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Genetic polymorphisms and periodontitis

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Abstract. Genetically complex diseases like periodontitis have been extensively studied over the past decade. With the pooled knowledge about genetic polymorphisms that were claimed to play a role in the predisposition to and the progression of aggressive and chronic periodontitis, it is now possible to identify candidate genes that could act as potential risk or protective factors for the disease. Genetic researches in periodontitis were generally focused on 1) inflammatory cytokines, 2) cell surface receptors and 3) enzymes and related factors. Our reports have indicated the positive relationship between IgG Fc receptor (Fcγ R) IIIB, Fcγ RIIB, interleukin-1 receptor antagonist (IL-1RN), IgA Fc receptor (Fcα RI) gene polymorphisms and aggressive periodontitis in Japanese. Moreover, variations in the Fcγ RIIIA, Fcγ RIIA, tumor necrosis factor (TNF) receptor 2 and IL-6 gene correlated with the severity of chronic periodontitis. Periodontitis may be explained not only by the presence of specific bacteria and environmental factors, but also by the several relatively common polymorphisms with cumulative high-susceptibility profiles.

Keywords: Gene; Polymorphism; Periodontitis; Susceptibility

1. Introduction

Periodontitis is a complex disease of the tooth supporting structures. It is highly prevalent, affecting 10–15% of the population and is considered as the most common cause of tooth loss among adults [1]. Currently, there are two major forms of the disease—chronic and aggressive periodontitis [2]. Its aetiology consists of the interplay between

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anaerobic bacteria found in the biofilm, subgingival dental plaque and exaggerated immune responses in susceptible individuals. Periodontopathic bacteria initiate and repeatedly attack the host stimulating an immune response and in some susceptible individuals, this host–parasite interaction progresses to periodontal attachment and alveolar bone loss that later appear as clinical signs of periodontitis. Although bacterial plaque is essential for its initiation, the amount of plaque does not necessarily correlate with the severity of periodontitis. Risk for this disease differs in each individual and at present, several risk and susceptibility factors are proposed, as discussed in the succeeding parts of this paper.

Microbial and other environmental factors that are usually inherent to the host are believed to initiate and modulate the progression of periodontitis. However, the vast number of available scientific articles supports the indispensable role of genes in the initiation and progression of the disease. Specifically, different allelic variants can result to variations in tissue structure (innate immunity), antibody responses (adaptive immunity), and inflammatory mediators (non-specific inflammation) [3]. Genetic variations could also act as protective or risk factors for certain conditions including periodontitis [4]. For this reason, periodontitis is considered as a complex genetic disease whose phenotype is determined by both the genetic makeup and the environmental influences on the affected individual.

Results obtained from numerous genetic reports could be utilized to identify the candidate genes for periodontal disease profiling in both aggressive and chronic periodontitis. Genetic variants at multiple loci synergistically contribute to the overall disease process. Our pooled data suggest that genetic polymorphisms affect both the qualitative and quantitative aspects of host response.

The succeeding parts of this paper are divided into three sections; genetics and major forms of periodontitis, issues for candidate gene approach in periodontitis and conclusion, respectively.

2. Genetics and major forms of periodontitis

2.1. Aggressive periodontitis

Aggressive periodontitis (AP) (former name; early onset periodontitis) is a specific type of periodontitis with clearly identifiable clinical findings, rapid attachment loss and alveolar bone destruction that make it distinct from chronic periodontitis. AP is subdivided into pre-pubertal, juvenile, and rapidly progressive periodontitis.

Many reports have documented the familial pattern of AP. In a phenotypic assessment by Boughman et al. [5], they found that AP-affected siblings had higher percentage of decreased neutrophil chemotaxis and positive serum *A. actinomycetemcomitans* antibody probands than the unaffected subjects. Moreover, 11 out of the 39 AP-affected sibships had both generalized and localized AP. The convincing evidence for autosomal dominant inheritance in AP was reported by Marazita et al. [6]. This large-scale segregation analysis enrolled 104 probands with AP. The authors concluded that the disease is most likely passed through an autosomal dominant mode of inheritance in both African-American and Caucasian subjects. The penetrance was 70% in African-American and 73% in Caucasians.