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NEUROFIBROMAS IN THE EXTERNAL AUDITORY CANAL OF A PATIENT WITH TYPE 1 NEUROFIBROMATOSIS- A CASE REPORT

Otolaryngology

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ABSTRACT

Neurofibromas are common tumours of nervous system, but only few cases have the external auditory canal involvement. Here we are reporting one such case of neurofibroma in 32 year old male patient who presented with bilateral swelling in the external auditory canal and conductive hearing loss on the left side.

KEYWORDS

Neurofibroma, External auditory canal, Neurofibromatosis type 1.

INTRODUCTION:

Neurofibromatosis (NF) comprises a distinct genetic disorder characterized by benign growths of peripheral nerve sheaths, Neurofibromas and cafe au lait macules (CALMs), associated with various other cutaneous and systemic manifestations.[3] The national institute of health (NIH) U.S.A held a consensus development Conference which clarified the 2 main types of neurofibromatosis. Type 1 and type 2 neurofibromatosis.[3]

Neurofibromatosis type 1 (NF1) was delineated by von Recklinghausen in late 19th centuary.[1] It is also known as Von Recklinghausen's disease which is a multisystem disorder without special predilection for race and sex occurring at a frequency of 1 per 2500-3000 births. NF 1 is inherited as an autosomal dominant trait with complete penetrance by the age of 5 years. The gene for NF 1 is located on chromosome17 q11.2 and encodes a protein called neurofibromin.[3]

Diagnostic criteria for Nf1

1. Six or more CALMs, >5mm in greatest diameter at prepubertal age and > 15mm in greatest diameter in adults.
2. Two or more neurofibromas of any type or one plexiform neurofibroma.
3. Axillary or inguinal freckling.
4. Optic glioma.
5. Two or more Lisch nodules.
6. A distinctive osseous lesion: thinning of cortex of long bones with or without pseudoarthrosis.
7. First degree relative with NF1 (parent, sibling, offspring).[3\]

The diagnosis of NF1 is made on the basis of clinical features requiring the presence of at least two of the above criterias.

Plexiform neurofibromas (PNs) also known as plexiform neuroma, pachydermatocele or neurofibromatous elephantiasis has been classified as benign tumour of nerve sheath involving multiple nerve fascicles.[2] They are commonly seen along the branches of the trigeminal nerve and cervical nerves and consists of a large, diffuse, elongated lesion with a bag of worm feel.[1] PNs are slow growing highly vascularised, locally invasive non metastatic tumour. PNs are one of the important complications of NF1. Although frequently associated with NF1, it is not pathognomonic of NF1. The most common area involved is trunk (43%), head and neck (42%) and limbs (15%). PNs may give rise to malignant nerve sheath tumours referred to as neurofibrosarcomas and malignant schwanommas.[2]

CASE REPORT:

A 32 year old middle age man presented to the ENT outpatient department of Sri Siddhartha Medical College, Tumkur, with a history of bilateral swellings in the External auditory canal since 2 years, decreased hearing on left side since 6 months. Initially the swelling was pea sized and gradually progressed to the present size of approximately 1.5*1cm. There was no sudden increase or decrease in the size of the swelling. No history of pain or discharge from the swelling. No history of giddiness, ringing sensation, pain, discharge or bleeding from the ear, no history of ear trauma, no history of co-morbidities like diabetes mellitus, hypertension, bronchial asthma, tuberculosis, epilepsy etc. No history of previous ear surgeries.

Patient has Neurofibromatosis type 1 diagnosed at the age of 1 year by a paediatrician in his home town and is the first case in his family.

Patient is married and has a daughter aged 4years and no significant history among family members.

On examination of the Ear: A solitary ovoid swelling of approximately 1.5*1cm seen obliterating the left EAC, (Figure 1) Swelling was slightly mobile, non tender. Similar shaped swelling of approximately 0.5cm seen in the right EAC (Figure 2). Multiple neurofibromas are seen in the post-auricular region on both the sides. Tympanic membrane was not visualized on left side. Right side tympanic membrane was normal. Tuning fork test with 512Hz revealed conductive hearing loss on left side. Pure tone audiogram with bone conduction threshold revealed 40dB. Bilateral facial nerve motor function was normal. Nose and throat examination was normal.

Patient has nodular growths suggestive of neurofibromas on face, neck, trunk, abdomen, upper and lower limbs along with cafe-au-lait macules. (Figure 3). Clinical examination of nose, oral cavity and larynx was normal. Patient also had a large plexiform neurofibroma on the right foot. (Figure 4)

