Available online at http://www.journalcra.com

International Journal of Current Research
Vol. 10, Issue, 08, pp.72734-72737, August, 2018

INTERNATIONAL JOURNAL OF CURRENT RESEARCH

ISSN: 0975-833X

RREVIEW ARTICLE

NEUROFIBROMATOSIS I WITH FACIAL PLEXIFORM NEUROFIBROMA AND MANDIBULAR HYPOPLASIA. A CASE REPORT WITH REVIEW OF LITERATURE

[1]Dr. Preeti Patil, [2]Dr. Venkatesh G. Naikmasur, [2]Dr Kirty Nandimath and [3]Dr. Karthik Kabbur

[1]Senior Lecturer, Department of Oral Medicine and Radiology Dayananda Sagar College of Dental Sciences, Bangalore Karnataka, India
[2]Professor, Department of Oral Medicine and Radiology Shri Dharmasthala Manjunatheshwara Dental College, Dharwad-580009, Karnataka, India
[3]Professor, Department of Orthodontics and Dentofacial Orthopaedics, Dayananda Sagar College of Dental Sciences, Bangalore Karnataka, India

ARTICLE INFO

Article History:
Received 08th May, 2018
Received in revised form
14th June, 2018
Accepted 10th July, 2018
Published online 30th August, 2018

Key Words:
Neurofibromatosis type1,
Plexiform neurofibroma,
Mandibular hypoplasia,
Café au lait spots.

ABSTRACT

Neurofibromatosis type 1 (NF1, or Von Recklinghausen's disease) is a genetic disorder transmitted as an autosomal dominant trait with variable expressivities and complete penetrance. It is characterized by a variety of clinical manifestations, including multiple neurofibromas that are associated with a high risk of sarcomatous transformation. This article reports a case of facial plexiform neurofibromatosis with mandibular hypoplasia and reviews various oral soft tissue and hard tissue manifestations of neurofibromatosis I.

Citation: ***Dr. Preeti Patil, Dr. Venkatesh G. Naikmasur, Dr Kirty Nandimath and Dr. Karthik Kabbur*****, 2018.** "Neurofibromatosis I with facial plexiform neurofibroma and mandibular hypoplasia. A case report with Review of Literature", *International Journal of Current Research*, 10, (08), 72734-72737.

INTRODUCTION

Neurofibromatosis type 1 (NF1) is a clinically and genetically distinct disease involving both neuroectodermal and mesenchymal derivatives. Head and neck are a common location for NF; however other sites may be involved less frequently including nasal cavity, paranasal sinus, nasopharynx, orbit, larynx, maxilla, and mandible (Shapiro, 1984). The frequency of oral manifestations is debated in the literature. Some authors report a frequency of 4-7% of cases (Geist et al., 1992), whereas others suggest that these manifestations are present in up to 72% of cases (Shapiro, 1984). Facial plexiform neurofibromas are not common and may cause asymmetry, disfigurement and usually arise from the trigeminal nerve.

Corresponding author:* **Dr. Preeti Patil,
Senior Lecturer, Department of Oral Medicine and Radiology Dayananda Sagar College of Dental Sciences, Bangalore Karnataka, India
DOI: https://doi.org/10.24941/ijcr.31848.08.2018

Intraorally, Neurofibromatosis may manifest in the form of soft tissue neurofibromas, developmental defects, or dental abnormalities. In this paper we present a case of neurofibromatosis I with facial plexiform neurofibromas and mandibular hypoplasia. We also review soft tissue and hard tissue manifestations of NF1 in the oral cavity.

Case Report

A 20year old female patient reported to the department with complaint of pain in her lower right back tooth since a week. Pain was sudden in onset, intermittent, moderate throbbing type and localized. Medical history revealed she had been diagnosed with neurofibromatosis1 8 years back and patient did not undergo investigations or treatment for the condition. Family history revealed that father and brother also suffered from the same condition. On general physical examination patient was moderately built and nourished and had normal gait. There were multiple sessile, round to oval nodular lesions on hands

(Figure 1) and trunk. On extra oral examination, multiple soft, non-tender sessile nodular swelling were seen on face ranging from 0.5cm to 2cm in size. Overhanging folds of soft tissue nodular growths were seen on left lower 1/3rd offace, neck and shoulder extending up to the chest region causing gross disfigurement of face and neck suggestive of plexiform neurofibroma (Figure 2).

