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Neurofibromatosis type –I, A rare genetic entity in adolescent patient: A case report and literature review.

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Abstract

A patient presented to our department of oral and maxillofacial surgery, IDS, Bareilly, with the chief complaint of painless swelling over right lateral nose and left temporal region along with multiple nodules. OPG was non-remarkable, whereas, MRI confirmed the presence of soft tissue mass in the right lateral region of nose and left temporal region. Treatment was planned to surgically excise the lesion in-toto followed by primary closure. MRI played an important role in establishing the diagnosis of rare case of neurofibromatosis apart from the clinical evaluations and the treatment resulted in good prognosis with no further signs of recurrence on follow-ups.

Keywords: Neurofibroma, soft tissue mass, MRI, surgical excision, primary closure.

Introduction

Neurofibroma is a benign non-odontogenic tumor that is uncommon. Neurofibromas may appear as solitary lesions or as part of a larger neurofibromatosis or von-Recklinghausen's disease (VRD) of the skin syndrome. Neurofibromatosis is a disease entity with two distinct

variants that vary genetically, histologically, and clinically.[1] Neurofibromatosis type -1 (NF-1), also known as VRD, is one of the most common autosomal dominant inherited diseases, with an incidence of one in every 3,000 people.[2] The wide spectrum of symptoms associated with this neurocutaneous condition puts the patient at a higher risk of morbidity and mortality.[3]

We report a case of involving the right lateral nasal region and left temporal region, diagnosed on the basis of clinical history and findings in MRI.

Case report

A 17 years old boy reported to the department of Oral and Maxillofacial Surgery with the chief complaint of painless swelling over right side of nose and left temporal region since birth (Fig. 1a, b). Patient gives no history of any adverse habits and past medical history was non - contributory. Family history revealed that his 3 brothers (Fig 2 a, b), mother and grandmother (Fig 3 a, b, c) also suffering from multiple solitary, generalized swellings. On systemic examination all parameters were within normal limits. Café-au-lait spots were seen on shin region and multiple solitary nodules were also seen at trunk, back, upper and lower limbs. The inspection revealed a solitary diffused swelling with well demarkable, imperceptible margins along the lateral aspect of the nose of right side and on the left side above the temporal region extending over the forehead region, Palpation was confirmatory of the inspection findings and revealed a painless swelling which was fixed to the underlying subcutaneous tissue structures. Mouth opening of the patient was normal.Upon further investigations, the orthopantomogram (OPG) was found to be non- remarkable. Magnetic resonance imaging (MRI), confirmed the presence of cystic soft tissue mass in the right lateral region of nose and left temporal region. (Fig. 2a, b). Biopsy was done for the lesion which revealed hypocellular myxoid stoma with fine fibrillar cells and fusiform spindle cells suggestive of neurofibromatosis type-1. Surgical management was instituted under general anesthesia as surgical excision (Fig. 3 a, b, Fig 4 a, b) of the lesion in-toto followed by primary closure which resulted in good prognosis with no further signs of reccurence on follow-ups (Fig. 5a, b).

Discussion

Solitary neurofibroma is a benign non-odontogenic tumour that is highly rare. Neurofibromas may appear as a single lesion or as part of a more widespread syndrome. Neurofibromatosis is a disorder that has two distinct forms that are genetically, histologically, and clinically distinct.3 One of the most common autosomal dominant inherited disorders, neurofibromatosis type-1 (NF-1), also known as von-Recklinghausen's disease of the skin, is neurofibromatosis type-1 (NF-1). The NF-1 gene is a large complex with several different manifestations in terms of gene organization and expression when it is mutated.4 The protein encoded by this gene (neurofibromin) is a GTPase activator that is expressed in a variety of tissues; its absence causes significant developmental abnormalities.5 Neurofibromatosis type-2 (NF-2), also known as central neurofibromatosis, is an autosomal dominant disorder that accounts for a very small percentage of all cases of neurofibromatosis. The hallmark of NF-2 is the presence of bilateral vestibular schwannomas.1,6 Neurofibromas, which are typically associated with VRD, are generally present as numerous lesions and very rarely as a single tumour, as in our case. The plexiform neurofibroma, on the other hand, is an unusual entity in NF-2, and rapid growth of a plexiform neurofibroma usually indicates that it will turn into a neurofibrosarcoma.7 On clinical examination neurofibromas usually presents as pedunculated or sessile nodules, with slow growth and usually painless in nature. Treatment is surgical and the prognosis is excellent. For illustration a rare case of

