*ejpmr, 2020,7(11), 600-603*

SJIF Impact Factor 6.222

EUROPEAN JOURNAL OF PHARMACEUTICAL AND MEDICAL RESEARCH

www.ejpmr.com

Case Study

ISSN 2394-3211

EJPMR

# NEUROFIBROMATOSIS – A RARE ENTITY

**Farheen Jahan[1*], Rashmi Sapkal[2] and Vinod V. C.[3]**

[1]Post Graduate Student, Department of Oral Medicine and Radiology, M.A. Rangoonwala College of Dental Sciences and Research Centre, Pune.

[2]Reader, Department of Oral Medicine and Radiology, M.A. Rangoonwala College of Dental Sciences and Research Centre, Pune.

[3]Professor and Head Department of Oral Medicine and Radiology, M.A. Rangoonwala College of Dental Sciences and Research Centre, Pune.

***Corresponding Author: Dr. Farheen Jahan**
Post Graduate Student, Department of Oral Medicine and Radiology, M.A. Rangoonwala College of Dental Sciences and Research Centre, Pune.

**Article Received on 06/09/2020** **Article Revised on 26/09/2020** **Article Accepted on 16/10/2020**

**ABSTRACT**

In 1882, Von Recklinghausen was first described Neurofibromatosis as a genetic disease. Neurofibromatosis comes under neuroectodermal abnormality that shows systemic manifestations that mainly affect nervous system, skin, bones, eyes and many other systemic organs. According to literature, Neurofibromatosis features vary from individual to individual. This paper reported a case of 40 year old female with the chief complaint of pain in upper right back region of the jaw since 4 days. On detailed medical history, family history and clinical examinations were confirmed the diagnosis of neurofibromatosis type I.

**KEYWORDS**: café-au-lait patches, Lisch nodules, Neurofibromatosis.

## INTRODUCTION

In 1882 by Friedrich Daniel Von Recklinghausen was first descirbed a genetic disease called neurofibromatosis (NF). It is a neuroectodermal abnormility that found clinical and systemic symptoms of set forth. Neurofibromatosis features vary from individual to individual, one and the same family. Neurofibromatosis mainly affects nervous system, skin, bones, eyes and many other systemic organs.[1,5]

## CASE REPORT

A female age 40 year reported to the Department of Oral Medicine and Radiology, with the chief complaint of pain in upper right back region of the jaw since 4 days. Patient was apparently asymptomatic 4 days back, when she experienced pain in right upper back region of jaw. Patient gives history of removal of her lower teeth of right and left side, she complains of mild pain. Pain was dull aching and continuous in nature with intense since a day. She took medication for relief of pain, and contents she is not aware off. She also gives history of multiple growths on her body since childhood and its increase in size and number since then. Past medical history: She has history of multiple growths on her body since childhood, and history of removal of one painful growth on her chest 5years ago without any history of complications. She lost her files & documents related to surgery. When history goes towards the dental and personal history that revealed, patient has undergone removal of her lower 4 teeth since 2months. Oral hygiene and personal history were not contributory with diagnosis. Habit history: Patient has habit of tobacco application 2 to 3times a day, since 20 years, and finally family history that was most important for this case revealed patient mother and aunt had same spots and growth on their skin.

### General, Extraoral examination

General examination revealed a moderately built and nourished with steady gait and satisfactory vital signs. There were no signs of icterus, clubbing. Both ears were normal on auditory evaluation. There was no evidence of neurologic symptoms.

Facial symmetry: Symmetrical (Fig.1)Growth noted on face, both side of chin and jaws (inferior labial surface), on forehead right and left side, preauricular area of left side of face. The patient reported that the face growth started at 8 years of age and continuously increased in number since then. (Fig 2, 3, 4) Neck examination revealed single large and multiple diffuse and soft growths in the neck region with ill defined edges. Multiple small nodules in the right side of neck with ill defined edges with no redness, tenderness. There was no evidence of palpable cervical lymph nodes. There was evidence of single large diffuse growth that was tender on examination. A bulging mass on the posterior aspect of her scalp. The mass was first observed at the age of 20 years, and it had slowly increased in size since then.

