Case Report

A Case Report on Neurofibromatosis Associated with Infective Mass Tumor

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ABSTRACT

A rare genetic disorder with many non-cancerous (benign) tumors of the skin (neurofibrom as) and nerves is known as Neurofibromatosis. This disorder includes type 1-neurofibromatosis 1 (NF1), type 2-neurofibromatosis 2 (NF2), and schwannomatosis as complex genetic abnormalities that may affect many different organ systems in the body. When alterations in the gene NF1, located in the 17th chromosome occur, it causes NF1 type disorder. NF1 gene controls the synthesis of a specific protein called neurofibromin that functions as a tumor suppressor. The predisposition to form tumors, often on the nerves, spine, brain, and skin, is a common characteristic of these disorders. But the types and severity of symptoms vary widely among individuals. Here, we present a case of NF-1 with a severe infective tumor lesion on the left hand. A 56-year-old male was admitted to the dermatology department with a chief complaint of a mass lesion on the hand that was painful, severely itchily, and progressively increased in size. Multiple neurofibromas were found on his body that are soft, fleshy tumors arising from a peripheral nerve sheath near the surface of the skin, or on the skin, and he had already been diagnosed in his childhood as neurofibromatosis.

Keywords: Neurofibromatosis type 1, Neurofibromas, Schwannomatosis

Neurofibromatosis type 1 (NF1) is the most frequent single-gene disorder affecting the human nervous system, an estimated prevalence of 2 to 3 cases per 10,000 people. He inherited this disorder autosomally dominant, with an equal sex incidence. The neurocutaneous abnormalities like axillary freckling, cafe-au-lait spots, iris hamartomas (Lisch nodules), and cutaneous neurofibromas were diagnosed in individuals with NF-1 type disorder [1]. Neurofibroma consists of a varied mixture of Schwann's cells, perineurial-like cells, and fibroblasts, which is a benign tumor deriving from peripheral nerve sheathing [2]. NF1 causes mutations in the NF1 gene, the neurofibromin-encoded 60-exon tumor suppressor locus of 17q11.2. Many malignancies of NF1 were observed for heterozygotic loss (LOH) in chromosome 17 (including locus NF1) [3].

NF1 can be evaluated by a clinical examination and by a family history of the patient. The diagnosis of NF1 is based on the diagnostic criteria of the national health institutes (NIH). These criteria typically appear in the following predictable order: café-au-lait macules, axillary freckling, Lisch nodules, and neurofibromas [4]. The distribution of the affected nerve is evident for cutaneous neurofibroma in the skin palpating, tenderness to touch, and tingling. Malignant changes occur rarely, and if removal intended, should consult expert assistance either from a soft tissue tumor/peripheral nerve surgeon, and the removal results occasionally in a neurological deficit [5]. The present study describes a case of an infective mass tumor over the left hand in an NF1 patient.

CASE REPORT

A 56-year-old male had been admitted to the dermatology department, government general hospital, Kadapa, with a chief complaint of mild fever, swelling lesion on the hand which is progressively increasing in size, painful and severe itchy for three months. The patient had a history of multiple raised non-itchy neurofibromas over the hands, followed by similar lesions on the face, legs, and body folds at two years of age. There was no family history of NF. There were no neurological abnormalities, skeletal malformations noted. Multiple cafe-au-lait spots disseminated on his body, several soft, fleshy tumors arose from a peripheral nerve sheath near the skin surface diagnosed in clinical examination. He was diagnosed in his childhood with neurofibromatosis. On cutaneous clinical examination, it revealed a severe infective mass tumor (**Fig. 1**) over the left forearm, the biopsy was not performed. Associated pruritus that was generalized moderate to severe in intensity with no diurnal variation and relieved by treatment with oral anti-histamines and topical calamine lotion. On physical examination, the patient have a high blood pressure of 170/90 mmHg that was newly diagnosed. On laboratory investigation, C-reactive protein values found to be increased 18mg/L (0.3- 10mg/L). The patient was admitted to the

hospital for seven days and treated with medications shown in **table 1**. After seven days the patient was discharged with the following medications Tab. Amoxycillin and Potassium clavulanate 625mg, Oint. Fusidic acid 2%, Tab. Chlorpheniramine Maleate 2mg OD and asked to review for surgery after 1 week

