

PENTA E VARIANT ALLELES OBSERVED IN THE JAPANESE POPULATION

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Penta E in the PowerPlex® 16 kit is a pentanucleotide tandem repeat marker located on chromosome 15, containing an AAAGA repeat motif. It has been reported that it is highly polymorphic among various populations, especially Asian populations. Out of 132 Japanese individuals, capillary electrophoresis detected two kinds of variant alleles, fragments of which were reproducibly sized as approximately one-base-shorter compared to alleles 19 and 20 in the allelic ladder marker respectively. The genotypes carrying the variant alleles were 17/19 variant, 11/20 variant, 12/20 variant and 16/20 variant. Each variant allele was isolated and collected from each sample using the Wave system, which utilizes DHPLC (denaturing high-performance liquid chromatography) in conjunction with a DNA separation cartridge. Sequence analysis revealed that both the variant alleles had a partial repeat motif, resulting in one-base-shorter alleles compared to known alleles. Following ISFG recommendations, the variant alleles were designated as 18.4 and 19.4 respectively. Despite the relatively large amplicon sizes of Penta E, an accurate allele assignment can be reliably made by capillary electrophoresis. However, alleles differing in size by one base (e.g. 18.4 and 19) were not separated and appeared as a single broad peak. Genotyper® software assigned one of the component alleles to this peak. Therefore, such broad peaks require careful interpretation so as to not overlook the other component allele contained by the peak. The ratio of peak area to peak height was a useful index to recognize a peak containing two alleles.