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Radiology

Neurofibroma in Unusual Localisation: Rare Case

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Abstract **Case Report**

Neurofibroma is a rare entity, with only few cases reported in the literature so far. Magnetic resonance imaging (MRI) is the key to the diagnosis. We report a 23 year-old women with an antero medial thigh mass diagnosed as neurofibroma on the basis of the imaging findings on US and MRI examinations. The differential diagnosis can be excluded by the fine imaging analysis always associated with the clinical context.

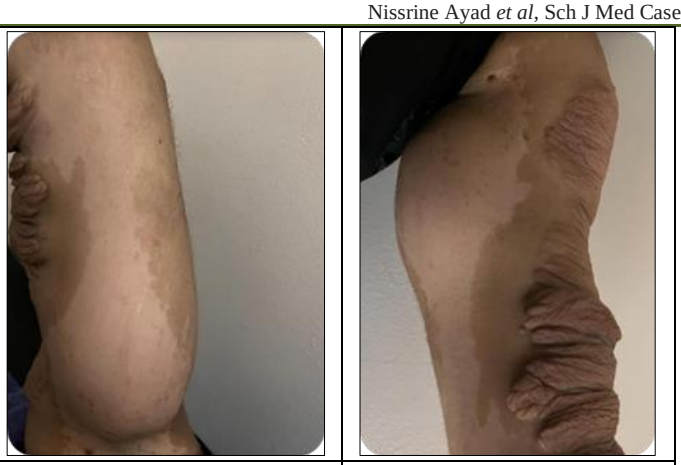
Keywords: Neurofibroma, Rare localisation, MRI.

INTRODUCTION

Neurofibroma is one of the benign peripheral nerve sheath tumors that's commonly being a part of the various signs of the multisystem genetic disorder called Von Recklinghausen's disease or neurofibromatosis type 1 (NF-1). The world Health organisation (WHO) had subdivided this entity into two wide categories: dermal and plexiform. The first one arise from a single peripheral nerve, while the second is associated with multiple nerve blundles [1]. The diagnosis is based on imaging and in some cases, histological examination is necessary to rule out the differential diagnosis. Ultrasound (US) and magnetic resonance imaging (MRI) are the reference modalities. Computed tomography (CT) is rarely performed because of its low resolution for soft tissues [2]. The risk of malignancy increases with the duration of disease as well as according to size of lesion. The commonest malignancy in neurofibromatosis is Malignant Peripheral Nerve Sheath Tumors (MPNST) [3]. For illustration, à rare case of isolated neurofibroma of a cutaneous branch of the femoral nerve is presented.

