

CASE REPORT

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Gingival Enlargement with Marfan Syndrome: A Case Report

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ABSTRACT

Marfan syndrome (MFS) is an autosomal dominant and multisystemic disorder affecting the connective tissues. Patients with MFS may exhibit characteristic oral features including maxillary protrusion, high palate, crowded teeth, and fragility of the temporomandibular joint. Periodontal manifestations may include a higher prevalence of gingivitis and/or periodontitis owing to the high concentration of elastic fibres in the periodontal ligament. This case report describes the management of gingival enlargement associated with MFS. The patient was successfully managed with non-surgical and surgical periodontal therapy. In patients with oral manifestations of systemic disorders, patient motivation and effective treatment planning are able to achieve the optimum periodontal and oral health outcomes.

Keywords: *Gingival enlargement; gingivectomy; Marfan syndrome; oral health; periodontal disease*

INTRODUCTION

Marfan syndrome (MFS) is an autosomal dominant disorder affecting the connective tissues. It is associated with mutations in the fibrillin-1 (FBN1) gene located on chromosome 15q21.1 that are responsible for alterations of the glycoprotein fibrillin-1, a major component of the 10–12 nm microfibrils present in the connective tissue matrices (Staufenbiel *et al.*, 2013; Pepe *et al.*, 2016). These microfibrils and elastin form the elastic fibres found in various tissues, including the suspensory ligament

of the lens, skeletal system, lungs, blood vessel walls, and the skin. Thus, MFS is a multisystemic disease where localisation and degree of symptoms are individually different. The reported prevalence of MFS was 6.5/100,000 in the Danish population, with a wide age range at diagnosis of zero to seventy-four years (Groth *et al.*, 2015). MFS was found to be the highest number of patients among the top 10 rare diseases in Malaysia (Shafie *et al.*, 2020).

MFS is considered a serious disorder because of its cardiovascular complications, which

cause 95% of deaths in patients with the condition and reduce life expectancy by up to 40%. This condition can also include mitral valve prolapse, dilatation of the main pulmonary artery in the absence of valvular or peripheral pulmonary stenosis, and calcification of the mitral annulus in the patient younger than 40. As for skeletal involvement, a tendency toward tall stature with long, slim limbs, pectus carinatum, muscle hypotonia, joint hypermobility, and a typical narrow facial appearance with a highly arched palate can be present (De Coster *et al.*, 2002). The main features in the ocular system include eye lens subluxation and severe myopia, found in 60% of the affected patients.

Patients with MFS may exhibit characteristic oral features, including maxillary protrusion, high palate, crowded teeth, and fragility of the temporomandibular joint. Periodontal manifestations such as a higher prevalence of gingivitis and periodontitis may be occurred due to a high concentration of elastic fibres in the periodontal ligament (Staufenbiel *et al.*, 2013). This case report describes the oral management of MFS presented with periodontal disease associated with gingival enlargement.

CASE REPORT

A 24-year-old patient was referred to the Periodontal Clinic, Hospital Universiti Sains Malaysia (HUSM) to manage gum swelling and bleeding during toothbrushing, which worsened over the past few months. The patient was diagnosed with MFS since birth and later, scoliosis when she was 12 years old. No similar history of MFS in the family was reported. She is under regular follow-up for MFS and will undergo treatment for scoliosis correction at HUSM.

Extraorally, she presented with cutaneous hyperpigmentation over her chin, mandible,

and arachnodactyly (overgrowth of fingers). The characteristic of the Walker-Murdoch sign (full overlap of the distal phalanges of the thumb and fifth finger when wrapped around the contralateral wrist) and the Steinberg sign (distal phalanx of the thumb fully extends beyond the ulnar border of the hand when folded across the palm) were also present (Pepe *et al.*, 2016) (Fig. 1). She has competent lips, but mild facial asymmetry deviated to the left upon smiling.

