

# EUROPEAN DNA PROFILING (EDNAP) MEETING

Oslo 2 September 1997

Host: Prof Björn Olaisen

Chairman: Dr Niels Morling

A list of participants is attached at Annex 10. Finland was not represented at the meeting.

## REPORT OF THE MEETING

Peter Schneider, on behalf of the organising committee, thanked participants who took part in the Ethics meeting at Mainz on 18/19 October 1996 and distributed reports of the meeting together with a photograph of the EDNAP members who attended.

### 1. Update on EDNAP exercises

#### *DYS385*

Peter Schneider presented the results of the exercise (see Annex 1). The results from 12 of the 14 laboratories were consistent. There was one obvious transpositional error but it is not clear where the transposition occurred. One laboratory reported some inconclusive results but stated that these were not final assignments.

The population data were very homogeneous (see Annex 2) and there appears to be a gradient of frequencies from north to south Europe. Peter Schneider considered that if this work is extended to include more Y haplotypes, it could prove valuable in the identification of markers in male stain analysis.

The discussion which followed the presentation included a number of issues concerned with future developments in Y chromosome marker analysis. While it was understood that discrimination could only be achieved between males, it was also appreciated that there could be some real value in the interpretation of results from mixtures of body fluids and in the analysis of old stains.

Angel Carracedo presented some data from allelic ladders obtained from the *DYS19*, *DYS389-1*, *DYS389-2*, *DYS390* and *DYS393* loci. The frequency of alleles from some loci are remarkably similar for all populations tested but others show considerable differences. The component alleles have been sequenced and the results show that previously published material includes inconsistencies in the nomenclature. Although there will be other scientists who are well advanced with studies of Y loci and could experience difficulty with a change in nomenclature, there could be some advantage in collaboration. EDNAP now has some valuable information concerning population studies for Y loci and it was agreed that it might be a sensible time to seek collaboration with other scientists to prepare a comprehensive publication.

Peter Schneider volunteered to contact other relevant laboratories to determine a way forward bearing in mind that questions of nomenclature and the publication of results would have to be viewed against the background of the STADNAP application.

#### *DIS1656*

Angel Carracedo presented a report of the exercise which included the results of allelic designations, a proposed structure and a sequence composition (see Annex 3). The results were correct except for two deviations associated with the existence of irregular repeats. Angel Car-

racedo suggested that the repeat nomenclature should be used although irregular repeats are common in this system. It was decided that this locus should be included in the publication, along with other loci, which is being prepared by Peter Gill.

At present no EDNAP laboratories are using this locus in casework but the Herman Schmitter informed that the BKA might well include it in future examinations.

#### *MtDNA(HVI region)*

Angel Carracedo also presented a report of the results of this exercise (see Annex 4). All participating laboratories reported the same correct results despite using different methods. It was suggested that EDNAP should collect population data for mtDNA and that this could be combined and placed on the Internet together with existing data. Angel Carracedo makes a compilation of the data for circulation.

#### *SE33/ ApoAII/ D11S554 and D12S391*

Peter Gill circulated a draft publication on the exercise concerned with the above loci (see Annex 5). Fourteen laboratories participated in the exercise and all used ABD 377 sequencers.

The best results were obtained from SE33; ApoAII showed some variation and there was greater variation in the results from D11. Although it was not possible to determine a single reason for this variation, an interesting point arose during the exercise. Inconsistent results were obtained, in collaborative work on the D11S554 locus, between the FSS and the Oslo laboratory. The source of the discrepancy was traced to variation between batches of primers; one batch had an extra base added. It is therefore recommended that all new batches of primers are checked against those in current use.

The results of the exercise demonstrated the robustness of the SE33 locus. Björn Olaisen has looked at the results from 1500 males and found no mutations and he therefore recommended the use of this locus for paternity work as well as mainstream forensic science. He considered that it would be a pity if ApoAII was not used especially as the results from Oslo and the FSS showed such good agreement.

