POPULATION GENETICS OF THREE STRs: TH01, CSF1PO AND TPOX IN SOUTHERN SPAIN

ANDRES MI, PRIETO V, FLORES IC, SANZ P.

National Institute of Toxicology. Seville. Spain.

Human systems under study

TH01: Tyrosine hydroxylase gene (11p15.5); AATG repeat; alleles 5, 6, 7, 8, 9, 9.3, 10, 11.

CSF1PO: c-fms protooncogene for CSF-1 receptor gene (5q33.3-34); AGAT repeat; alleles 8, 9, 10, 11, 12, 13, 14.

TPOX: Thyroid peroxidase gene (2p13); AATG repeat; alleles 8, 9, 10, 11, 12.

Population and sample size: Andalusian (Southern Spain) N:150.

Methods:

Blood samples from casework and staff. Extraction: Chelating Agent (Sigma).

Amplifications: simultaneous amplification using the Gene Print STR multiplex from Promega.

Typing: 6% denaturing gel electrophoresis followed by silver staining according to Promega protocols (1994).

Results

Table 1 TH01 observed genotypes

Gen.	N	Freq.	Gen.	N	Freq.	Gen	N	Freq.
6-6	3	0.0203	**7-7**	8	0.0541	**8-9**	7	0.0473
6-7	8	0.0541	**7-8**	8	0.0541	**8-9.3**	11	0.0743
6-8	9	0.0608	**7-9**	6	0.0405	**9-9**	7	0.0473
6-9	13	0.0878	**7-9.3**	13	0.0878	**9-9.3**	13	0.0878
6-9.3	16	0.1081	**7-10***	4	0.0270	**9-10***	1	0.0068
6-10*	3	0.0203	**8-8***	6	0.0405	**9.3-9.3**	12	0.0811

X^2=8.1088 df=7 p=0.3231 % Heterozygosity= 75.68 * pooled classes

Discrimination Power= 0.9312 Average Power of exclusion= 54.71 %

Table 2 TPOX observed genotypes

Gen.	N	Freq.	Gen.	N	Freq.	Gen	N	Freq.
6-9*	1	0.0067	**8-12†**	4	0.0268	**10-10‡**	1	0.0067
8-8*	32	0.2148	**9-9†**	2	0.0134	**10-11**	2	0.0134
8-9	14	0.0940	**9-10†**	2	0.0134	**11-11**	18	0.1208
8-10	13	0.0872	**9-11‡**	3	0.0201	**11-12**	5	0.0336
8-11	51	0.3423	**9-12‡**	1	0.0067			

X^2=7.5555 df=3 p=0.0561 % Heterozygosity= 64.43 *,†,‡ different pooled classes
Discrimination Power= 0.8027 Average Power of exclusion= 38.13 %

Table 3 CSF1PO observed genotypes

Gen.	N	Freq.	Gen.	N	Freq.	Gen	N	Freq.
8-9*	2	0.0136	**10-10**	8	0.0544	**11-13**	10	0.0680
8-10*	1	0.0068	**10-11**	22	0.1497	**11-14‡**	1	0.0068
8-11*	1	0.0068	**10-12**	24	0.1633	**12-12‡**	19	0.1293
9-10*	2	0.0136	**10-13**	2	0.0136	**12-13**	8	0.0544
9-11*	3	0.0204	**10-14†**	1	0.0068	**12-14°**	2	0.0136
9-12*	1	0.0068	**11-11†**	15	0.1020	**12-15°**	1	0.0068
9-14*	1	0.0068	**11-12**	21	0.1429	**13-14°**	2	0.0136

X^2=7.2439 df=3 p=0.0645 % Heterozygosity=71.43 *,†,‡,° different pooled classes
Discrimination Power= 0.8912 Average Power of exclusion= 48.05 %

Table 4 TH01 allele frequencies

Allel.	N	Freq.	Allel.	N	Freq.	Allel.	N	Freq.
5	0	0.0000	**8**	47	0.1588	**10**	8	0.0270
6	55	0.1858	**9**	54	0.1824	**11**	0	0.0000
7	55	0.1858	**9.3**	77	0.2601			

Table 5 TPOX allele frequencies

Allel.	N	Freq.	Allel.	N	Freq.	Allel.	N	Freq.
6	1	0.0034	**9**	25	0.0839	**12**	10	0.0336
7	0	0.0000	**10**	19	0.0638	**13**	0	0.0000
8	146	0.4899	**11**	97	0.3255			

Table 6 CSF1PO allele frequencies

Allel.	N	Freq.	Allel.	N	Freq.	Allel.	N	Freq.
7	0	0.0000	**10**	68	0.2313	**13**	22	0.0748
8	4	0.0136	**11**	88	0.2993	**14**	7	0.0238
9	9	0.0306	**12**	95	0.3231	**15**	1	0.0034

Comparison with other caucasian populations:

TH01 genotype frequencies were compared with two Spanish published data (Philipe, 1994; Lorente, 1994) using a Rx C contingency table X^2 test for homogeneity. The allele distributions were similar ($p=0.8$ and $p=0.3$ respectively). Up to date, enough genotype frequency data were not available for TPOX and CSF1PO in other Spanish caucasian populations.

Examples from casework

1.- PATERNITY CASE: After the implementation of STRs an old conflicting paternity case was reconsidered. The two alleged fathers were brothers of the mother. Paternity could not be solved at the time of the study: RFLPs could not be applied because blood samples, inadequately conserved, were deteriorated on arrival at the laboratory, DNA was highly degraded and no more blood samples could be obtained. Only DQA1 and D1S80 were added to the set of conventional genetic markers. No exclusions were found due to a high degree of consanguinity within the family group for several generations. The probabilities of paternity were 98.62% for alleged father 1 and 94.26% for alleged father 2. With the investigation of STRs (TH01, CSF1PO and TPOX) the probability of paternity for alleged father 1 increased to 99.89% and one exclusion was detected for alleged father 2 in TH01 (two additional exclusions were found when investigating other STR loci).

2.- SEXUAL ASSAULT: Samples: Victim and suspect blood samples, vaginal swab, blood stain on the suspect's underpants. Pubic hair obtained from pubic combing of the victim. The following results were obtained:

	DQA1	D1S80	CSF1P0	TPOX	TH01
Victim	1.3,2	T24T29	10-12	8-11	6-7
Suspect	2,3	T20T28	11-13	8-11	6-9
Spermatozoa	2,3	ND	11-13	8-11	6-9
Blood Stain	1.3,2(3)	T24T29 (T20T28)	10-12	8-11	6-7(9)
Pubic hair	2,3	ND	11-13	8-11	6-9

ND= not detected ( )= weak signal

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