Journal of Chemical Health Risks

www.jchr.org

JCHR (2024) 14(3), 1741-1745 | ISSN:2251-6727

Case of Von-Recklinghausen disease – A clinical report on the Diagnosis and Prosthodontic rehabilitation.

[1]Dr. Md. Rabiul Islam, [2]Dr. Samiran Das,[3]Dr. Kritika Rajan, [4]Dr. Ishan Roy Choudhury, [5]Dr. Avishek Bhandari

[1]Assistant Professor, Department of Prosthodontics and Crown & Bridge, Dr. R. Ahmed Dental College and Hospital, Kolkata.

[2]Professor, Department of Prosthodontics and Crown & Bridge, Guru Nanak Institute of Dental Science and Research, Kolkata.

[3]Assistant Professor, Department of Prosthodontics and Crown & Bridge, Dr. R. Ahmed Dental College and Hospital, Kolkata.

[4]Assistant Professor, Department of Prosthodontics and Crown & Bridge, Dr. R. Ahmed Dental College and Hospital, Kolkata.

[5]BDS, MDS (Oral Pathology), Private Practitioner.

Corresponding author – Dr. Kritika Rajan

Assistant Professor, Department of Prosthodontics and Crown & Bridge, Dr. R. Ahmed Dental College and Hospital.

Email ID – kritikarajan91@gmail.com

Mob no - +91-90073 04332

(Received: 08 February 2024 Revised: 11 March 2024 Accepted: 08 April 2024)

KEYWORDS

Neurofibromatosis, Removable Prosthesis, Oral manifestations

ABSTRACT:

Neurofibromatosis is a neurocutanous-skeletal syndrome caused by mutations in the NF1 tumor suppressor gene located at 17q11.2. It has a prevalence of ~ 1:2000–1:3000. The diagnosis of NF is based on criteria outlined by the National Institute of Health Consensus Development Conference in 1987. Cutaneous neurofibromas and café au lait spots are the hallmarks of NF1 on the skin. The oral involvement has been reported to occur in 3.4–92% of adult patients. Usually discrete, non-ulcerated nodules, varying from normal mucosal colour to red or even yellow, they are rarely seen occurring in 2% or possibly as many as 7% of cases. Since NF1 is not curable, diagnosis of such syndromes is of utmost importance such that proper and timely prosthodontic rehabilitation is offered to the patient for long term success of the prosthesis through multidisciplinary management. Dentists should be aware of the possible oral manifestations of neurofibromatosis because some of these lesions may become large enough to interfere with functions of mastication and speech. It is also imperative to put these patients on a long-term follow up for early detection of any possible signs of malignant transformation. Special attention should be paid to oral hygiene and regular follow up. This paper presents the diagnosis and prosthetic management for a case of Neurofibromatosis NF-1 in Recklinghausen disease.

Introduction -

The term neurofibromatosis (NF) is used for a group of genetic disorders that primarily affects the cell growth of neural tissues. It was first described by Robert Smith in 1849, but its name comes from its first publication in 1882, by Von Recklinghausen.[1] A total of eight forms of Neurofibromatosis has been recognized out of which two forms of neurofibromatosis are most prevalent. These are - Neurofibromatosis type 1 (NF1) and Neurofibromatosis type 2 (NF2). Neurofibromatosis type 1 (NF1), also known as Von Recklinghausen´s disease, is the most common type of NF and accounts for about 90% of all cases.

The inheritance is autosomal dominant and it is caused by a mutation in a tumour suppressor gene which maps to chromosome region 17q11.25.6. The hallmark of NF type 2 (NF2) consists of bilateral acoustic neuromas. NF2 is caused by mutation in a tumour suppressor gene which maps to chromosome region 22q12.2.[2]

Oral involvement has been reported to occur in 3.4–92% of adult patients, and in about 40% of children with NF1. Oral soft tissue manifestations in NF1 have been reported in 74% of patients, the most common

Journal of Chemical Health Risks

www.jchr.org

JCHR (2024) 14(3), 1741-1745 | ISSN:2251-6727

findings being prominent lingual papillae, overgrowth of gingival soft tissue, and mucosal tumours.[3]

Diagnostic criteria for Neurofibromatosis type 1 (NF1) [4]

The patient should have two or more of the following:

1. Six or more café-au-lait spots more than 0.5 cm in the greatest diameter in prepubertal individuals and more than 1.5 cm in post-pubertal individuals,
2. Two or more neurofibromata of any type or one or more plexiform neurofibroma,
3. Freckling in the axillary or inguinal regions (Crowe's sign),
4. An optic pathway tumour,
5. Two or more Lisch Nodules (iris hamartomas),
6. A distinctive osseous lesion, such as sphenoid wing dysplasia or thinning of the cortex of the long bones (with or without pseudoarthrosis) and
7. A first-degree relative with NF1 by the above criteria.

