

ghep-isfg

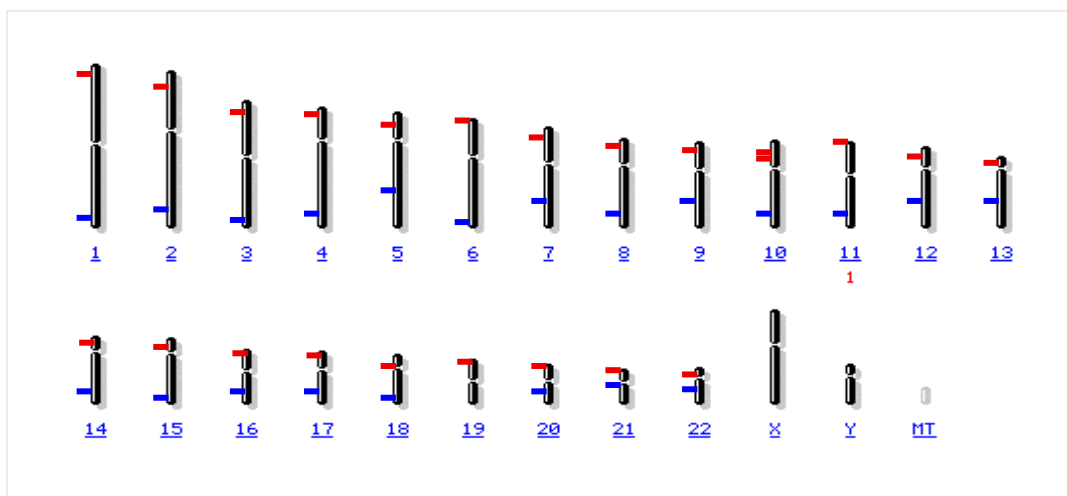
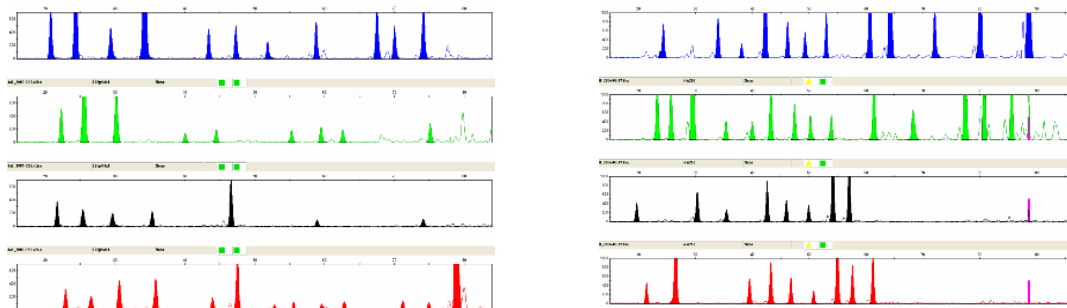
Ejercicio de genotipado de SNPs autosómicos

FASE 2: 52plex

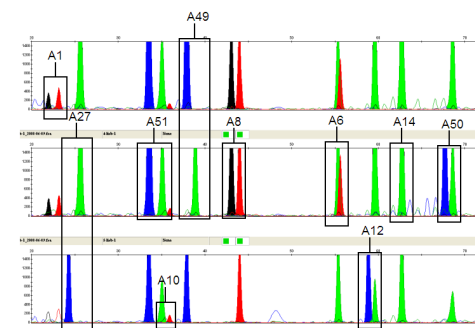
FASE 1: 10Plex

Reacción multiplex PCR + Reacción multiplex de SNaPshot

Human Identification 52plex



SFI code	rs-number	Ref allele (A1)
1	rs1490413	G
6	rs1029047	T
8	rs763869	C
10	rs735155	G
12	rs2107612	G
14	rs1454361	A
27	rs2111980	A
49	rs1005533	G
50	rs8037429	C
51	rs891700	A



Resultados del Genotipado

participants	total analysis	correct	reversed	incorrect	no results
18	540	369	89	47	35

