



GRUPO DE HABLA ESPAÑOLA Y PORTUGUESA DE LA ISFG
GRUPO DE LINGUAS ESPANHOLA E PORTUGUESA DA ISFG
SPANISH AND PORTUGUESE-SPEAKING WORKING GROUP OF ISFG

Cromossomas sexuais: discussão dos resultados

XVII JORNADAS DO GHEP-ISFG

San Andrés - Islas (Colombia)

4-7 de Junho de 2012

Leonor Gusmão

NIVEL BÁSICO: Módulo de parentesco



M1-M4

20172	AMELOGENINA	X/X	X/Y	X/X	X/Y
20181	AMELOGENINA	X	XY	X	XY
20182	AMELOGENINA	X/X	X/Y	X/X	X/Y
20183	AMELOGENINA	X/X	X/Y	X/X	X/Y
20184	AMELOGENINA	X	X/Y	X	X
20194	AMELOGENINA	X	XY	X	XY
20199	AMELOGENINA	X	X/Y	X	
20202	AMELOGENINA	X	XY	X	XY
20210	AMELOGENINA	X	X/Y	X/Y	X/Y
20211	AMELOGENINA	XX	XY	XX	XY
20212	AMELOGENINA	X/X	X/Y	X/X	X/Y
20233	AMELOGENINA	X/X	X/Y	X/X	X/Y
20236	AMELOGENINA	X	X/Y		X
20238	AMELOGENINA	X/X	X/Y	X/X	X/Y
20242	AMELOGENINA	X	XY	X	XY
20249	AMELOGENINA	X/X	X/Y	X/X	X/Y
20255	AMELOGENINA	XX	XY	XX	XY
20269	AMELOGENINA	X	XY	X	XY
20271	AMELOGENINA	XX	XY	XX	XY
20279	AMELOGENINA	X	X/Y	X	
25152	AMELOGENINA	X/X	X/Y	X/X	X/Y

22 em 110

Nomenclatura!

NIVEL BÁSICO: Módulo de parentesco



Cromossoma Y

Marcador	M2	erros	M4	erros	S/ result.	Labs
DYS_19	80		76			
DYS_385	78	1	75	3		20231/20254/20194
DYS_389_I	80		75		1	20231
DYS_389_II	80		75		1	20231
DYS_390	80		76	1		20219
DYS_391	80		76			
DYS_392	80		76		1	20231
DYS_393	80		76			
DYS_437	79		75			
DYS_438	79		75			
DYS_439_GATA_A4	79		75		1	20231
DYS_448	62		59			
DYS_456	61		58			
DYS_458	62		59	1		23177
DYS_460_GATA_A71	5		5			
DYS_461_GATA_A72	4		4			
DYS_635_GATA_C4	64		61		1	20231
GATA_A10	5		5			
GATA_H4	65		62			

NIVEL BÁSICO: Módulo de parentesco



Cromossoma Y

Observações:

Lab sem resultados para M4: 20184*/20199**/20249***/20279

*apres. resultados para cr. X e Autossomicos com Amel X (contaminação M3)

20184	AMELOGENINA	X	X/Y	X	X
-------	-------------	---	-----	---	---

**apres. resultados para Autossomicos (perfil incompleto)

***apres. resultados para Autossomicos (tudo ok!)

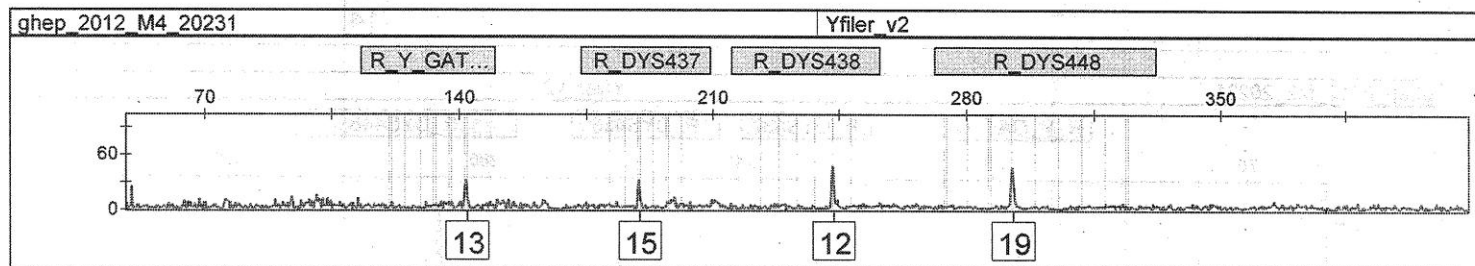
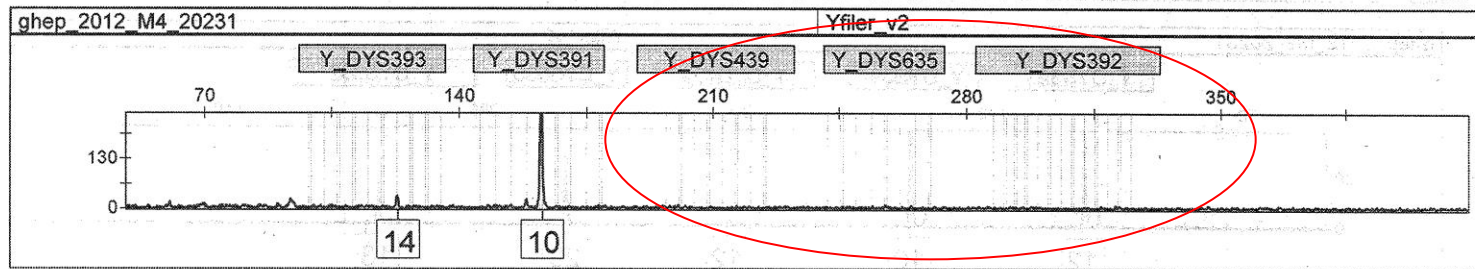
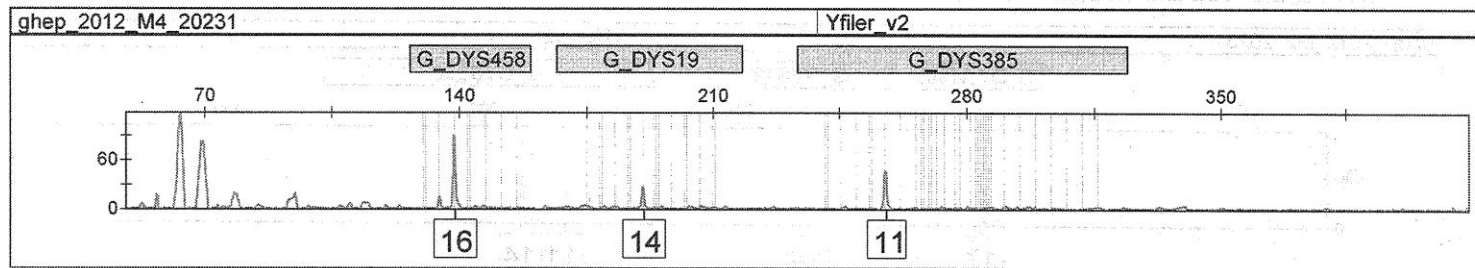
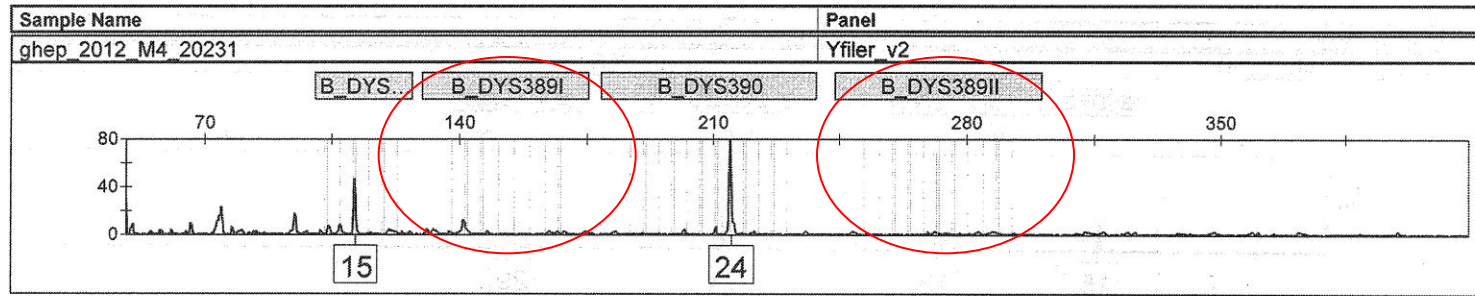
20249	AMELOGENINA	X/X	X/Y	X/X	X/Y
-------	-------------	-----	-----	-----	-----