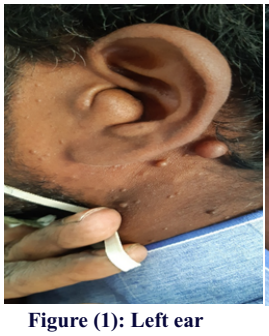


Figure (1): Left ear

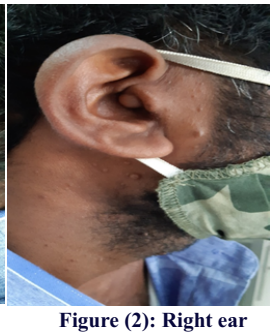


Figure (2): Right ear

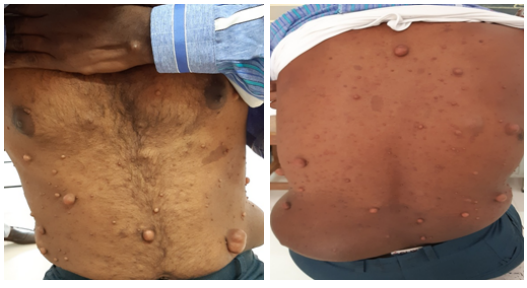
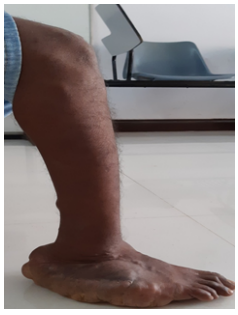


Figure (3): Multiple neurofibromas on chest, abdomen and back along with cafe-au-lait macules.



Figure(4): Plexiform neurofibromatosis of right foot.

Ophthalmic examination revealed bilateral Iris Lisch nodules (Figure 5). Visual acuity was normal.

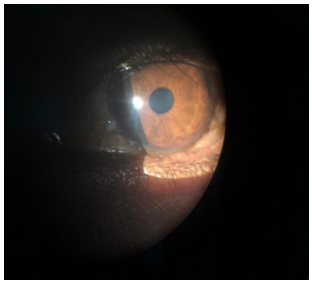


Figure (5): Iris Lisch nodules.

High resolution CT scan of temporal bone showed high riding of right jugular bulb suggestive of jugular bulb dehiscence (Figure 6). Middle ear, inner ear and mastoid appeared normal on CT scan on both sides. MRI scan with gadolinium contrast of brain did not show any evidence of optic glioma and other intracranial pathologies. Histopathology of external auditory canal swelling demonstrated neurofibroma with lobulated pattern, lobules showing bundles of spindle cells in myxoid stroma (Figure 7).

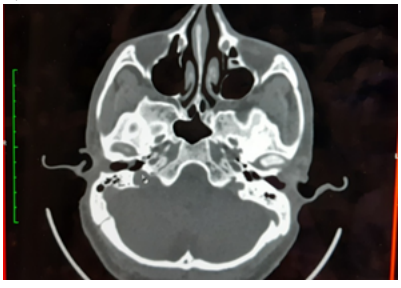


Figure (6): HRCT temporal bone showing high riding of right jugular bulb.

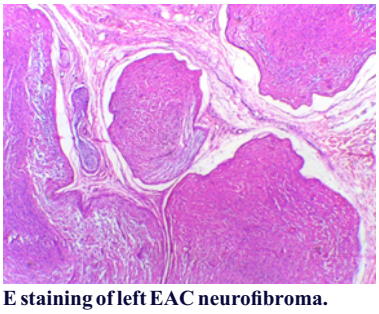


Figure (7): H & E staining of left EAC neurofibroma.

DISCUSSION

Otolaryngologic manifestations of NF1 can be divided into two categories: extracranial and intracranial. Facial bone configuration and size are abnormal in upto 30% of patients. Sphenoid bone and temporal bone dysplasia occurs in 4% of patients.[4] MRI of brain and HRCT temporal bone did not reveal any dyplasia of sphenoid or temporal bone in our patient. Optic glioma is the most common intracranial tumour in NF1 patients, identified on CT scan. Other intracranial tumours, including meningiomas, neurofibromas, and astrocytomas can also occur. All patients with NF1 should have gadolinium enhanced MRI scan of the brain to rule out intracranial tumours.

Extracranial findings include Iris Lisch nodules which are pigmented iris hamartomas occurring in 90% of the affected adults, cafe-au-lait spots in 95% , and plexiform neurofibroma in 17%. Other features like kyphoscoliosis, visceral neurofibromatosis, short stature, precocious puberty, acromegaly, hyperparathyroidism and macroglossia can also be seen in NF1 patient.[1]

Plexifrom neurofibromas can occur anywhere as they originate from peripheral nerves. They can grow in enormous proportions, resulting in damage to surrounding structures.

GENETIC COUNSELLING

Neurofibromatosis type 1 can occur as a result of either spontaneous mutation (50% 0f cases) or as a result of transmission of the autosomal dominant gene from an affected parent. Genetic counselling is necessary because 50% of the descendants will become affected.[4] A child with no family history of neurofibromatosis cafe-au-lait spots alone are insufficient to make diagnosis of NF1. The child must be followed clinically to check for other manifestations of NF1 as the definitive diagnosis can be made with in 5 years of age.[5] The expressivity of this disorder is variable, and patients with same mutation may present with very severe disease or mild symptoms.[1]

SUMMARY

Neurofibromas are non capsular, slow growing benign tumour. As external auditory canal is an uncommon site for neurofibroma, there may be functional impairment, cosmetic problems and a sense of fullness along with mild to moderate conductive hearing loss.

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