MED).

15.3 Different short tandem repeat polymerase chain reaction chemistries/multiplexes

15.3.1 For searches against profiles generated using different PCR chemistries/multiplexes, demonstrable consideration shall be given to high stringency searching and searching to accommodate for profile anomalies such as allele mis-designation or omission and the rarer events of non-concordance and somatic mutations (the Codes 20.18.4 and ISO/IEC 17025 ref 5.4.5).

15.3.2 For a partial search profile determined to be unsuitable for N-1 searching as per [15.2.4] then high stringency matching shall be undertaken.

15.3.3 For a discriminating search profile (for example, 11 to 16 alleles present) then high stringency matching with an N-1 routine should be undertaken.

15.3.4 For a highly discriminating search profile (for example, 17 or more alleles present) then high stringency matching with an N-1 routine shall be undertaken and an N-2 routine should also be undertaken. N-2 means as a minimum that the search profile and the retained elimination database profile contains a single designated allele difference at one locus, of which the loci could be different for each profile and is not position specific (i.e. either high or low molecular weight allele). The relevance of two differences in a crime stain profile of interest compared with an elimination profile should be considered as a targeted N-2 search condition (for example, for use on search profiles derived from component[s] of interest in interpreted mixtures). The N-2 routine could automatically produce N-1 matches.

16. REPORTING MATCHES

16.1.1 A match to a crime stain/reference DNA profile shall be reported directly to the search requester, typically the DNA profiling provider/forensic science provider (FSP) or central/national operator where appropriate, on generation of the match.

16.1.2 Where a match against a profile from a consumables manufacturer is observed, the manufacturer should also be notified. See Table 1.

Target profile source	Match against	Match report sent to single point of contacts
Undetected crime stains Reference samples Unsourced (negatives)	**LED** (CED), for example, laboratory staff i.e. DNA profiling lab/FSP/police force	• Target profile owner • Lab/FSP/ police force • (Target profile provider if match generated from CED- PED subset)
	PED, for example, police officers, CSIs, contractors i.e. force	• Target profile owner • Force • Target profile provider
	MED, for example, manufacturing and kit assembly staff i.e. consumable suppliers	• Target profile owner • Manufacturer • Target profile provider
	MEDExD, for example, forensic pathology, medical and nursing staff – i.e. force contracted to SARC, Department of Health, etc.	• Target profile owner • Force/SARC/pathology unit as pre-determined as the staff profile owner on MEDExD • Target profile provider
Unsourced (negatives)	**MED – unsourced**	• Target profile owner • DNA profile providers (target and matched profiles) • CED • NDNAD data integrity

Table 1: Guide to where matches should be sent for investigation and follow up.

16.1.3 All demographic information, except for the individual's name (if it is held), and the search profile submitted alongside the matching loci (including the N-1, N-2 near match) of the nominated profile shall be provided with a unique match reference number, to allow for the distinction between repeat profile searches and for audit, tracking and follow-up purposes.

16.1.4 Following the investigation (see section [18]) of the match the organisation shall provide feedback as to the outcome of its investigation to relevant stakeholders.

17. MANAGEMENT INFORMATION

17.1.1 Records shall be maintained (ISO/IEC 17025 ref 4.13.2, 5.4.7 and the *Codes of Practice and Conduct for Forensic Science Providers and Practitioners in the Criminal Justice System,* (20.18) of all reported matches and outcomes of the investigations (see [9.1.4] and [17.1.3]). Reviews of contamination rates and trends shall be periodically undertaken as appropriate within the laboratory/forensic science provider (FSP) quality management review meeting (ISO/IEC 17025 ref 4.15).

17.1.2 Records shall be maintained of the matches and outcomes of the investigations and made available on request to the Forensic Science Regulator/designated representative or nationally authorised forensic assurance unit for appropriate reviews/analysis/monitoring of contamination rates and trends.

17.1.3 The details recorded and reported for trend analysis and management information purposes should include the following:

a. single source or mixture profile result;
b. full or partial profile match i.e. match probability/confidence;
c. the type of event i.e. person to person, person to item, item to item;
d. direct or indirect transfer i.e. primary or secondary transfer;
e. the stage, process or place in the process where the contamination event occurred i.e. the consumable, equipment, environment, recovery, packaging, examination, sampling, extraction, polymerase chain reaction (PCR) and post-PCR;

f. time line i.e. especially where indirect contact/secondary transfer is the only feasible explanation;

g. other relevant information to aid trend analysis, understand mechanisms of transfer and improve anti-contamination good practice, for example, staff repeat incidences, faulty air flow, cleaning regime, storage conditions, skin condition, etc.

18. INVESTIGATION PROCESS

18.1 Match investigations

18.1.1 All instances where a match against an elimination database profile is observed shall be investigated. The default position is that there is an innocent explanation for the match.

18.2 Match of reference (Police and Criminal Evidence Act 1984) sample to a laboratory staff elimination profile record

18.2.1 An investigation shall be undertaken to determine if contamination occurred during processing of the Police and Criminal Evidence Act 1984 (PACE) sample, and full records shall be maintained of all instances, investigative steps taken, conclusions drawn and subsequent corrective actions taken. Throughout the investigation, the individual member of staff should be kept fully informed of progress. Investigations may include one or both of the following steps, depending on the circumstances:

a. the investigation may include processing the second sample (swab), where this still exists due to the constraints of PACE as modified by the Protection of Freedoms Act 2012, as a quality assurance measure; and

b. if the match was against a member of staff involved in the processing of reference samples, they should not be involved in the reprocessing of the second sample.