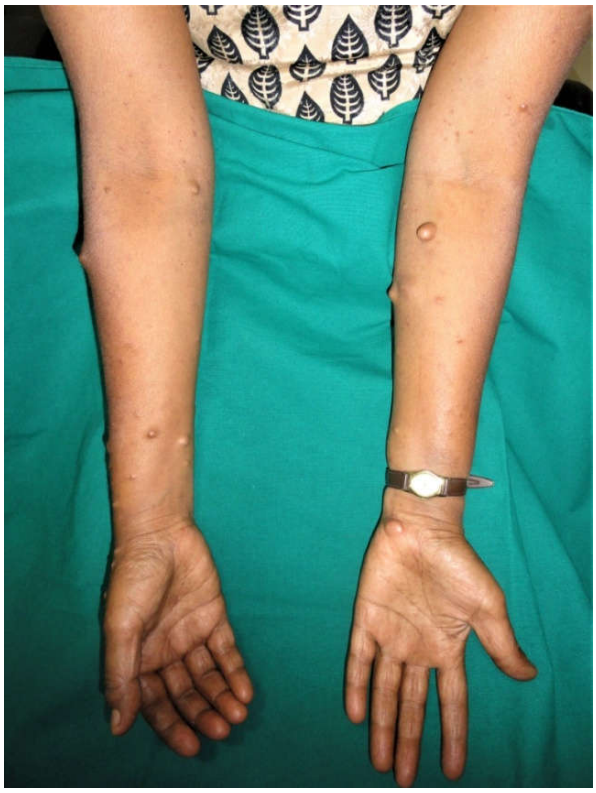

Figure 1. Nodular lesions on arms

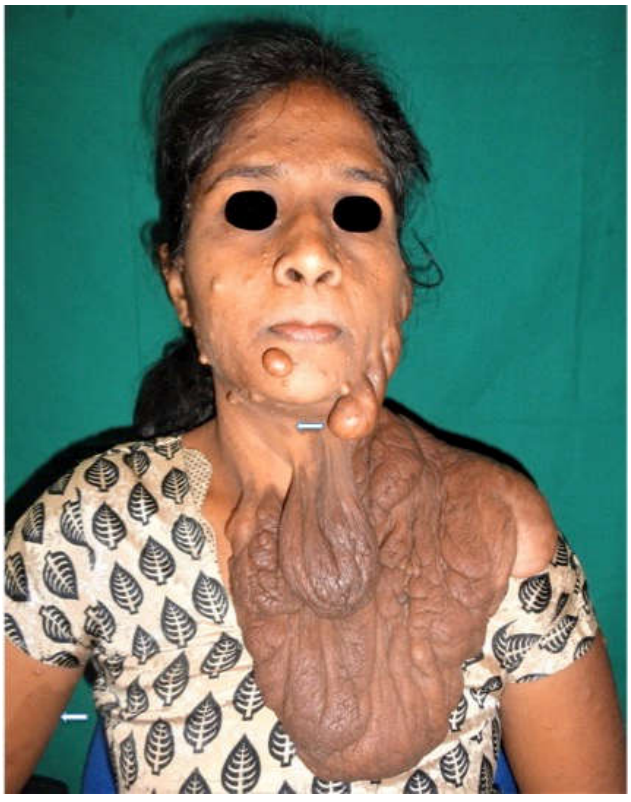

Figure 2. Folds of soft nodular growths causing gross disfigurement

Symptoms of paresthesia or numbness were not present. There was deformity of the left ear (Figure 3). Café au lait spots were seen on neck and forearm (Figure 2, arrow mark).

