EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

GLASGOW 4 – 5 APRIL 2005

Host: Matin Fairley
Chairman: Niels Morling

A list of participants is attached as Annex 1.

1. Welcome
Martin Fairley welcomed members to Glasgow.

2. Update on publications Niels Morling

In late 2004, Peter Gill circulated the draft of a manuscript, 'EDNAP/ENFSI note on evolution of DNA databases'. Please submit comments to Peter Gill who will circulate an updated version.

3. Update on EDNAP exercises

3.1 mtDNA from head hair samples – II Niels Morling
Although a large amount of work has been invested in the project – latest by the Innsbruck laboratory – too much information is still lacking, and it is not possible to generate the lacking information. The group decided to close the activities.

3.2 FSS SNPs + NIST STR exercise Peter Gill
Peter Gill summarized the results of the exercise.
Reagents were sent by Peter Gill and colleagues at the FSS and by John Butler and colleagues at the NIST. In summary,

- 2 degraded blood and 2 degraded saliva samples were prepared by incubating stains in humid tubes for 2-16 weeks
- The material was sent to 11 participating laboratories for
    - conventional STR typing
    - Low Copy Number STR typing
    - typing with short STRs from NIST (miniNCO1 and miniSGM)
    - typing with the autosomal SNP package from the FSS (21 SNPs with amplicons < 150 bp, LCN sensitivity, fully validated and ready for publication)
- Results were received from 9 laboratories.

ANOVA showed significant influence of lab-id, ref-id, sample-type and degradation time.
Small DNA fragments performed better than long DNA fragments.

The miniSGM and miniNCO1 STR typing systems performed better than conventional STR typing and the FSS SNP-plex, in general. The 10 best SNPs performed almost as the short STRs.
The conclusions of the exercise were:

- Short STRs and 34 cycle PCR seems to be the best option to maximise sensitivity (note importance of minimising cycle number to avoid stochastic effects).
- EDNAP suggests to the ENFSI group that short STRs are the best way forward.
- EDNAP suggests that the miniNCO1 loci are included as additional European loci.

3.3 Telogen hair STR typing Hermann Schmitter
Herman Schmitter offered to circulate the latest updates on procedures, invited to collaboration and repeated his invitation to help with the method.

4. Update on planned publications

4.1 Further standardisation of STRs in Europe - Considerations on how to do it
Peter Gill will circulate an updated version of the draft of the manuscript, 'EDNAP/ENFSI note on evolution of DNA databases'. It is considered to submit the final manuscript as a 'letter to the editor'.

4.2 Report of the short STRs and SNPs exercise
Peter Gill will write a draft of the report and circulate it to EDNAP members.

4.3 Suggestions of new STR loci
Presently, the following 7 STRs are recommend by Interpol based on the original recommendations from EDNAP and ENFSI:

D3S1358
D8S1179
D18S51
D21S11
HUMFGA/FIBRA
HUMVWA
HUMTH01

When selecting new STR loci, EDNAP give high priority to short STRs. The miniNC01 from NIST performed well in the EDNAP exercise, and EDNAP suggests that the NIST miniNC01 be used as standard markers. The loci and lengths of the amplified DNA fragments of the miniNC01 are:

D10S1248: 83 – 123 bp
D14S1434: 70 – 98 bp
D22S1045: 76 – 109 bp.

For further information, see Coble MD, Butler JM. Characterization of new miniSTR loci to aid analysis of degraded DNA. J Forensic Sci 2005; 50: 43-53 (can be downloaded from http://www.cstl.nist.gov/div831/strbase/NISTpub.htm).

Other loci were considered as optional loci, including

D1S1656
D12S391
HumTPOX.

D1S1656 and D12S391 were successfully used in previous EDNAP exercises and are reasonably polymorphic. Due to limited polymorphism, HumTPOX is not the best choice, but it is in general use in the forensic community.

The plan is to suggest the loci to the ENFSI group. If agreed, the suggestions will be given to the manufacturers who will meet with ENFSI during the ENFSI-meeting.

It is considered to write a manuscript with suggestions of new STR loci and the considerations behind the suggestions. If the ENFSI Philosophy Group agrees, Peter Gill will write a draft of a manuscript and circulate it to EDNAP and ENFSI Philosopy Group members.

5. Updates from other groups

5.1 ISFG — Peter Schneider

5.1.1 *Nomenclature of Y-chromosome STRs*
An ad hoc established group of scientist has finished the work on recommendations on the nomenclature of STRs on the Y-chromosome, and the manuscript has been sent to the Forensic Science International.

5.1.2 *The DNA Commission*
Peter Schneider informed that the DNA Commission will convene during the ISFG meeting in September 2005 at the Azores in order to discuss the possibility of formulating recommendations on the evaluation of the weight of the evidence of DNA results from mixtures of DNA, Low Copy Number DNA results and other special situations.

5.1.3. *The Paternity Testing Commission*
The PTC held a meeting in 2004 in order to make recommendations on biostatistics in paternity and immigration testing. The group will continue the work in 2005.

5.2 ENFSI
The EDNAP members expressed that the collaboration with the ENFSI Philosophy Group is fruitful. The EDNAP members are in favour of having future EDNAP meetings in collaboration with ENFSI.