18.2.2 If the profile obtained from the second sample does not match the profile from the first, attempts should be made to determine the point at which contamination occurred, by re-extracting and re-amplifying sample 1 and/or re-amplifying the DNA extract from sample 1.

18.2.3 If the profiles from the first and second samples match, this indicates that contamination may not be the only explanation. The DNA profile result shall be loaded to the National DNA Database® (NDNAD) once the NDNAD Assurance Service (NAS) has confirmed that the match is not due to the individual being a donor of a quality assurance (QA) sample used in an NAS blind trial.

18.3 Match of scene of crime profile to a Laboratory Elimination Database profile record

18.3.1 An investigation shall be undertaken to determine if contamination occurred during processing of the scene of crime (SOC) sample within the laboratory environment, and full records shall be maintained for each investigative step taken, conclusions drawn and subsequent corrective actions taken.

18.3.2 Investigations may include some or all the following steps depending on the circumstances:

a. the investigation may include reworking the original material, for example, by re-electrophoresis, re-amplification or re-extraction from the stored extract component;
b. if the match was against a member of staff involved in the processing of SOC samples they should not be involved in the reworking; and
c. where appropriate, a deep clean should be conducted of the laboratories where the contamination may have occurred and where any re-processing takes place, before re-processing is undertaken.

18.3.3 If the profile obtained from the rework no longer matches the original profile (if a single source) or in the case of a DNA mixture no longer contains the components that matched, the rework result may be used for casework analysis or for NDNAD applications, provided all the other required quality criteria are met.

18.3.4 If the profile remains unchanged on the reworking of the original material, then the original item should be re-examined and, where possible, attempts should be made to re-sample, i.e. take a new previously unprocessed part of the

material, for example, a different area of the same stain or other discrete source of biological material.

18.3.5 If the re-sampled material no longer matches the original elimination profile record the rework result may be used for casework analysis or for NDNAD applications, provided that all other required quality criteria are met.

18.3.6 If the re-sampled material still provides a profile that matches against the original elimination profile record, another item linked to the same case or a different stain from the same item should be sought and processed, if these options exist.

19. FOLLOW-UP ACTIONS IN THE EVENT OF A CRIME STAIN MATCH AGAINST A LABORATORY ELIMINATION DATABASE PROFILE RECORD

19.1 Actions where contamination is a feasible explanation for the observations

19.1.1 In virtually all circumstances where the profile from an individual matches that from an exhibit that they have had either direct or indirect exposure to, it is reasonable to believe that this has arisen through innocent means, of which contamination is the likely cause.

19.1.2 The investigation process outlined in [18.3.2] is designed to elicit information regarding the probable mechanism by which contamination may have occurred. However, not all investigations into instances of matches against the laboratory staff elimination profile can be completed, for example, if insufficient material remains to enable rework to be undertaken, or only a partial profile can be generated. Under these circumstances the conclusions drawn should also be that contamination is the likely cause, but that it cannot be proven. As a guide the following actions should be considered.

a. Notify all relevant staff (for example, the individual involved, their line manager, the quality leader, and other senior managers as dictated by the severity of the impact of the contamination) on the outcome of the investigation. It is not necessary to disclose the name of the person

involved to staff or senior managers that are not relevant to the investigation.

b. Inform the person involved in the match, or where this person's sample has been anonymised, the organisation for which they work.

c. Document that contamination may have occurred on the case-file, together with the summary of the investigation (ISO/IEC 17025 ref 4.13.2).

d. Record the incident in the laboratory contamination log (ISO/IEC 17025 ref 4.13.2). This should be regularly reviewed to identify trends in contamination and potential improvements to reduce the risk of recurrence. These actions should be captured within the improvement and corrective action process, the operation of which is a requirement of ISO/IEC 17025 (ref 4.11).

19.2 Actions where contamination is not a plausible explanation for the observations

19.2.1 Circumstances in which contamination is not a plausible explanation for a match are extremely rare.

19.2.2 Typically this is where a discrete item, such as a piece of chewing gum or blood stain, is of sufficient size and DNA yield to enable re-sampling from a separate part of the same item, and this repeatedly yields a full DNA profile matching against the Laboratory Elimination Database (LED). Under these circumstances, assuming it is not a quality assurance sample used in an National DNA Database® (NDNAD) Assurance Service (NAS) blind trial, the following actions may be required, with the individual in question, where known, kept fully informed throughout.

a. Disclosure to the investigating police force of the name of the individual concerned where this is known, for example, a member of staff of the forensic science provider (FSP).

b. Disclosure of the organisation for which the matching individual works, where their name has not been provided to the FSP, for example, sub-contractors.

c. Depending on the circumstances of the case, the police may wish to make further inquiries with the individual in question in order to eliminate them from the investigation.

d. Disclosure of the incident to senior managers within the relevant FSP, with subsequent actions according to the organisation's Human Resources procedures.

e. At the conclusion of the investigation a decision should be made in conjunction with the police force on whether the crime profile should be entered on the NDNAD.

20. BROADER CONSIDERATIONS IN CONTAMINATION INVESTIGATION

20.1.1 Knowledge regarding the mechanisms by which DNA contamination can occur is still developing and will continue to do so in line with the evolution of increasingly sensitive DNA profiling technology.

20.1.2 Investigations into contamination events should not just focus on the processing of exhibits for DNA analysis and the events within the rooms in which samples have been processed, but should take a wider view of activities within the entire building. For example, any activities including inspection, cleaning or maintenance of air management systems within the same building, even if remote from DNA clean areas, or any other kind of structural perturbation of the building, increases the risk of contamination occurring. This risk should be addressed by additional non-routine deep cleaning and environmental monitoring as required.

