Hereditary Gingival Fibromatosis: A Family Study

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Abstract

Hereditary gingival fibromatosis is considered a rare disease, which appears in the oral cavity. Its prevalence is 1:750000. A gradually progressive of the gingival characterizes the condition. Usually hereditary gingival fibromatosis appears with autosomal-dominant inheritance but it can also appear with autosomal-recessive inheritance. The most common mutation is the Son of Sevenless-1 gene. It can be either non-Syndromic or Syndromic. The onset is usually with the eruption of permanent dentition. All treatment modalities aim to the removal of the excessive gingival tissues. However, there is recurrence of the condition after miscellaneous period of time. This study is about a three generation appearance of Hereditary Gingival Fibromatosis, occurred only in female members of the family. Its biological mechanism, diagnosis and treatment are described.

Keywords: Hereditary Gingival Fibromatosis; Biological Mechanism; Diagnosis; Treatment

Abbreviations

HGF: Hereditary Gingival Fibromatosis

Introduction

Hereditary gingival fibromatosis (HGF) is a rare oral disease, characterized by a gradually progressive enlargement of the maxillary and mandibular gingiva, with a prevalence of about 1:750000. HGF is considered an autosomal-dominant disease, however, there are studies demonstrating that it can also be an autosomal-recessive inheritance [1]. Different mutations have shown association with HGF [2]. One of the most common is a single nucleotide polymorphism (SNP) in the Son of Sevenless-1 gene (SOS-1) [3].

In order for diagnosis to be placed, histological examination is needed to affirm the gingival condition. Histologically HGF is characterized by fibrous enlargement of the gingiva. The connective tissue appears highly collagenized, avascular, with sparse differentiated fibroblasts and very few inflammatory infiltrates [4].

HGF can occur as an isolated disease (non-Syndromic) limited only at gingival or as a part of a syndrome [1]. Zimmerman-Laband, Cowden's, Murray-Puretic-Drescher, Rutherford, Ramon, Jones, Cross, Cantu, Schinzel-Giedion, Sweet-like, Amelogenesis imperfecta, Prune-belly syndrome are the most common syndromes, which HGF can be part of. Moreover, HGF can be in conjugation with hypertrichosis, epilepsy, mental retardation, hearing deficiencies, hypertelorism, supernumerary teeth and deficiency of growth hormone caused by lack of growth hormone release factor [5-7].

The onset of the gingival overgrowth usually coincides with the eruption of permanent incisors. Furthermore, the presence of teeth seems to be necessary for HGF to occur, because the condition disappears or recedes with the tooth loss [8].

As far as the HGF clinical expression is concerned, it is very heterogeneous. The gingival enlargement is fibrotic and may interfere with speech, mastication, occlusion and facial appearance. Although the gingival enlargement does not directly affect the alveolar bone, pseudopockets can exist, which facilitate plaque accumulation and subsequent bone loss [4].

Different treatment options have been suggested in the literature. All modalities aim to remove the excess tissue and can be conventional surgery, electro surgery, an apical positioned flap and lasers [9].

Thus, this aim of this study is to add a family study of this rare condition to bibliography.

**Case Report**

A 9-year-old female, with nothing significant at her medical record, showed up at the Postgraduate Clinic of Periodontology of the Aristotle University of Thessaloniki, Greece with HGF. HGF appeared as non-Syndromic and only her gingival were affected. Clinical examination revealed the presence of pseudopockets, especially in the anterior teeth leading to secondary inflammation. Clinical crowns were extremely short. Figures 1, 2 and 3 demonstrate the initial examination of the girl (Figures 1-3).