Physical examination revealed a soft 4x3cm skin colored mass with mild tenderness on the occipital scalp. (Fig 5)

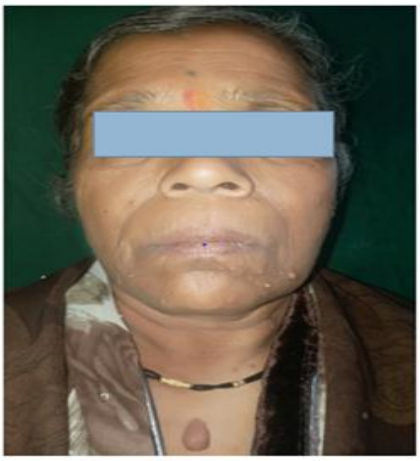

Fig 1: facial profile

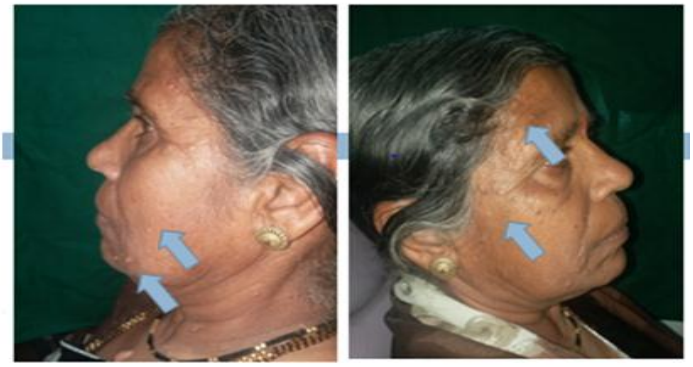

Fig 2, 3: Right & Left Lateral profile

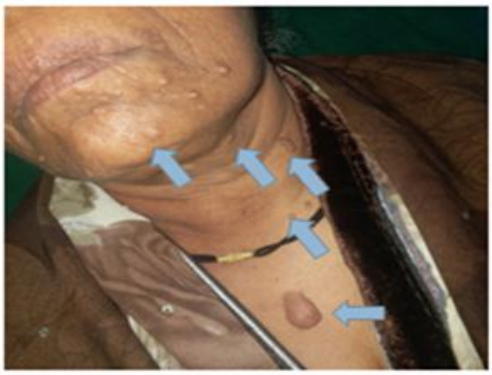

Fig 4: Neck examination

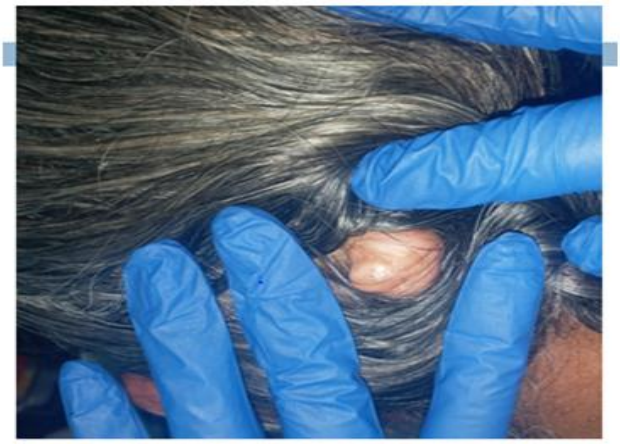

Fig 5: Examination of posterior aspect of scalp

Fig 6, 7: Lisch nodules

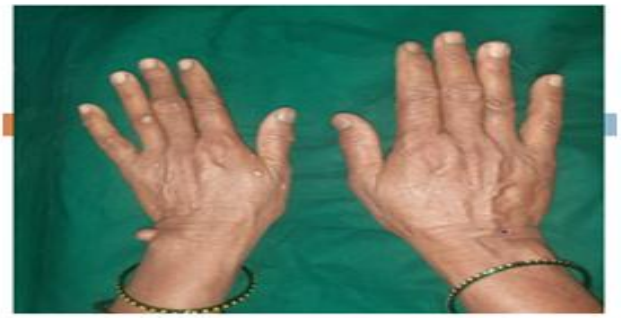

Fig 8 : Neurofibromas

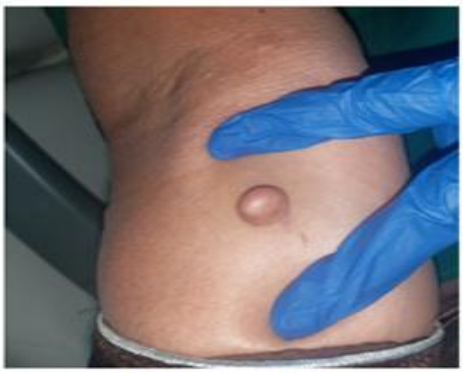

Fig 9: Well-circumscribed Neurofibromas

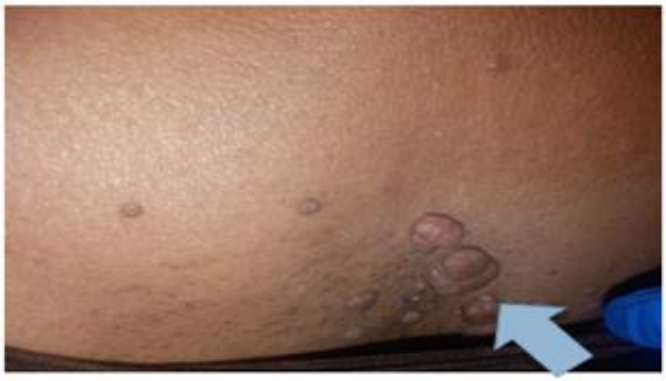

**Fig 10: Examination of back**

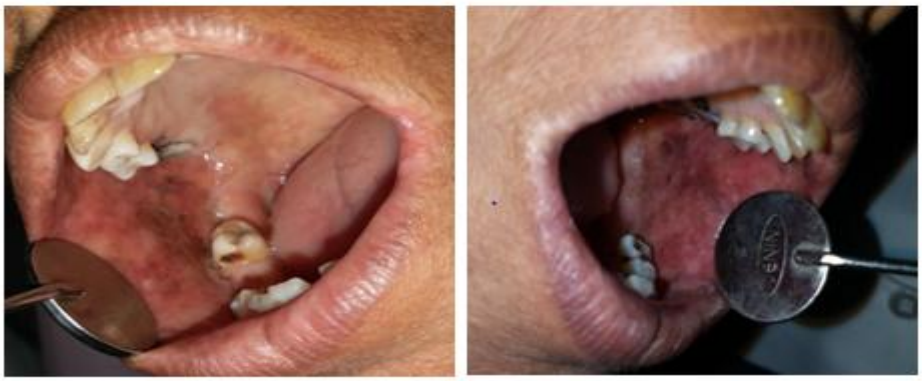

**Fig 11, 12: Intraoral examinations**

Lisch nodules: She has multiple reddish brown spots in the lower pole of the iris that are slightly raised proliferations of melanocytes and fibroblasts. (Fig 6, 7) She has at least 6 raised, soft, well-circumscribed, pedunculated Neurofibromas. The patient also has a prominent rounded macule, which is a café-au-lait spot. (Fig 8, 9)

