

XVIII Jornadas del GHEP SEVILLA 2013



SPANISH AND PORTUGUESE-SPEAKING
WORKING GROUP OF ISFG

Discusión de resultados Cromosomas Sexuales

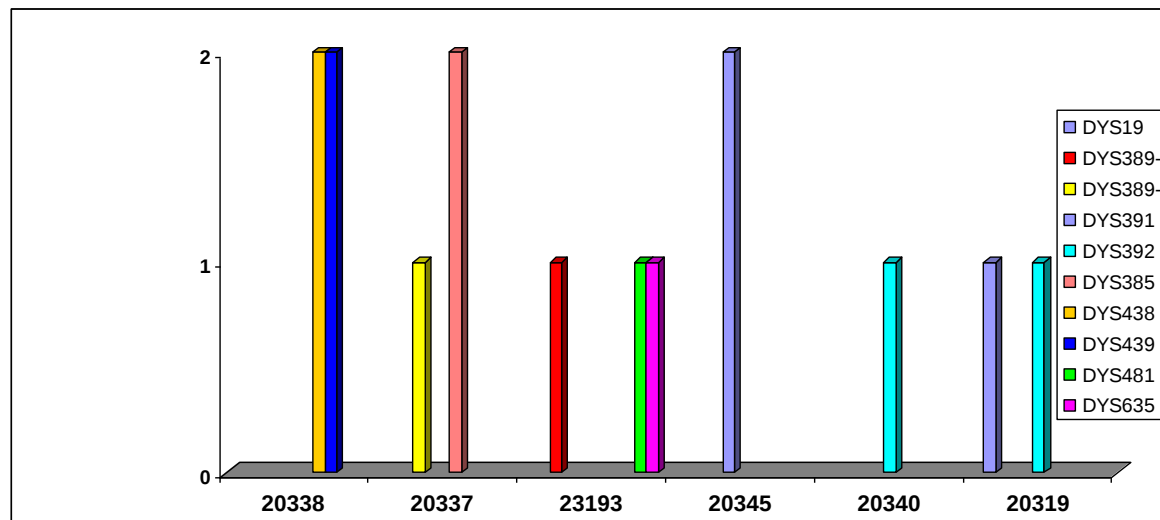


**Dra. Andrea Sala
Servicio de Huellas Digitales Genéticas-FFyB
Universidad de Buenos Aires-Argentina**

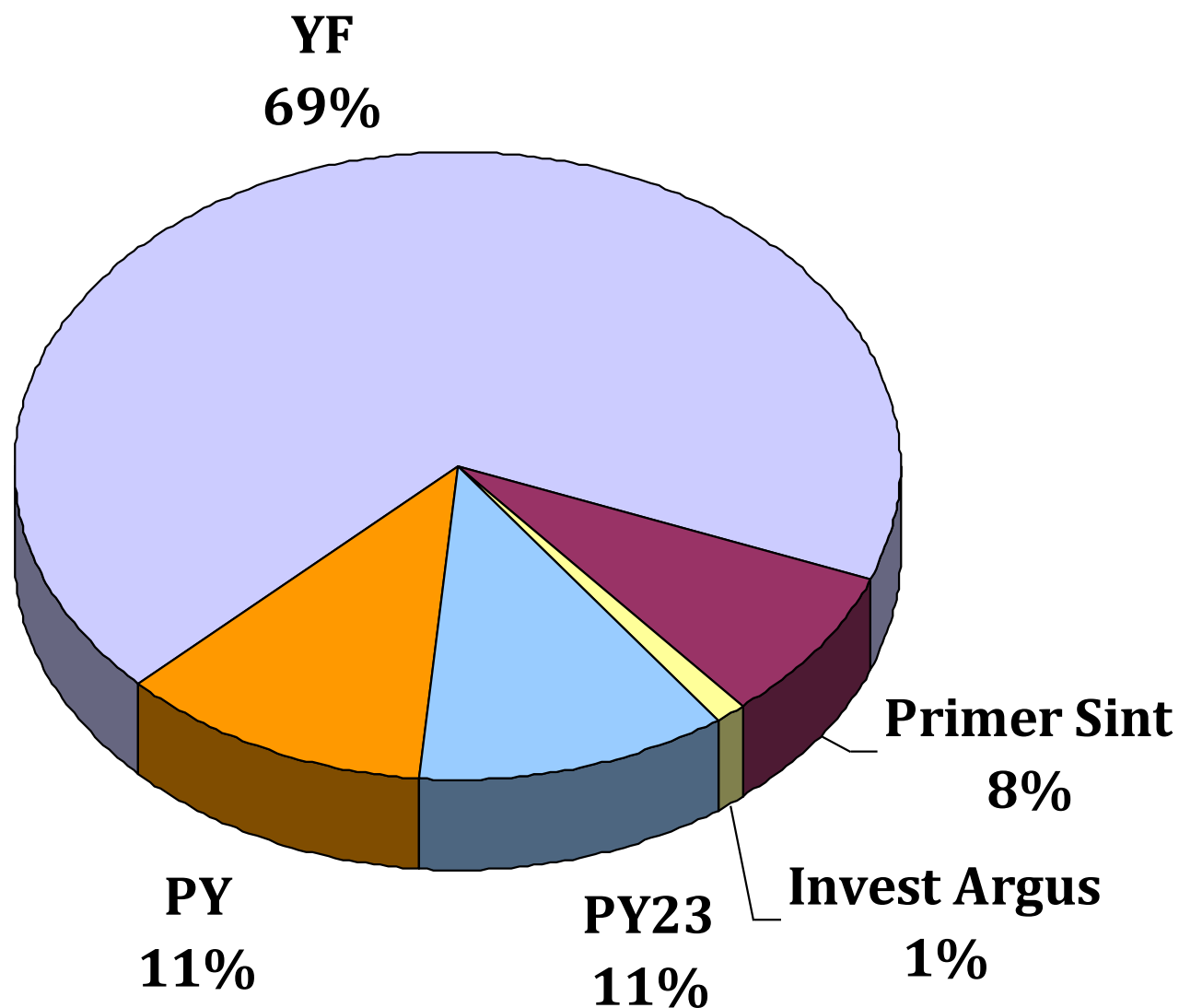
Cromosoma Y

Básico (M1 y M3)

- ✓ Número de Laboratorios: 91
- ✓ Número de discrepancias: 15
- ✓ Número de errores de transcripción: 4
- ✓ Error de nomenclatura: 2 (- x /)
- ✓ Empleo de nomenclatura ISFG (GATA H4): 2



Metodología

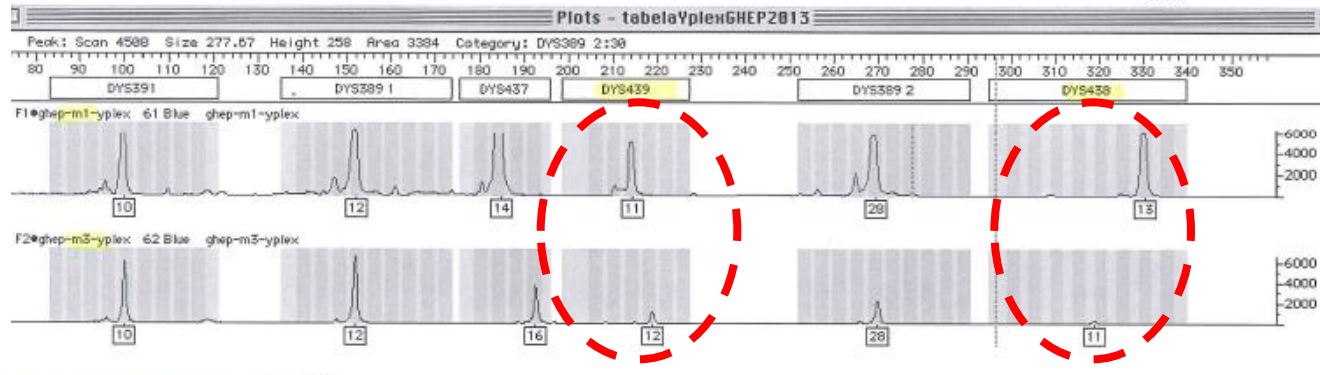


PRECINTO 20338 (PL01; ABI 310)

20338



SPANISH AND PORTUGUESE-SPEAKING
WORKING GROUP OF ISFG



DYS439

DYS438

M1 (12)

M1 (12)

M3 (13)

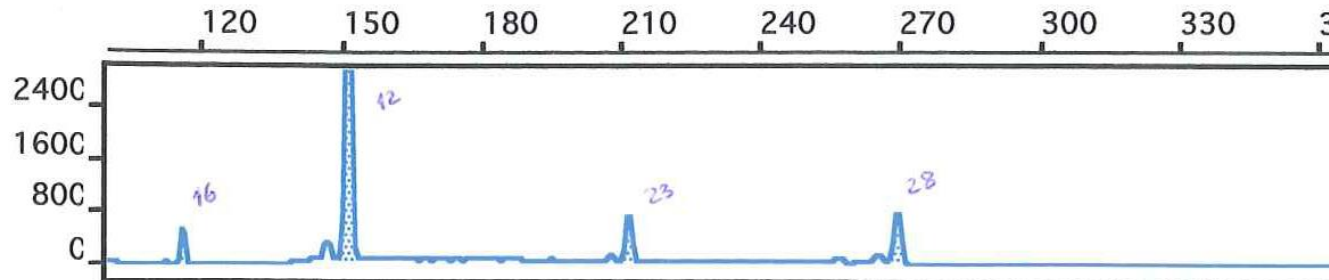
M3 (10)



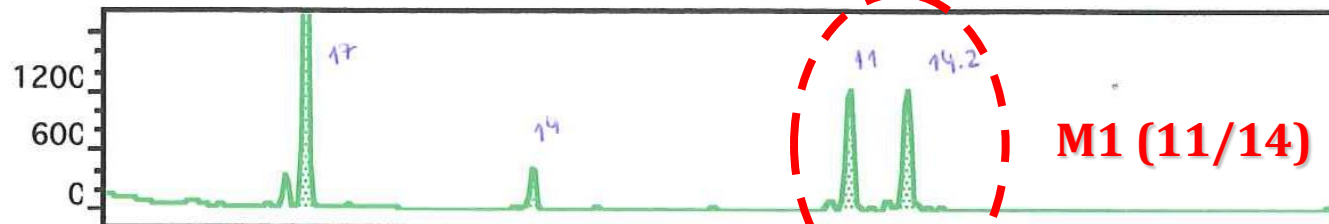
GeneScan® 3.1.2

20337

GeneScan™ Project-22.03.13 Displa

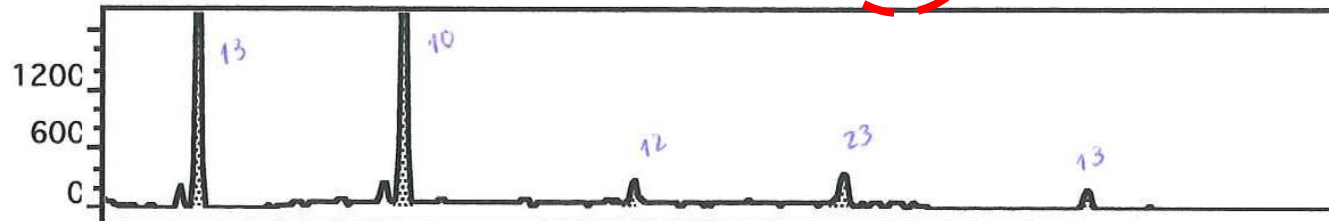


1B:20337 M1 2013 YFILER / DYS456, DYS389I, DYS390, DYS389II



1G:20337 M1 2013 YFILER / DYS458, DYS19, DYS385

M1 (11/14)



1Y:20337 M1 2013 YFILER / DYS393, DYS391, DYS439, DYS635, DYS392

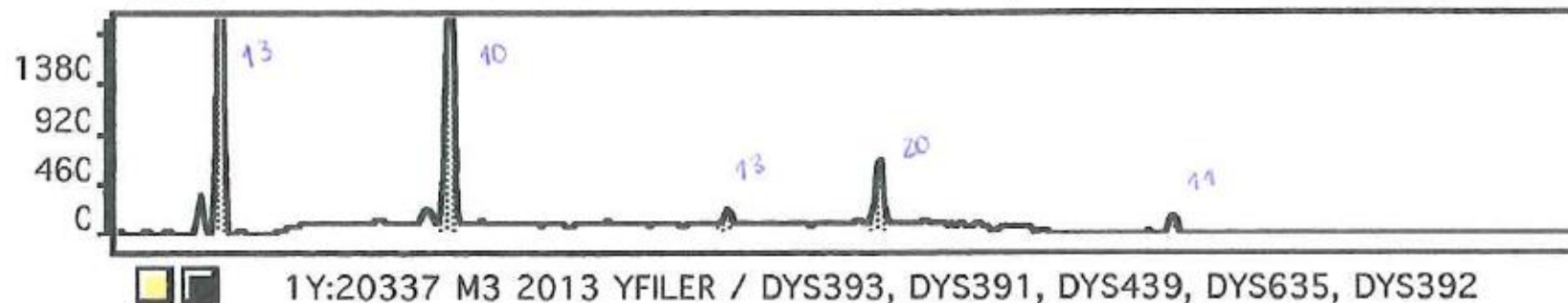
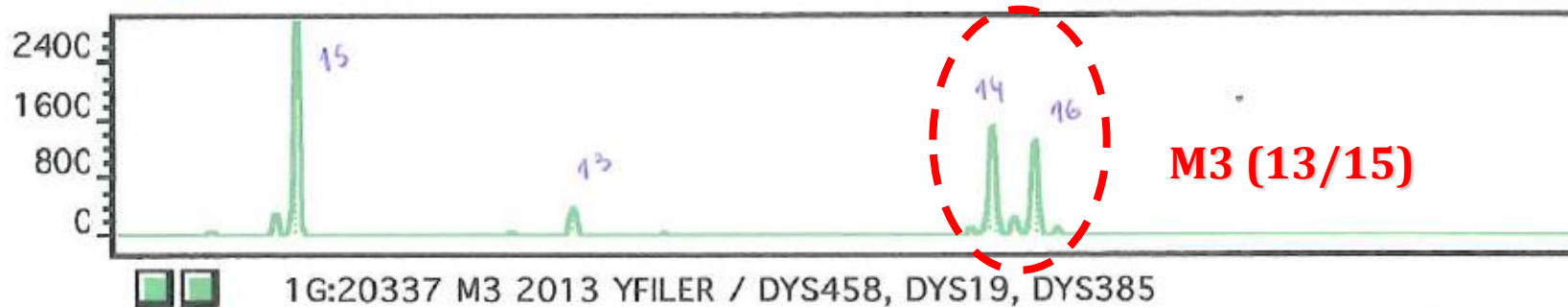
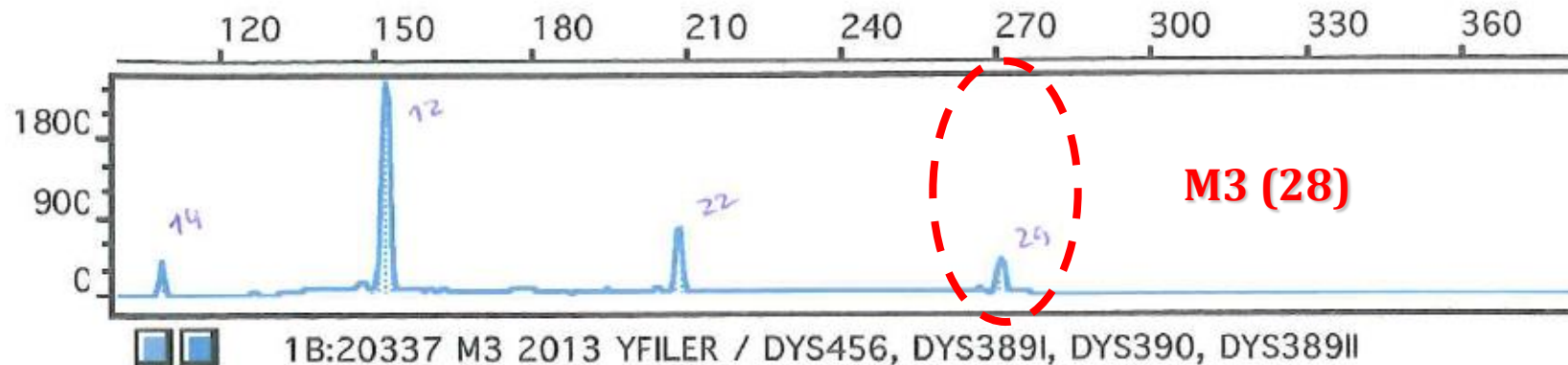
PRECINTO 20337 (YF; ABI 310)



