EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

PRAGUE, Czech Republic

22 APRIL 2008

Host: Roman Hradil.
Chairman: Peter Schneider.
A list of participants is attached.

Welcome
Roman Hradil welcomed members to Prague.
Peter Schneider excused Niels Morling who was unfortunately unable to attend the meeting. Furthermore, he announced that Peter Gill has moved from the Forensic Science Service to the Dept. of Pure and Applied Chemistry, University of Strathclyde (Glasgow). Peter proposed to invite this group as a new member into the EDNAP group. The proposal was unanimously adopted by the members.

Update on publications Peter Schneider
The manuscript about the collaborative SNPforID SNP exercise testing the 29-plex by SNaPshot analysis is in press and a proof has been produced. The paper will be published in the 3rd issue of FSI Genetics

Update on exercises and other activities
EMPOP Walther Parson
Walther Parson announced that release 2.0 of the EMPOP database will be launched at the upcoming Ancona "DNA in Forensics" meeting (May 27-30). More than 5,000 new sequences have been added. In contrast to the previous version holding 5.173 haplotypes from West Eurasian populations, version 2.0 will concentrate on haplotypes mostly from East Asia. Furthermore, new alignment tools based on phylogenetic strategies will be made available.

Collaborative exercise on AB MiniFiler Peter Gill
As agreed at the last meeting in Rome, a second round of the Minifiler exercise was carried out at the beginning of this year. A total of 7 forensic DNA samples were shipped by Copenhagen and Cologne to four participants. These samples were either heavily degraded or showed inhibition in PCR as previously demonstrated by SE-Filer or SGM-plus STR typing. The samples were tested using both the Minifiler and the SGM-plus kits using both 28 and 30 cycles, as far a sufficient sample DNA was available. Based on data from three of the four labs, the results clearly show that the Minifiler gave more complete results compared to SGM-plus regarding the total number of alleles detected. When the final results have been compiled, a short publication of these data would be appropriate.

Updates from other groups
ENFSI Ingo Bastisch
Ingo Bastisch reported on the activities of the various subgroups. A document on the establishment and management of databases is in preparation and is expected to be approved soon. National databases are operational based on the Prüm treaty in DE, AT, LU and ES. A discussion about the best approach for a combined presentation of autosomal and Y-STR results has been initiated. The DVI concept is close to being finalized. The EU will fund the

project „Improving the efficiency of European DNA data exchange“ for 3 years with 300 k€. The money will be used to support the ESS extension, database operation recommendations, and QA, as well as cooperation with other groups and organisations. The ENFSI website has been moved to www.enfsi.eu. At present, most of the content is restricted to members, but the public part of the website will be enhanced soon.
Subsequently, a discussion about possibilities for a DNA contamination database took place. At present, DNA profile data from negative controls are not collected systematically, and if so, it is done locally based on altruism, i.e. without any funding. However, the establishment of a European "unsourced profile database" would be highly desirable, not necessarily to trace back the origin (there may be privacy issues involved), but to monitor contaminations at the European level. Also, there is a need to define a forensic standard for consumables. The usefulness of establishing a European "NIST" dealing with these an other issues was recognized.

Australia Vanessa Ientile

Vanessa Ientile from the Queensland Health Scientific Services (QHSS), and working at the Forensic and Scientific Services in Brisbane, better known as the John Tonge Centre, reported about forensic DNA casework in Queensland, Australia.

ICMP Tom Parsons

Tom Parsons gave an update on the work of the ICMP laboratory in Sarajevo, which has a maximum capacity to process 105 bone samples per day. Up to now, 14,000 match reports have been issued. It is planned to continue working on cases from the former Yugoslavia until 2010. Furthermore, international collaborations have been started, e.g. with Colombia on missing persons. The ICMP will also continue to provide a training programs, and to serve as an resource center providing excess typing capacity for international casework.

NIST John Butler

A PowerPoint presentation sent by email from John Butler was shown by Peter Schneider (attached as pdf file).

ISFG Peter Schneider

On behalf of the ISFG board, Niels Morling has written a letter to William Linton, the owner of Promega, to explain the European need for new kits, and to inquire about the licensing policy regarding the STR patents held by Promega. The ISFG board hopes to get a positive reaction from Promega so that the completely dissatisfying situation regarding the development of new STR kits following the decision about new ESS loci may eventually improve.

SNPforID Ch. Phillips, P. Schneider

For this topic, Lisa Schade and Jörg Willuhn from Applied Biosystems were present as well. Chris Phillips reported about the continuation of the work carried out in Santiago de Compostela about identification and ancersy-informative SNPs. The SNP*for*ID Browser website located at http://spsmart.cesga.es/ has been greatly enhanced by adding numerous features, as well as adding search and frequency comparison functions for HapMap, Perlegen, and CEPH data in addition to the data for the 52 SNP ID and the 34 SNP AIM sets. In addition, two dedicated sets were developed – an Eurasianplex (28 SNPs) and an Americanplex (26 SNPs) – all SNaPshot assays. In Santiago, SNPs are used as supplementary markers to STRs in challenging identification cases and where single one-step exclusions are found.

