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Case Report

Utility of MRI neurography in neurofibromatosis type I: Case example and review of MRI neurography literature

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Abstract

Background: Neurofibromatosis is an autosomal dominant disorder of the nerves, resulting in café-au-lait spots, axillary freckling, macules, and neurofibromas throughout the nervous system. Diagnosis of this condition has in the past been mainly clinical, but the usage of magnetic resonance imaging neurography (MRN) is a new diagnostic modality. Here, we report on a case of neurofibromatosis type I (NF-1) that was diagnosed using MRN after a protracted clinical course.

Case Description: A 23-year-old female presented with several months of worsening right upper and lower quadrant abdominal pain. The patient underwent computed tomography (CT) of the abdomen and pelvis demonstrating multiple neurofibromas involving the psoas muscle and mesentery of the lower abdomen. Subsequent total neuronal axis magnetic resonance imaging (MRI) using the neurography protocol (MRN) showed multiple neurofibromas in both the right brachial plexus and lumbar plexus.

Conclusion: We present a case of NF-1 that was diagnosed using MRN following a protracted clinical course. MRN is a diagnostic modality for NF-1 and other peripheral nerve disorders.

Key Words: Lumbar plexus, MRI neurography, neurofibromatosis type I

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INTRODUCTION

Neurofibromatosis is an autosomal dominant disorder characterized by café-au-lait spots, axillary freckling macules, and neurofibromas scattered throughout the nervous system. While neurofibromatosis type I (NF-1) is typically diagnosed with genetic testing, it is often missed on clinical examination because of its variable expressivity. Here, we report on a patient with NF-1 who was diagnosed using magnetic resonance imaging neurography (MRN) after a protracted clinical course. MRN is a new useful diagnostic modality that can be used in patients at high suspicion for NF-1 and other peripheral nerve sheath tumors. It can also be used for preoperative surgical planning and postoperative surgical monitoring.

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CASE ILLUSTRATION

A 23-year-old female presented with several months of worsening right upper and lower quadrant abdominal pain with radiation to the right anterior thigh accompanied by paresthesia and numbness. She also reported right-sided neck pain with numbness radiating into the right arm involving the ring and middle finger. Four months before this visit, she was treated with a cholecystectomy to address her abdominal pain: the gallbladder proved to be normal. Next, a computed tomography (CT) of the abdomen and pelvis demonstrated multiple lesions within the psoas muscle and mesentery of the lower abdomen [Figures 1 and 2]. She was referred to the neurosurgery clinic and on examination demonstrated hyperpigmentation in the axilla with mild freckling. Additionally, she had diminished sensation in the right C8 distribution. She underwent neural axis magnetic resonance imaging (MRI) using the neurography protocol (MRN). It revealed multiple neurofibromas involving the right brachial and lumbar plexuses [Figures 3–5]. Within the lumbar plexus, neurography demonstrated multiple neurofibromas involving the nerve roots throughout the plexus and involved both the femoral and sciatic nerves bilaterally. Given the extent of involvement, the patient was medically managed.

DISCUSSION

Genetics of NF-1

NF-1 is an autosomal dominant genetic disorder caused by a deletion mutation of the *neurofibromin* gene on chromosome 17.[13] While roughly half of the cases of NF-1 are genetically inherited, the other half arise due to *de novo* mutations.[5] Loss of the *neurofibromin* gene leads to a loss of regulation of DNA replication, resulting in uninhibited growth of the characteristic cutaneous neurofibromas, plexiform neurofibromas, and optic gliomas.[9] NF-1 has complete penetrance but variable expressivity, resulting in a wide range of phenotypes.[4] Common manifestations include café-au-lait spots, axillary freckling, lisch nodules in the iris, optic gliomas, and neurofibromas (peripheral, cutaneous, plexiform, or nodular).

