International Journal of Otorhinolaryngology and Head and Neck Surgery
Saleh MM et al. Int J Otorhinolaryngol Head Neck Surg. 2019 Jul;5(4):1095-1098
http://www.ijorl.com
pISSN 2454-5929 | eISSN 2454-5937

Case Report

DOI: http://dx.doi.org/10.18203/issn.2454-5929.ijohns20192737

Neurofibroma in the external auditory canal

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Received: 02 March 2019
Revised: 17 May 2019
Accepted: 22 May 2019

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ABSTRACT

Neurofibromas are relatively common tumours of the nervous system, but only few cases involving the external ear have been reported. We are reporting here a case of a 30-year-old male with neurofibroma of the external auditory canal. The primary complaint was cosmetic deformity and mild conductive hearing loss. There was total occlusion of the external auditory canal. The swelling was excised by postauricular approach. Surgery resulted in an superb purposeful and cosmetic outcome. Neurofibromas of the head and neck do not seem to be uncommon, however they seldom have an effect on the external ear and only a few such published reports are available.

Keywords: Neurofibroma, External ear, Benign tumor

INTRODUCTION

Neurofibromatosis, first described by the German pathologist Frederich von Recklinghausen, is a hereditary neurological disorder. Without special predilection for race or sex and with an incidence of 1:2500-3000 live births, neurofibromatosis type 1 (NF1), or peripheral neurofibromatosis, is the most common form of autosomal dominant phakomatoses. Although several cases are inherited, approximately 30–50% arise de novo from spontaneous mutations. The genetic defect has been linked to chromosome 17q11.2.[1, 2]

Neurofibromatosis 2 (NF2), or central neurofibromatosis, has been shown to be related to a gene found in chromosome 22. It is far less common than NF1, with an incidence believed to be about 1 in 50,000 live births. NF2 is often called bilateral acoustic neurofibromatosis because acoustic neuromas, seen in about 90% of those affected, are by far the most common central tumors seen.[3]

Genetics

The NF1 gene has been mapped to chromosome 17q11.2 and cloned. The gene is large, spanning over 350 kb of genomic DNA, and contains 60 exons. Neurofibromin, the protein product encoded by the gene, encompasses 2818 amino acids and has an estimated molecular mass of 327 kDa. Neurofibromin is expressed in several tissues, including brain, kidney, spleen, and thymus. Mutations in the NF1 gene result in loss of functional protein, causing the wide spectrum of clinical findings including NF1-associated tumors.[4-6]

Clinical features of NF1

The diagnosis of NF1 is made on the basis of clinical features requiring the presence of at least two of the following major criteria: 6 or more cafe´-au-lait spots (CALS) >5 mm in diameter in prepubertal and >15 mm in diameter in postpubertal individuals; for each lesion, the longest diameter is measured; axillary or inguinal freckling; two or more cutaneous neurofibromas; one

plexiform neurofibroma; characteristic bony defects; optic glioma; two or more iris Lisch nodules; or a first-degree relative with NF1(parent, sibling, or offspring).[7]

It is a progressive condition with variable complications occurring over the time course of the disease. CALS, pseudarthrosis and externally visible plexiform neurofibromas can generally be identified during infancy.

Freckling, optic gliomas and severe scoliosis occur within the first decade of life. On progression, cutaneous neurofibromas and iris Lisch nodules usually appear during the second decade.[8]

Lisch nodules characteristically are raised, often pigmented hamartomas of the iris and represent a relatively specific finding for NF1. These do not affect vision in any manner.[9]

In a child with a negative family history and no other diagnostic features, CALS alone are insufficient to make a diagnosis of NF1. These children should be followed clinically for the appearance of other manifestations, as a definitive diagnosis usually can be made by the time the child is four years of age. Alternatively, genetic testing can be considered to make a molecular diagnosis.[10]

Skin manifestations of neurofibromatosis type 1

CALS are well circumscribed, uniformly light to dark brown macules with an average size of 2–5 cm in adults. The frequency of five or more CALS in normal individuals is rare and thus an excess of this number provides a significant diagnostic feature for NF1. Diffuse freckling is common in NF1 but clustering hyperpigmentation on the axilla and inguinal area is very unusual, except in NF1.

