Case Report

Solitary giant neurofibroma of thigh

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Abstract Neurofibromas are rare, benign, nerve sheath tumors in the peripheral nervous system. The solitary type is found in those who do not have neurofibromatosis. Solitary neurofibromas are too rare in the giant type. We report a rare case of a solitary giant neurofibroma of the anterior right thigh. The diagnostic criteria, characteristics of imaging studies, and operative approach are represented.

Key Words: Giant, lower extremity, neurofibroma, solitary, thigh

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INTRODUCTION

Neurofibromas are benign, nerve sheath tumors in the peripheral nervous system. They evolve anywhere along a nerve from the dorsal root ganglion to the terminal nerve branches.[1,2] Neurofibromas are commonly, but not always, associated with neurofibromatosis type 1 (NF-1).[3] The solitary form occurs in those who do not have neurofibromatosis.[4]

Solitary neurofibroma is common in young adults, without a gender preference. Neurofibromas have been known to occur mostly in the trunk and head compared to other body surfaces. A solitary neurofibroma has been reported in the following locations, the spine, retroperitoneal space, mandible, cheek mucosa, nose, bladder, male breast, subungal area, abdominal wall, lower lip, and scrotum.[5]

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To the authors' knowledge, a solitary giant neurofibroma in the thigh has not been described previously. Electronic searches for reports of similar cases using the keywords, 'neurofibroma,' 'solitary,' and 'giant,' were conducted in the Pubmed and Scopus databases for all English articles with abstracts. Solitary giant neurofibromas in uncommon sites have been reported on the oral cavity, neck, scalp, presacral space, paraspinal area, retroperitoneum, labia, and appendix.

CASE REPORT

A case is presented of a 31-year-old female who complained of a painless, growing, palpable mass in her left thigh, associated with erythema of the adjacent skin [Figure 1]. No other findings at the physical examination or any other symptomatology were reported. Her family history and past medical history were not significant. For diagnosis, magnetic resonance imaging (MRI) of her left thigh was performed. On the MRI of the left thigh, a discrete, large, well-defined homogeneous tumor measuring 7 × 15 × 21 cm was described in the anteromedial compartment of the thigh, by displacing all muscles of the medial compartment. On MRI, the tumor showed an isointense signal on T1-weighted images

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and high-signal intensity on T2-weighted images, suggestive of myxoma [Figures 2 and 3]. A core needle biopsy was performed and a microscopically pathological examination showed a neoplastic proliferation of all elements of the peripheral nerve (axons, Schwann cells, and fibroblasts) with a diffused pattern. Elongated nuclei with a wavy serpentine configuration and pointed end, without evidence of malignant transformation were also evident, which confirmed the diagnosis of neurofibroma. The tumor cells were negative for epithelial membrane antigen (EMA), smooth muscle actin, and S100 protein.

The patient underwent surgical excision of the mass and a discrete, large, capsulated mass lesion was exposed and completely removed. There was no adhesion to the adjacent tissue or vascular involvement [Figure 4].

Pathological findings confirmed the neurofibroma diagnosis and revealed tumor-free margins on all specimens. In the specimen taken from the adjacent skin, hyperpigmentation of the basal layer with neurofibromatous change in the dermis was present [Figures 5 and 6].

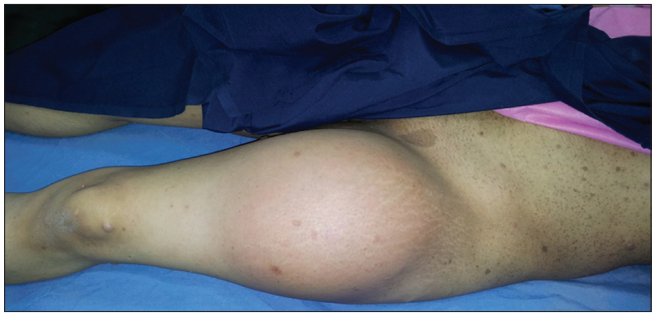

Figure 1: Large palpable mass in the left thigh of patients associated with erythema of the adjacent skin

