Plexiform Neurofibromatosis with Abnormal Breast Shape (Pachydermatocele): a Case Report

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Abstract

Neurofibromatosis type 1 (NF1), also known as von Recklinghausen'as disease, is the most common type of NF. The hallmarks of NF1 are the multiple café-au-lait macules and associated with cutaneous neurofibromas. The condition is called segmental NF1 when clinical features are limited to one area of the body. We report the case of a 23 years old male patient who presented with leftbreastdisfigurement.Physical examination showed severe disfigurement (fibrosis swelling) over left breast with multiple different size café-au-lait spots. Ophthalmology and MRIbrain showed no evidence of glioma. The diagnosis of plexiform neurofibromatosis based on clinical &histopathological findings.

Keywords: Plexiform, Neurofibromatosis, Disability.

Abbreviations: BRCA1; Breast Cancer 1. NF1; Neurofibromatosis Type 1.MPNST;Malignant Peripheral Nerve Sheath Tumors.NCCN; American National Comprehensive Cancer Network.

Introduction

NF1 is an autosomal dominant genetic disorder with an incidence of approximately 1 in2600to3000Individuals[1,2].

Approximately one-half of the cases are familial (inherited). The remainder are the result of de novo (sporadic) mutations [2]. The de novo mutations occur primarily in paternally derived chromosomes, and the likelihood of de novo NF1 increases with advanced paternal age [3]. The incidence of segmental NF1is estimated at 1 in 36,000 to 40,000[4]**.**

Plexiform neurofibromatosis, also known as pachydermatocele, caused by excessive growth of the neural tissue in the subcutaneous fat.It causes cosmetic as well as functional disability. The optimal management of plexiformneurofibroma is

not well-definedand surgery is often delayed.

Case report

a 23-year-old male patient came with Lt breast disfigurement andcafé-au-lait spots. He had no family history of neurofibromatosis.

General Examination: normal adult male with good health.Locally revealed a mass of 20x14x2.5cm in diameter covering his left breast to the areola, involving the cutaneous and subcutaneous tissues (plexiformneurofibroma).It presented as a subcutaneous mass which feels like a "bag of worms" [Fig 1]. Other two small lesions were also noted in close proximity to his left nipple [Fig 1]and one at the rightthigh[Fig 2]. Ophthalmology examination also done and reveled no glioma or any disease affectsvision. MRI brain was normal.

Ultrasound for both breast and axilla reveal BIRADS I. He underwent an excisional biopsy of the left breast neurofibromatosis [Fig 3]and the defect was closed by rotation flap. Post-operative period passed smoothly [Fig 4]. Patient confirmed the diagnosis on histological grounds.

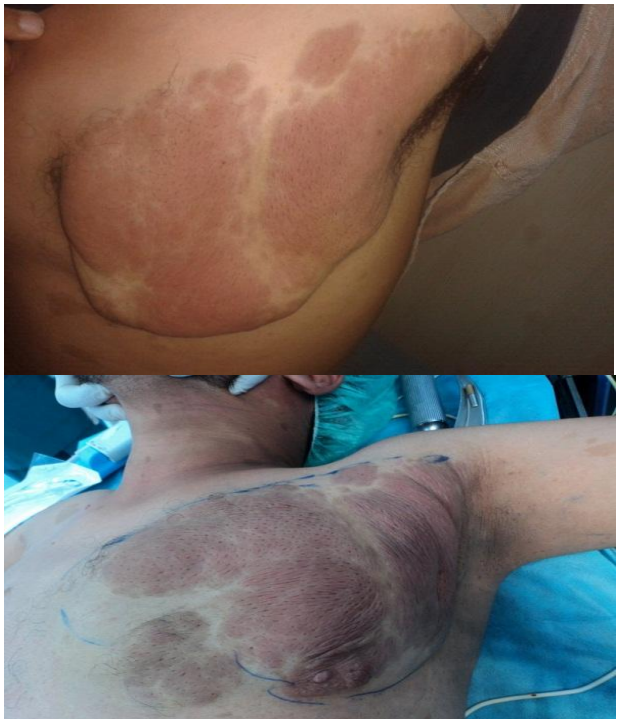

Figure 1: Shows subcutaneous mass which feels like a "bag of worms'' (neurofibromas).

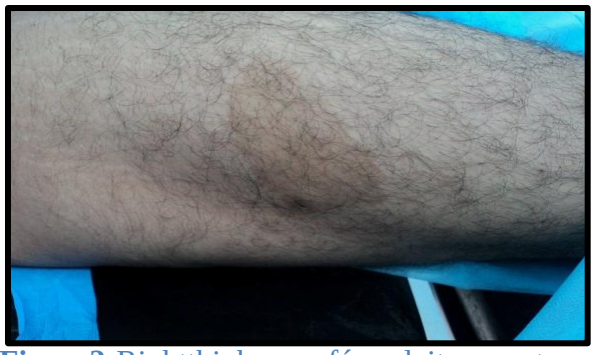

Figure2: Rightthigh café-au-lait spots (small neurofibroma).