There are four types of skin neurofibromas : cutaneous, subcutaneous, nodular plexiform, diffuse plexiform. Skin neurofibroma, a benign tumor derived from the cutaneous or subcutaneous nerve sheath, is comprised of Schwann cells, fibroblasts, perineural cells, mast cells, axons and blood vessels. Some erupt on a broad front at the base of skin whereas others form pedunculated lesions.

Cutaneous neurofibromas, the most common type, consist of soft fleshy tumors arising from cells in the peripheral nerve sheath. In general, these dermal lesions begin to appear just before or during adolescence. They tend to increase in size and number with age. They vary in number from just a few to several hundred, with the highest density occurring over the trunk.[11]

Cutaneous neurofibromas are benign and do not carry an inflated risk of developing malignant transformation. However, they often represent a major cosmetic problem in adults. Pruritus associated with accelerated growth of neurofibromas may be a prominent and distressing symptom.

Pregnancy can affect the number and size of neurofibromas, suggesting that these tumors have a hormone-responsive component.[12]

Subcutaneous neurofibromas usually become apparent at the start of adolescence or early adulthood. These lesions present as firm, tender nodules along the course of peripheral nerves.

Nodular plexiform neurofibromas appear as complex clusters along proximal nerve roots and major nerves. They are similar to subcutaneous neurofibromas. These lesions can be a significant challenge in surgical treatment and pain management, especially with progressive growth along the spinal column that can cause vertebral erosion and may result in compression of the spinal cord.[13]

Although diffuse plexiform neurofibromas often are not apparent in infants, they are thought to be congenital lesions. In infants, hyperpigmentation of the skin may overlie an area where a lesion will develop and enlarge steadily with age.

Diffuse-type neurofibroma (DN) presents as a plaque-like elevation of the skin, and often arises in the head and neck region; however, it is uncommon in the external auditory canal. This diffuse type of neurofibroma has ill-defined, infiltrative, cell-rich tumors that occupy the whole dermis and extend into the subcutaneous tissue.[14]

Plexiform neurofibromas (PN) represent a major cause of morbidity and disfigurement in individuals with NF1, and symptomatic PN are associated with increased mortality. These lesions usually involve multiple nerve fascicles, with serpiginous growth and significant vascularity, rendering complete surgical resection extremely difficult, if not impossible, to accomplish.[13,15,16]

Histologically, plexiform neurofibromas are similar to cutaneous neurofibromas except for an increase in the extracellular matrix and greater vascularization. Tumors may be discrete, homogeneous and well circumscribed, or diffuse, heterogeneous and infiltrative. Facial dysmorphisms with visual acuity loss are not uncommon in large facial plexiform neurofibromas.[17]

Skeletal problems

Abnormal skeletal development occurs in 10–20% of NF1 patients. Bony dysplasia, bony erosion, demineralizing osteoporosis, nonossifying fibromas and scoliosis are all features. Fractures and deformity of bones is present in 5–10% of young patients, especially boys.[18-20]

Vascular lesions

NF1 patients exhibit vascular complications that include arterial stenosis, aneurysm and arteriovenous fistulas

involving the abdominal aorta and its branches. We have reported an NF1 family with multifocal stenosis in intracranial arteries with complete occlusion of the left middle cerebral artery causing a fatal brain ischemia.[21]

Neurological disorders in neurofibromatosis type 1

Neurofibromas originating from the spinal roots may cause pain, weakness, muscle atrophy and depressed tendon reflexes. Radiculopathy associated with myelopathy can occur in the dumbbell-shape tumors. Occasionally, images of the multiple root neurofibromas look like a Christmas tree with many hanging gifts (Figure 1). Involvement of the lumbar roots causes back pain that is aggravated by exertion and coughing. There can be massive proliferation of tumors with infiltration of the bladder and compression of the uterus, rectum and ureters.[17]

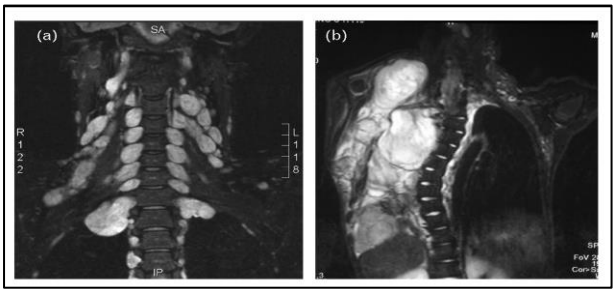

Figure 1: Magnetic resonance image of patients with neurofibromas.[17] (a) An individual with multiple root neurofibromas. (b) thoracic plexiform neurofibromas transforming to malignant peripheral nerve sheath tumors.

