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A VNTR polymorphism in human 5′ H19 flanking regions in Japanese and German populations

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In mammals, imprinted genes are preferentially expressed from either the maternal or paternal allele. Several recent observations show that DNA methylation plays an important role in the imprinted inheritance of the gene. Generally, an individual inherits two alleles together from his parents and the origin of each allele can be determined by typing the parents. The ultimate aim of this study is to detect a paternally or maternally derived allele from one person by using the methylation difference in the imprinted region. For the purpose, a useful probe within the region is requisite as the analytic target.

In this study, the forensic utility of the VNTR locus, which is located approximately 7.6 kb upstream of the H19 gene that is maternally expressed, was evaluated. The human 5′ H19 flanking sequence was searched from the DDBJ. The allele frequency of the VNTR locus was examined in a total of 199 unrelated Japanese and 171 unrelated German individuals. In the Japanese samples, 7 alleles and 22 genotypes were identified. The heterozygosity and polymorphism information content (PIC) were 0.749 and 0.669, respectively. By contrast, 9 alleles and 27 genotypes were detected in the German samples. The heterozygosity and PIC were 0.969 and 0.705, respectively. Thus, the frequency distribution of the two populations showed different profiles (Table 1). In a Japanese case study, this genetic typing was successfully applied to the personal identification of decomposed remains as a forensic sample. These results indicated that the VNTR in the H19-5′ flanking region is highly polymorphic and useful for personal identification of both Japanese and German DNA samples. This suggests the VNTR has become a useful probe for the aimed method.

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Table 1

Allele (repeat)	Japanese ($n = 199$)	German ($n = 171$)
14	–	0.006
13	–	0.020
12	0.060	0.085
11	0.083	0.167
10	0.091	0.023
9	0.472	0.184
8	0.015	0.020
7	0.221	0.436
6	0.058	0.059