GeneScan® 3.1.2

20337

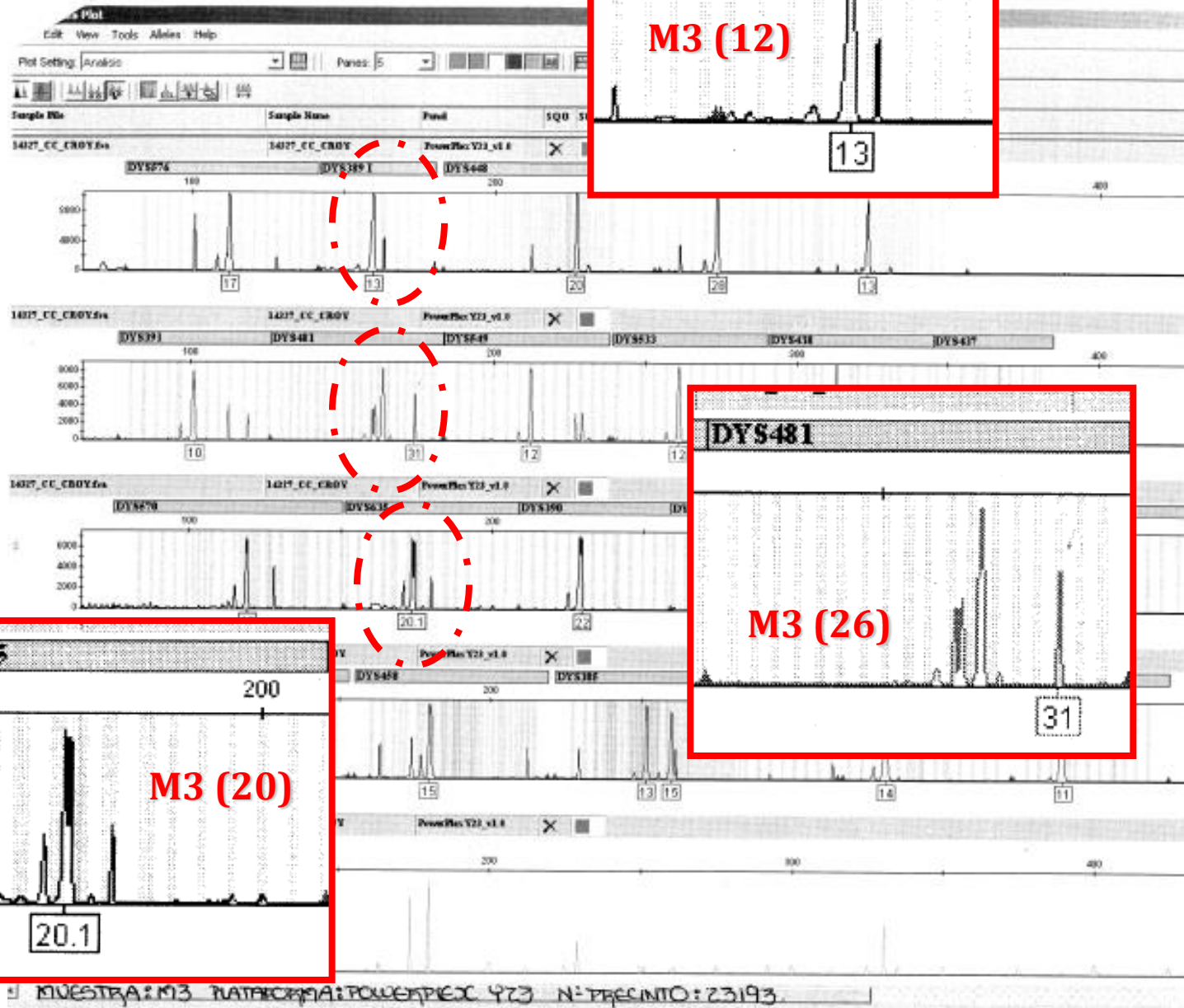
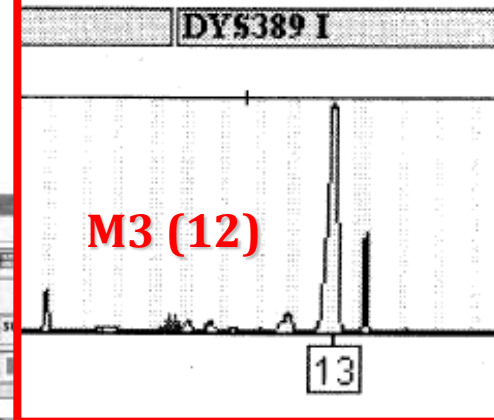
GeneScan™ Project-04.04.13 Display-1



144C

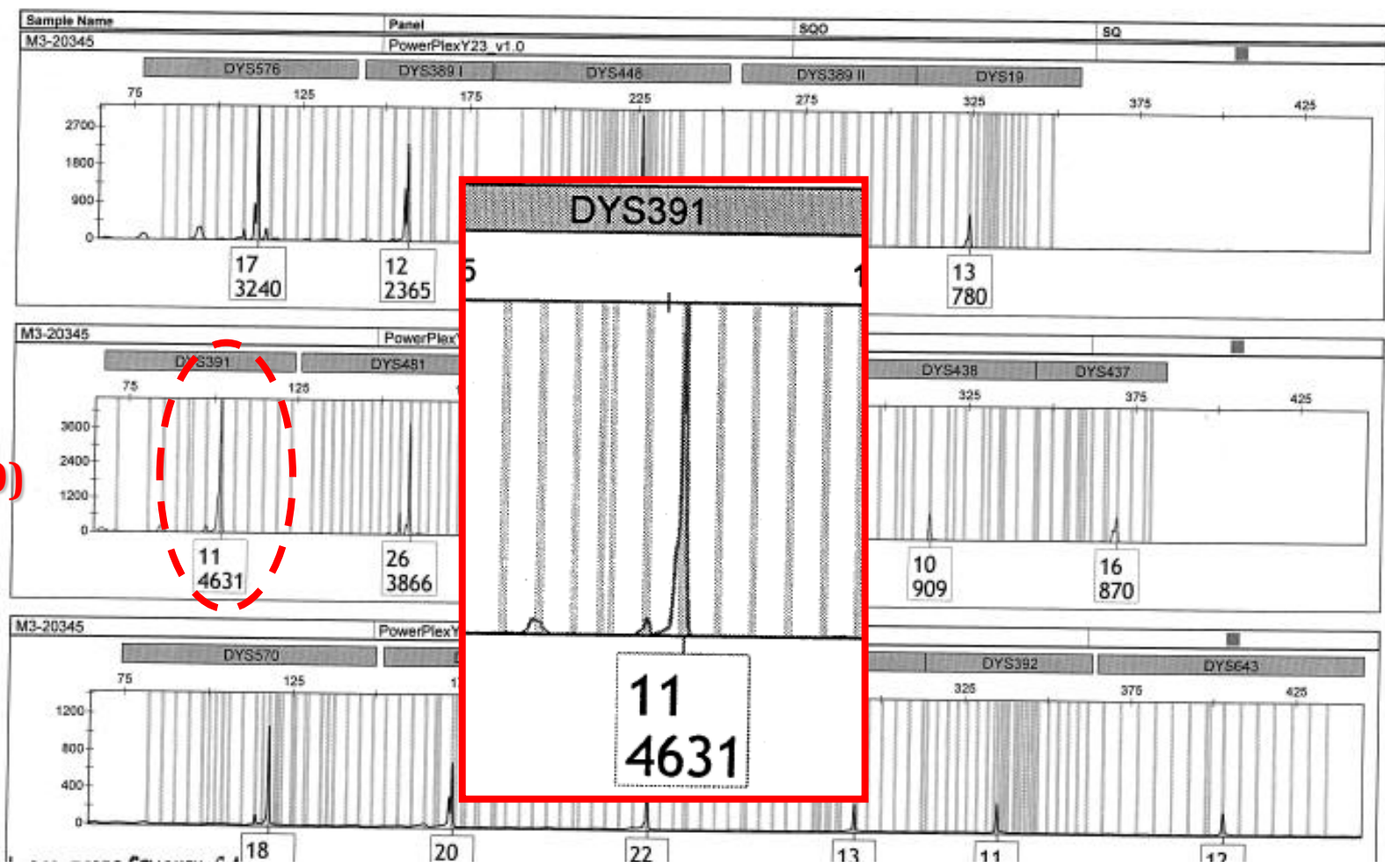
PRECINTO 20337 (YF; ABI 310)

PRECINTO 23193 (PY23; ABI 3100)



MUESTRA: M3 PLATAFORMA: POWERPIX 473 N° PRECINTO: 23193





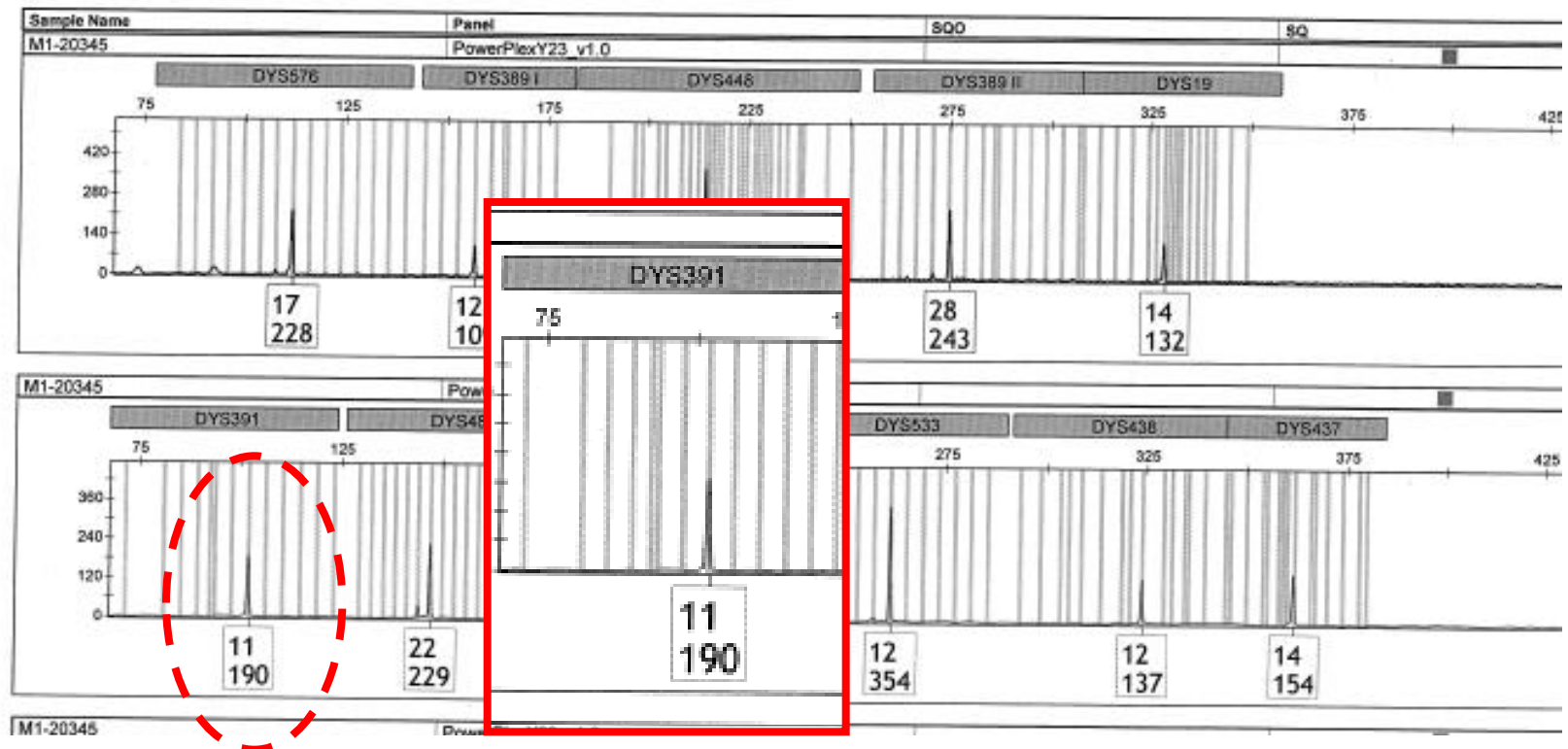
M3 (10)