neurofibroma in the right nasal region as well as in left temporal region is presented. Plexiform neurofibromas are poorly circumscribed tumours that present as tortuous cords along the segments and branches of a nerve with a proclivity to develop centrally.8 This tumour is said to be representative of VRD, even though it may be the only manifestation of the disease.9,10 Neural sheath tumours are uncommon in the oral cavity, despite their high prevalence in the head and neck. Oral signs are seen in just 4-7 percent of patients with neurofibromatosis.11 Since neurofibromas normally have numerous lesions, the entire body, as well as the larynx and trachea, must be examined in a patient with oral neurofibroma, since lesions in the upper airway can cause respiratory obstruction. Yamada et al. identified a seven-month-old baby who had breathing difficulties and a sublingual mass. The patient died of respiratory failure, and autopsy showed severe plexiform neurofibromas affecting the vagal, chronic laryngeal, and phrenic nerves, as well as laryngeal submucous plexiform neurofibromatous nodules.12 In our patient, we did not encounter any lesion in the upper airway. The treatment of such lesions is generally surgical and the diagnosis can only be confirmed after histological examination. The family members of the index case should also be examined, since intragenic microsatellite markers were reported to be highly informative for familial neurofibromatosis in Turkish families.13Familial café-au-lait spots is a disorder with an uncertain relationship to NF-1. Studies described families whose affected members had multiple café-au-lait macules (CALMs). It's important to distinguish a neurofibroma from a schwannoma histopathologically, since von Recklinghausen's neurofibromatosis-associated neurofibroma has a higher risk of malignant transformation, ranging from 5 to 16 percent.14,15 Neurofibromas have extensive vascularity and tend to bleed during surgery. Therefore, excessive bleeding should be kept in mind while attempting surgical removal. In our patient, despite a poor cleavage plane and bleeding, we were able to accomplish total extirpation of the lesion preserving the surrounding tissue. The resection bed was primarily closed. Early detection of recurrences and the appearance of other manifestations of VRD, particularly central nervous system tumours, is critical in such a patient, and these patients need ongoing follow-up throughout their lives to detect recurrences and the appearance of other manifestations of VRD, particularly central nervous system tumours (namely acoustic schwannomas and optic nerve gliomas).6Fortunately, there were no signs of recurrence or other manifestations of VRD during the follow-up period of our patient.

Table 1: Showing reported cases of neurofibromatosis till date and symptoms (Note: Our case is the fifth reported case in 17 year age group male patient and fifth study to report asymptomatic type -1 neurofibromatosis

Authors	Year	Cases	Symptoms
Kariyappa M. *etal*	2015	11/M	Headache with tonic clonic convulsions
Urbizu A. *etal*	2013	Series of 382 cases with CMI	11.8% with asymntomatic neurofibromatosis type 1
Ghosh P. *etal*	2012	Series of 396 children with NF1	CMI (n=3), headache
N. Plumpe*etal*	2010	9/M	Inappetance, an increased need to sleep and recurrent headaches
D.SantosGracia*et al*	2007	60/F	Asymptomatic