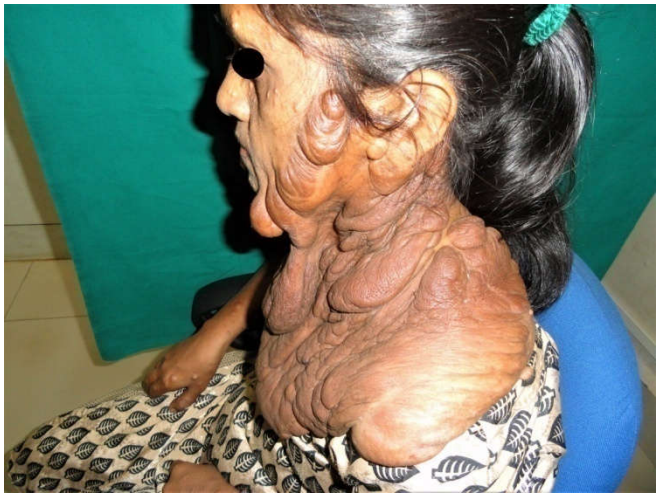

Figure 3. Deformity of left ear

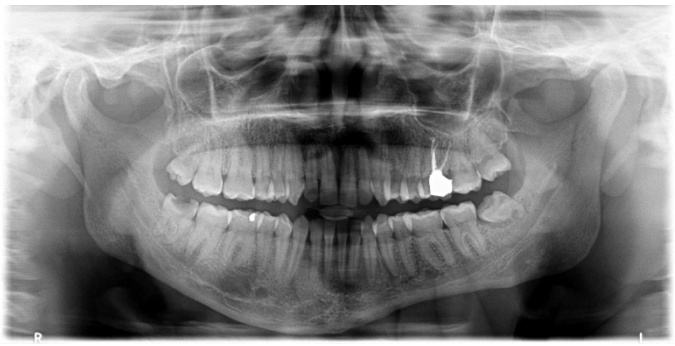

Figure 4. Mandibular hypoplasia on left side, enlarged mandibular canal and mental foramen

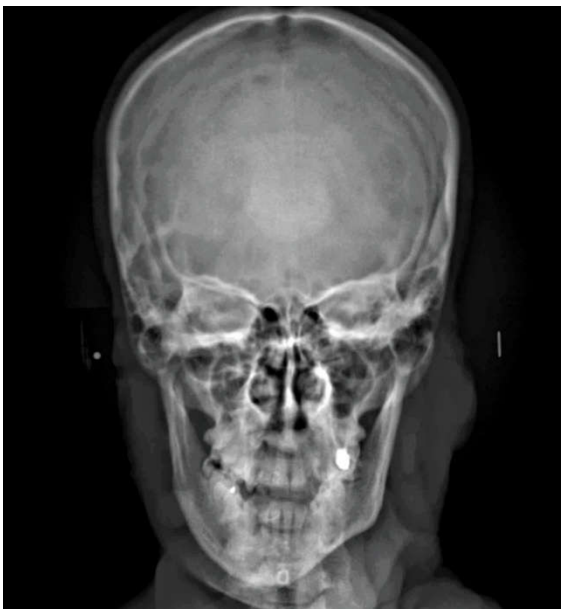

Figure 5. Mandibular hypoplasia on left side

Intraorally, there was deep dentinal caries with 48which was tender on percussion and 38 was partially erupted. No other abnormality was noted. Provisional diagnosis of apical periodontitis with 48 was given. OPG revealed chronic periapical abscess with 48.

There was hypoplasia of left side of the mandible, with reduction in the height of the mandibular symphysis, parasypmhysis and body of mandible (Figure 4). There was widening of the inferior alveolar nerve canal and enlargement of the mental foramen on left side. There was impacted 38 with malformation of roots. PA mandible was advised to see the asymmetry in the mandible that showed hypoplasia of left mandible giving scooped out appearance (Figure 5). OPG of patient's father also was taken which revealed marked widening of mental foramen and mandibular canal (Figure 6). Patient was advised extraction with 48. Patient was referred to neurosurgeon for further investigation and needful treatment for the cutaneous lesions.

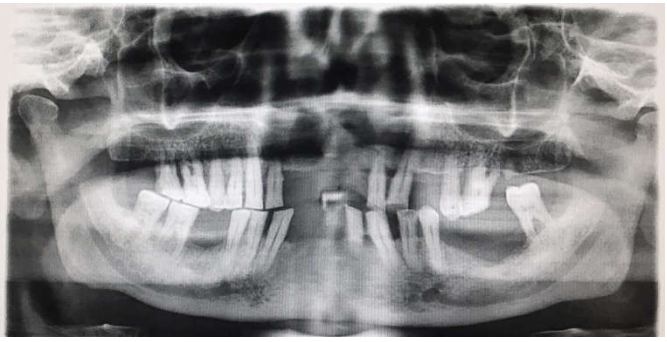

Figure 6. Widening of mandibular canal and mental foramen

DISCUSSION

Neurofibromatosis, as identified by Von Recklinghausen, is an autosomal dominant neurocutaneous syndrome, characterized by multiple neurofibromas, cafe-au-lait spots, and iris Lisch nodules (D'Ambrosio, 1988), Other features which can be associated with Von Recklinghausen's neurofibromatosis are macrocephaly, short stature, seizures, hypertension, deafness, constipation, kyphoscoliosis, developmental and learning disorders, cosmetic disfigurement and neurofibrosarcomas, among others (Riccardi et al., 1986). Plexiform neurofibroma is a non-circumscribed, thick, and irregular benign tumor of the peripheral nerve sheath. It is a virtually pathognomonic and often disabling feature of neurofibromatosis type I. Oral manifestations of neurofibroma may occur in about 72-92% of all cases, and commonly affected sites are the tongue and buccal mucosa (Gómez-Oliveira, 2004).

