

Chronic periodontitis with familial aggregation and discordant identical twins

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Abstract

Chronic periodontitis is an advancement of gingivitis that involves progressive attachment loss, pocketing, and bone loss around the teeth. It has been linked to both genetic and environmental factors, making it a multifactorial disease. This case examines a family of eight siblings with a 50% prevalence of chronic periodontitis. A strong genetic influence is not likely, since one identical twin in this family has chronic periodontitis, while the other twin is unaffected. Environmental risk factors, including smoking, emotional stress, and poor oral hygiene, are present in all affected individuals, thus appearing to play a significant role in the development of periodontitis in this family. It is important for a dentist to understand the etiology of the family's chronic periodontitis in order to provide proper treatment and management of risk factors so that good oral health can be maintained.

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Background

Periodontal diseases are a broad category of inflammatory conditions that affect the tissue and supporting bone that surround the teeth. Gingivitis and periodontitis are the most common periodontal diseases [1]. Gingivitis is a milder form, marked by inflammation of the gingiva. The inflammation is a result of the host immune response to bacteria in the oral cavity, as plaque is the initiating factor of periodontal disease [1]. Numerous other factors contribute to the development of gingivitis, including a genetic predisposition, use of certain medications, smoking, pregnancy, puberty, and the presence of systemic disease. Signs of gingivitis can progress from bleeding on probing to erythema and changes in

texture of the gingiva. Gingivitis is reversible, but if left untreated, can progress to periodontitis [1]. Periodontitis has multiple forms. This study will focus on the most common form, chronic periodontitis. Chronic periodontitis is defined as long term inflammation of the supporting tissues of the teeth with progressive attachment loss, pocketing, and bone loss around the teeth [1, 2]. It typically occurs in adults and progresses slowly, and treatment is crucial otherwise bone loss will continue. Plaque on the teeth and gingiva is the primary initiating agent, and specific bacteria including *Porphyromonas gingivalis*, *Aggregatibacter actinomycetemcomitans*, *Tannerella forsythia*, *Prevotella intermedia*, and *Fusobacterium nucleatum* have been linked to chronic periodontitis [2]. Genetic

factors do not seem to play as much of a role in chronic periodontitis as in other forms of periodontal disease. However, there is still genetic influence and possible heritable risks for more aggressive periodontal breakdown. For example, genetic variation of Interleukin -1 (IL-1) is associated with increased susceptibility to aggressive forms of chronic periodontitis [1, 3]. Environmental factors appear to have the largest role in the development and progression of chronic periodontitis. These factors include emotional stress, smoking, diabetes, increasing age, and the presence of other systemic disease [2]. Periodontal therapy aims to remove plaque and manage these risk factors in order to halt disease progression and restore oral health [1].



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Case

This case examines a family of eight siblings, four of which currently have or have had chronic periodontitis. The siblings' mother had chronic periodontitis during her lifetime. She was a heavy smoker until age 60, when she suffered a heart attack and quit smoking. She continued to suffer from chronic obstructive pulmonary disease (COPD), obesity, and other health issues as her age increased. Chronic periodontitis was one of these issues. She eventually lost or had all of her teeth extracted, becoming a complete denture patient around age 65. She was a mother to eight

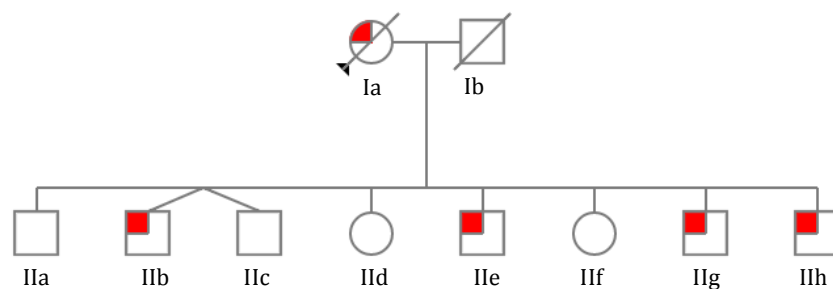
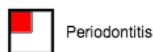


Figure 1. Pedigree of chronic periodontitis prevalence over two generations. Affected individuals marked in red. IIb and IIc represent identical twins; IIb is affected with periodontitis and IIc is not.

children, including one set of identical twin boys. Four sons are affected with chronic periodontitis, while two sons and two daughters remain unaffected, as shown in the Figure 1 pedigree. Individual IIb is 45 year-old lifetime smoker. He was recently diagnosed with periodontitis due to periodontal pocketing exceeding 8mm and mobility of multiple teeth. He has since had two teeth extracted and undergone scaling and root planing. He is the identical twin of IIc; however, individual IIc remains

unaffected. Individual IIc is a lifetime smoker with liver disease but without periodontitis. Individual IIe is 52 year-old lifetime smoker with a history of colon cancer and periodontal disease. Individual IIg is a 58 year-old lifetime smoker currently undergoing treatment for chronic periodontitis. The final individual affected, IIh, is a 60 year-old lifetime smoker with a history of drug abuse and periodontitis. He had full-mouth extractions at age 58 and began wearing complete dentures. He no longer has periodontitis and continues to maintain his oral health. Individuals IIa, IIc, and IIf remain unaffected; they are all non-smokers with good oral hy-

giene and minimal emotional stress.

Discussion

This case demonstrates the multifactorial nature of chronic periodontitis. Although genetic factors may make the children of this family more susceptible to periodontal disease, they do not seem to be the major determinant. The set of twins, IIb and IIc, support this claim. The twins are identical, meaning they share the same DNA, yet IIb has chronic periodontitis and IIc

does not. Thus, there must be additional factors contributing to the development of the disease in individual IIb. These factors are most likely environmental factors, including smoking, poor oral care and emotional stress. Since all four affected individuals are males and their mother was affected, X-linked inheritance could be considered. However, this has not been shown to be a common mode of inheritance of chronic periodontitis, as the specific genetic influences of chronic periodontitis still remain uncertain. The possible environmental risks of the affected individuals of this case have been specifically linked to chronic periodontitis. Furthermore, three of the four unaffected individuals do not display any of the environmental risk factors of chronic periodontitis. The environmental factors thus seem to play a significant role in the development of periodontal disease in this family, with genetics possibly contributing additional susceptibility or risks. It is important for a dentist to understand the etiology of the family's chronic periodontitis in order to provide proper treatment and management of risk factors so that oral health can be maintained. If the environmental factors are not managed, long-term treatment will not be successful. Bone and tooth loss will continue unless the periodontal disease is successfully treated [1]. Successful treatment involves mechanical debridement via scaling and root planing, in addition to managing environmental factors that may be contributing to the disease. Depending on the severity, some effects of the periodontal destruction may be irreversible. Patient specific treatment planning must be completed in order to determine the best treatment option, which may include tissue or bone grafts, extractions, and tooth replacement.

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