Neuromuscular Choristoma of the Sciatic Nerve

Abstract

Neuromuscular Choristomas are rare benign tumors characterized by their composition as well as their location. They are typically composed of mature skeletal muscle tissue elements that are intermixed with mature neural tissue elements. This condition is extremely rare as is noted by its prevalence. To date, only about forty cases of neuromuscular choristomas have been reported with only five specific to the sciatic nerve. Neuromuscular choristomas often present with progressive neuropathy, undergrowth of the affected limb, and limb length discrepancy. In the sciatic nerve (the most common location of NMC), this may also lead to specific manifestations in the distal limb, including progressive neuropathy, a shortened atrophic limb, dorsal contracture of the toes, and a cavovarus foot.

Introduction

Neuromuscular choristomas of the sciatic nerve are unique in nature. This presents diagnostic challenges as well as opportunities for diagnosis. While these tumors are a mix of musculoskeletal tissue as well as neural tissue, the neural tissue in particular is generally composed of both myelinated and unmyelinated tissues. These lesions are composed of striated skeletal muscle fibers that are admixed with mature nerve fibers entering around and between nerve fascicles. They are also known by other names such as: neuromuscular hamartoma’s, peripheral nerve tumors and benign triton tumors. Terminology depends largely on the location of the tumor and the one doing the research. The term choristoma refers to normal tissue found in an abnormal location. Hamartoma is the term describing abnormal tissue found in a normal
They present typically as solitary lesions in childhood and have been reported to be associated with cranial nerves or large peripheral nerves such as the brachial plexus, median nerve, and sciatic nerve. Most often these lesions are found in major nerves. However, smaller peripheral nerves and cranial nerves can be affected. Signs and symptoms of NMC in the sciatic nerve include:

- Leg length discrepancy
- Weakness
- Sensory abnormality
- Neuropathic pain
- Ipsilateral cavus foot
- Gait abnormality
- Paresthesia

With so few cases, the pathogenesis of NMC is controversial and unclear. Open biopsy has been the most common method for diagnosis. However, this can lead to fibromatosis and even more difficulties for the patient.

**Fibromatosis**

Fibromatosis is a non-inflammatory and non-metastasizing generation of mature fibrous tissue. It is characterized by production of intercellular collagen and proliferation of fibroblasts, and grossly resembles scar tissue. It is also known as a desmoid tumor and is one of the rarest tumors. Diagnosis of aggressive fibromatosis is made by Magnetic Resonance Imaging. It
presents as an irregular, locally infiltrated mass. It may progress along the fascial planes and long axis of the nerve.

The most common method for diagnosing NMC is through a biopsy of the lesion. According to several studies, these biopsies may lead to or be a catalyst for the development of fibromatosis.\textsuperscript{2,6} There is evidence to support the notion that fibromatosis and NMC has a direct relationship. This is found in a study performed by Hebert-Blouin et al,\textsuperscript{6} in their observation that all patients with NMC who developed an aggressive fibromatosis did so at the site of their NMC. Hebert-Blouin et al\textsuperscript{6} identified ten patients with NMC. Of the six patients who did not develop this tumor, only one was adequately followed up. Of these cases, four had definite or suspected aggressive fibromatosis. Diagnosis of these four cases were made by open biopsy or CT-guided biopsy. “Between 4 months to 8 years postoperatively, all patients presented with a new enlarging mass in the vicinity of the NMC and the biopsy site.” \textsuperscript{6(p2)}

Another study performed by Niederhauser et al\textsuperscript{2}, discusses cases where the patients were diagnosed with NMC through biopsy. Of the six cases observed, five received post-biopsy Magnetic Resonance Imaging (MRI). These follow-ups ranged from six months to thirteen years after diagnosis. A fibrosmatosis developed in four of the five cases. The case that did not develop this mass had received a subtotal resection of involved brachial plexus and while this did not produce fibromatosis, it did result in neurological dysfunctions.\textsuperscript{2}

