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Solitary Giant Neurofibroma of Right Thigh A Report of a Rare Case

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ABSTRACT
Neurofibromas, typically associated with neurofibromatosis, are benign tumors originating from nerve sheaths. Solitary giant neurofibromas, particularly those emerging from the lower limbs, are relatively uncommon. Here, we present a case of a giant neurofibroma located in the right thigh of a 35-year-old female patient who presented with a 5-year history of a gradually enlarging swelling in the anterior aspect of the right thigh, diagnosed as neurofibroma following MRI and Trucut biopsy. Local excision was performed, with uneventful postoperative recovery. This case underscores the clinical importance of considering neurofibroma in the differential diagnosis of thigh swellings and highlights the efficacy of local excision in managing such lesions

Keyword: *solitary neurofibroma, giant neurofibroma, giant, neurofibroma, neurofibromatosis, mri, surgery..*

1. INTRODUCTION

Neurofibromatosis is a complex neurocutaneous disorder characterized by the involvement of multiple organ systems [1]. It is classified into two major subtypes: neurofibromatosis type 1 (NF1), also known as peripheral neurofibromatosis, and neurofibromatosis type 2 (NF2), referred to as the central type. NF1, commonly known as Von Recklinghausen disease, is an autosomal disorder resulting from mutations in the NF1 gene, which encodes the protein neurofibromin [2]. Patients with NF1 typically exhibit widespread development of multiple neurofibromas, along with a 10% risk of developing malignant peripheral nerve sheath tumors [3,4]. Additionally, they may present with characteristic features such as Lisch nodules, café au lait spots, hypertension, macrocephaly, and an elevated risk of other malignancies [2,5]. In contrast, NF2 primarily manifests with central nervous system involvement, including meningiomas, ependymomas, acoustic neuromas, and ocular abnormalities, with neurofibromas occurring less frequently [6].Solitary neurofibromas, distinct from neurofibromatosis, occur sporadically and are not associated with the genetic mutations seen in NF1 or NF2 [7]. These tumors typically arise in young adults and exhibit no gender preference. Solitary neurofibromas can occur at various anatomical sites, including the retroperitoneal space, cheek mucosa, mandible, spine, nose,urinary bladder, subungual area, abdominal wall, lower lip, and scrotum. Despite their sporadic occurrence, giant solitary neurofibromas are exceptionally rare, particularly those located on the thigh [8]. Consequently, there is limited literature documenting cases of giant solitary neurofibromas in this anatomical region. Therefore, the present study aims to contribute to the existing body of knowledge by reporting a unique case of giant solitary neurofibroma located on the right thigh, highlighting its rarity and clinical significance.

Case Presentation

A 35-year-old female presented to our outpatient department with a complaint of a swelling measuring 15 x 10 cm over the anterior aspect of the right thigh, present for the past 5 years. The patient first noticed a small swelling 5 years ago, which had a gradual onset and had progressively increased in size over time. He reported no history of movement restriction, no

neurological deficits distally, pain, discharge, fever, unintentional weight loss or any other swellings elsewhere in the body. The patient had no relevant past medical, surgical, or family history.Clinical examination revealed a firm, non-tender swelling measuring 10 x 15 cm was noted in the anterior and medial compartments of the right mid-thigh. The swelling extended approximately 10 cm below the pubic symphysis to around 15 cm above the knee joint. It exhibited mobility in both medial to lateral and vice versa directions. No signs of inflammation or skin involvement was observed, and the patient did not present with any other symptoms or signs. To establish a diagnosis, an MRI of the right thigh was performed followed by a Trucut biopsy. The MRI findings were suggestive of a well-defined solid mass. Routine blood investigations were within normal limits.The MRI revealed a heterogeneously enhancing mass measuring 11 x 12 x 15 cm located in the upper thigh, involving the anterior and medial compartments. The imaging also indicated cystic degeneration/necrosis within the mass and its infiltration into the sartorius muscle (Figure 1).The swelling is seen displacing the adjacent muscles and neurovascular bundle. These findings were indicative of a likely benign neoplastic mesenchymal tumor.