CASE REPORT

A 30 year male patient presented to the ENT department with left ear fullness & left mild conductive hearing loss. He was previously misdiagnosed as otitis externa few months ago. There was a soft swelling at the concha extending into cartilaginous part of external auditory canal. The swelling was occludig the external auditory canal totally. The tympanic membrane was not seen from the swelling.

There was no pain. There was no history of bleeding. His vision was normal. He did not have family history of neurofibromatosis. The swelling was excised by postauricular approach under general anaesthesia (Figure 2 and 3). The mass was adherent to the ear canal. The entire mass was removed. A successful cosmetic result was achieved.

Histopathological examination of the mass revealed neurofibroma. It was 2×2 cm in size whitish and soft. Microscopic examination showed a growth made up of proliferating spindle cells arranged in bundles and fascicles with storiform pattern and whorls formation. No atypia was noted.

An informed consent was signed by the patient.

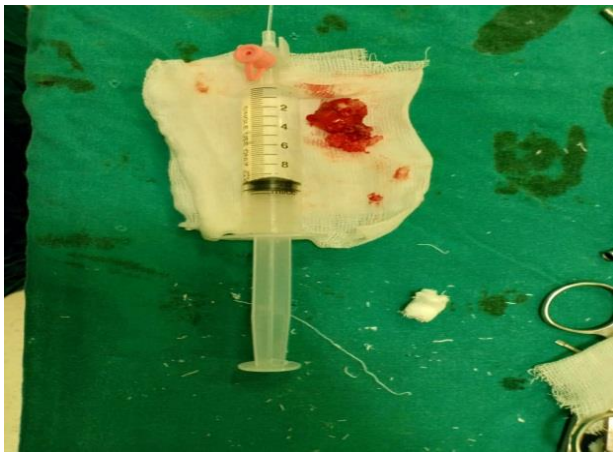

Figure 2: The mass after excision.

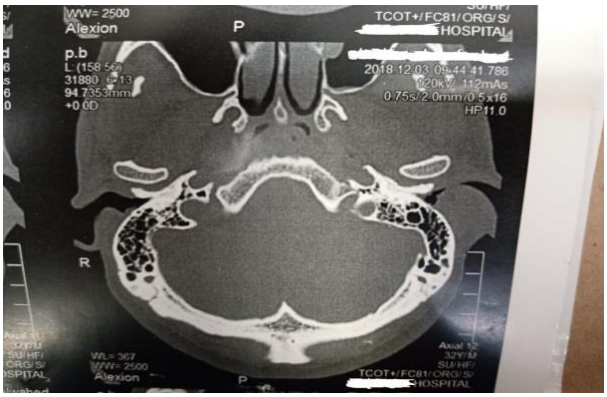

Figure 3: CT petrous temporal bone (axial view) the mass appeared in the cartilagenous portion of left external auditory canal.

DISCUSSION

In our case there was no family history. A single or few neurofibromas alone do not indicate NF; some people who do not have NF could have a neurofibroma. The present case did not have any other features of NF 1.[22]

Indications for surgical removal of neurofibromas include cosmetic disfigurement or compression effects.The indication in this case was both for cosmetic disfigurement and improvement in hearing.

Surgical management is the only viable therapy when a neurofibroma causes disability, intractable pain, or disfigurement.[23]

The surgical management of neurofibromas generally entails a concern: the extent of resection with respect to the probability of recurrence and loss of function.[23]

To minimize the postoperative recurrence rate, it is important to resect the tumor to the greatest extent possible without causing postoperative functional loss.

The most intricate surgical aspect in this patient was the infiltration of the tumors into the dermis and subcutaneous tissue involving the EAC.

