Available online at www.sciencedirect.com
ScienceDirect
Forensic Science International: Genetics Supplement Series 1 (2008) 105–106
www.elsevier.com/locate/FSIGSS

Research article

Population data for MiniNC01 in a population sample from North-eastern Italy and their use in neoplastic tissues fixed in formalin and embedded in paraffin

N. Cerri [a], L. Caenazzo [b], A. Verzeletti [a,*], F. Gasparini [a], E. Ponzano [b], F. Ceola [b], P. Tozzo [b], F. De Ferrari [a]

[a] Department of Forensic Medicine, University of Brescia, Piazzale Spedali Civili 1, 25123Brescia, Italy
[b] Department of Environmental Medicine and Public Health, Legal Medicine, University of Padua, Italy

Received 20 August 2007; accepted 9 October 2007

Abstract

To establish a database for the three MiniNC01 loci D10S1248, D14S1434, D22S1045 in a population sample from North-eastern Italy, 102 unrelated individuals were typed. DNA was amplified in a multiplex reaction with subsequent automatic detection using capillary electrophoresis. The obtained data give a contribution to the definition of Italian population miniSTRs allele frequencies for the three analysed loci. These three MiniSTRs were tested on 21 neoplastic tissues and the obtained genotypes were compared to those obtained from normal tissue. Only 3 cases (14.28%) gave a different genotype suggesting a better performance of these markers than traditional STRs.

Keywords: MiniNC01; North-eastern Italy; Population data; Neoplastic tissues

1. Introduction

A number of studies demonstrated that successful analysis of degraded DNA specimens from mass disaster or forensic evidence improves with smaller sized PCR products (MiniSTRs) [1].

Because of the few population data regarding MiniNC01 loci in Italy, 102 unrelated individuals from North-eastern Italy were typed for the three MiniNC01 loci D10S1248, D14S1434 and D22S1045. To verify the value of these markers in neoplastic tissues, they were tested on different neoplastic specimens fixed in formalin and embedded in paraffin (8 breast cancer, 7 gastric cancer, 6 primary colorectal cancer and their metastasis) that sometimes are used for forensic purposes. For each case the genetic profile obtained from the neoplastic specimen was compared to that obtained from normal tissue.

2. Materials and methods

Genomic DNA was extracted using the Chelex-100 procedure from whole blood, buccal swabs or from neoplastic tissue and their normal counterpart. PCR was performed in a GeneAmp PCR System 2400 (PE) using the protocol suggested by Coble (www.cstl.nist.gov./biotech/strbase/miniSTR). The amplification products were loaded on the ABI Prism 310 Genetic Analyser and analysed by GeneMapperID V3.2.

3. Results and discussion

This work provides a picture of allele frequencies for three mini-STRs loci in a population sample from North-eastern Italy (Table 1). As expected the distribution of the allele frequency in our population sample is close to that found in the Caucasian population; no microvariants previously described were found [1,2].

Neoplastic tissues show a great variety of genetic alterations, such as allelic deletions (loss of heterozygosity LOH), allelic insertions (microsatellite instability, MSI) or chromosomal instability [3]. In this study the typing of the neoplastic tissues was found incorrect in 3 cases on 21 (14.28%), 2 of which

* Corresponding author. Tel.: +39 030 3995838; fax: +39 030 3995839.
E-mail address: verzelet@med.unibs.it (A. Verzeletti).

1875-1768/$ – see front matter
doi:10.1016/j.fsigss.2007.10.088

Table 1
Allele frequencies and statistical evaluation

Allele	D10S1248	D14S1434	D22S1045
9			0.1225
10			0.0049
11	0.0049		0.0147
12	0.0098		0.0441
13	0.0245		0.3873
14	0.2598	0.2157	0.2941
15	0.3284	0.0735	0.1275
16	0.2059	0.0490	0.0049
17	0.1373	0.3088	
18	0.0245	0.2941	
19	0.0049	0.0392	
20		0.0049	
21		0.0147	
N	102	102	102
Observed *H*	0.7843	0.7745	0.7451
Expected *H*	0.7620 ± 0.0421	0.7620 ± 0.0421	0.7300 ± 0.0440
PE	0.5703	0.5527	0.5014
PD	0.8998	0.8972	0.8791
PI	2.3181	2.2174	1.9615
PIC	0.7238	0.7242	0.6877

H: Rate of heterozygosity, PE: power of exclusion, PD: power of discrimination, PI: paternity index, PIC: polymorphic information content.

