EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

Brussels, Belgium

6 April 2011

Host: Tom Heylen/Fabrice Noël
Chairman: Niels Morling.

A list of participants is attached.

Welcome
Tom Heylen welcomed members to Kiev.

Update on exercises and other activities
mRNA exercises C Haas/J Ballantyne
The manuscript of the 2nd exercise on mRNA was recently accepted by FSI: Genetics.

mRNA exercise no 3: Cordula Haas presented the results obtained in the laboratories (attachment).

There is uncertainty in some laboratories concerning the reason for reduced sensitivity and other problems. Therefore, the group accepted an offer from Cordula Haas for an exercise with the two best markers for semen and the two best markers for saliva. Singleplex primers and samples will be sent from Zürich to the participating laboratories.

Cordula Haas and Jack Ballantyne will try to identify the best markers for menstrual blood, epithelial cells and vaginal secretion/cells for the following exercise.

Updates from other groups
EMPOP Walther Parson
Walther Parson gave an update on EMPOP. Some new developments are compiled in the recently published special issue of *Forensic Science International Genetics* **Haploid DNA markers in Forensic Genetics** (Volume 5, Issue 2, Pages 77-154; March 2011). This includes i) the new EMPOP search engine[1] that is immune to alignment and therefore guarantees that a matching sequence in the database is found regardless of annotations, ii) the provision of a new west Eurasian filter file for quasi-median network analysis supporting more sensitive data quality control[2], iii) the report on the GHEP-EMPOP collaborations that resulted in the expansion of the EMPOP database and significantly improved success rates in GHEP mtDNA proficiency testing[3], iv) a discussion on the detection and exclusion of closely maternally related samples in mtDNA datasets to avoid bias due to convenience sampling[4], and v) the evaluation of length heteroplasmy detection with direct Sanger sequencing and amplicon size analysis[5].

The EMPOP project was funded by the Austrian Science Fund for a period of 3 years (FWF Translational Research L397; Nov. 2007 - Nov. 2010). This project was extremely successful which is not only evident by numerous publications (19 peer reviewed original articles) and lecture invitations to international conferences; within this period the EMPOP database advanced to the prime mover of quality control in mtDNA research and its forensic

application. Some of the striking achievements are a recent declaration of the German Federal Court of Justice in favour of EMPOP's court acceptance, its explicit endorsement by the International Society for Forensic Genetics, its central role for quality control of mtDNA datasets prior to peer-review submission to the two leading forensic journals and its intensive collaborative interaction with academic and governmental organisations around the globe. Numerous continuing collaborations have meanwhile started and a new project application has been granted by the National Institute of Justice to support ongoing research for another 5 years.

References:

[1] Röck A, Irwin J, Dür A, Parsons T, Parson W (2011) SAM: String-based sequence search algorithm for mitochondrial DNA database queries. Forensic Sci Int Genet 5: 126-132

[2] Zimmermann B, Röck A, Huber G, Krämer T, Schneider PM, Parson W (2011) Application of a west Eurasian-specific filter for quasi-median network analysis: Sharpening the blade for mtDNA error detection. Forensic Sci Int Genet 5: 133-137

[3] Prieto L, Zimmermann B, Goios A, Rodriguez-Monge A, Paneto GG, Alves C, Alonso A, Fridman C, Cardoso S, Lima G, Anjos MJ, Whittle MR, Montesino M, Cicarelli RM, Rocha AM, Albarran C, de Pancorbo MM, Pinheiro MF, Carvalho M, Sumita DR, Parson W (2011) The GHEP-EMPOP collaboration on mtDNA population data - A new resource for forensic casework. Forensic Sci Int Genet 5: 146-151

[4] Bodner M, Irwin JA, Coble MD, Parson W (2011a) Inspecting close maternal relatedness: Towards better mtDNA population samples in forensic databases. Forensic Sci Int Genet 5: 138-141

[5] Berger C, Hatzer-Grubwieser P, Hohoff C, Parson W (2011) Evaluating sequence-derived mtDNA length heteroplasmy by amplicon size analysis. Forensic Sci Int Genet 5: 142-145.

ENFSI DNA Working Group Ingo Bastisch

Ingo Bastisch reported on the activities of the various subgroups (presentation attached). The issue concerning the weight of the evidence of a match in a crime DNA database was discussed. The German Stain Commission has recently published recommendations in the German language. The recommendations have caused some debate.

ISFG Congress 2011 in Vienna Niels Morling

Members are encouraged to communicate the congress on 29 August - 3 September 2011, and the educational workshops to be held on 28 - 29 August 2011. The workshop 'Interpretation of complex STR results' will be an advanced course for experienced colleagues.

ISFG Commission on DNA of limited quality and/or quantity Peter Schneider

Nothing new since the last meeting.

ISFG Commission of Non-Human DNA Typing in Criminal Investigations Adrian Linacre

Adrian Linacre gave an overview of the background and recommendations agreed upon by the members of the commission (summary attached). The recommendations are in press in FSI: Genetics (preprint attached).

NIST John Butler

John Butler updated on the activities of the NIST group (presentation attached).

