TECHNICAL NOTE

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Population Genetics of HPRTB, F13B, and LPL in Pernambuco, Northeast Brazil

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ABSTRACT: One hundred thirty-four unrelated Northeast Brazilian individuals were typed for the HPRTB, F13B, and LPL short tandem repeats (STRs). DNA was amplified by specific primers and identified by silver staining of polyacrylamide gels. The allelic frequencies of these loci were in agreement with Hardy-Weinberg proportions. The most frequent alleles were HPRTB*13, F13B*10, LPL*10. The combined probability of paternity and the discrimination power of these 3 STRs were high, permitting their utilization for forensic science purposes.

KEYWORDS: forensic science, short tandem repeat, HPRTB, F13B, LPL, population genetics, DNA typing, Brazil

The short tandem repeat (STR) loci are composed of tandemly repeated sequences of 4 base pairs (bp) in length and are abundant in the human genome. The HPRTB, F13B, and LPL STRs are highly polymorphic in every population studied (1,2). These STRs are generally less than 350 bp, and therefore suitable for amplification by the polymerase chain reaction (PCR). The amplified products of STRs can be resolved to single bases by separation on denaturing polyacrylamide gels, and the discrete data can be analyzed by population genetic methods.

Currently, there are few data regarding the allelic frequency of variable number of tandem repeat (VNTR) loci in Brazilian populations, particularly in the trihybrid population of the Northeast region. The striking feature of this population is the intermingling of individuals of Caucasian, Black and American Indian origin, in a fashion making it very difficult to identify a native with pure phenotype characteristics of any of these groups. This study describes the HPRTB, F13B, and LPL allelic frequencies and some parameters of a Brazilian population from Pernambuco (Northeast region) of Brazil that can be used for forensic science purposes.

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Material and Methods

Whole blood from 134 unrelated healthy Northeast Brazilian individuals, obtained by venipuncture, and was collected by the staff of the Human Molecular Genetics Laboratory of the Genetic Department of the Federal University of Pernambuco. The DNA was extracted using a salting out method as described elsewhere (3).

The amplification of STRs was performed using GenePrint monoplex kits (Promega, Madison, WI). The amplification reaction was carried out in a 25 μL reaction volume containing 10 ng of template DNA and 0.2 units of Taq DNA polymerase (BRL, Bethesda, MD). The reactions were performed using a Perkin-Elmer 2400 thermal cycler (Foster City, CA). Eight μL of loading buffer (10 mM NaOH, 95% formamide, 0.05% bromophenol blue, 0.05% xylene cyanol) were mixed with 2 μL of the amplification product, previously denatured at 95°C for 5 min. A total of 10 μL of the mixture was loaded into each slot of the denaturing polyacrylamide gel (7% T, 5% C, 10 cm long, and 1 mm thick), containing 7M urea and Tris-borate-EDTA buffer (TBE). The gels were submitted to an electrophoresis procedure in an EPS 300 power supply apparatus (Pharmacia Biotech, San Francisco, CA) at room temperature, 250 V, for 3 h for LPL, and 4 h for the HPRTB and F13B STRs. The specific amplification products were revealed with silver staining. Allele assignment was determined by the comparison with an allelic ladder supplied by Promega (Madison, WI).

The allele and genotype frequency was calculated by direct counting. Heterozygosity was calculated from the observed numbers of heterozygotes and homozygotes within each sample set. Possible departure from Hardy-Weinberg equilibrium was determined by the exact test; the matching probability and power of exclusion were estimated as previously reported (4,5).

Results and Discussion

This is the first report on STRs loci in the Brazilian Northeast population. The observed allele frequencies and the genotype frequencies for the three loci are shown in Tables 1 and 2. The comparisons between the observed and expected genotype frequencies of the HPRTB, F13B, and LPL loci were in agreement with Hardy-Weinberg proportions (Exact test $P = 0.3912$; $P = 0.7453$; $P = 0.5964$, respectively). For the HPRTB locus, the most frequent allele in the population studied here was the HPRTB*13 (gene frequency = 0.319). The most frequent allele for the F13B locus was the F13B*10 (0.294). The LPL*7 and LPL*14 alleles were fre-

TABLE 1—*Allelic frequencies for the STR loci HPRTB, F13B, and LPL in a population from the Northeast region of Brazil.*

Loci	HPRTB (n = 254)		F13B (n = 248)		LPL (n = 268)	
Alleles	Number	Proportion	Number	Proportion	Number	Proportion
6	—	—	49	0.198	—	—
7	—	—	22	0.089	3	0.011
8	—	—	37	0.149	1	0.004
9	4	0.016	63	0.254	19	0.071
10	5	0.020	73	0.294	97	0.362
11	37	0.146	4	0.016	65	0.242
12	75	0.295	—	—	64	0.239
13	81	0.319	—	—	16	0.060
14	42	0.165	—	—	3	0.011
15	6	0.024	—	—	—	—
16	4	0.016	—	—	—	—

TABLE 2—*Genotype frequencies of the STR loci HPRTB, F13B, and LPL in a population of Northeast region of Brazil.*

Loci	HPRTB (n = 127)		F13B (n = 124)		LPL (n = 134)	
Genotypes	Number	Proportion	Number	Proportion	Number	Proportion
6,6	—	—	4	0.032	—	—
6,7	—	—	4	0.032	—	—
6,8	—	—	6	0.048	—	—
6,9	—	—	10	0.081	—	—
6,10	—	—	21	0.169	—	—
7,7	—	—	—	—	1	0.008
7,8	—	—	4	0.032	—	—
7,9	—	—	7	0.056	—	—
7,10	—	—	6	0.048	1	0.008
7,11	—	—	1	0.008	—	—
8,8	—	—	2	0.016	—	—
8,9	—	—	12	0.097	—	—
8,10	—	—	12	0.097	1	0.008
9,9	—	—	9	0.073	1	0.008
9,10	—	—	14	0.113	9	0.068
9,11	2	0.016	1	0.008	4	0.030
9,12	1	0.008	—	—	3	0.022
9,13	1	0.008	—	—	1	0.008
10,10	—	—	9	0.073	16	0.119
10,11	—	—	2	0.016	24	0.179
10,12	1	0.008	—	—	22	0.164
10,13	2	0.016	—	—	7	0.052
10,14	2	0.016	—	—	1	0.008
11,11	2	0.016	—	—	7	0.052
11,12	16	0.126	—	—	19	0.142
11,13	10	0.079	—	—	3	0.022
11,14	4	0.032	—	—	1	0.008
11,15	1	0.008	—	—	—	—
12,12	13	0.102	—	—	8	0.060
12,13	22	0.173	—	—	4	0.030
12,14	9	0.071	—	—	—	—
13,13	12	0.094	—	—	—	—
13,14	18	0.142	—	—	1	0.008
13,15	3	0.024	—	—	—	—
13,16	1	0.008	—	—	—	—
14,14	2	0.016	—	—	—	—
14,15	2	0.016	—	—	—	—
14,16	3	0.024	—	—	—	—

quently detected in our group of individuals (both = 0.011), but only in very low frequencies in African-American individuals from North America (6).

The combined matching probability for the three loci was about 1 in 1000. The combined discrimination power for the three STRs was 0.9991. The typical paternity indexes were: HPRTB, 2.193; F13B, 2.632; LPL, 2.000. Taken together, the results reported here show that the STR used in this study are suitable for forensic identification in this trihybrid population of the Northeast Region of Brazil.

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