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A Large Retroperitoneal Neurofibroma with Lumbar Scoliosis and the First Lumbar Vertebral Subluxation: A Rare Case Report

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Abstract

Retroperitoneal neurofibromas are rare tumors, with an incidence of 3-5% for all neurofibromas. The Schwann cells in the peripheral nerve give rise to these histologically benign tumors. They can be hard to diagnose and treat because of where they are located and their potential to invade the surrounding area and press on nerve roots or other structures. Herein, we report a 17-year-old male patient, who had a complaint of low back pain and pain radiating to the legs, especially on the right side, for one year. MRI and CT scans showed a fairly big (9.7 cm × 4.3 cm) retroperitoneal mass on the right side of the aorta. He also had associated lumbar scoliosis and first lumbar vertebral subluxation. Subsequently, CT-guided core biopsy of the mass provided pathological confirmation indicative of a spindle cell neoplasm consistent with neurofibroma. Retroperitoneal neurofibromas are characterized by their slow growth, and therefore the tumor may not present clinically for a very long time and may only present when it has grown to a considerable size and is exerting pressure on structures in the retroperitoneum. Here, the macroscopic characteristic of a giant size and retroperitoneal location of the tumor gave rise to the low back pain and the radicular leg pain, as it resulted in the compression of spinal nerves. MRI and CT scans were useful in the staging of the tumor prior to surgery, and histologic examination of the biopsy specimen confirmed the diagnosis. This case should serve as a reminder that any rare retroperitoneal neurofibromas, especially those that do not fit traditional clinical presentations, should be approached in a more general and extensive collaboration with clinicians, radiologists, and pathologists. As such, the acute phase must be identified early and treated adequately to avoid other complications and improve the outcomes of the treatment.

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Introduction

Neurofibromas are a form of benign tumoral growth that arise from the nerve sheath cells.[1] These tumors can be sporadic or associated with NF1 – an inherited neurocutaneous syndrome that results from mutation of the NF1 gene. Retroperitoneal neurofibromas are relatively rare growths that occur in the retroperitoneal space and are found in 3 – 5 % of all cases of

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Neurofibromas.[2] The retroperitoneum is a large part of the anatomy of the human body, with important organs including the kidneys, pancreas, ureters, blood vessels, and nerves. Despite being nonspecific in nature, a large retroperitoneal mass can apply substantial pressure on these structures, which results in a variety of symptoms.[2,3] Thus, the diagnosis and the treatment of the retroperitoneal neurofibromas should involve clinicians, radiologists, as well as pathologists. A 17-year-old male with a large retroperitoneal neurofibroma with lumbar scoliosis and L1 vertebral subluxation is discussed in this case report. The present case exemplifies the multifaceted diagnostic and therapeutic approach of these rare neoplasms, emphasizing the focus on a synopsis of clinical history, radiological imaging, and histopathological examination of the tissue.[4,5]

Case Description

The patient experienced the complaints of low back pain that are accompanied by lower limb pain and more right-sided than left for few months. A normal physical examination was conducted on the patient. Further examination of the patient was done through radiological tests. MRI of the lumbar spine showed multiple round, well-defined hyperintense T2 signal intensity lesions in the right paraspinal muscles between the T12 and L3 vertebrae. Also, for the MRI dynamics, the following comments could be made: a slight curve of the lumbar spine – scoliosis, the convexity of which is directed to the right; subluxation of the L1 vertebra to the right. Further CT scans showed a large retroperitoneal mass measuring about 9.7 × 4.3 cm in size positioned on the right side of the aorta. Upon failure to definitively diagnose the nature of the mass, a CT-guided core biopsy of the lesion was done. In the light of histopathological examination of the biopsy specimen, a diagnosis of spindle cell neoplasm was made, and it was in favor of a neurofibroma. In conclusion, radiological studies, MRI, and CT, as well as the histopathological examination of the biopsy sample, were helpful in coming to the diagnosis of this 17-year-old patient presenting with low backache with radiation to the leg.