Intraorally, her buccal and labial mucosa and floor of the mouth appeared to have prominent dilated blood vessels (Fig. 2). She has a high-arched, deep palate with notable dental crowding, especially on her upper dentition. Occlusion assessment showed a Class 1 relationship with mild crowding and crossbite between teeth 21 and 31 due to the palatally incline of tooth 21. Gingival enlargement was mainly at the lower anterior teeth, which appeared edematous, erythematous, and bleeding upon probing, with a soft consistency and absence of stippling. Mild enlargement is also seen at the labial interdental papilla of tooth 21. Multiple restorations, retained roots and carious teeth were also noted. She has poor oral hygiene with generalised plaque and calculus accumulation on all teeth. The full mouth plaque and bleeding score was 100% at the initial visit. False pocketing at the gingival enlargement region showed a maximum probing pocket depth of 8 mm. Intraoral clinical features of the patient are shown in Fig. 3.

The orthopantomogram radiograph taken during her first visit showed no abnormality on either condyle, maxillary sinuses, or inferior dental nerve canal. No significant bone loss and periodontal ligament space widening observed on both maxillary and mandibular teeth. Multiple retained roots and radiolucency areas were noted on the crown indicating carious teeth (Fig. 4).



Fig. 1 Extraoral characteristic of patient with Marfan syndrome. (a) Cutaneous hyperpigmentation over the chin and mandible with mild facial asymmetry deviated to the left upon smiling. (b) Overgrowth of the fingers is generally a subjective finding (arachnodactyly). (c) Distal phalanx of the thumb fully extends beyond the ulnar border of the hand when folded across the palm (Steinberg or thumb sign). (d) Full overlap of the distal phalanges of the thumb and fifth finger when wrapped around the contralateral wrist (Walker-Murdoch sign).

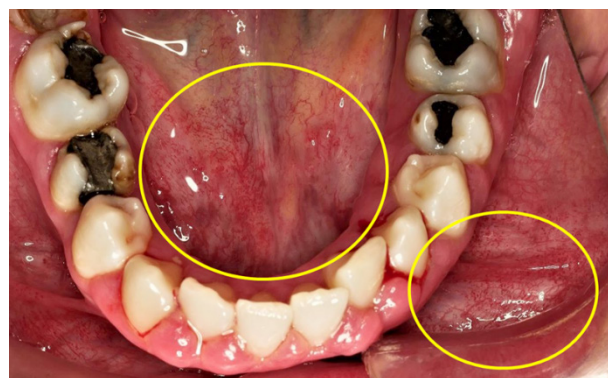


Fig. 2 Intraoral clinical photograph of the patient showed prominent dilated blood vessels of the buccal mucosa and floor of the mouth (yellow circle).

