

Extensive Gingival Enlargement in Siblings

A case report

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ضخامة اللثة الشاملة عند الأشقاء

تقرير حالة

كومار بوشبانشو، راتشنا كوشيك، آراس ساداوين، رافي أثاويل

المخلص: يتميز وُرامُ اللثة الليفي بدرجات متفاوتة من فرط نمو اللثة التليفي الذي يمكن أن يسببه عوامل متنوعة. النوع الوراثي من وُرام اللثة الليفي هو اضطراب نادر، يتميز بكونه حميدا، وينمو ببطء في الخلايا المتقرنة للثة. يمكن أن تنتج الحالة من اضطرابات صبغية جسدية سائدة أو متنحية، والنوع الأول أكثر انتشارا. وعادة ما يكون اضطرابا معزولا، غير أنه يمكن أن يكون ضمن متلازمة تشمل أعضاء عديدة في الجسم. وتبعاً لذلك يمكن تقسيم وُرام اللثة الليفي الوراثي إلى نوعين: وجوده مع المتلازمة أو بمفرده بدون متلازمة. كما يمكن أن تكون ضخامة اللثة موضعية أو عامة، ولكنها عادة ما تصيب كلا القوسين. هذا وصف لحالة من وُرام اللثة الليفي الوراثي الشديد المنفرد، شاملا قوسي الفك العلوي والسفلي في أخوين. يركّز هذا التقرير على تشخيص وعلاج المرض والسيطرة عليه. كما تم التأكيد على النمط الوراثي والخصائص النسيجية المرضية.

مفتاح الكلمات: جسدية سائدة، وُرامُ اللثة الليفي، وراثي، الأشقاء، تقرير حالة، الهند.

ABSTRACT: Gingival fibromatosis is characterised by varying degrees of fibrotic gingival overgrowth that can be caused by a variety of aetiological factors. Hereditary gingival fibromatosis (HGF) is a rare genetic disorder, characterised by a slowly progressive, benign enlargement of keratinised gingiva. The condition may be found in an autosomal dominant or autosomal recessive mode of inheritance, the former being more common. It usually develops as an isolated disorder but can be one feature of a multisystem syndrome. Accordingly, HGF has been divided into two forms: non-syndromic and syndromic. The gingival enlargement can be localised or generalised, but usually involves both arches. The authors describe a case of non-syndromic generalised severe HGF, involving the maxillary and mandibular arches in two brothers. This report focuses on the diagnosis, treatment, and control of the disease. The pattern of inheritance and histopathologic characteristics are also emphasised.

Keywords: Autosomal dominant; Gingival fibromatosis; Hereditary; Siblings; Case report; India.

GINGIVAL HYPERPLASIA IS AN UNUSUAL condition which interferes with speech, mastication, occlusion, lip continence, and facial appearance of the affected individual, causing aesthetic, functional, psychological, and masticatory disturbances. Drug-induced gingival enlargements are the commonest form. The inherited condition in which the gingival tissue spontaneously and progressively enlarges is identified as hereditary gingival fibromatosis (HGF).¹ It is a rare condition, affecting only one in 750,000 people, with males and females being equally affected.² HGF is characterised by a slowly progressive, non-haemorrhagic fibrous enlargement of gingival tissue, occurring peripheral

to the alveolar bone, which does not involve the periodontal ligament. The hyperplastic gingival tissue usually presents with a normal colour and has a firm consistency with abundant stippling.^{3,4} Furthermore, HGF usually develops as an isolated disorder but can be one feature of a syndrome or a chromosomal abnormality. Accordingly, it has been divided into two forms: non-syndromic and syndromic.⁵

The gingival enlargement usually begins with the eruption of the permanent or deciduous dentition.¹ The growth may worsen throughout adolescence, suggesting an influence of growth hormones.⁶ The clinical presentation of HGF is highly variable, both in the number of teeth involved and degree of