PRECINTO 20345 (PY23; ABI 3130)

PRECINTO 20345 (PY23; ABI 3130)

Applied Biosystems
GeneMapper ID v3.2

EJERCICIO GHEP 2013 Y23

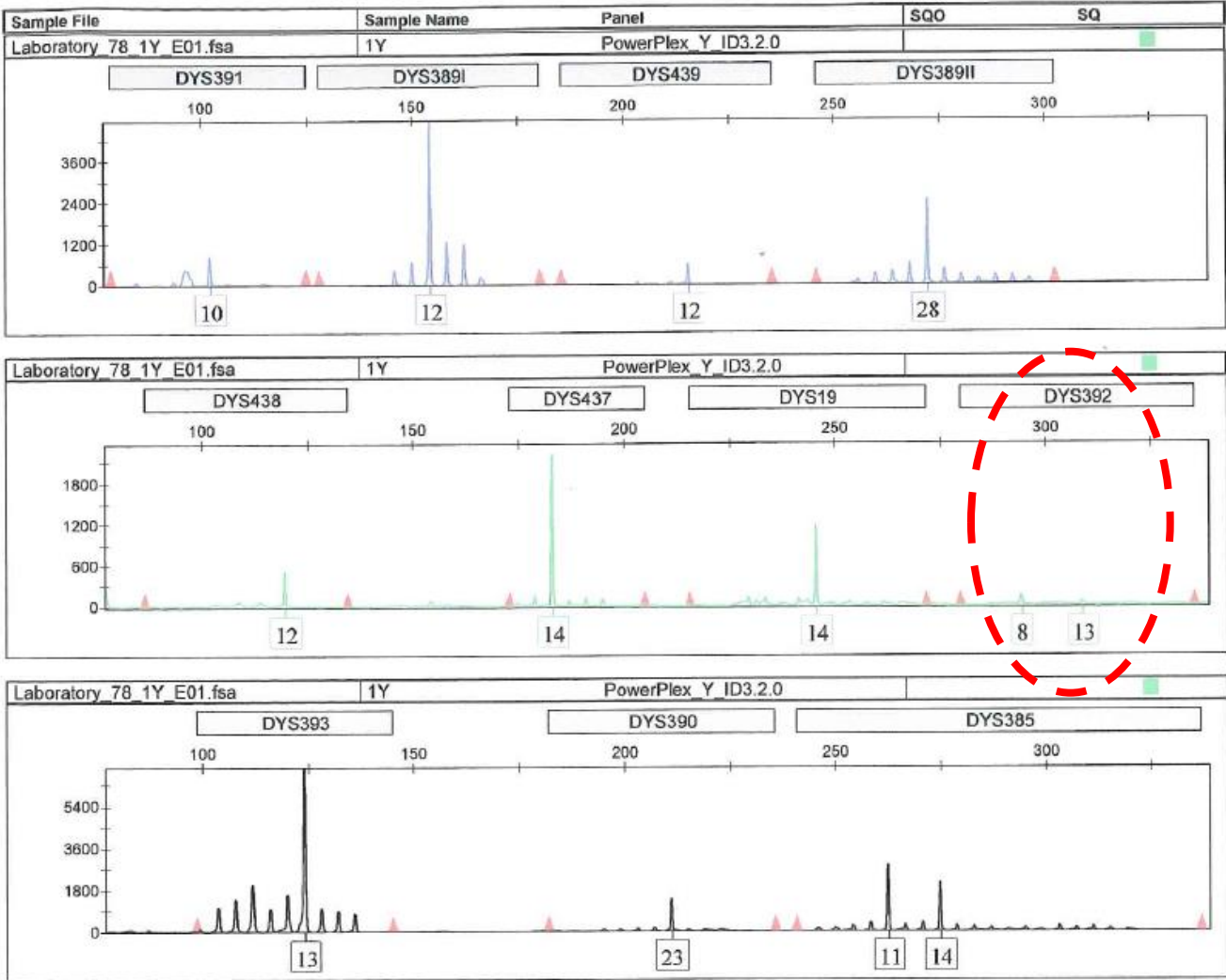


M1 (10)

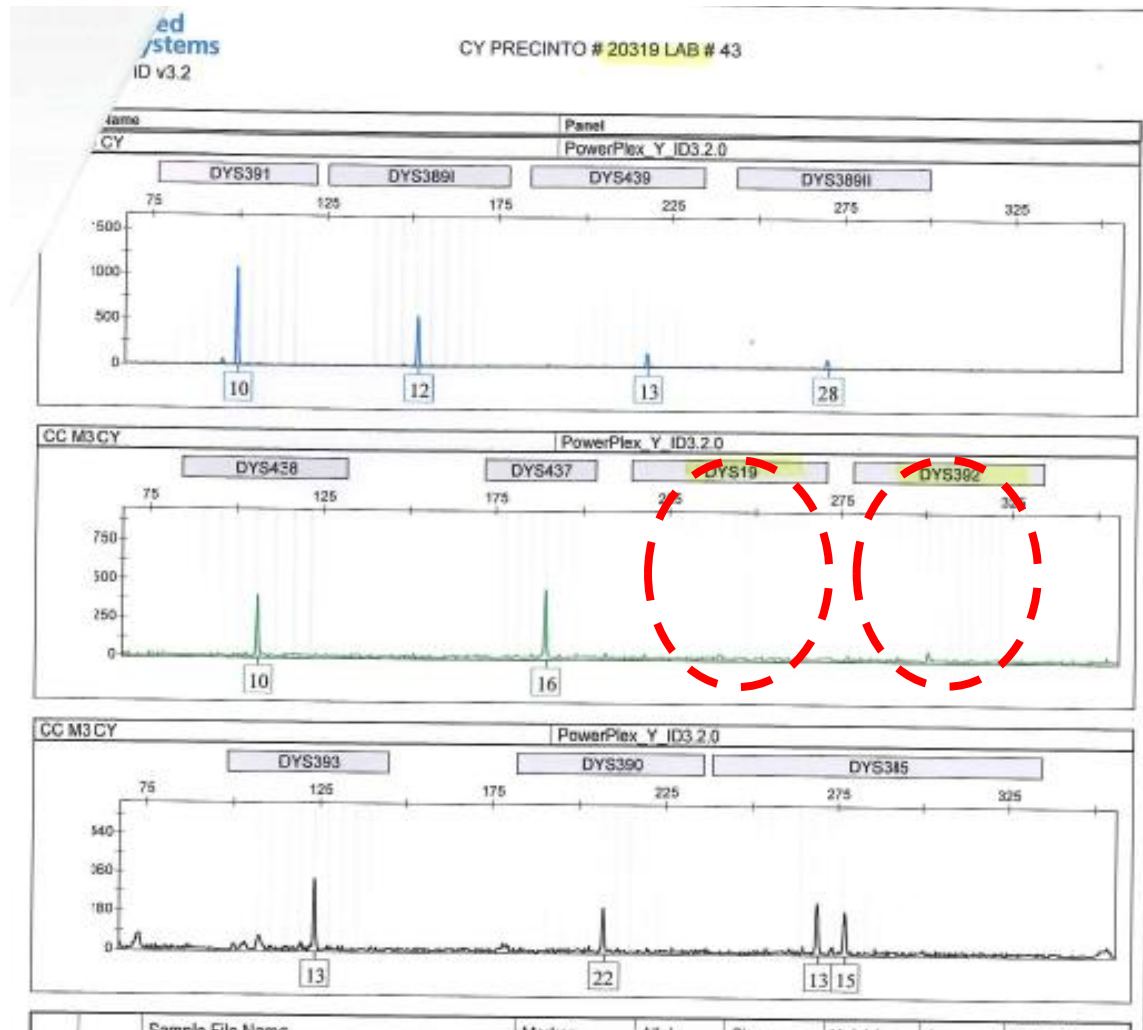
M1 | 20340

Applied Biosystems
GeneMapper ID v3.2

Powerplex Y STRs



M1 (13)



PRECINTO 20319 (PPY; ABI 310)

PRECINTO 20304 (YF, PPY; ABI 3130/xl)



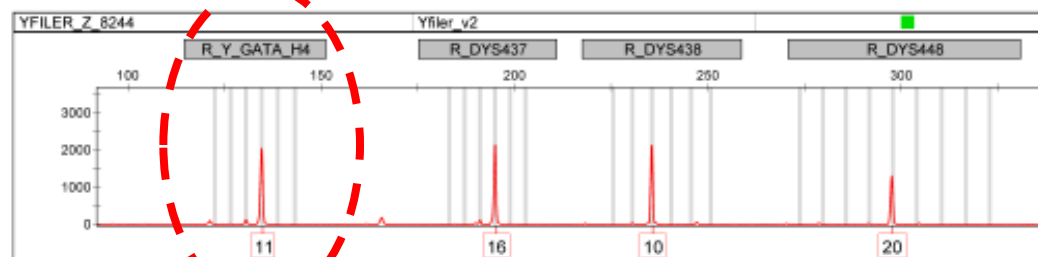
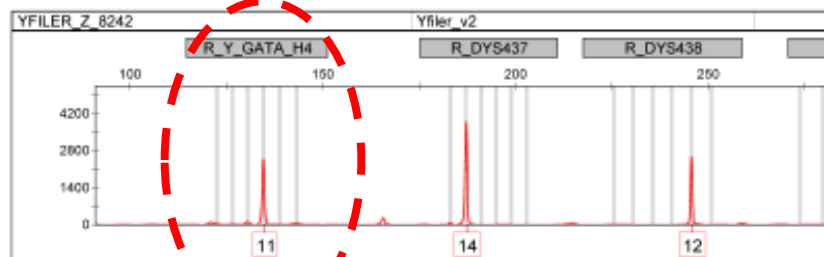
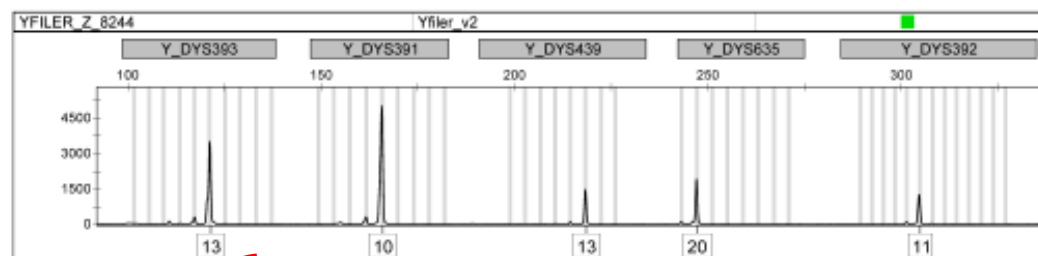
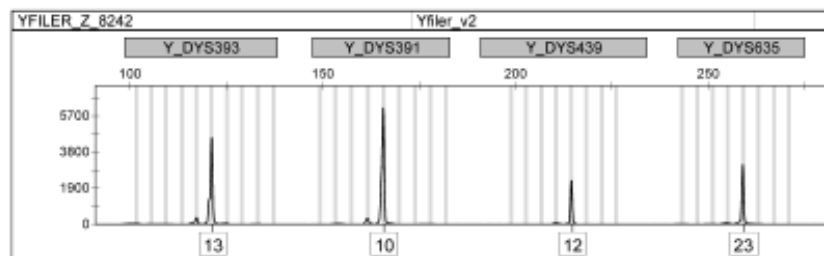
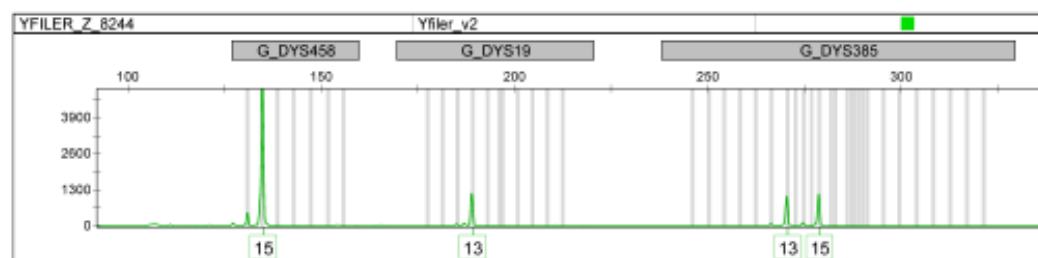
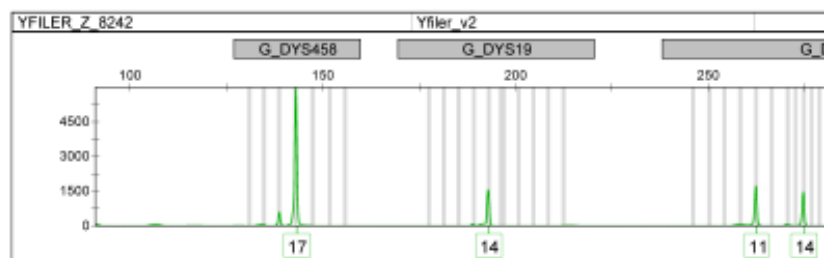
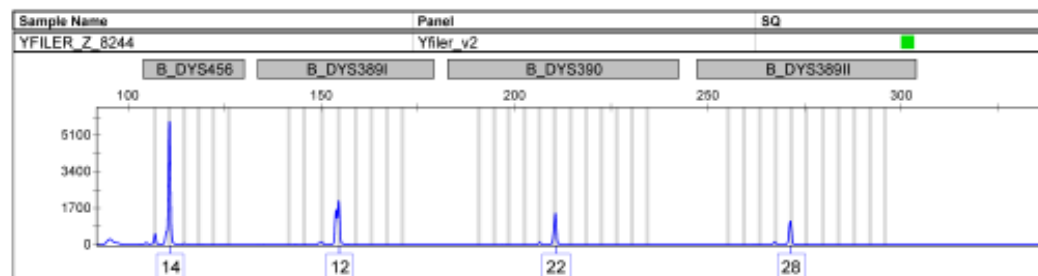
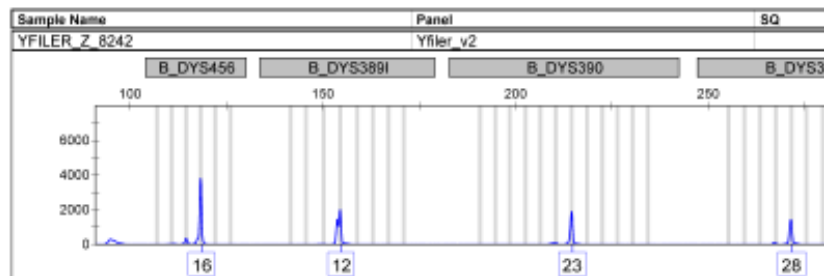
Biosystems
GeneMapper ID v3.2

