GRUPO DE HABLA ESPAÑOLA Y PORTUGUESA DE LA ISFG

GRUPO DE LÍNGUAS ESPANHOLA E PORTUGUESA DA ISFG

Ministerio de Justicia
INSTITUTO NACIONAL DE TOXICOLOGÍA Y CIENCIAS FORENSES

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INTERCOMPARISON PROGRAM

"ANALYSIS OF DNA POLYMORPHISMS IN BLOODSTAINS AND OTHER BIOLOGICAL SAMPLES" MASSIVE PARALLEL SEQUENCING MODULE (MPS)

BASIC LEVEL EXERCISE EIADN MPS Nr 1 (2023) DEADLINE: 15/05/2023

2023/Kinship Module
M1 to M3: reference items
2023/Forensic module
M4: forensic unknown item
M5: hair sample

Seal number

Approach:

2023/Kinship Module – Basic level

Practical Kinship study

- **M1, M2, M3:** reference items for genetic profiling.

2023/Forensic Module – Basic level

Practical Forensic study

- **M4:** forensic item for genetic profiling.
- **M5:** hair for mitochondrial DNA analysis.

Methodology to be used

The analyses will be performed by using the markers and with the Massive Parallel Sequencing (MPS) methods used by the laboratory: autosomal STRs, Y- STRs, X-STRs and mitochondrial DNA analysis. The items must be processed as real casework and, if possible, as blind samples.

INDEX

1. Methodology *Read carefully the instructions provided before filling in this section*

1.1 Workflow methodology

1.1.1 Library preparation

TABLE 1A

Marker	Kit	Others (specify)	Instrument	Others (specify)
A-STR				
Y-STR				
X-STR				
Mitochondrial DNA				

See Appendix 2023 MPS for codes

1.1.2 Library quantification

TABLE 1B

Marker	Kit	Others (specify)	Instrument	Others (specify)
A-STR				
Y-STR				
X-STR				
Mitochondrial DNA				

See Appendix 2023 MPS for codes

1.1.3 Template preparation

TABLE 1C

Marker	Kit	Others (specify)	Instrument	Others (specify)
A-STR				
Y-STR				
X-STR				
Mitochondrial DNA				

See Appendix 2023 MPS for codes

1.1.4 Sequencing

TABLE 1D

Tipo de marcador	Kit	Others (specify)	Chip/Flow Cell	Others (specify)	Instrument	Others (specify)
A-STR						
Y-STR						
X-STR						
Mitochondrial DNA						

See Appendix 2023 MPS for codes

1.1.5 Reference Sequence used.STR markers

1.1.6 Data analysis

TABLE 1E

Marker	Raw Data	Others (specify)	Allele/Variant Calling	Others (specify)	Analysis	Others (specify)
A-STR						
Y-STR						
X-STR						
Mitochondrial DNA						

See Appendix 2023 MPS for codes

1.2 Other considerations regarding methodology different to reported in the preceding tables

2. Practical studies results:

Read carefully the instructions provided in order to fill in the results tables and the rules of participation in order to know the establishment of assigned values and the evaluation of results https://ghep-isfg.org/en/proficiency/participation/

2.1 Library enrichment and quantification (if relevant)

TABLE 3

	Library amplification volumen (µL)		Individual Library quantification (pM)		Pool Library quantification (pM)	
Items	**STR**	**mtDNA**	**STR**	**mtDNA**	**STR**	**mtDNA**
M1						
M2						
M3						
M4						
M4.1 (if relevant)						
M4.2 (if relevant)						
M5						

2.2 Chips or flow cells data

TABLE 4

	STR	mtDNA
Instrument		
Markers		
Chip / Flow Cell		

	STR	mtDNA
Addressable Wells (Total reads)		
Usable reads (Final library ISPs)*		
Usable reads (%)		
No. Samples per chip / flow cell		
Exercise samples reads		
M1		
M2		
M3		
M4		
M4.1 (if relevant)		
M4.2 (if relevant)		
M5		
Number of markers		-
Chip equalization*		

** Ion Torrent.*

2.3 STR results

2.3.1 Autosomal STR and amelogenine

TABLE 5A-M1

					Nomenclature*	
LOCUS	**Alelle** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
AMEL						
D8S1179						
D21S11						
D7S820						
CSF1PO						
D3S1358						
TH01						
D13S317						
D16S539						
D2S1338						
D19S433						
vWA						
TPOX						
D18S51						
D5S818						
FGA						
Penta D						
Penta E						
D10S1248						
D22S1045						
D2S441						
D1S1656						
D12S391						
SE33 (ACTBP2)						

					Nomenclature*	
LOCUS	**Alelle** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
FES/FPS						
F13A01						
F13B						
LPL						
Penta C						
D6S1043						
D12ATA63						
D14S1434						
D1S1677						
D2S1776						
D3S4529						
D4S2408						
D5S2800						
D6S474						

* It can be filled one of them or both.