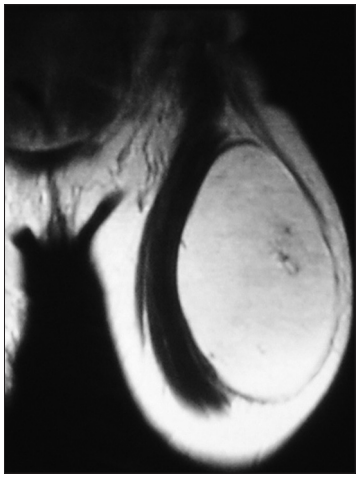

Figure 3: Coronal T2-weighted images show a well-defined homogeneous mass lesion

Soon after the surgery, the patient was re-examined, and there were no circumscribed or segmental features of NF-1.

DISCUSSION

There are three types of peripheral neural sheath tumors (PNST) including schwannoma, neurofibroma, and neurogenic sarcomas.[4] Schwannomas and neurofibromas, as benign PNSTs, account for 10% of benign soft-tissue tumors.[2,6]

Neurofibromas can develop in any peripheral encapsulated nerve of the body, which can be invaded by the neurofibroma and can be divided into: (1) Solitary or isolated neurofibromas, which more commonly originate from cutaneous nerves; (2) diffused, most likely to arise from the nerves in the subcutaneous tissues of the head and neck; (3) plexiforms, which

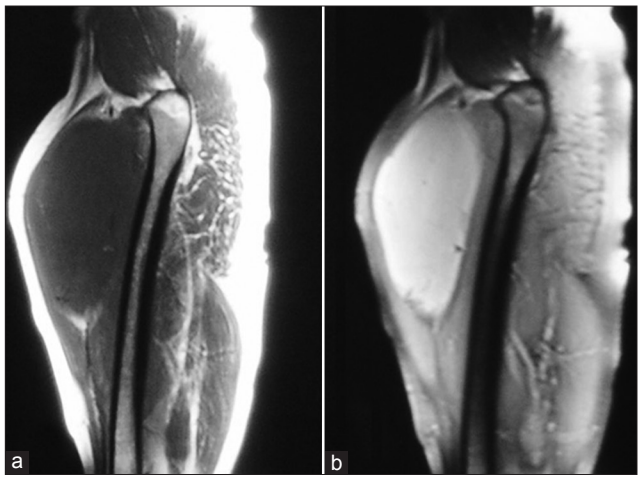

Figure 2: Sagittal T1-weighted images (a) and T2-weighted images (b) Show a fusiform shape of a slightly isointense to muscle intensity on T1 images and homogenously hyperintense on T2 images

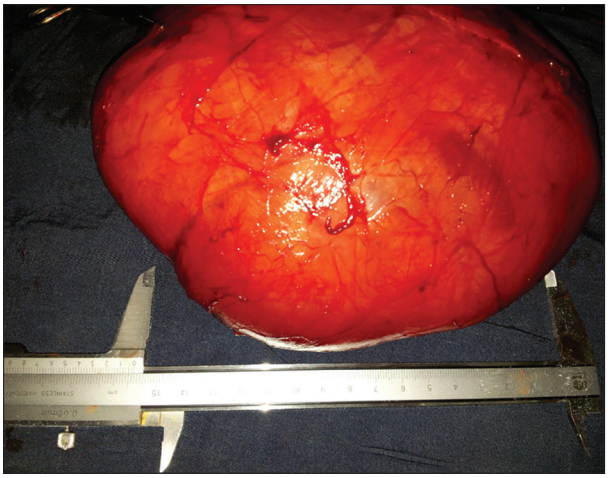

Figure 4: Giant encapsulated tumor after resection, having no adhesion to the adjacent tissue or vascular involvement