Diagnostic criteria for Neurofibromatosis type 2 (NF2) [4]

The patient should have the following clinical criteria:

1. Bilateral vestibular schwannomas (VS) or
2. Family history of NF2 (first degree family relative) plus

a. Unilateral VS < 30 years or

b. Any two of the following: meningioma, glioma, schwannoma, juvenile posterior sub-capsular lenticular opacities/juvenile cortical cataract.

Usually discrete, non-ulcerated nodules, varying from normal mucosal colour to red or even yellow, they are rarely seen occurring in 2% or possibly as many as 7% of cases. They are in the soft tissues such as the cheek, palate, tongue, floor of the mouth and lips. The tongue is the most common affected site. If the neurofibromas are deep, the tongue appears enlarged (Macroglossia); if they are superficial, it appears fissured (Scrotal).[5] Tumours also occur on the gingival and buccal mucous membrane. Neurofibromas may occur within the periodontal membrane and lead to the migration of teeth. It can also develop intra-osseously, resulting in well-demarcated unilocular, but occasionally multilocular, radiolucent lesions.[6]

Surgery is currently the only treatment option for most of the lesions in NF1. Chemotherapy is helpful with visual pathway tumours. Radiation therapy should be avoided except with malignant tumours because it can stimulate the growth of plexiform lesions. Surgical removal of cutaneous neurofibromas is indicated if they are causing discomfort or are visible and stigmatizing. Resection of plexiform lesions should be considered when there is cosmetic disfigurement, pain, or compromise of function.[7]

This paper presents the diagnosis and prosthetic management for a case of Neurofibromatosis NF-1 in Recklinghausen disease.

Case report –

I) Diagnostic phase -

A 68-year-old woman reported to the Department of Prosthodontics for oral rehabilitation. She had short stature, and was moderately built. She presented with several pendulous tumour masses of skin, which were loose overgrowths hanging in folds. (Figure 1)

Figure 1 - Extra-oral photograph

They were sessile, elevated smooth surface nodules of variable sizes and scattered all over the body, the trunk, face, extremities, scalp and also present intraorally. More than 20 café-au-lait spots were noted on the trunk and upper arms which were flat and brown pigmented. On intraoral examination, it was found that the patient was partially edentulous, she had a poor oral hygiene with lots of deposits of plaque, calculus and stains. (Figure - 2, 3) There was bleeding on probing (Grade 1), there was presence of discrete nodular fibrotic growths, of the same colour as that of the normal mucosa, which resembled gingival hyperplasia or gingival fibromatosis. They were scattered throughout the entire alveolar ridge buccally, lingually as well as in the palate. Written consent was obtained from the patient to avoid ethical conflicts in documenting her case.

Journal of Chemical Health Risks

www.jchr.org

JCHR (2024) 14(3), 1741-1745 | ISSN:2251-6727

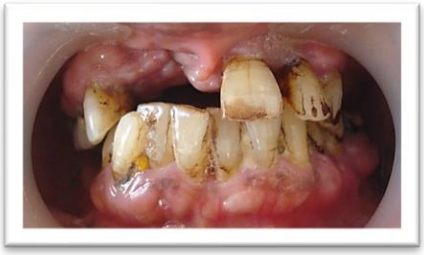

Figure 2 - Lesions present on the alveolar ridge

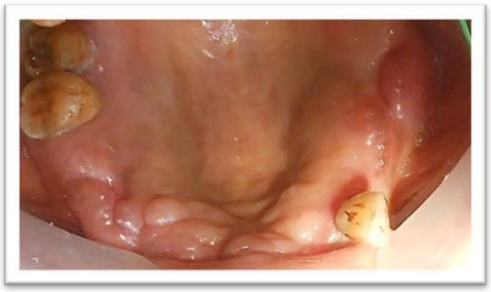

Figure 3 - Lesions present on the maxillary ridge

The orthopantomography revealed enlarged mandibular canal, mandibular foramen and mental foramen along with decreased mandibular angle and alteration in coronoid notch morphology. (Figure - 4)