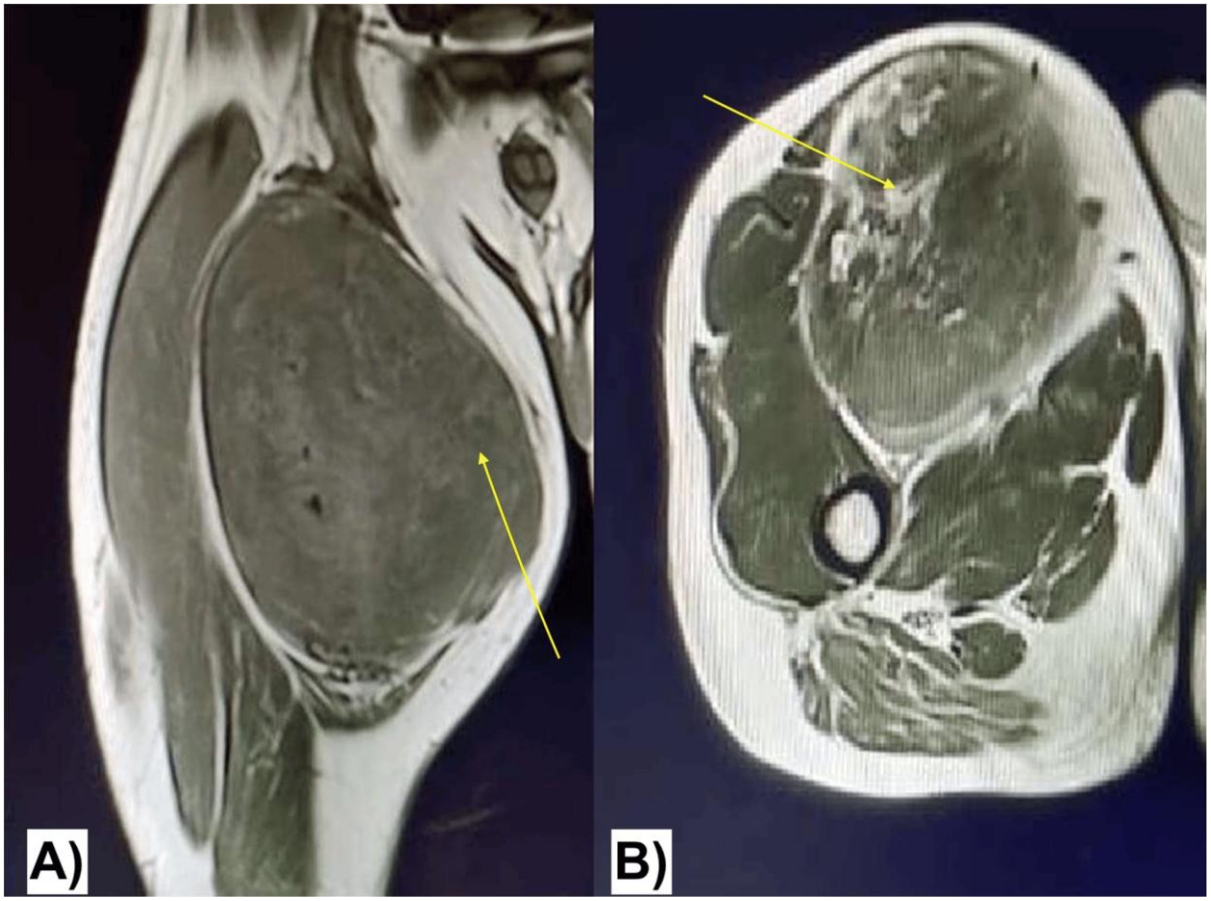

Figure 1: MRI films showing heterogenosly enhancing mass occupying the anterior compartment of thigh.

Magnetic Resonance Imaging (MRI) depicting a heterogeneously enhancing mass measuring 11 x 12 x 15 cm located in the upper thigh, involving the anterior and medial compartments. The swelling is seen displacing the adjacent muscles and neurovascular bundle. A) MRI image in coronal plane; B) MRI image in axial plane.