Radiation and chemotherapy, multiple operations, and even amputation is often required for treatment of aggressive fibromatosis.\textsuperscript{2} While it is true that fibromatosis can occur in patients without surgery, as noted above it is believed that open biopsy is a preventable risk factor for fibromatosis in attempting to diagnose NMC. As is also stated above, the association between NMC and fibromatosis is causally related to open biopsy. It is believe that open biopsy acts as a
catalyst to the development of fibromatosis, making it a preventable risk factor. “Biopsy should not be performed lightly or out of curiosity, because its risks are not insignificant. We recommend a ‘no-touch’ approach to NMC.”7(p2)

MRI

To minimize the risk of developing fibromatosis while still being able to diagnose NMC with confidence, MRI should be used in all pre-operative cases as well as the primary diagnosing modality for NMC. This also complies with the “no touch” recommendation outlined above. In fact, in multiple studies examined that were attempting to diagnose NMC, both T1 and T2 weighted MRI studies were used.2-4,8,9 T1-weighted images are useful for assessing central nervous system tissue, identifying fatty tissue and for post-contrast imaging. T2-weighted images are more indicative of magnetization decay and this image weighting is useful for detecting edema, axon fibers, and assessing where zones of similar kinds of tissue intersect. The unique nature of the composition of NMC tissue (muscle, fat, axons, and various other tissues as described previously) makes this type of imaging indispensable in diagnoses. A typical MRI reading for NMC should include a description of the strands of fat, thickened neurological tissue, fibrous tissues, and striated muscle tissues all of which are commonly intertwined in NMC but with definitive borders as well. No other imaging method can describe NMC with this much clarity and that is why it is so critical to the diagnoses.

Case Report

History
A 10-year-old female presented with progressive right lower extremity deformity manifest by a malformed foot including the beginnings of muscle atrophy. The patient first noticed that one shoe was much tighter than the other and asked her mother to offer her opinion. Her mother inspected it and noticed that the left foot was about a shoe size and a half larger than the right. Upon closer examination, the right foot appeared to have a higher arch, and the toes on the right foot were curved strangely when compared to the left foot. The only discomfort the patient mentioned at the time was that the left shoe did not fit correctly. About two years later, the patient noticed weakness and little feeling in her toes. She had no history of scoliosis, back pain, or sensory disturbance.

**Examination**

The patient’s right lower limb was 1 inch shorter than its left counterpart. The patient had shortening of the forefoot, and noticeable pes cavus with contractures of the toes. A small amount of atrophy to the gastrocnemius muscle was noted. Inversion and eversion were difficult on the right foot. Quadriceps and hamstring strength were normal. Knee flexion and extension also appeared normal. The patient was able to walk on her toes but had difficulty walking on her heels. Normal capillary refill to her toes was observed. There was no obvious dysmetria or ataxia noted. Vibratory sense on the right foot was less than the left and some decreased touch sensation in non-specific patterns was also detected.

Attempting to further investigate the specifics of this lesion, MRI imaging was used with both T1 and T2 weighted techniques being used. A fusiform thickening of the sciatic nerve continued down into the mid-thigh. At that junction the nerve returns to normal size and appearance (see Figure 1 and 2). Follow up MRI interpreted results were as follows: “Magnetic
resonance imaging revealed right sciatic nerve abnormalities that began in the nerve roots of the lumbosacral trunk and extended through the nerve roots of S1, S2 and extending along the sciatic nerve over the piriformis into the posterior thigh” (MRI report, 11/06/2012). It was noticed that diffuse atrophy and subtle T2 hyperintensity of the right gluteal musculature, the posterior thigh musculature, the hamstring portion of the adductor magnus, and the quadratus femoris, piriformis, and aburator internus muscle. The right S2 nerve root sleeve was enlarged.

Further confirming the lesion, an electromyography (EMG) was performed. The EMG was used to assess the control of the nerve cells to the muscle and the overall health of the muscle. Electrical signals were sent by the motor neurons which caused the muscle to contract. These signals were then translated into graphs, sounds and numerical values and presented for interpretation. For this patient the study was essentially normal. Nonspecific findings of possible chronic neurological changes in the right tibialis anterior muscle with an absent central nerve response in the right leg were both demonstrated by the EMG.

