Case study

Plexiform neurofibroma of arm: a case report

Abstract

Plexiform neurofibroma (PNF) is a rare benign nerve tumor. It is usually considered pathognomonic of neurofibromatosis type 1 (NF1 or Von Recklinghausen disease). MRI is the examination of choice in the exploration of this pathology. It provides decisive arguments for the positive diagnosis, the evaluation of the prognosis and the follow-up of the lesions. Histological confirmation is sometimes necessary, especially in cases other than NF1. We report the observation of a 22-year-old woman who consulted for a mass of the left arm.

Key words: Neurofibromatosis, plexiform, MRI, anatomy pathology

Introduction

Neurofibromas are benign tumors that develop from the roots and plexuses of spinal nerves. They can be uni or bilateral, sometimes stepped, superficial or deep. There are 3 types of neurofibromas: plexiform neurofibromas (nodular form - diffuse form), cutaneous and nodular. Plexiform neurofibromas correspond morphologically to a more or less long segment of tortuous dilatation of a nerve and its branches, producing the appearance of a bundle of string. Plexiform neurofibromas are pathognomonic of NF1. They are generally slow growing tumors [1]. Their symptomatology is variable depending on their topography. We report the case of a 22-year-old woman who consulted for a mass of the left arm.

CASE REPORT

She was a 22-year-old woman with a father suffering from neurofibromatosis type I. Since childhood, she presented with a mass in the upper 1/3 of the anterolateral aspect of the left arm, which was painless, soft, and gradually increasing in size. For some time, the patient reported a sensation of heaviness with paresthesia of the left upper limb. Clinical examination revealed a mass on the anterolateral aspect of the arm with a soft, painless consistency, well limited and without inflammatory signs, measuring 10 cm in length (figure 1). Skin examination revealed the presence of multiple café au lait spots on the trunk and limbs (figure 2). In view of this very suggestive clinical picture and the patient's past history, the diagnosis of neurofibromatosis type I was evoked. An MRI of the left arm was performed in order to characterize the mass described above. It revealed the presence of an oblong tissue mass in the subcutaneous soft tissues of the upper-external aspect of the left arm, with regular contours, in T1 hypersignal (Figure 3) and T2 heterogeneous hypersignal (Figure 4), discreetly enhanced after injection of gadolinium (Figure 5), measuring 68x15mm extended over 114mm, respecting the muscular and vascular

structures and the bone opposite. The diagnosis of plexiform neurofibroma of the left arm was retained.

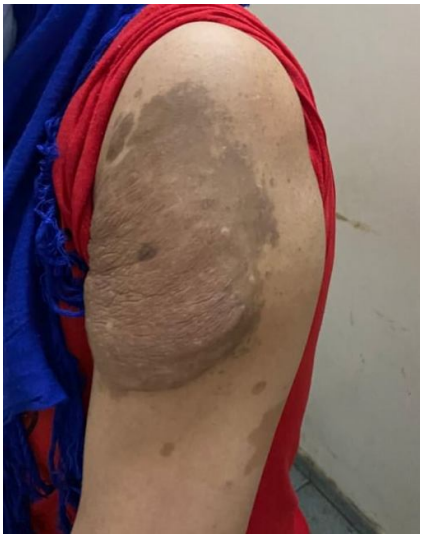

Figure 1: Profile view of the mass on the left arm.

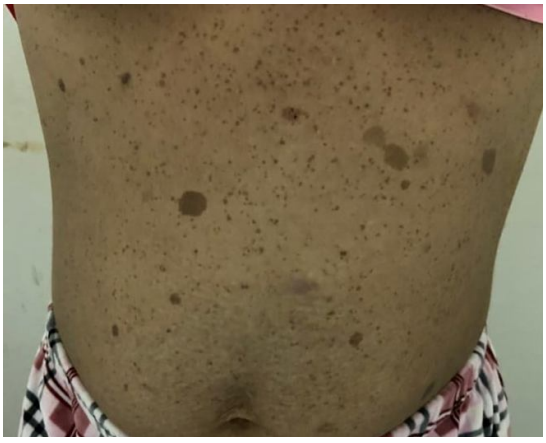

Figure 2: Café au lait stains on the trunk.