Figure 1

Figure 2

The relatives of the patient have also been diagnosed with HGF. The grandmother and mother, 70 and 46 years old respectively, reported undergoing gingivectomy during their childhood. Especially, the mother underwent a second surgery of gingivectomy because of tissue relapse after the first surgery. Furthermore, the sister of the patient, a 6-year-old girl, showed clinical characteristics of the disease. Diagnosis was based on the family history and the clinical appearance and HGF diagnosis was placed (Figures 4-6).
Treatment plan in this case consisted of three phases of periodontal therapy. Phase I included etiological therapy with oral hygiene instructions, scaling and root planing. After reevaluation of the first phase of therapy, periodontal surgery was also performed as part of phase II treatment. Gingivectomy with open flap debridement and extraction of deciduous teeth were done during periodontal surgery (Figure 7-9). After completion of phase II therapy, the patient was scheduled for maintenance or phase III therapy. During maintenance, the patient presented with significant relapse of gingival overgrowth (Figure 10, 11) and a second gingivectomy-open flap debridement took place.
Peripheral blood was also collected by standard venipuncture from the 9 year old girl, the mother and the grandmother, who all presented with signs of HGF. White blood cells were isolated and RNA was purified. The RNA was reversed transcribed to cDNA. The cDNA products were used as templates for partial amplification of SOS-1 mRNA by using the PCR primers reported by Hart, et al [3]. The PCR products were subsequently sequenced. The single nucleotide insertion mutation in codon 1083 of the SOS-1 gene was not detected.
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Discussion

Despite the fact that HGF may be part of a syndrome, this case is non syndromic. Examples of syndromes which HGF noticed more frequently are Juvenile Hyaline Fibromatosis, Amelogenesis imperfecta Zimmermann-Laband Syndrome, Jones Syndrome, Klippel-Trenaunay-Weber Syndrome, Ramon Syndrome and hypetrichosis [7].

The SNP in the SOS-1 gene was not detected, suggesting that another mutation might be linked with the disease. This familial case of HGF had incomplete penetrance, since not all members of the family showed characteristics of the disease. The fact another male sibling had normal gingival might imply that another mutation in the X chromosome can be associated with HGF. Other studies attempted to find another mutation in the GINGF loci with no positive results [10-12]. Nonetheless, Hakki., et al. found a CMG-2 mutation at their cases [13].

Fibromatosed gingival may hinder tooth eruption, mastication, oral hygiene, speech, lip closure, chewing and even patient’s self-esteem [14]. In this case the patient experienced only speech impairment. Thus, all treatment approaches aim to remove surgically the excessive gingival tissues. These approaches can be conventional surgery, electro surgery, laser surgery, apically positioned flap. Goyal., et al. suggest that external bevel gingivectomy is the most preferable method, especially when there is no bone loss [6]. A periodontal flap procedure can be used if there are large areas of gingival overgrowth or attachment loss and periodontal lesions [15]. Finally, the use of diode laser technique presents advantages to a better visibility during the procedure, minimal post-operative discomfort and pain, and better esthetic results. Nevertheless, the high cost of the equipment and the time consumption remain the major disadvantages [16]. In this case gingivectomy with open flap debridement took place.

Moreover, a second surgery was conducted because of presented relapse of the gingival overgrowth. According to Emerson the best time points for surgical intervention are at the ages of 3, 6, 12 years. Since the recurrence rate after the surgery is high, he also recommended that the best time for retreatment is after completion of the permanent dentition [17].

Conclusions

Hereditary Gingival Fibromatosis in the presented case is familial with incomplete penetrance. Thus, since exact etiology and pathogenesis have not been yet established, there is a need for further research regarding genetic inheritance of the disorder.

Patient’s motivation with oral hygiene instructions and frequent recall appointments is of outmost importance in order to eliminate inflammation and bone loss due to plaque accumulation in the areas of excessive gingival tissues. Tooth prognosis is usually good, although partial relapse of the disease is frequent.

In the future identification of all the mutations that can cause HGF and genetic screening of families that present with HGF can be very important in treatment of HGF and prevention of bone loss.

Conflict of Interest

No conflict of interest exists.

Bibliography


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