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RESEARCH ARTICLE

AN UNCOMMON COMBINATION OF NEUROFIBROMATOSIS TYPE 1 WITH INTRAORAL HAEMANGIOMA AND PORT WINE STAIN: A CASE REPORT

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Abstract

Neurofibromatosis (NF) is a group of genetic disorders that primarily affect neural tissues. NF type I also known as Von Recklinghausen's diseaseis the most common type of NF, it accounts for about 90% of all cases, with a prevalence rate of one in 3,000 births. The disease is often characterized by complex and multicellular neurofibroma which may lead to different complications for an affected individual throughout life, so it is important to throw light on type I NF.

Introduction:-

NF is a benign neural tumour derived from peripheral nerve sheath and it consists of Schwann cells, perineural fibroblast, or both.[1] Von Recklinghausen coined the term Neurofibroma in the year 1881. Neurofibroma can occur alone or as a part of NF.[2]

NF is an autosomal dominant hereditary disorder to a variety of related syndromes which is characterized by the neuroectodermal tumour.[3]Prevalence of NF type I is about 1/3,000.[1] NF has been classified into eight different types by Riccardiwhich is NF1-NF8.[2]Only 4%-7% of oral involvement of NF has been reported,[1] so it is of utmost importance to know different oral manifestations in NF to avoid complications.In this article, we present a case of NF type I with intraoral haemangioma and port-wine stain in a 62-year-old male patient.

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Case Report:
A 62-year-old male patient reported to the Department of Oral Medicine and Radiology with the complaint of multiple painless swellings on the middle and lower third of the face for 35 years. The patient noticed single painless swelling on the lower lip 35 years ago and had undergone surgery, after which there was a recurrence of the swelling in the same region and on other parts of the face. The patient also noticed pigmentation in the chest region at the same time. Past medical and dental history was not contributory. No other members in the family had a similar malformation. The patient had a habit of smoking beedi and paan chewing for 30 years, 3-4 times a day. The patient was moderately built and nourished, all vital signs were under the normal limit. As a part of routine protocol, informed consent has been obtained. Extraoral examination revealed multiple nodules on the middle and lower third of the face including right and left ears of varying size, largest nodule measuring 3x3 cm, and smallest nodule measuring 0.5x0.5 cm, round to ovoid in shape, smooth surface, pedunculated with a bag of worm appearance inconsistency on palpation,dark brown pigmentation noted on the middle and lower third of the face bilaterally.[Figure 1] Café au lait pigmentation is noted on the center of the chest measuring 4x4 cm.[Figure 2] Intraoral examination revealeddiffusely swollen lower lip, pinkish-brown in color, non-tender, soft in consistency on palpation. Solitary nodule noted on the right side of the floor of the mouth, underneath the tongue, ovoid, measuring approximately 2x2 cm, pinkish-red in color, smooth surface, non-tender, firm in consistency, pedunculated. Diffuse swelling was noted on the right of the ventral aspect of the tongue, extending till the midline, purplish-red with a well-defined margin, pulsatile, and the diascopy was positive. [Figure 3A] Port-wine stain was noted on the right side of the palate extending 5cm from the incisive papillae up to the palatoglossal arch which does not cross the midline, and the diascopy was positive. [Figure 3B]Based on history and clinical features provisional diagnosis is given as Neurofibromatosis with intraoral haemangioma.

Contrast-enhanced computed tomography of the face, neck, and Non-contrast computed tomography was done to rule out cranial involvement, which reveals heterogeneously enhancing diffuse en plaque skin thickening with multiple pedunculated lesions in the face and neck suggestive of diffuse cutaneous neurofibromas. [Figure 4] No abnormalities were noted in the brain.Based on investigations, the final diagnosis was given as Neurofibromatosis type I with intraoral haemangioma. Schwannoma, peri neuroma was considered a differential diagnosis.

No treatment has been done as recurrence after surgery was seen in this case, the long-term periodic follow-up has been advised to screen for the development of other lesions.