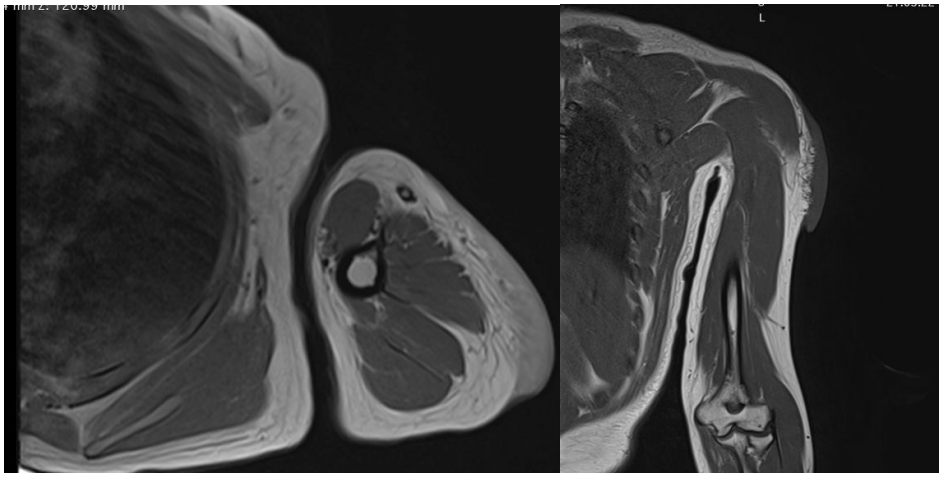

Figure 3: MRI of the left arm, axial and coronal section in T1-weighted sequence without injection of gadolinium: lesion process of the superior-external aspect of the left arm well limited, with regular contours in T1 hypo signal.

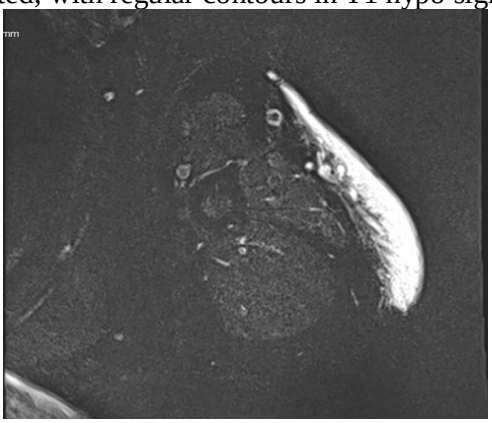

Figure 4: MRI of the left arm, axial section in T2-weighted FSTSE sequence: lesion process of the superior-external aspect of the arm with regular T2 hypersignal contours.

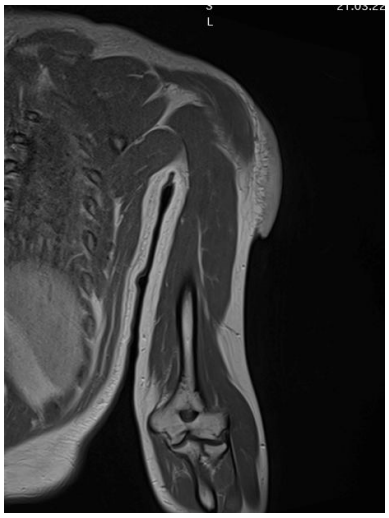

Figure 5: MRI of the left arm, coronal section in T1-weighted sequence after gadolinium injection: the mass is discreetly enhanced after gadolinium injection.

Discussion

Plexiform neurofibromas are pathognomonic of NF1, affecting between 27 and 40% of NF1 subjects. NFP belongs to the four types of neurofibromas encountered in NF1 according to the 1988 National Institute of Health Development consensus conference classification. The diagnosis of the disease is based on the presence of at least two of the following criteria: At least six café au lait spots larger than 5mm before puberty and larger than 15mm after puberty; Two or more neurofibromas, one or more plexiform neuromas; Lentiginous spots of the axillary or inguinal region; Two or more iris hamartomas (Lisch nodules),one optic tract glioma; A characteristic bone lesion (pseudarthrosis of a long bone, spheno-orbital dysplasia, cervical kyphosis). Isolated plexiform neurofibromas may be observed outside the context of NF1, the diagnosis of which must remain a diagnosis of elimination. It would appear that some of these cases are part of a segmental neurofibromatosis (NF5) [2,3]. The NF1 gene responsible for the disease is located on chromosome 17 at locus 17q11.2 that codes for the protein neurofibromin. The frequency of neomutations is particularly high and almost half of the cases are sporadic. NF1 is characterized by a wide variability of clinical expressions, even

