EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

BLED, SLOVENIA

26 – 27 APRIL 2006

Host: Aleksander Regent
Chairman: Niels Morling

A list of participants is attached.

Welcome
Aleksander Regent welcomed members to Bled.

Update on publications in press Niels Morling
Two publications are available as electronic publications on the web:

Gill P, Fereday L, Morling N, Schneider PM. New multiplexes for Europe-Amendments and clarification of strategic development. Forensic Sci Int. 2006 Jan 16; [Epub ahead of print].

Dixon LA, Dobbins AE, Pulker HK, Butler JM, Vallone PM, Coble MD, Parson W, Berger B, Grubwieser P, Mogensen HS, Morling N, Nielsen K, Sanchez JJ, Petkovski E, Carracedo A, Sanchez-Diz P, Ramos-Luis E, Brion M, Irwin JA, Just RS, Loreille O, Parsons TJ, Syndercombe-Court D, Schmitter H, Stradmann-Bellinghausen B, Bender K, Gill P. Analysis of artificially degraded DNA using STRs and SNPs-results of a collaborative European (EDNAP) exercise. Forensic Sci Int. 2005 Dec 9; [Epub ahead of print].

Update on other activities
AB mini-STRs – verification test update Helle Smidt Mogensen
Walther Parson
Before ISFG/EDNAP/ENFSI had published the recommendations on miniSTRs, AB had designed an adjunct miniSTR kit with 8 STRs from the Identifiler kit + amelogenin. The purpose of the kit was to overcome problems with degradation and inhibition. AB selected the markers because they are the most likely Identifiler STRs not to give results with degraded DNA. The fragment lengths of the STRs are reduced compared to those in Identifiler, and a new, enhanced buffer is used. The markers are:

Amelogenin
D2S1338
D7S820
D13S317

D16S539
D18S51
D21S11
CSF1PO
FGA.

The recommended number of PCR cycles is 30 compared to 28 for SGM Plus and Identifiler.

Innsbruck has tested two versions of the AB miniSTR kit and Copenhagen has tested the later version. The results obtained in both labs with the last version of the kit showed that the miniSTR kit gives more results with partly degrade DNA than SGM Plus and Identifiler and that the miniSTR is less sensitive to inhibitors than SGM Plus and Identifiler.

The AB miniSTR kit is not in accordance with the recommendations offered by the ISFG/EDNAP/ENFSI. EDNAP suggest that the principles of the new buffer is used for the existing kits, including SGM Plus.

Selection of more 'standard' STRs Peter Gill

The 7 common Interpol STRs are:

D3S1358
D8S1179
D18S51
D21S11
HUMFGA/FIBRA
HUMVWA
HUMTH01

In two joint papers from ISFG/EDNAP/ENFSI, ways forward were suggested, including the use of:

Group I miniSTRs:	Group II minSTRs
D2S441 (replacing D14S1434)	D12S391
D10S1248	D1S1656
D22S1045	TPOX

Due to limited polymorphism, HumTPOX is not the best choice, but it is in general use in the forensic community.

EDNAP members realize that the development of kits that fulfil all wishes is unrealistic to take place within the first years. Therefore, realistic priorities must be found. No final conclusion is presently offered, and EDNAP members realize that the final decision is up to the producers. Suggestions included:

- SGM Plus as it is + 3 miniSTRs from group 1 (likely time frame: 18 – 24 months)
- mini-SGM Plus + 5 miniSTRs
- 5 miniSTR + 3 Interpol loci

During the joint meeting between EDNAP and the ENFSI Philosophy Group, it was stressed that ISFG/EDNAP/ENFSI has expressed the need for at least 3 new miniSTR loci, and that

the 7 Interpol STR loci must be typed with the most sensitive methods. No decision concerning the precise loci was made. Members will discuss the options in details in an email discussion group that will be initiated by Peter Gill.

ISFG/EDNAP/ENFSI has offered manufacturers to help with validation of new miniSTRs, and the EDNAP group feels that it should be done on voluntary basis.

During the joint meeting between EDNAP and the ENFSI Philosophy Group, a total of 15 EDNAP and ENFSI laboratories expressed interest in taking part in a validating exercise.

Update on exercises

EMPOP Walther Parson

Walther Parson presented an update of the EMPOP database project. A new software set that allows quality checks on mtDNA datasets has been implemented. The software set includes:

Haplogroup-ID: A software that allows for the identification of errors at haplogroup-diagnostic positions.

Phylocheck: highlights arteficial recombinants between wrongly assembled HVS-I and HVS-II segments.

Strong compatibility: A new software that is capable of finding errors at stochastic (private) mutations in the mtDNA control region. This is achieved by filtering sites with high mutation frequencies from the dataset and use the reduced set for reporting a network. Reticulations in the network indicate parallel mutations and/or errors in the dataset that can be checked on an individual basis.

The software set helps to improve the quality of data extracted from the literature (to be loaded onto the database). It has also been successfully applied to check the correctness of mtDNA data in the course of the reviewing process.