Discussion:-

NF is a benign slowly progressing genetic disorder affecting the skin and the nervous system.[4] The classic description of this is given by a German pathologist, Friedrich Daniel Von Recklinghausen who described the variety of findings as a single entity in the year 1882, thus the condition is often referred to as Von Recklinghausen disease.[3]

NFI is an autosomal dominant disorder caused by an alteration in the NFI gene which is a tumor suppressor located on the long arm of chromosome 17 (17q11.2)[5] Riccardi (1982) classified NF into eight types, designation, clinical features, and pattern of inheritance are listed in Table 1.

The diagnostic criteria for NF1 were proposed by the National Institute of Health Consensus Development Conference in 1988. The patient should have two or more of the following findings.

1. Five or more café au lait macule
2. Two or more Neurofibroma of any type or one plexiform neurofibroma.
3. Axillaryor Inguinal freckling.
4. Optic glioma
5. Lisch nodule
6. Distinctive osseous lesions such as sphenoid dysplasia
7. Family history of first degree relative with NF

Neurofibromas in the oral cavity often show involvement of trigeminal and upper cervical nerve, whereas the mandibular branch of trigeminal nerve involvement is seen in our case. Intraorally neurofibroma occurs most commonly on the tongue, lip, gingiva, major salivary gland, and maxillary bone.[1]It is rarely seen in the palate and floor of the mouth.[7]Other oral manifestations include macroglossia, enlarged fungiform papillae, bony deformities, wide inferior alveolar canal, and enlarged mandibular foramen.[4]In our case, it is seen on the floor of the

mouth.Along with NF other conditions can also be seen intraorally like, epulis, ossifying fibroma, pyogenic granuloma, odontogenic fibroma, giant cell granuloma, haemangioma, and lipoma.[7]In our case,haemangiomawas noted on the ventral aspect of the tongue.

Investigations that are helpful in the diagnosis of NF1 are Magnetic Resonance Imaging (MRI) to identify the extent and size of plexiform neurofibromas, optic pathway gliomas, structural abnormalities, and tumours of the brain. Conventional radiographs to demonstrate the skeletal anomalies in NF1 patients.Positron emission computed tomography and biopsy are helpful in distinguishing benign and malignant peripheral nerve sheath tumors.[8]

Three-dimensional computed tomography (3D CT) is useful in indicating the extent of the lytic lesion and identifying additional bony deformities of the mandible.[9]Immunohistochemistry to detect S-100 protein which is specific for neural crest cells.[4]

Treatment involvesregular appointments to the multidisciplinary clinic which includes a primary care physician, dentist, and dermatologist.[1] Cutaneous neurofibromas can cause transient stinging, itching, and aesthetic problem which leads to decreased sociability.[10]Such neurofibromas should be treated by a skilled surgeon and plastic surgeon.[11]Tumors larger than 4cm should be surgically excised, tumors less than 2cm should be excised by CO2 laser therapy, Photocoagulation can be done to the tumor less than 1cm, Electrodesiccation to very small tumor less than 5mm, rapid surgery like Radiofrequency ablation/diathermy loop can also be followed.[12]

Table 1:- Riccardi classification of the Neurofibromatosis.[6]

Category	Description	Phenotypic Features
NF-I	Von Recklinghausen	Heritable, diffuse café -au-lait spots, diffuse neurofibromas, Lisch nodules, CNS-derived neural-crest tumors
NF-II	Acoustic	Heritable, few and large diffuse café-au-lait spots, few neurofibromas, bilateral acoustic neuromas, no Lisch nodules
NF-III	Mixed: Mixture of primary features of NF1 and NF2	Heritable, diffuse café-au-lait spots, variable to few neurofibromas, multiple CNS brain and spinal cord tumors (hallmark)
NF-IV	Variant	Diffuse café-au-lait spots and neuromas
NF-V	Segmental	Café-au-lait spots and neurofibromas with distribution limited to a given region of the body
NF-VI	Café-au-lait spots	Diffuse café-au-lait spots, no neural fibromas, no other neural-crest tumors
NF-VII	Late-onset	Diffuse neurofibromas (cutaneous or deeper) in the 3rd decade of life or later
NF-VIII	Not otherwise specified	All other forms of NF that do not fit into the above categories

