Neurofibroma (NF) is a rare benign non-odontogenic tumor of the oral cavity. NF may present either as solitary lesions or as part of the generalized syndrome of NF or von Recklinghausen's disease of the skin. The heterogeneous nature of NF was established by Riccardi et al. and he recognized at least seven types of NF. Among these proposed classifications of the disease, two distinct forms are generally accepted namely, a peripheral form known as NF Type I (NF-I), and a central form known as NF-II. NF-I represents the classic form of this disease, described by Recklinghausen in 1882. Clinically, oral NF usually appears as slow growing, painless, pedunculated or sessile nodules. For illustration, a case of a NF with oral findings is been presented.

Keywords: Neurofibromatosis Type-1, Neurofibroma, Mucocutaneous, Oral manifestations

spots on the iris. Oral manifestations may occur in about 72-92% of all cases, and commonly affected sites are the tongue and buccal mucosa. Oral radiographic findings include an enlarged mandibular canal, mandibular foramen, and mental foramen. If NF-I cases develop intraosseously, it can result in well demarcated unilocular, but occasionally multi-locular, radiolucent lesions. In this paper, we report a case of NF-I with significant oral manifestations.

INTRODUCTION

The term neurofibromatosis (NF) was first described by Von Recklinghausen and Festscher refers to a group of genetic disorders that primarily affect the cell growth of neural tissues. Among the eight different forms of NF recognized the most common is the NF Type 1 also known as Von Recklinghausen’s disease (NF-I). NF-I accounts for about 90% of all cases as reported by Cunha et al. It is usually transmitted as an autosomal dominant trait and mutations usually occur in the long arm of chromosome number 17 (17q11.2). In the disease process, due to the defect in the embryonic neural crest cells ectodermal and mesodermal derivatives are usually affected. 50% of NF-I patients presents with positive family history of the disease while the rest 50% represent spontaneous mutations.

The most common skin manifestations are multiple NF that can occur anywhere in the body. Other characteristic features include the presence of café au lait spots (coffee with milk) pigmentation, Lisch nodules (pigmented hamartomas of the Iris), and translucent brown pigmented

CASE REPORT

A 45-year-old male patient (Figure 1) reported to the Department of Oral Medicine and Radiology with the chief complaint of deposits on teeth. Deposits on teeth noticed since last few months, and the patient has bleeding during brushing. The past dental and medical history was non-contributory, but the family history of the patient reveals that his grandfather had a history of multiple nodular swellings on skin and face. On general examination, the patient was of moderate built and nourishment and all vital signs were within the normal limits. Extraoral examination revealed the presence of multiple nodules on the face, trunk, back, upper (Figures 1 and 2), and lower extremities. These were round to oval in shape of size varying from millimeters to centimeters, with a smooth surface and skin over and surrounding nodules appeared normal. Intraoral examination of the patient shows two nodular swellings on the posterior aspect of the tongue (Figure 3). On palpation, these were sessile, soft to firm and non-tender, and not fixed to underlying tissues. A provisional diagnosis of generalized gingivitis with NF-I

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was given. Panoramic radiographic examination revealed the presence of widened inferior alveolar canal with enlarged right mental foramina reaching a size of 1.5 cm × 0.5 cm and the right mandibular foramen attaining a size of 3 cm × 4 cm which is typical of NF-I (Figure 4). On the basis of the family history, clinical, and radiographic examinations, the case was diagnosed as NF-I.

DISCUSSION

NF-I is a multi-systemic disorder with a protean expression of cutaneous, neurologic, skeletal, visceral, and ocular manifestations. The mutations of NF-1 gene, which is a large complex, gives rise to diverse manifestations in terms of gene organization and expression.

Despite the advances of molecular biology, the diagnoses of NF-I and NF-II are still based on clinical criteria. The National Institutes of Health Consensus Development Conference has suggested clinical criteria diagnostic of NF-I and NF-II.