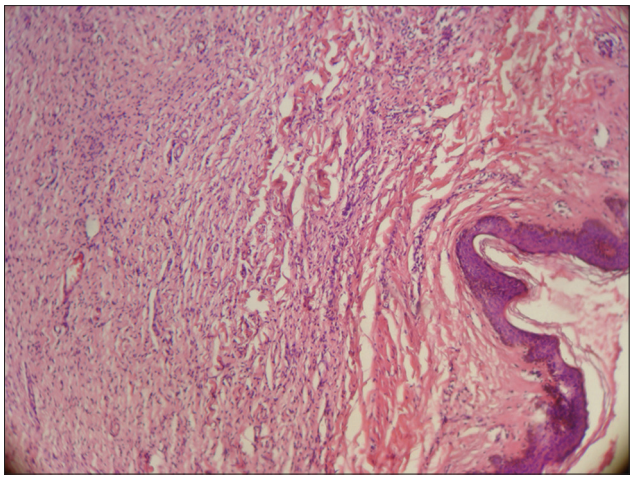

Figure 5: On microscopic examination, a combined proliferation of all the elements of a peripheral nerve (axons, schwann cells, and fibroblasts) are seen, most have elongated nuclei, with wavy serpentine configuration and a pointed end (H and E, ×100)

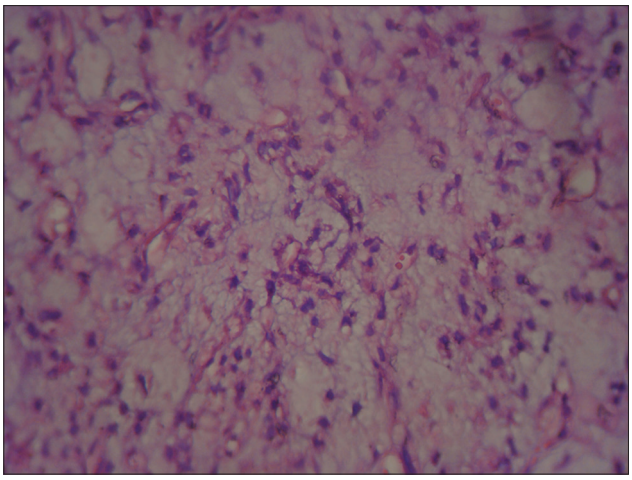

Figure 6: In some areas, mucinous changes were prominent, which could be mistaken for myxoma or myxoid liposarcoma (H and E, ×400)

are diffused masses with tortuous expansion along the branches of the parent nerve.[2,4]

Although 10% of the lesions are isolated, up to 10% of the neurofibromas are associated with neurofibromatosis. In the setting of NF-1, neurofibromas tend to be larger and have a higher propensity of malignant transformation. The lesions may be deep or superficial, and can involve small or cutaneous nerves or large main nerve trunks. Superficial neurofibromas characteristically present with painless masses, while manifestation of deeper types are mainly neurological symptoms.[2,4,6]

In case of encountering a neurofibroma, the clinician should search for other manifestations of generalized neurofibromatosis. Neurofibromatosis type I (NF-1), formerly known as von Recklinghausen disease is an autosomal dominant condition which is clinically characterized in part by pigmented skin lesions known as café-au-lait spots, benign cutaneous and subcutaneous tumors known as neurofibromas, characteristic bone lesions, and focal malformations of the iris.[3]

On gross appearance, a neurofibroma is a firm mass with variable size, from a few millimeters to a few centimeters. Pathologically, diagnosis of neurofibromas is made by detection of the cells in the nerve sheath including: Schwann cells, perineurial cells, fibroblasts, and mast cells.[1] They too are similar to schwannomas. The differentiating features include the presence of neurofibromatosis, nerve fibers, and more myxoid tissue (Antoni B tissue) in the neurofibromas, while schwannomas are well-capsulated, and the matrix is more compact, associated with fascicular formation and Antoni A cells.[3,7]

In contrast to schwannomas, neurofibromas are closely connected to their nerve of origin and cannot be separated from the nerve fibers. Therefore, resection needs sacrificing of the parent nerve.[2,7]