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GeneMapper ID v3.2

M1 Precinto 20304

M3 Precinto 20304



M1 (11) 20*

M3 (11) 20*



R44: 114256 haplotypes

Search

Analyse

Research

Loci

References

Amelogenin Y

deletions

Metapopulations

YSNPs

Contribute

Meet

Y-GATA-H4

General Informations

»There are differences in allele designations at the GATA H4 marker between those recommended in the Applied Biosystems AmpF/STR® Yfiler™ polymerase chain reaction amplification kit (Applied Biosystems, Foster City, CA, United States) and the ISFG recommendations. The nomenclature for the GATA H4 marker in the Yfiler kit is based on the allele repeat structure defined by the National Institute of Standards and Technology Standard reference material (SRM) 2395 and the work of [Butler et al. 2002](#). In the Yfiler kit the variable core repeat (TAGA) is used to designate the alleles (such as alleles 8–13 in the allelic ladder of the Yfiler kit) (see [NIST Y-STR Web site](#) and [Mulero et al. 2006](#)). Furthermore, the Yfiler kit primers amplify the region now designated as the GATA H4.1 locus as defined by [Gusmao et al. 2002](#) and recommended by the ISFG DNA Commission ([Gusmao et al. 2006](#)). The GATA H4.1 locus structure consists of a core repeat region designated as AGAT and nonvariable tetranucleotide repeats ((AGAT)₄ CTAT(AGAT)₂ (AGGT)₃ (AGAT)_n) that are considered for allele number designation under the ISFG recommendations. Thus, there is a difference in allele nomenclature that depends on whether or not the nonvariable region is included.« (taken from [Mulero et al. 2006](#))

This database is using the Applied Biosystems AmpF/STR® Yfiler™ nomenclature. In order to convert the allele nomenclature according to the ISFG recommendations ([Gusmao et al. 2006](#)) add +9 repeat units ([Mulero et al. 2006](#)).

GATA H4 (YF+16)



GATA H4.1 (YF+9)



GATA H4.2

Y-File - NIST (TAGA) (Butler et al, 2002, For. Sci. Int.)

ISFG (YF+9) (Gusmao et al, 2002 y 2006, For Sci Int.)

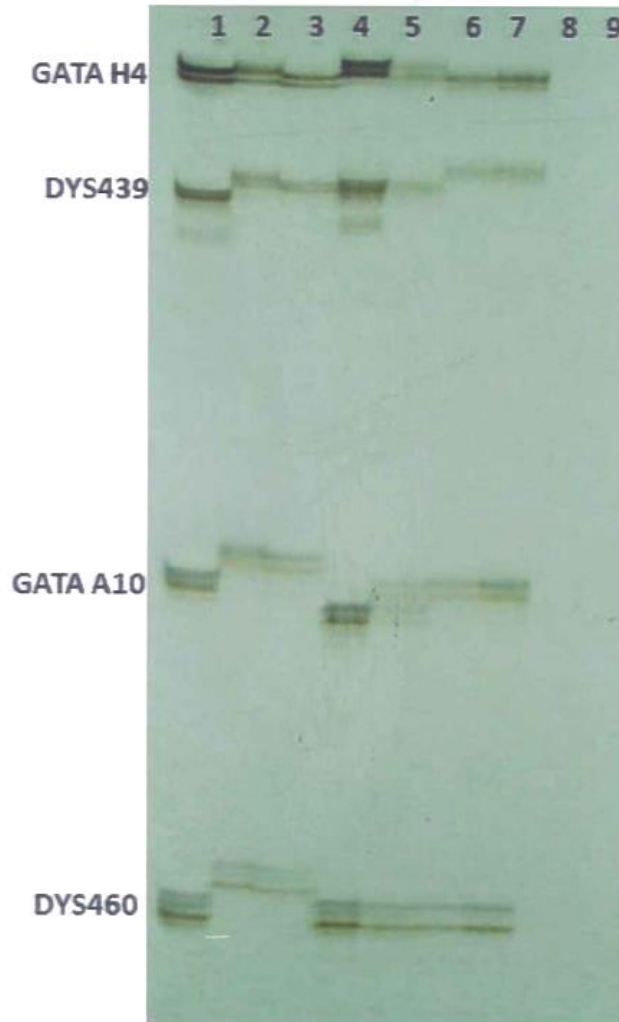
(Mulero et al, 2006, J. For Sci.)

XVI Jornadas GHEP-ISFG (Austria)



STRs CROMOSOMA Y

GEPY II



Muestras:

1. JJ (+) LAB
2. M1
3. M1
4. LM (+) LAB
5. Escalera
6. M3
7. M3
8. (-) EXT
9. (-) PCR

GATA H4 M1 : 27
M3 : 27

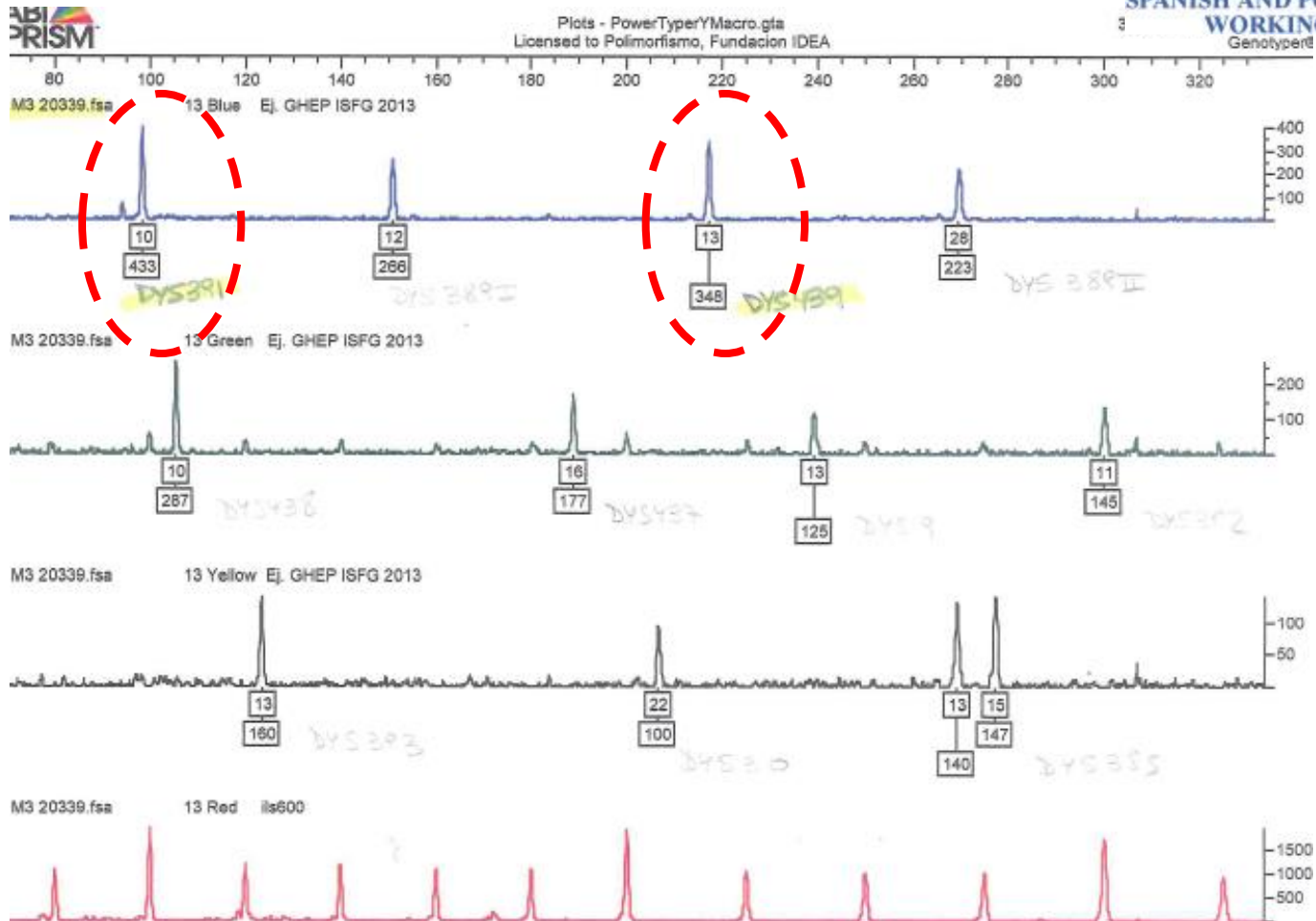
N° de Precinto

20318

N° laboratorio

41

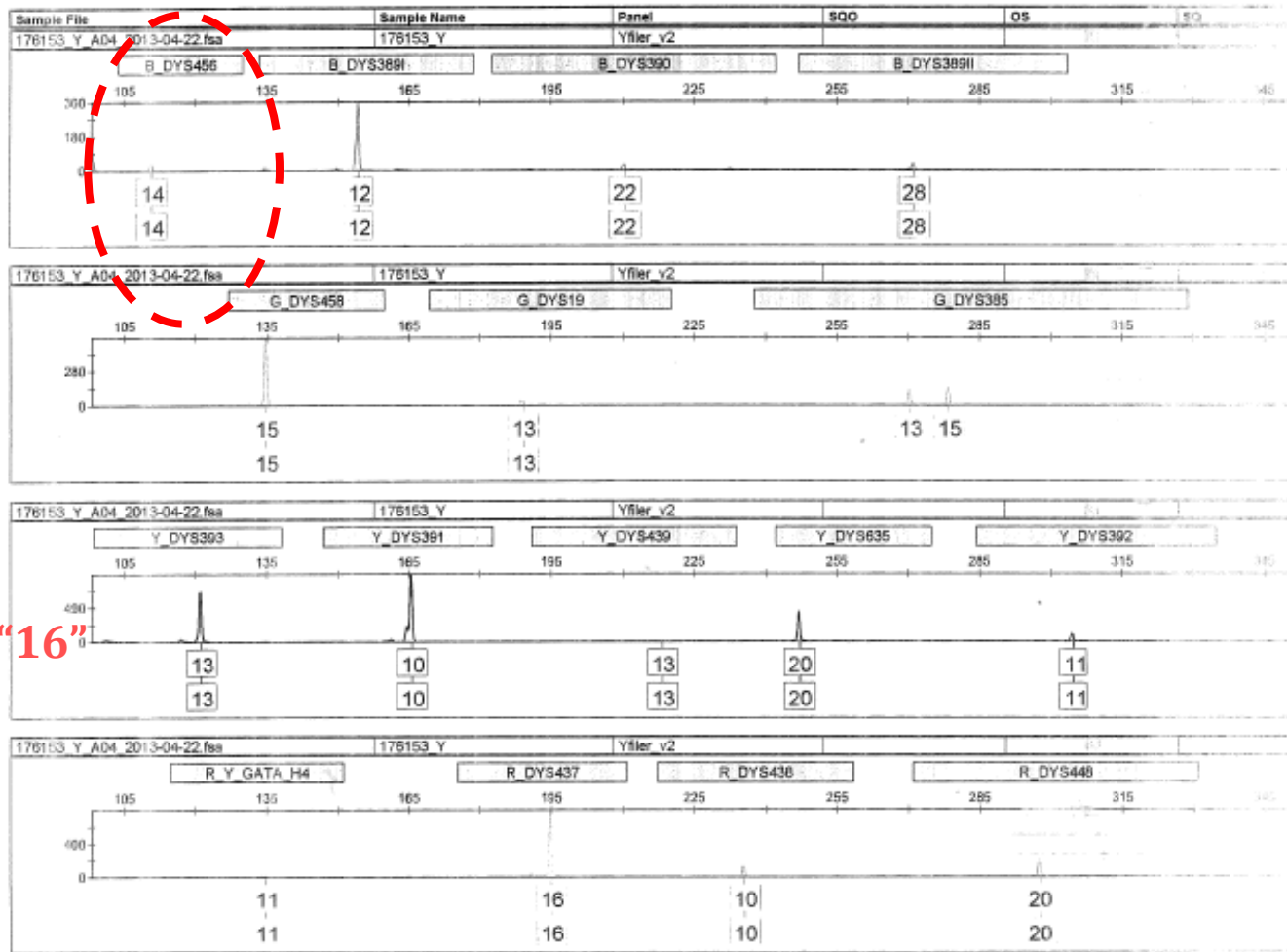
PRECINTO 20318 (PL00; tinción Ag)



PRECINTO 20339 (PY; ABI 310)

**Error de transcripción
DYS391 13
DYS439 10**

GeneMapper ID v3.2



Mon Apr 22, 2013 10:58AM, COT

Printed by: gmid

20314

Page

DYS456

M3 (14)

Error T
informa "16"