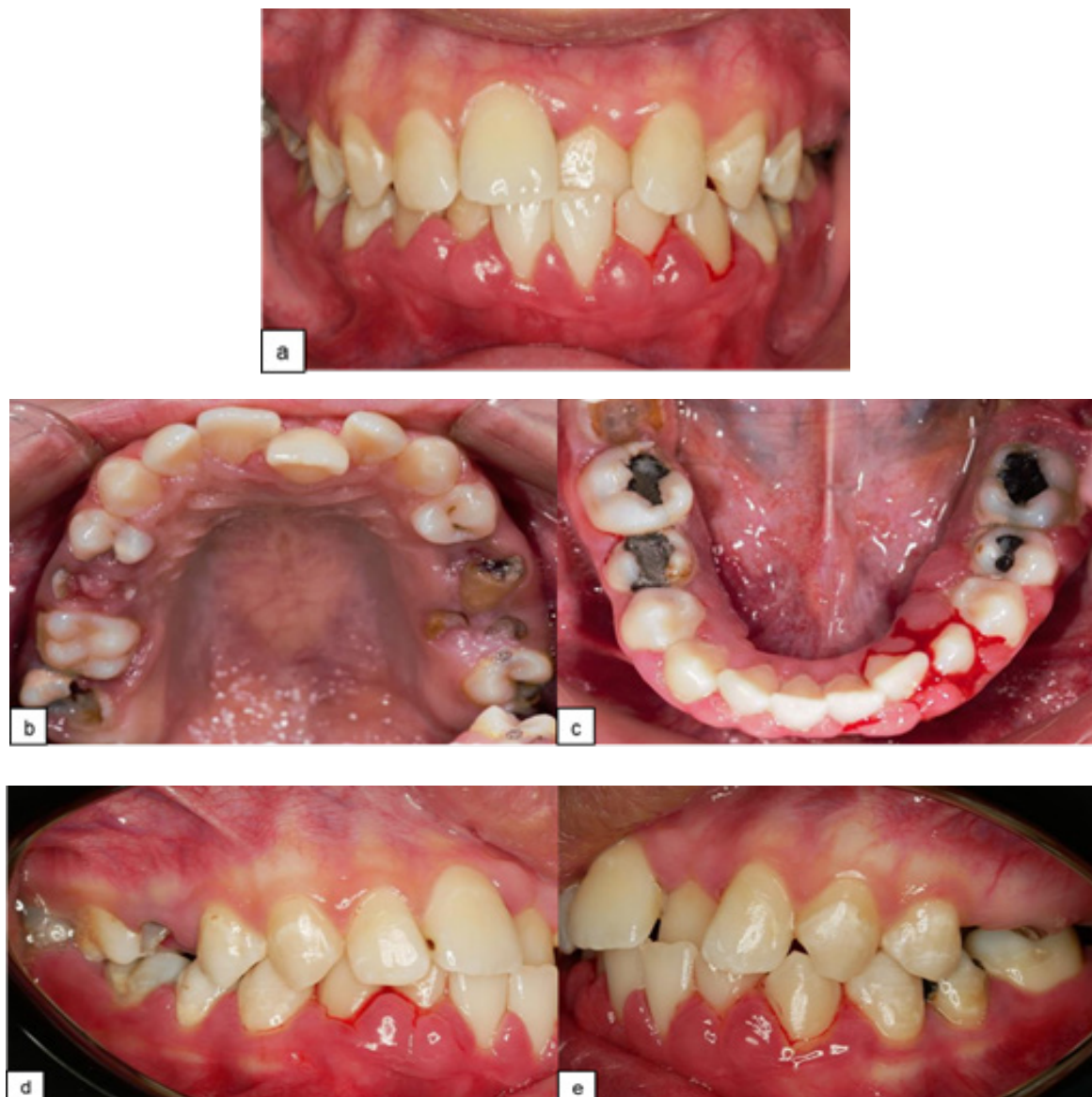


Fig. 3 Intraoral clinical photograph of a patient at baseline. Note that presence of gingival enlargement at lower anterior region which is edematous and easily bleeds upon probing. Multiple carious and retained roots of upper teeth can be seen. (a) Frontal view. (b) Maxillary occlusal view. (c) Mandibular occlusal view. (d) Right buccal view. (e) Left buccal view.



Fig. 4 Orthopantomogram radiograph of patient at the initial visit.

The patient was then diagnosed with generalised gingivitis and gingival enlargement associated with MFS. The therapeutic goals for her were to improve oral hygiene, remove local factors that favour plaque retention, control periodontal inflammation and restore the aesthetic and masticatory function. Non-surgical periodontal therapy was characterised by oral hygiene coaching and motivation, scaling, root debridement and local irrigation of the pockets using chlorhexidine gluconate 0.2% at staged appointments. Restorations of carious teeth, topical fluoride application and tooth extractions for teeth that are irrational to treat were carried out to eliminate existing infection and arrest the progression of the disease.

Upon a few visits of initial therapy and improvement of her oral hygiene, the gingival enlargement remained despite a reduction of plaque (15%) and bleeding scores (3%). Thus, gingivectomy was carried out under local anaesthesia (Mepivacaine 2% with Adrenalin 1:100,000) by using the combination of a scalpel blade and electrosurgery (Servotome, Acteon) due to the profuse bleeding tendency upon manipulation of soft tissues. Excess gingival tissues were removed by external bevel incision, and the gingiva was contoured according to the scalloping borders of teeth. The hemostasis was achieved, and Gengigel paste was applied to the surgical area before the periodontal dressing with Coe-Pak™ (GC, America). The patient was prescribed Tab mefenamic acid 500 mg thrice daily

for five days and Chlorhexidine mouthwash 0.12% twice daily for one week. The gingivectomy procedure is shown in Fig. 5.