Diagnostic criteria of NF-I:

1. Six or more café au lait macules over 5 mm in greatest diameter in pre-pubertal individuals and over 15 mm in greatest diameter in post-pubertal individuals
2. Two or more NF of any type or one plexiform NF
3. Freckling in the axillary or inguinal regions (Crowe’s sign)
4. Optic glioma
5. Two or more Lisch nodules (iris hamartomas)
6. A distinctive osseous lesion such as sphenoid dysplasia or thinning of long bone cortex with or without pseudoarthrosis
7. A first-degree relative (parent, sibling, or offspring) with NF-I by the above criteria.

If two or more of the features listed are present, then the case can be diagnosed as NF-I. In our case, the patient had multiple nodular swellings in body and the patient was a first degree relative of NF-I patient.

NF-I mostly appears in the third decade of life without any sex predilection and association with any immunocompromising diseases. NF-I can present with both extraoral and intraoral manifestations with the prevalence of oral soft tissue manifestation as reported by

Figure 1: Extraoral examination

Figure 2: Lesions on hand

Figure 3: Intraoral examination

Figure 4: Orthopantamograph
The presence of impacted, enlarged mandibular canal, as well as mental foramen, canal, mandibular foramen, and mental foramen. An Oral radio migration of teeth. occur within the periodontal membrane and lead to the they are superficial, it appears fissured (Scrotal). NF may of deep NF the tongue appears enlarged (Macroglossia); if
in our case where two nodular lesions were present on the eighth, ninth, and tenth nerve. A plexiform NF is a peripheral nerve sheath tumor that may involve multiple nerve branches or can extend for some distance along a nerve, this type of NF are a major source of morbidity associated with NF-I mainly due to their tendency disfigurement by growing to large sizes. The cranial nerves most involved in plexiform NF are the fifth, ninth, and tenth nerve. Oral manifestations of NF-I include discrete, non-ulcerated nodules, varying from normal mucosal color to red or even yellow, seen in 2% or possibly as many as 7% of cases. These nodular lesions usually appear in soft tissues such as the cheek, palate, tongue, floor of the mouth, and lips. The most common oral finding of NF-I reported in the literature is enlargement of the fungiform papillae of the tongue that occurs in about 50% of cases. This was consistent with our case where two nodular lesions were present on the tongue. In tongue it can present in two ways, i.e. in case of deep NF the tongue appears enlarged (Macroglossia); if they are superficial, it appears fissured (Scrotal). NF may occur within the periodontal membrane and lead to the migration of teeth.

Oral radiographic findings include an enlarged mandibular canal, mandibular foramen, and mental foramen. An enlarged mandibular canal, as well as mental foramen, was present in our case. NF has also been noted in the intraosseous tissues within the jaw as well they appear as radiolucencies in radiographic examination and may present. A diagnostic problem. The presence of impacted, displaced or missing teeth particularly in the mandible is also a recognized oral manifestation. Abnormalities in the cranial and jaw bones in the form of macrocephaly are frequently identified in patients with NF. NF-I can affect sphenoid bone causing sphenoid dysphasia, including absence of the wings, enlargement of sella turcica, and resorption of the anterior clinooid processes. As a result of soft tissue tumor growing against or within the bone, osseous alterations can occur resulting in hypoplasia or resorption. It has also been proposed that bone growth may by stimulated by the adjacent neural tumor leading to hypoplasia.\textsuperscript{18}

CONCLUSION

From the above case, it is evident that NF-I presents with significant oral manifestations, hence a dentist may be a pioneer in the diagnosis of this condition. Although cases of oral NF are well documented, it never comes forefront in mind while dealing with swellings of the oral cavity. Hence, thorough examination and trained eye are required to utilize the opportunity to diagnose and categorize NF cases. The timely intervention by oral diagnosticians can act as the key to prevent complications arising from NF. In order to achieve this, a life-long evaluation of the patients for newly forming or pre-existing lesions is important. Hence, for all swellings in the oral cavity a thorough medical history coupled with an equally thorough medical examination must be undertaken which will enable clinicians to arrive at a proper diagnosis of the condition.

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