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Case Report

Neurofibromatosis with Conductive Hearing Loss - A Rare Presentation

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Abstract

Background: Neurofibromatosis (NF1) is an autosomal dominant, multisystem disorder. Trials are ongoing to discover and test medical treatments for the various manifestations of NF1, primarily plexiform neurofibromas, learning disabilities, and optic pathway gliomas, which are a significant cause of morbidity in these patients. Penetrance approaches 100% by age 20; if the patient has the mutation, he or she will exhibit manifestations, although expressivity is highly variable.

Objective: This case highlights the importance of early diagnosis and management in case of plexiform neurofibromatosis, to rectify conductive hearing loss and for aesthetic corrections.

Methods: Here we report a case of 22 year old female with right side conductive hearing loss with growth present over right external auditory canal.

Result: Computed tomography and histopathology revealed that this is case of plexiform neurofibromatosis which was managed by surgical excision.

Conclusion: By the help of this study, we emphasized the role of early intervention and treatment in case of plexiform neurofibromatosis to control its further growth and spread.

Keywords: Plexiform Neurofibromatosis, Cafe-au-lait Spots; Axillary Freckling; Mustarde's Suture

Introduction

Neurofibromatosis (NF1) is an autosomal dominant, multisystem disorder affecting approximately 1 in 3500 people. The various manifestations of NF1, primarily plexiform neurofibromas, learning disabilities, and optic pathway gliomas. The earliest historical evidence first appeared in the 13th century but it wasn›t until Friedrich Daniel von Recklinghausen published his landmark paper [1]. While no medical therapies are currently available, trials are ongoing to discover and test medical treatments for the various manifestations of NF1, primarily plexiform neurofibromas, learning disabilities, and optic pathway gliomas, which are a significant cause of morbidity in these patients. In 2007, a disorder with cutaneous findings (multiple café-au-lait macules and axillary freckling) similar to NF1 was described [2] although the causative gene (SPRED1) was different.

NF1 is an autosomal dominant disorder, with a nearly even split between spontaneous and inherited mutations. Penetrance approaches 100% by age 20; if the patient has the mutation, he or she will exhibit manifestations, although expressivity is highly variable [3,4]. The NF1 gene is located on chromosome

17q11.2 and encodes for the protein neuro fibroma. This large gene (60 exons and >300 kilobases (kb) of genomic DNA) has one of the highest rates of spontaneous mutations in the entire human genome [5].

Here we report an extremely rare variant of neurofibromatosis type 1 with plexiform neuro fibroma on right Pinna (Figure 2 and 3) over the external auditory canal in a young female patient.

Case Report

A 22 year old female patient presented with complaints of reduced hearing from right ear since 6 years which progressive, unilateral, and growth over right ear since last 5 years. The right ear was also low set i.e., it was at lower level than left ear, which appeared normal. The mass was non tender, non discharging, cystic, approximately 5 cms x 5 cms, temperature normal over the swelling which was mobile and not adherent to underlying structures or skin over it.

Figure 1: Shows "buttonhole sign" of neurofibromas.

Figure 2: Shows extension of neurofibroma in post aural region of right side.

History of incision and removal of mass was given by patient when she was 5 years old, though no scar mark was seen over the swelling. Nose and oral cavity appeared normal. Tympanic membrane of right side could not visualised as the mass was blocking the external auditory canal, but left tympanic membrane appeared normal. There were multiple cutaneous neurofibromas all over her body with café au lait macules (Figure 4) on fore arm and chest with axillary freckling and a plexiform neurofibroma over right ear.

Figure 3: Shows neurofibroma of right, Ear occluding the right EAC.

Figure 4: Shows multiple café-au-lait macule over neck during general examination.

The tympanic membrane along with the external auditory canal of right side in this patient look healthy with cone of light present. Pure tone audiometry was done for the patient which revealed 50 db hearing loss moderate conductive type of right ear. Tympanometry revealed 'B' type curve with loss of stapedial reflex. X-ray skull showed soft tissue swelling over right mastoid and right auricular region. MRI brain revealed multiple well defined lesions in the scalp involving subcutaneous planes likely neurofibromas. Similar intensity irregular lesion in right posterior auricular region infiltrating into right external auditory canal likely infiltrative plexiform neuro fibroma. Overall features of neurofibromatosis type 1 seen. After proper clearance from ophthalmology and medicine departments, the patient was taken up for surgery for Pinna deformity and excision of the plexiform neuro fibroma over right post auricular region and right external auditory canal under general anaesthesia in May 2022.An elliptical incision was taken over the swelling and blunt and sharp dissection was done to see the extension of the mass and it was excised. Excess skin was removed. The same procedure was followed post aural with removal of the mass and obliteration of the cavity was done. Mustarde's (chonco -mastoid) suture was taken up to pull up the Pinna and set it to normal position. Haemostasis was achieved. Pressure dressing was applied over right Pinna, a right mastoid region. Regular dressing was done.

Histopathological examination report stated hypo cellular areas consisting of short spindle shaped cells with slender wavy nuclei with pointed ends and indistinct cytoplasmic borders interspersed with variably sized collagen bundles and mast cells. All places of focal area of serous acini and lymphoid tissue comprising of germinal centre, lymphoid follicles and lymphocytes seen. Findings are consistent with neurofibroma.

Figure 5: Post operative right side post aural suture of excised neurofibroma.

Figure 6: Post operative right side stump of excised neurofibroma after surgery.

Discussion

Plexiform neurofibroma(PNF),a rare variant of neurofibromatosis type 1 (NF-1), which is a subtype of benign nerve sheath tumours, in neurofibromas (NF). It is a result of development of proliferation in all areas of peripheral nervous system and may cause functional damage with deformities and pain along with considerable mortality and morbidity and increased risk of malignant transformation in some case. They are benign, but can be disfiguring, may lead to erosion of adjacent bone or blood vessel, may cause obstruction in the respiratory or GI tract [5] or they can progress to malignant transformation, producing malignant peripheral nerve sheath tumours (MPNSTs), one of the topmost cause of premature death amongst people with NF1 [6]. Currently, surgical intervention is the choice for treatment for PNF patients, which due to invasion by tumor with massive growth, and chance of postoperative recurrence, becomes difficult. Neurofibromatosis 1 was described as a clinical entity during 1882 [6], but accepted diagnostic criteria were unestablished until [7]. The diagnosis of neurofibromatosis 1 is made on the basis of clinical criteria, although the gene responsible was identified 10 years prior [8-10]. The National Institutes of Health (NIH) Diagnostic Criteria was re-evaluated during 1997, and their continued use was recommended with no modification [10]. Clinical experience shows that the National Institutes of Health and Diagnostic Criteria are both highly specific and sensitive in adults having neurofibromatosis 1. These criteria are very specific for children too, but many children later are seen to have neurofibromatosis 1 who do not meet the National Institutes of Health Diagnostic Criteria in the first years of life [11-14]. The diagnosis of neurofibromatosis 1 cannot surely be made with certainty before first year of life in almost 50 per cent of affected children with a negative family history for the same [11].

Conclusion

Patients inflicted with plexiform neurofibromatosis should be subjected to all possible investigations and imaging regarding status of cp angle and its relation to hearing loss in case of such patients. Consent should be taken before surgery for unsatisfactory result and recurrence. Results are good if post operative evaluation with regular dressing is good and neuro fibroma removal is complete including surrounding tissue.

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