	A1	A27	A51	A10	A49	A8	A6	A12	A14	A50
	rs1490413	rs2111980	rs891700	rs735155	rs1005533	rs763869	rs1029047	rs2107612	rs1454361	rs8037429
Consenso	G/G	G/G	A/G	A/A	A/G	C/T	A/T	A/A	A/A	C/T
Lab 1	C/C	G/G	A/G	T/T	G/A	C/T	T/A	A/A	A/A	A/G
Lab 2	G/G	G/G	A/G	A/A	A/G	C/T	A/T	A/A	A/A	C/T
Lab 3	G/G	G/G	G/A	A/A	G/A	C/T	A/T	A/A	A/A	C/T
Lab 4	G/G	G/G	A/G	A/A	A/G	C/T	A/T	A/A	A/A	C/T
Lab 5	T/T	G/G	A/G	T/T	A/G	C/T	A/T	A/A	A/A	A/G
Lab 6	C/C	A/G	A/G	C/T	A/G	C/T	T/A	G/G	A/A	G/G
Lab 7	C/C	G/G	G/A	C/T	G/A	C/T	A/T	A/A	A/A	G/A
Lab 8	C/C	G/G	A/G	C/T	A/G	C/T	A/T	A/A	A/A	A/G
Lab 9	G/G	G/G	A/G	A/G	A/G	C/T	A/T	A/A	A/A	T/C
Lab 10	C/C	G/G	A/G	?	G/G?	C/T	A/T?	A/A?	A/A?	A/G
Lab 11	C/C	G/G	A/G	T/T	A/G	C/T	A/T	A/A	A/A	A/G?
Lab 12	C/C	G/G	A/G	T/T	A/G	C/T	A/T	A/A	A/A	A/G
Lab 13	-	-	-	T/T	-	C/T	A/T	-	-	-
Lab 14	C/C	G/G	A/G	T/T	G/G	C/T	A/T	A/G	A/A	A/G
Lab 15	C/C	G/G	G/G	C/T	A/G	C/T	T/T	A/A	-	G/G
Lab 16	C/C	G/G	A/G	T/T	A/G	C/T	A/T	A/A	A/A	A/G
Lab 17	G/G	G/G	A/G	A/A	A/G	C/T	A/T	A/A	A/A	C/T
Lab 18	C/C	A/G	A/G	T/T	A/G	C/T	A/T	A/A	A/A	A/G

FASE 2: 52Plex

Electrophoresis 2006, 27, 1713–1724

1713

Juan J. Sanchez¹
 Chris Phillips²
 Claus Børsting¹
 Kinga Balogh³
 Magdalena Bogus³
 Manuel Fondevila²
 Cheryl D. Harrison⁴
 Esther Musgrave-Brown⁴
 Antonio Salas²
 Denise Syndercombe-Court⁴
 Peter M. Schneider³
 Angel Carracedo²
 Niels Morling¹

Research Article

A multiplex assay with 52 single nucleotide polymorphisms for human identification

A total of 52 SNPs reported to be polymorphic in European, Asian and African populations were selected. Of these, 42 were from the distal regions of each autosome (except chromosome 19). Nearly all selected SNPs were located at least 100 kb distant from known genes and commonly used STRs. We established a highly sensitive and reproducible SNP-typing method with amplification of all 52 DNA fragments in one PCR reaction followed by detection of the SNPs with two single base exten-



ELSEVIER

Available online at www.sciencedirect.com



ScienceDirect

Forensic Science International: Genetics 1 (2007) 186–190



www.elsevier.com/locate/bsfig

Forensic validation of the SNPforID 52-plex assay

Esther Musgrave-Brown^{a,*}, David Ballard^a, Kinga Balogh^b, Klaus Bender^b, Burkhard Berger^c,
 Magdalena Bogus^b, Claus Børsting^d, María Brion^e, Manuel Fondevila^e, Cheryl Harrison^a,
 Ceylan Oguzturun^a, Walther Parson^c, Chris Phillips^e, Carsten Proff^f, Eva Ramos-Luis^e,
 Juan J. Sanchez^d, Paula Sánchez Diz^e, Bea Sobrino Rey^e, Beate Stradmann-Bellinghausen^b,
 Catherine Thacker^a, Angel Carracedo^e, Niels Morling^d, Richard Scheithauer^c,
 Peter M. Schneider^f, Denise Syndercombe Court^a

^a Centre for Haematology, ICMS, Barts & the London, Queen Mary's School of Medicine & Dentistry, London, United Kingdom

