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Neurofibromatosis Type 1 (NF1) or Von Recklinghausen Disease. Filing a case

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Abstract

Introduction*: Von Recklinghausen`s disease or Type I Neurofibromatosis is an autosomal dominant disease that presents a varied clinical expression, with manifestation ranging from café-au-lait spots on the skin to severe aesthetic and functional complication affecting bone and nerve tissues. Objective: We present clinical case treated in Gaborone, Republic of Botswana by a multidisciplinary team made up of a plastic surgeon, neurosurgeon and pathologist from the Republic of Cuba. Result: The patient showed neurofibromatosis skin lesions and family history of the disease, as well as bone changes due to an exaggerated syphosis. No damage to the intellect. Surgery was performed for the exacerbated back injury, obtaining an acceptable cosmetic result and no malignancy of the injury was reported.*

Keywords*: neurofibromatosis, Von Recklinghausen disease, neurofibroma.*

Introduction

Neurofibromatosis is a genetic disease of the nervous system that affects the development and growth of neural cell tissues. There are two variants called type 1 and type 2 characterized by a mutation on chromosomes 17 and 22 respectively. These conditions cause tumors in the nerves, as well as skin changes and bone deformities. Type I NF, or Von Recklinghausen disease, accounts for 95% of all cases. It presents with a broad spectrum of phenotypic expression and unpredictable evolution.

Type I NF was initially called peripheral neurofibromatosis because some of the symptoms, such as skin spots and tumors, appeared to be limited to the peripheral nervous system. This name is not correct, because tumors of the central nervous system can also occur. This growth is caused by the mutation in a "suppressor" tumor growth gene in 17q11.2 encoding neurofibromin: a protein that acts as a tumor suppressor under normal conditions that regulates another cellular protein that stimulates cell growth and proliferation. The characteristic lesion, neurofibroma, originate from Schwann cells and fibroblasts from peripheral nerve sheakhs.

It begins in childhood and in most patients the clinical picture is complete by the age of 5, worsening during puberty, pregnancy, and menopause. 1

Von Recklinghausen's disease or Neurofibromatosis Type I is an autosomal dominant disease that presents a varied clinical expression, with manifestations ranging from café-latte-like spots on the skin to severe aesthetic and functional complications affecting bone and nervous tissues. 2

NF types 1 and 2 occur in all racial groups. NF 1 occurs in 1 in 3,000 births, and NF 2 in 1 in 40,000. Mortality from NF 2 is higher than from NF 1. [1]

Below is a clinical case treated in Gaborone, Republic of Botswana by a multidisciplinary team consisting of a plastic surgeon, neurosurgeon, and pathologist from the Republic of Cuba. For its clinical interest due to the size of its lesions is described below.

Presentation of the Case

Female patient of 18 years of age, African black race, nullipara. With a history of manifesting the disease from a very young age, although it does not specify the age. Personal pathological history: Since childhood presents nodular lesions of variable size, diffusely located on the face, neck, trunk, back and extremities.

Family pathological history: her mother and brother also suffer from it. On physical examination

Disseminated dermatosis is observed on the face, trunk, upper and lower limbs, armpits and inguinal regions. In addition, multiple skin tumors multiple dedicated pink nodules, with a vascularized base ranging from 5 millimeters to a giant plexiform tumor, in the left dorsal region, 15 cm x 11 wide x 3 cm thick.

Associated with this on examination of your SOMA is a deformity of the spine with the excessive pronunciation of syphosis. No harm was found at any other level. The patient was kept studying with a seemingly normal coefficient. He even kept a job.

The radiological study showed: congenital scholastic deformity with multiple neurofibromatosis lesions at different levels of the dorsal rachis without medullary involvement.