Nomenclatura H4: 62 labs usam nomenclatura do Yfiler

NIVEL BÁSICO: Módulo de parentesco



Cromossoma Y



NIVEL BÁSICO: Módulo de parentesco

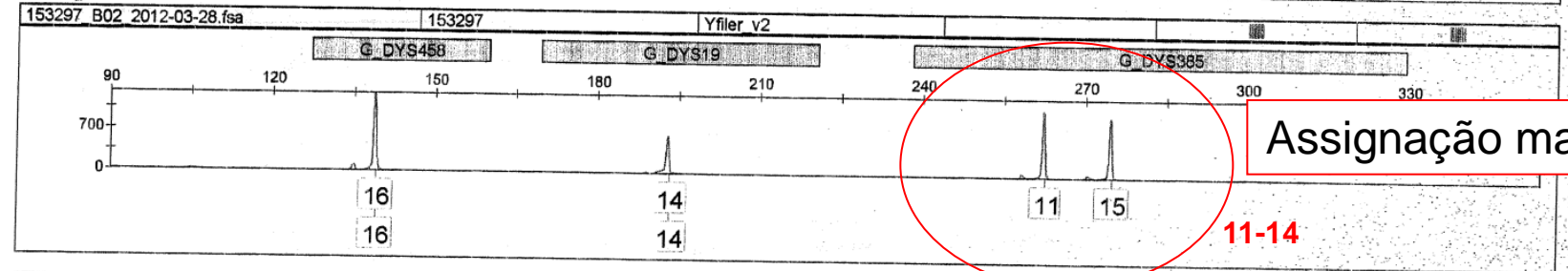
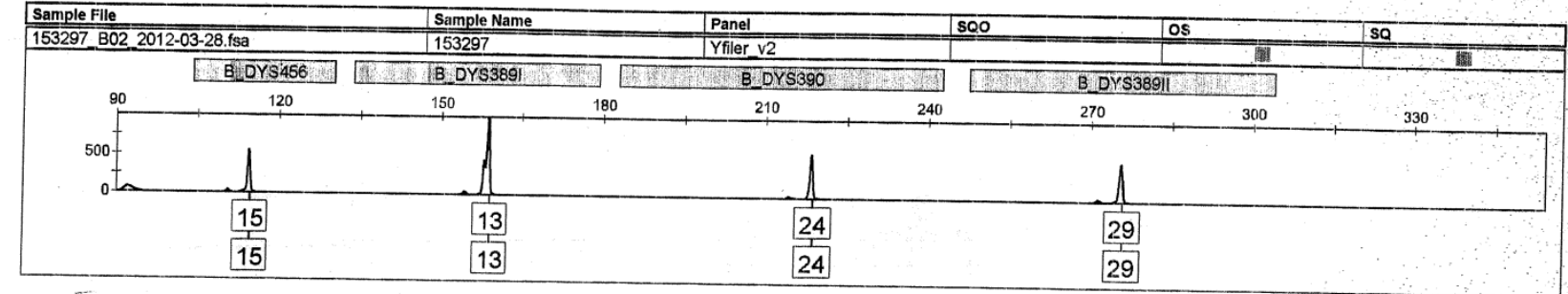
Cromossoma Y



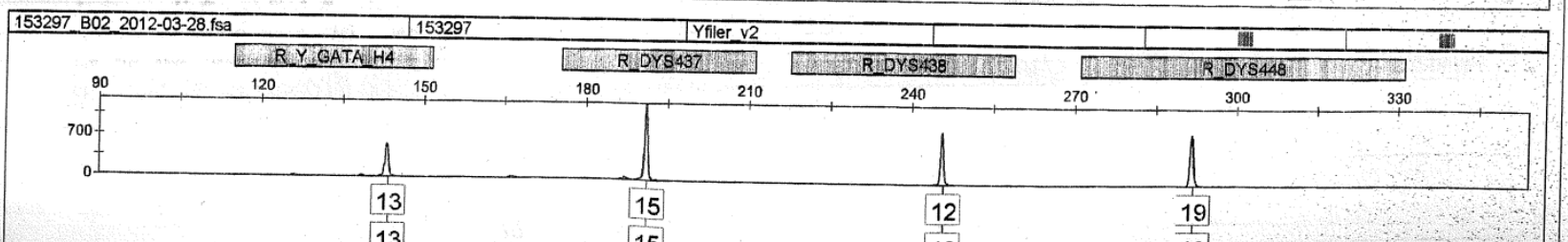
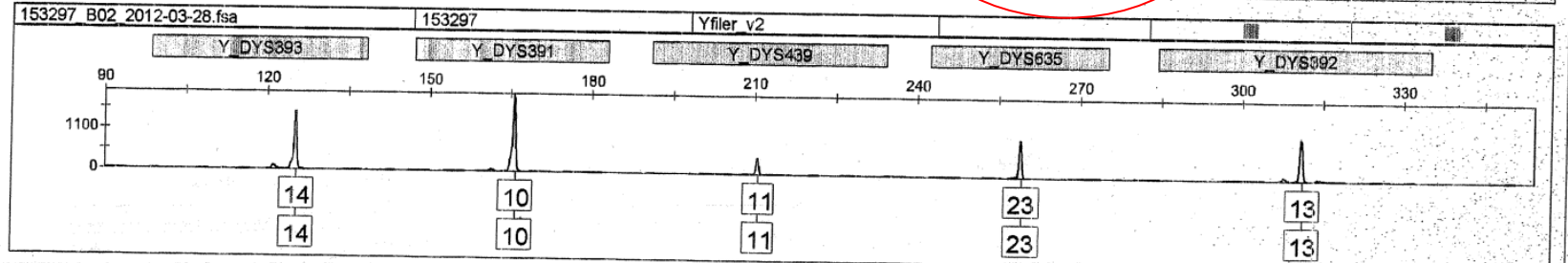
Applied Biosystems
GeneMapper ID v3.2