Regarding the location and size of tumor, the symptoms are variable. Discoloration of skin accompanied with disfiguration in the cutaneous form of NF is expected, whereas, deeper masses can cause neurological symptoms and destruction as a result of compression of the neighboring organs. Neurofibromas are often asymptomatic, but can cause debilitating pain and motor sensory dysfunction. They are usually benign and grow slowly, but sometimes degenerate to become malignant.[3,6,8]

Computed tomography (CT) depicted a relatively hypodense, well-defined mass in contrast to muscle density. Neurofibromas are usually low-signal intensity to isointense in muscles on T1-weighted MR images and slightly hyperintense on T2-weighted MR images. They are variable with contrast enhancement, but are nonspecific in terms of their signal intensity. Signal on T2-weighted images can be either homogeneously hyperintense or show a characteristic target sign. A target sign is more suggestive of neurofibromas than schwannomas, which has a central area of low T2 SI histologically corresponding to a fibrocollagenous tissue; whereas, the outer area of high T2 SI corresponds to a myxomatous tissue. The typical feature is a fusiform shape oriented longitudinally in the nerve distribution, often revealing tapered ends that are contiguous with the parent nerve. The 'split fat sign' can be seen in both neurofibromas and schwannomas. As the tumor grows, an encompassing rim of normal fat is maintained.[2,9,10]

The management of a neurofibroma depends on the symptoms and is indicated when there is pain, progressive neurological deterioration, compression of the adjacent tissues, with loss of functions and cosmetic consideration, as well as suspected malignant degeneration.[11]

In summary, we have presented a case of a solitary giant neurofibroma in the left thigh, which was rare in size and location. The differential diagnosis of a solitary neurofibroma included myxoma, myxoid liposarcoma, and schwannoma.

REFERENCES

1. Weiss SW, Goldblum JR: Benign tumors of peripheral nerves. In: Weiss SW, Goldblum JR: Enzinger and Weiss's Soft Tissue Tumors. 5th ed. St. Louis, Mo: Mosby, 2008, p. 769-84.
2. Lin J, Martel W. Cross-sectional imaging of peripheral nerve sheath tumors: Characteristic signs on CT, MR imaging, and sonography. AJR Am J Roentgenol 2001;176:75-82.
3. Carpenter B, Stickney N. Neurofibroma of the Anterior Leg: A Case Report. The Foot and Ankle Online Journal 5:2.
4. Chennakeshaviah G, Ravishankar S, Maggad R, Manjunath GV. Solitary Giant Intramuscular Myxoid Neurofibroma Resulting in an above Elbow Amputation. Case Rep Pathol 2012;2012:353215.
5. Lee YB, Lee I, Park HJ, Cho BK. Solitary Neurofibromas: Does an Uncommon Site Exist? Ann Dermatol 2012;1:100-1.
6. Banks KP. The target sign: Extremity. Radiology 2005;234:899-900.
7. Ismail N, Shehu B, Lasseini A, Hassan I, Shilong D, Obande J, *et al*. Solitary Giant Neurofibroma of the Scalp with Calvarial Defect in a Child. J Surg Tech Case Rep 2010;2:24-6.
8. Barajas-Gamboa J, Flórez-Salamanca L. Solitary neurofibroma in the abdominal wall of a patient without neurofibromatosis: Case report. Biomédica 2009;29:501-5.
9. Subhawong TK, Fishman EK, Swart JE, Carrino JA, Attar S, Fayad LM. Soft-tissue masses and masslike conditions: What does CT add to diagnosis and management? AJR 2010;194:1559-67.
10. Wu JS, Hochman MG. Soft-tissue tumors and tumorlike lesions: A systematic imaging approach. Radiology 2009;253:297-316.
11. Gottfried ON, Viskochil DH, Fults DW, Couldwell WT. Molecular, genetic, and cellular pathogenesis of neurofibromas and surgical implications. Neurosurgery 2006;58:1-16.

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