TABLE 5A-M2

					Nomenclature*	
LOCUS	**Allele** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
AMEL						
D8S1179						
D21S11						
D7S820						
CSF1PO						
D3S1358						
TH01						
D13S317						
D16S539						
D2S1338						
D19S433						
vWA						
TPOX						
D18S51						
D5S818						
FGA						
Penta D						
Penta E						
D10S1248						
D22S1045						
D2S441						
D1S1656						
D12S391						
SE33 (ACTBP2)						
FES/FPS						
F13A01						

					Nomenclature*	
LOCUS	Allele (CE)	Coverage	Sequence	Flanking regions variations Ex:rs9546005	Bracketed Repeat Region Ex [TATC]8	ISFG
F13B						
LPL						
Penta C						
D6S1043						
D12ATA63						
D14S1434						
D1S1677						
D2S1776						
D3S4529						
D4S2408						
D5S2800						
D6S474						

* It can be filled one of them or both

TABLE 5A-M3

					Nomenclature*	
LOCUS	Allele (CE)	Coverage	Sequence	Flanking regions variations Ex:rs9546005	Bracketed Repeat Region Ex [TATC]8	ISFG
AMEL						
D8S1179						
D21S11						
D7S820						
CSF1PO						
D3S1358						
TH01						
D13S317						
D16S539						
D2S1338						
D19S433						
vWA						
TPOX						
D18S51						
D5S818						
FGA						
Penta D						
Penta E						
D10S1248						
D22S1045						
D2S441						
D1S1656						
D12S391						
SE33 (ACTBP2)						
FES/FPS						
F13A01						
F13B						
LPL						

					Nomenclature*	
LOCUS	**Allele** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
Penta C						
D6S1043						
D12ATA63						
D14S1434						
D1S1677						
D2S1776						
D3S4529						
D4S2408						
D5S2800						
D6S474						

* It can be filled one of them or both

TABLE 5A-M4

					Nomenclature*	
LOCUS	**Alleles totales** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
AMEL						
D8S1179						
D21S11						
D7S820						
CSF1PO						
D3S1358						
TH01						
D13S317						
D16S539						
D2S1338						
D19S433						
vWA						
TPOX						
D18S51						
D5S818						
FGA						
Penta D						
Penta E						
D10S1248						
D22S1045						
D2S441						
D1S1656						
D12S391						
SE33 (ACTBP2)						
FES/FPS						
F13A01						
F13B						
LPL						
Penta C						

					Nomenclature*	
LOCUS	**Alleles totales** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
D6S1043						
D12ATA63						
D14S1434						
D1S1677						
D2S1776						
D3S4529						
D4S2408						
D5S2800						
D6S474						

* It can be filled one of them or both

2.3.2 Y-STR

TABLE 5B-M1

					Nomenclature*	
LOCUS	**Allele** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
DYS456						
DYS389 I						
DYS390						
DYS389 II						
DYS458						
DYS19						
DYS385						
DYS393						
DYS391						
DYS439						
DYS635						
DYS392						
GATAH4						
DYS437						
DYS438						
DYS448						
DYS460						
DYS461						
GATAA10						
DYS388						
DYS576						
DYS481						
DYS549						
DYS533						
DYS570						
DYS643						
DYS627						
DYS518						

					Nomenclature*	
LOCUS	**Allele** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
DYS449						
DYF387S1						

It can be filled one of them or both.

TABLE 5B-M2

					Nomenclature*	
LOCUS	**Allele** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
DYS456						
DYS389 I						
DYS390						
DYS389 II						
DYS458						
DYS19						
DYS385						
DYS393						
DYS391						
DYS439						
DYS635						
DYS392						
GATAH4						
DYS437						
DYS438						
DYS448						
DYS460						
DYS461						
GATAA10						
DYS388						
DYS576						
DYS481						
DYS549						
DYS533						
DYS570						
DYS643						
DYS627						
DYS518						
DYS449						
DYF387S1						

* It can be filled one of them or both

TABLE 5B-M3

					Nomenclature*	
LOCUS	**Allele** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
DYS456						
DYS389 I						
DYS390						
DYS389 II						
DYS458						
DYS19						
DYS385						
DYS393						
DYS391						
DYS439						
DYS635						
DYS392						
GATAH4						
DYS437						
DYS438						
DYS448						
DYS460						
DYS461						
GATAA10						
DYS388						
DYS576						
DYS481						
DYS549						
DYS533						
DYS570						
DYS643						
DYS627						
DYS518						
DYS449						
DYF387S1						

* It can be filled one of them or both.

