Case Report and Review Literature
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Oral Rehabilitation in Neurofibromatosis Type 1- A Six Year Follow-Up Case Report

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Abstract

Neurofibromatosis is an autosomal dominant disorder that affects the bone, the nervous system, soft-tissue, and the skin. The expressivity of the disease is extremely variable, with manifestations ranging from mild lesions to several complications and functional impairment. The penetrance, otherwise, is 100%. Approximately 72% of Neurofibromatosis Type1 patients present with oral manifestations. These patients have an estimated 3-15% additional risk of malignant disease in their life-time. The following report presents a 30 year old lady diagnosed with Neurofibromatosis Type 1 in childhood who presented to department with complain of bleeding gums. Radiograph showed shallow sigmoid notch on left side, obtuse and thinned out left angle of mandible which are some additional features. A multidisciplinary approach of dental and medical specialties was taken here for oral rehabilitation and to manage and treat periodontal manifestations and other oral complaints. Case was followed for more than 6 years. Neurofibromatosis has no cure hence when such patients report to dental professionals it is up to them to act as primary care giver and instill positive frame of mind to help reduce stressful life events.

Keywords: Multidisciplinary Approach, Neurofibromatosis Type 1, Oral Manifestations, Oral Rehabilitation, Periodontal Manifestations, Shallow Sigmoid Notch

1. Introduction

The term Neurofibromatosis (NF) is used to a group of genetic disorders that primarily affect the cell growth of neural tissues.

1.1 Two Major Subtypes Exist

- Neurofibromatosis type 1(NF1), which is referred to as peripheral neurofibromatosis
- Neurofibromatosis type 2(NF2), which is referred to as central neurofibromatosis
- A third variant known as segmental neurofibromatosis-limited to a single body region.

Regardless of the race, age, sex or ethnic background NF1 is seen on average one in every 2,500-3,300 individuals[1]. NF1, also known as von Recklinghausen's disease, is the most common type of NF and accounts for about 90% of all cases[2]. For establishing a diagnosis of neurofibromatosis two or more of the criteria described by Gutman *et al.*,[3] needs to be fulfilled, which includes:

- Six or more *cafe au lait* macules:
  - Diameter ≥1.5 cm in post-pubertal individuals
  - ≥0.5 cm in pre-pubertal individuals
- Two or more neurofibromas of any type, or one plexiform neurofibroma
- Multiple freckles in the axillary area or groin
- Optic glioma
- Two or more Lisch nodules (pigmented hamartomas of iris)
- A distinct osseous lesion, such as

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  - Sphenoid dysplasia
  - Thinning of the long bone cortex with or without pseudoarthrosis
- A first-degree relative (parent, sibling or offspring) who meets the above criteria for NF1.

We report a case of NF1 with some additional OPG findings, periodontal manifestations its management and a follow up of more than six years. A discussion about NF1 oral manifestations is also presented.

1.2 Case Report

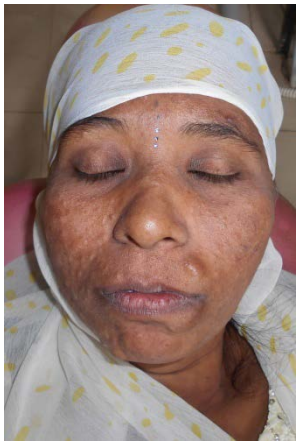
(Figure 1) & (Figure 2) present a 30-year-old lady diagnosed with NF1 in childhood. Patient reported to dept. of Periodontology in year 2014 with complain of loose teeth and bleeding gums since past few years.

Extraoral examination revealed multiple round densities all over the body. Comparatively large nodule like structure was noticeable on upper and lower lip and right side of chin on the face.

Intraoral examination revealed poor oral hygiene, moderate calculus, grade III mobile 11, 22, missing 21 and rotated lower anteriors. A large diffuse lobulated swelling approx. 7×4 cm in size was seen over dorsum of tongue (Figure 3). Gingival examination presents generalised gingival inflammation, enlargement of interproximal, marginal, and attached gingiva, suppuration and bleeding on probing (Figure 4). Periodontal examination revealed generalised pocket along with generalised gingival recession, furcation involvement with 46 & 47 and generalised tooth mobility.

At her first visit patient was reserved and apprehensive about the extraoral and intraoral examinations and the number of visits it would take until completion of treatment. This could be attributed to the fact that young female NF patients are likely to be insecure due to negative life experiences. Family members also reported of patient being fearful, nervous and hesitant to strangers. Hence the above mentioned examinations and the following investigations and treatments were done with utmost care in presence of her family member's to build a positive doctor patient relationship.