There was evidence of Café-au-lait spot and pedunculated cutaneous Neurofibromas. She has at least 21 or more, raised, soft, well-circumscribed, pedunculated neurofibromas on back and chest. The patient also has a prominent rounded macule, which is a café-au-lait spot. A single mass was first observed at the time of birth and it had slowly increased in size and number since then. Physical examination revealed a soft 4x3cm large one and 1x2 cm in diameter smaller ones skin colored mass. (Fig 10)

**Intra oral examination**

Intraoral examination categorized into hard tissue and soft tissue examinations,
Hard tissue examination:
Root piece with 16,17,26,27
Missing with 37, 45, 46
Stain ++, Calculus +
Intraoral soft tissue examination revealed Melanin pigmentation is noted in relation to right and left buccal mucosa. Anterio-posteriorly: Right and left side corner of mouth to retromolarpad area. Superio-inferiorly: right and left upper buccal mucosa to lower one. All other aspect e.g. palatal, labial, lingual appears to be normal. (Fig 11, 12)

On the basis of clinical examination and history involvement our working provisional diagnosis were Root piece with 16, 17, 26, 27, false partial anodontia with 37, 45, 46, Tobacco induced melanosis and Neurofibromatosis (type 1). For chief complaint patient advised to take IOPA with 16,17,26,27 as radiographic investigation.