10 cm over the anterior aspect of the right thigh, present for the past 5 years. The patient first noticed a small swelling 5 years ago, which had a gradual onset and had progressively increased in size over time. He reported no history of movement restriction, no neurological deficits distally, pain, discharge, fever, unintentional weight loss or any other swellings elsewhere in the body. The patient had no relevant past medical, surgical, or family history.Clinical examination revealed a firm, non-tender swelling measuring 10 x 15 cm was noted in the anterior and medial compartments of the right mid-thigh. The swelling extended approximately 10 cm below the pubic symphysis to around 15 cm above the knee joint. It exhibited mobility in both medial to lateral and vice versa directions. No signs of inflammation or skin involvement was observed, and the patient did not present with any other symptoms or signs. To establish a diagnosis, an MRI of the right thigh was performed followed by a Trucut biopsy. The MRI findings were suggestive of a well-defined solid mass. Routine blood investigations were within normal limits.The MRI revealed a heterogeneously enhancing mass measuring 11 x 12 x 15 cm located in the upper thigh, involving the anterior and medial compartments. The imaging also indicated cystic degeneration/necrosis within the mass

and its infiltration into the sartorius muscle.

The local excision of the swelling was done during which the tumor was carefully dissected from the surrounding structures-femoral vessels and nerve were identified and medicalised away from the swelling and the anterior compartment of the thigh (Figure 2). The excision was performed completely, including the removal of the sartorius muscle along with the tumor. The excised specimen was then sent for histopathological examination (HPE). HPE examination of the Trucut biopsy sample confirmed the presence of spindloid cells with wavy fibrillary cytoplasm, consistent with a diagnosis of neurofibroma.Following the procedure, the patient was carefully observed during postoperative period which was uneventful. The patient was finally discharged on the seventh day after surgery.Follow up : 12 month follow up was taken and no local recurrence seen both clinically and by ultrasonography.