21. COLLABORATIVE CONTAMINATION CHECKS BETWEEN FORENSIC SCIENCE PROVIDERS

21.1.1 Where an accredited DNA profiling provider/forensic science provider (FSP) is undertaking analysis on material previously examined by a different DNA profiling provider, it may be necessary to check any new profiles generated against the Laboratory Elimination Database (LED) of the original examining FSP. Where this is undertaken, the crime stain profile shall be provided to the original examining DNA profiling provider/FSP for an LED search. Where a match is observed, release of the information is limited to the alleles shared with

the crime profile, provided the consent form has explicitly allowed for such disclosure (sections [10][and [25]).

22. MATCH OF REFERENCE (POLICE AND CRIMINAL EVIDENCE ACT 1984) SAMPLE OR CRIME STAIN TO A POLICE ELIMINATION DATABASE PROFILE RECORD

22.1.1 The Investigating Officer (IO) and where appropriate the Scientific Support Manager (SSM) shall be informed that a match to a crime stain/reference DNA profile has been obtained against a police staff elimination profile, disclosing demographic and matching profile information as agreed in the consent form.

22.1.2 It is the responsibility of the police force(s) involved in the match to investigate and advise the reporting DNA profiling provider/forensic science provider (FSP) whether or not contamination is the accepted explanation for the match and agree any follow-up actions and reporting requirements as necessary.

23. MATCH OF REFERENCE (POLICE AND CRIMINAL EVIDENCE ACT 1984) SAMPLE OR CRIME STAIN TO A MANUFACTURING STAFF ELIMINATION PROFILE RECORD

23.1.1 The Investigating Officer (IO) and the police force single point of contact (SPOC) shall be informed that a match to a crime stain/reference DNA profile has been obtained against a manufacturing staff elimination profile, providing matching profile information as agreed in the consent form.

23.1.2 Consideration should be given to whether contamination is the accepted explanation, particularly if the consumable manufacturer is not UK-based. The DNA profiling provider/forensic science provider (FSP) shall advise the police force if any follow-up investigation should be undertaken for the match, and agree any follow-up actions and reporting requirements as necessary.

23.1.3 The manufacturer shall be informed that a match to a crime stain/reference DNA profile was obtained against one of their staff elimination profiles, providing matching profile information as agreed in the consent form, to enable the manufacturer to investigate and feedback. The outcome of the investigation

should be used for continuous review and improvement of its quality and staff training procedures.

24. CONTAMINATION REPORT

24.1.1 Should the police require a contamination report (ISO/IEC 17025 ref 5.10) it should be provided.

24.1.2 Where a contamination report has been prepared it is revealable and disclosed to the Crown Prosecution Service (CPS) and should be notified to the police for inclusion in any schedule of unused material prepared for the purposes of criminal proceedings. A summary on the schedule in similar terms to that outlined below might assist the prosecutor in determining whether a report is required, or whether there is a need to disclose.

a. The contamination report must not identify the person involved by name.
b. The report should explain the principles by which the appropriate Laboratory Elimination Database (LED), Police Elimination Database (PED) or Manufacturers Elimination Database (MED) operates and the nature of investigations undertaken when a match occurs.
c. The investigation undertaken should be outlined in the report, identifying the root cause of the observed match and corrective actions taken where appropriate.
d. The report should include wording along the lines of:
"The result/components of the DNA profile obtained from item x has matched a DNA profile held on the Laboratory Elimination Database/Police Elimination Database/Manufacturers Elimination Database. As the result/the component of the mixture relates to an individual involved with the laboratory analysis/sample handling/manufacturing process, the profile/component of the mixed profile can be assumed to be the result of contamination at the laboratory/scene/manufacture. As such, it has been treated as having no evidential value and has not contributed to my interpretation".

25. ELIMINATION DATABASE CONSENT FORM

25.1.1 The principles and basis for obtaining consent for elimination samples for inclusion on an elimination database is set out in section [10]. An example template that can be customised as appropriate is set out below.

25.2 Elimination database consent form template – visitor example

1. I recognise that in the course of my employment or during my attendance at a scene or visiting a facility processing forensic material, I may come into contact with *(select as appropriate)*:
    a. *items, samples or extracts on which DNA analysis may be required*;
    b. consumables to be used in the collection, storage and processing of samples for DNA analysis.
2. As such there is a possibility that I could inadvertently contaminate these with my own DNA and this could give misleading results.
3. I therefore volunteer to provide a *buccal/saliva* sample for DNA profiling, and I agree to this profile being held on the *Laboratory/Police/Medical/Manufacturers /Central (select as appropriate)* Elimination Database.
4. I also agree to this database being used by the *Laboratory/Police/Medical/Manufacturers /Central (select as appropriate)* Elimination Database administrators and authorised forensic science providers to check against profiles generated for intelligence or evidential purposes for contamination, where I have had an opportunity to cause contamination, prior to and/or after their being loaded on to the National DNA Database® or used for casework reporting purposes.
5. I understand that routinely this will involve the checking of each profile generated for criminal justice purposes against relevant staff, scene attendee, contractor and visitor profiles from site or sites where the item, or sample derived from it, or consumables used in the processing of the sample, have been handled.

6. I understand that should I withdraw my consent, my profile will be removed and destroyed six[12] months after I cease to pose a contamination risk as defined in paragraph 1 above: it will not be transferred to an archive database and I will be notified of its destruction in writing.

