EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

NICOSIA - 15 - 16 APRIL 2004

Host: Marios Coriolou Chairman: Niels Morling

A list of participants is attached as Annex 1.

1. Welcome

Marios Coriolou welcomed members to Nicosia.

2. Update on publications

2.1 mtDNA from head hair samples -1

A paper has been published:

Tully G, Barritt SM, Bender K, Brignon E, Capelli C, Dimo-Simonin N, Eichmann C, Ernst CM, Pfeiffer H, Salas A, Schneider PM and Staalstrom E, Results of a collaborative study of the EDNAP group regarding mitochondrial DNA heteroplasmy and segregation in hair shafts, Forensic Sci Int 2004; 140: 1-11.

2.2 Degraded DNA

A paper has been published:

Schneider PM, Bender K, Mayr WR, Parson W, Hoste B, Decorte R, Cordonnier J, Vanek D, Morling N, Karjalainen M, Marie-Paule Carlotti C, Sabatier M, Hohoff C, Schmitter H, Pflug W, Wenzel R, Patzelt D, Lessig R, Dobrowolski P, O'Donnell G, Garafano L, Dobosz M, De Knijff P, Mevag B, Pawlowski R, Gusmao L, Conceicao Vide M, Alonso Alonso A, Garcia Fernandez O, Sanz Nicolas P, Kihlgreen A, Bar W, Meier V, Tevssier A, Coquoz R, Brandt C, Germann U, Gill P, Hallett J, Greenhalgh M. STR analysis of artificially degraded DNAresults of a collaborative European exercise. Forensic Sci Int. 2004; 139: 123-34.

A paper with details concerning the artificial degradation of the DNA has been published: Bender K, Farfan MJ, Schneider PM. Preparation of degraded human DNA under controlled conditions. Forensic Sci Int 2004;139: 123-34.

2.3 EMPOP – quality assessment

A paper has been published:

Parson W, Brandstätter A, Alonso A, Brandt N, Brinkmann B, Carracedo A, Corach D, Froment O, Furac I, Grzybowski T, Hedberg K, Keyser Tracqui C, Kupiec T, Lutz-Bonengel S, Mevag B, Ploski R, Schmitter H, Schneider P, Syndercombe-Court D, Sørensen E, Thew H, Tully G and Scheithauer R. The EDNAP mitochondrial DNA population database (EMPOP) collaborative exercises: organisation, results and perspectives. Forensic Sci Int 2004; 139: 215-226.

Niels Morling

Niels Morling

Walther Parson

3. Update on EDNAP exercises

3.1 mtDNA from head hair samples -2Niels Morling/Walther Parson Innsbruck has sequenced a number of hairs. There are difficulties in obtaining sequences from some hairs, and hairs sent from one laboratory seem to have unexpected sequences. Walther Parson and Niels Morling will look into this.

3.2 EMPOP

The errors detected in the first collaborative exercise were in agreement with the findings of a posterior phylogenetic analysis of existing mtDNA datasets (Bandelt and Parson, Rechtsmedizin, in press). The EMPOP software package was presented including the LIMS software, Haplogroup-ID and Phylocheck, which can be used for the detection of errors in mtDNA analysis. The EMPOP database consists of more than 2,500 haplotypes now. Search engine Version 2.0 has been programmed in Innsbruck. This work will be finished soon and evaluated by the group later.

3.3 Y chromosome SNPs - Santiago II **Chris Phillips** A manuscript was circulated just before the meeting. Participants are asked to comment as son as possible.

3.4 Population database compilation The compilation is progressing but not finished.

3.5 Feedback on SNaPshot experience All participants A little more than 50 % of the laboratories have experience with SNP analysis with SNaPshot.

3.6 Telogen hair STR typing

Herman Schmitter gave an update of the results from Wiesbaden and repeated the invitation to help with the method.

4. Considerations

4.1 Contamination theory - the problems and solutions Peter Gill 4.2 Testing strategy - which test to use when -Peter Gill given limited sample size

Please see the attached summaries of the presentations.

5. Updates from other groups

5.1 ENFSI

ENFSI had invited EDNAP representatives to a meeting with the ENFSI philosophy group in order to discuss and coordinate work.

5.2 SNPforID

SNPs have been selected according to the following criteria:

- a minimum of 100 kb between a SNP and a gene (or STR)
- approximately 1 Mb distance or more between SNPs on the same chromosome
- a 0.28 lower limit for the minor allele frequency in at least one major population
- a 0.18 lower limit for the minor allele frequency in all reported populations, corresponding to a 40 % and 30 % heterozygosity value respectively

Hermann Schmitter

Chris Philips

Walther Parson

Denise Syndercombe Court

• an emphasis on informative allele frequency distributions in European population samples in preference to other populations.

Validation studies of the first 23 SNPs indicate that the allele frequency estimates reported in the online databases are a reliable guide to the actual frequencies in most cases.

Fifteen of 52 SNPs showed marked contrasts in allele frequencies in different populations. These SNPs with allele distributions that may be characteristic of certain populations are being investigated.

Please see the attached summary of Chris Phillips for further details.

5.3 SWGDAM

No SWGDAM member attended the meeting.

5.4 <u>NIST</u>

John Butler presented work done by the National Institute of Standards and Technology (NIST) including results obtained with STRs with 'short' amplicons, increased number of PCR cycles, degraded DNA, SNP typing of autosomal, Y-, and mtDNA SNPs as well as results of various methods for DNA quantitation. The results are presented in further details in the attached 'pdf'-file that can be found at the web site:

http://www.cstl.nist.gov/biotech/strbase/pub_pres/EDNAP-Cyprus.pdf.

Please notice that <u>http://www.cstl.nist.gov/</u> offers other valuable information.

5.5 Interpol

Richard Scheithauer

The DNA activities of Interpol were briefly presented. The information can be found on the website http://www.interpol.int/forensic/DNA.

6. Future activities

6.1 <u>New exercises</u>

Peter Gill will organise an exercise concerning comparison of the performance of short STRs from NIST, LCN with STR kit(s) and the autosomal SNP package from the FSS. Some logistics must be sorted out first, and the exercise will probably not be ready before the second part of 2004.

The SNPforID group plans to offer an SNP exercise in the second part of 2004.

6.2 Joint meetings with ENFSI

Members were in favour of organising future EDNAP meetings so that meetings with the ENFSI philosophy group in order to discuss and coordinate work.

7. EDNAP website update (www.isfg.org/ednap/ednap.htm)

Please contact Peter Schneider if you have suggestions for the EDNAP web site.

8. Next meeting

The next EDNAP meeting will be held in Dublin on Thursday 2 September 2004 in conjunction with the next ENFSI meeting on 3 - 4 September 2004.

9. Any other business

There was no other business and the meeting closed with sincere thanks to Elena Ioannidou and Mario Cariolou.