Australia and New Zealand John Scheffer

John Scheffer briefly mentioned that the lab in Victoria is preparing the upgrade to new kits, the work on interpretation and biostatistics, the exploration of expert systems for interpretation of STR profiles, standardisation of methods, work on species identification, problems related to secondary transfer of DNA and the surveillance of the efficiency of the forensic service.

University of Central Florida Jack Ballantyne

Jack Ballantyne presented the research activities of the group (presentation attached).

Thousand Genomes Project Chris Phillips

The major interim data release of late 2010 (629 complete genomes), an open access browser developed at Santiago and potential forensic applications.

The USC 1000 Genomes browser, ENGINES, is available at http://spsmart.cesga.es/engines.php

The 1000 Genomes project was briefly outlined: Pilot 1 comprises low coverage whole genome sequencing (2-6x) of 270 individuals from three populations, now expanded to 629 from 12 populations; Pilot 2 comprises high coverage (>60x) sequencing of an African trio and a European trio, across multiple platforms and centres. There is a browser for the six Pilot 2 genomes, but no browser exists for Pilot 1 to allow users to explore SNP variation in different populations. We have developed an open-access browser termed ENGINES (ENtire Genomes INterface for Exploring SNPs) to allow users to find established and new SNP sites in defined segments or genes, or by submitting a list of rs-numbers - allowing a user to browse positions, allele frequencies and other genome details of a variant or list of variants.

Some potential forensic applications were demonstrated. These include: exploring forensic SNPs (e.g. SNPforID or Kiddlab loci) where HapMap data was not previously available, exploring low frequency SNPs in genes of interest (e.g. MC1R) and exploring SNPs that might interfere with primer binding sites that were previously uncharted or confined to particular populations. 1000 Genomes also presents a good opportunity to catalogue SNPs in STR repeat units or in primer binding sites. Unfortunately the short read limitations of high-throughput next-gen sequencing means all STR repeat unit SNPs have been removed from 1000 Genomes as unreliable calls and furthermore novel discoveries such as tri-allelic SNPs - only detectable by re-sequencing have also been considered to be spurious genome features and are now excluded. STRs as part of the CNV catalogue are due to be curated and released in 2011.

We aim to catalogue primer binding site SNP variability and to track this growing SNP repository to help pre-empt problems with allele dropout or kit discordance seen in certain loci in certain population groups that continue to arise in routine use of STRs.

Capillary electrophoresis with AB sequencers John Butler

John Butler discussed the challenges that the forensic genetic society meets due to the monopoly of AB of production of multicolour fluorescence DNA sequencers, capillaries, etc.

Reassessment of the NIST D12-vWA linkage disequilibrium data John Butler

John Butler presented the reanalysis of the NIST data. The first conclusion was that there was statistically significant linkage disequilibrium between D12 and vWA. In collaboration with two other groups, the data was reanalyzed and the conclusions were that the alleles of D12 and vWA are without significant linkage disequilibrium. The problem with the first analysis seems to be due to (1) inclusion of the paternal haplotypes twice and (2) uncertainty of some paternal haplotypes. The second set of analyses included (1) only one paternal haplotype per father-son pair and (2) either exclusion of uncertain haplotypes or maximum likelihood estimates of the uncertain paternal haplotypes.

A simulation study demonstrates that potential linkage disequilibrium between vWA and D12 loci has negligible significance Peter Gill

Peter Gill presentated a study of linkage disequilibrium by means of simulations (attached).

Development of open source (Forensim) software to interprete low-template DNA profiles – a discussion on the way forward Hinda Haned/Peter Gill

Hinda Haned explained the philosophy and principles of open source software, R and the software Forensim. Peter Gill informed about the software developing part of the EU project EUFORFORGEN.

EU Funding Peter Schneider

An application for a network of excellence has been submitted in December 2010 by a consortium of 12 partners from eight EU countries (led by Peter Schneider and Angel Carracedo) in response to a call for proposals in the framework of the SECURITY programme. The proposal has received a positive review, and an invitation for contract negotiations is expected to arrive within the next 2-3 months. If negotiations can be completed successfully, further details will be given at the next EDNAP meeting in October 2011.

EDNAP web site update (www.isfg.org/EDNAP) Peter Schneider

New content is being added about current and past EDNAP meetings. In addition to that, more information with links to relevant websites has been added to the ISFG "Software Development" website http://www.isfg.org/Software.

Any other business

There was no other business.

Next EDNAP meeting

The next EDNAP meeting will most likely be held in October 2011 in Athens in conjunction with the next ENFSI DNA Working Group Meeting.

Closing of the meeting

The meeting closed with sincere thanks to Tom Heylen and Fabrice Noël and colleagues.

Attachments

- List of participants
- Presentations by
  - Cordula Haas/Jack Ballantyne: mRNA exercises no 3
  - Walther Parsons: EMPOP report
  - Ingo Bastisch: ENFSI report

- John Butler: Update from NIST
- John Scheffer: Australia/New Zealand update
- Jack Ballantyne: Update on research at the National Center for Forensic Sciences
- John Butler: Reanalysis of possible linkage disequilibrium of D12-vWA
- Peter Gill: Simulations of linkage disequilibrium
- Adrian Linacre: Non Human DNA Commission: Summary and preprint
- Hinda Haned: Open Source
- Peter Gill: Open Source