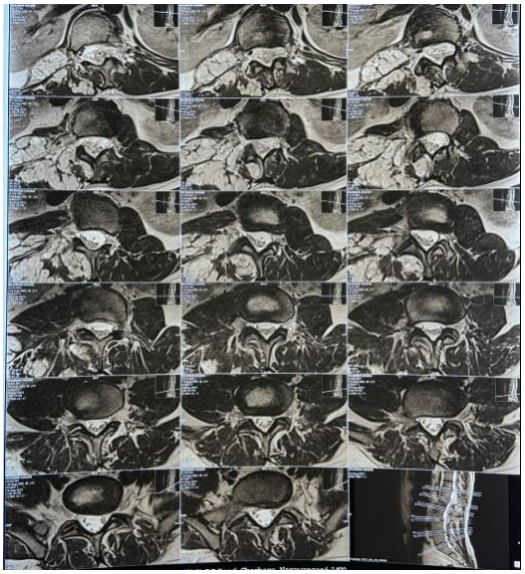

Fig. 1: Axial MRI of Lumbo-Sacral Spine

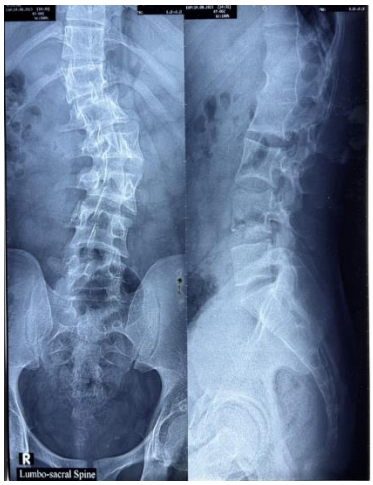

Fig. 2: Radiographic Imaging of the Lumbo-Sacral Spine

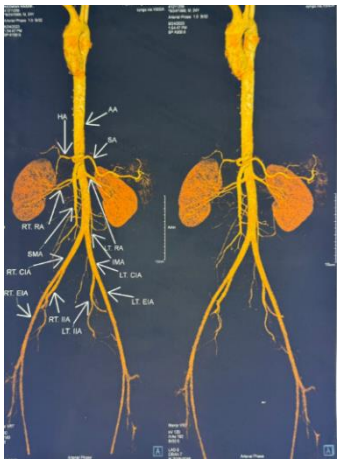

Fig. 3: Arterial Phase Imaging of the Abdominal Aorta and Renal Arteries

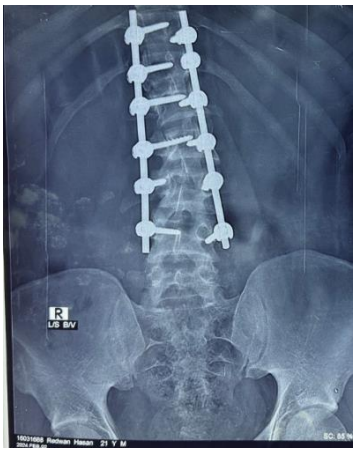

Fig. 4: Post-operative Radiographic Imaging of the Lumbo-Sacral Spine

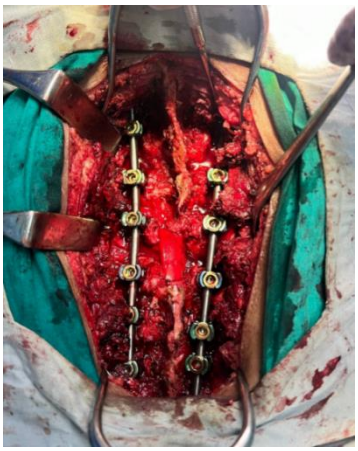

Fig. 5: Post-operative Lumbo-Sacral Spine

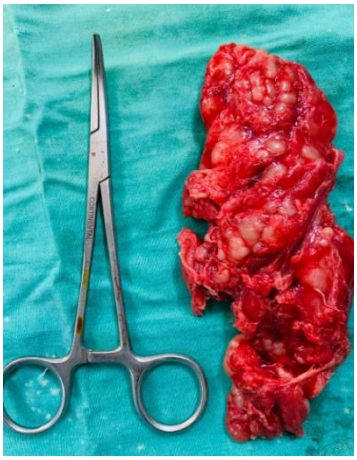

Fig. 6: Excised Large Retroperitoneal Neurofibroma

Discussion

Retroperitoneal neurofibromas are rare and can present a diagnostic and therapeutic problem primarily because of their infiltrative nature and the ability to compress adjacent structures. Some of these tumors may not cause symptoms at all for a long time, but rather present clinically when they have attained considerable size or when they start to invade or compress on structures derived from the neighboring organs and tissues.[6,7] In the present case, signs such as low back pain and radiating leg pain were expected due to the pressure that the tumor exerted on spinal nerves and adjacent structures.[8] A diplomatic approach is usually employed in the management of retroperitoneal neurofibromas, with radiological imaging being an important diagnostic tool in the preoperative period.[9] MRI may show the size and location of the tumor and its relative position to critical structures, while CT scans may give details of the density of the tumor and its interaction with vessels. Furthermore, biopsies are mandatory in order to allow

histopathologic examination of a biopsy specimen in order to confirm the diagnosis of neurofibroma and exclude the presence of more aggressive or malignant lesions.[10]

Superficial or retroperitoneal neurofibromas, however, comprise only 3-5% of all reported neurofibromas. These tumors are frequently discovered incidentally, and therefore diagnosis and treatment are difficult for the physicians. In the present case, however, the primary tumor was bulky and was (9.7 × 4.3) cm in size and situated in the retroperitoneum, which may explain considerable symptoms in the patient. There were two main manifestations observed during the patient's initial consultation: low back pain, which was reproduced with radiation towards the legs, mostly on the right side. These symptoms are most probably due to a mass effect caused by the massive retroperitoneal neurofibroma and subsequent compression of spinal nerves and the related structures.