The patient was then reviewed one week and every week for up to a month, followed by monthly visits. The healing was uneventful, and complete tissue healing was observed after one month of review appointment, as shown in Fig. 6. There was also a reduction of pocket depth to less than 4 mm, and mild swelling of the interdental papilla of tooth 21 was still present, but no bleeding upon probing was noted. The patient was then scheduled for orthodontic treatment to manage cross bite and mild crowding. However, based on a discussion with a medical specialist, it was put on hold as she will undergo surgery to correct her scoliosis. Options for replacing missing teeth post orthodontic treatment were also discussed with the patient. Based on periodontal risk assessment (PRA) and considering the history of multiple carious teeth and retained roots, she was put under supportive periodontal therapy (SPT) at three months intervals in the first-year recall schedule (Lang & Tonetti, 2003). Upon review at one year in SPT, her periodontal health was satisfactory, by which the patient could maintain good oral hygiene status with less than 10% plaque and bleeding score (Fig. 7). Therefore, further review for periodontal evaluation was set at six monthly intervals. Patient is satisfied with her current oral health status and looking forward to the regular check-up as planned.

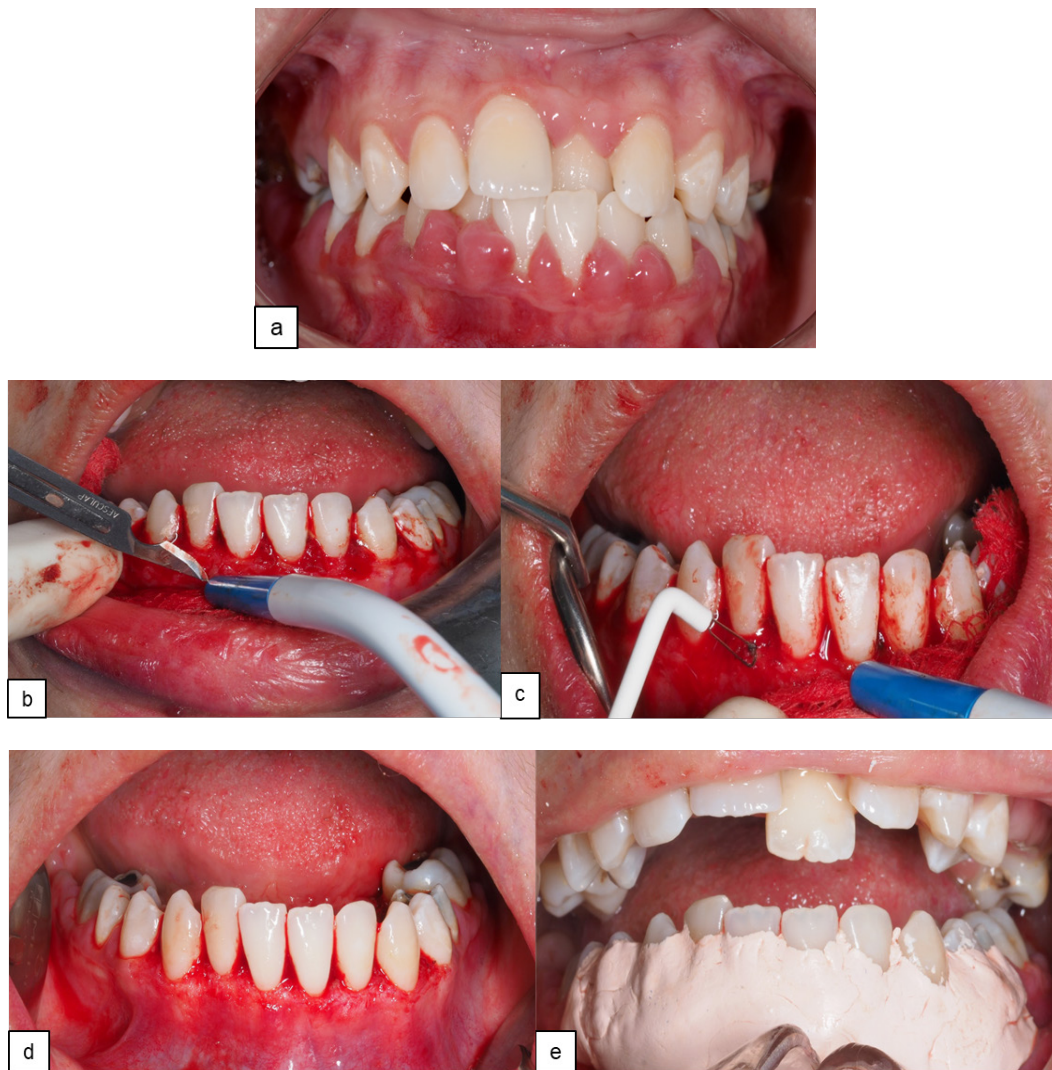


Fig. 5 Gingivectomy procedure using scalpel blade and electrocauterization done under local anaesthesia. (a) Baseline appearance of the gingival enlargement at lower anterior region after non-surgical periodontal therapy prior to surgery. (b) Gingivectomy done by using scalpel blade #15C. (c) Cauterisation of surgical area with electrocauterization to reduce bleeding. (d) Surgical area immediately after gingivectomy. (e) Periodontal dressing was placed on the surgical site.



Fig. 6 The gingiva well healed at one-month after gingivectomy procedure.

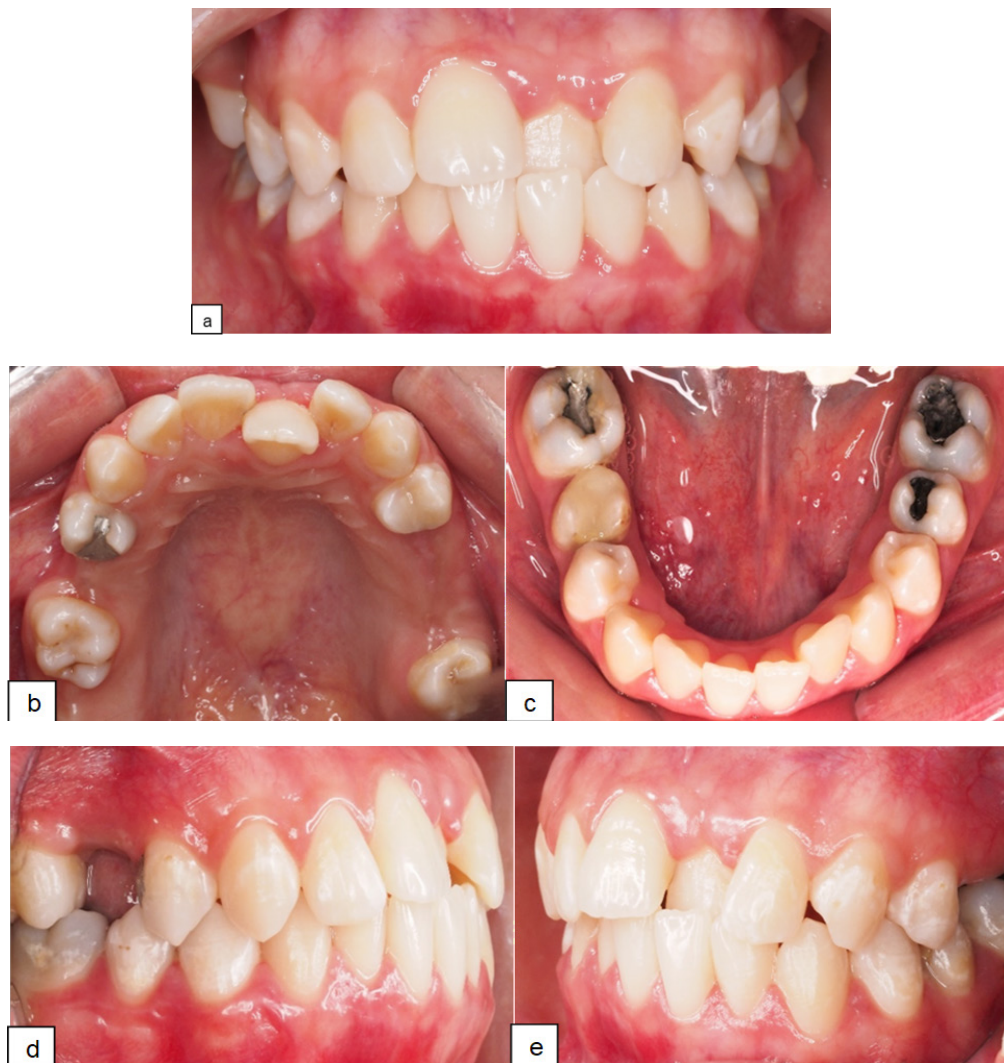


Fig. 7 The healthy gingival appearance of the patient during recall visit at one-year supportive periodontal therapy. (a) Frontal view. (b) Maxillary occlusal view. (c) Mandibular occlusal view. (d) Right buccal view. (e) Left buccal view.