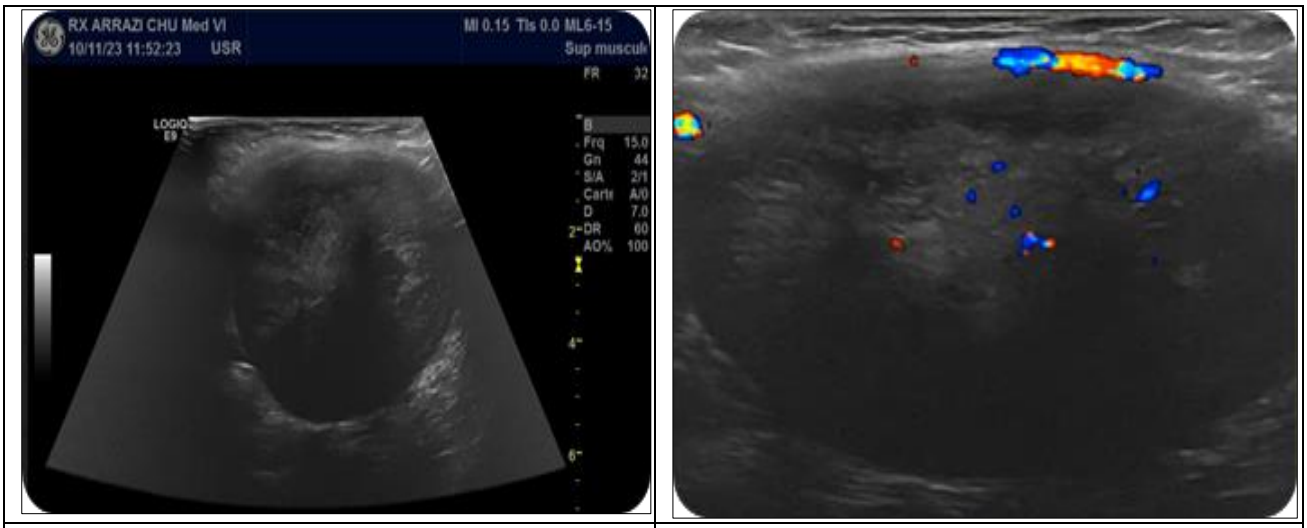
CASE REPORT

A 23 year-old patient was complaining chronical left arm pain with no history of fever or trauma. The physical examination found a mobile mass in the left arm wich generated an exquisite pain (Figure 1). Therefore, the patient were send to our department to conduct an US examination of soft tissue. It showed a fusiform and well defined mass, hypoechoic heterogeneous with sharp borders that seem to continue a linear nervous structure. This swollen area measures: 9 x 6 cm (Figure 2) and it's moderately vascular on doppler color flow. For a better characterization and locoregional extension assesment of the mass, an MRI was performed. The protocol consisted of axial T2 weighted, fat-suppressed T2 weighted, T1-weighted scan and contrast-enhanced fat-suppressed, axial and coronal T1-weighted images using a 1.5-Tesla system. It objectified a small elongated antero medial thigh mass centered to the nerve root wich appears in isosignal on T1 weighted sequence with moderate enhancement after intravenous gadolinium injection (Figure 3). On the basis of these imaging findings, and remains clinical data, specially with « cafe-au-lait » spots, the diagnosis of a neurofibroma was strongly suggested, and finally confirmed by histological examination (Figure 4).



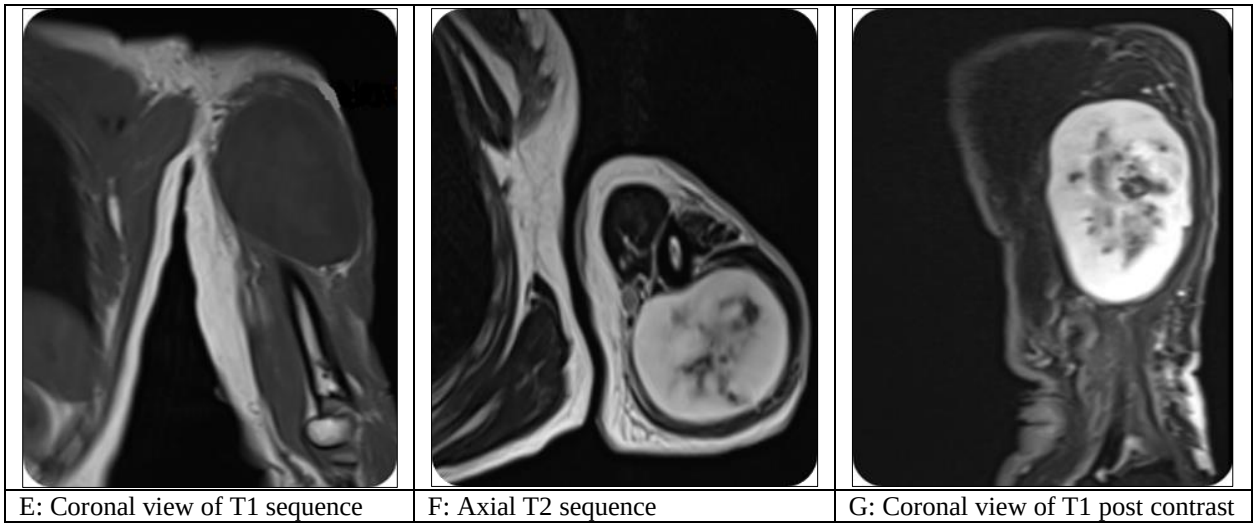
A B

Figure 1: mobile mass in the left arm with (café-au-lait stains).