SNPforID SNP exercise Niels Morling

Kits for (1) amplification of the 52 autosomal SNPs and (2) single base extension of 29 of the SNPs were sent to 12 laboratories together with a macro for the analysis of the results. A total of 9 laboratories have returned results in form of phenotypes. Considering that this is a complicated kit that has not been through a commercial customization, the results are good. However, a number of discrepancies were observed.

Participants are encouraged to send the original Genotyper files to Copenhagen where the results will be checked and the laboratories will be asked to go through discrepant results. Clerical errors are to be expected because the process included a large amount of human interpretations and transfers of information done by hand. Copenhagen has established rules for the interpretation of results obtained in the Copenhagen lab. Similar rules must be established in each laboratory. In order to help with this process, Copenhagen will collate the results and, if possible, establish general, basic rules for interpretation that can be used as the starting point for local implementation of SNP interpretation rules.

During the discussion, a number of points were raised, including the need for automation in order to avoid clerical errors, rules for calling of heterozygotes and homozygotes (accepting that some results will be categorized as unclear).

Laboratories are encouraged to send the Genotyper data files as soon as possible to Juan Sanchez in Copenhagen.

Update on planned publications

DNA mixtures interpretation Peter Gill

The DNA Commission of the ISFG has produced recommendations on the interpretation of DNA mixtures. Peter Gill gave an overview of the recommendations that have been submitted to the FSI. The manuscript has been circulated to EDNAP members as a pdf-document. The manuscript will be published together with an editorial in FSI in August 2006. The ISFG will open a discussion forum on the web site to promote discussion. Members are encouraged to involve local statisticians in discussions. The DNA Commission is kept active because it is expected that the recommendations will need to be updated within the near future.

During the ENFSI meeting, it was decided to set up an email discussion group in order to implement policies for interpretation e.g. as clarifications to ISO 17025. The email group will be initiated by Peter Gill.

Updates from other groups

ENFSI Peter Gill

ENFSI members shortly mentioned the work that is going on in the various groups, see the attached agenda from the ENFSI meeting.

SWGDAM & FBI

No member could help with update.

NIST

John Butler was unable to attend the meeting and had kindly sent a pdf-file with slides summarising the present status (attached) that was presented.

SNPforID Peter Schneider

Project status after three years of funding (2002-2005):

- Selection of SNPs completed
- Population genetic validation completed
- Assessment of technology platforms continued
- Forensic validation in progress
- Final report in preparation

Major results:

- Y-chromosomal 29-plex for major Y haplogroup assigment (Brion et al, Electrophoresis, 2005)
- Autosomal 52-plex for human identification (Sanchez et al. Electrophoresis, 2006)
- Autosomal population-specific 32-plex for prediction of the population of origin (Africans, Europeans, Asians) (Phillips et al., in prep.)
- Definition of 45 mtDNA coding SNPs for hg H* subtyping (50% of samples reveal subgroup) (Brandstätter et al., Electrophoresis, in press)

Current activities:

- Forensic validation study on 52-plex using SNaPshot typing on:
- Reference DNA samples for sensitivity testing
- Artificially and naturally degraded DNA
- Simulated casework samples
- SNP exercise among EDNAP labs
- Collaborations with companies expressing interest to use 52-plex panel for development of forensic SNP typing kit

The SNPforID website (http://www.snpforid.org) with:
- publications
- supplementary data on the 52-plex (full primer and sequence information, frequency data)
- link to the SNPforID browser (population data from SNPforID, linked with relevant dbSNP & HapMap SNP data)

Disaster Victim Identification Peter Schneider
The ISFG has made a Disaster Victim Identification (DVI) commission has been established. The first meeting will be held in the middle of May 2006.

Interpol
No member could help with update.

Future activities
New mtDNA SNP exercise Niels Morling
Walther Parson will contact Angel Carracedo, Antonio Salas and others to discuss the possibility to make a joint exercise with the two mtDNA SNP multiplexes from Innsbruck and Santiago de Compostela.

EDNAP web site update (www.isfg.org/ednap/ednap.htm) Peter Schneider
The STADNAP information is now placed at the EDNAP part of the ISFG web site. A new ISFG web site is planned.

Joint meeting with ENFSI Philosophy Group Niels Morling
The points to be discussed were identified.

After the presentation of the EDNAP website, it was clear that the EDNAP website for some time has signalled that ENFSI members can participate in EDNAP meetings.

During the EDNAP-ENFSI meeting, it was agreed that members of the ENFSI Philosophy Group, now called 'Technology and Interpretation', will be invited for future EDNAP meetings.

Any other business
Genemapper Walther Parson
The group recognises the many problems with AB Genemapper and the urgent needs for improvements. During the EDNAP-ENFSI meeting, it was agreed to set up an email discussion group initiated by Walther Parson.

During the ENFSI meeting, it was agreed with the AB European Manager, Marco Piccinini that Walther Parson and Niels Morling will inform about problems that need to be addressed.

Next meeting
The next EDNAP meeting will be held in conjunction with the next ENFSI meeting on 11- 12 September 2006 in Tallin, Estonia.

Closing of the meeting
The meeting closed with sincere thanks to Alexander Regent for hosting the meeting.

Attachments

- List of participants
- Two ISFG-EDNAP-ENFSI papers on mini-STRs and database-STRs
- ENFSI DNA WG Agenda
- NIST-update.