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Case Report

A Case of Giant Scalp Neurofibroma

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Abstract

Objective: To report a rare case of giant scalp Neurofibroma in eighteen year male presenting with ulceration of overlying skin with recurrent bleeding.

Case Report: A eighteen year male presented with giant neurofibroma in front temper occipital region with ulceration in occipital region due to pressure sore with difficulty in sleeping in supine position. He had family history of similar lesions in his mother and sister suggestive of hereditary origin. CT head showed calvarial defect in torcular heropili.

Conclusion: Genetic counselling is necessary before getting married and having children because neurofibromatosis is an inherited condition that runs in the autosomal dominant gene family. Decisions regarding indications for and scope of surgery need to be tailored to the tumour extent, location radiologic features, and within the context of the individual patient's overall health.

Keywords: Giant Neurofibroma; Neurofibromatosis; NF1; Plexiform

Introduction

The term "neurofibromatosis" (NF) refers to a set of hereditary conditions that mainly impact the development of brain tissues at the cellular level. Numerous café-au-lait macules (CALMs), skinfold freckling (more accurately referred to as lentiginous macules since they occur in non-sun exposed areas), iris Lisch nodules, tumors of the nervous system, and other characteristics are indicative of neurofibromatosis type 1 (NF1; OMIM 613113) [1]. It is an autosomal dominant condition brought on by a variety of mutations in the NF1 gene, which is found at the 17q11. [2] chromosome. Among human genetic illnesses, it has one of the highest rates of spontaneous mutation. Only 50% of those with NF1 have a confirmed family history of the condition. The remaining patients are examples of unintentional mutations. The disease's expressivity is incredibly variable, with symptoms ranging from minor lesions to a number of consequences and functional impairment. Otherwise, the penetration is 100% [2].

Surgery is the main type of treatment for neurofibromas, but following partial removal of big plexiform neurofibromas, recurrence rates are significant. Due to the numerous pathways involved in the growth of NF1-related tumors, there is no agreement on the best course of action in the case of NF1-related malignancies. Targeted therapy can have a significant effect [3]. Because Ras-GTP expression is increased in neurofibromas, anti-Ras medications are the best option. In phase II clinical trials, plexiform neurofibromas were treated with drugs targeting Ras signaling and other pathways, including tipifarnib, pirfenidone, sirolimus, pegylated interferon alfa-2b, and imatinib. Promising treatment approaches include pharmacologically decreasing kit activity and 41 adhesion as well as targeting downstream effectors of the Ras signaling cascade, such as drugs that block MEK and PI3K. According to a recent clinical research, children with NF1 and inoperable plexiform neurofibromas respond partially to the MEK inhibitor selumetinib [4].

We are presenting here a case of giant scalp neurofibroma without associated skin phacomatiosis which is rare presentation in NF1.

Case Report

A nineteen year boy presented with painless swelling in the scalp since birth which was progressively growing in size. Swelling gradually increased in size to cover the front teporoparietal and occipital region of scalp. Similar skin lesions was seen in his mother and his sister. There was loss of hair in the affected part and followed by ulceration of the involved skin with on and off bleeding from the ulcer. No headache, vomiting, loss of consciousness. Examination revealed 30cm x 26 cm soft irregular swelling with ulceration in occipital region as seen in figure. CT scan head showed large scalp mass with calvarial defect in midline in occipital region which enhanced with contrast. Laboratory investigation were within normal limits.

Surgery was planned under general anaesthesia and intra operative findings was highly vascular yellowish tissues with multiple venous sinuses with erosion of occiput exposing the torcular herophili. Excision of the neurofibroma and primary scalp closure was done after placing sub galeal drains. Postoperative period patient received three units of blood and was discharged on seventh post operative day.

Discussion

A mutation in the NF-1 gene is the root cause of NF-1. Neurofibromin, a cytoplasmic protein that is mostly expressed in neuron, Schwann cells, oligodendrocytes, and leukocytes, is produced by the NF1 gene. It is a multi domain protein that has the power to control a number of cellular functions, including the ERK/MAP kinase cascade, the RAS cyclic AMP pathway, adenylyl cyclase, and cytoskeletal assembly [5].

Beginning in childhood, the illness often gets worse with time. It has been demonstrated that NF1 patients have a 15-year shorter life expectancy than the general population. Death in NF1 individuals under the age of 40 has been strongly linked to malignant tumors and vascular disease [6].

Malignant peripheral nerve sheath tumors are more common in some people who also have greater rates of intellectual disability, developmental delay, dysmorphic facial characteristics, and early cutaneous neurofibromas (MPNSTs). Missense variations in codons are linked to a severe phenotype that includes plexiform neurofibromas, spinal neurofibromas, ocular gliomas, skeletal dysplasia, and malignant transformation [7,8].

The distinctive clinical characteristics would be noticeable in early childhood or adolescence. Ninety percent of cases of NF-1, the most prevalent kind, are associated with several café-au-lait spots and a tendency toward neurofibromas along peripheral nerves. Neurofibromas typically develop later than café au lait discolouration, and their prevalence rises with age. The largest sessile or dome-shaped masses of cutaneous neurofibromas are soft, fleshy pink tumors. Other clinical features include Lisch's nodules (melanocytic pigmented iris hamartomas) and mouth lesions [9].

Involvement of the skeleton is observed in nearly 40% of NF-1 patients. The most prevalent skeletal pathology is scoliosis. There are also numerous neurological disorders, such as auditory nerve tumors, hamartomas of the eye, and central nervous system cancers, macrocephalies, (gliomas, glioblastomas), and mental disability (in 40 percent of cases) [10]. Our case had no such associated lesions however there was calvarial defect in the midline.

In a study, it was found that 66 percent of his NF1 patients had at least one intraoral symptom and 58 percent experienced symptoms in the mandible and maxilla that were seen on panoramic radiographs [11]. In our study, the patient did not have oral any oral lesions.

A multidisciplinary strategy is needed to address the multiorgan incidence of NF1. Since NF1 has no known medical cure, care must focus on preventing and controlling consequences. NF1 neoplasms have a modest (3-5%) probability of malignant development, but they can nevertheless compromise a patient's aesthetic and functional quality of life. Therefore, surgical treatment is not always successful because it is exceedingly challenging to completely remove large and many lesions. When a patient's function is compromised, surgery is recommended. Consideration should be given to the risk, potential complications, and anticipated benefits of such procedures [5]. In this patient, surgery was done as the lesion was cosmetically disfiguring as well as patient was unable to sleep supine due to the lesions and had developed pressure sores.

Patients with NF1 should be informed that the illness is autosomal dominant and that both sexes are equally likely to inherit it. Because of the potential for local problems and the possibility of

malignant transformation, it is crucial to carry out a long-term follow-up. The likelihood of malignant transformation must be taken into account in cases when the neurofibroma's size is increasing quickly and discomfort is present [12].

Conclusion

Genetic counselling is necessary before getting married and having children because neurofibromatosis is an inherited condition that runs in the autosomal dominant gene family. Decisions regarding indications for and scope of surgery need to be tailored to the tumour extent, location radiologic features, and within the context of the individual patient's overall health. Validated preclinical models and meaningful clinical trial designs and outcome measures are available to guide clinical development of novel therapies. The clinical implementation of therapies for NF1 requires careful consideration of multiple factors and should be done with the input of a multidisciplinary team experienced in NF1.

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