^b Institute of Legal Medicine, Johannes Gutenberg University, Mainz, Germany

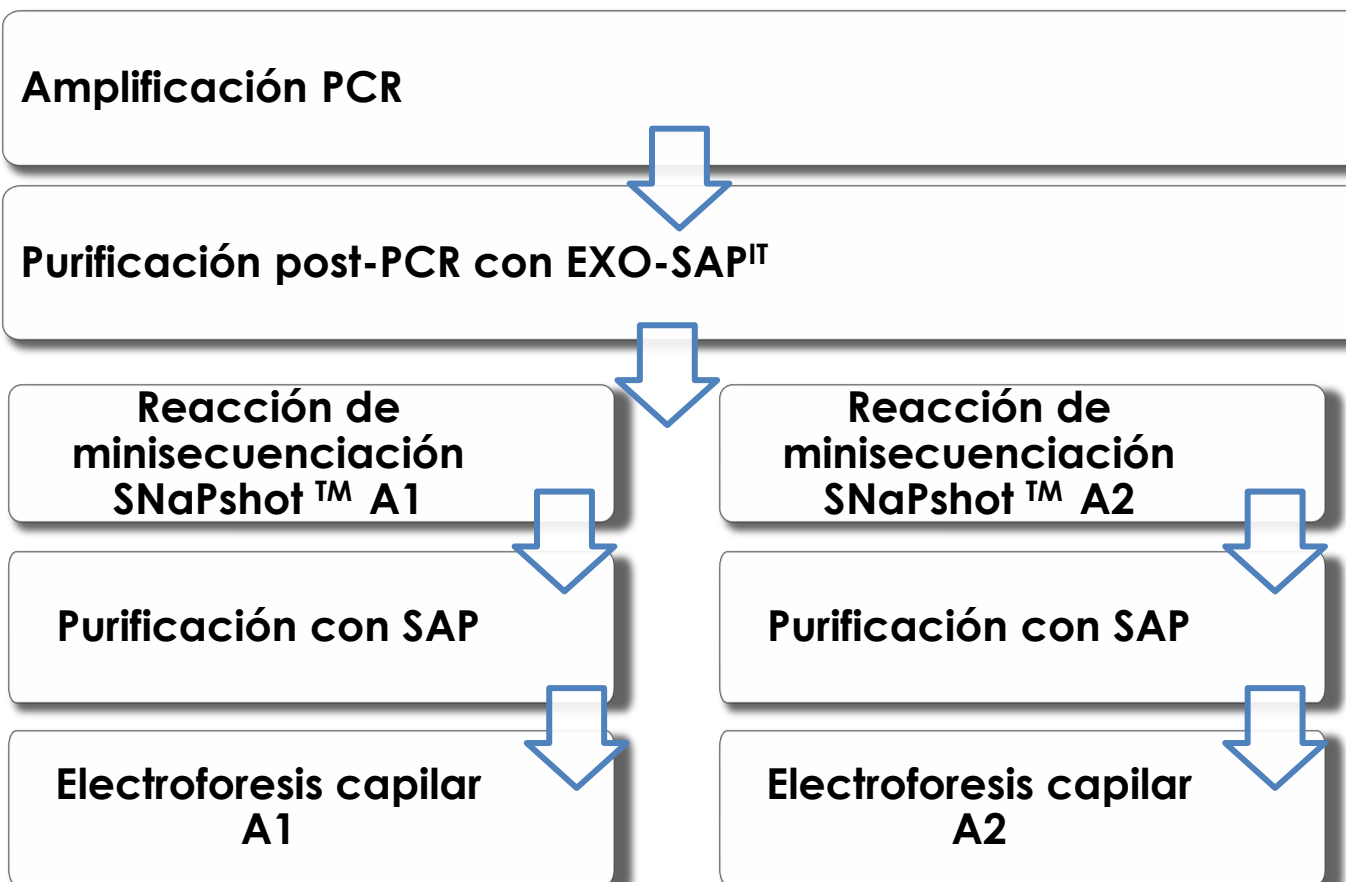
^c Institute of Legal Medicine, Innsbruck Medical University, Innsbruck, Austria

^d Department of Forensic Genetics, Institute of Forensic Medicine, University of Copenhagen, Copenhagen, Denmark

