International Congress Series 1239 (2003) 213–218

Paternity testing analysis—allelic distribution, heterozygosity and power of exclusion of commonly used SLPs and STRs in the Brazilian Caucasoid population

L.F. Jobim [a,b,*], M.R. Jobim [a], F. Gamio [a], G. Ewald [a], M. Jobim [a], L. Fernandes [a]

[a] *Laboratório DNA Reference Ltda, Porto Alegre, Brazil*
[b] *Hospital de Clínicas de Porto Alegre, Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil*

Abstract

Analysis of allelic frequencies, heterozygosity and power of exclusion of commonly used SLP and STR markers in the Brazilian Caucasoid population was performed based on 6000 paternity testings. The SLPs (RFLPs) used were G3; YNH24, MS205, MS43 and MS31 and the STRs were TH01, VWA31, FES, F13A, CD4, FGA, D1S1656, D3S1358, D5S818, D7S820, D8S639, D8S1179, D12S391, D13S317, D18S51 and D21S11. The allele frequencies of the STRs are in the range of published Caucasian Portuguese population frequencies and in Hardy Weinberg equilibrium. The SLPs are in the same range as other Caucasian frequencies as well as their heterozygosity and power of exclusion. The power of exclusion of the 5 SLPs and 10 of the STRs was the same (99.99%). The STR with the highest power of exclusion (81%) and heterozygosity (89%) in the studied population was D12S391. We also present results from 197 fatherless cases analyzed according to the number of family members tested. The exclusion rate was 21.25%. Analysis with different family compositions will be presented. In 29 cases in which the alleged father was exhumed, we obtained a success rate of 83%. The exclusion rate of the successful cases was 25%. Furthermore, the exclusion rate of regular cases in court was 24%, lower than the exclusion rate in private requests of 32%.

Keywords: Paternity testing; DNA in Brazil

* Corresponding author. Laboratório DNA Reference Rua Padre Chagas 140, Porto Alegre 90570-080, Brazil. Tel.: +55-51-32221350.
E-mail address: drjobim@dnareference.com.br (L.F. Jobim).
URL: http://www.dnareference.com.

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PII: S0531-5131(02)00335-7

Table 1
STRs allelic frequency

Allelle	THO1	VWA31	FES	F13A	CD4	FGA	D1S1656	D3S1358	D5S818	D7S820	D8S639	D8S1179	D12S391	D13S317	D18S51	D21S11
3.2				0.100												
4.2					0.040											
4				0.070												
5				0.210	0.290											
5.2					0.020											
6	0.150			0.240	0.240											
7	0.170			0.320					0.010	0.020						
8	0.130		0.002	0.010	0.020				0.020	0.150				0.110		
9	0.160		0.007	0.003	0.040				0.040	0.130		0.013		0.090		
9.3	0.370															
10			0.280		0.270		0.010		0.070	0.280		0.070		0.050		
10.3																
11			0.400	0.008	0.040		0.050		0.340	0.210		0.071		0.310		
12		0.002	0.240				0.130		0.330	0.170		0.130		0.280	0.130	
13		0.004	0.040				0.090		0.170	0.040		0.290		0.100	0.110	
14		0.090					0.090	0.100	0.020			0.250		0.030	0.170	
15		0.120					0.150	0.260				0.110	0.050		0.150	
15.3							0.160									
16		0.240					0.040	0.250				0.030	0.030		0.140	
16.2						0.002										
16.3							0.050									
17		0.240				0.002		0.210				0.005	0.090		0.110	
17.3							0.130									
18		0.200				0.014		0.140					0.160		0.070	
18.3							0.060									
19		0.080				0.070		0.006					0.160		0.050	
19.3																

20		0.020				0.120						0.150		0.020		
21						0.160						0.090				
22						0.170						0.090				
22.2						0.002										
23						0.150					0.002	0.080				
24						0.140					0.020					
24.2															0.005	
25						0.110					0.008					
25.2																
26						0.030					0.160					
26.2																
27						0.013					0.240				0.020	
28											0.190				0.160	
29											0.140				0.210	
30															0.220	
30.1											0.080				0.030	
30.2															0.030	
31															0.070	
31.1											0.020					
31.2															0.100	
32															0.010	
32.2															0.100	
33																
33.2															0.030	
34																
34.2																
N	107	1461	206	107	100	897	192	896	894	896	192	900	358	895	900	898
Heterozygosity (%)	77	79	72	66	70	87	72	79	73	80	72	79	89	80	88	85
Power of exclusion (%)	64	62	64	65	64	73	68	54	57	69	70	68	81	60	75	74

1. Introduction

The aim of this work was to present the allele frequencies, heterozygosity and power of exclusion rate for STRs and SLPs in the Caucasian population of Brazil. The results from 6000 paternity tests are shown regardless of whether the alleged fathers were available or not. In the last situation, we tested their ascendant or descendent members or performed exhumation of the deceased alleged father. In these cases, we extracted DNA from bones or teeth. The power of exclusion of each locus was calculated. We also studied the results found with different analyses according to the number of family members.

