

FOR THE RECORD

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Variation of the Hypervariable Region-1 of Mitochondrial DNA in Central-Eastern Italy

POPULATION: Blood samples were collected from unrelated individuals living and born in central-eastern Italy, in the Abruzzo region ($n = 50$). Informed consent was obtained from all donors.

KEYWORDS: forensic science, Abruzzo, mitochondrial DNA, hypervariable region-1

DNA samples were extracted from fresh blood (5 mL in EDTA tube) by alkali bursting of blood cells followed by protein digestion with proteinase K. After a phenol-chloroform extraction, DNA was precipitated with absolute ethanol and dried at room temperature. The DNA pellet was diluted in 100 μ L of TBE buffer (Tris HCl, 10mM, pH 7.5; EDTA 1mM). PCR amplification was carried out by using a Perkin Elmer 2400 thermal cycler in 25 μ L reaction volume. The product of amplification was purified with a High Pure Product Purification Kit (Roche Diagnostics GmbH, Mannheim, Germany). The sequencing reactions were performed separately on each strand with the primers L15990 and H16401. The hypervariable region-1 (HVR1) sequence analysis was carried out using the A.L.F. semi-automated sequencer (Pharmacia Biotech, Uppsala, Sweden). We observed a total of 44 different haplotypes out of 50 individuals examined, 41 of which were singletons. Only 8 haplotypes (no. 09, 13, 29, 39, 40, 42, 44, and 50) have been already observed among the Italian populations database available at www.hvrbase.de. The parameters of intrapopulation diversity (haplotype diversity 0.990 ± 0.008 ; mean number of pairwise differences 4.979 ± 2.463) fall into the range of values reported for

Italian populations (1). The values of Tajima ($1,944, p = 0.015$) and Fu's tests ($-25.552, p = 0.00$) are those expected for populations who have had a large and sudden expansion (2). Applying an exact test for population comparisons implemented in the Arlequin software (3), we obtained statistically significant p -values for the comparison between our sample and those from Tuscany (0.004) Sardinia (0.039). Complete data are available at the e-mail address of the corresponding author upon request.

References

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