7. I attach the following conditions to my agreement.

    a. My DNA profile must not be used for any other purpose than for the detection of accidental contamination.

    b. Should my DNA profile match that of a sample from a scene of crime, this will have to be disclosed to the Investigating Officer, who will assume it to be the result of contamination if this is a reasonable explanation.

    c. Access to information to link my DNA profile with me must be on a strict need to know basis, and all reasonable steps must be taken to eliminate any adventitious match with my DNA profile by analysis at additional loci.

    d. Once I cease to pose a contamination risk as defined in paragraph 1 above, my profile shall either be (*a) permanently deleted, or (b) transferred to a secure archive restricted for cold cases, appeals and judicial reviews (delete as appropriate).*

    e. Should the *Laboratory/Police/Medical/Manufacturers /Central (select as appropriate)* Elimination Database cease to operate, then my DNA profile should be (a) *transferred to another approved elimination database/authorised archive, or (b) removed and destroyed (select as appropriate),* and I will be notified of this in writing.

Donor (name) _____Witness[13] (name) _____

Signature _____ Signature _____

Date _____ Date _____

[12] Retention time is dependent on the role, access, risk and processing timescales for submission and analysis.

[13] Witness is anyone confirming the continuity of the sample taken against the details of the individual, this is usually someone involved in the sample collection or work.

26. REVIEW

26.1.1 This document is subject to review at regular intervals.

26.1.2 If you have any comments please send them to the address as set out on the Internet site at: www.gov.uk/government/organisations/forensic-science-regulator or email: FSREnquiries@homeoffice.gsi.gov.uk

27. ACKNOWLEDGEMENTS

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f. Key Forensics;
g. LGC Forensics;
h. National DNA Database® Unit;
i. Orchid Cellmark Forensic Services;
j. Scottish Police Services Authority.[14]

28. REFERENCES

Forensic Science Regulator (2014) Codes of Practice and Conduct for Forensic Science Providers and Practitioners in the Criminal Justice System. London: Home Office, Office of the Forensic Science Regulator. Available at: https://www.gov.uk/government/collections/forensic-science-providers-codes-of-practice-and-conduct [Last accessed September 2014].

ISO/IEC 17025:2005 General Requirements for the Competence of Testing and Calibration Laboratories.

[14] The Scottish Police Services Authority has now become part of the Scottish Police Authority.

PAS 377:2012 *Specification for consumables used in the collection, preservation and processing of material for forensic analysis: Requirements for product, manufacturing and forensic kit assembly*.

29. ABBREVIATIONS

Abbreviation	Meaning
ACPO	Association of Chief Police Officers of England, Wales and Northern Ireland
BS	British Standard
CED	Central Elimination Database
CJS	criminal justice system
CODIS	Combined DNA Index System: the USA national DNA Database
CPIA	Criminal Procedure and Investigations Act 1996
CPS	Crown Prosecution Service
CSI	Crime Scene Investigator
DNA	Deoxyribonucleic Acid
EN	European Standards
ENFSI	European Network of Forensic Science Institutes
ESS	European Standard Set of Loci
FSP	forensic science provider
HR	Human Resources
ICO	Information Commissioner's Office
IEC	International Electrotechnical Commission
IO	Investigating Officer
ISO	International Organisation for Standardization: A network of the national standards institutes of 157 countries

LED	Laboratory Elimination Database
MED	Manufacturers Elimination Database
MedExD	Medical Examiners Elimination Database
NAS	National DNA Database® Assurance Service
NDNAD	National DNA Database®
PACE	Police and Criminal Evidence Act 1984
PAS	publicly available specification
PCR	polymerase chain reaction
PED	Police Elimination Database
QA	quality assurance
QC	quality control
SARC	Sexual Assault Referral Centre
SIO	Senior Investigating Officer
SOC	scene of crime
SPOC	single point of contact
SSM	Scientific Support Manager
STR	short tandem repeat

30. GLOSSARY

Crime sample: An item or sub-item recovered and believed to provide evidence to investigate or prosecute a criminal offence, i.e. crime-related.

DNA contamination: The introduction of DNA, or biological material containing DNA, to an exhibit during or after its recovery from the scene of a crime or a person.

Data controller: A person who (either alone or jointly or in common with other persons) determines the purposes for which and the manner in which any personal data are, or are to be, processed.

Data processor: Any person (other than an employee of the data controller) who processes the personal data on behalf of the data controller.

DNA-sensitive area: Area in which appropriate DNA contamination prevention measures shall be maintained at all times.

Elimination database: Collection of DNA profiles held in a searchable format from staff whose access/role/activities are deemed to be a potential DNA contamination risk. The profiles are used to identify instances of inadvertent contamination.

Forensic science provider: Organisation that undertakes any part of the DNA sample recovery and analytical process on behalf of the police or other criminal justice system customers, police evidence recovery labs are also included.

Human DNA-free: Human DNA is not detectable by the most sensitive DNA profiling techniques currently in use.

Partial profile: An incomplete profile obtained from the profiling system used.

Police and Criminal Evidence Act 1984 samples : Reference DNA samples taken under the provisions of the Police and Criminal Evidence Act 1984 (PACE) and accompanying codes of practice, that provide the core framework of police powers and safeguards around stop and search, arrest, detention, investigation, identification and interviewing detainees.

The Prüm Treaty: The Prüm Treaty is an international police co-operation agreement signed by Austria, Belgium, France, Germany, Luxembourg, the Netherlands and Spain on 27 May 2005, which has now become part of the legislative framework of the EU. The agreement involves police co-operation and information exchange on DNA profiles, fingerprints and vehicle number-plates.

Reference sample: A biological sample obtained from a known person with the purpose of creating a DNA profile for comparison.

Unsourced contaminant: A DNA profile identified as a contaminant i.e. following all relevant elimination database checks of which the source has not been identified. No template (negative) controls and quality control batch tests are considered as having originated from the manufacturing supply chain, historically most have been found to come from manufacturing staff.