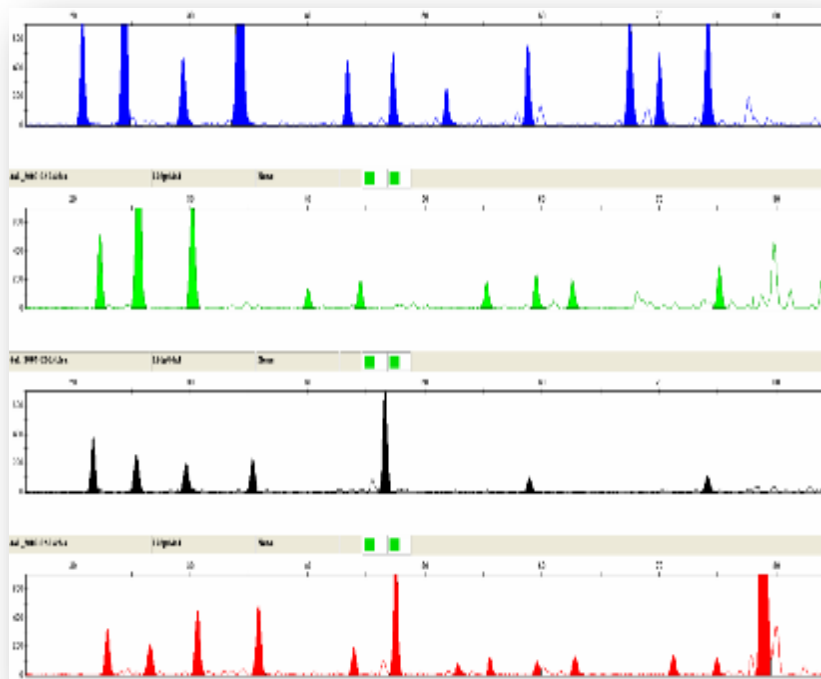
^e Institute of Legal Medicine, University of Santiago de Compostela, Santiago de Compostela, Spain

^f Institute of Legal Medicine, University of Cologne, Cologne, Germany

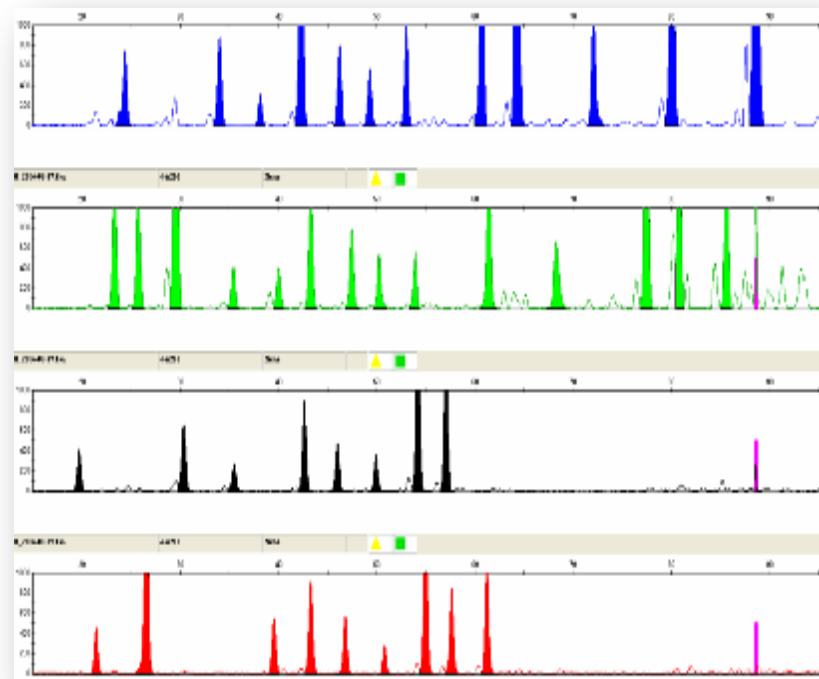
Muestras: M1, M2 y M3 del ejercicio de intercomparación **2012**



Auto1- 23plex



Auto2- 29plex



NOMENCLATURA

Marker code	NCBI rs No.	SBE primer size	Detection orientation	Detected genotype	Report genotype
a1	rs1490413	18	Reverse	T/C	A/G
a2	rs876724	24	Forward	C/T	C/T
a3	rs1357617	29	Reverse	T/A	A/T
a4	rs2046361	78	Reverse	T/A	A/T

Los resultados de todos los SNPs se darán en sentido FORWARD, de manera que en aquellos casos en los que las sondas estén diseñadas sobre la cadena reverse, el genotipo a reportar será la base complementaria a la detectada en el electroferograma.

Plazo de Inscripción en el ejercicio: Hasta el 30 de Octubre de 2011

Para cualquier consulta las direcciones de contacto son:

lourditasmt@gmail.com

vanesa.alvarez@usc.es