#### *D12S391*

The results from the D12S391 exercise were updated. The results demonstrated agreement although there were a few minor problems. In some cases the 18.3 allele had been recorded as 19 and Peter Gill recommended that the window should be set at  $\pm 0.5$  bp. At the last EDNAP meeting, it was agreed that these data should be included in the report, along with the data of the other loci, which is being prepared by Peter Gill.

EDNAP members should provide comments to Peter Gill on the draft publication by 1st October 1997. The next draft will be produced in the first week of October.

## **2. Update on ENFSI activities**

Ate Kloosterman presented an account of the recent meeting in Parma, Italy of the DNA Working Group of ENFSI. Innsbruck and Copenhagen have applied for membership, and Richard Scheithauer and Niels Morling were present at the meeting. Bruce Budowle was also present.

(i) An exercise in which samples were analysed by various systems currently used by ENFSI members, produced results which were disparate and difficult to compare.

(ii) A second exercise was prepared by the FSS in which the FSS multiplex (SGM) was used together with Taq gold. Thirty five (35) laboratories were sent a set of samples, reagents and allelic ladders. Twenty six (26) replies were received and the majority of the results were correct.

(iii) The laboratories had submitted an example of a statement according to their usual procedure including:

- an explanation of the technology
- a table of results
- a DNA profile.

Inevitably, there were some differences in the way in which the reports were written and it was not clear how to progress this initiative.

(iv) ENFSI is currently pursuing an information gathering exercise to determine:

- the legal situation in each country
- how many forensic science laboratories exist
- how many samples are received etc.

(v) There is an Interpol objective to have a pan-European database of paedophiles. (This has been endorsed by senior politicians at a meeting in Stockholm.)

ENFSI, in response to an Interpol request, recommended that a standard set of loci would include - HUMTHO1, VWA, D21S11 and FGA. The 4 loci were chosen for pragmatic reasons. The loci are used by the great majority of the European laboratories and it is expected that the FBI package will contain the same loci. It will be the responsibility of ENFSI to organise and make recommendations on the management and structure of the Interpol database.

(vi) A study will be undertaken to decide on  $F_{ST}$  values to be used when calculating frequencies from the Interpol database. Data will be collected from those laboratories who achieved good results in the SGM exercise and statisticians at the FSS will calculate the relevant  $F_{ST}$  values.

(vii) A Quality Control working party will discuss the necessary requirements to ensure that error-free results are transmitted to the database.

The next ENFSI meeting will be in Lausanne on 17-20 September 1997. The agenda will include a discussion on the interpretation of mixed stain results. There will also be the first meeting of the European Academy of Forensic Scientists.

There was a debate which followed the presentation and this centered on the apparent competing roles of ENFSI and EDNAP. There is no doubt that ENFSI has a higher profile and to some extent is driven by external politics but, with the STADNAP initiative, EDNAP should achieve a more public position. Although there was obvious concern, EDNAP members who attend ENFSI meetings gave assurances that they were also keen to ensure the survival of EDNAP.

### 3. STADNAP exercise

Peter Schneider presented this agenda item. Positive signals have been received from Brussels (see Annex 6, only the first page enclosed) and there is now a framework document for EDNAP to receive funding and to start work. There is a contract of work and a requirement for milestones (see Annex 7) which must form the basis of the initiative.

With regard to the budget, the co-ordinating laboratory will receive the bulk of the money and reagents etc. will be centrally purchased and distributed to participating laboratories. Money for travel and other expenses will be allocated to member laboratories who should set up a bank account which can be audited.

A budget will be available for:

- inviting external scientists to meetings to explain points of interest
- inter-laboratory exchange of staff for training etc.
- preparation of a newsletter and an Internet website

Two major points of interest emerged:

- (i) it is essential to retain all receipts
- (ii) all allocated money must be spent

Two documents, a Thematic Network contract (Annex 8, only the first page enclosed) and a Contract of Association (Annex 9, only the first page enclosed) must be completed in triplicate and sent to Angel Carracedo (Peter Schneider has already circulated the work contract).

Angel Carracedo will send the relevant documents to members with instructions for their completion.

### Work Packages

The whole project is defined in terms of Work Packages (WP) and each WP will require a Work Package Manager (WPM). It is recommended that 4 or 5 laboratories collaborate on each WP.