31. FURTHER READING

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The Forensic Science Regulator

5 St Philip's Place

Colmore Row

Birmingham

B3 2PW

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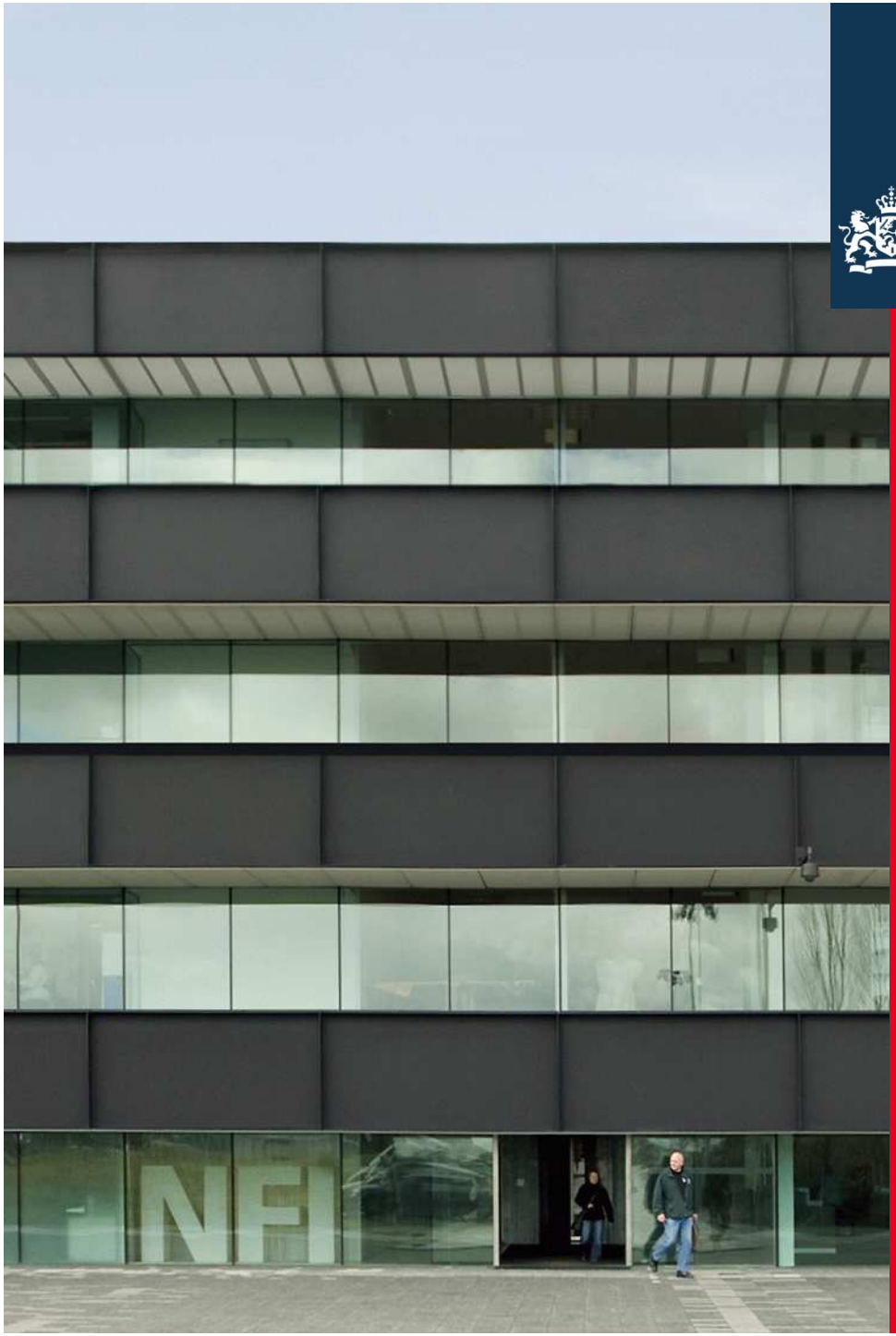


Netherlands Forensic Institute
Ministry of Security and Justice

A SNaPshot targeting common mtDNA mutations

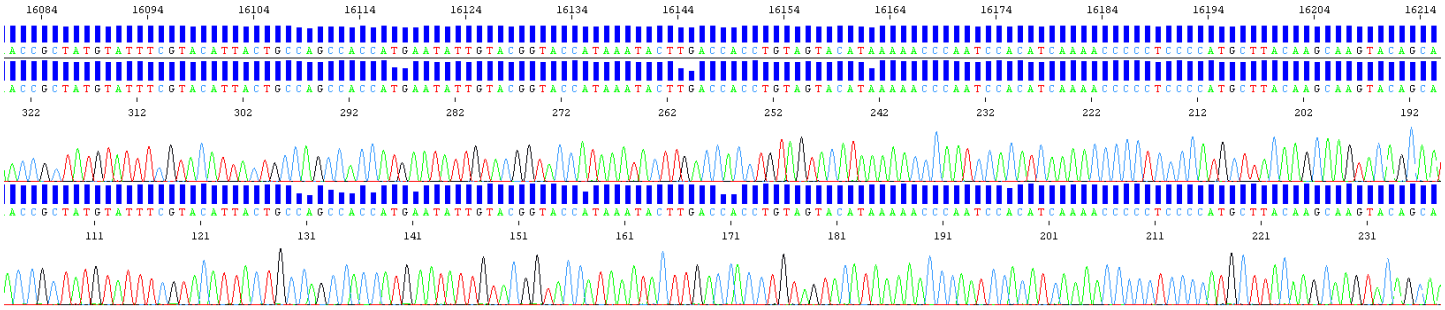
19 November 2014

Current method is time consuming

Mini-mtDNA method: 10 amplicons in 2 multiplexes

Sequencing reaction: 10x forward + 10x reverse = 20 sequencing reactions for 1 sample