Soft tissue manifestations

Oral soft tissue 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 (Gutteridge, 1991). Neurofibromas of the oral soft tissue have been reported in the literature; the tongue, being the mostcommon site, usually is macroglossic. Other areas of oral soft tissue involvement include the buccal mucosa, the alveolar ridge, gingiva, lips, the palate, the floor of the mouth, and the pharyngomaxillary space (Arendt, 1987). Localized oral neurofibromas usually appear as asymptomatic nodules covered by normally colored mucosa (Cunha, 2004 and Bekisz, 2000). However, when they are adjacent to the cranial nerves, they can impair motor function of the facial or hypoglossal nerves or the sensitivity of the trigeminal nerve (Shapiro, 1984). Shapiro et al. stated that gingival involvement is 5% (Shapiro, 1984). K S G Cunha reported acase of NF1 patient with a gingival neurofibroma in the attached gingiva of the lingual aspect of the lower central incisors. It presented as a lesion was nodular, with sessile base, non-ulcerated, non-painfulgrowth (Cunha, 2004). Vandana Shekaret al report a rare case of gingival neurofibroma in maxillary posterior region in NF1 patient with no radiographic changes (VandanaShekar, 2015). In the literature the most common oral finding of NF-I reported is enlargement of the fungiform papillae of the tongue that occurs in about 50% of cases (Cherrick, 1971), Shapiro SDet al reported 24 patients with neurofibromatosis in which he noted enlargement of the fungiformpapillae in seven patients (31.89%) and oral soft tissueneurofibromas in six patients (27.2%) (Shapiro, 1984). In the present case there were no intraoral manifestation of NF1.

Hard tissue manifestation

Radiographic findings of NF1in the jaws include an enlarged mandibular canal, mandibular foramen, mental foramen and hypoplasia areas of the orofacial complex. Intraosseously, it can result in well demarcated unilocular, but occasionally multi-locular, radiolucent lesions. A radiographic study by D' Ambrosio and coworkers of 38 patients with neurofibromatosis revealed that 4 had intraosseous lesions (D'Ambrosio, 1988). Shapiro and co-workers found from radiographs that four of 24 patients had intrabonylesions (Shapiro, 1984). Hypoplastic areas of the orofacial complex have been shown inpatients with neurofibromatosis (D'Ambrosio, 1988). Areas noted to be hypoplastic include the maxillae, the zygomatic bone, the temporomandibular joint, and the ramus of themandible. Shortening of the ramus, notching of the in ferior border of the mandible, and enlarged lingual openings are other hard tissue findings reported in the literature. Intracranial nerves also can be involved in NF-I. Cranial nerve VII and IX involvement results in decreased (taste and gag reflexes). If cranial nerves V or VII are affected, the tongue can deviate to one side and the patient may experience altered sensation (Arendt, 1987). Sigillo et al. reported the orofacial manifestations of NF1 observed in 6 pediatric patients (between 4 and 15 years of age) where three patients had intrabony lesions where one had maxillary involvement extending to floor of orbit and rest two had mandibular hypoplasia on affected side (Sigillo, 2002). Raghavendra Kini et al reported a case of NF1in a 10 year old girl who had hypoplasia of mandibular condyle and ramus (Raghavendra Kini, 2012). Gloria Molinsetal reported acase of 18 years old teenager with neurofibromatosis type 1 with right hemifacial hypoplasiaand occlusion of the left common carotidartery from its origin (Gloria Molins, 2017).

Mubeen Khan et al reported neurofibromatosis in family where three siblings had variety of oral manifestations. One of the siblings had oral neurofibroma of tongue and diffuse melanin pigmentation of tongue and elder sibling had neurofibroma involving anterior mandible manifesting as a radiolucency. Also there was widening of mandibular canals, enlarged mental foramen, deepening of sigmoid notch and shortening of ramus (Mubeen Khan, 2011). The present case also had radiographic manifestations like hypoplasia of mandible, enlargement of inferior alveolar nerve canal and mental foramen. C. Cervellera et al reported a case of neurofibromatosis 1with four impacted third molars and 6 impacted super neumararyteeth (Charline, 2017). In the present case there was an impacted 38 with malformation of root. E. Bardellin reported Oral findings in 50 children with neurofibromatosis type 1 where outof 50 children, 14 (28%) patients showed radiographic features of NF.