Table 2
Genotypes observed in neoplastic tissues regarding 21 individuals (In bold the observed alterations)

Tissue	D10S1248	D14S1434	D22S1045
Gastric 1*N*	11–14	**14–15**	10–13
Gastric 1*P*	11–14	**14–14**	10–13
Gastric 2*N*	15–17	**12–14**	14–14
Gastric 2*P*	15–17	12–14	14–14
Gastric 3*N*	14–15	14–17	11–15
Gastric 3*P*	14–15	14–17	11–15
Gastric 4*N*	15–15	18–18	13–14
Gastric 4*P*	15–15	18–18	13–14
Gastric 5*N*	15–15	18–18	13–13
Gastric 5*P*	15–15	18–18	13–13
Gastric 6*N*	13–16	16–17	13–13
Gastric 6*P*	13–16	16–17	13–13
Gastric 7*N*	16–16	17–18	13–14
Gastric 7*P*	16–16	17–18	13–14
Colon 8*N*	15–16	15–17	9–13
Colon 8*P*	15–16	15–17	9–13
Colon 8*met*	15–16	15–17	9–13
Colon 9*N*	14–16	17–18	**13–14**
Colon 9*P*	14–16	17–18	**13–14**
Colon 9*met*	14–16	17–18	**13–13**
Colon 10*N*	14–14	14–14	14–15
Colon 10*P*	14–14	14–14	14–15
Colon 10*met*	14–14	14–14	14–15
Colon 11*N*	16–17	14–17	14–14
Colon 11*P*	16–17	14–17	14–14
Colon 11*met*	16–17	14–17	14–14
Colon 12*N*	15–17	**15–18**	12–13
Colon 12*P*	15–17	**15–15**	12–13
Colon 12*met*	15–17	**15–15**	12–13
Colon 13*N*	13–15	14–14	14–15
Colon 13*P*	13–15	14–14	14–15
Colon 13*met*	13–15	14–14	14–15
Breast 14*N*	14–18	14–17	12–14
Breast 14*P*	14–18	14–17	12–14
Breast 15*N*	15–17	14–16	13–15
Breast 15*P*	15–17	14–16	13–15
Breast 16*N*	15–15	17–18	12–12
Breast 16*P*	15–15	17–18	12–12
Breast 17*N*	14–16	16–18	14–14
Breast 17*P*	14–16	16–18	14–14
Breast 18*N*	15–16	17–18	13–14
Breast 18*P*	15–16	17–18	13–14
Breast 19*N*	14–15	15–17	13–14
Breast 19*P*	14–15	15–17	13–14
Breast 20*N*	14–14	16–17	13–14
Breast 20*P*	14–14	16–17	13–14
Breast 21*N*	15–15	17–18	9–14
Breast 21*P*	15–15	17–18	9–14

N: Normal tissue; *P*: pathological tissue, *met*: metastasis.

regarding colon tumors and 1 regarding a gastric cancer; no differences were found in breast cancer cases (Table 2). Locus D14S1434 has been the most prone to alterations (2 cases on 21, 9.52%) while D10S1248 gave no differences. Although STRs alterations in neoplastic tissues seem to be higher then those found in this study (Vauhkonen et al. reported a rate of 68% [4]; Ceccardi et al. reported a rate of 54.4% [5]) tumor tissues should only be used in forensics with great care, since any exclusion in identification or paternity testing may be due to alteration events in the tumor.

Conflict of interest

None

References

[1] M.D. Coble, J.M. Butler, Characterization of new miniSTR loci to aid analysis of degraded DNA, J. Forensic Sci. (50) (2005) 43–53.
[2] P. Martin, O. Garcia, C. Albarran, P. Garcia, I. Yurrebaso, A. Alonso, Allele frequencies of six miniSTR loci (D10S1248, D14S1434, D22S1045, D4S2364, D2S441 and D1S1677) in a Spanish population, Forensic Sci. Int. (169) (2007) 252–254.
[3] C. Lengauer, K.W. Kinzler, B. Vogelstein, Genetic instability in human cancers, Nature (396) (1998) 643–649.
[4] H. Vauhkonen, M. Hedman, M. Vauhkonen, M. Katja, P. Sipponen, A. Sajantila, Evaluation of gastrointestinal cancer tissues as a source of genetic information for forensic investigations by using STRs, Forensic Sci. Int. (139) (2004) 159–167.
[5] S. Ceccardi, M. Alù, F. Lugaresi, G. Ferri, C. Bini, T. Balbi, F. Ingravallo, S. Pelotti, Evaluation of reliability of STR typing in different types of cancerous tissues used for identification purpose, in: A. Amorin, F. Corte-Real, N. Morling (Eds.), Progress in Forensic Genetics, vol. 11, Elsevier, 2006, pp. 672–675.