- Time consuming
- Labour intensive
- Expensive
- Example: Case with 30 hairs → 600 sequencing reactions! (2011.09.15.067)



SNP analysis for mtDNA screening

Need for a quicker examination of mtDNA samples

- Selection of mtDNA samples for sequencing analysis
- Increasing sample throughput

Chemale et al. (2013) published a mtDNA screening tool

- SNaPshot assay targeting common SNPs in mtDNA HVS fragments
- Feasibility for degraded DNA?
- Focus on Brazilian population

Aim of project

Develop and optimise a SNaPshot assay targeting common mtDNA mutations in HVS fragments relevant to the Dutch Criminal casework, reflecting the individuals present in the National DNA database

Project carried out by:

Titia

Natalie

Gerda

Same PCR product for SNaPshot and mini-mtDNA

<u>Mini-mtDNA</u>

DNA (*5µl*)

↓

2x 5-plex PCR amplification (*50µl*)

↓

Purification of PCR product

↓

Sequencing PCR

↓

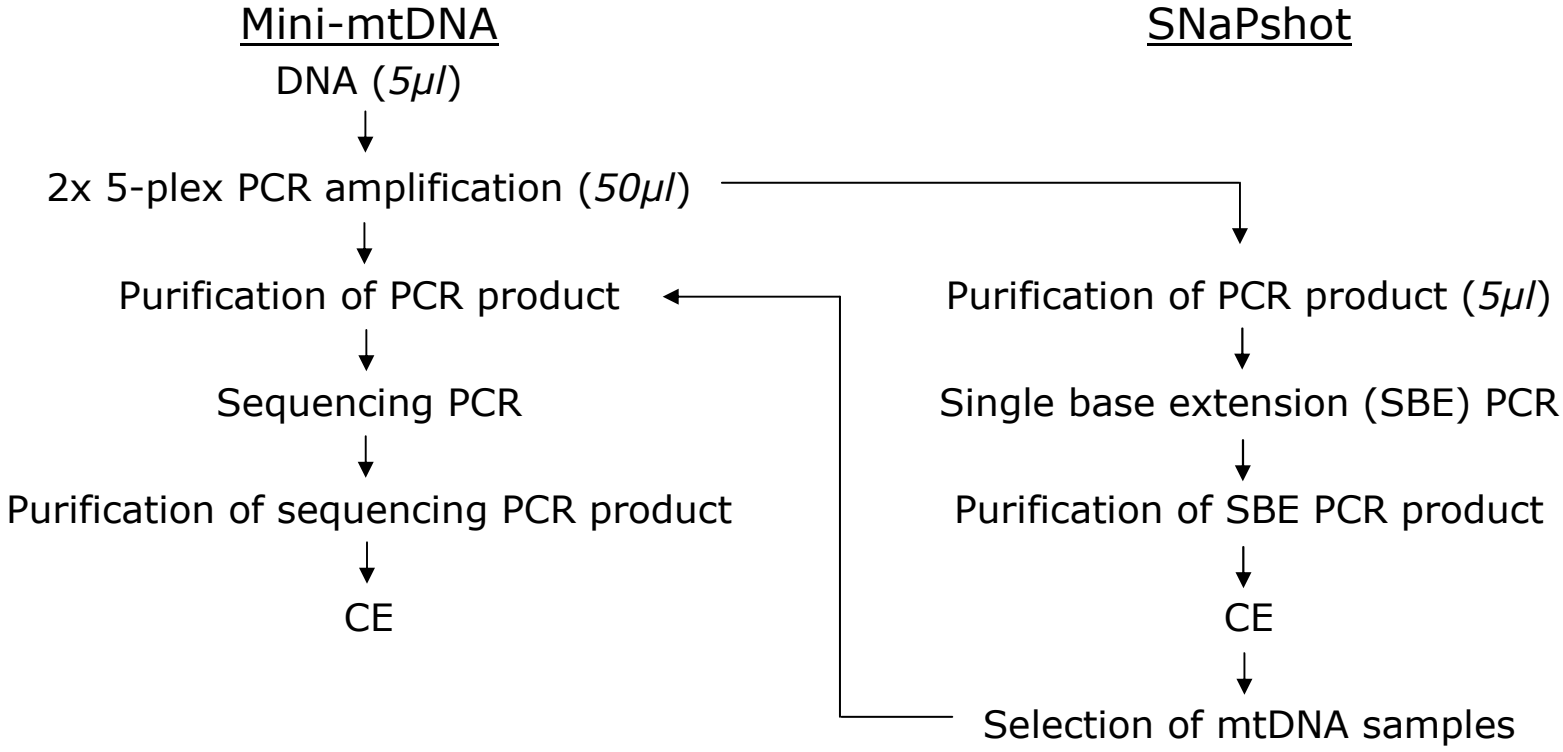
Purification of sequencing PCR product

↓

CE

Example: Case with 30 hairs → 600 sequencing reactions! (2011.09.15.067)

Same PCR product for SNaPshot and mini-mtDNA



Example: Case with 30 hairs → 600 sequencing reactions! (2011.09.15.067)

SNaPshot: Selection of 3 hair samples → 60 sequencing reactions

SNP selection

Selection criteria:

1. HVS fragments (mini-mtDNA)
2. Limited number of SNPs
3. High discrimination power
4. Haplogroup information
5. Non redundant SNPs
6. SNPs with high and low frequency in Dutch population