20254

YFILER_280312



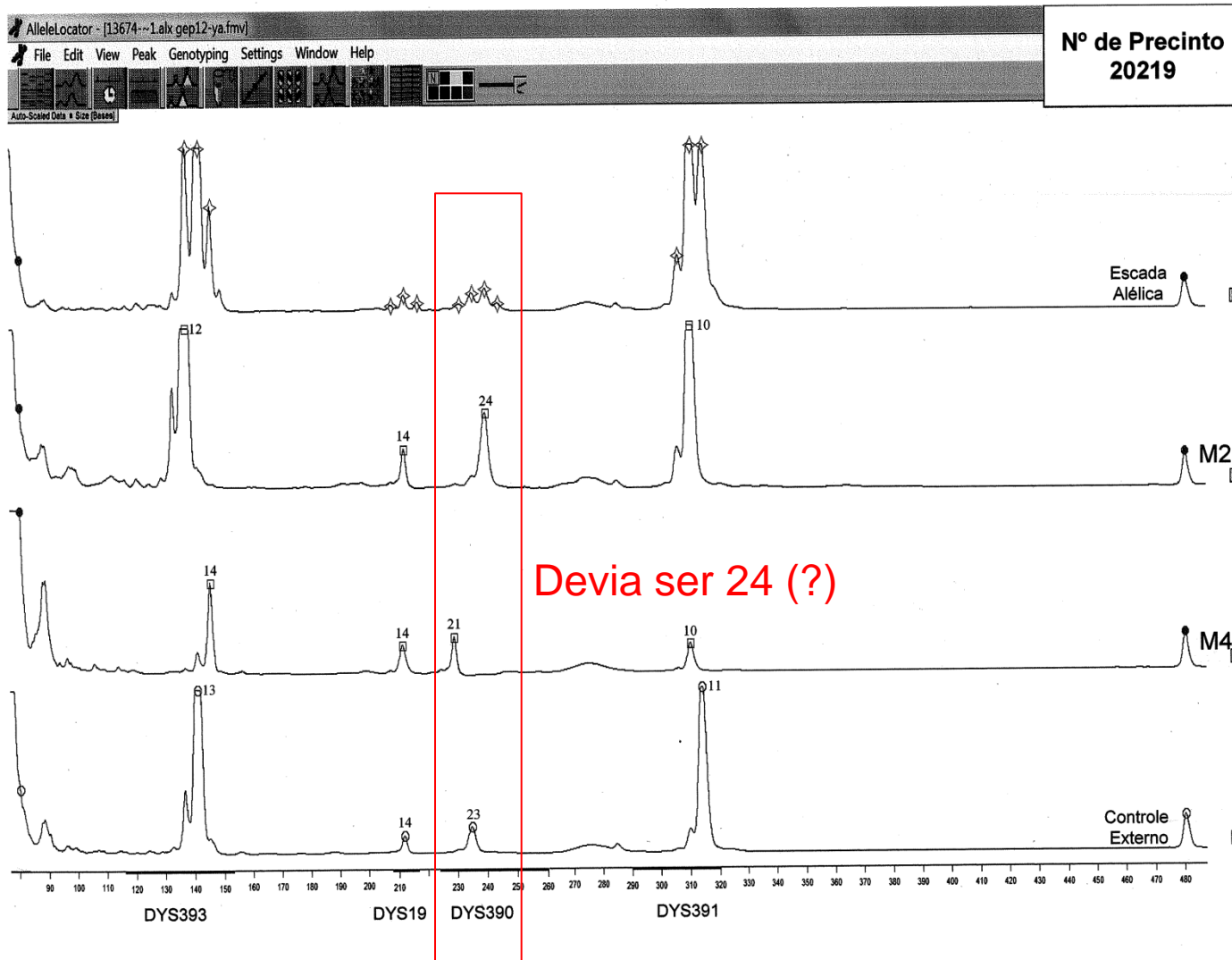
Assinação manual?



NIVEL BÁSICO: Módulo de parentesco



Cromossoma Y

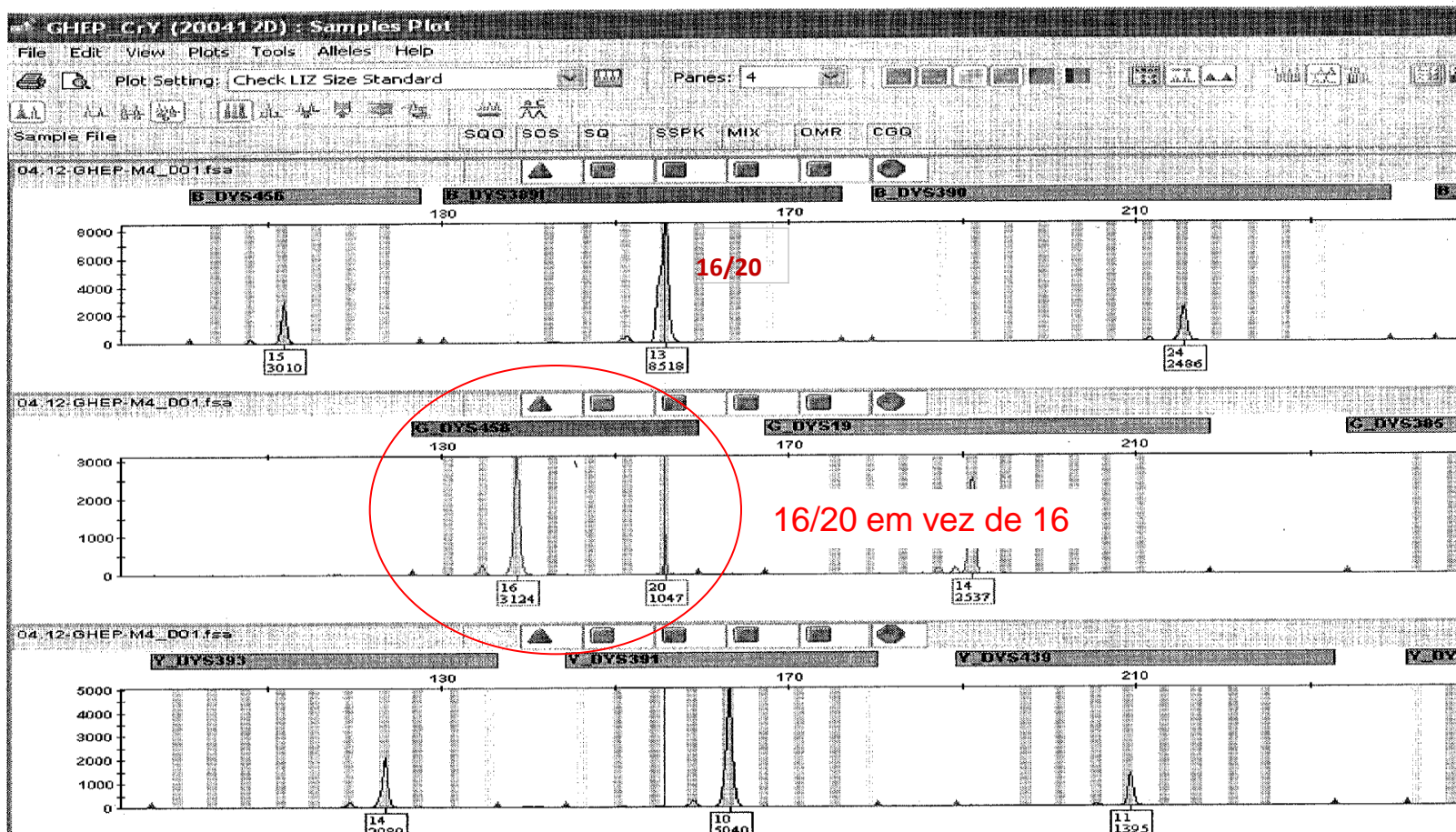


NIVEL BÁSICO: Módulo de parentesco

Cromossoma Y



23177



NIVEL BÁSICO: Módulo de parentesco



Cromossoma X

M1-M4

Marcador/Marker	M1-M3	M4	M1	M2	M3	M4	Labs.
DXS8378		35	34	1		1	20184
DXS9898		29	28	1	5	1	20184(M1-M4)/M2:20226/20232/20248/25152
DXS7133		29	28			1	20184
GATA31E08		27	26			1	20184
GATA172D05		30	29			1	
DXS7423		35	34			1	20184
DXS6809		29	28	25		1	20230(M3)/20184(M4)
DXS7132		34	33	1	1	1	25152(M1-M3)/20184(M4)
DXS9902		27	26			2	20184/20248
DXS6789		29	28			1	20184
DXS10103		10	10				
DXS10134		10	10	1			20174
DXS10074		11	11				
DXS10101		10	10				
DXS10135		11	11			1	20231
DXS10146		10	10				
DXS10079		11	11				
DXS10148		10	10	1			20231
HPRTB		11	11				

20199 – S/resultado para M4

Determinações - 1582

NIVEL BÁSICO: Módulo de parentesco

Cromossoma X



Marcador	M1	M2	M3	M4
AMELOGENINA	X	X/Y	X	X
D8S1179	15	11/12	12	12
D21S11	28/31	29/30	28/29	28/29
D7S820	9/10	10/11	8/11	11
CSF1PO	11/12	11/13	10/11	10/11
D3S1358	15/17	17	14/16	14/16
TH01	6/9.3	8/9	9/9.3	9/9.3
D13S317	11/12	8/13	12	12
D16S539	11	11/12	11	11
D2S1338	17/20	17/19	20	20
D19S433	13/16	13/15	14	14
VWA	18	15/17	16/18	16/18
TPOX	8/9	8	9/11	8/11
D18S51	13/17	12/14	16/17	16/17
D5S818	11/13	12	11/13	11/13
FGA	22/25	21/22	22/23	22/23

