

ABSTRACT

Gene and haplotype polymorphisms of the Prion gene (PRNP) in Japanese Brown, Japanese native and Holstein

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Polymorphisms in the prion protein gene (*PRNP*) are known to be associated with transmissible spongiform encephalopathies in human, sheep and goats. There is tentative association between *PRNP* promoter polymorphism and bovine spongiform encephalopathy (BSE) susceptibility in cattle. In this study, we genotyped for six bovine *PRNP* polymorphic sites including a 23-bp indel in the promoter, a 12-bp indel in the intron 1, two nonsynonymous single nucleotide polymorphisms (SNPs), octapeptide repeats in the coding region and a 14-bp indel in the 3'-untranslated region in 178 animals representing Japanese Brown, Kuchinoshima feral, Mishima, Japanese Shorthorn and Holstein. In 64 Japanese Brown cattle, three indel sites were polymorphic. All of the six sites were monomorphic in Kuchinoshima. The 23-bp and 12-bp indel sites were polymorphic in Mishima cattle. The 23-bp and 14-bp indel sites were polymorphic in Japanese Shorthorn cattle. Both SNP sites were monomorphic in all cattle examined in this study. At the 23-bp indel site, the genotype frequencies of Japanese Brown and Holstein breeds were similar to that of BSE affected cattle. We estimated 12 different haplotypes from these genotypic data. A '23-12-K6S14+' haplotype was the major haplotype in all populations, whose frequencies ranged from 0.50 to 1.00.

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