Final selection: 18 SNPs

Divided in two multiplex systems

-mp1: 9 SNPs (set1 mini-mtDNA)

-mp2: 9 SNPs (set2 mini-mtDNA)

SNP	Base change	Frequency	Haplogroup
73	A>G	0.5483	HV, H, V
146	T>C	0.0917	
	T>a	0.0001	
150	C>T	0.1023	
	C>g	0.0001	
152	T>C	0.2018	
182	C>T	0.0089	
185	G>A	0.0548	
	G>t	0.0031	
	G>c	0.0004	
195	T>C	0.1963	
	T>a	0.0002	
489	T>C	0.1091	M / J
497	C>T	0.0434	K
16126	T>C	0.1821	
16129	G>A	0.0662	
	G>c	0.0112	
16223	C>T	0.1285	
16270	C>T	0.0891	
16278	C>T	0.0657	
16294	C>T	0.1077	
	C>a	0.0003	
	C>g	0.0002	
16311	T>C	0.1692	
16362	T>C	0.0700	
16519	T>C	0.6441	

High power of discrimination for mtDNA

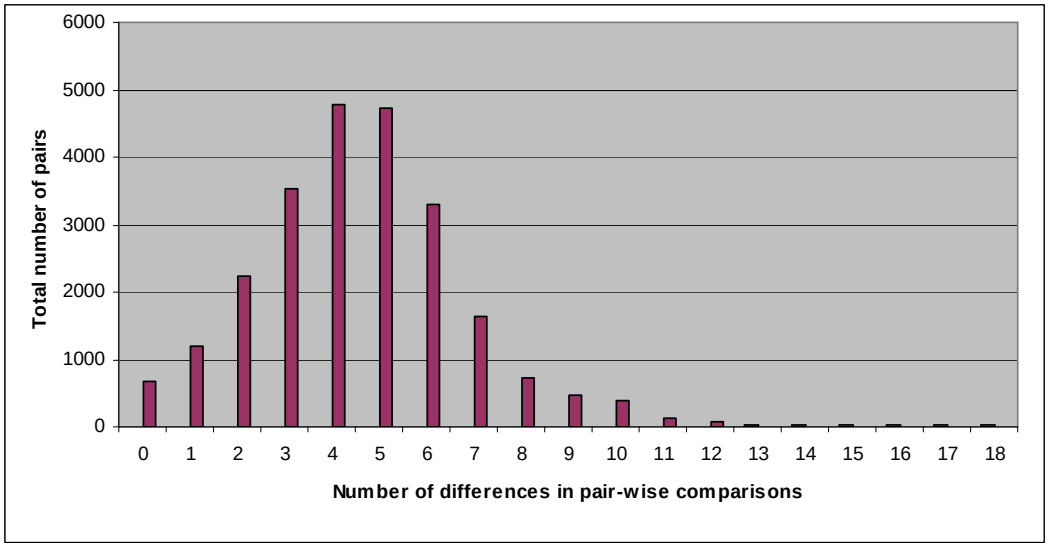
Power of the SNaPshot assay to discriminate mtDNA samples using the 18 SNPs selected

- Pair-wise comparisons between sequence data of 155 unrelated samples from NFI elimination dataset