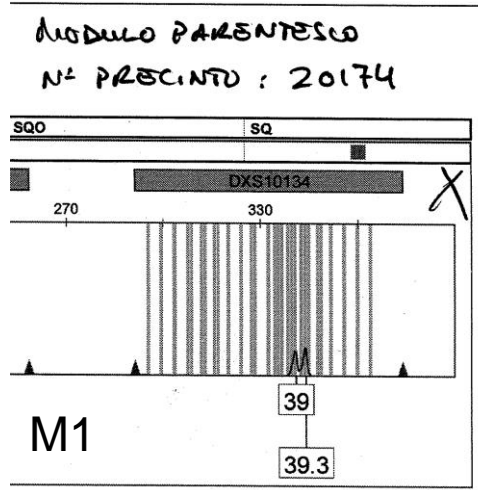
Precinto 20184

**Contaminação
ou
troca de amostras**

NIVEL BÁSICO: Módulo de parentesco



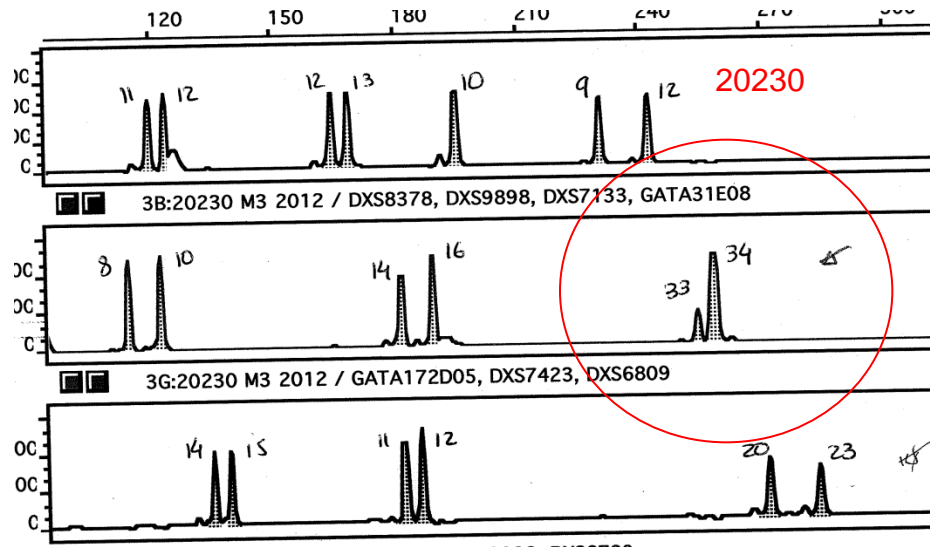
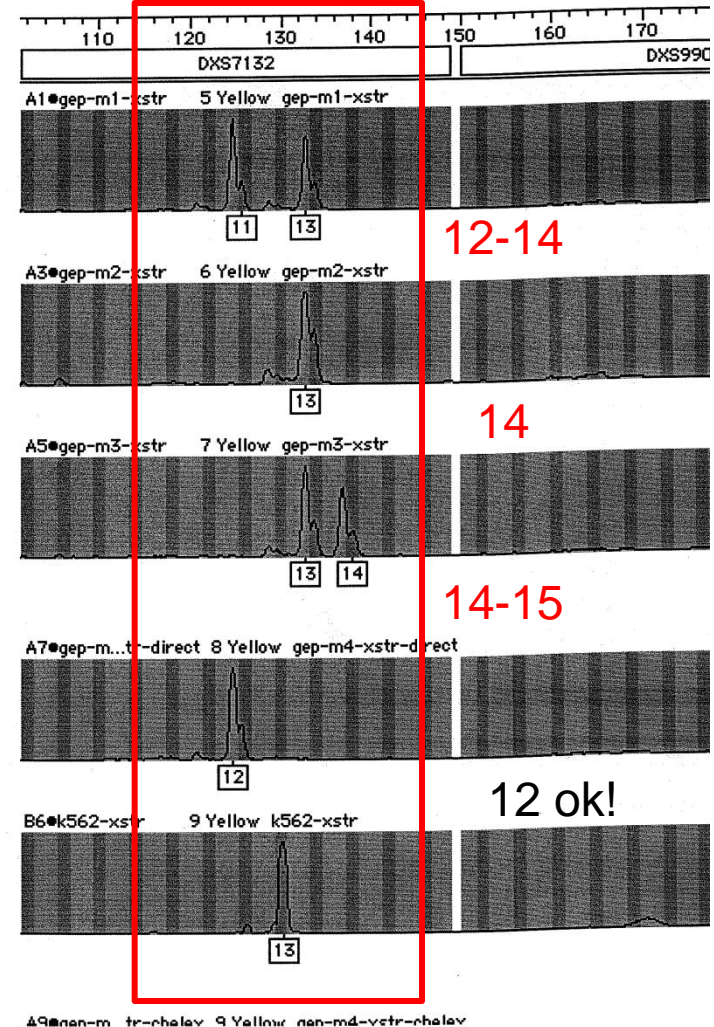
Cromossoma X



Reporta: 30-39.3

Erro de transcrição

25152 – DXS7132



NIVEL BÁSICO: Módulo de parentesco

Cromossoma X



DXS9898

Precinto	M1	M2	M3	M4
20184	11	8	11/12	11
20226	12	8	12/13	12
20232	12	9	12/13	12
20248	12	8	12/13	12
25152	12	9	12/13	12

12	8.3	12/13	12
----	-----	-------	----

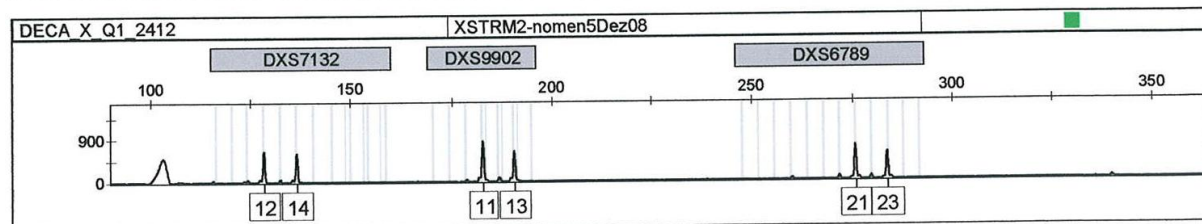
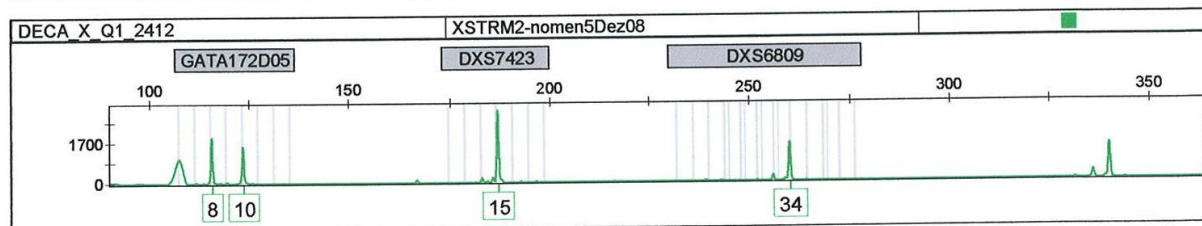
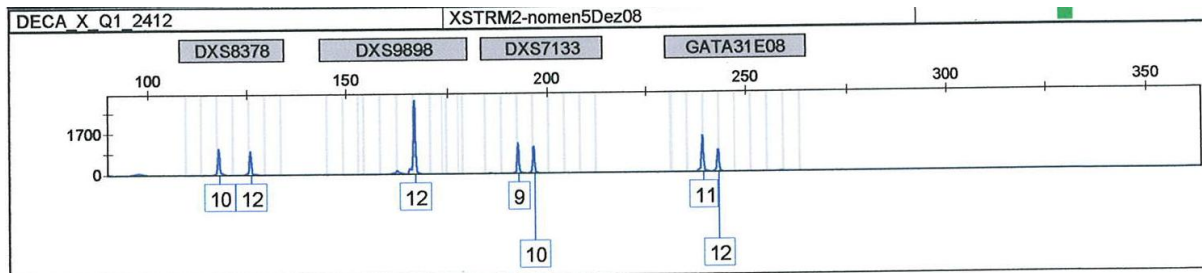
LADDER ALÉLICO!