Peter Schneider summarized the efforts about developing the Genplex typing assay for 48 ID SNPs plus Amelogenin in collaboration with Applied Biosystems. Following up in his presentation at the last EDNAP meeting, he described the requirements for running the Genplex assay on a CE instrument: a "31xx" 4- or 16-capillary sequencer running on data collection software 3.0 and equipped for POP-7 polymer, as well as GeneMapper 4.0 software installed on a separate PC for data analysis. Furthermore, some results from a validation study of the Genplex assay on 286 individuals from Copenhagen were presented indicating identical results for all but one genotype (of 13,156) compared to an accredited version of the SNaPshot assay (see enclosed pdf version of the presentation). Furthermore, the sensitivity of the Genplex assay was excellent and identical for sample amounts between 250 and 500 pg of DNA, and still good for 100 pg.
Lisa Schade informed the participants that AB has finally introduced the SNP*for*ID 48-plex plus Amelogenin as a commercial kit (Part number 4383547; 500 tests; US$ 1,750). The kit contains separate PCR primer sets which need to be mixed prior to use following a protocol provided in the manual, as well as specific oligo ligation reaction mix. The other components for running the assay are identical to those used for the SNPlex assay. The full protocol (version 2.0.2) for the "GenPlex™ HID System" can be downloaded from the AB website. There will only be limited support, as the assay is sold as a "custom product ... that has not undergone any in-house validation". A discussion on using the Genplex assay for a larger collaborative exercise was held later (see below).

German Stain Commission Peter Schneider

Peter Schneider announced that the English version of the Recommendations of the German Stain commission on mixture interpretation is now in press in the Intl. J. of Legal Med. A pdf version is enclosed with the minutes. Then he gave a presentation about the errors rates for STR systems commonly used for databasing in Europe, i.e. the ESS loci, the three additional STRs from the SGM plus kit, and ACTBP2 (SE33), currently only used for the German database. Data were obtained from the results of the GEDNAP exercises in the period 2002-2006 (pdf enclosed). The overall error rates were between 0.8 and 1.6%. It was found that errors are not related to a particular STR system, and that increased error rates also depend on the occurrence of unusual alleles not representing the expected allelic range, such as D18S51 17.3, D19S433 6.2, and D21S11 28.3 and 33.1, whereas the error rate for SE33, a system well known for many unusual length variants, was not particularly high. When the "rare" alleles were removed from the analysis, overall error rates were between 0.6 (THO1) and 1% (D21S11 and D3S1358). Finally, it has to be considered that during each GEDNAP exercise, there are 1-2 "newcomer" laboratories that accumulate the majority of errors across all loci. If these are disregarded, the overall error rate is further reduced.

Interpol Richard Scheithauer

Richard Scheithauer briefly introduced the scope of the Interpol DNA Monitoring Expert Group (DNA-MEG; http://www.interpol.int/Public/Forensic/dna/dnameg.asp). It has currently 10 members representing all world regions. The ISSOL (Intl. standard set of loci) will be increased in parallel with the ESS. Interpol prtovides a DNA data gateway that accepts results from all kits and all currently used loci. Matches are based on all overlapping loci between the DNA profiles, with ISSOL being the minimum no. of loci. Member states can upload any profile, including missing persons, unidentified bodies, and decide upon matching with whom, how often, and removal, but always remain the owner of the profile. Currently, 65,000 profiles are held from 47 countries. A revised version of the Interpol DVI Guide will be finalized, also based on the recently published ISFG guidelines. Furthermore, the 2nd edition of the "Interpol Handbook on DNA Data Exchange and Practice" is available as pdf file from the Interpol website.

DNA interpretation pitfalls following the Omagh judgement Peter Gill

Peter Gill shared his thoughts about the consequences following the Omagh judgement in the UK. In this case, DNA evidence was considered by the judge as not sufficiently validated both by the scientific community in general, as well as by the laboratory using it in this case. The main point was the use of "low copy number" PCR by the FSS on some of the samples – although not all evidence profiles in this case were actually obtained using LCN PCR. Subsequently, the UK Forensic Regulator had commissioned a review of this method which was recently published (circulated by Niels Morling to EDNAP members prior to the Prague meeting). Peter Gill suggested that the term "LCN" needs to be abandoned and to be replaced by a completely different descriptive approach for DNA analysis that may give rise to complex profiles exhibiting stochastic effects. They key is not the amplification protocol, since such profiles may also occur at 28 cycles, but to find an universal approach for interpretation. The presence of a profile derived from a contact trace does not tell us how it got there, and whether it is directly connected with the crime event. Bayesian networks may serve as an approach for such scenarios. However, it might be difficult to present complex reasoning in court. If the court doesn't accept (or won't listen to) a scientific explanation of the evidence then it is difficult to defend any rationale. The second issue relates to the judge's quote: "The absence of an agreed protocol for the validation of scientific techniques prior to their being admitted in court is entirely unsatisfactory", and leads to the question, what is regarded as "scientific consensus" both from the scientist's *and* from a court's perspective.