After all confirmatory findings, our final diagnosis were

- Root piece with 16, 17, 26, 27
- False partial anodontia with 37, 45, 46
- Tobacco induced melanosis.
- **Neurofibromatosis (type 1)**

Treatment planning under various phases, in which first one was emergency phase that advice stoppage of habit. Tobacco cessation counseling, patient education, motivation and oral prophylaxis come under etiotrophic phase. Patient was advised extraction of 16,17,26,27 as surgical phase. Prosthetic and maintenance phase as per scheduled. For treatment of neurofibromatosis patient referred to dermatology department of near government hospital.

## DISCUSSION

Neurofibromatosis affecting more than one system of bodily organs. In literature epidemiology shows both genders and all races were equally affected. Incident rate of neurofibromatosis is 1 case per 3000 population in which half cases shows hereditary history and other half shows mutational changes. Neurofibromatosis is an autosomal dominant disease with characteristic hereditary pattern.[1, 3, 5]

### Classifications of Neurofibromatosis

**1. RICCARDI 1982**[2]

Type 1: Neurofibromatosis type 1 (NF 1)
Type 2: Acoustic (NF 2)
Type 3: Mixed (NF 3)
Type 4: Variant (NF 4)
Type 5: Segmental (NF 5)
Type 6: Familial CLS (NF 6)
Type 7: Late onset (NF 7)
Type 8: Unspecified (NF 8)

**2. Carey *et al. (1986) proposed that***

NF is classified into only five types, based on Clinical features and genetic implications for the patient, as follows:[1, 2]

NF1 – classical, NF2-acoustic, NF3-segmental, NF4 - CALM - familial

NF5- NF Noonan phenotype.

**3. Huson (1994):** (on the basis of clinical manifestation)[1]

- Major NF
- Minor NF

### Diagnostic Criteria of Neurofibromatosis[1,3,4]

1. **Café-au-lait:** Six and more café-au-lait > 5mm in larger diameter.
2. **Freckling:** Two or more freckles in axillary and inguinal regions.
3. **Neurofibromas:** Two or more of any type
4. **Glioma**
5. **Lisch nodules:** Two or more nodular Hamartoma in the iris
6. **Osseous dysplasia**
7. **Familial history**

### Prognosis

Neurofibromatosis shows higher mortality rate as compare to general population. Malignant transformation of tumor caused death of individuals. Mortality rate higher in female as compare to male, mortality age reported in literature as 10 to 40 years old patients. For accurate diagnosis of neurofibromatosis, advance radiographic investigations such as CBCT, CT, MRI, PET-SCAN and immunohistochemical (S-100 protein test).

## CONCLUSION

Neurofibromatosis has different types; this article reported neurofibromatosis type 1 with detailed examination of the growth. Accurate diagnosis and dimensions of the growth will help in reduction of recurrence rate. Oral physician and maxillofacial radiologist play important role in early diagnosis and reduce recurrence rate of the disease with the help of advance radiographic investigations and newer diagnostic aids.

**Financial support and sponsorship:** Nil.
**Conflicts of interest:** There are no conflicts of interest.

## REFERENCES

1. Antonio JR, Goloni-Bertollo EM, Trídico LA, "Neurofibromatosis: chronological history and current issues" An Bras Dermatol, 2013; 88(3): 329-43.
2. Riccardi VM. Historical background and introduction. In: Friedman JM, Gutmann DH, MacCollin M, Riccardi VM (Eds). Neurofibromatosis - Phenotype, Natural History, and Pathogenesis. 3 ed. Baltimore: The Johns Hopkins University Press, 1999: 1-28.
3. Ferner RE. Neurofibromatosis 1. In: Ferner RE, Huson SM, Evans DG (Eds.) Neurofibromatoses in clinical practice. 1 ed. London: Springer- Verlag London Limited, 2011: 1-46.
4. Boyd KP, Gao L, Feng R, et al. Phenotypic variability among cafe-aulait macules in neurofibromatosis type 1. J Am Acad Dermatol, 2010; 63: 440-447.
5. Brosius S. A history of von Recklinghausen's NF1. J His Neuro Sci, 2010; 19: 333-48.