TABLE 5B-M4

					Nomenclature*	
LOCUS	**Alleles totales** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
DYS456						
DYS389 I						
DYS390						
DYS389 II						
DYS458						
DYS19						
DYS385						
DYS393						

					Nomenclature*	
LOCUS	**Alleles totales** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
DYS391						
DYS439						
DYS635						
DYS392						
GATAH4						
DYS437						
DYS438						
DYS448						
DYS460						
DYS461						
GATAA10						
DYS388						
DYS576						
DYS481						
DYS549						
DYS533						
DYS570						
DYS643						
DYS627						
DYS518						
DYS449						
DYF387S1						

* It can be filled one of them or both

2.3.3 X-STR

TABLE 5C-M1

					Nomenclature*	
LOCUS	**Allele** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
HPRTB						
DXS8378						
DXS9898						
DXS7133						
GATA31E08						
GATA172D05						
DXS7423						
DXS6809						
DXS7132						
DXS9902						
DXS6789						
DXS10103						
DXS10134						
DXS10074						
DXS10101						
DXS10135						

					Nomenclature*	
LOCUS	**Allele** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
DXS10146						
DXS10079						
DXS10148						

* It can be filled one of them or both

TABLE 5C-M2

					Nomenclature*	
LOCUS	**Allele** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
HPRTB						
DXS8378						
DXS9898						
DXS7133						
GATA31E08						
GATA172D05						
DXS7423						
DXS6809						
DXS7132						
DXS9902						
DXS6789						
DXS10103						
DXS10134						
DXS10074						
DXS10101						
DXS10135						
DXS10146						
DXS10079						
DXS10148						

* It can be filled one of them or both

TABLE 5C-M3

					Nomenclature*	
LOCUS	**Allele** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
HPRTB						
DXS8378						
DXS9898						
DXS7133						
GATA31E08						

					Nomenclature*	
LOCUS	**Allele** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
GATA172D05						
DXS7423						
DXS6809						
DXS7132						
DXS9902						
DXS6789						
DXS10103						
DXS10134						
DXS10074						
DXS10101						
DXS10135						
DXS10146						
DXS10079						
DXS10148						

* It can be filled one of them or both

TABLE 5C-M4

					Nomenclature*	
LOCUS	**Alleles totales** (CE)	**Coverage**	**Sequence**	**Flanking regions variations Ex:rs9546005**	**Bracketed Repeat Region Ex [TATC]8**	**ISFG**
HPRTB						
DXS8378						
DXS9898						
DXS7133						
GATA31E08						
GATA172D05						
DXS7423						
DXS6809						
DXS7132						
DXS9902						
DXS6789						
DXS10103						
DXS10134						
DXS10074						
DXS10101						
DXS10135						
DXS10146						
DXS10079						
DXS10148						

* It can be filled one of them or both

2.4 Mitochondrial DNA results

In Table 6A, report the initial and final positions of the edited regions and in Table 6B report the haplotypes in the order requested in the instructions**. Remember ONLY to analyze the Control Region.**

TABLE 6A

ITEMS	Edited regions	Coverage	Haplogroup
M1			
M2			
M3			
M4			
M5			

TABLE 6B

ITEMS	HAPLOTYPE
M1	
M2	
M3	
M4	
M5	

3.Practical Studies Conclusions

3.1 Massive Parallele Sequencing (MPS) Module

3.1.1 Remarks about items M1, M2, M3, M4 and M5

Indicate any comments or remarks, you consider, about the analyzed items. It is not necessary to investigate a genetic relationship among them.

4. Remarks about this exercise

5. Suggestions for subsequent exercises

6.Compromises to be met by the participant

The analyses, both, the generated results and their statistical evaluation have been performed in the facilities of the participating laboratory and by its own staff, following working protocols used in routine casework together with safety precautions. **In accordance with the donors' consent, these items will be processed anonymously for the Intercomparison Exercise INTCFM/GHEP-ISFG. Additionally they could be used as a reference material and/or quality control for 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 that would not provided sensitive information about the donor: illnesses, pathologies or other genetic information which could infringe his/her privacy.**

Name of the person in charge

Date and signature

Note.- In order to receive the certificate of participation you must return this form duly signed.