2. Materials and methods

STRs in single amplifications (not multiplexed) were used when family members were not available and following phenol/chloroform extraction of DNA from bones and teeth from the deceased father. SLP probes (Cellmark Laboratory) were used in cases when the family of the deceased alleged father (parents, sons, brothers and sisters) was present for analysis. In all regular cases, with alleged father, mother and child present for sampling, we used several multiplexed, ''in house'' STRs as well as the ones in the Profiler Plus kit (Applied Biosystems, USA). The salting out extraction method (Miller, 1988) was used for all DNA prepared from blood. All STRs were analysed using either an ABI 310 (Applied Biosystems) or an ALF Express (Pharmacia). The paternity and frequencies calculations were done with the computer software DNA View (Charles Brenner, USA).

3. Results and conclusions

The allele frequencies of STRs were in a range comparable to other Caucasian Portuguese populations [1–4] and were in Hardy Weinberg equilibrium (Table 1). Similarly, SLP gave values which were in a range comparable to other Caucasian frequencies [5] with the following heterozygosity values and powers of exclusion—G3: 95% and 94.21%; YNH24: 95% and 94,62%; MS205: 91.66% and 95.02%; MS43: 84.84% and 92.75%; MS31: 87.75% and 94.28%. The combined power of exclusion of the SLPs and of the STRs was the same at: 99.999%.

The exclusion rate in regular cases, with alleged father, mother and child present and using STRs, was different if the case was private (32%) or court directed (24%) (Fig. 1). The lower exclusion rate in court can be explained by the fact that mothers go to the court when they are more confident of an inclusion result. The cost of going to court is a discouraging factor if they believe that an exclusion will be the probable result.

In fatherless examinations, the court cases also had less exclusions (26%) than the private ones (31%). When we tested mother, son, widow and one son of the alleged father, the probable exclusion rate was 29.41% (paternity index 0.087). In cases with mother, son, widow and two sons of the alleged father, the exclusion rate was 26.67%. When the mother, son, widow and three sons of the alleged father were tested, the exclusion rate was 29.41%. In cases where the widow was not available, but the mother, possible son and two

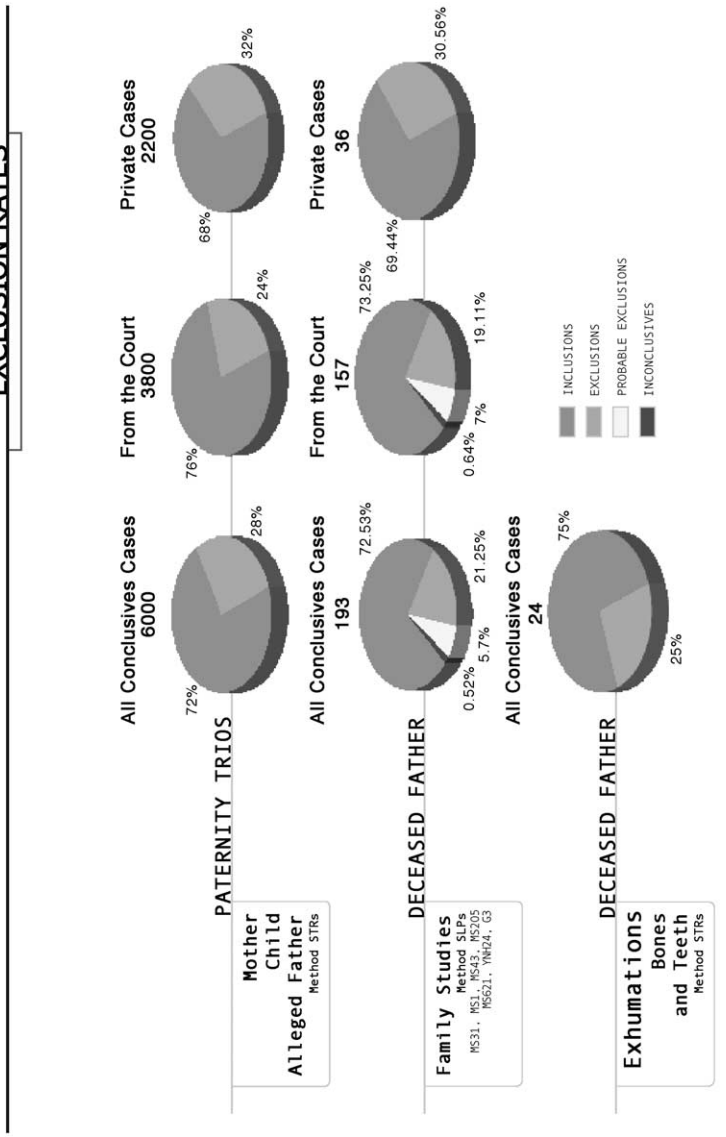

Fig. 1. Exclusion rates.

sons of the alleged father were present for analysis, the exclusion rate was 23.81%. Again, with the presence of mother, child and three possible uncles, the exclusion rate was 25%. In cases where both parents of the alleged father were analysed, the exclusion rate was 8.82%.

We had inconclusive cases (0.64%), mainly when the samples were from the mother, the child and one possible brother only. The exclusion assessment is impossible with such sampling. In some cases, however, the paternity index is so low that exclusion is a strong possibility (29%). In cases where an inclusion was reached (64.71%), the paternity index ratio was 15,800. In cases where two or more children of the alleged father were available for testing, we had 26% of exclusions and the mean paternity index was greater than 100,000.

We examined 29 exhumation cases with a success rate of 83% (24). From the successful testing, we obtained 25% of exclusions.

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