Complete blood investigations, Intra-oral Periapical x-ray with respect to maxillary posteriors and mandibular posteriors and Orthopantomogram (OPG) were done. Patient was further referred to Department of Dermatology for examination of cafe au lait spots, Department of ENT for audiometry test



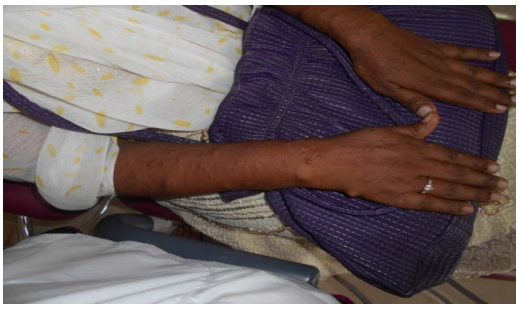


Figure 1. and Figure 2. Showing multiple nodules of varying diameters on face and hands.

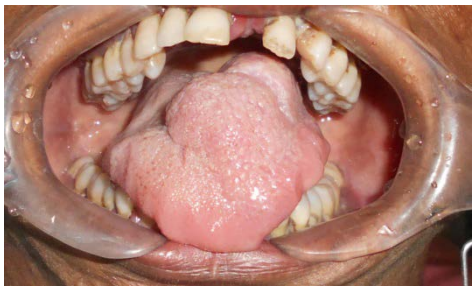


Figure 3. Neurofibroma on tongue.

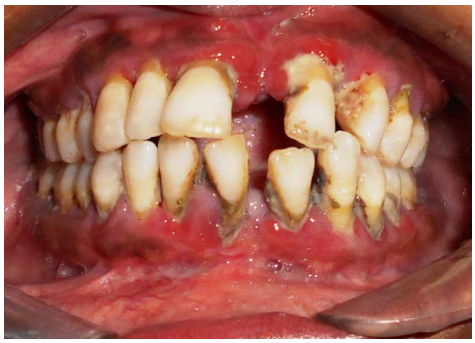


Figure 4. Gingival status.

to detect any possible hearing loss and Department of ophthalmology for eye signs.

In the present case, patient had poor oral hygiene so after supragingival scaling, oral hygiene maintenance instructions were given and scaling was completed in subsequent appointments. 0.2% chlorhexidine mouth wash was prescribed as home care regimen. Before planning periodontal surgical phase all hopeless teeth were extracted and indicated endodontic restorations were completed. Oral hygiene maintenance was evaluated for up to 3 months and then Open Flap Debridement was performed with maxillary and mandibular quadrants, intrasurgical assessment shows grade III furcation involvement with 46 (Figure 5). After 1-month follow-up of surgical phase patient was referred to dept. of prosthodontics for prosthesis with missing teeth and it was decided that a flexible removable partial denture was more suitable according patients clinical condition. The present case was followed up to a time period of six years. After this when she showed self confidence it was discussed with her family and referred positively to institutes psychiatrist for psychotherapeutic interventions.

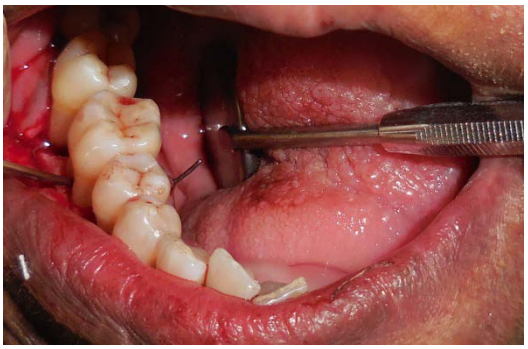


Figure 5. Intrasurgical view of grade III furcation with 46.

2. Discussion

NF1 is considered a dermatologic disorder but intraoral signs of neurofibromatosis occur quite commonly. Periodontitis is enlisted as one of the dental manifestations of NF1[4]. The present case showed classical signs and symptoms of periodontitis which are generalized alveolar bone loss, pathologic tooth migration, mobility and subsequently loss of teeth. Other dental features associated with NF 1 are usually seen on OPG which are discussed below. D'Ambrosio *et al.,* in their study reported buccal mucosa, alveolar ridge, gingiva, lips, palate and floor of mouth as the most commonly involved intraoral sites in NF 1[5]. Marx RE, Stern D in their report on diagnosis of NF 1 reported orofacial bone malformations may involve temporomandibular joint[6].

Shapiro *et al.* and D'Ambrosio et al. reported frequency of oral manifestations in NF1 patients to be 72% and 66% respectively[7,5]. The most commonly reported oral finding of NF1 is enlargement of the fungiform papillae of the tongue that occurs in about 50% of cases. Majority of oral neurofibromatoma occur in the tongue[7,8]. Our patient also presented with enlarged fungiform papilla and neurofibromatoma on tongue.