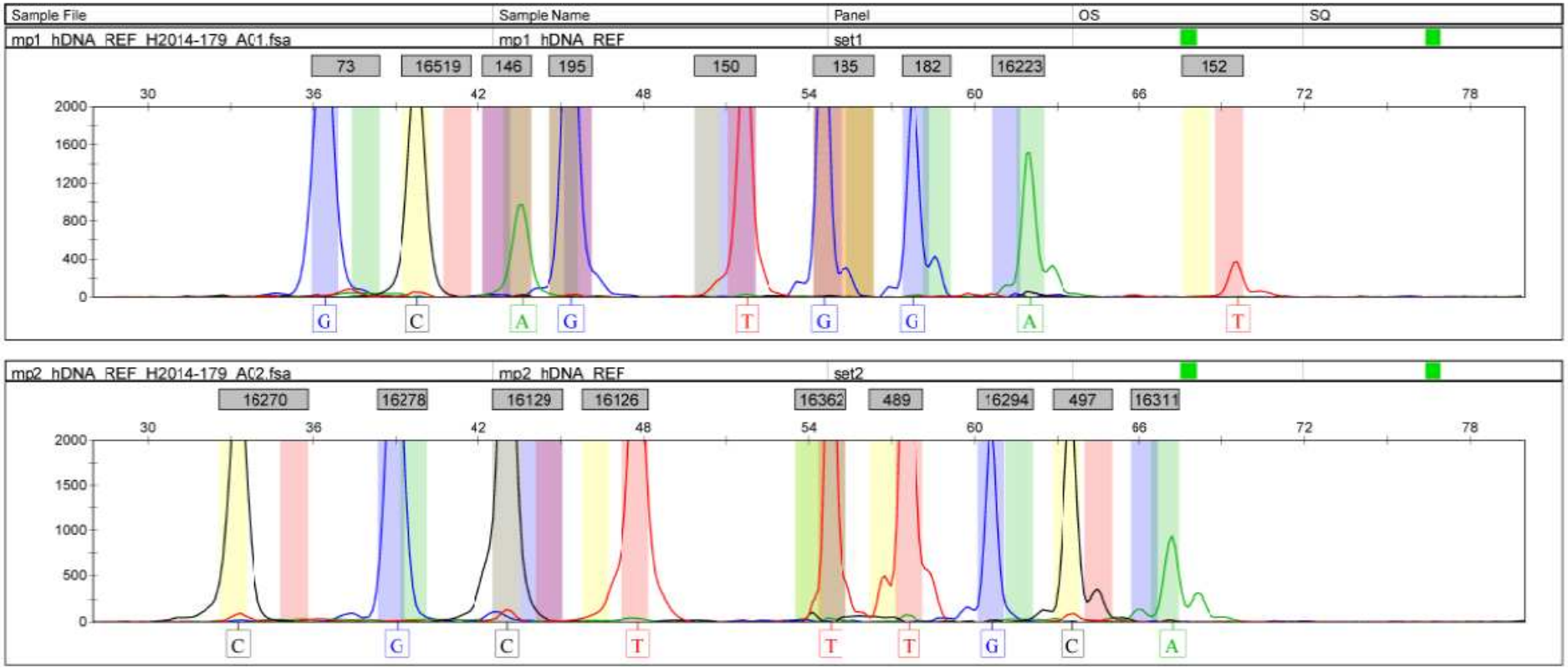
- Number of differences:
  0 – 15

Power of discrimination:

> 97.2%

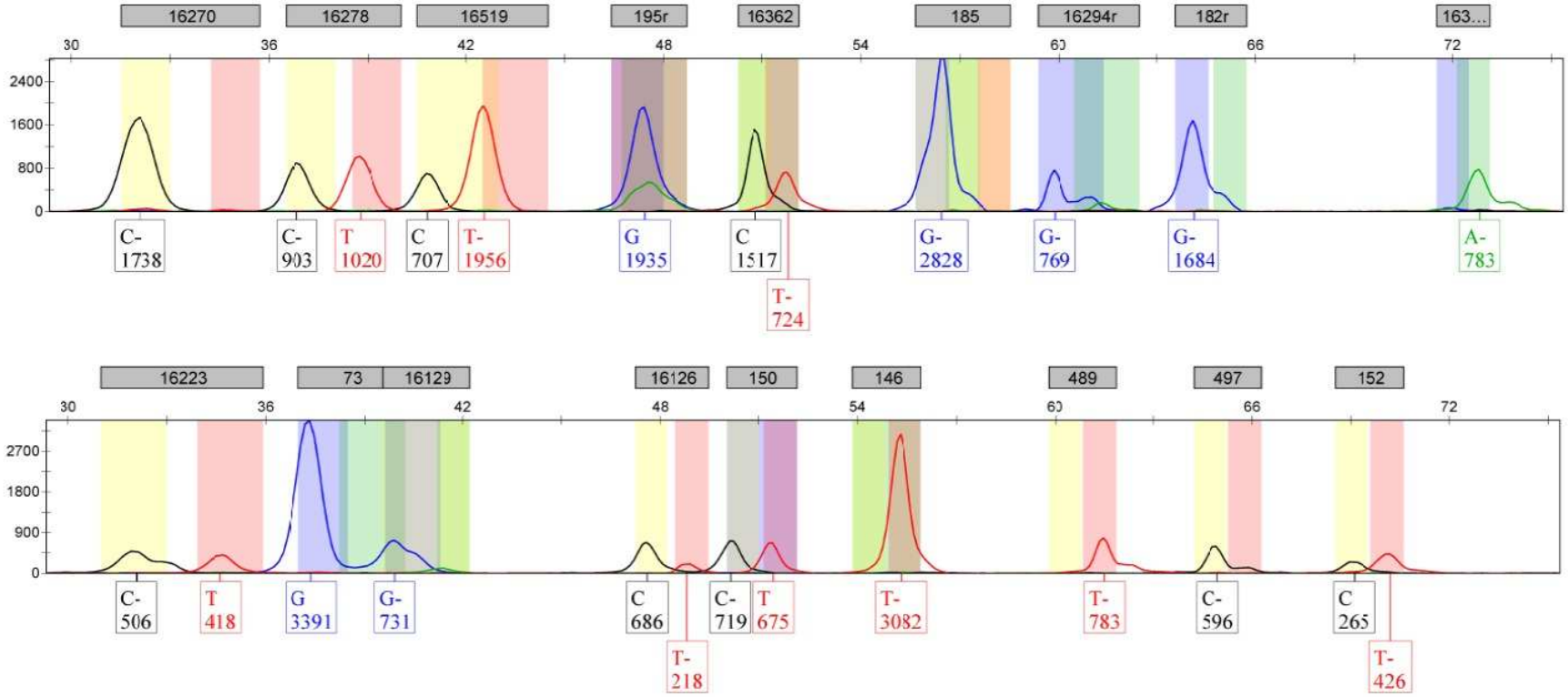


Optimised SNaPshot assay



Note: some extension primers have degenerate bases

SNaPshot of mixture



Optimised SNaPshot assay, summary

Reproducibility	✓
Inhibited samples	✓
Mixtures	✓
Concordance	✓
Manuscript	in preparation

Conclusion

MtDNA SNaPshot is a fast and efficient screening tool to discriminate mtDNA samples and facilitates the selection of samples for subsequent mtDNA sequencing

MtDNA SNaPshot can be incorporated into the existing workflow

MtDNA SNaPshot does not consume extra DNA extract

EDNAP Exercise proposal

Excercise on 10 samples (10 x SNaPshot, 2 x Sanger)

NFI provides:

- Protocols
- Primers
- Samples

Labs provide:

- All other chemistry

2015 Q1: start
2015 Q2: data collection
2015 Q3: data analysis, preparation of manuscript

Interested in joining the EDNAP excercise?

Contact:

Titia Sijen
T.sijen@nfi.minvenj.nl

Arnoud Kal
A.kal@nfi.minvenj.nl