Here, the ISFG should play a more prominent role. It is proposed to have a DNA Commission explaining the process of consensus building in a scientific community which is clearly not synonymous with everyone following exactly the same protocol for a particular sample. Similarly, the ISO 17025 guidelines do not prescribe any details for a method, but provide a framework for establishing and validating the methods used in a given lab. Similar to the guidelines published by the ISFG Paternity Testing Commission, the proposed DNA Commission should provide a specific framework for forensic DNA labs on the background of the ISO 17025 guidelines. The scientific guidelines may then be adopted first at the European level, and then by national bodies to provide guidelines as the final basis for accreditation that should also take into account local requirements. The discussion was continued at the ENFSI meeting.

Future activities - *two areas were discussed for new collaborative exercises:*

1. mRNA profiling for the identification of body fluids

Cordula Haas presented data from Zürich about their work on the identification of body fluids such as peripheral blood, menstrual blood, saliva, seminal fluid and vaginal secretion with mRNA. Using primer pairs for approx. 20 genes, they were able to obtain specific signals both using endpoint and realtime PCR. Furthermore, samples can be extracted in parallel to obtain both mRNA and DNA for subsequent genetic analysis. The sensitivity was the same or better compared to conventional tests for specific detection of body fluids (blood, saliva, semen). Thus, advantages are the posibility of simultaneous DNA isolation without loss of material, that all body fluids can be detected in one multiplex PCR, and that additional body fluids or tissues could be identified using additional markers. Disadvantages are that RNA is not very stable and requires special precautions in the lab, and that the assay is more time-consuming and expensive. Nevertheless, great interest was expressed by EDNAP members to participate in a collaborative exercise which, according to Cordula Haas, could be organized during the next meeting. The following labs indicated that they would like to participate: Münster, Strasbourg, Wiesbaden, Birmingham, Lyon, Innsbruck, Brussels, Bratislava, Copenhagen, Cologne.

2. Genplex SNP typing exercise

Following earlier presentations, the feasibility of a collaborative Genplex SNP typing exercise was discussed. Due to the fact that SNP analysis is only applicable in special forensic cases where reference samples are availaible, the scope of the exercise should be extended to include also labs working in relationship and immigration testing. There are as well a number of labs in the US and in South America who might be interested. AB has indicated that they are willing to support such an exercise with reagents, as it would be a way forward to spread the technology with the help of the labs in Cologne, Copenhagen, London and Santiago who have already experience with the Genplex system. The following laboratories expressed interest: ICMP/Sarajevo, Wiesbaden, Strasbourg (both Legal Medicine and Codgene), Lyon, Zürich, Oslo, Lisbon, London, Santiago, Copenhagen, Cologne. At the upcoming Ancona meeting, further details will be discussed to develop more specific plans regarding the scope of such an exercise including the number of participants and samples to be typed to make a specific request with AB for support.

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

Peter Schneider demonstrated the new EDNAP website, which is online since the end of last year. It features extended pages on activities, publications, the history of EDNAP, and a list of all EDNAP meetings since 1988 with major topics, as well as a summary and the final report from the STADNAP network project.

Any other business

On October 16, 1988, the first meeting of a group of European DNA laboratories and a handful of companies met in Sunbury, UK. This was the birth date of the EDNAP group, an acronym which was selected at the second meeting in Münster in 1989. Thus the 20th anniversary will be celebrated at the next meeting in Zürich. It was observed that Ate Kloosterman and Hermann Schmitter are the only current members still active who were already present at the first meeting. They were asked by the group to prepare a presentation about the EDNAP activities and achievements during the last 20 years. Furthermore, Ate Kloosterman was asked to contact Peter Martin, the first EDNAP secretary, to invite him to the anniversary meeting. The current secretary was asked to contact to organizers of the Zürich meeting about the possibility to have a formal dinner with the group on this occasion.

Next meeting

The next EDNAP meeting will be held in conjunction with the next ENFSI DNA Working Grouping Meeting in Zürich– most likely 23-24 September 2008.

Closing of the meeting

The meeting closed with sincere thanks to Roman Hradil for hosting the meeting.

Attachments

- List of participants (Word)
- Genplex slides (Peter Schneider, pdf)
- Presentation on database STR error rates from GEDNAP (Peter Schneider, pdf)
- English version of the Recommendations of the German Stain commission on mixture interpretation (Int. J. Legal Med., in press, pdf)