Oral radiographic findings include enlarged mandibular canal, mandibular foramen and mental foramen, condylar hypoplasia, elongated coronoid process and notching in posterior border of ramus[9,10]. Friedrich RE et al., reported some unique radiographic features in their study such as narrowing and rarefaction of coronoid and articular process and deepening of sigmoid notch[11]. OPG of present case showed enlarged mandibular foramen and mandibular canal predominantly on left side which are similar to other reported cases. However, some atypical unreported radiographic features as spiked root resorption with mesial root of 37 and associated arc shaped bone loss, obtuse mandibular angle, shallow sigmoid notch, resorption of ramus and thinning of angle of mandible were also seen (Figure 6). Patient reported no history of trauma to mandible on either side till date. A follow up OPG was done after six years to evaluate for any further changes (Figure 7).

NF 1 cases associated with Periodontitis can present with gingival thickening, increased gingival fibroblast with extracellular matrix, overgrown attached gingiva with no signs of inflammation and neurofibroma in gum in 5% of cases as per report by Mahajan A *et al.*[12] Doufexi A *et al.*, reported high plaque index leading

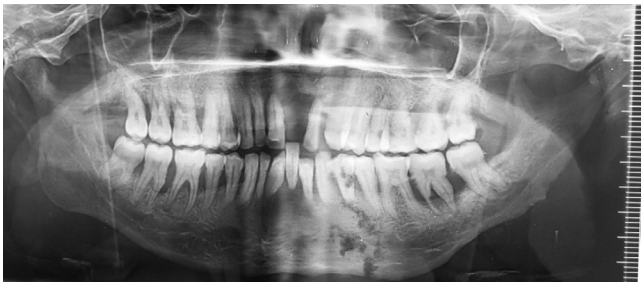


Figure 6. OPG showing obtuse mandibular angle, shallow sigmoid notch, resorption of ramus and thinning of angle of mandible.

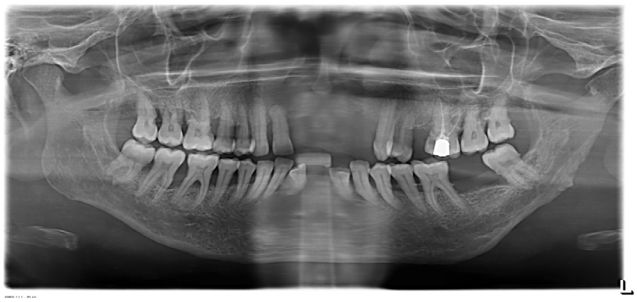


Figure 7. Six year follow up OPG showing no new lesions or progression of disease.

to attachment loss in NF 1 patients when compared to control group[13]. This can be due to brushing difficulties, teeth malpositioning and poor oral hygiene maintenance. Freidrich and Reul showed lowest DMFT index in NF1 group when compared with reference group solely in mandibular nerve region. They reported missing teeth were more frequently registered with NF1 group[14]. In present case localized severe attachment loss and bone loss was seen with mandibular region when compared with maxillary region thus showing a predilection towards mandibular tooth loss. Other physical features that can be associated with NF 1 are macrocephaly, short stature, deafness, kyphoscoliosis. Sometimes developmental and learning disorders are also present[15]. However present case did not present any of these features.

3. Conclusion

Presently there is no cure for NF1 hence it is only humane to understand the mental status of such patients. They show more symptoms of depression and anxiety, high stress levels and low levels of self esteem[16]. They have stressful life events and gloomy life experiences related to health and rejections by others. Even if only one member has the condition, NF affects entire family. Body image is an important link between disease visibility and psychological well being. The present case was approached with multidisciplinary management and followed up for more than six years which helped to reduce the stress levels, increase self esteem and self confidence to face negative life events. The present case is unique in terms of atypical radiographic findings which are not yet reported and also in terms of such long follow up of six years to monitor for any malignant changes which is rarely done. The positive findings of this case are oral rehabilitation was done including periodontal and prosthetic concerns, patient showed immense co-operation for follow up visits, good oral hygiene maintenance for such a long time which is key to success of periodontal procedures, no recurrence of periodontal disease or no new intraoral lesions (Figure 8), follow up OPG shows comparatively equal findings of initial visit. The present case shows even a periodontist can be acting as "key worker" and play a valuable role in managing and effectively supporting people with such complex condition.

