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Efficient human paternity testing with a panel of 40 short insertion-deletion polymorphisms

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ABSTRACT. We developed a panel of 40 multiplexed short insertion-deletion (indel) polymorphic loci with widespread chromosomal locations and allele frequencies close to 0.50 in the European population. We genotyped these markers in 360 unrelated self-classified White Brazilians and 50 mother-child-probable father trios with proven paternity. The average heterozygosity (gene diversity) per locus was 0.48, and the combined probability of identity (matching probability) for the 40-locus set was 3.48 x 10^{-17}. The combined power of exclusion of the indel panel was 0.9997. The efficiency of the 40 indel set in the exclusion of falsely accused individuals in paternity casework was equivalent to the CODIS set of 13 microsatellites. The geometric mean of the paternity indices of the 50 mother-child-probable father trios was 17,607. This panel of 40 short indels was found to have excellent performance. Thus, especially because of its simplicity and low cost, and the fact that it is composed of genomic markers that have very low mutation rates, it represents a useful new tool for human paternity testing.

Key words: Paternity testing; Insertion-deletion polymorphisms; Single nucleotide polymorphisms; DNA; Indels