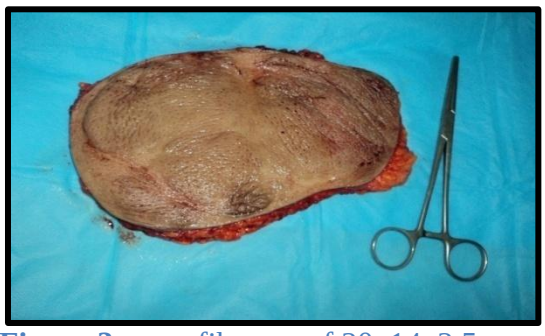

Figure 3:neurofibroma of 20x14x2.5 cm

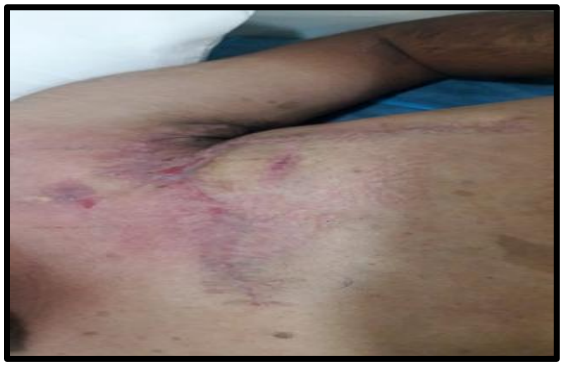

Figure 4: shows post operative healing with no traction.

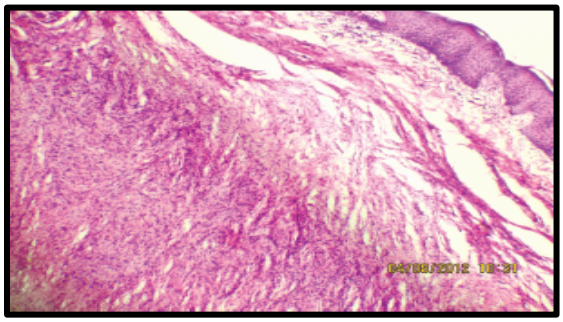

Figure 5: Shows extensive permeation of neuronal cells around fat cells.

Microscopic findings of specimens showedan ill-defined neoplastic proliferation of benign peripheral nerve sheath tumor involving the whole dermis and subcutaneous tissue,characterized by extensive permeation of neuronal cells around fat cells[Fig5] and adnexalskin without destruction. The neoplastic cells displaysmall oval to elongated and wavy nuclei (coma shaped). Additionally,no cellular atypiaappeared.

Discussion

NF-1 is a hamartomatous disorder,caused by a genetic defect localizedon the long arm of chromosome 17 (17q11.27).Plexiformneurofibromafound in up to 26.7% of patients with NF-1 is considered an uncommon skin tumor [5], usually presenting at birth or during the first several years of life. They are non-encapsulated, poorly circumscribed tumors that diffusely infiltrate the nerve and the adjacent fat and muscle. As a result,they are at high risk of malignant transformation. NFare usually unresectabletumors, where tumor resection is impossible without sacrificing the nerve tissue. Fusiform enlargement of multiple nerve fascicles and branches are characteristic. Plexiformneurofibromas contain a mixture of Schwann cells, fibroblasts, reticulin and collagen fibers and a loose mucoid matrix interspersed between the axons of the parent nerve. They typically affect the trunk and extremities, but may also involve the head-neckand bladder [5]. Leading to a variety of problems, including disfigurement and functional impairment.

Plexiformneurofibroma is an uncommon variant of neurofibroma, a benign tumor of peripheral nerves (WHO grade I). The NCCNguidelines on Genetic/Familial high risk assessment in breast cancer screening doesn't include what to do in patients with

NF1. But it recommends early (as early as 18 years of age) and more frequent (twice a year) screening in patients with familial breast cancer disorders [9].

Madinka recommended NF1 women >18 years old to have annual clinical breast examination by a trained practitioner[6]. Though no specific age is recommended, because breast cancer in NF1 occurs at younger age, experts agree on earlier age for screening.

Treatment of neurofibromas is by surgical excision. Care should be taken in placing the incision and an inferior periareolar incision is preferred. Solitary neurofibromasin general are associated with a low local recurrence rate if completely excised [7,8]. In none of the previously reported cases of a neurofibroma in the breast, has recurrence occurred.

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