DISCUSSION

MFS is a rare disease which has pleiotropic clinical manifestations and frequently debilitating. Early clinical diagnosis enables better clinical management with a multidisciplinary approach and may prevent possible fatal complications (Pepe *et al.*, 2016; Shafie *et al.*, 2020). Fortunately, this patient has been regularly under follow-up with a medical specialist for her condition, and no complications were detected except scoliosis. She is currently in the queue for surgical treatment. Nevertheless, she cooperates throughout the periodontal

treatment without facing difficulties during procedures on a dental chair. Scoliosis is an abnormal lateral curvature of the spine and is one of the major musculoskeletal manifestations of MFS (Erkula *et al.*, 2002; Tinkle *et al.*, 2023).

This patient is concerned about the appearance of her swollen lower gum associated with bleeding during toothbrushing. Initially, the patient was not aware of her oral health condition. Upon history taking and clinical assessment, she displayed high caries risk, evident by the moderately filled dentition and history of

extraction due to unrestorable caries and retained roots. However, after a proper explanation, she understood and was happy to adhere to the regime and treatment plan to improve the condition. It was reported that the socio-behavioural aspect of a person has been acknowledged as a risk factor for periodontal disease and its progression (Thomson *et al.*, 2012). Thus, instilling motivation and awareness about oral diseases and the contributing factors should be taught during the initial phase of periodontal therapy.

Various oral manifestation related to MFS occur, including severe periodontitis due to abnormal connective tissues resulting in the degradation of the periodontal ligament (Cervino *et al.*, 2020). There is insufficient evidence regarding gingival enlargement as an oral manifestation of MFS. Local risk factors such as plaque and calculus may exacerbate periodontal disease and gingival enlargement as observed in this patient. She has been treated with a few visits of initial periodontal therapy to reduce the inflammatory burden. However, gingival enlargement and deep periodontal pockets at the labial aspect of the lower anterior region did not resolve after scaling and root debridement despite improvement in her oral hygiene. Therefore, gingivectomy procedures were undertaken with a combination of scalpel blade and electrosurgery because of bleeding tendency during or post-surgery that may be relevant to hemostatic deviation in patients with MFS (Kornhuber *et al.*, 2019). Electrosurgery may help clot the blood by cauterisation effect, thus allowing the gingiva to re-contour. Generation of excess heat should be avoided when using this technique, which could lead to thermal damage to the tissues. Laser treatment is another alternative method for gingivectomy procedures which has been shown to produce similar healing capacity (Kumar *et al.*, 2015). The healing was uneventful and resulted in a completely resolved false pocket with a healthy gingival appearance at the surgical area.

Orthodontic therapy is required to correct the crossbite of the upper anterior teeth, which will be carried out after the surgical therapy of scoliosis. Although the final condition of the gingiva in the area is under control, the position of tooth 21 might promote plaque accumulation and become a risk of gingivitis development in future. Meticulous oral hygiene and optimal periodontal conditions will enable the successful outcome of an overall treatment. Orthodontic correction may help improve periodontal health by eliminating plaque retentive factors and facilitating oral hygiene procedures by the patient (Andrade *et al.*, 2014).

SPT commenced when the patient had achieved consistent good plaque control. Local and modifying factors must be monitored to prevent infection recurrence (Trombelli *et al.*, 2020). Based on PRA, a three-month interval for SPT is necessary for first-year recall to maintain her periodontal condition and general health (Lang & Tonetti, 2003). If the patient has an adequate level of plaque control and comparing present with past measurements indicates stability, the interval can be prolonged up to six months by increments of one month from visit to visit (Mombelli, 2019). Besides that, clinical follow-up for MFS is very important to evaluate any disease progression and adverse manifestation for early intervention or therapy.

CONCLUSION

The present case showed that oral manifestation of MFS can be presented as gingival enlargement requiring surgical intervention. Patient awareness and motivation and good treatment regime compliance yield satisfactory periodontal outcomes. Long-term maintenance should be emphasised to maintain good periodontal health, apart from monitoring her systemic conditions for better quality of life.

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