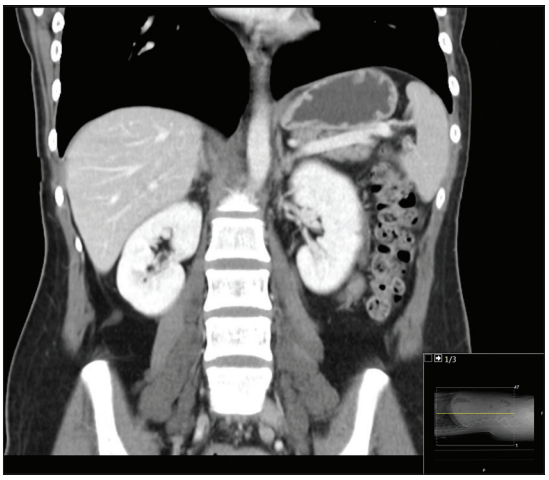

Figure 1: Coronal CT with contrast

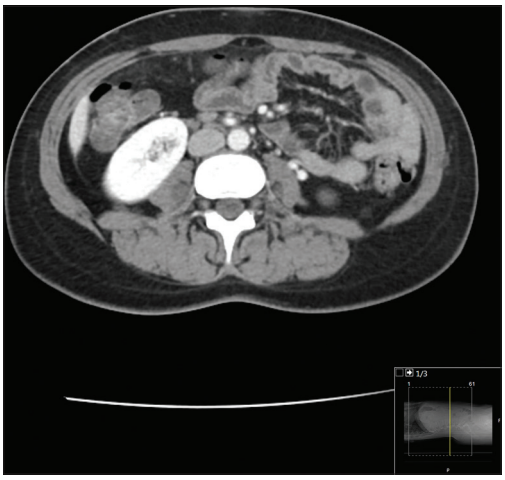

Figure 2: Axial CT with contrast

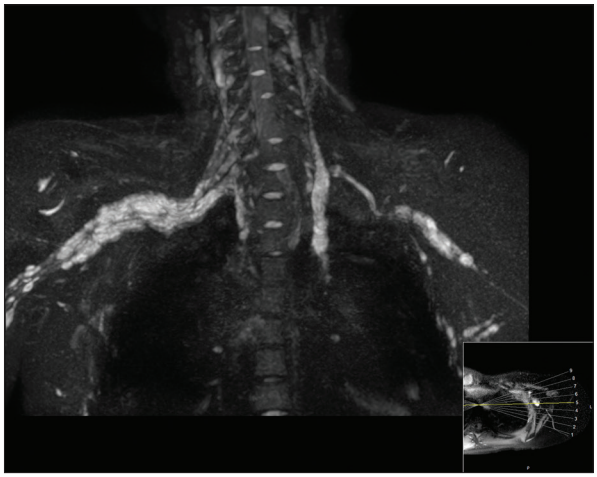

Figure 3: Coronal brachial plexus MRN 3D reconstruction

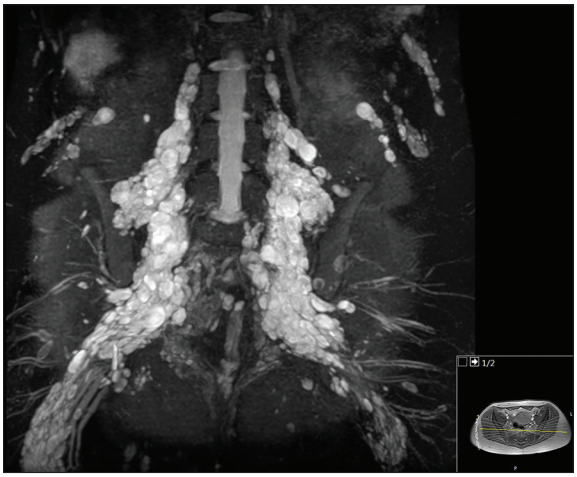

Figure 4: Coronal lumbar plexus MRN 3D reconstruction

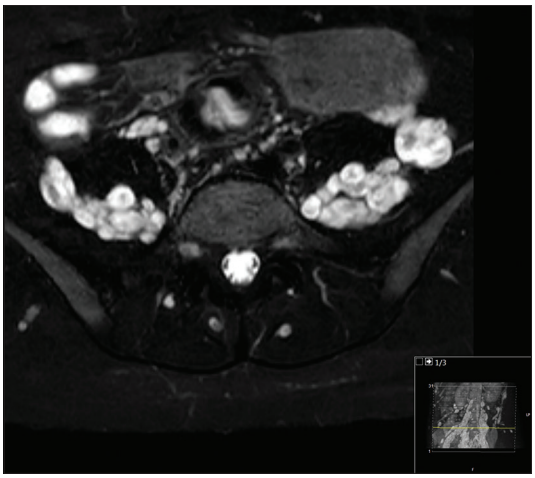

Figure 5: Axial lumbar plexus MRN

MRN

Over two decades, MRN has slowly become an important resource for the imaging and diagnosis of many peripheral nerve disorders. Additionally, three-dimensional reconstructions of neurography images had markedly improved over this period, and is now being used more frequently.[3] As opposed to conventional MRI, MRN better distinguishes between neural surrounding soft tissue.[8] MRN identifies disruption of the fascicular pattern, the etiology of T2-weighted MR signal hyperintensity, deviations of nerve course, and enlargement of the nerve itself.[7,11] MRN uses mainly T1-weighted and fluid-sensitive fat-suppressed T2-weighted images to assess peripheral nerve damage. In neuropraxia, the MRN will show hyperintensity on T2-weighted imaging with enlargement of the nerve. With axonotmesis, reflecting complete rupture of the nerve, the distal segment undergoes Wallerian degeneration, while the proximal segment remains viable. With neuropraxia, the MRN will show enlargement of individual nerve fascicles,[10,12] while with neurotmesis, the MRN will demonstrate complete disconnection between the proximal and distal segments of the nerve, with fibrosis and granulation tissue best seen on T2-weighted images.[10,12]

MRN as a diagnostic modality

MRN-based radiographic signs have been named, such as the "bag-of-worms" in the case of plexiform neuromas.[2,6] Other signs, such as the "split fat" sign on T1-weighted MR and the "target tail" sign on T2-weighted MR have also been described. MRN can be reliably used to diagnose a neurogenic tumor and differentiate this from a schwannoma.[1] NF-1 and plexiform neurofibromas are at significant risk for transformation to malignant peripheral nerve sheath tumors (MPNSTs). MPNSTs have been shown to display key characteristics that can be indicative of malignancy.[2,6,15] These include infiltration into the surrounding tissue, areas of necrosis, disrupted fascicular patterns, enhanced peripheral tumor tissue, intratumoral cystic lesions, and an increased size >5 cm, and perilesional edema.[14]

CONCLUSION

MRN is an imaging modality specifically designed for visualization of peripheral nerves. Here, MRN identified NF-1 in a patient presenting with multiple neurofibromas in both the right brachial plexus, and lumbar plexus. On the basis of multiplicity of disease, it was elected to pursue nonsurgical management in this patient.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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