Eighteen children (36%) showed increase in dimension of the coronoid notch and deformity of the condylar head; increase in bone density and enlarged mandible foramen were observed in 6 children (18%); in 2 cases (4%) there was a decreased mandibular angle. No intraosseous neurofibromas were detected (Bardellini, 2011).

Conclusion

Oral manifestations of NF1 are common and may be seen in soft tissues or in the jaws. Facial plexiform neurofibroma is one of the manifestations of NF1 and has disfiguring nature that can cause physical as well as psychological morbidity. Early diagnosis and intervention is important as they have tendency for malignant transformation. Since it is a genetic condition, genetic counseling of patients and families are highly recommended.

REFERENCES

Arendt DM, Schaberg SJ, and Meadow Js T 1987. Multiple radiolucent areas of the jaw. *J Am Dent Assoc*., 115:597-99, 1987.

Bardellini, E., Amadori, F. P. Flocchini, G. Conti, G. Piana, A. Majorana. 2011. Oral findings in 50 children with neurofibromatosis type 1. A case control study. European *Journal of Paediatric Dentistry.,* 12(4):256-60

Bekisz O, Darimont F, Rompen EH. 2000. Diffuse but unilateral gingival enlargement associated with von Recklinghausen neurofibromatosis: A case report. *J ClinPeriodontol*., 27:361–5.4

Charline Cervellera, David Del Pin, Caroline Dissaux. 2017. Neurofibromatosis type I and supernumerary teeth: up-to date review and case report. *Med Buccale Chir Buccale* 23:84-8

Cherrick, H.M., Eversole, L.R. 1971. Benign neural sheath neoplasm of the oral cavity. Report of thirty-seven cases. *Oral Surg Oral Med Oral Pathol*, 32:900-9

Cunha, K.S., Barboza, E.P., Dias, E.P., Oliveira, F.M. 2004. Neurofibromatosis type I with periodontal manifestation. A case report and literature review. *Br Dent J.*; 196:457–60.

D'Ambrosio, J.A., Landlais, R.P, Young, R. S: 1988. Jaw and skull changes in neurofibromatosis. *Oral Surg.,* 66: 391-96.

Geist JR, Gander DL, Stefanac SJ. 1992. Oral manifestations of neurofibromatosis types I and II. *Oral Surg Oral Med Oral Pathol.*, 1992; 73:376-382

Gloria Molins, AdaiaValls, Lorena Silva, JordiBlasco, Federico Hernández-Alfaro. Neurofibromatosis type 1 and right mandibular hypoplasia: unusual diagnosis of occlusion of the left common carotid artery. *Journal of Clinical Anesthesia* 42 (2017) 98–99

Gómez-Oliveira G, Fernández-Alba Luengo J, Martín-Sastre R, Patiño-Seijas B, López-Cedrún-Cembranos JL. Plexiformneurofibroma of the cheek mucosa. *A case report. Med Oral*., 2004;9:263-7

Gutteridge DL. 1991. Neurofibromatosis: an unusual oral manifestation. *Br Dent J.,* 170:303-4.

Mubeen Khan, NeeraOhri. Oral manifestations of Type INeurofibromatosis in a family. *J ClinExp Dent.*, 3(5): e483-6.

Raghavendra Kini, Vathsala Naik, Ashwini Baliga, Anjali P Shetty, Y R Girish. 2012. Neurofibromatosis type 1 with unusual oral manifestations., 24(3); 226-229.

Riccardi, V.M., Eichner, J.E. 1986. Neurofibromatosis: Phenotype, Natural History, and Pathogenesis, 1st ed. Baltimore, MD: The John Hopkins University Press, 37-55

Shapiro, S. D., K. Abramovitch, M. L. Van Dis et al., 1984. "Neurofibromatosis: oral and radiographic manifestations," *Oral Surgery, Oral Medicine, Oral Pathology,* 58(4): 493–8.

Sigillo, R., H. Rivera, MS N.G. Nikitakis, J.J. Sauk, MS. 2002. Neurofibromatosis type 1: a clinicopathological studyof the orofacial manifestations in 6 pediatric patients: Pediatric Dentistry., 24(6):575-580

VandanaShekar, Vishwanath Rangdhol, W. John Baliah, and Sivasankari Thirunavukarasu. 2015. An unusual oral manifestation of type 1 neurofibromatosis: A case report and review of literature. *J Nat Sci Bio Med.,* Jan-Jun; 6(1): 261–263.

****