Felicio AC et al	2007	31/F	Right side hemifacial spasm
Herrero A. et al	2007	23/F	Headache
Hara H.,Arakawa H.*et al*	2005	29/F	Gait disorder, sensory and urinary disturbances
Tubbs RS *et al*	2004	Series of 198 cases of CMI	8.6% of cases with asyptomatic NF-1
Chakravarty A. et al	2002	22/F	Optic nerve glioma, scoliosis, syringomelia
Guistini S. et al	2002	2 cases	Asymntomatic (case1) and hydrocephalus (case 2)
Batissela PA *et al*	1996	11/M	Headache
Dooley J et al	1993	16/M	Asymptomatic
Tominga T et al	1991	1 case	Headache, hydrocephalus
Afifi AK et al	1988	2 cases	Hydrocephalus (both)
Parkinson D, Hay Ret al	1986	1 case	Rhinorrhea, fistula

Conclusion

Neurofibroma is although a rare benign lesion of maxillofacial region, but it should be considered as differential diagnosis of swelling over maxillofacial regions and its syndromic association should also be taken into consideration. Genetic and family predisposition in such patients is seen very often. MRI play an important role in the diagnosis of Neurofibromatous cystic lesion. Though being a benign lesion it can be treated with surgical excision followed by primary closure, the chances of recurrence are also minimal.

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Legend Figure

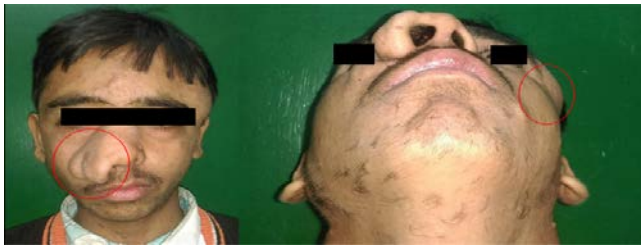


Fig. 1: a Diffused swelling extending to right lateral part of nose. b:Worm's eye view showing swelling in right base of the ala and left temporal region.

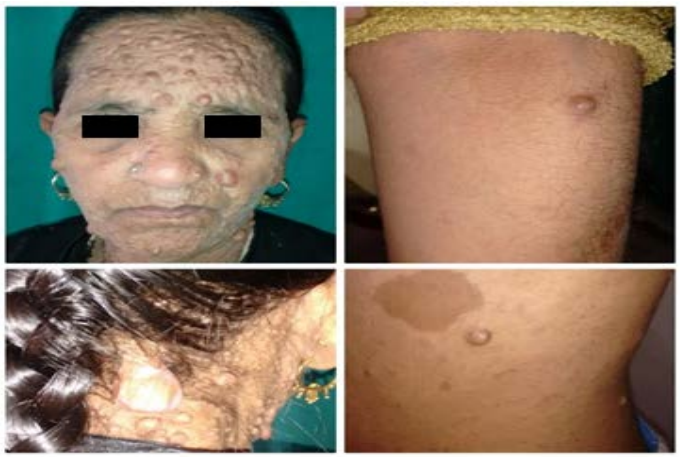
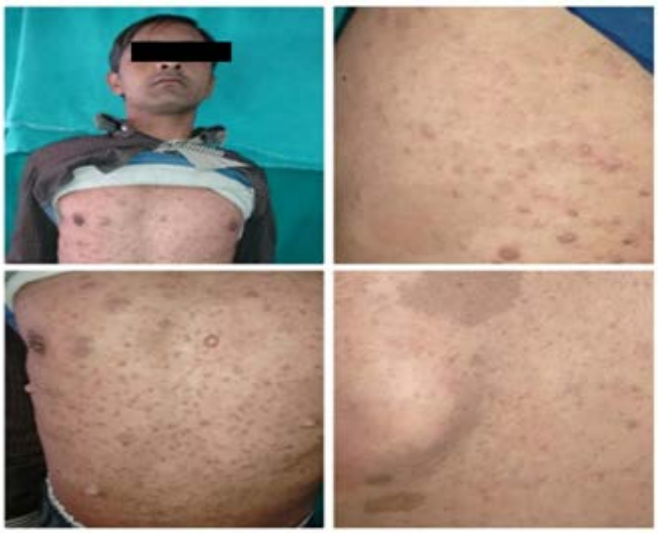


Fig 2 a: Familial disposition showing photos of patient's brother, lesions at upper chest, trunk, arm and shin region.



b:Familial disposition showing photos of patient's brother, lesions at upper chest, trunk, arm and shin region.

