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Analysis of 124 SNP loci included in HID Ampliseq identity panel in a small population of Rio de Janeiro, Brazil

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ABSTRACT

Implementation of massively parallel sequencing platforms can bring a great contribution to Forensic Genetics field, with a great saving of time and costs, as well as allowing reliable results to be obtained from small or extremely degraded samples. The aim of this work was to analyze 124 SNP loci (90 autosomal and 34 Y-SNP) included in HID-Ion Ampliseq Identity Panel in a small sample from Rio de Janeiro state, Brazil. Samples from 12 non-related individuals were amplified with HID-Ion Ampliseq Identity Panel and sequenced on the Ion Torrent PGM platform (Thermo Fisher Scientific); genotypes were generated with HID SNP Genotyper plugin and forensic parameters were calculated with PowerStats v.12. All samples were successfully genotyped and were used to calculate allele frequencies, homozygosity, heterozygosity, random match probability (RMP) and exclusion power for all 90 autosomal SNP loci. Using the formula proposed by Budowle et al. (1996), only 4 of the 90 *loci* genotyped (4,4%) showed allele frequencies below the minimum required. It means that although a small set of individuals was used on this study, it may have shown a good perspective of Rio de Janeiro state allele frequencies. Among the 11 male samples analyzed, a prevalence of the haplogroup R1b of Y chromosome was observed, followed by the haplogroups E, Q and J. Such distribution reflects the results demonstrated in other studies for the population of Rio de Janeiro. All results together demonstrate the usefulness and applicability of SNP analysis on Ion Torrent PGM.

1. Introduction

Massively parallel sequencing (MPS) platforms allow the simultaneous analysis of thousands to millions of DNA fragments, generating large amounts of data in a relatively short time frame compared with traditional sequencing methods. Implementation of such analysis would allow reliable results to be obtained from small or extremely degraded samples [1]. First applications of MPS on Forensic Genetics were published in early 2010's with single nucleotide polymorphisms (SNPs) analysis [2–4].

SNPs are point mutations on DNA (substituions, insertions or deletions) that are usefull on genotyping degraded samples, as they require smaller fragments to be amplified and analysed. They are distributed throughout the human genome and can be divided in 4 categories: identity, lineage informative, ancestry informative or fenotipic [5]. A panel of 124 SNP *loci* (90 autossomal and 34 Y-SNPs) were mapped by Kidd et al [6] and the SNPforID consortium [7] and incorporated on HID-Ion AmpliSeq Identity Panel™ (Thermo Fisher Scientific), a multiplex kit for genotyping samples on Ion Torrent PGM.

The aim of this study was to analyze 124 SNP loci (90 autosomal and 34 Y-SNP) included in HID-Ion Ampliseq Identity Panel in a small sample from the state of Rio de Janeiro, Brazil.

2. Methodology

Buccal swabs from 12 non-related individuals from Rio de Janeiro state were collected on IPPGF/PCERJ. DNA was extracted by organic [8] or Chelex® method [9] and quantified by qPCR with Quantifiler HP kit (Thermo Fisher Scientific). DNA amplification was performed with HID-Ion Ampliseq Identity Panel™ (Thermo Fisher Scientific), followed by library preparation with HID-Ion Ampliseq Library ™ kit and Ion Xpress Barcode Adapters™ kit (Thermo Fischer Scientific). Emulsion

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Table 1
Forensic parameters for a sample of 12 individuals from Rio de Janeiro state based on 90 autosomal SNPs included in HID-Ion Ampliseq Identity Panel.

Parameter	Values
Allele frequencies (range)	0.042–0.958
Homozygosity (mean; min – max)	0.453 (0.083–0.833)
Heterozygosity (mean; min – max)	0.450 (0.083–0.522)
Random Match Probability (RMP) (combined; min – max)	7.643×10^{-32} (0.333–0.847)
Power of Exclusion (PE) (combined; min – max)	0.999999998518661 (0.006–0.662)

Table 2
Frequencies of Y haplogroups found in a set of 11 individuals from Rio de Janeiro state genotyped with HID-Ion Ampliseq Identity Panel.

Haplogroup	#	Biogeographical origin
R1b	5 (45.5%)	European
E	3 (27.3%)	African / Mediterranean
Q	2 (18.2%)	Native American
J	1 (9.0%)	European / Middle East

PCR and enrichment was performed with Ion PGM™ Hi-Q™ OT2 kit (Thermo Fischer Scientific) and the libraries were sequenced in Ion Torrent PGM™ with Ion PGM™ Hi-Q™ Sequencing kit and chip 318 v2 (Thermo Fischer Scientific).

Genotypes were generated with HID SNP Genotyper plugin and forensic parameters were calculated with PowerStats v.12. Estimation of minimum allele frequencies was performed as described by Budowle et al [10].

3. Results and discussion

All samples were successfully genotyped and were used to calculate allele frequencies and forensic parameters for all 90 autosomal SNP loci (Table 1). Using the formula proposed by Budowle et al [10], only 4 of the 90 *loci* genotyped (4,4%) showed allele frequencies below the minimum required. It was possible to verify that although a small set of individuals was used on this study, it may have shown a good perspective of Rio de Janeiro state allele frequencies. Results are also similar to those published in a previous study for Brazilian population [11].

Among the 11 male samples analyzed, a prevalence of the haplogroup R1b of Y chromosome was observed, followed by the haplogroups E, Q and J (Table 2). Such distribution reflects the results demonstrated in other studies for the population of Rio de Janeiro state [12].

4. Conclusion

All results together demonstrate the usefulness and applicability of SNP analysis on Ion Torrent PGM for Rio de Janeiro state population.

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Declaration of Competing Interest

None.

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