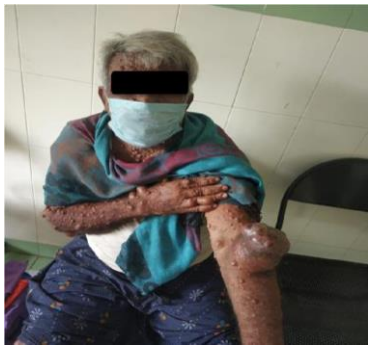

Fig. 1 Neurofibromas associated with infective mass tumor over left forearm

Table 1: Drugs prescribed to the patient

Drug name	Dose	Route	Frequency	Duration
Inj. Amoxycillin and Potassium clavulanate	1.2g	IV	BID	6 d
Inj. Diclofenac Sodium	75mg	IV	BID	4 d
Inj. Pantoprazole	40mg	IV	BID	6 d
Oint. Fusidic acid	2% w/w	Topical	TID	7 d
Tab. Chlorpheniramine Ma	4 mg	Oral	OD	6 d

Note: IV - Intravenous BID-Twice a day; TID-Three times a day; OD-Once a day

DISCUSSION

NF1 is an inherited neurocutaneous condition and characterized by a multi-system tumor with a risk of malignant transformation across the skin in the central nervous system. Neurofibromas are a distinctive feature of the NF1, which are benign tissue-based tumors that occur at the periphery of the nerves of Schwann cells. They comprise fibroblasts, macrophages, and mast cells in addition to neoplastic Schwann cells [6]. 52.5 % of individuals with pruritus had localized in one or more cutaneous neurofibromas. Pruritus pathogenesis is not well known in NF1. It hypothesized that Mast cells and components produced from their degranulation were considered the main source of pruritus, cNF micro-environment was known to have mast cells, which may contribute to tumor initiation, progression, and angiogenesis [7]. Type 1 neurofibromatosis is described as a benign tumor disorder nevertheless, in 2% or 4.2% of patients older than 21 years malignant transformation was reported [8]. There is currently no effective treatment method for people with type 1 neurofibromatosis. One approach is surgical excision, although it is usually impossible to remove all lesions due to excess of NF-1 and the progression of the disease. Surgery is required when suspected of malignancy. Operation indicated if tumors pressure on other organs [9]. In preoperative, perioperative, and postoperative conditions, radiation treatment is used. Adjuvant radiation treatment produced a statistically significant decrease in rates of recurrence of local diseases [10].

CONCLUSION

Benign (harmless) tumors of neurofibromatosis do not need treatment. But subcutaneous tissues are often affected by superficial tumors and may evoke significant infection or cellulitis. The dermatologist, neurologist, and general surgeon should follow these patients routinely to ensure no mass lesions were developing.

Note: Written informed consent was taken from the patient who participated in this study.

REFERENCES

1. North K. Neurofibromatosis type 1. Am J Med Genet. 2000; 97(2):119-127.
2. Nallanchakrava S, Mallela MK, Jeenepalli VS, et al. A rare case report of neurofibromatosis type 1 in a 12-year-old child: A 15-month follow-up. J Oral Maxillofacial Pathol. 2020; 24:106-109.
3. Rasmussen SA, Overman J, Thomson SA, et al. Chromosome 17 loss-of-heterozygosity studies in benign and malignant tumors in neurofibromatosis type 1. *Genes Chromosomes Cancer*. 2000; 28(4):425-431.
4. Bergqvist C, Servy A, Valeyrie-Allanore L, et al. Neurofibromatosis 1 French national guidelines based on an extensive literature review since 1966. Orphanet J Rare Dis. 2020; 15(1):37.
5. Ferner RE, Huson SM, Thomas N, et al. Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. J Med Genet. 2007; 44(2):81-88.
6. Karaconji T, Whist E, Jamieson RV, et al. Neurofibromatosis type 1: review and update on emerging therapies. *Asia Pac J*Ophthalmol. 2019; 8(1):62-72.
7. Ortonne N, Wolkenstein P, Blakeley JO, et al. Cutaneous neurofibromas: current clinical and pathologic issues. Neurology. 2018; 91:5-13.
8. Rallis E, Ragiadakou D. Giant plexiformneurofibroma in a patient with neurofibromatosis type I. Dermatol Online J. 2009; 15(5):7.
9. Fijałkowska M, Antoszewski B. Clinical picture and treatment of cutaneous lesions in patients with neurofibromatosis type 1. Postepy Dermatol Alergol. 2020; 37(5):781-784.
10. Ferner RE, Gutmann DH. International consensus statement on malignant peripheral nerve sheath tumors in neurofibromatosis. Cancer Res. 2002; 62(5):1573-1577.

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