[11] The lumbar scoliosis and L1 vertebral subluxation seen in this case should be considered as mass effect-related complications of the tumor since spinal deformities may occur due to distortion and displacement of normal vertebral columns by large space-occupying lesions.[12] MRI and CT scans are essential diagnostic tools in the differentiation of retroperitoneal neurofibromas and in surgical planning preoperatively.[13] MRI is particularly helpful in defining the margins of the tumor and outlining the interdependence of the spinal cord, nerve roots, and large vessels.[14] Magnetic resonance imaging produces great soft tissue contrast, which helps in the evaluation of the tumor signal intensity, uniformity, and presence or absence of cystic or hemorrhagic components.[7] CT imaging offers the criterion to the size, position, density, and the relations of the mass to the adjacent bony structures. This information is valuable in surgical operation planning since it defines the likelihood and modality of the resection procedure and the risks of such possible complications as intraoperative hemorrhaging.[15] In conclusion, radiological imaging is used in the diagnosis and preoperative assessment of retroperitoneal neurofibromas, but histopathological examination confirms the diagnosis using a biopsy of the of the tumor. The lesion in this case was a benign neurofibroma, and, by performing the basic core biopsy, the diagnosis was made and ruled out the malignant-appearing spindle-shaped cells that are typically associated with malignant schwannomas.[14] Retroperitoneal neurofibromas are complex tumors, and their management should involve clinicians, radiologists, and pathologists. Patients with these rare and complex tumors require thorough clinical examination and imaging, as well as definitive histopathological examination to assist in their management.

Conclusion

This case underscores the need for the appropriate focused management approaches to rare retroperitoneal neurofibromas that involve clinical history, radiology investigations, and histopathologic evaluation. As important as the diagnosis is the timely control of diabetes to minimize risk factors and maximize likely benefits to the patient. Early interventions are possible and crucial for the favorable outcome of both diseases and a successful patient treatment process. A holistic approach to formulating diagnoses and offering tailored treatment is essential when dealing with these multiple and interrelated medical presentations.

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