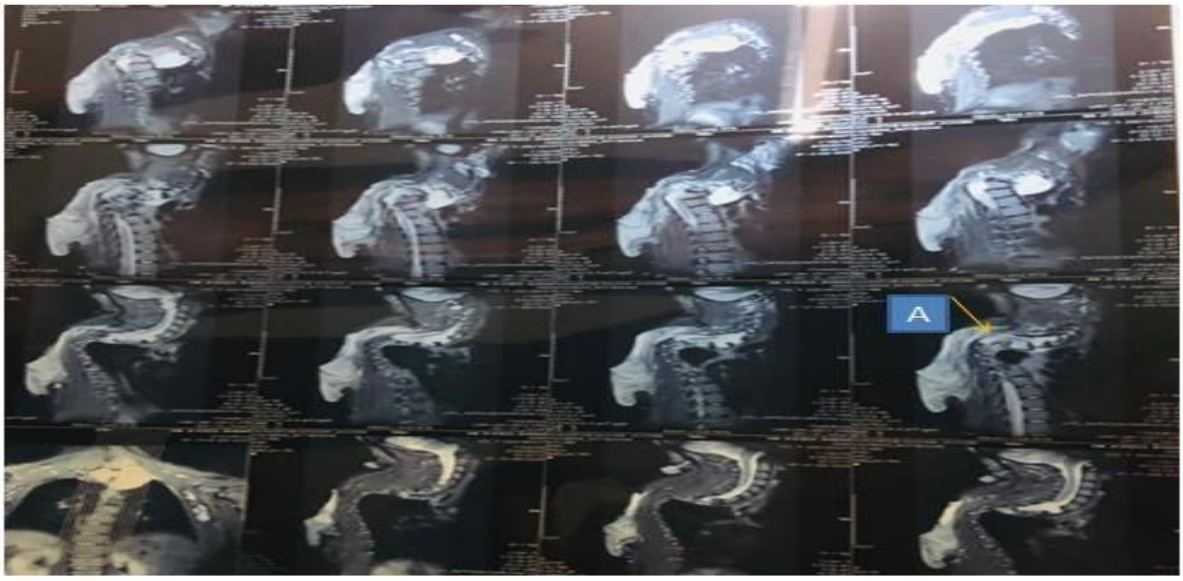

Figure 1. Radiological study of spine. Note scholastic deformity and neurofibromatose lesions.

Figure 1. Radiological study of vertical column. Note scholastic deformity A.

Surgical removal: The lesion was removed with a wide elliptical lesion, up to the fascia in depth, without penetrating the paravertebral muscles. The edges were faced prior decolado of the same and sutured by planes with suture vikryl 2.0, we left drainage. See Figure 2.

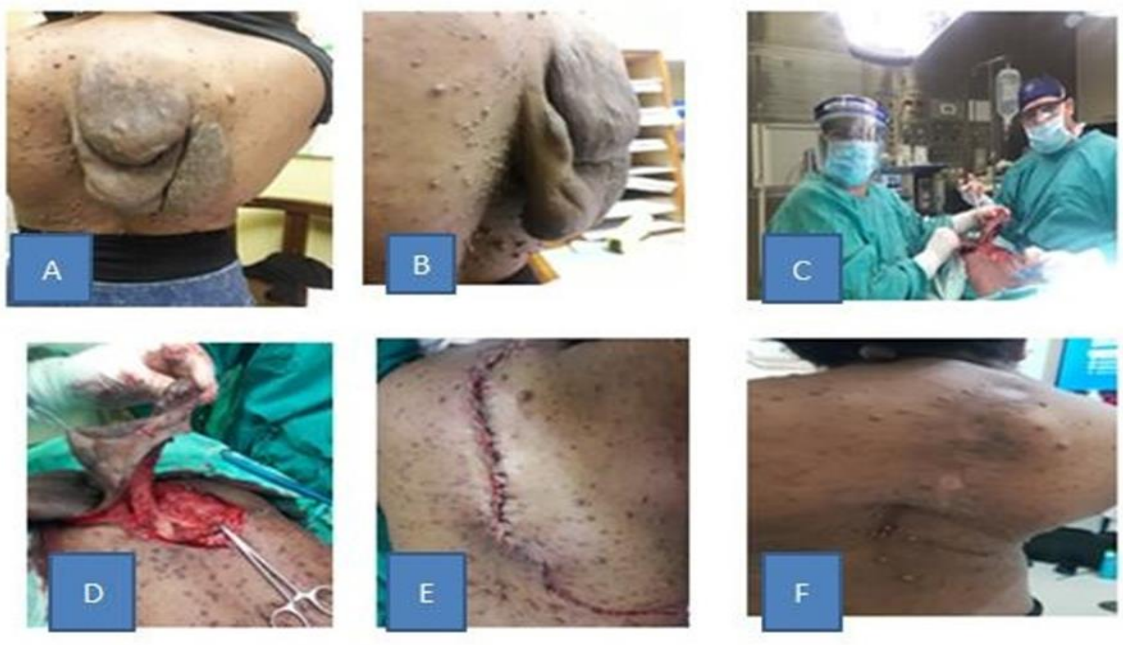

Figure 2. A and B. Preoperative back and profile. C and D. Transoperative. E Immediate postoperative. F. Postoperative per year.

Anatomohistopathological findings: see Figure 3

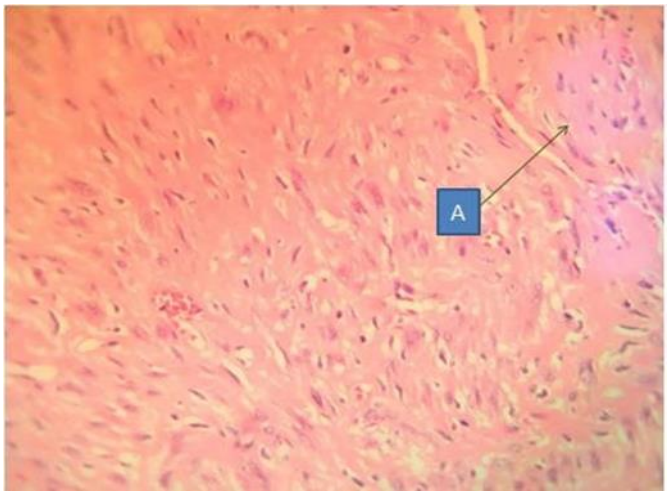

Figure 3. Histopathological picture. A. Ftumor ormations. Staining with hematoxylin and eosin.