within a given family. Majority of patients can be diagnosed only after thorough physical examination [2,3]. They are recognized as congenital, slow growing and often remain asymptomatic. NPs can develop from superficial or deep nerve plexuses, essentially in the trunk and extremities, all spinal segments are affected. They are most often multiple, multi-radicular and tend to expand along the nerve sheaths. Often asymptomatic (40-60% of cases), they may reveal themselves by a varied clinical symptomatology, pain and/or radicular irritation, paresthesias, sensory-motor deficits, and even signs of spinal cord compression, which may vary according to the affected plexus or nerve trunk [4]. Clinically, they present as diffuse, patchy, bumpy, irregular tumors, covered with skin that is sometimes atrophic, flabby, wrinkled, and of the color of normal skin or sometimes bluish and pigmented. Their palpation is usually painless and gives a sensation of "bundle of strings" and inside the soft tumor, hard fibrous cords can be found on palpation [4,5].

Imaging is of great help in the diagnosis, allowing the characterization of lesions for a positive diagnosis, the search for possible associated lesions, the evaluation of the prognosis and the follow-up [6].

On ultrasonography, NPs appear as tortuous lobulated masses with a clustered, hypoechoic appearance, with well-defined contours oriented along the axis of the nerve trunk on longitudinal sections and characterized by the target sign on transverse sections with an echogenic center and a hypoechoic periphery, which correlates with the ultrastructure of neurofibromas with a fibrocollagenous center surrounded by a myxomatous periphery. Cystic formations can be seen within the mass as well as posterior enhancement encountered in 70% of cases. On color Doppler, there may be moderate, central or predominantly peripheral vascularization. Some may be poorly vascularized [5,7].

On CT, plexiform neurofibromas appear spontaneously isodense compared to muscle. They take up contrast only slightly, less than muscle, which gives them a hypodense appearance, differentiating them from schwannomas and neurofibrosarcomas [8].

The NP appears on magnetic resonance imaging sequences, which is the reference radiological examination, by a relative T1 hypo signal in relation to the muscle, T2 hyper signal and when it is voluminous, it may contain a central hypo signal producing a characteristic cocoon aspect. The enhancement is variable: central, diffuse, peripheral, in target [6,8,9].

The diagnosis of NFP is essentially anatomopathological, especially outside the context of neurofibromatosis type I. NFP differs from other types of neurofibromas by the importance of its schwannian component [10,11]. The risk of sarcomatous degeneration justifies, whenever technically possible, the removal of the lesion as completely as possible. Finally, these patients should be monitored clinically and radiologically by a multidisciplinary team, at least once a year and then on a regular basis, in order to assess any recurrence or malignant transformation [3,12]. The Treatment for NF depends on the patient's age, health, and medical history, symptoms, which nerves are affected and the expected progression of the disease. Although there is no known cure for NF, surgery and other treatments can help to relieve symptoms [13,14]. Surgery, radiation, and monitoring are the three main treatment approaches. The U.S. Food and Drug Administration (FDA) has approved selumetinib for use in patients with inoperable plexiform neurofibromas. In the clinical trials, over 70% of NF patients with inoperable plexiform neurofibromas saw tumor size reduction anywhere from 20 to 60% in size. In addition to both visible and actual tumor reduction, patients reported higher-quality physical function, reduced pain, improved mobility, and enhanced emotional and psychological status [2,14].

Conclusion

Plexiform neurofibromas are rare benign tumors of the peripheral nerves. They are pathognomonic tumors of NF 1 which is a multi-systemic pathology with clinical and radiological polymorphism. MRI is the examination of choice in the exploration of this pathology. It provides decisive arguments for the positive diagnosis, the evaluation of the prognosis and the follow-up of the lesions.

Consent

As per international standard or university standard, patients' written consent has been collected and preserved by the author(s).

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