CONCLUSION

Neurofibromas are usually benign. Malignant transformation has been reported to occur in 2 to 16% of cases. Neurofibromas of the head and neck don't seem to be uncommon, however they seldom have an effect on the external ear and only a few such published reports are available.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. von Recklinghausen. Neurofibromatosis. N Engl J Med. 1982;306(19):1177-8.
2. Friedman JM. Epidemiology of neurofibromatosis type 1. Am J Hum Genet. 1999;89(1):1-6.
3. Rouleau GA, Wertelecki W, Haines JL, Hobbs WJ, Trofatter JA, Seizinger BR, et al. Genetic linkage of bilateral acoustic neurofibromatosis to a DNA marker on chromosome 22. Nature. 1987;329(6136):246-8.
4. Ledbetter DH, Rich DC, O'Connell P, Leppert M, Carey JC. Precise localization of NF1 to 17q11.2 by balanced translocation. Am J Hum Genet. 1989;44(1):20-4.
5. Feldkamp MM, Gutmann DH, Guha A. Neurofibromatosis type 1: piecing the puzzle together. The Canadian J Neurological Sci Le J Canadien Des Sci Neurologiques. 1998;25(3):181-91.
6. Fain PR, Barker DF, Goldgar DE, Wright E, Nguyen K, Carey J, et al. Genetic analysis of NF1: identification of close flanking markers on chromosome 17. Genomics. 1987;1(4):340-5.
7. National Institutes of Health Consensus Development Conference Statement: neurofibromatosis. Bethesda, Md, USA, July 13-15, 1987. Neurofibromatosis. 1988;1(3):172-8.
8. Tonsgard JH. Clinical manifestations and management of neurofibromatosis type 1. Seminars in Pediatr Neurol. 2006;13(1):2-7.
9. Lewis RA, Riccardi VM. Von Recklinghausen neurofibromatosis. Incidence of iris hamartomata. Ophthalmology. 1981;88(4):348-54.
10. Korf BR. Diagnostic outcome in children with multiple café au lait spots. Pediatrics. 1992;90(6):924-7.
11. Lott IT, Richardson EP, Jr. Neuropathological findings and the biology of neurofibromatosis. Advances in Neurol. 1981;29:23-32.
12. Dugoff L, Sujansky E. Neurofibromatosis type 1 and pregnancy. Am J Hum Genet. 1996;66(1):7-10.
13. Hummel T, Anyane-Yeboa A, Mo J, Towbin A, Weiss B. Response of NF1-related plexiform neurofibroma to high-dose carboplatin. Pediatric blood & cancer. 2011;56(3):488-90.
14. Lustig LR, Jackler RK. Neurofibromatosis type I involving the external auditory canal. Otolaryngol Head Neck Surg. 1996;114(2):299-307.
15. Prada CE, Rangwala FA, Martin LJ, Lovell AM, Saal HM, Schorry EK, et al. Pediatric plexiform neurofibromas: impact on morbidity and mortality in neurofibromatosis type 1. J Pediatrics. 2012;160(3):461-7.
16. Serletis D, Parkin P, Bouffet E, Shroff M, Drake JM, Rutka JT. Massive plexiform neurofibromas in childhood: natural history and management issues. J Neurosurgery. 2007;106(5 Suppl):363-7.
17. Overdiek A, Feifel H, Schaper J, Mayatepek E, Rosenbaum T. Diagnostic delay of NF1 in hemifacial hypertrophy due to plexiform neurofibromas. Brain & development. 2006;28(5):275-80.
18. Riccardi VM, Womack JE, Jacks T. Neurofibromatosis and related tumors. Natural occurrence and animal models. The Am J Pathol. 1994;145(5):994-1000.
19. Crawford AH, Schorry EK. Neurofibromatosis in children: the role of the orthopaedist. J Am Acad Orthop Surg. 1999;7(4):217-30.
20. Gilbert A, Brockman R. Congenital pseudarthrosis of the tibia. Long-term followup of 29 cases treated by microvascular bone transfer. Clinical orthopaedics and related research. 1995;14(3):37-44.
21. Tang SC, Lee MJ, Jeng JS, Yip PK. Novel mutation of neurofibromatosis type 1 in a patient with cerebral vasculopathy and fatal ischemic stroke. J Neurol Sci. 2006;243(1-2):53-5.
22. Merker VL, Murphy TP, Hughes JB, Muzikansky A, Hughes MR, Souter I, et al. Outcomes of preimplantation genetic diagnosis in neurofibromatosis type 1. Fertility and sterility. 2015;103(3):761-8e1.
23. Manolidis S, Higuera S, Boyd V, Hollier LH. Single-stage total and near-total resection of massive pediatric head and neck neurofibromas. J Craniofac Surg. 2006;17(3):506-10.

Cite this article as: Saleh MM, George PE, Mohsen MM, Eldakiky AM, Makarious IG. Neurofibroma in the external auditory canal. Int J Otorhinolaryngol Head Neck Surg 2019;5:1095-8.