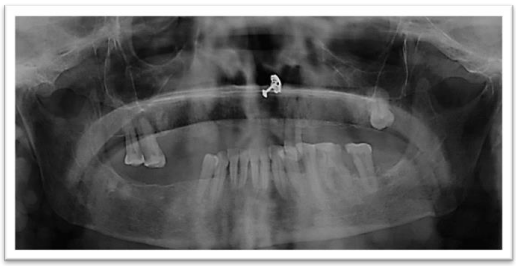

Figure 4 - Orthopantomogram showing enlarged mandibular canal and mental foramen

A fibromatous lesion was excised from the pre-molar region on the left side of the lower arch and sent for histopathological examination. Under magnification of: 10x, 40x and 100x it showed a clear case of Neurofibroma Type 1 consisting of elongated fibroblasts with sinuous nuclei separated by fine and equally sinuous collagen fibres. (Figure – 5, 6)

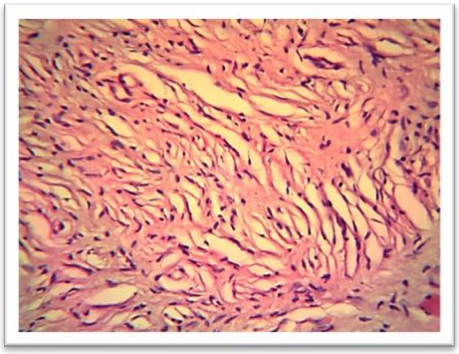

Figure 5- Photomicrograph showing (H-E stained; Magnification:40x) Neurofibroma consisting of elongated fibroblasts with sinuous nuclei separated by fine and equally sinuous collagen fibres.

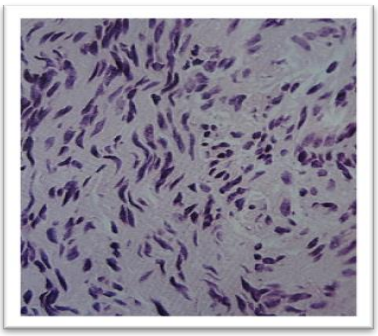

Figure 6 - Photomicrograph showing (H-E stained; Magnification:100x) Neurofibroma: consisting of elongated fibroblasts with sinuous nuclei separated by fine and equally sinuous collagen fibres.

II) Pre-prosthetic phase –

After histopathological examination, oral prophylaxis and extraction of hopeless tooth, which was carious 13, root stumps in 14 and 15, patient reported to the Department of Prosthetic Dentistry for prosthodontic rehabilitation.

The prosthetic treatment options for her dental reconstruction were either a conventional removable prosthesis or an implant-supported removable protheses. The patient was given explanation regarding the advantages and limitations of both these prostheses and the conventional removable prosthesis was chosen due to hygiene as well as financial constraints. Pre-prosthetic surgery was recommended in certain areas in order to remove the nodular masses on the edentulous arches to improve the denture foundation.

III) Prosthodontic rehabilitation phase -

Journal of Chemical Health Risks

www.jchr.org

JCHR (2024) 14(3), 1741-1745 | ISSN:2251-6727

Preliminary impressions were made using alginate with the help of suitable stock trays. (Figure - 7) Primary casts were made and the areas with tissue overgrowth was relieved with wax. Special trays with complete spacer design were fabricated with autopolymerising resin. Special trays were checked in the patient's mouth and border moulding was done using low fusing green stick compound and the secondary impressions were made using low viscosity alginate impression material.

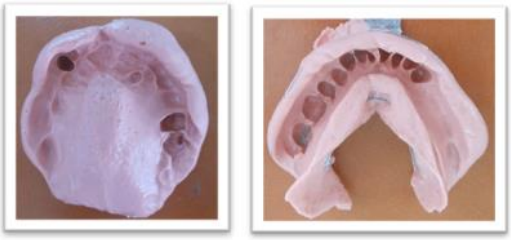

Figure 7 - Maxillary and Mandibular Preliminary impressions

Master cast (Figure - 8) was obtained and was used to fabricate a temporary denture base of autopolymerising resin, on which an occlusal rim was made out of modelling wax. The vertical dimension was determined using Niswonger's technique, phonetics and aesthetics. The patient was guided into centric relation and the relation was recorded. Once the teeth arrangement was completed, trial was done and corrections were made accordingly. (Figure - 9) The prosthesis was polished and inserted into the patient's mouth. Necessary corrections and adjustments were made. (Figure - 10)