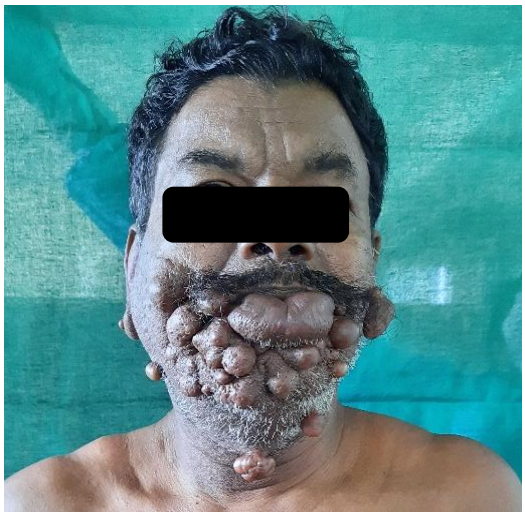

Figure 1:- Multiple nodules on the middle third and lower third of the face.

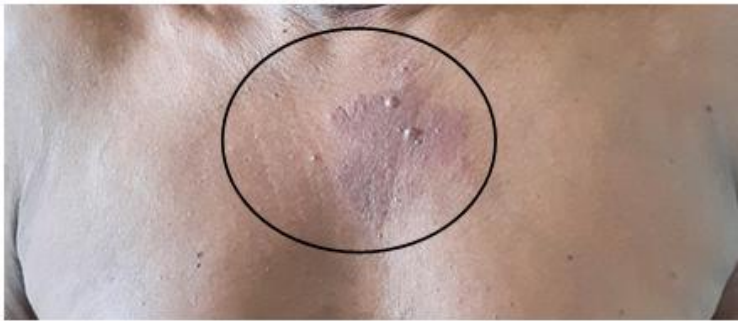

Figure 2:- Café au lait macule on the chest.

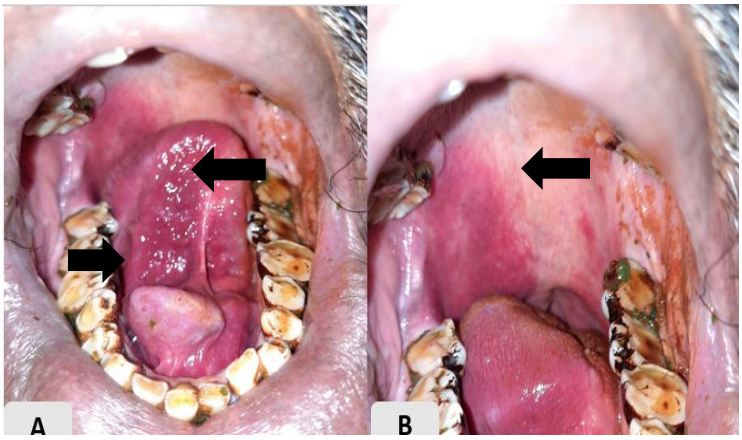

Figure 3A:- Neurofibroma was noted on the floor of the mouth, haemangiomawas note on the ventral half of the tongue on the right side.
Figure 3B:- Port wine stain on the right half of the hard and soft palate.

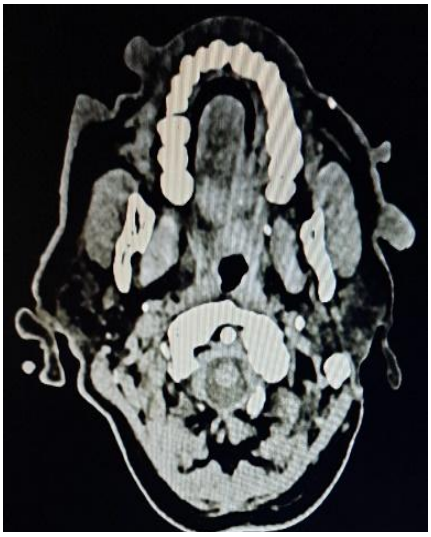

Figure 4:- Computed tomography image showing neurofibroma on the middle third of the face.

Conclusion:-

NF1 is a common neurocutaneous condition with an autosomal dominant pattern of inheritance. Because of the varying features and clinical diversity inherent to this disorder, patients can present to different medical specialists and dentists. Thus, for prompt diagnosis and to provide optimum care for patients, clinicians must be aware of the diverse clinical features of this disorder. Reliable clinical and radiological assessment will be helpful for the treatment plan.

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