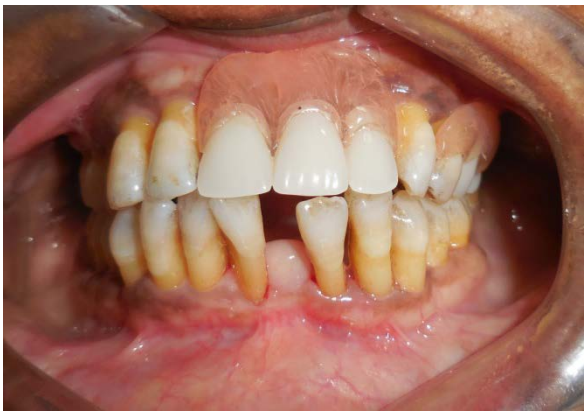


Figure 8. Six year follow up clinical view.

4. References

1. Friedman JM. Neurofibromatosis 1. Clinical Genetics. In: Friedman JM, Gutmann DH, MacCollin M, Riccardi VM, editors. Neurofibromatosis: Phenotype, Natural History and Pathogenesis. 3rd ed. Baltimore: Johns Hopkins University Press; 1999. p. 110–8.
2. Gorlin RJ, Cohen MM, Levin LF. Syndromes of the head and neck. p. 353–416. Oxford: Oxford University Press, 1990.
3. Gutmann DH, Aylsworth A, Carey JC et al. The diagnostic evaluation and multidisciplinary management of neurofibromatosis 1 and neurofibromatosis 2. JAMA 1997; 278: 51–57. https://doi.org/10.1001/jama.1997.03550010065042
4. Eiti Singh, Khushboo Singh, Sunita Gupta, Mayank Shailat. Use of advanced imaging in diagnosis of neurofibromatosis 1. J Oral Maxillofac Radiol 2018; 6: 36–41. https://doi.org/10.4103/jomr.jomr_15_18
5. D'Ambrosio JA, Langlais RP, Young RS. Jaw and skull changes in neurofibromatosis. Oral Surg Oral Med Oral Pathol 1988; 66: 391–396. https://doi.org/10.1016/0030-4220(88)90252-6
6. Marx RE, Stern D. Oral and Maxillofacial Pathology: A Rationale for diagnosis and treatment. 2nd ed. Illinois: Quintessence Publishing Co Inc; 2003. p. 422–7

7. Shapiro SD, Abramovitch K, Van Dis ML et al Neurofibromatosis: oral and radiographic manifestations. Oral Surg Oral Med Oral Pathol 1984; 58: 493–498. https://doi.org/10.1016/0030-4220(84)90350-5
8. Neville BW, Damm DD, Allen CM et al Oral and Maxillofacial Pathology. Philadelphia: WB Saunders Company, 1995, pp. 381–383.
9. Visnapuu V, Peltonen S, Tammisalo T, et al. Radiographic findings in the jaws of patients with Neurofibromatosis 1. J Oral Maxillofac Surg., 2012; 70: 1351–1357. https://doi.org/10.1016/j.joms.2011.06.204
10. Krishnamoorthy B, Singh P, Gundareddy S.N, et al. Notching in the posterior border of the ramus of mandible in a patient with Neurofibromatosis Type I- A case report. J Clin Diagn Res., 2013; 7: 2390–2391. https://doi.org/10.7860/JCDR/2013/5952.3534
11. Friedrich RE, Giese M, Schmelzle R, Mautner VF, Scheuer HA. Jaw malformations plus displacement and numerical aberrations of teeth in neurofibromatosis type 1: a descriptive analysis of 48 patients based on panoramic radiographs and oral findings. J Cranio Maxillofac Surg. 2003; 31: 1–9. https://doi.org/10.1016/S1010-5182(02)00160-9
12. Mahajan A,Dixit J and Bharadwaj A. Gingival enlargement in Neurofibromatosis Type I: A Case report and literature review. J. Contemp. Dent. Pract., 2010; 11: 57–63. https://doi.org/10.5005/jcdp-11-2-57
13. Doufexi A, Mina M and Ionnidou E. Gingival overgrowth in children: Epidemiology, Pathogenesis and Complications. A literarture review. J Periodontol, 2005; 76: 3–10. https://doi.org/10.1902/jop.2005.76.1.3
14. Reinhard E. Freidrich and Anika Reul. Decayed, missing and restored teeth in patients with Nuerofibromatosis Type I, J Clin Exp Dent, Feb 2018; 10(2): e107–115.
15. Riccardi VM, Eichner JE. Neurofi bromatosis: Phenotype, Natural History, and Pathogenesis. 1sted. Baltimore: John Hopkins University Press; 1986, p. 37–55.
16. Daphne L. Wang, Kelly B. Smith, Sonia Esparza. *et al.* Emotional functioning of patients with neurofibromatosis tumour suppressing syndrome. Genet Med. Dec 2012; 14 (12): 977–982. https://doi.org/10.1038/gim.2012.85

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