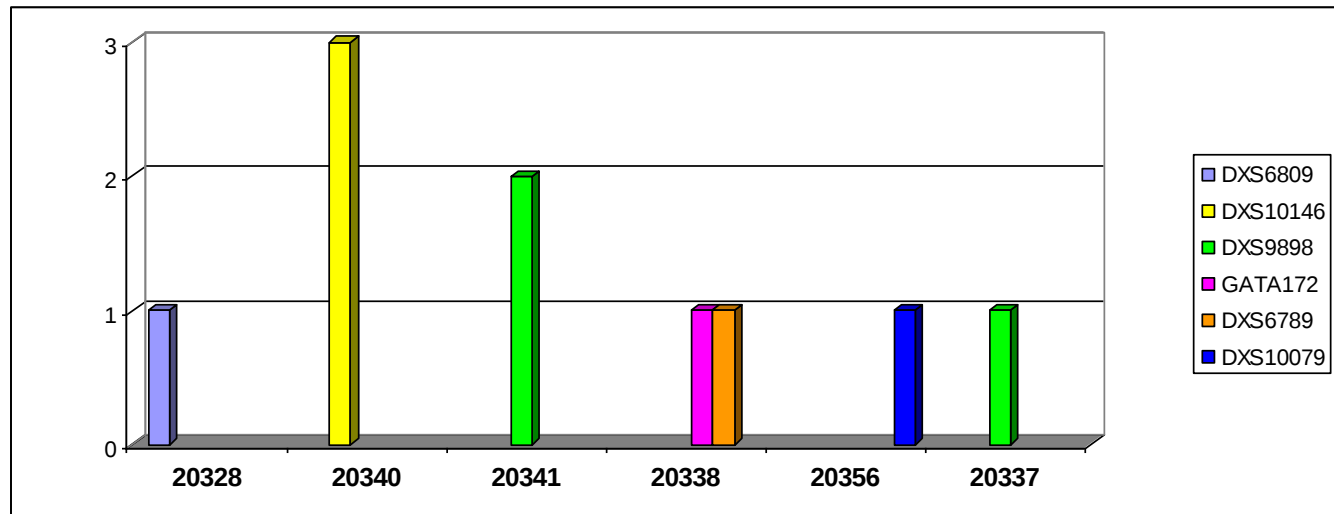
PRECINTO 20314 (YF)

Cromosoma X

Básico (M1, M2 y M3)

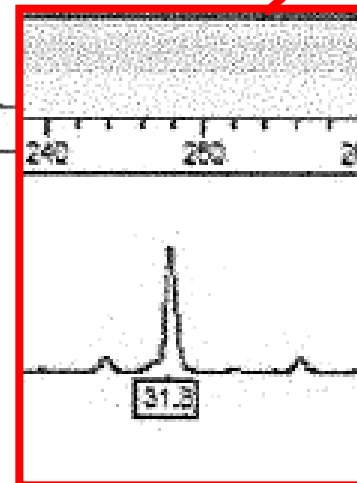
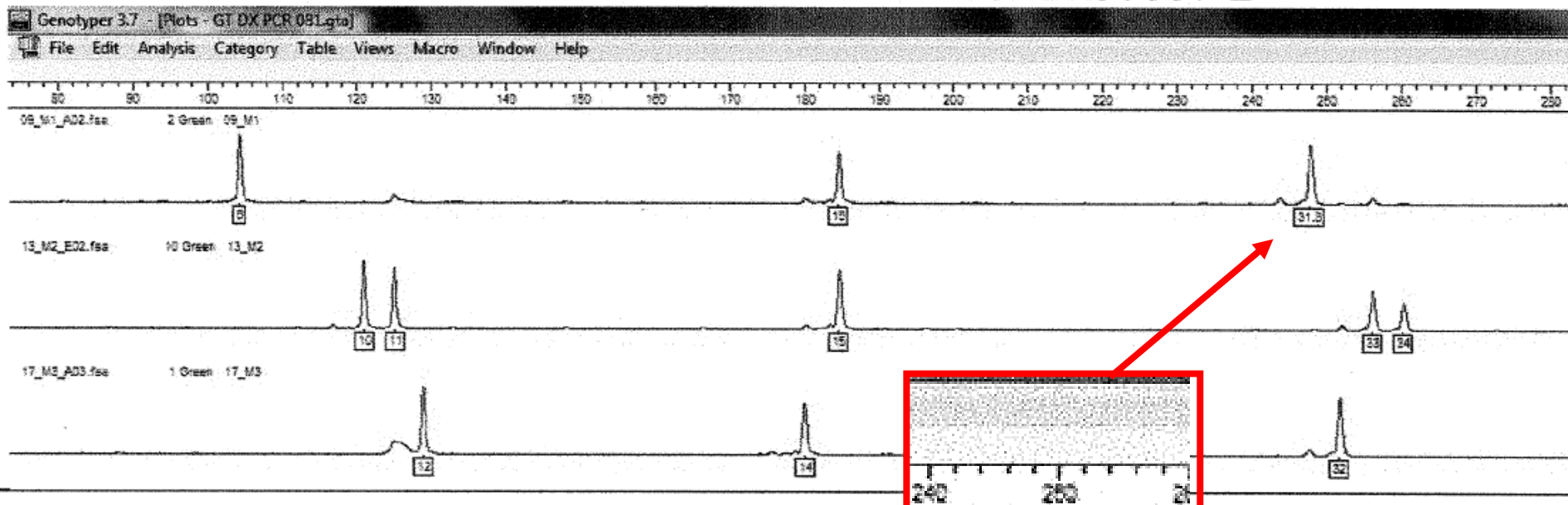
- ✓ Número de Laboratorios: 39 (61% X-STRs; 39% Invest Argus)
- ✓ Número de discrepancias: 10
- ✓ Número de errores de transcripción: 2

Laboratorios que presentaron discrepancias



PRECINTO 20328 (Pr sint; ABI 3130/3130 xl)

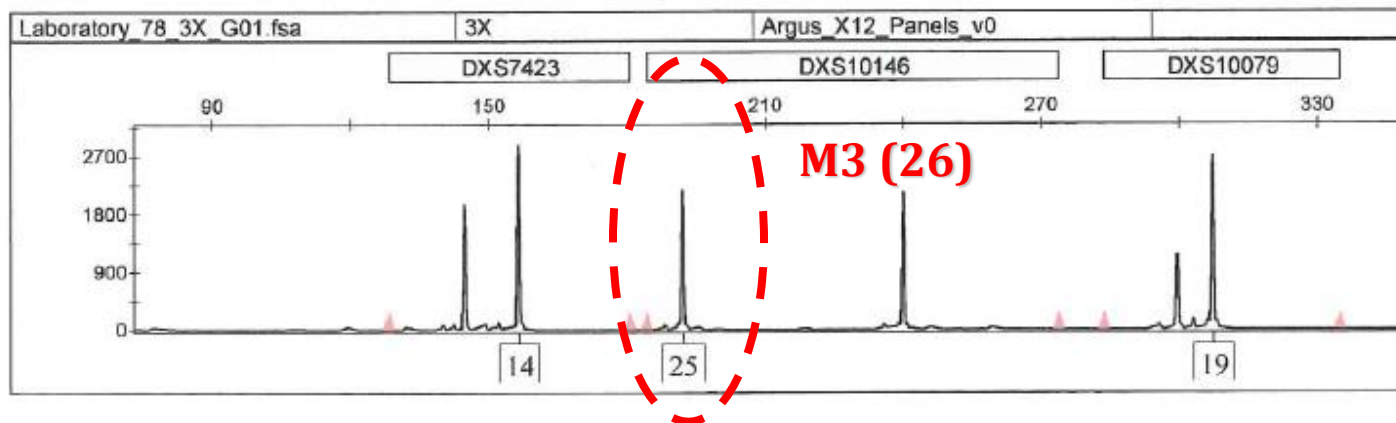
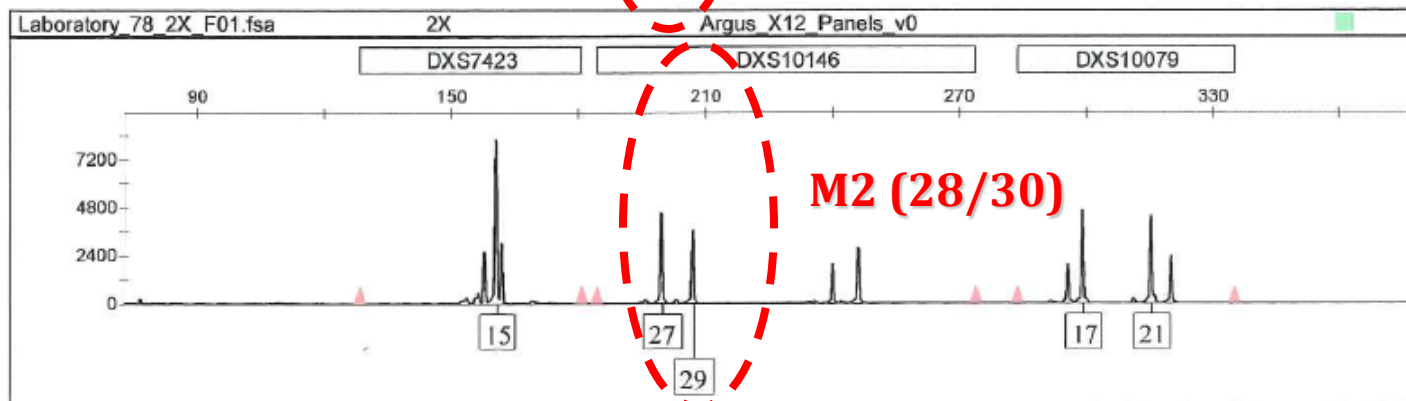
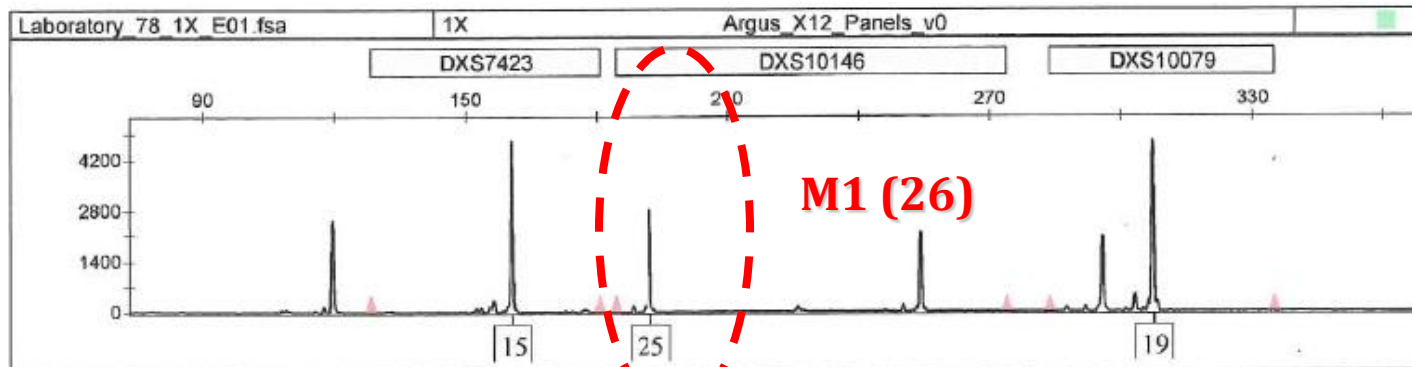
GREEN: GATA172D05/ DXS7423/ DXS6809 2



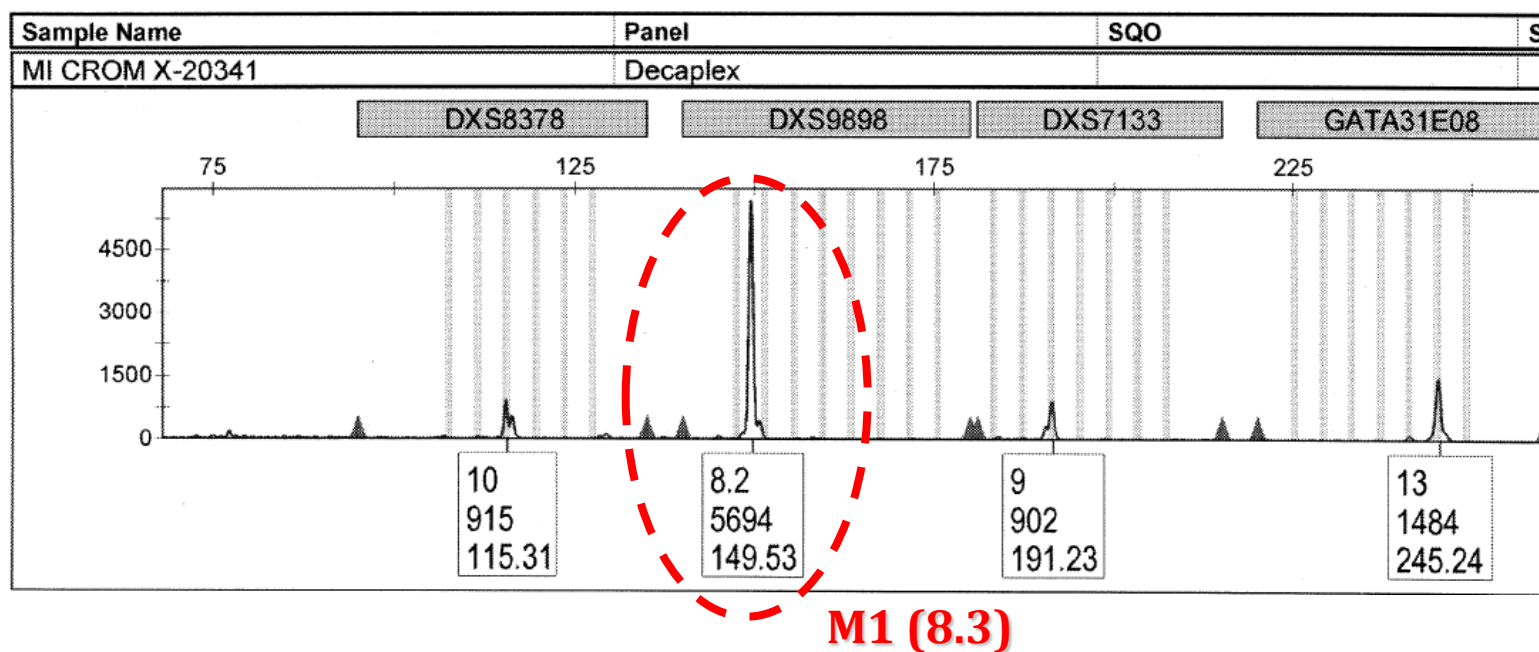
M1 (31)

PRECINTO 20340 (Argus X12; ABI 3130/3130 xl)