NIVEL BÁSICO: Módulo de parentesco



Cromossoma X- DXS6809

25 laboratórios	34
20243	34/54
20277	34/X
20278	34/>38
20282	34/53.1



NIVEL BÁSICO: Módulo de parentesco



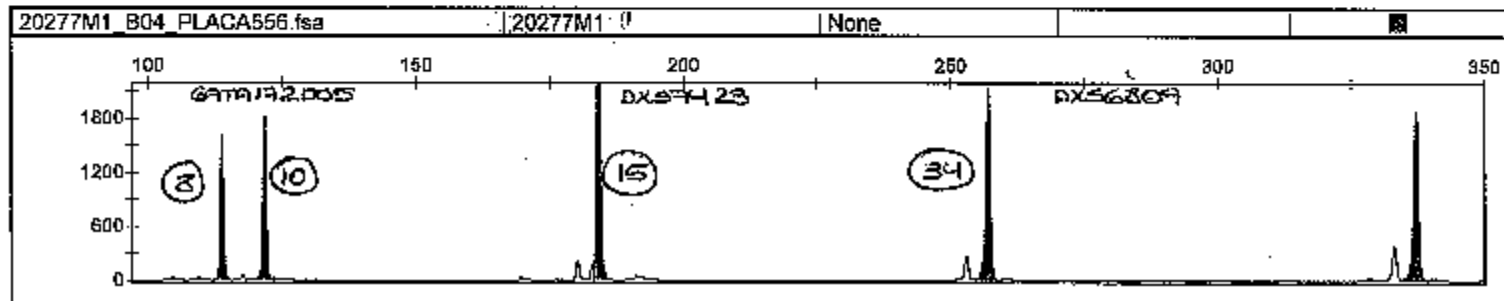
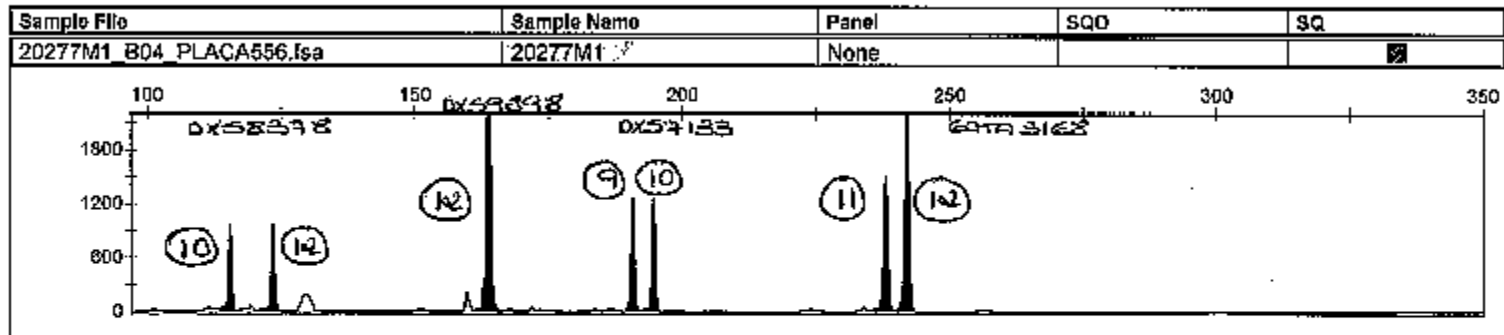
Cromossoma X- DXS6809

25 laboratórios	34
20243	34/54
20277	34/X
20278	34/>38
20282	34/53.1

Silent Allele!



20277.M1-M4(X-STRs)

