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Neurofibromatosis Type I Plexiform Geante of the Buttock in a Surgical Setting in a Second Referral Hospital

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Abstract **Case Report**

Neurofibromatosis type I (NFI) or Von Reccklinghausen disease is a rare pathology in the surgical setting. We report the observation of a giant plexiform neurofibromatosis type I (NFI) of the buttock in a 61-year-old patient. The pathology was responsible for functional discomfort, unaesthetic and a problem of personal hygiene (after the stool). The she, unaesthetic and a problem of body hygiene (after the stools). The diagnosis of neurofibromatosis was confirmed according to the diagnostic criteria of the National Institue of Health (NIH) consensus conference. Surgical excision was performed, the anatomo-pathological examination of the surgical specimen was a benign tumor, the long-term evolution was satisfactory without local recurrence and a resumption of the activities of daily life.

Keywords: Neurofibromatosis NFI, plexiform, Size, Surgery.

INTRODUCTION

The neurofibromatoses cover very distinct entities, having in common only certain cutaneous signs due to a common embryogenesis.

The term includes at least two different diseases with autosomal dominant transmission, neurofibromatosis type I (NFI) or Von Recklinghausen disease and neurofibromatosis typeII(NFII)[1,2].

Neurofibromatosis type I, is the most frequent autosomal genodermatosis, this phacomatosis is due to germline mutation of the tumor suppressor gene [2]. The gene is located on the long arm of chromosome 17 (17q11.2) and causes damage to several organs predisposing to the development of benign but also malignant tumors [3]. Approximately 50% of neurofibromatoses are due to sporadic mutations and the others are familial forms[4]. Its incidence is estimated at one birth in 3000 to 4000 individuals [5]. The penetrance of the NF1 gene is almost 100% at the age of 8 years and de novo mutations represent about half of the cases [1]. The expressiveness of the disease is variable from one patient to another, even within an affected family, and in the same individual according to his age [6, 7]. NF1 presents clinical manifestations including nervous tissue, bone and soft tissue abnormalities [8]. The different cutaneous manifestations are, cafe au lait lentiginous spots, cutaneous neurofibromas, plexiform neurofibromas[9]. Plexiform NFI exists in two forms, the diffuse form or "royal tumor" are skin and subcutaneous swellings that can reach several centimeters in size, with a soft and irregular consistency and functional impairment of the use of a limb that may be hypertrophied as a whole or in segments [10, 11]; the nodular form, which may be superficial or deep and responsible for compression as well as growth abnormalities and/or bone osteolysis with a non-negligible risk of degeneration [11, 12].

OBSERVATION

We report the case of a 61-year-old female patient, housewife, referred by dermatologists to the general surgery department of the regional hospital of kayes for giant plexiform neurofibromatosis type I of the buttock (Cf. fig 1). This patient had a family history of neurofibromatosis (her mother) and no other medical or surgical history.

Since the age of sixteen she had skin nodules of variable size on the trunk and face, which then generalized to the whole body.

The skin nodule on the left buttock progressively increased in size to its present size. The presence of this mass was responsible for discomfort when walking, preventing daily activities of life and a problem of personal hygiene (especially after bowel movements).

On physical examination, the mass presented two hemorrhagic ulcerations on contact, it extended from the buttock to the anal margin without reaching the vulva, the presence of a Lisch nodule on ophthalmological examination, the café au lait spots were rather dark brown on the body, there was no cardiopulmonary pathology, a deformation of the dorsal spine in the form of kyphosis linked to neurofibromatosis NFI. She had a hemoglobin level of 9.6 g/dl, which required blood transfusion during the operation.

The patient was operated on under general anaesthesia, and we performed the removal of the mass (see Fig. 2). The postoperative course was simple with no sensory or motor disturbance of the left lower limb and no anal sphincter disturbance. She was discharged from hospital after 7 days of hospitalization. Fifteen days after the operation the wound was practically healed (Cf. fig 3).

Anatomopathological examination confirmed the diagnosis of neurofibromatosis type I with healthy excisional margins without malignant cells.

The long-term evolution was satisfactory with no local recurrence, resumption of daily activities and good personal hygiene.

Fig-1: Presentation of the pathology before surgery

Fig-2: Surgical resection

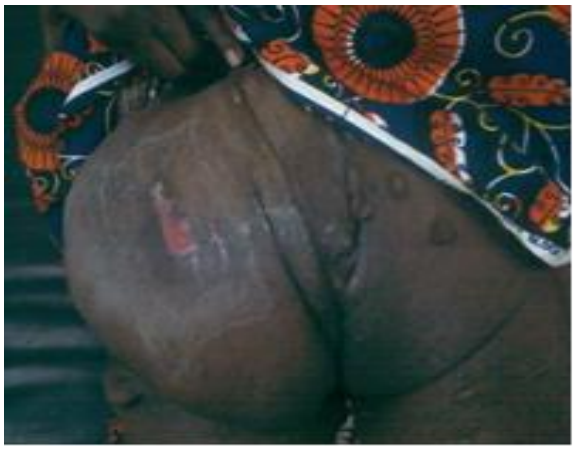

Fig-3: 15 days after surgery

DISCUSSION

We report the case of a giant plexiform neurofibromatosis type I of the buttock with a satisfactory long-term evolution without local recurrence.

This pathology is not frequent in the surgical setting except for a complication related to the size of the tumor or its malignant transformation.

Neurofibromatosis type I has been described by other authors [13-15], in particular the plexiform form. Our clinical case met the diagnostic criteria of the NFI of the consensus conference of the National Institute of Health (NIH)[16]. The age of onset of the first cutaneous signs of IFN at 16 years in our study is in accordance with the literature where the penetrance of the gene is almost 100% at the age of 8 years[1] .The cutaneous form is the most frequent in the literature, accounting for 95% of cases in adults[10]. Spinal involvement is 10 to 30% in the literature [17, 18].

CONCLUSION

Neurofibromatosis type I is an infrequent pathology in the surgical setting, surgical intervention is performed in case of malignant degeneration, signs of compression (organs, vessels, nerves), functional or aesthetic discomfort, the place of surgery is not negligible in the management of certain forms of NFI because it considerably improves the quality of life of patients.

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