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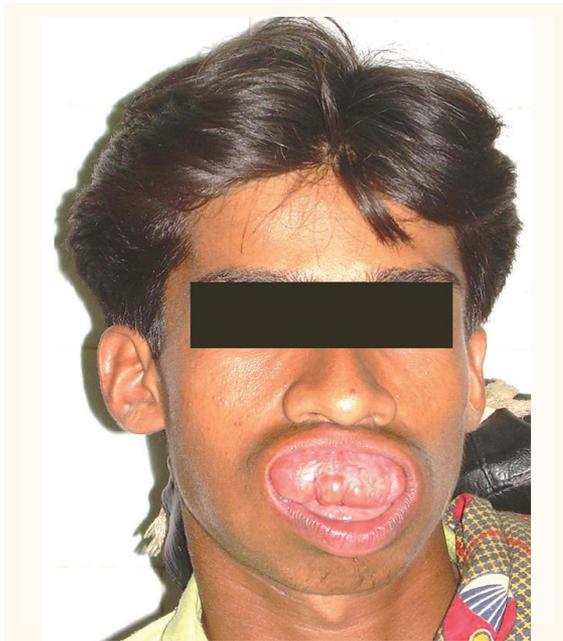


Figure 1: Severe gingival enlargement protruding out of the oral cavity, preventing lip closure.

severity. Gingival enlargement may be generalised to all gingival areas, or quite focal and limited in its distribution. The severity may range from slight enlargement to total coverage of the dentition. Buccal and lingual tissues of both maxilla and mandible may be involved, and the degree of hyperplasia may vary between individuals within the same family.^{2,7}

We report a rare clinical presentation of an extensive hereditary gingival fibromatosis in two siblings. The report focuses on the clinically relevant aspects of HGF, highlighting an unusually severe non-syndromic gingival fibromatosis. Diagnosis was based on family history, and clinical and radiographic assessments. Family history suggested an autosomal dominant inheritance.

Case Report

A 19-year-old unmarried male reported enlarged gums from the time that he was six years old. According to the patient, the swelling appeared at the time of eruption of permanent teeth without any associated pain; however, it caused functional and masticatory difficulty and the patient was unhappy with the appearance of his gingiva. His medical history was unremarkable. He denied taking any medications and did not appear to have any mental impairment. His family history was of significance,

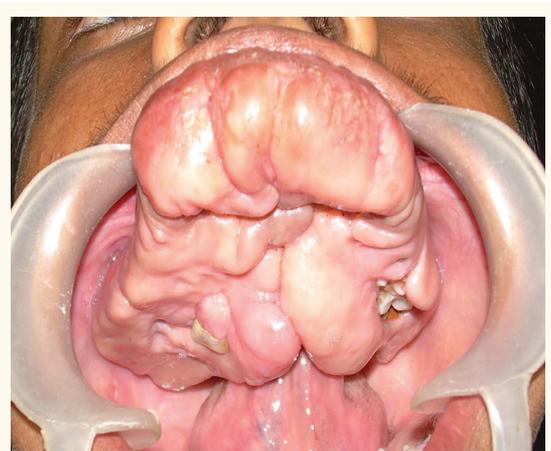


Figure 2: Gingival enlargement seen obliterating the palatal vaults space and completely embedding maxillary teeth.

since his brother had a similar condition. Genetic history revealed that the patient's deceased mother had a similar gingival condition; however, her medical and dental records were not available to confirm this. Neither the patient's maternal aunt nor the patient's sister had the disease.

Extraoral examination showed facial disfigurement with incompetent protruding lips. Excessive gingival growth was observed protruding out of the oral cavity, which interfered with lip closure [Figure 1]. Intraoral examination revealed severe gingival overgrowth, reddish pink in colour, involving both the maxillary and mandibular arches on both buccal and the lingual/palatal sides. Gingival tissue was firm, dense and fibrotic in consistency, and was non-haemorrhagic and non-tender. The enlargement in the maxillary arch obliterated the vestibular as well as palatal vault spaces and caused obliteration of the maxillary teeth and palatal displacement of the right maxillary second molar tooth [Figure 2].

Examination of the patient's 24-year-old brother revealed generalised gingival enlargement, though of a lesser severity. Based on the above findings and in combination with the patient's family history and absence of any relevant medication history, a provisional diagnosis of HGF was made.