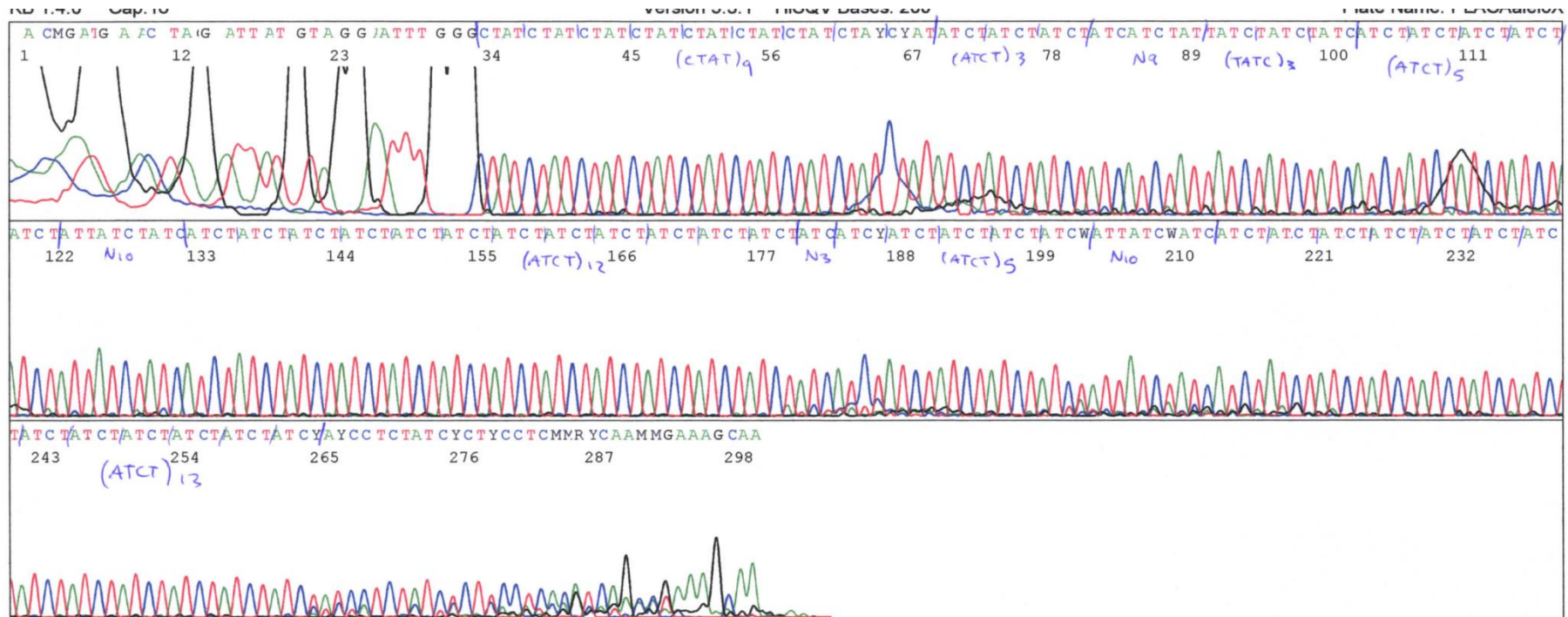


NIVEL BÁSICO: Módulo de parentesco



Cromossoma X- DXS6809

20277: 34/X



(CTAT)₉ (ATCT)₃ N₉ (TATC)₃ (ATCT)₅ N₁₀ (ATCT)₁₂ N₃ (ATCT)₅ N₁₀ (ATCT)₁₃

X-STRs

DXS6809

Al.	Allele structure: (CTAT) _n (ATCT) ₃ N ₉ (TATC) _n (ATCT) _n N ₁₀ (ATCT) _n	n	Sequenced in
27	Pr1 (22bp) 47bp (CTAT) ₂ <u>A</u> TAT (CTAT) ₃ (ATCT) ₃ N ₉ (TATC) ₄ (ATCT) ₅ N ₁₀ (ATCT) ₉ 14bp (20bp) Pr2	1	AFR
27	Pr1 (22bp) 47bp (CTAT) ₇ (ATCT) ₃ N ₉ (TATC) ₃ (ATCT) ₃ N ₁₀ (ATCT) ₁₁ 14bp (20bp) Pr2	2	CAU
28	Pr1 (22bp) 47bp (CTAT) ₂ ATAT (CTAT) ₃ (ATCT) ₃ N ₉ (TATC) ₄ (ATCT) ₅ N ₁₀ (ATCT) ₁₀ 14bp (20bp) Pr2	1	AFR
28	Pr1 (22bp) 47bp (CTAT) ₇ (ATCT) ₃ N ₉ (TATC) ₃ (ATCT) ₃ N ₁₀ (ATCT) ₁₄ 14bp (20bp) Pr2	2	CAU
29	Pr1 (22bp) 47bp (CTAT) ₂ ATAT (CTAT) ₃ (ATCT) ₃ N ₉ (TATC) ₄ (ATCT) ₅ N ₁₀ (ATCT) ₁₁ 14bp (20bp) Pr2	1	AFR
29	Pr1 (22bp) 47bp (CTAT) ₆ (ATCT) ₃ N ₉ (TATC) ₄ (ATCT) ₆ N ₁₀ (ATCT) ₁₀ 14bp (20bp) Pr2	1	AFR
29	Pr1 (22bp) 47bp (CTAT) ₇ (ATCT) ₃ N ₉ (TATC) ₃ (ATCT) ₃ N ₁₀ (ATCT) ₁₃ 14bp (20bp) Pr2	1	CAU
29	Pr1 (22bp) 47bp (CTAT) ₈ (ATCT) ₃ N ₉ (TATC) ₄ (ATCT) ₅ N ₁₀ (ATCT) ₉ 14bp (20bp) Pr2	1	CAU
30	Pr1 (22bp) 47bp (CTAT) ₂ ATAT (CTAT) ₃ (ATCT) ₃ N ₉ (TATC) ₄ (ATCT) ₅ N ₁₀ (ATCT) ₁₄ 14bp (20bp) Pr2	1	AFR
30	Pr1 (22bp) 47bp (CTAT) ₇ (ATCT) ₃ N ₉ (TATC) ₃ (ATCT) ₃ N ₁₀ (ATCT) ₁₄ 14bp (20bp) Pr2	1	AFR
30	Pr1 (22bp) 47bp (CTAT) ₈ (ATCT) ₃ N ₉ (TATC) ₄ (ATCT) ₅ N ₁₀ (ATCT) ₁₀ 14bp (20bp) Pr2	1	CAU

Alelo 34

48bases- (CTAT)₉ -(ATCT)₃ -9bases-(TATC)₃ -(ATCT)₅ -10bases-(ATCT)₁₄

Alelo >38

48bases- (CTAT)₈ -(ATCT)₃ -9bases-(TATC)₃ -(ATCT)₅ -10bases-(ATCT)₁₂ -ATC-(ATCT)₅ -10bases- (ATCT)₁₄

Seria alelo 31 + 3bases + 10bases + (ATCT)₁₉ = 31 + 3.1 + 19 = 53.1

32	Pr1 (22bp) 47bp (CTAT) ₈ (ATCT) ₃ N ₉ (TATC) ₃ (ATCT) ₅ N ₁₀ (ATCT) ₁₃ 14bp (20bp) Pr2	2	AFR, CAU
32	Pr1 (22bp) 47bp (CTAT) ₉ (ATCT) ₃ N ₉ (TATC) ₃ (ATCT) ₅ N ₁₀ (ATCT) ₁₄ 14bp (20bp) Pr2	3	AS, CAU
32	Pr1 (22bp) 47bp (CTAT) ₁₀ (ATCT) ₃ N ₉ (TATC) ₃ (ATCT) ₅ N ₁₀ (ATCT) ₁₁ 14bp (20bp) Pr2	1	AS
32.1	Pr1 (22bp) 47bp (CTAT) ₉ (ATCT) ₃ N ₉ (TATC) ₆ <u>T</u> N ₁₀ (ATCT) ₁₄ 14bp (20bp) Pr2	1	AFR
33	Pr1 (22bp) 47bp (CTAT) ₁₀ (ATCT) ₃ N ₉ (TATC) ₃ (ATCT) ₅ N ₁₀ (ATCT) ₁₄ 14bp (20bp) Pr2	1	AFR
33	Pr1 (22bp) 47bp (CTAT) ₉ (ATCT) ₃ N ₉ (TATC) ₃ (ATCT) ₅ N ₁₀ (ATCT) ₁₃ 14bp (20bp) Pr2	3	AS, CAU
33	Pr1 (22bp) 47bp (CTAT) ₁₁ (ATCT) ₃ N ₉ (TATC) ₃ (ATCT) ₅ N ₁₀ (ATCT) ₁₁ 14bp (20bp) Pr2	1	AS
33.1	Pr1 (22bp) 47bp (CTAT) ₁₀ (ATCT) ₃ N ₉ (TATC) ₆ <u>T</u> N ₁₀ (ATCT) ₁₄ 14bp (20bp) Pr2	4	AFR
33.1	Pr1 (22bp) 47bp (CTAT) ₉ (ATCT) ₃ N ₉ (TATC) ₆ <u>T</u> N ₁₀ (ATCT) ₁₅ 14bp (20bp) Pr2	1	AFR
34	Pr1 (22bp) 47bp (CTAT) ₉ (ATCT) ₃ N ₉ (TATC) ₃ (ATCT) ₅ N ₁₀ (ATCT) ₁₄ 14bp (20bp) Pr2	6	AFR, CAU, AS

NIVEL BÁSICO: Módulo Forense



	M5 (2004) +	M2 (2012)	Consensuado		M5-1	M5-2
DXS8378	10	12	10/12	12	23	14
DXS9898	12	8.3	8.3/12	8.3	19	11
DXS7133	9	9	9	9	19	11
GATA31E08	13	13	13	13	18	11
GATA172D05	6	6	6	6	19	11
DXS7423	14	17	14/17	17	23	14
DXS6809	31	33	31/33	33	19	11
DXS7132	12	14	12/14	14	23	15
DXS9902	13	12	12/13	12	19	11
DXS6789	21	22	21/22	22	19	11
DXS10103	16	19	16/19	19	7	4
DXS10134	35	37	35/37	37	7	4
DXS10074	16	14	14/16	14	8	5
DXS10101	33	34.2	33/34.2	34.2	7	4
DXS10135	20.1	22.1	20.1/22.1	22.1	7	4
DXS10146	28	29	28/29	29	7	4
DXS10079	18	20	18/20	20	8	5
DXS10148	24.1	24.1	24.1	24.1	7	4
HPRTB	12	14	12/14	14	7	4