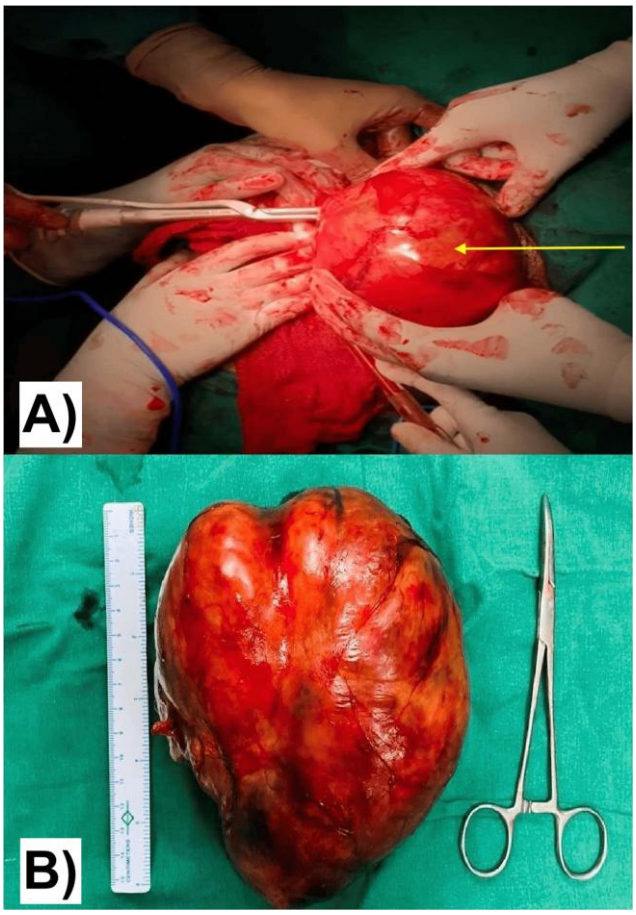

Figure 2: Showing the intra-operative findings and specimen after excision

A) Intra-operative image; B) Excised specimen

2. DISCUSSION

Peripheral nerve sheath tumors encompass various types, including schwannomas, neurofibromas, and neurogenic sarcomas [2]. Approximately 10% of benign soft tissue tumors consist of schwannomas and neurofibromas [9]. NF1, characterized by the development of multiple neurofibromas along with other symptoms, has an incidence of approximately 1 in 3000 live births and is associated with various malignancies such as malignant peripheral nerve sheath tumors (MPNSTs), gliomas, leukemias, and gastrointestinal stromal tumors (GISTs) [10].Neurofibromas associated with NF1 tend to exhibit a larger size and have a higher potential for malignancy. MPNSTs, a type of malignant tumor arising from peripheral nerves, can occur both sporadically and as part of NF1 [2]. Neurofibromas can develop in any peripheral encapsulated nerve of the body, and they can be classified into three main types: solitary or isolated neurofibromas, diffused neurofibromas typically originating from cutaneous nerves, and plexiform neurofibromas characterized by their diffused, tortuous expansion along the branches of the parent nerve [7]. In cases of solitary neurofibromas, it is important to assess for signs of NF1, such as café au lait spots, other neurofibromas, bone lesions, and Lisch nodules. The symptoms of solitary neurofibromas vary depending on factors such as the size and location of the tumor and any pressure effects caused by deeper tumors [5].The pathogenesis associated with solitary neurofibromas independ of NF1 remains incompletely understood. A study by Storlazzi et al., in 2005, reported that translocation mediated inactivation of the NF1 gene locus on chromosome 17, results in an increased production of

neurofibromin [11]. Neurofibromin further regulates the RAS-mediated cell proliferation pathway, and its increased production leads to elevated levels of activating proteins p2 and p13, consequently promoting cellular proliferation of Schwann cells associated with neurofibroma.Gross examination typically reveals encapsulated firm masses of varying sizes.Histopathologically, neurofibromas consist of nerve sheath cells such as perineural, myxoid, Schwann cells, or mast cells [7,12]. Immunohistochemical studies utilizing markers such as S100 or CD34, or both, aid in distinguishing neurofibromas from other soft tissue tumors [13]. For instance, leiomyomas and myofibroblastic tumors are positive for SMA, whereas S100 is a preferred marker for nerve cell tumors. There are very few reported cases of Giant Neurofibroma , typically for a swelling to be called it as a giant it needs to be in greater than 5 cm in any dimension [14].

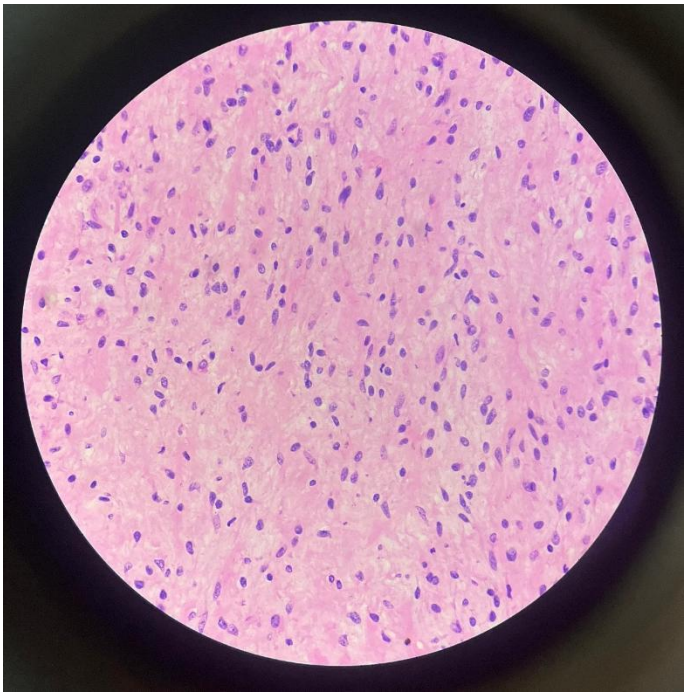

Figure 3: 40X Histopathological image of neurofibroma

Treatment of solitary benign neurofibromas is dependent on factors such as tumor location, size, and involvement of surrounding structures.

3. CONCLUSION

Solitary giant neurofibromas that occur sporadically, without any association with neurofibromatosis, are rare, particularly in the thigh region.Complete excision after identification of neural structure without causing nerve injuries is important as it happened in our case. Only a few reported cases of giant solitary neurofibromas in the thigh have been documented in the medical literature. This rarity underscores the unique nature of such occurrences and highlights the importance of reporting and documenting cases to enhance our understanding of this uncommon presentation of neurofibromas.

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