Radiographic examination revealed generalised moderate bone loss [Figure 3]. An incisional biopsy was performed and the tissue was examined by an oral pathologist. The histopathological examination revealed a parakeratinised stratified squamous acanthotic epithelium with thin long rete ridges



Figure 3: Panoramic radiograph of 19-year-old patient showing moderate generalised bone loss.

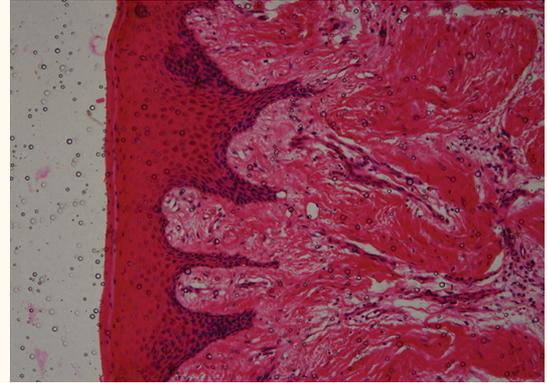


Figure 4: Well-structured epithelium displaying elongated rete pegs and dense connective tissue (original magnification x10).

extending into the connective tissue [Figure 4]. The underlying connective tissue showed dense wavy bundles of collagen fibres, containing numerous fibrocytes and fibroblasts. Deep connective tissue was characterised by dense, mature parallel collagen bundles. A mild chronic inflammatory cell infiltrate was also noted. These microscopic findings supported the diagnosis of HGF.

A multidisciplinary approach to treatment was considered. Treatment included full-mouth gingivectomy, extraction of mobile teeth, and prosthetic rehabilitation with upper and lower partial dentures. Quadrant by quadrant external bevel gingivectomy was planned in association with gingivoplasty followed by 0.2% chlorhexidine oral rinses twice a day for two weeks after each surgery. However, the patient did not report back to the hospital for the treatment and was lost to follow-up.

Discussion

Generalised gingival fibromatosis can be caused by a number of factors, including inflammation, leukaemic infiltration, and medication use such as phenytoin, cyclosporine, or nifedipine.² It can also be inherited (HGF). Gingival enlargement results in both aesthetic and functional problems in affected individuals. The most common effects are diastemas, malpositioning of teeth, prolonged retention of primary dentition, delayed eruption, cross and open bites, prominent lips, and open lip posture.¹ Severe overgrowth can result in crowding of the tongue, speech impediments, and difficulty with mastication.⁸ Although the gingival enlargement does not directly affect the alveolar

bone, the gingival swelling may increase the bacterial plaque accumulation, causing periodontitis, bone resorption, and halitosis.¹

In the present report, both patients had functional discomfort, and they were unhappy with the appearance of their gingiva. HGF is mainly inherited in an autosomal-dominant manner, although autosomal-recessive inheritance has also been reported.⁵ HGF as an isolated feature is believed to be expressed as an autosomal dominant trait.⁹ According to the genetic history of the present case, autosomal-dominant inheritance is a feasible diagnosis because family members of both sexes were affected, and the condition was present in two successive generations (mother and two sons); however, there was no history of consanguinity in the family.

HGF can develop as an isolated disorder affecting only gingiva, or more rarely as part of a syndrome such as Murray-Puretic-Drescher syndrome (multiple hyaline fibromas), Rutherford syndrome (corneal dystrophy), Laband syndrome (ear, nose, bone and nail defects with hepatosplenomegaly), Jones syndrome (progressive deafness), and Cross syndrome (microphthalmia, mental retardation, athetosis and hypopigmentation).² Syndromic gingival fibromatosis has been associated with ancillary features such as hypertrichosis, mental retardation, epilepsy, progressive sensorineural hearing loss, and abnormalities of the extremities, particularly of the fingers and toes.¹⁰⁻¹¹