C D

Figure 2: Fig-1 : An US image on longitudinal section shows the hypoechoic fusiform and well defined mass with sharp borders that seem to continue a linear nervous structure.



E: Coronal view of T1 sequence | F: Axial T2 sequence | G: Coronal view of T1 post contrast

Figure 3: Individualization at the level of the posterior compartment of the left arm centered on the triceps muscular body of a sub-aponeurotic lesion formation, oval in shape, well limited in T1 isosignal, heterogeneous T2 STIR hypersignal with central restriction in diffusion, enhancing heterogeneously and centripetal after injection of gadolinium.

DISCUSSION

The term neurofibromatosis describes a group of genetic disorders that primarily affect the cell growth of neural tissues. At least eight forms of neurofibromatosis have been recognized, the most common form being neurofibromatosis type I (NF-I) [4]. Neurofibromas are the most common benign tumors of NF-I. These can develop at any point along a nerve and often form by late adolescence. Three subtypes of neurofibromas exist: cutaneous, subcutaneous and plexiform. The plexiform variety is specific for the disease [5]. Plexiform neurofibroma is an irregular, thick and non circumscribed tumor of peripheral nerve sheath which can involve multiple nerve fascicles. These are slow growing, painless and locally infiltrating tumors. The consistency of the lesion is compared to „bag of worms" [6]. The disease is manifested by developmental changes in bone, skin and nervous system. Its incidence is estimated to be 1/2500 births per year and its penetrance is almost complete by 5 years of age. The NF1 gene responsible for the disease is located on chromosome 17 at locus 17q11.2 that codes for protein neurofibromin [7, 8]. The pattern of inheritance is autosomal dominant. The size of lesion can increase during pregnancy and puberty [2]. The term "plexus" refers to a combination of interlaced parts or a network. Plexiform neurofibromas are uncommon and occur almost exclusively in about 515% patients with neurofibromatosis-I. Two types of plexiform neurofibromas have been recognized: (a) a diffuse type/ elephantiasis neurofibromatosa and (b) a nodular type [9, 10]. The cranial nerves most commonly involved in plexiform neurofibromas are the fifth, ninth, and tenth. These masses can be quite disfiguring, and hemifacial hypertrophy can occur. These tumors are known to cause symptoms ranging from minor discomfort to extreme pain. These lesions sometimes demonstrate a vascular nature, and they may cause dangerous bleeding and complicate surgical procedures. The size of these tumors may increase during puberty and pregnancy [11]. These lesions manifest early in life and tend to transform to malignant peripheral nerve sheath tumors (MPNST). Malignant progression is generally considered the main cause of mortality, occurring in 2% to 16% of cases [12]. The role of imaging is important for a variety of reasons, including delineating the extent of involvement and effect on adjacent structures, exposing associated anomalies and last but not least, for predicting possible malignant transformation. The role of sonography as a primary modality remains to be recognized not only for the exclusion of simulating conditions at the earliest instance of imaging, but also because a radiation free technique is preferable in the younger population; The earliest description of the sonographic appearances of plexiform neurofibroma was perhaps by Reuter *et al*., who described these tumors as comprising of hypoechoic nodules, which needed to be distinguished from an abscess and a vascular malformation [13]. However, a more definitive description was emphasized by Hong *et al*., who described this entity as a poorly marginated tumor, comprising of multiple hypoechoic nodules on a hyperechoic background with significant vascularity [14].

CONCLUSION

Neurofibromas are neural tumors which occur characteristically in patients with NF1. The tumors which originate from nerve sheath, are large, lobulated masses and demonstrate characteristic imaging features of simultaneous involvement of subcutaneous and cutaneous tissues along with infiltrative invasion of deeper structures. Imaging plays an important role in confirming the diagnosis, delineating involved structures, excluding simulating conditions and malignant transformation.

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