Skin biopsy and paravertebral TCS.

Macroscopic description: We receive mass of tissue formed by skin and TCS that measures 15x11x3cm of grayish brown color with multiple nodular lesions whose diameters range between

0.8 mm and 10 mm, to the cut of whitish gray color, fibrous appearance, hard consistency, some extend to the deep dermis. Surgical margins colored orange.

Microscopic description: The skin biopsy and CT examined shows multiple well-delimited, hypocellular tumor formations formed by intertwined fascicles of fusiform cells with wavy nuclei whose background shows varying degrees of collagen and minimal cellular pleomorphism.

Immunohistochemistry S100 is indicated to be positive.

Diagnosis: Diffuse neurofibromas associated with neurofibromatosis type 1 (Disease ofVon Recklinghausen).

Discussion

To diagnose a patient affected by NF1 from the clinical point of view must meet diagnostic criteria and it is stipulated that they must meet two or more of the following: [2.3]

1.Six or more spots "milkcafé" larger than 5 mm in diameter in people before puberty and more than 15 mm if measured after puberty.

2.Two or more neurofibromas of any type.

3.Freckles in the armpits and/or in English.

4.Gliomas in optical pathways.

5.Two or more Lisch nodules (benign hamartomas of the iris).

6.A characteristic bone lesion (such as scoliosis).

7.Affected first-degree relative of NF1 (parent or sibling).

The patient was diagnosed as Neurofibromatosis Type I or Von Recklinghausen's disease because she met three of the diagnostic criteria for it:

Presence of nodular lesions of more than 5mm with characteristics for the disease, Figure 2, the history of two members of the first-line family affected, with maternal inheritance in addition to the characteristic bone deformity such as scoliosis described in the Rx report, Figure 1, and verifiable in the pre- and postoperative photos. Figure 2.

NF 1 is an autosomal dominant disease with 100% penetrance and variable expressivity. It has been found that the expressiveness of this disease is more severe if it is inherited maternally. [4]

It has been reported absence of dominant inheritance in certain families with NF 1, explainable by the lack of diagnosis in a given relative, by not being biological child of the parents socially or familially attributed to the index subject, or by de novo mutations. [4]

It is not possible to predict the severity with which the disease will occur, even among affected members of the same family. [2]

No harm was found at any other level. The patient was kept studying with a seemingly normal coefficient. He even maintained a job, however, other authors have reported that among the most frequent complications are growth problems, learning difficulties, precocity or delayed puberty, hypertension, increased head circumference and tumors. [2.3]

Regarding the surgical treatment, which was performed for aesthetic purposes, we can state that this objective was achieved, see Figure 2, since as other authors comment currently there is no curative treatment for neurofibromatosis, there are only therapies that can improve some of the symptoms.

Surgical treatment is indicated for the resection of neurofibromatose lesions when they interfere with function and aesthetics, or when there are obvious possibilities of malignancy. [2.3]

The treatment of neurofibromatosis is symptomatic as it is a disease of a progressive nature. There is no specific treatment, this being focused on the prevention or treatment of complications. According to the literature, clinical follow-ups and surgical intervention should be performed when the lesions compromise aesthetics or function. 3.5 or when there is an obvious possibility of malignancy. [3]

The removed sample was biopsied without signs of malignancy and if a typical histological picture corroborated the diagnosis since it coincided with the description made by other authors.

Tumors that have grown rapidly should be studied with biopsy and removed immediately, since they can become malignant, this being one of the complications of NF1. If there are no complications, the life expectancy of people with NF is almost normal. [6]

In this sense, there is literature that supports that the belief that the removal of a neurofibroma leads to its malignancy is mistaken and in cases of neurofibromas, surgical treatment can be used to improve the quality of life. [6]

The removed sample was biopsied without signs of malignancy and if a typical histological picture corroborated the diagnosis since it coincided with the description made by other authors. [7]

The neurofibromas characteristic of NF1 have histological variations such as: benign intradermal neurofibromas, which are the most frequent, approximately 95% of cases; nodular neurofibromas, which affect the peripheral nerves and do not infiltrate the surrounding tissues, although they can generate a phenomenon of compression due to their large size; and, finally, plexiform neurofibromas, which are usually congenital and occur in about one third of cases. [7]

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