5.3 SWGDAM — Ingo Bastisch
Ingo Bastisch attended the latest SWGDAM meeting. The main subjects were

- Interpretation of autosomal and Y-chromosome STR results
- RNA investigations (age determination, tissue, etc.)
- Contamination issues, including staff databases
- Training
- Quality and accreditation issues.

5.4 NIST — Mike Coble

Mike Coble presented the status of the work at NIST. Mike Cobble's PowerPoint presentation can be seen at the NIST web site:
http://www.cstl.nist.gov/biotech/strbase//pub_pres/Coble_EDNAP_Apr2005.pdf.

5.5 EMPOP Walther Parson

Walther Parson presented an update from the EMPOP database project. The errors detected in the first collaborative exercise (Parson et al 2004, FSI), which were in agreement with the findings of *a posteriori* phylogenetic analysis of existing mtDNA datasets were addressed by redundant analysis of the data. The EMPOP database is available now on request. Please contact Walther for more details and access.

5.6 Interpol Richard Scheithauer

The implementation of a forensic DNA database has been delayed but, now, there should be no more obstacles. Interpol has bought the electronic version of the DVI System (Disaster Victim Identification), which is made by Plass Data Software (www.plass.dk). Presently, the DNA database will not hold DNA profiles from DNA mixtures nor DNA profiles of family members.
The annual DNA users' meeting will be held 14 November 2005, and the DNA Monitoring Expert Group will later this year meet in Beijing.

5.7 SNPforID Peter Schneider

A set of *29 Y-chromosomal SNPs* that identifies major Y haplogroups has been selected and a SNP multiplex created. The manuscript has been sent to Electrophoresis.

A *52 autosomal SNP set* has been developed. Multiplex amplification is performed in a single 52-plex. SNaPshot minisequencing can be performed in a 23-plex and a 29-plex reaction with double injection so that the electrophoresis and the subsequent data analysis can be done in one session. Studies using MALDI TOF MS, Nanogen electronic microarrays and spotted SBE-TAG arrays are continued.

Other SNP sets: Non-binary SNPs exhibiting 3 alleles, population-specific SNPs and SNPs with skewed allele frequency distributions have been identified.

Nanogen Capture Down Arrays: 24 SNPs (amplicons) are hybridized simultaneously to microarray with specific stabilizer (capture) probes. SNP-specific unlabelled reporter probes are hybridized together with labelled universal reporter probes. "Touch down" thermal discrimination ensures high stringency. The assay will be expanded to 50 SNPs.

Phylogeny for mtDNA haplogroup H: A total of 41 SNPs were selected to define more than 20 haplogroup H sublineages. The method is being implementated with SnapShot, SNPlex, MALDI and alternative technologies.

Exchange of information: The SNPforID consortium is bound by the EU contract to protect the intellectual property rights on knowledge produced. New knowledge will be protected by material transfer agreements for an interim period until the information has been published and, thereby, made generally available in the public domain.

Future activities will include establishing functional SNP multiplex for the various typing platforms, forensic validation using reference DNA, artificially and naturally degraded DNA, simulated casework samples, and SNP exercise among EDNAP laboratories.

6. Automation of forensic genetic investigations Herman Schmitter

Niels Morling mentioned the problems that arise if DNA analysis software is not sufficiently flexible to allow the kind of interpretation rules that are needed in a given situation and mentioned 'Genemapper' from Applied Biosystems as an example.
Hermann Schmitter mentioned the problems that arise in real life work, if too stringent interpretation rules are employed. In Thailand, US and Australian guidelines for the interpretation of STR typing results were used on degraded material saying that an STR profile cannot be combined from two electropherograms. This makes conclusions difficult if the DNA is partly degraded and, therefore, gives weak results with large DNA fragments and strong results with short DNA fragments. In such cases, two electrophoreses with (1) an usual amount of amplicon to detect short DNA fragments and (2) a high amount of amplicon to detect partly degraded, large fragments may be necessary.

7. Future activities

7.1 New exercises

7.1.1 *SNPforID SNP exercise* Niels Morling
Copenhagen offers a SNP exercise with amplification of the 52 autosomal SNP multiplex and minisequencing detection of 29 SNPs. The reagents and the instructions will be sent to laboratories after the ISFG meeting in September 2005 in the Azores. The following laboratories signed up for the exercise:

Birmingham
Copenhagen
London
Mainz/Cologne
Münster
NIST
Santiago
Strasbourg
Vanta, Finland
Wiesbaden.

7.1.2 *mtDNA SNP exercise* Walther Parson/Angel Carracedo
The mtDNA SNP set is not ready for an EDNAP exercise on mtDNA-SNP typing.

8. EDNAP website Peter Schneider
URL of the web site: www.isfg.org/ednap/ednap.htm.
Instructions and passwords for the member site can be obtained by e-mailing Peter Schneider.
Peter Schneider will update the site with a summary of activities, including the STADNAP report as pdf-file.
Please contact Peter Schneider if you have suggestions for the EDNAP web site.

9. Next meeting

The next EDNAP meeting will be held in conjunction with the next ISFG Congress September 2006 at the Azores.

10. Any other business

There was no other business and the meeting closed with sincere thanks to Martin Fairley.