

Population and formal genetics of the STRs TPO, TH01 and VWFA31/A in North Portugal

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Systems and loci

TPO, repeat (AATG), intron 10 of thyroid peroxidase gene (2p23-pter)

TH01, repeat (TCAT), intron 1 of tyrosine hydroxylase gene (11p15.5-p15)

VWFA31/A, repeat (TCTR), intron 40 of von Willebrand factor gene (12p12-pter).

Population and sample sizes:

North Portugal. TPO: N=273; TH01: N=197; VWFA31/A: N=255

Methods:

Samples: blood or buccal swabs from unrelated individuals and mother/child pairs; DNA extraction: method of Lareu *et al.* (1994); electrophoresis according to Luis and Caeiro (1995); silver staining: Budowle *et al.*(1992); genotyping was performed by side-by-side comparison with an allelic ladder made up from previously typed samples.

TPO: primers (Anker *et al.*, 1992); amplification conditions: 5 min at 93°; 94°: 1 min; 63°: 0.5 min; 72°: 1.5 min; 27-35 cycles

TH01: primers (Gill *et al.*, 1994); amplification conditions: 5 min at 93°; 94°: 1 min; 54°: 1 min; 72°: 1 min; 27-35 cycles

VWFA31/A: primers (Kimpton *et al.*, 1992); amplification conditions: the same as for TH01.

Results:

Allele frequencies

TPO		TH01		VWFA31/A	
Allele	Freq.	Allele	Freq.	Allele	Freq.
6	0.005	6	0.193	12	0.002
7	0.004	7	0.191	13	0.002
8	0.511	8	0.140	14	0.121
9	0.084	9	0.201	15	0.110
10	0.060	9.3	0.256	16	0.214
11	0.291	10	0.019	17	0.300
12	0.042			18	0.176
13	0.002			19	0.061
				20	0.012
				21	0.002

Observed genotypes

TPO		TH01		VWFA31/A	
Genotypes	Obs.	Genotypes	Obs.	Genotypes	Obs.
6-8	1	6-6	8	12-17	1
6-9	1	6-7	13	13-16	1
6-11	1	6-8	13	14-14	4
7-8	2	6-9	17	14-15	6
8-8	73	6-9.3	19	14-16	9
8-9	17	6-10	2	14-17	25
8-10	17	7-7	11	14-18	10
8-11	83	7-8	8	14-19	3
8-12	12	7-9	16	14-20	1
8-13	1	7-9.3	19	15-15	4
9-9	2	7-10	1	15-16	19
9-10	3	8-8	5	15-17	12
9-11	17	8-9	11	15-18	6
9-12	4	8-9.3	15	15-19	3
10-10	1	8-10	1	15-20	2
10-11	10	9-9	9	16-16	11
10-12	1	9-9.3	19	16-17	35
11-11	21	9-10	2	16-18	15
11-12	6	9.3-9.3	16	16-19	6
		9.3-10	2	16-20	2
				17-17	14
				17-18	37
				17-19	14
				17-21	1
				18-18	9
				18-19	4
				19-20	1

P=0.9 P=0.9 P=0.7

Comments:

Mother-child pairs analyses (nr. of pairs: 159, 121 and 160 for each system, respectively) show no exclusions and no deviations to mendelian rules.

In TPO system two new alleles (7 and 13) were found, and frequencies estimated in this sample are similar to those reported up to now for Caucasians.

For TH01, allele 9 is more frequent than in other Caucasians, allele 9.3 being correspondingly rarer.

Concerning VWFA31/A, a new allele (12) with 130 bp was found; gene frequencies are similar to those described in other Caucasian populations.

References

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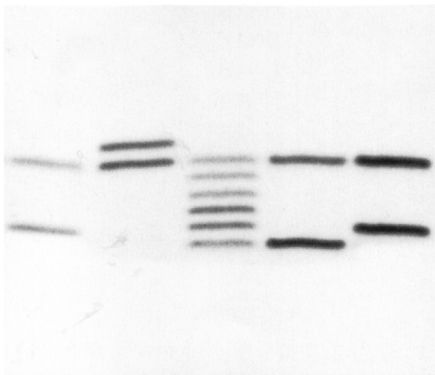


Fig 1. TPO genotypes

From left to right: lane 1: 8-12; lane 2: 7-8; lane 3: ladder; lane 4: 8-13; lane 5: 8-12;

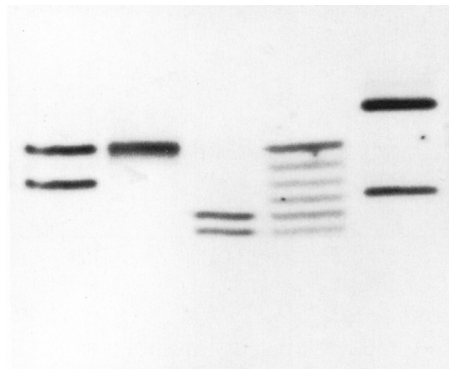


Fig 2. VWFA31/A genotypes

From left to right: lane 1: 14-16; lane 2: 14-14; lane 3: 18-19; lane 4: ladder; lane 5: 12-17;