**Surgical Intervention/Open Biopsy**

To confirm the suspicion from the imaging and for a definitive diagnosis, an open biopsy was recommended. After a lengthy discussion of the risks associated with the biopsy, consent was obtained and a biopsy was performed. An 8-cm incision was made distal to the gluteal crease and carried down to the facial plane. The gluteus maximus muscle belly was identified and was retracted proximally but not cut. The sciatic nerve was identified and neuroplasty was performed. The nerve was dissected out. Great care was taken to inflict minimal trauma on the nerve. The nerve was incised in line with its fibers and a small piece of atypical appearing non-fascicle mass was excised. This was sent to pathology. No single area of nodularity was noted;
however, the nerve was said to appear abnormal in all areas. Biopsy of the right sciatic nerve demonstrated evidence of possible chronic injury as well as ongoing denervation and re-innervation of the sciatic nerve. The biopsy also revealed “mature skeletal muscle in association with peripheral nerve axons” (biopsy report, 11/8/2010) consistent with neuromuscular choristoma of the sciatic nerve.

**Pathological**

The biopsy specimen was examined microscopically and was found to contain benign skeletal muscle with a focal increase in the soft tissue between some of the muscle fibers. This intramuscular stromal expansion appears to be due to prominent peripheral neural tissue in some areas, and to fibrous tissue in others. High-grade atypia was not seen. Immunohistochemical stain for S-100 protein was performed with appropriately reactive positive and negative controls. This highlights focally prominent peripheral nerve tissue between muscle fibers. Neuromuscular choristoma was diagnosed based on the histological features of the specimen.

**Treatment**

There is not a cure for neuromuscular choristoma and there is no effective treatment currently available besides serial MRI’s to monitor progression of the lesion. Several things that have been shown to help are appropriate pain management to keep the patient comfortable, physical therapy to continue to stimulate the limb to maintain tone and strength as tolerated, and counseling support to help the patient cope with challenges of this condition. It is typical for the clinicians to order an MRI once a year to monitor the sciatic nerve and make sure fibromatosis does not form. This patient also has had surgery to fuse her growth plates on the unaffected
lower leg to help maintain symmetry of both of her legs as she continues to grow and to minimize the leg length discrepancy. The pain in her foot also continues to worsen over time. To combat this, she uses lidocaine patches on the bottom her foot for some relief.

**Conclusion**

Neuromuscular choristomas are rare and debilitating tumors requiring good clinical examination, histology analysis, as well as appropriate imaging techniques to properly diagnose. This especially applies to neuromuscular choristomas of the sciatic nerve or “NMC.” The tissues in NMC are unique in the sense that they are not composed of abnormal neoplastic or foreign tissues like most tumors are, but are composed of ordinary nerve, muscle and fat tissues that are intertwined in nerves where they should not be. This causes problems in the entire area that the involved nerve is designed to innervate. In this case, the sciatic nerve, and the entire leg that the sciatic nerve is servicing. This also makes NMC’s challenging to diagnose. Open biopsy can definitively diagnose, but can also lead to fibromatosis causing even more problems for the patient. Therefore, T1 and T2 weighted MRI’s are the mainstays in imaging as they can identify these types of normal tissues and aid in properly diagnosing this challenging condition without the risk of creating fibromatosis. This condition is difficult to treat effectively as well, since any attempt to treat the normal muscle, nerves and fat tissues found in NMC would also harm the rest of the body. Early detection, MRI’s to assess the growth of the tumor, pain control for the patient, physical therapy to maintain muscle tone, specialized surgery and counseling can all help the patient in making the best of this difficult condition.
References


Figure 1. Magnetic resonance imaging obtained preoperatively of the lower extremity. Axial T1-weighted image of 10-year-old female demonstrates a right sciatic neuromuscular choristoma (arrow), which appears as a fusiform enlargement of the sciatic nerve.
Figure 2. Magnetic resonance imaging obtained preoperatively of the lower extremity. Axial STIR-weighted image of 10-year-old female demonstrates a right sciatic neuromuscular choristoma (arrow), which appears as a fusiform enlargement of the sciatic nerve.