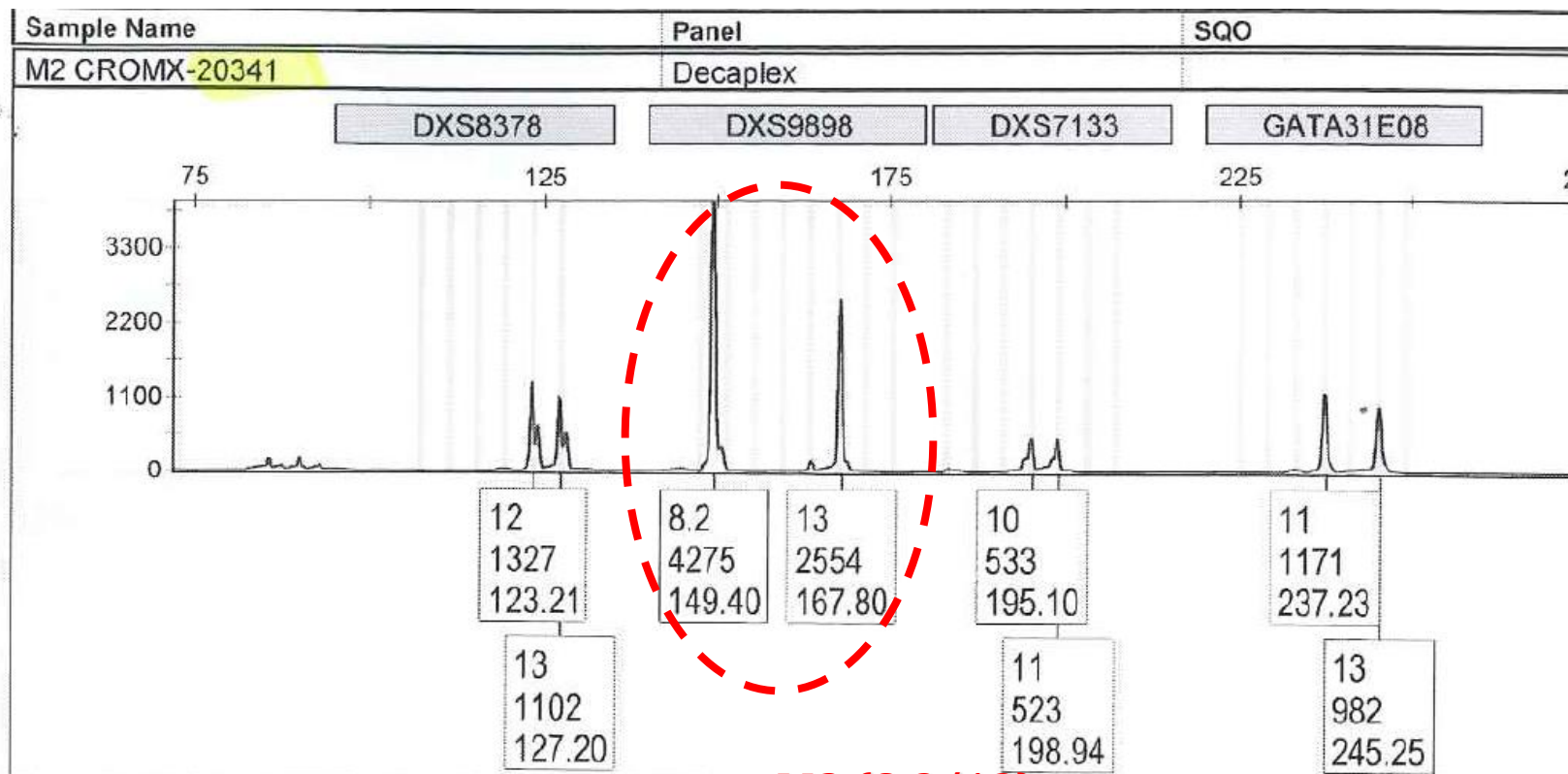
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PRECINTO 20341 (Decaplex; ABI310)



PRECINTO 20341 (Decaplex; ABI310)



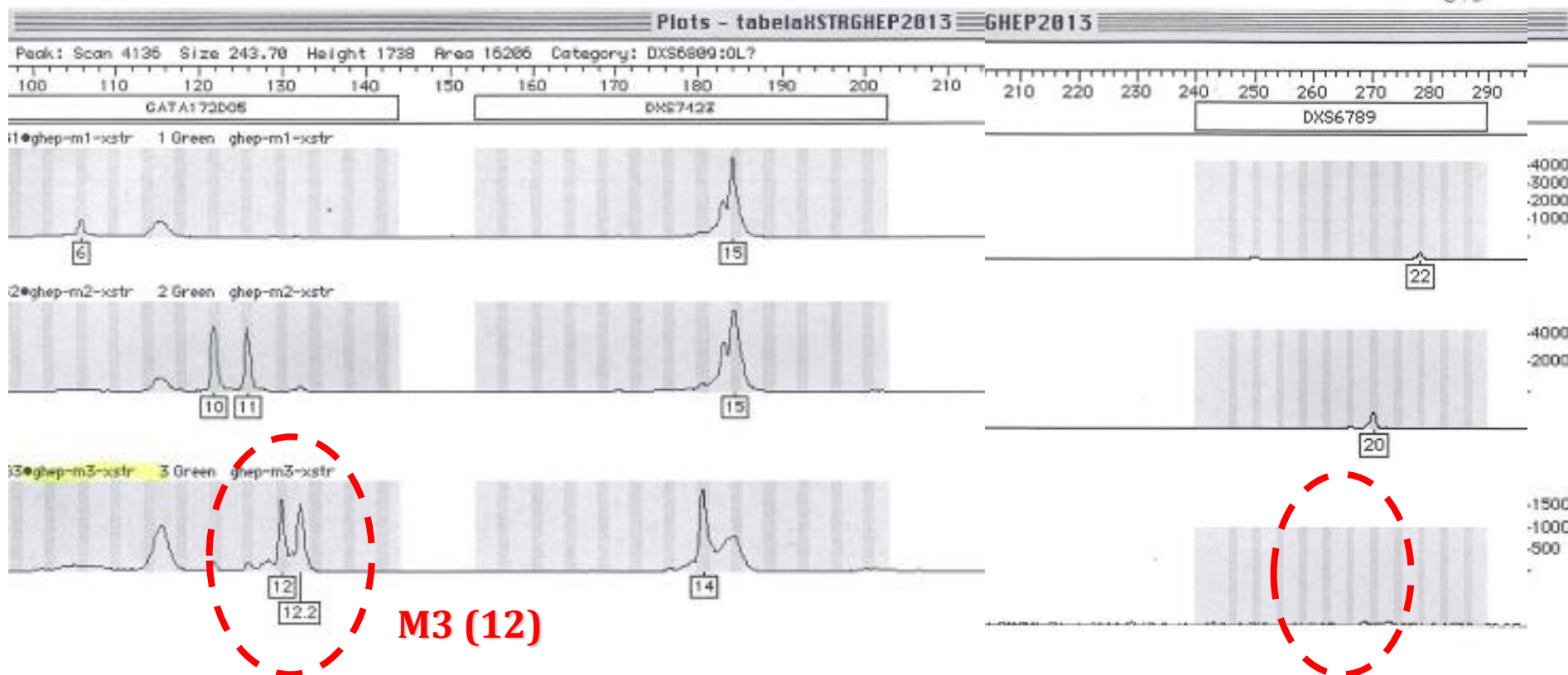
M2 (8.3/13)

PRECINTO 20338 (X-Strs; ABI 310)

20338

20338

073

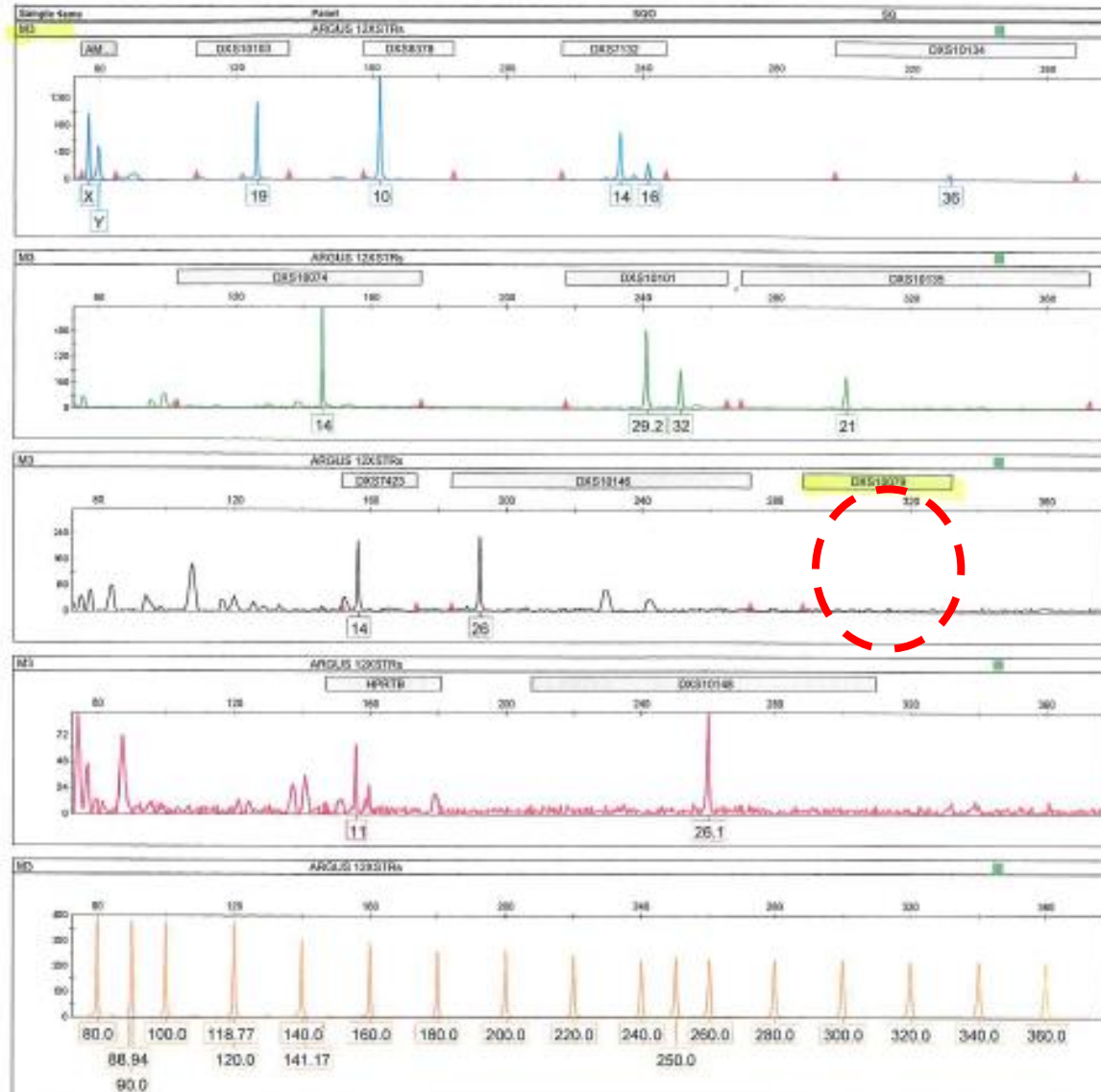


PRECINTO 20356 (Argus X12; ABI 3130/3130 xl)

