

Population genetics of the STRs TPO, TH01 and VWFA31/A in S.Tomé e Príncipe

M.J. Prata, A. Amorim, L. Gusmão and M. J. Trovoada

Inst. Antropologia, Univ.Porto and IPATIMUP, 4050 Porto, Portugal

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

S.Tomé e Príncipe (Gulf of Guinea)

TPO: N=147

TH01: N=143

VWFA31/A: N=133

Methods: Samples: blood from unrelated newborns; DNA extraction: method of Lareu *et al.* (1994); electrophoresis according to Luis and Caeiro (1995). Silver staining: Budowle *et al.* (1992);

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 allele | Freq. | TH01 allele | Freq. | VWFA31/A allele | Freq. |
|---------------|-------|----------------|-------|--------------------|-------|
| 6 | 0.065 | 6 | 0.091 | 13 | 0.026 |
| 7 | 0.034 | 7 | 0.357 | 14 | 0.083 |
| 8 | 0.388 | 8 | 0.346 | 15 | 0.184 |
| 9 | 0.197 | 9 | 0.150 | 16 | 0.286 |
| 10 | 0.075 | 9.3 | 0.049 | 17 | 0.199 |
| 11 | 0.214 | 10 | 0.007 | 18 | 0.135 |
| 12 | 0.027 | | | 19 | 0.056 |
| | | | | 20 | 0.030 |

Observed genotypes

| TPO genotypes | Obs. | TH01 genotypes | Obs. | VWFA31/A genotypes | Obs. |
|--------------------------|-------------|---------------------------|-------------|-------------------------------|-------------|
| 6-8 | 10 | 6-6 | 1 | 13-14 | 1 |
| 6-9 | 7 | 6-7 | 4 | 13-15 | 1 |
| 6-10 | 1 | 6-8 | 14 | 13-16 | 2 |
| 6-11 | 1 | 6-9 | 5 | 13-17 | 1 |
| 7-8 | 6 | 6-9.3 | 1 | 13-18 | 2 |
| 7-9 | 2 | 7-7 | 18 | 14-14 | 1 |
| 7-11 | 1 | 7-8 | 38 | 14-15 | 4 |
| 7-12 | 1 | 7-9 | 17 | 14-16 | 6 |
| 8-8 | 17 | 7-9.3 | 6 | 14-17 | 6 |
| 8-9 | 26 | 7-10 | 1 | 14-18 | 3 |
| 8-10 | 12 | 8-8 | 15 | 15-15 | 7 |
| 8-11 | 22 | 8-9 | 13 | 15-16 | 9 |
| 8-12 | 4 | 8-9.3 | 4 | 15-17 | 10 |
| 9-9 | 6 | 8-10 | 1 | 15-18 | 10 |
| 9-10 | 4 | 9-9 | 2 | 15-19 | 1 |
| 9-11 | 4 | 9-9.3 | 3 | 16-16 | 15 |
| 9-12 | 3 | 9-10 | 1 | 16-17 | 16 |
| 10-10 | 2 | | | 16-18 | 5 |
| 10-11 | 1 | | | 16-19 | 6 |
| 11-11 | 17 | | | 16-20 | 2 |
| | | | | 17-17 | 3 |
| | | | | 17-18 | 8 |
| | | | | 17-19 | 4 |
| | | | | 17-20 | 2 |
| | | | | 18-18 | 2 |
| | | | | 18-19 | 2 |
| | | | | 18-20 | 2 |
| | | | | 19-19 | 1 |
| | | | | 20-20 | 1 |

see Comments section

P=0.73

P=0.49

Comments:

Genotype distribution for TPO system is not in accordance with Hardy-Weinberg expectations. Sequential statistical analysis for each allele (Rand *et al.*, 1992) has demonstrated that only TPO*11 distribution is abnormal, with a large excess of homozygotes ($\chi^2 = 25.2$, 1d.f., $P < 0.001$). Since another sample studied by us (N.Portugal) is in equilibrium, this finding is probably due to a specific feature of this locus in this population. Sequencing of a subsample is under progress. A new allele (7) was found, and allele 6, which is rare in Caucasians, has a rather high frequency in this sample; no other data on Blacks are available for comparisons.