### Reports and Milestones (see Annex 7)

There will be a requirement to decide the progress of the projects and to set a schedule of events to supply the deliverables. Information will be provided via a Homepage on the Internet. At the conclusion of the 3 year programme it will be necessary to make recommendations regarding any outstanding work and any further research.

Perkin-Elmer (PE) and Pharmacia are listed as consulting commercial organisations and will be expected to supply information regarding current developments within their organisations. They will need constant updates on the progress of the work undertaken by EDNAP.

The project begins 3 months after the contract is signed and, as the aim is to start early in the new year, there should be an inaugural meeting in January 1998. In the meantime members should give serious consideration to their involvement in the various work package groups. Angel Carracedo, Peter Schneider and Niels Morling will offer suggestions on the structure of each WP group and nominate some WP managers.

All laboratories should have access to e-mail.

## **4. Proposal for socio-legal research group**

Peter Schneider has been contacted by Dr Ruth McNally at Brunel University in the UK as she is interested in a research project to discover how scientists interact and network on projects throughout Europe. She hopes to obtain EU funding for this project and needs feedback from EDNAP for her grant application.

There did not appear to be any advantage to EDNAP and members were opposed to Ruth McNally attending future meetings. It was conceded, however, that there might be the possibility of her making a formal presentation to a future meeting.

Peter Schneider will make contact for more information.

## **5. Interpretation of mixtures**

Due to time constraints Peter Gill had to truncate his presentation on this topic. He gave brief account of the procedure and criteria used by the FSS for the interpretation of stains of mixed origin. The following is a résumé of the points presented:

(i) Explanation of all peaks - consider all peaks above the threshold and eliminate all those which occur outside the guidelines.

(ii) Consider the characteristics of mixtures in the context of casework - make a visual estimate of all peaks.

(iii) Stutters have the following characteristics - 4bp less than a major allele, usually less than 15% of the size of the major allele (can rarely be up to 27%) and usually occur in pairs.

(iv) Heterozygous peaks - check that the minor peak is at least 60% (peak area) of the major allele.

(v) Determine the mixture proportion - component peaks from one origin can be measured as a proportion of all alleles at a particular locus. The mixture proportion will be similar across all loci within the mixture.

(vi) Compare the mixture proportion with the amelogenin peaks to ensure the inference is consistent.

(vii) When making estimates consider all possibilities for the denominator of a likelihood ratio - also consider whether bands can be allelic peaks or stutters, considering both possibilities.

(viii) List all of the possible genotypes and refer to the case circumstances e.g. eliminate the victim profile if this is justified and interpret the profile using the Evett model.

## **6. Future exercises**

Members should complete outstanding EDNAP exercises. There will be financial help in future to employ assistance for the preparation of samples etc. At the next meeting areas of interest should be presented with possible definitions for network research within the STADNAP framework. The following topics for further study were discussed:

- (i) mtDNA
- (ii) Y chromosome markers
- (iii) mixtures and artefacts
- (iv) degradation - there was no decision on how to obtain degraded material
- (v) population studies
- (vi) detection limits

Peter Gill will contact ABD and Pharmacia with a view to their attendance at a future meeting to inform on current developments. Their information might influence decisions on future EDNAP work.

## **7. EDNAP on the Internet**

Peter Schneider informed the meeting that the following have been added to the EDNAP website:

- (i) Conferences and announcements
- (ii) List of institutes represented at EDNAP
- (iii) STR population frequencies are under construction

## **8. Next EDNAP meeting**

Ann Jangblad and Ernesto d'Aloja offered Linköping and Rome, respectively, as the venue for the mid January meeting. Members elected for the meeting to be held in Rome. Details will be forthcoming.

Richard Scheithauer has offered Innsbruck as the venue for a June meeting of EDNAP and this can link with the Promega conference.

## **9. Any other business**

Some members have received a letter from Prof Elfreide Van den Eeckhout in Gent regarding EDNAP membership. Niels Morling will deal with the correspondence and Bernadette Hoste will make informal contact.