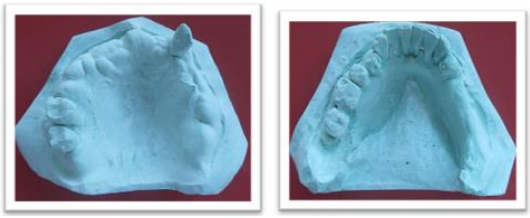

Figure 8 - Maxillary and Mandibular master casts

Figure 9 - Teeth arrangement on Articulator

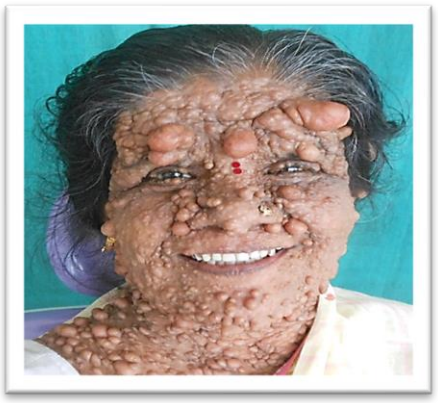

Figure 10 – Post Denture insertion

Discussion –

Conventional prostheses may not be appropriate because they are incompatible with excessive soft tissue contour and defective bone morphology. Despite the long-term success of the implant-supported fixed protheses, the placement of sufficient numbers of implants can be restricted due to financial constraints. Implant-supported removable prostheses terms of improvement of mastication, speaking ability, and quality of life.[8] Also, implant-supported overdentures will create pink interdental papilla better than implant-supported fixed prostheses and as well, they have flanges to rehabilitate the support-less soft tissue. To avoid peri-implantitis, in patients with poor oral hygiene, implant-supported removable prostheses have been mostly indicated because it's easy to keep them clean.

Since NF1 is not curable, multidisciplinary management is recommended.[9] Dentists should be aware of the possible oral manifestations of neurofibromatosis because some of these lesions may become large enough to interfere with functions of mastication and speech. It is also imperative to put these patients on a long-term follow up for early detection of any possible signs of malignant transformation. Special attention should be paid to oral hygiene and regular follow up.[10]

References -

1. Michele Callea, An unusual case: Neurofibromatosis Type 5; J Int Dent Med Res 2012; 5: (2), pp. 102-5.
2. Cunha K. S. G, Barboza E. P, Dias E. P, Oliveira F. M. Neurofibromatosis type I with periodontal

Journal of Chemical Health Risks

www.jchr.org

JCHR (2024) 14(3), 1741-1745 | ISSN:2251-6727

manifestation. A case report and literature review. Br Dent J 2004; 196(8):457-460.

3. Jouhilahti EM, Visnapuu V, Soukka T, Aho H, Peltonen S, Happonen RP, Peltonen J. Oral soft tissue alterations in patients with neurofibromatosis. Clin Oral Investig. 2012; 16: 551–8.
4. Gutmann DH, Aylsworth A, Carey JC, Korf B, Marks J, Pyeritz RE, Rubenstein A, Viskochil D. The diagnostic evaluation and multidisciplinary management of neurofibromatosis 1 and neurofibromatosis 2. JAMA. 1997 Jul 2;278(1):51-7.
5. K. S. G. Cunha, E. P. Barboza, E. P. Dias and F. M. Oliveira. Neurofibromatosis type I with periodontal manifestation. A case report and literature review; British Dental Journal Vol 196 no. 8, 2004.
6. Rupak S, Neurofibromatosis Type I with oral manifestations; e-Journal of Dentistry Jan - Mar 2012 Vol 2 Issue 1.
7. H. Tonsgard, Clinical Manifestations and Management of Neurofibromatosis Type 1; James, Semin Pediatr Neurol 13:2-7 2006.
8. Kokovic V, Lazic V, Petrovic M, Jelovac D, Todorovic A (2012) Implant Restoration in a Patient with Neurofibromatosis Type 1: A Case Report. Dentistry 2:124.
9. Wotjiuk F, Hyon I, Dajean-Trutaud S, et al. Dental Management of Neurofibromatosis Type 1: A Case Report and Literature Review. Int J Clin Pediatr Dent 2019;12(6):577–81.
10. M S, Joseph B, Nath SG, Sheejith B. An unusual case of multiple intraoral manifestations of neurofibromatosis type 1: case report with literature review. J Clin Diagn Res. 2014 Dec;8(12): ZD 20-2.