Evaluation of RANK/RANKL/OPG gene polymorphisms in aggressive periodontitis

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ABSTRACT

Background and Objective: Aggressive periodontitis (AgP) is a specific type of periodontal disease that is characterized by rapid attachment loss and bone destruction. While attempting to identify genetic polymorphisms associated with AgP, previous research has focused on candidate genes that may be involved in immune responses to microbial infections. In this study, the focus was on single nucleotide polymorphisms (SNPs) in the key mediators of osteoclast differentiation and activation, which involve receptor activator of nuclear factor-κB (RANK), RANK ligand (RANKL) and osteoprotegrin (OPG), in the Japanese population. The aim of this study was to evaluate the association of RANK/RANKL/OPG gene polymorphisms with AgP in the Japanese population.
Material and Methods: We examined 99 patients with AgP and 89 controls from the Japanese population to explore the possibility of RANK/RANKL/OPG loci as candidate regions associated with the disease. All exons and relevant exon–intron boundaries of these three candidate genes were amplified by polymerase chain reaction (PCR) using 19 primers, followed by direct sequencing. The polymorphisms were identified by comparing the sequences obtained from 48 subjects.

Results: We identified 27 SNPs in RANK, including 10 novel SNPs and seven SNPs each in both RANKL and OPG. A pairwise linkage disequilibrium analysis using the $r^2$ statistic showed that some SNP pairs from the three loci are in tight linkage disequilibrium.

Conclusion: An association analysis with allelotypes showed that SNPs identified in the RANK/RANKL/OPG genes have no significant association with AgP in the Japanese population.