Applied Biosystems
GeneMapper ID v3.2

GHEP-ISFG 20356 X-STRs

20356



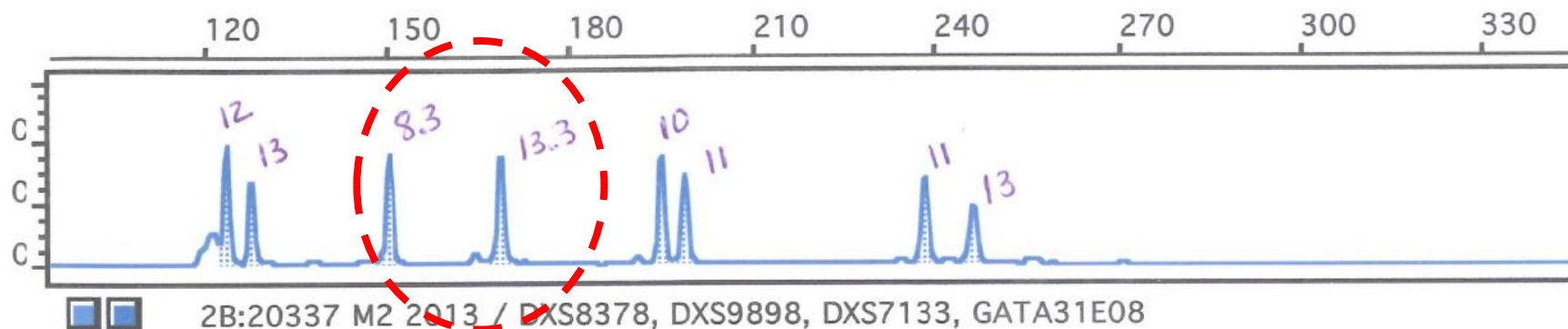
DXS10079



GeneScan® 3.1.2

GeneScan™ Project-11.04.13 Display-2

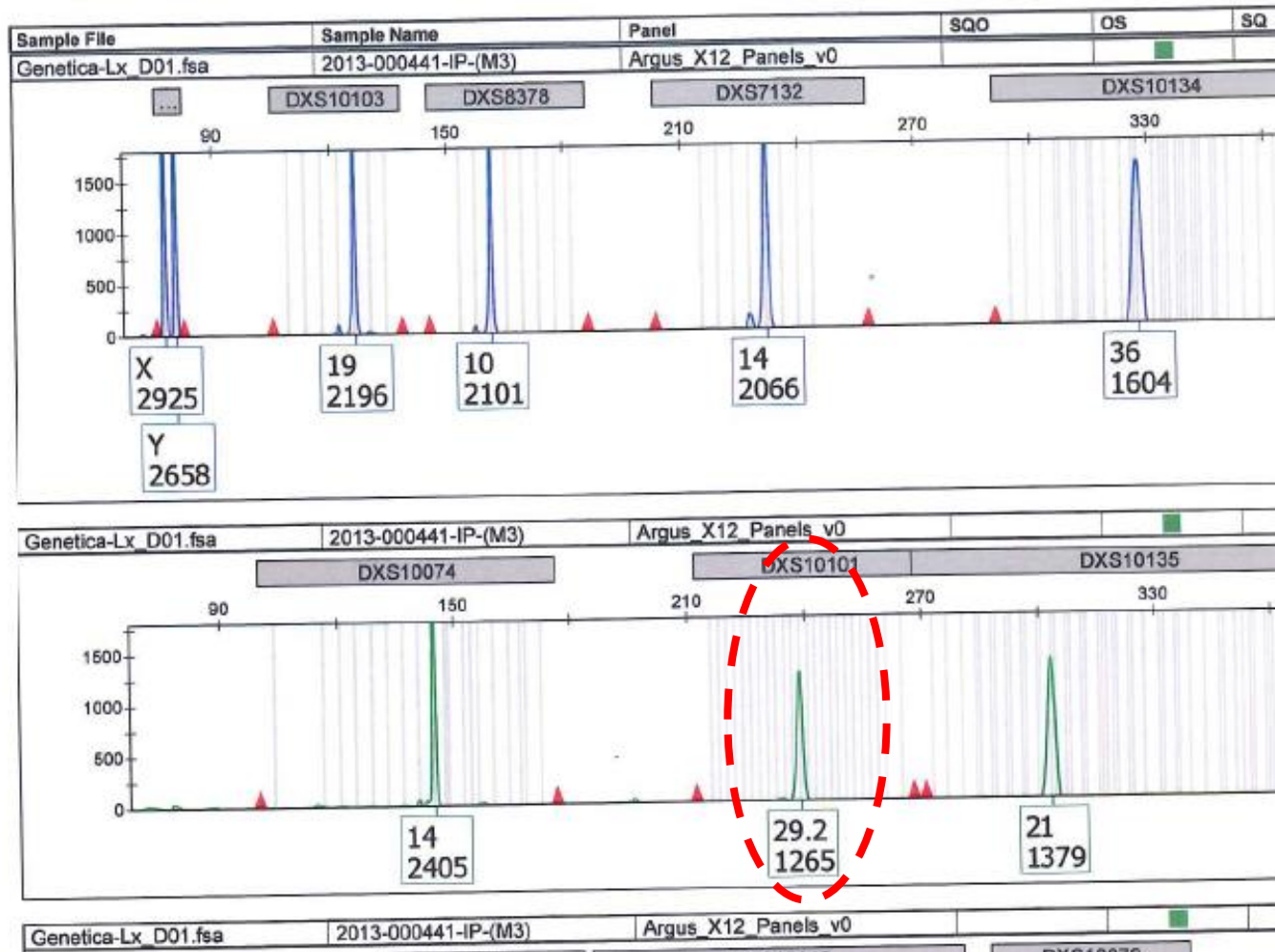
20337 Pag



M2 (8.3/13)

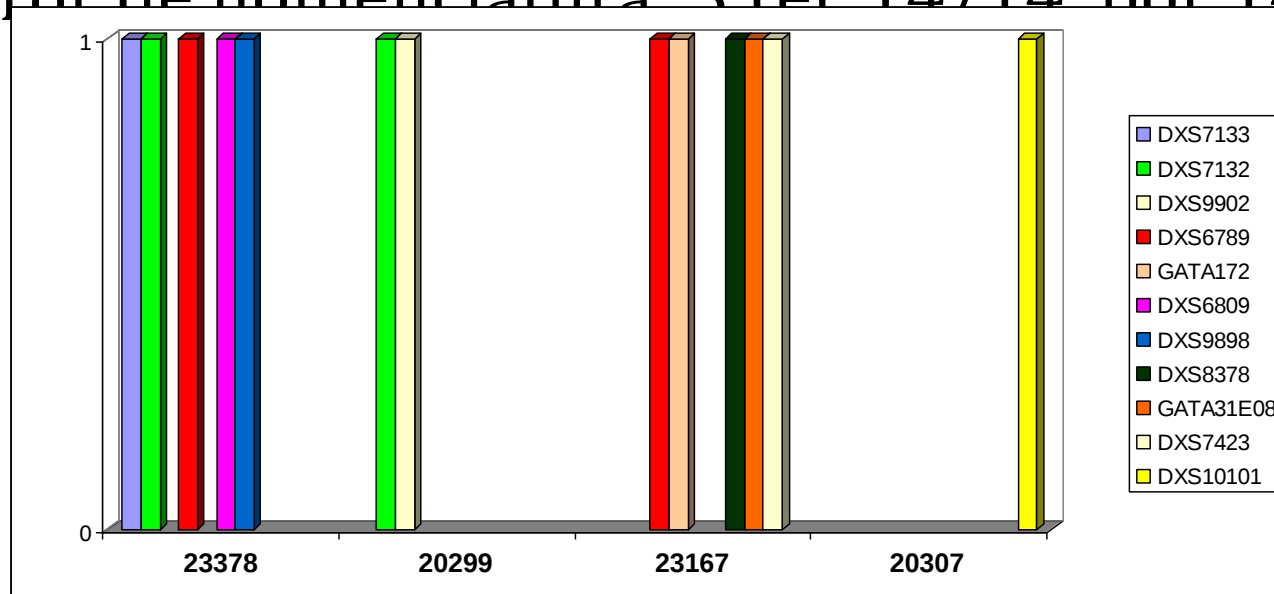
PRECINTO 20337 (X-STRs/ ABI 310)

111042013_Precinto_20307_IArgus-X-12



PRECINTO 20307
(Argus X12; ABI 3130/3130 xl)

M3 (29.2),
error transcripción informa "29"



PRECINTO 23378 (X-STRs/ ABI 3500)

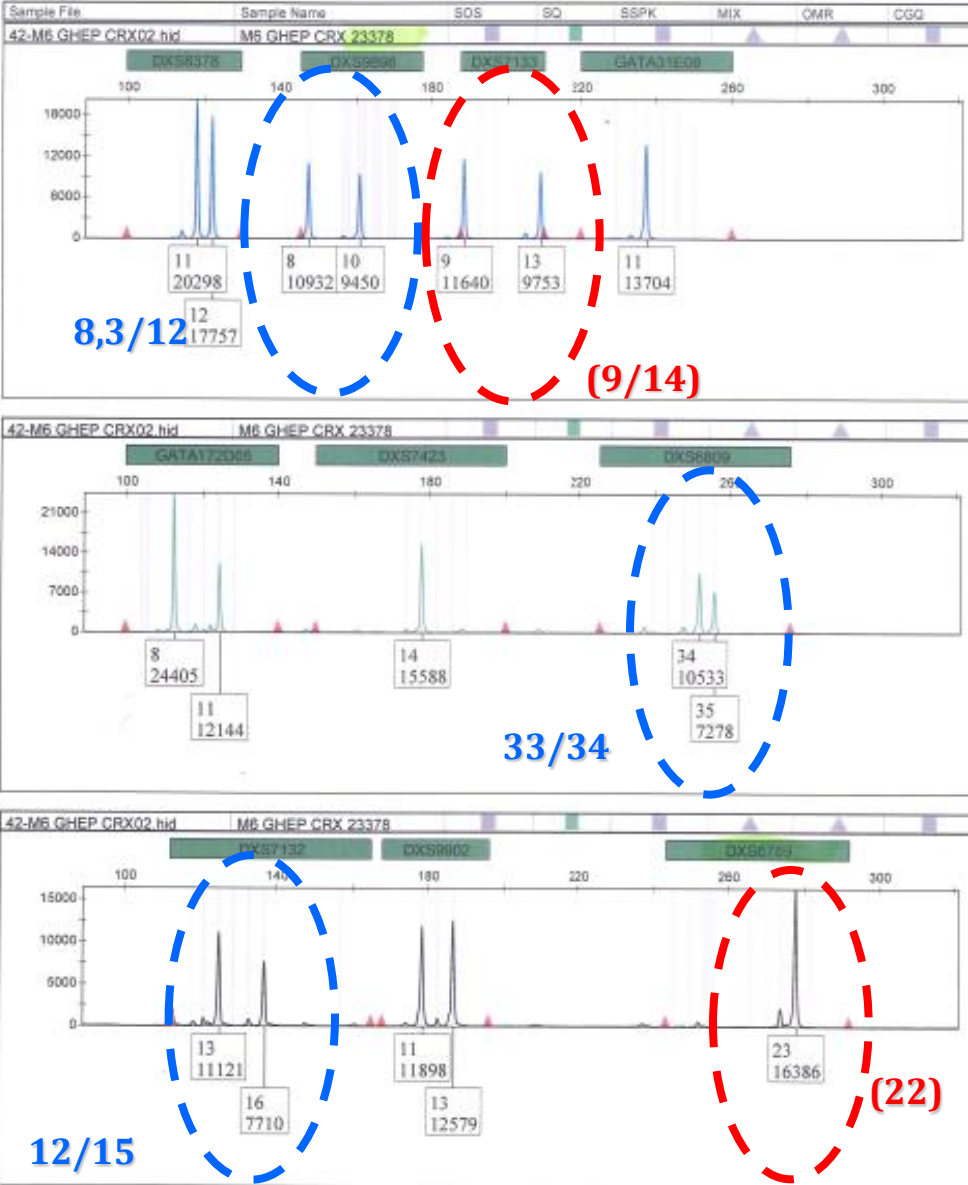


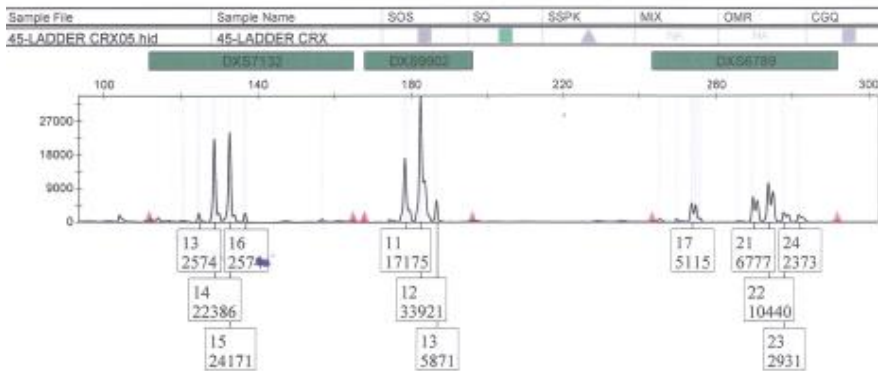
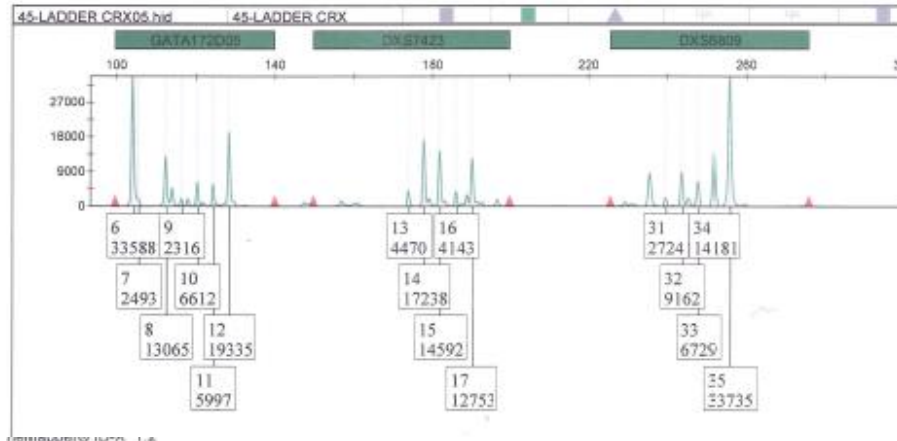
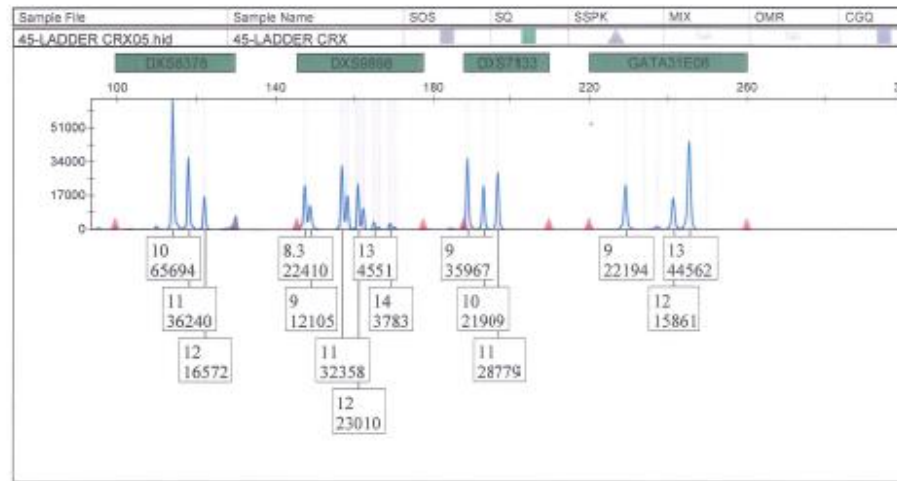
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Informado:

