

A case of 25-year-old giant neurofibromatosis

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ABSTRACT

In this paper, the authors presented a case report of a 39-year-old man taken to the Emergency Unit with a 25-year-old giant neurofibromatosis. Moreover, the treatment applied for this case and the physiopathology of the neurofibromatosis disease are also discussed.

Key words:

Giant neurofibromatosis; physiopathology; neurofibromas

INTRODUCTION

Neurofibromatosis is a neurocutaneous condition that can involve almost any organ system. Thus, the signs and symptoms presented may vary widely. Two major subtypes exist: neurofibromatosis 1 (NF-1), which is the most common subtype, referred to as peripheral NF, and neurofibromatosis 2 (NF-2), which is referred to as central NF^[1,2]

We report a case of a giant neurofibroma kept for 25 years without treatment.

CASE REPORT

A 39-year-old male patient arrived at the Emergency Department with a history of bleeding from a moll on his back for the last 3 days. The patient also sustained episodes of nausea with no associated vomit, weakness and sleepiness.

At examination, we found a giant mass on his sacococigeal region [Figures 1 and 2] with small areas of scarification with punctiform mild bleeding. Patient was mildly dispneic with taquicardia associated. No other symptoms were found.

According to the patient, the mass started to develop at the age of 14 years and he never felt compelled to have that examined by doctor. In fact, he regarded this as a phenomenon rendering him different from others.

Blood tests showed severe anemia (hemoglobin 6.9 g/dL). Computed tomography scan of thoracic and abdominal areas didn't reveal any tumors or abnormalities with the patient.

The patient was diagnosed with neurofibromatosis, the anemia was corrected with blood transfusion and the patient was taken to the operating room.

Through a direct elipse incision the tumor was dissected, appearing to be limited to the epidermic region [Figure 3].

While incising its base, we were not able detect any infiltration on the subjacent subcutaneous tissues. The tumor was completely removed and the surgery was completed with a simple closure of the lateral skin flaps [Figure 4]. The tumor weight was 5,276 g [Figure 5]. The patient was

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Figure 1: Lateral aspect of the tumor



Figure 3: Dissection of the tumor showing limitation to the epidermis



Figure 2: Posterior view of the tumor demonstrating much narrower base that allowed direct excision



Figure 4: Final aspect of the mass after removal with over 5 kg total weight

discharged 48 h later after an uneventful postoperative recovery. Histopathological analysis confirmed diagnosis of benign neurofibroma.

DISCUSSION

Neurofibromas are tumors composed of schwann cells, fibroblasts, mast cells, and vascular components. They can be found in nerves, independently of the location in its surface. Moreover, many authors consider neurofibromas the commonest benign tumors of the NF-1.^[3] Neurofibromas can be presented in three subtypes: cutaneous, subcutaneous,

and plexiform. Cutaneous lesions and subcutaneous lesions are circumscribed, however none of them have compatible features with NF-1.^[4]

During physical examination, neurofibromas may present a pathognomonic buttonhole invagination when pressed with a finger. However, their morphology is not always consistent, resulting in lesions either soft or firm to the touch. Plexiform neurofibromas are noncircumscribed, thick and irregular. This specific subtype for NF-1 can be associated with aesthetic deformation by entwining important supportive structures.^[5]

One of the main physical signs encountered along with neurofibromatosis diagnosis is the presence of café au lait spots. These rather unfamiliar pigmented patterns are brown macules irregularly shaped and evenly pigmented. Usually, when neurofibromatosis is diagnosed, subjects have 6 or more spots that are 1.5 cm or greater in diameter. However, in young children, 5 or more café au lait macules greater than 0.5 cm in diameter are indicative of neurofibromatosis and for which investigation is required. Less than 1% of healthy children have 3 or more of such spots, although 1 or 2 café au lait macules are commonly encountered in healthy individuals without disease. Due to the large volume of the mass presented in this case, it was difficult to determine if the alterations on the skin colour were café au lait spots or



Figure 5: Final aspect of the wound closed

decurrent from years of scarification under clothes.^[6]

Neurofibromatosis is a dominant autosomal neurogenetic disorder that tends to change and develop with time.^[7] Many authors demonstrated the development of this disease with increment of the nerve growth stimulation.^[8,9] NF-1 is a pathology that can be associated with different phenotypical manifestations where a group of patients often express cutaneous findings as first symptoms, whilst others might develop life-threatening or severely disfiguring complications. Moreover, amongst individuals from the same family, neurofibromatosis presents itself in different degrees of severity and incidence. The spontaneous mutation rate is 100 times greater than many other genes, and it is considered to contribute to approximately 30-50% of neurofibromatosis cases, however many different mutations in the neurofibromatosis gene have been described.^[10]

NF-1 is linked to a large gene on band 17q11.2. It encodes a protein called neurofibromin, which has a guanosine triphosphatase region that binds to Ras and positively modulates conversion of guanosine triphosphate to guanosine diphosphate. Several studies confirmed the negative regulation of Ras by this protein, allowing authors to

infer that the neurofibromin may act as a tumor suppressor.^[11]

Many individuals with this pathology are known to present below average intelligence and some types of learning disabilities, which may comprise neuromotor dysfunction, attention deficit hyperactivity disorder and deficits in visual-spatial processing.

It was also found that 25-40% of individuals with NF-1 might present learning disabilities and up to 5-10% have mental retardation.^[4]

It should also be noted that when the lesion has a benign aspect and there are no other signs of malignancy, the excisional biopsy is the treatment of choice and it is normally curative.

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Conflict of interest

There are no conflicts of interest.

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