LD 14
No LD 9

M5 – Cromossoma Y

LD: si / no

Decomposição M5-1/M5-2

NIVEL BÁSICO: Módulo Forense

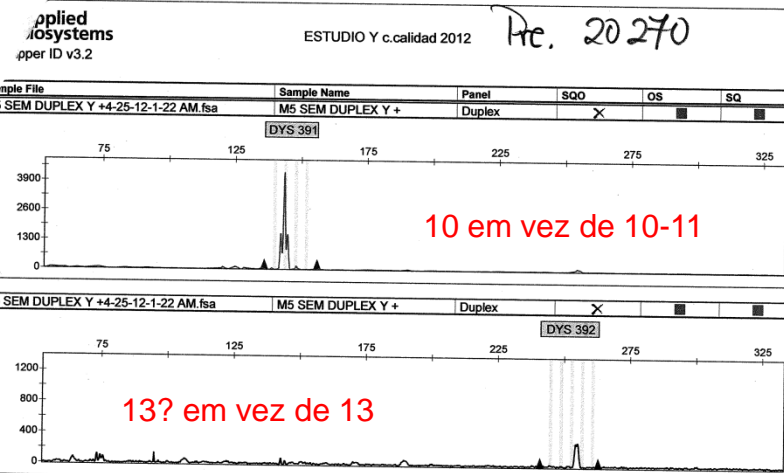


LD/DE	Marcador	Precinto	M5-1	M5-2	Vref 1	Vref-2
NO	DYS_385	20176	11/14		11/12/14	11/14
NO	DYS_389_II	20184	29		29/30	29
NO	DYS_385	20184	11/14		11/12/14	11/14
NO	DYS_391	20184	10		10/11	10
si	DYS_385	20197	11-14		11/12/14	11/14
SI	DYS_389_I	20207	13/14		13/14	13
NO	DYS_389_II	20230	29		29/30	29
NO	DYS_635_GATA_C4		23		23/24	23
SI	DYS_458	20238	16/17	17	16/17	17
SI	DYS_392		12/13	13	13	13
SI	DYS_385	20241	7/11/12/14	11/14	11/12/14	11/14
SI	DYS_389_I	20243	13/14	13/14	13/14	13
SI	DYS_458		16/17	16/17	16/17	17
SI	DYS_385		11/12/14	11/12/14	11/12/14	11/14
SI	DYS_393		12/13	12/13	12/13	12
SI	DYS_391		10/11	10/11	10/11	10
SI	DYS_635_GATA_C4		23/24	23/24	23/24	23
SI	DYS_437		14/15	14/15	14/15	15
SI	DYS_448		18/19	18/19	18/19	19
SI	DYS_389_I	20245	13/14	13/14	13/14	13
SI	DYS_458		16/17	16/17	16/17	17
SI	DYS_393		12/13	12/13	12/13	12
SI	DYS_391		10/11	10/11	10/11	10
SI	DYS_448		18/19	18/19	18/19	19
NO	DYS_385	20248	11-14		11/12/14	11/14
	DYS_389_I	20249	13		13/14	13
	DYS_389_II		29		29/30	29
	DYS_393		14		12/13	12
	DYS_391		10		10/11	10
	DYS_439_GATA_A4		11		12	12
	DYS_437		15		14/15	15
SI	DYS_458	20259	16/17	16/17	16/17	17
SI	DYS_393		12/13	12/13	12/13	12
SI	DYS_391		10/11	10/11	10/11	10
SI	DYS_635_GATA_C4		23/24	?	23/24	23
SI	DYS_437		14/15	14/15	14/15	15
SI	DYS_448		18/19	18/19	18/19	19
si	DYS_389_I	20261	13/14		13/14	13
NO	DYS_635_GATA_C4	20265	23		23/24	23
NO	DYS_635_GATA_C4	20269	23		23/24	23
si	DYS_391	20270	10	10	10/11	10
SI	DYS_392	20275	13/14	13	13	13
NO	DYS_389_I	23017	13	14	13/14	13

M5 – Cromossoma Y

LD: si / no

Decomposição M5-1/M5-2



NIVEL BÁSICO: Módulo Forense



LD/DE	Marcador/Marker	Precinto	M5-1	M5-2		
NO	DXS9898	20184	8/11		8.3/12	8.3
NO	DXS7423	20208	17		14/17	17
NO	DXS7132	20208	14		12/14	14
NO	DXS10103	20208	19		16/19	19
NO	DXS10074	20208	14		14/16	14
NO	DXS10101	20208	34.2		33/34.2	34.2
NO	DXS10135	20208	20.1/21.1/22.1		20.1/22.1	22.1
NO	HPRTB	20208	14		12/14	14
SI	DXS7132	20221	14	14	12/14	14
SI	DXS10079	20221	20	20	18/20	20
SI	DXS8378	20243	10/12	10/12	10/12	12
SI	DXS9898	20243	8.3/12	8.3/12	8.3/12	8.3
SI	DXS7423	20243	14/17	14/17	14/17	17
SI	DXS6809	20243	31/33	31/33	31/33	33
SI	DXS7132	20243	12/14	12/14	12/14	14
SI	DXS9902	20243	12/13	12/13	12/13	12
SI	DXS6789	20243	21/22	21/22	21/22	22
NO	DXS9898	20248	8/12		8.3/12	8.3
SI	DXS7132	20259	12/14	12/14	12/14	14
SI	DXS9902	20259	12/13	12/13	12/13	12
Si	DXS10103	20262	16/19		16/19	19
Si	DXS10134	20262	35/37		35/37	37
Si	DXS10074	20262	14/16		14/16	14
Si	DXS10101	20262	33/34.2		33/34.2	34.2
Si	DXS10135	20262	20.1/22.1		20.1/22.1	22.1
Si	DXS10146	20262	28/29		28/29	29
Si	DXS10079	20262	18/20		18/20	20
Si	DXS10148	20262	24.1		24.1	24.1
Si	HPRTB	20262	12/14		12/14	14

NIVEL AVANÇADO: Módulo Forense



M8 / M9

M8

LD 16

No LD 9

M9

LD 4

No LD 20

No contesta 1

Marcador	n	M8-1	M8-2
AMELOGENINA	19	X/Y	X/Y
AMELOGENINA	15	X/Y	
AMELOGENINA	5	X	X/Y

NIVEL AVANÇADO: Módulo Forense



Cromossoma Y

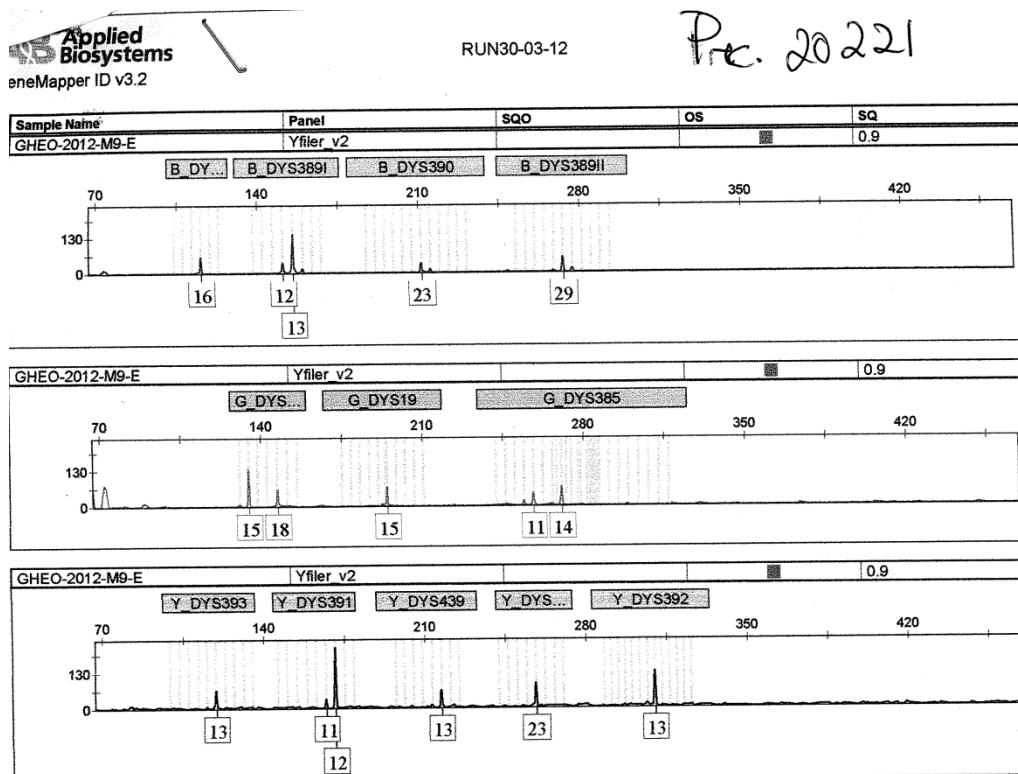
Marcador	M7	M8-1 (Mulher + M4)	M8-2 = M4	M8 (consenso)	M9 cont = M2_2011	M9 cont. Consensuado
DYS_19	-	14	14	14	15	15
DYS_385	-	11/14	11/14	11/14	11/14	11/14
DYS_389_I	-	13	13	13	13	13
DYS_389_II	-	29	29	29	29	29
DYS_390	-	24	24	24	23	23
DYS_391	-	10	10	10	12	12
DYS_392	-	13	13	13	13	13
DYS_393	-	14	14	14	13	13
DYS_437	-	15	15	15	15	15
DYS_438	-	12	12	12	12	12
DYS_439_GATA_A4	-	11	11	11	13	13
DYS_448	-	19	19	19	19	19
DYS_456	-	15	15	15	16	16
DYS_458	-	16	16	16	15	15
DYS_635_GATA_C4	-	23	23	23	23	23
GATA_H4	-	13	13	13	12	12

