Intercomparison Program **"ANALYSIS OF DNA POLYMORPHISMS IN BLOOD STAINS AND OTHER BIOLOGICAL SAMPLES"**
Massive Parallel Sequencing (MPS)

1.-SCOPE AND ITEMS

The scope using Massive Parallel Sequencing (MPS) covered in the Intercomparison Exercise Program "Analysis of DNA polymorphisms in blood stains and other biological samples" includes tests for genetic identification by autosomal STR markers, X-chromosome, Y-chromosome markers and mitochondrial DNA.

Items are the same as for The basic level of the intercomparison exercise program and although it serves as proof of competence to the laboratory performing this methodology **it is not included in the accredited scope. Items from the advanced level are not supposed to be analyzed by MPS** The analysis of items consists of two modules:

- **Kinship Module , which contains:**
  - **Practical Study**: it entails the genetic analysis of 2-4 reference items (blood and saliva).
- **Forensic module , which contains**:
  - **Practical Study**: it involves analyzing genetically 2 unknown forensic items: one consisting of a human biological fluid or of a mixture of body fluids with a maximum of two contributors, in a volumetric proportion not more than 1:2 (v/v) in case they are different fluids and in 1:3 if it is the same fluid. The other item consists of hair or hairshaft in which analysis of mitochondrial DNA is requested.

The following points **DEFINITIONS**, **TIMETABLE**, **PARTICIPATING LABORATORIES AND REGISTRATION, Compromises to be met by participants, INSTRUCTIONS AND DELIVERY OF ITEMS**, are the same described in the participation rules of the analysis of items of the basic level.

2. THE RESULTS FORMS AND HOW TO SEND THEM

The results have to be sent through an electronic form of the GHEP-ISFG group website which is different from the results form of the analysis of the basic level. The laboratory will receive by mail information about its availability, approximately, a month before the deadline for sending results. The laboratories, will also receive a reminder a week before the end of that term. The form will be disabled immediately after the date and time scheduled for the deadline, the results being not admitted if received subsequently.

It is compulsory to send a signed and complete copy of the form with all paper sheets.even they are not filled, by ordinary mail or by uploading it to the web in case of

electronic signature. By signing this form, the laboratory agrees to use the items anonymously for the Exercise. Additionally they can be used as a reference material and/or as a quality control of the laboratory either using the techniques required in the Exercise or other forensic techniques but always, for the purpose of human identification. Analyzing non coding regions or regions which do not provide any sensitive information of the donor: diseases, pathologies or other genetic information which could infrige his/her privacy. Therefore, it is essential to send this document to the coordinator in order to receive the certificate.

Digital and printed versions should have the same content. All along the evaluation of MPS analysis results, records could be required, as it considers necessary for the assessment.

It is the responsibility of the laboratory to identify these records as described in the instructions, eliminating any data which could allow the identification of the laboratory and compromise anonymity.

It will be also reminded in the instructions to the participants that all comments made in the spaces provided will be published in the final report. So it is recommended not to give any information that might compromise the laboratory's anonymity.

Corrections of data and/or results will not be allowed after the deadline.

For any query related to the documentation sent, participants should contact the addresses and telephone numbers which are indicated in the instructions.

3. STATISTICAL DESIGN AND ESTABLISHMENT OF ASSIGNED VALUES

Definitions

Assigned value: Evaluation will be carried out with regard to assigned values. These values can be established by consensus from participants or by consensus value from expert laboratories.

Consensus value from participants results: to agree on a result there must be a minimum participation of 5 laboratories and concordance of results in at least 70% of the participants.

Consensus value from expert laboratories: the assigned value will be established from at least two expert laboratories.

4. EVALUATION OF RESULTS AND ISSUING OF CERTIFICATES

Regarding autosomal , X-chromosome and Y Chromosome results assessment will be performed taking into account the consensus value obtained from expert laboratories whereas in the case of mitochondral DNA assessment will be performed against the consensus value obtained from participants.

Mitochondrial DNA: In the case of the expression of results, both nomenclature medical (ex. A73G) and forensic (ex. 73G) will be accepted. Laboratories can submit results for all regions of mitochondrial DNA that are edited, however the haplotypes evaluated on the certificate will be the **ones regarding the control region**. The length variants present in the homopolymeric tracts will not be assessed. This includes insertions or deletions in the length variants that may appear in the positions: 16193, 309, 455, 463 and 573. In case of genetic analysis of mixtures, mitochondrial DNA haplotypes will not be evaluated for the certificate.

Certificates of evaluation will be issued

The following points **REPORTS OF RESULTS, WORKING GROUP MEETING CONFIDENTIALITY, QUALITY ASSURANCE and COMPLAINTS,** are the same described in the participation rules of the analysis of items of the basic level.