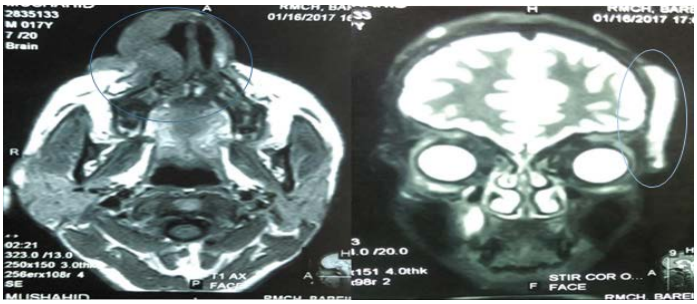


Fig. 3: a Axial section of MRI showing Epithelial cystic mass at right Lateral part of nose. b: Coronal section of MRI showing mass at left temporal region.

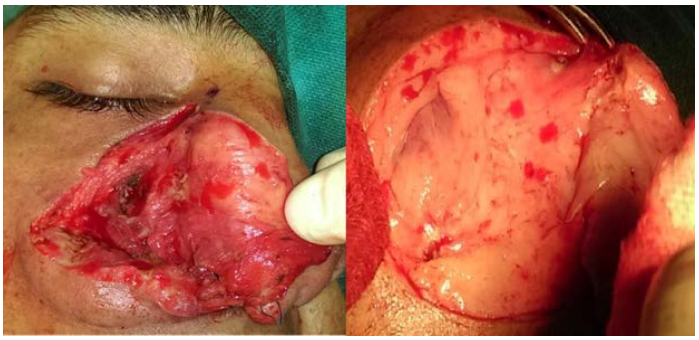


Fig. 4: a Showing intra operative excision of mass

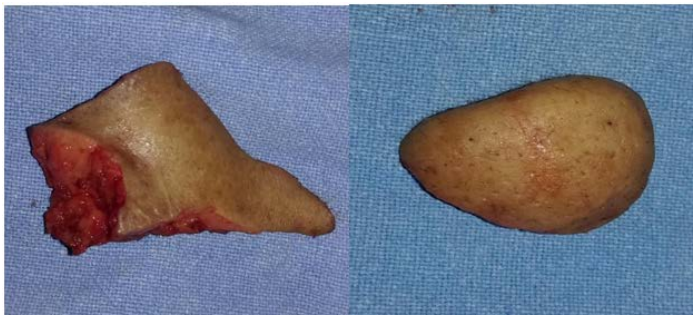


Fig. 4 b Showing excised mass of lesion at right lateral nasal region and left temporal region.

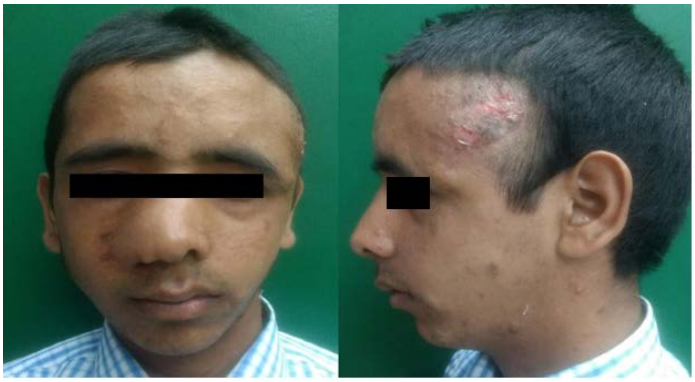


Fig. 5: a, b: Showing the lesion sites at postoperative follow up of the Patient.