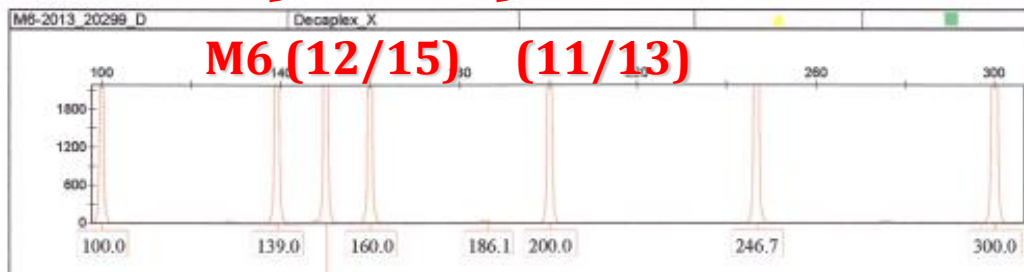
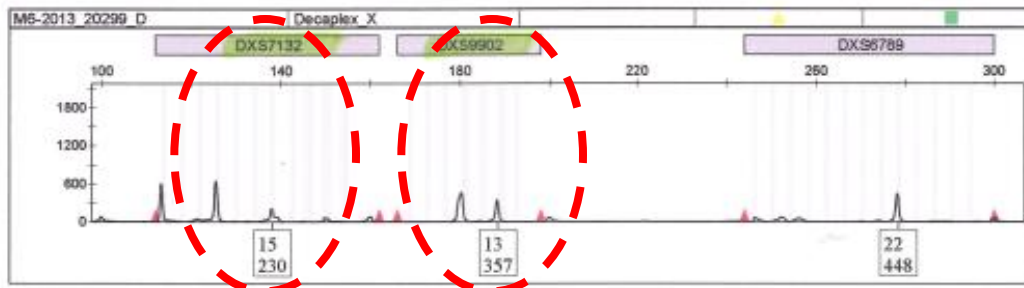
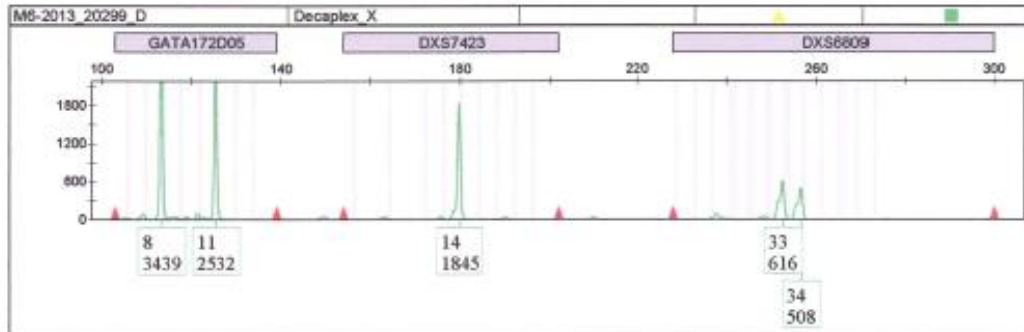
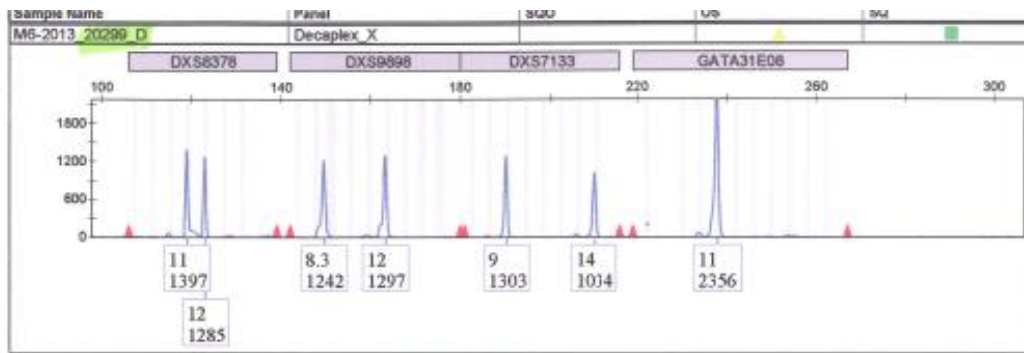
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23378	DXS6809_M6	33/34
23378	DXS7132_M6	12/15
23378	DXS7133_M6	9/13
23378	DXS7423_M6	14
23378	DXS8378_M6	11/12
23378	DXS9898_M6	8.3/12
23378	DXS9902_M6	11/13
23378	GATA172D_M6	8/11
23378	GATA31E08_M6	11

2 marcadores fuera de consenso
3 errores de transcripción, en consenso??



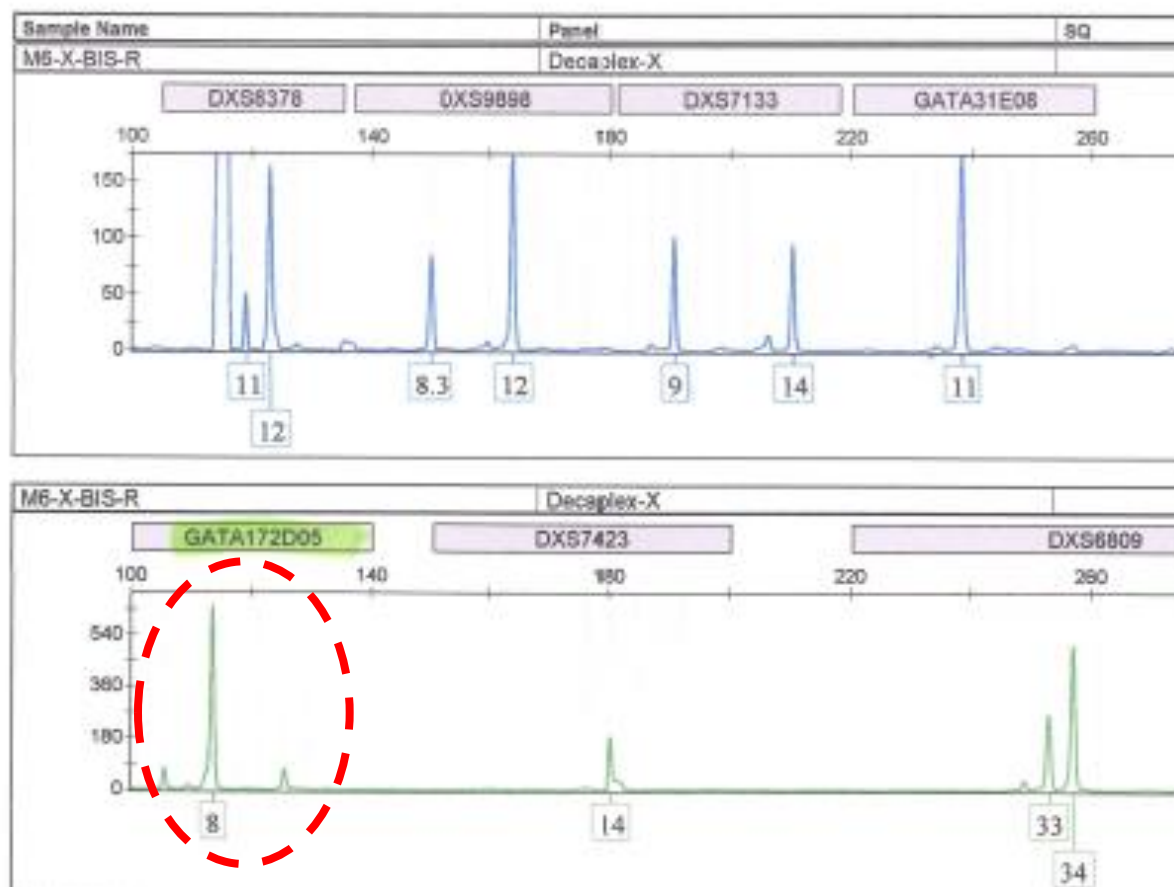


Ladder Cromosoma X
Precinto 23378

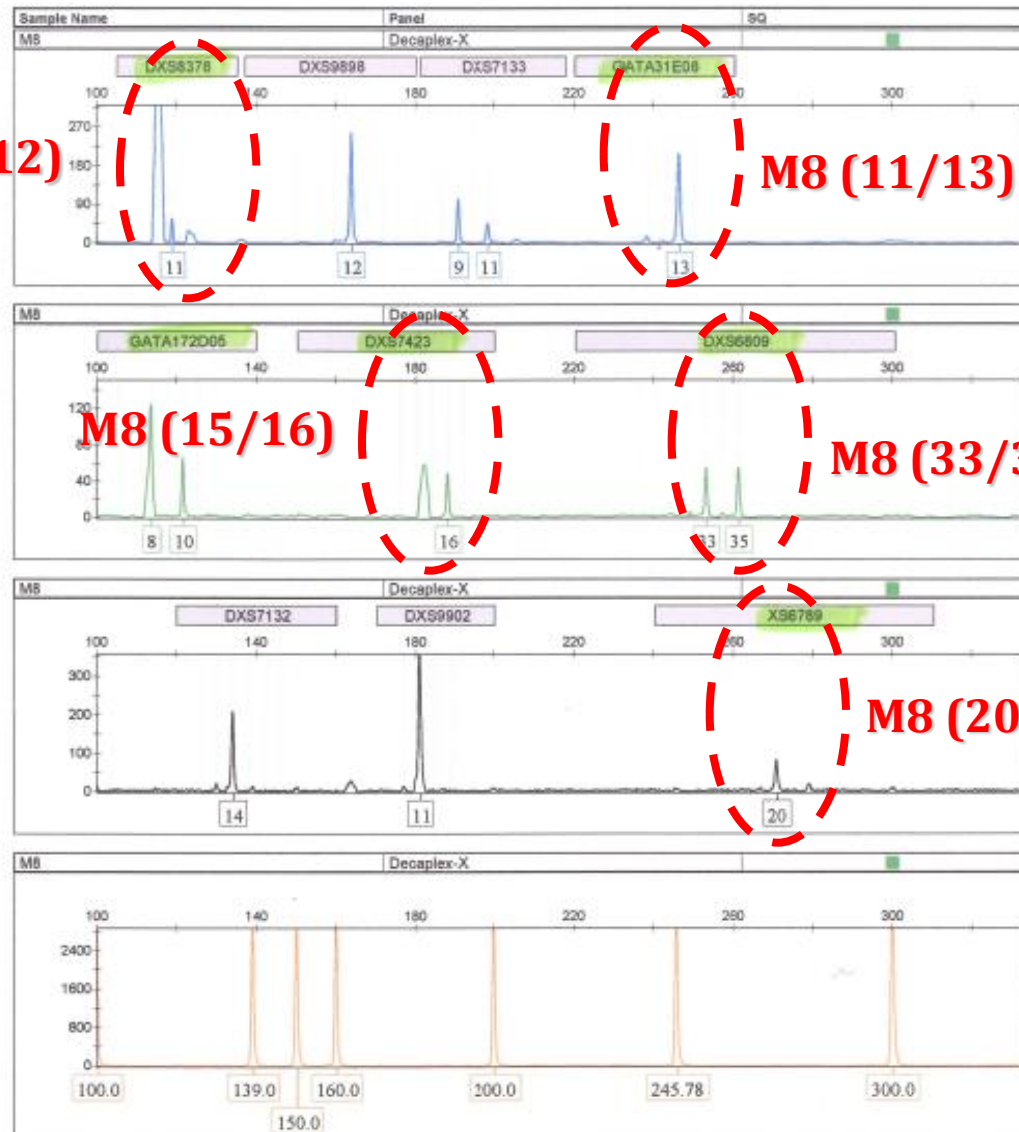


PRECINTO 20299
(X-STRs; ABI 3130/3130 xl)

DXS7132, DXS9902



PRECINTO 23167 M6, GATA172 (8/11)
(X-STRs; ABI 3130/3130 xl)



M8 (11/12)

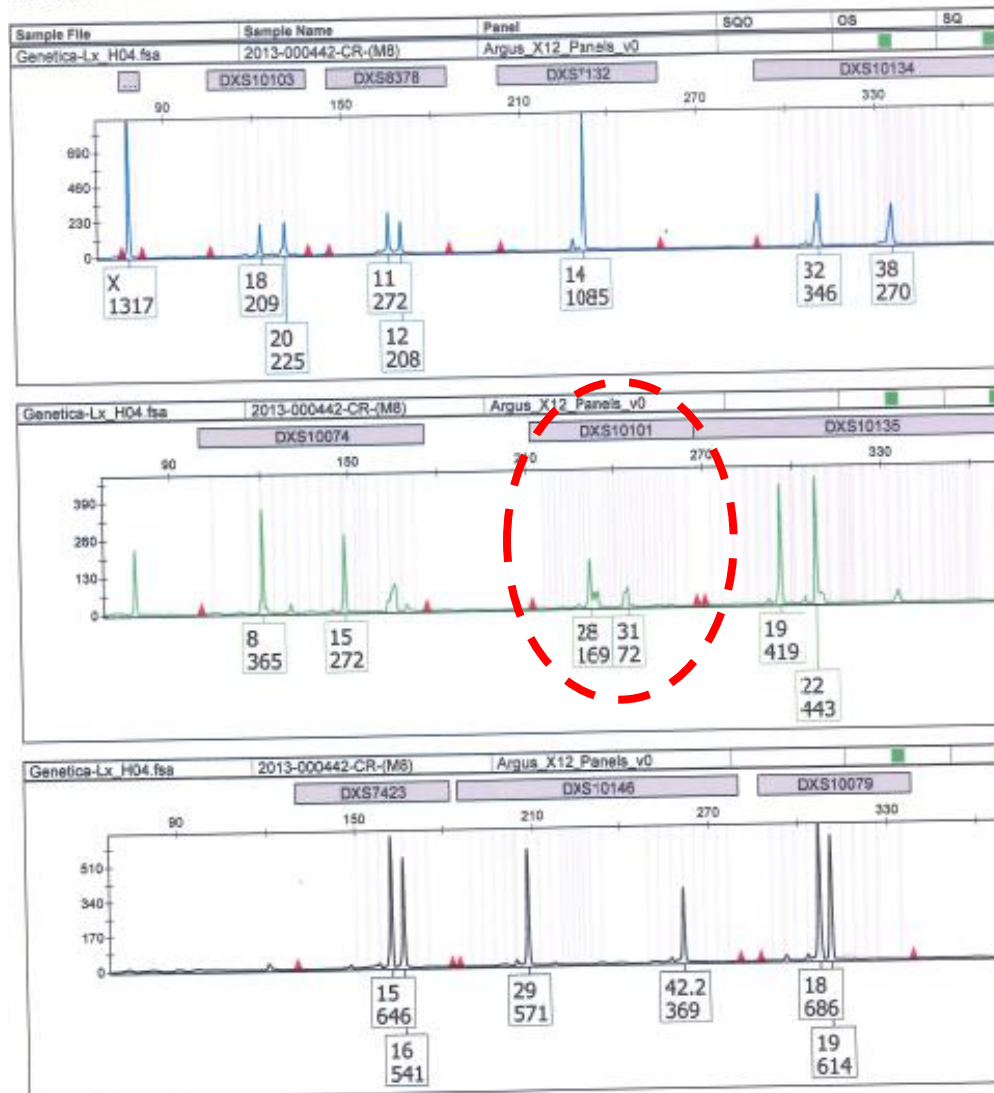
M8 (11/13)

M8 (15/16)

M8 (33/35) informa 33/34 (error T)

M8 (20/22)

PRECINTO 23167 - M8



M8 (28.2/31)

DXS10101

PRECINTO 20307
(Argus X12; ABI 3130/3130 xl)

Conclusiones

- Si bien el número de discrepancias es alto están concentradas en un reducido número de laboratorios
- Mayor número de discrepancias en Cr-X que en Cr-Y
- Envío de electroferogramas con la corrida del Size y Ladders.
- Chequear controles (Ladders o muestras controles)

Muchas gracias!!