Concerning TH01, a very low frequency for the allele 9.3 (still lower than in USA Blacks) was found; on the other hand, allele 8 exhibits a very high frequency.

In VWFA31/A system alleles 12 and 21 were not observed but allele 13, a rare allele both in Caucasians and USA Blacks, reached polymorphic proportions (2.6%).

References

- Anker R, Steinbrueck T, Donis-Keller H (1992) Tetranucleotide repeat polymorphism at the human thyroid peroxidase (hTPO) locus. *Hum Mol Genet* 1: 137
- Budowle B, Chakraborty R, Giusti AM, Eisenberg AJ, Allen RC (1991) Analysis of the VNTR locus D1S80 by the PCR followed by high-resolution PAGE. *Am J Hum Genet* 48: 137-144
- Gill P, Kimpton C, D'Aloja E, Andersen JF, Bar W, Brinkmann B, Holgersson S, Johnson V, Kloosterman AD, Lareu MV, Nellemann L, Pfitzinger H, Phillips CP, Schmitter H, Schneider PM, Stenersen M (1994) Report of the European DNA profiling group (EDNAP) towards standardisation of short tandem repeat (STR) loci. *Forensic Sci Int* 65: 51-59
- Kimpton C, Walton A, Gill P (1992) A further tetranucleotide repeat polymorphism in the vWF gene. *Hum Mol Genet* 1: 287
- Lareu MV, Phillips CP, Carracedo A, Lincoln PJ, Court DS, Thomson JA (1994) Investigation of the STR locus HUMTH01 using PCR and two electrophoresis formats: UK and Galician Caucasian population surveys and usefulness in paternity investigations. *Forensic Sci Int* 66: 41-52
- Luis JR, Caeiro B (1995) Application of two STRs (VWF and hTPO) to human population profiling. A survey in Galicia. *Human Biology* (in press)
- Rand S, Puers C, Skowasch K, Wiegand P, Budowle B, Brinkmann B (1992) Population genetics and forensic efficiency data of 4 AMPFLPs. *Int J Legal Med* 104: 329-333

Acknowledgements This work was partially supported by JNICT (Junta Nacional de Investigação Científica e Tecnológica, BD/2849/93-ID and PBIC/C/CEN/1174/92) and CNCDP (Comissão Nacional para as Comemorações dos Descobrimientos Portugueses, research contract nº 70).

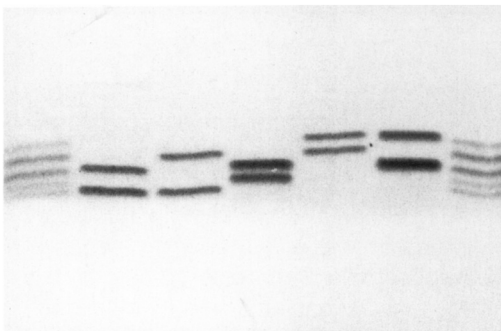


Fig.1. TH01 genotypes

From left to right: lane 1: ladder; lane 2: 8-9.3; lane 3: 7-9.3; lane 4: 8-9; lane 5: 6-7; lane 6: 6-8; lane 7: ladder.

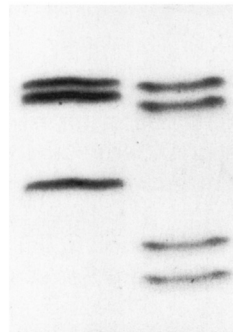


Fig.2. Distinction of TH01 9, 9.3 and 10 alleles and heteroduplex analysis.

From left to right: lane 1: 9-9.3; lane 2: 9-10