

## CASE REPORT

# Segmental neurofibromatosis-1 post-chemotherapy in an adult patient

Dana Khoury, M.D, Serena Saade M.D, Mazen Kurban M.D, Ossama Abbas M.D

*American University of Beirut Medical Center, Beirut, Lebanon*

## ABSTRACT

Segmental neurofibromatosis (SNF) represents a rare, atypical phenotypic NF1 variant, diagnosed in the presence of unilateral/localized pigmentary abnormalities and/or neurofibromas, with no systemic involvement and a negative family history of NF. We report an extraordinary case of SNF occurring on the scalp in an adult patient after receiving chemotherapy.

KEYWORDS: Segmental neurofibromatosis, neurofibroma

## INTRODUCTION

Neurofibromas are the most prevalent benign peripheral nerve sheath tumors.<sup>1, 2</sup> Most occur sporadically while 10% are associated with neurofibromatosis (NF), a heterogeneous group of disorders involving the skin, the nervous system or both. Here, we report a peculiar case of segmental NF-1 occurring on the scalp in an adult patient after receiving chemotherapy.

## REPORT OF A CASE

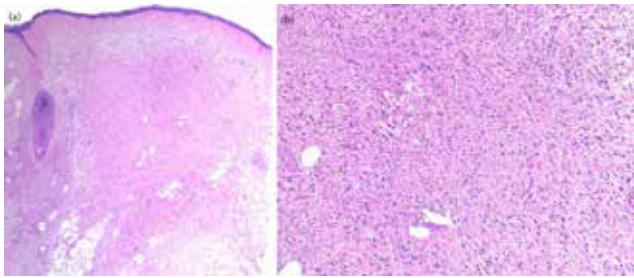
A 53-year-old lady presented with several weeks history of asymptomatic, grouped, skin-colored, soft papules of different size over left parietal scalp, the largest of which measured 0.5 cm (Fig. 1). The lesions were noticed by the patient after she started chemotherapy with Docetaxel and Carboplatin for a new diagnosis of left-sided ER/PR-positive, and HER2-negative breast cancer,



**Fig. 1** Multiple discrete grouped skin-colored papules on the left parietal scalp

status post left total mastectomy. A punch biopsy (Fig. 2) exhibited dermal proliferation of haphazardly arranged uniform, bland spindle cells with wavy nuclei in a myxoid stroma with numerous small blood vessels and scattered mast cells. Based on the clinical presentation, diagnosis of segmental NF1 was made.

**Correspondence:** Dr, Ossama Abbas, Professor, Department of Dermatology, American University of Beirut Medical Center, Riad El Solh/ Beirut 1107 2020, Beirut, Lebanon, P.O.Box 11-0236, Email : [ossamaabbas2003@yahoo.com](mailto:ossamaabbas2003@yahoo.com) Phone : +961-1-350000, ext. 7915, Fax : +961-1-745320



**Fig. 2** Histopathological examination showed dermal bland spindle cells proliferation in a myxoid stroma with numerous small blood vessels. H&E (a) X 40, (b) X 200

## DISCUSSION

Segmental neurofibromatosis (SNF) represents a rare, atypical phenotypic variant of NF1, classified as NF type 5 by Riccardi.<sup>1,2</sup> It is diagnosed in the presence of unilateral and localized pigmentary abnormalities (freckles and/or café-au-lait macules) and/or neurofibromas, with no systemic involvement and a negative family history of NF, as was the case in our patient.<sup>1-3</sup> The most common presentation is neurofibromas alone, usually in a dermatomal distribution with a predilection for cervical segments. SNF can affect all age groups with a slightly greater prevalence in males.<sup>1-4</sup> SNF was then further segregated into four categories: true segmental, localized with deep involvement, hereditary segmental, and bilateral segmental.<sup>3</sup> The cutaneous findings in SNF happen because of postzygotic somatic NF1 gene mutations in primitive neural crest cells, a manifestation of mosaicism, which is why SNF is also referred to as mosaic localized NF1.<sup>1-3</sup> The initial evaluation of patients with SNF is directed at ruling out generalized disease. It is fundamental to counsel patients that the risk of developing disease-related complications is low; however, they should be made aware of the small but existent risk of having a child with generalized NF1. The exact probability of having gonadal involvement seems to be proportionate to the degree of

body surface area affected.<sup>1</sup>

Some authors have put forward the observation that during puberty and pregnancy, neurofibromas tend to increase in dimension, quantity, and malignancy potential in plexiform neurofibromas, thus advocating for a hormonal influence on these tumors. As a matter of fact, progesterone receptors (PR) and G-protein coupled estrogen receptor 1 (GPER-1) are commonly expressed in cutaneous neurofibromas and a high PR expression is associated with an increase in the proliferation index.<sup>5</sup> Extrapolating from this hypothesis, ER/PR-positive breast cancers might also contribute to the growth and development of neurofibromas, which could explain the appearance of these tumors in our case. It is worth mentioning that women with NF1 are at a greatly increased risk of developing breast cancer before 50 years of age, frequently associated with unfavorable tumor markers including HER2.<sup>2</sup>

To our knowledge, this is the first case reporting the sudden onset of segmental neurofibromatosis on the scalp of a patient recently diagnosed with ER/PR-positive breast cancer. It highlights the potential hormonal influence on the development and growth of neurofibromas and the need for dermatologists to consider this differential diagnosis in such situations. Additional studies are needed to further investigate the association between neurofibromas and hormonal receptors, especially in the setting of hormone-sensitive breast cancers.

## REFERENCES

1. Abbas O, Chedraoui A, Kibbi AG, Zayour M, Ghosn S. Multiple asymptomatic skin-coloured papules on the left great and second toes. *Clin Exp Dermatol*. 2009; 34(8):933-34.
2. Ly KI, Blakeley JO. The Diagnosis and Management

- of Neurofibromatosis Type 1. *Med Clin North Am.* 2019; 103(6):1035-54.
3. Adigun CG, Stein J. Segmental neurofibromatosis. *Dermatol Online J.* 2011; 17(10):25.
  4. Poornimambaa M, Puthussery PV, George S, Narayanan B, et al. Segmental neurofibromatosis: An unusual association with ocular, skeletal, and cerebral anomalies. *Ind Dermatol Online J.* 2017; 8(1):65-67.
  5. Rozza-de-Menezes RE, Almeida LM, et al. A Clinicopathologic Study on the Role of Estrogen, Progesterone, and Their Classical and Nonclassical Receptors in Cutaneous Neurofibromas of Individuals With Neurofibromatosis 1. *Am J Clin Pathol.* 2021; 155(5):738-47.