Gene polymorphism of B- defensin - 1 is associated with susceptibility to periodontitis in Japanese

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Abstrak

Periodontitis is a multifactorial disease associated with genetic and environmental factors. Single-nucleotide polymorphisms (SNPs) are associated with susceptibility to common diseases such as diabetes and periodontitis. Although the oral cavity is exposed to various organisms, the conditions are well controlled by innate and acquired immune systems. Antimicrobial peptides (AMPs) play an important role in the innate immune system; however, the association of AMP-SNPs with periodontitis has not been fully elucidated. This study investigated the relationship between AMP-SNPs and periodontitis in Japanese. One hundred and five Japanese subjects were recruited, which included patients with aggressive, severe, moderate and mild periodontitis, and age-matched healthy controls. Genomic DNA was isolated from peripheral blood and genotypes of SNPs of β-defensin-1 and lactoferrin genes (DEFB1: rs1799946, rs1800972 and rs11362; and LTF: rs1126478) were investigated using the PCR-Invader assay. Protein level of AMPs in gingival crevicular fluid (GCF) was quantified by ELISA. Case–control studies revealed that the −44 CC genotype of DEFB1 (rs1800972) was associated with periodontitis (OR 2.51), particularly with severe chronic periodontitis (OR 4.15) and with combined severe and moderate chronic periodontitis (OR 4.04). No statistical differences were found in other genotypes. The β-defensin-1 concentrations in GCF were significantly lower in subjects with the −44 CC genotype of DEFB1 than in those without this genotype. No significant differences between GCF concentrations of AMPs and other genotypes were detected. The −44 CC genotype of the β-defensin-1 gene (DEFB1 rs1800972) may be associated with susceptibility to chronic periodontitis in Japanese.