Clinically, HGF is characterised by a firm, painless gingival growth, which is not especially liable to trauma.¹ The hyperplastic gingiva usually

presents a normal colour with abundant stippling.³ The clinical manifestations of both the cases reported here were consistent with most descriptions in the literature. HGF can vary from focal sites of gingival enlargement to generalised involvement, with the degree of overgrowth varying from slight to severe.¹² Unlike drug-induced gingival overgrowth, HGF is not influenced by plaque, and the incidence and severity of the disease appears to depend on the penetrance of the mutated gene.⁸ The patients reported here exhibited a generalised and severe gingival overgrowth. Furthermore, the condition seemed to become evident with the emergence of the permanent dentition. It is evident from the previous reported cases that the condition usually begins at the time of eruption of the permanent dentition, but it can develop with the eruption of the deciduous dentition and is even more rarely seen at birth.¹² In a study involving 17 family members with gingival fibromatosis, Fletcher reported that the most extensive enlargement appeared to occur either during loss of the deciduous teeth or in early stages of eruption of the permanent dentition.⁴ He noted that the enlargement seemed to progress rapidly during "active" eruption and decrease with the end of this stage. He also reported that the presence of teeth appears to be necessary for HGF to occur because the condition is not seen before the eruption of teeth and disappears or recedes with the loss of teeth.⁹

The histologic features of the gingiva were also consistent with previously reported cases. The gingival tissues were composed of fibrous connective tissue, with elongated and thin papillae.^{5,12} Histologically, HGF is benign, the main feature being accumulation of mature collagenous connective tissue.⁷ The results of the histopathologic evaluation of the biopsied tissues of our patient were consistent with those for fibrous gingival hyperplasia.¹¹ However, the histologic features of gingival fibromatosis are usually nonspecific and a definitive diagnosis should be established based on family history and clinical findings.

HGF cannot be cured, but can be controlled with varying degrees of success. When the enlargement is minimal, good scaling of teeth and home care may be all that is required to maintain good oral health. As the excess tissue increases, appearance and function indicate a need for surgical intervention.² Many techniques have been used for the excision

of the enlarged gingival tissues, including external or internal bevel gingivectomy in association with gingivoplasty, an apically positioned flap, electrocautery, and carbon dioxide laser.¹ In consideration of the severity of the involvement in this case, external bevel gingivectomy in association with gingivoplasty was planned. Several authors have reported the recurrence of hyperplastic tissue following gingivectomy, necessitating a repeat of the procedure.^{13,14} This often causes a further increase in the psychological and emotional stress of parents and patients; hence, psychological counselling is a must for both parties.¹⁵ Although recurrence is unpredictable, it is most often seen in children and teenagers rather than adults.¹ The correct physiologic contour of marginal gingiva and good plaque control are important to prevent recurrence. Normally, recurrence is minimal or delayed if good oral hygiene is achieved by a combination of monthly examinations with professional cleaning and oral hygiene instructions.¹⁵ Although there is a large consensus on the modality of treatment to be performed in HGF patients, there are controversies with regard to the exact period in which it should be accomplished.¹⁵ According to several authors, the best time is when all of the permanent dentition has erupted, because the risk of recurrence is higher before then.³ However, in some cases, a delay in surgical treatment may result in significant consequences for the patient, such as primary dentition retention with a delay in the eruption of permanent teeth, difficulties in mastication and phonation, malpositioning of teeth, aesthetic effects, and psychological problems for the patients and relatives.^{5,15} Thus, if the patient is unhappy with his/her gingival condition or if the enlarged tissue interferes with normal functioning, the treatment should be performed once the patient is cooperative and shows good oral hygiene.¹ Cosmetic concerns aside, a compromised oral cavity may cause several functional and periodontal problems. In addition, social consequences can be dramatic, forcing patients to lead an isolated, reclusive life. The local and psychological benefits, even temporary, must not be underestimated and may outweigh the probability of recurrence.¹

Conclusion

We have reported two siblings affected with HGF, with an autosomal dominant mode of inheritance. The diagnosis was confirmed by the typical presentation, family history and histopathological features. Although the clinical and histopathological aspects of HGF are well understood, the pathogenic mechanism still remains unclear. Surgical intervention is the usual treatment of HGF, but the patients still have to deal with the risk of recurrence. Once the correlations between gene mutations, molecular changes, histology, and clinical situation are clear, they can be applied clinically, providing novel methods for disease prognosis and diagnosis and targets for disease prevention and treatment.

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