NIVEL AVANÇADO: Módulo Forense

Cromossoma Y



LD/DE	Precinto	Marcador/Marker	M8-1	M8-2	M9	m8-1 R	m8-2 R	m9-r
SI	20221	DYS_390		24	23/24	24	24	23
SI	20221	DYS_458		16	15/18	16	16	15
no	20266	DYS_393	15		13	14	14	13
No	20264	DYS_392	13		**	13	13	13

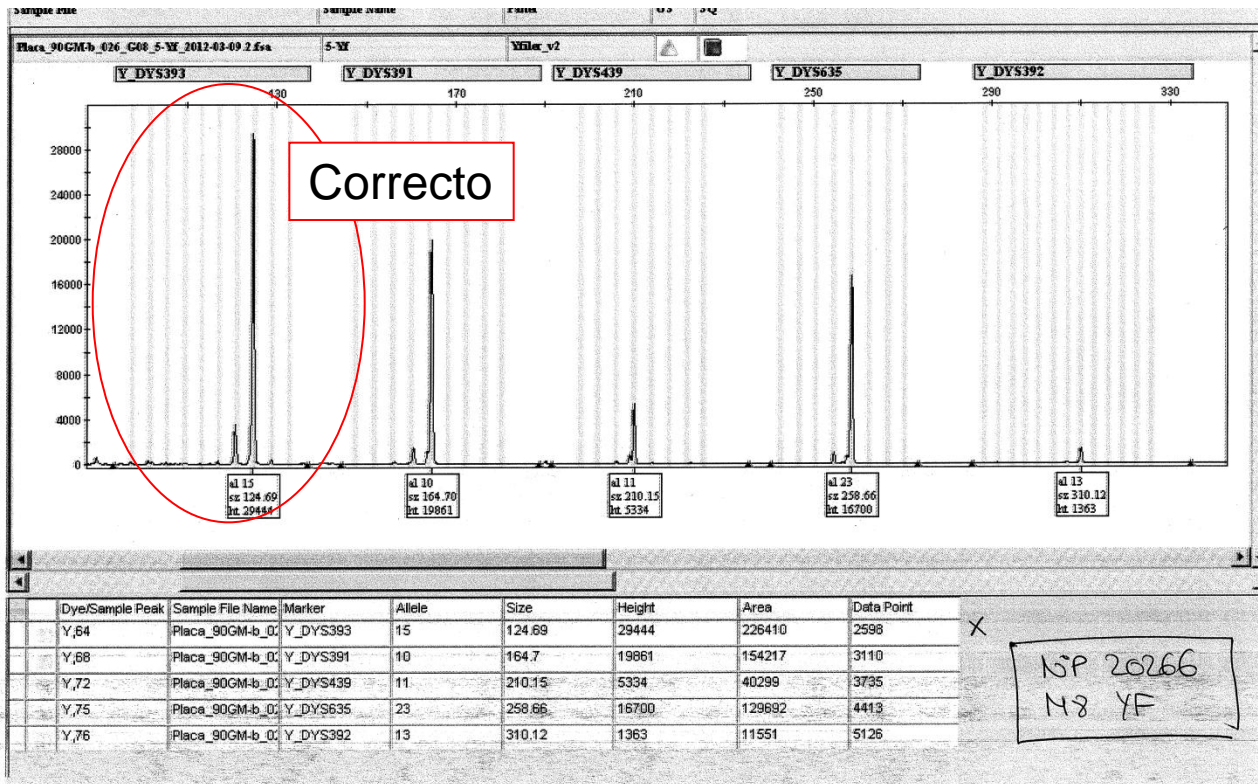


NIVEL AVANÇADO: Módulo Forense

Cromossoma Y



LD/DE	Precinto	Marcador/Marker	M8-1	M8-2	M9	m8-1 R	m8-2 R	m9-r
SI	20221	DYS_390		24	23/24	24	24	23
SI	20221	DYS_458		16	15/18	16	16	15
no	20266	DYS_393	15		13	14	14	13
No	20264	DYS_392	13		**	13	13	13



NIVEL AVANÇADO: Módulo Forense



Cromossoma X

Marcador/Marker	M8-1 (Mulher + M4)	M8-2 = M4	M8 (consenso)	M9 cont = M2_2011	M9 cont. Consensuado
DXS8378		10	10/12	10	10
DXS9898		12	12/13	11	11
DXS7133		11	11	11	11
GATA31E08		12	11/12	9	9
GATA172D05		11	8/11	6	6
DXS7423		14	14/16	13	13
DXS6809		32	32/34/35	34	34
DXS7132		12	12/13	14	14
DXS9902		11	11/12/13	11	11
DXS6789		20	20/21	22	22
DXS10103		16	16/19	18	18
DXS10134		32	32/36	35	NC
DXS10074		15	NC	18	NC
DXS10101		31	31/31.2/32	30	NC
DXS10135		22	19/22/31	20	NC
DXS10146		27	27/29/30	29	NC
DXS10079		17	17/18/19	18	NC
DXS10148		28.1	18/19/28.1	25.1	NC
HPRTB		12	12/15	13	NC

NIVEL AVANÇADO: Módulo Forense



Cromossoma X

		M8-1	M8-2	M8 (consenso)
DXS9898	20209	8.3/12	12	12/13
DXS6789	20209	19/20	20	20/21
GATA31E08	20266	12/12		11/12
DXS7423	20221	14	14	14/16
DXS10074	20221	13/15	15	13/15/16
DXS10074	20261	13/15/16/18		13/15/16
DXS10135	20261	19/21/22/31		19/22/31

20254

DXS8378	10
DXS9898	12
DXS7133	11
GATA31E08	12
GATA172D05	11
DXS7423	14
DXS6809	32
DXS7132	12
DXS9902	11
DXS6789	20

M8-1

=M4 = M8-2

NIVEL AVANÇADO: Módulo Forense



Cromossoma X

Precinto	M8-1	M8-2	M9
----------	------	------	----

DXS10134

20170	32/36	32	35
20221	32/36	32	
20261	32/36		
20264	32/36		35
20278	32/36	32	

Correcto ou consenso?

DXS10074

20170	13/15/16	15	18
20221	13/15	15	
20261	13/15/16/18		18
20264	13/15/16		18
20278	13/15/16	15	

Marcador/Marker	M9 cont = M2_2011	M9 cont. Consensuado
DXS10134	35	NC
DXS10074	18	NC
DXS10101	30	NC
DXS10135	20	NC
DXS10146	29	NC
DXS10079	18	NC
DXS10148	25.1	NC
HPRTB	13	NC

NIVEL AVANÇADO: Módulo Forense



Cromossoma X

DXS6809	M8-1	M8-2	M9
20171	32/34/35	32	34
20244	32/34/35		
20254	32		
20259	34/35	32	
20264	32/34/35		34
20266	32/34/35		34
20268	34/35	32	34
20278	32/34/35	32	

Que é que estamos a avaliar?

LD: sim ou não?

Metodologia LD: separa bem ou não?

Indica correctamente ou não? nomenclatura