Addendum: Evidence supports a “no-touch” approach to neuromuscular choristoma

To The Editor: Our article called attention not only to an association between these 2 rare disorders, neuromuscular choristoma (NMC) and fibromatosis, but also to the role of biopsy in that association (Hébert-Blouin MN, Scheithauer BW, Amrami KK, et al: Fibromatosis: a potential sequela of neuromuscular choristoma. Clinical article. J Neurosurg 116:399–408, February 2012). Since the publication of this article, the association has been further strengthened by additional evidence provided by 2 patients evaluated at our institution as well as information from a radiological review and a case report.

An 11-year-old boy (Case 1) presenting at another institution with a several-year history of buttock and lower-extremity pain was found to have a unilateral sciatic neuropathy, hammer toes, and a smaller leg and foot. Imaging revealed a sciatic nerve lesion (Fig. 1A and B) extending over 30 cm in length. He underwent first an ultrasound-guided biopsy, then a CT-guided biopsy (Fig. 1C and D), and finally an open biopsy, the last biopsy confirming a histological diagnosis of NMC. This patient was recently reported on by the original group for his NMC, but follow-up was not provided. We then saw him in consultation and have witnessed aggressive fibromatosis in evolution. At our initial evaluation (6 months postbiopsy), MRI findings concerning for early development of fibromatosis were seen along the previous biopsy track and surrounding the sciatic nerve in that area (Fig. 1E and F). We recently reviewed 18-month postbiopsy MR images demonstrating aggressive fibromatosis, with the epicenter being at the biopsy site (Fig. 1G–I).

Fig. 1. Case 1. A and B: Axial T1-weighted (A) and T2-weighted (B) MR images with fat suppression at the level of the upper thigh showing the lesion associated with the right sciatic nerve. Note the band of low signal surrounding the lesion suggesting fibrous tissue (solid white arrows). The normal left sciatic nerve (open arrows) is included for comparison. C: Axial image from a CT-guided biopsy showing the needle within the sciatic lesion in the upper thigh. The image is reversed as the patient was imaged in the prone position for the biopsy. D: Axial CT image obtained 1 month after open biopsy showing the surgical defect to the skin with air bubbles (arrows). E and F: Axial T1-weighted MR images obtained before (E) and after (F) administration of contrast medium with fat suppression in the upper thigh at the level of the biopsies showing enlargement of the lesion and increasing areas of low signal compatible with fibrous tissue surrounding the sciatic nerve (arrows). Note enhancing fibrous tissue along the surgical approach reflecting fibrosis (arrowheads). Focal enhancing fibrous tissue within the lesion is also seen (asterisk). G and H: Axial T1-weighted MR images with fat suppression obtained before (G) and after (H) administration of contrast medium 1 year after E and F, showing the florid increase in soft tissue within and around the lesion in the sciatic nerve with increasing areas of low signal, all consistent with progressive fibrosis (asterisks). Note the prominent enhancement on the postcontrast images along the original surgical tract (arrowheads). The normal fascicular appearance of the original lesion has been lost (compare with A). I: Sagittal T1-weighted postcontrast MR image with fat suppression showing the longitudinal extent of the enhancing fibrous tissue with encasement of the nerve (asterisk). The original surgical approach is still visible with enhancement along the tract (arrowhead).
A 10-year-old girl (Case 2) had a similar presentation: a short foot (Fig. 2A), sciatic neuropathy, an enlarged sciatic nerve on MRI (Fig. 2B and C) and a diagnosis of NMC after an open biopsy performed at another institution 2 months prior to our evaluation. She is being followed up closely for the development for fibromatosis. Curiously, both of these patients developed keloid following biopsy. The keloid formation in these 2 patients is reminiscent of that described by our group in patients operated on for lipomatosis of nerve (LN).\(^3\) We believe that the keloid formation (Fig. 3A and B) is an external manifestation of the same abnormal fibroproliferation involved in the formation of the fibromatosis.

Based on our recognition of a strong association of NMC and fibromatosis after biopsy, we recently analyzed our own cases and those reported in the literature to identify radiological features that could be used to diagnose NMC without the inherent risks of a biopsy and distinguish it from LN, its major differential diagnosis.\(^5\) This study revealed that both conditions have characteristic clinical and imaging features. The MRI features of LN have been well described and are pathognomonic.\(^4\) Neuromuscular choristoma also has characteristic MRI features, which include fusiform enlargement, less than 50% intralesional fat component, good demarcation from surrounding structures, MRI signal closely mimicking muscle on all sequences, and the absence of cystic spaces or nodular enhancement on prebiopsy imaging. Both clinically and on imaging, NMC is associated with nerve-territory soft tissue and/or bony undergrowth, whereas LN is associated with overgrowth.

An intriguing case report has described the association of mandibular desmoplastic fibroma—the bony counterpart of soft-tissue fibromatosis—with multiple foci of neuromuscular hamartoma.\(^7\) This case, together with one described by Oeppen et al.\(^6\) (despite its not being fully substantiated\(^1\)), suggests that these intracranial lesions, similar to those in the periphery, may also develop fibromatosis.

The association between NMC and fibromatosis is causally, not coincidentally, related. We believe that percutaneous or open biopsy acts as a catalyst to the development of fibromatosis. Thus, it is 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, and for that matter, for LN. Both conditions are benign and can be diagnosed confidently when characteristic clinical and imaging features are considered. For NMC, a “no-touch” approach may minimize the risk of fibromatosis either in the short or long term. The data in this follow-up letter are compelling: a temporal and spatial relationship between the biopsy and the development of fibromatosis is evident.

Marie-Noëlle Hébert-Blouin, M.D.
Kimberly K. Amrami, M.D.
Robert J. Spinner, M.D.
Mayo Clinic
Rochester, Minnesota

Disclosure

Dr. Spinner reports that he is a consultant to Mayo Medical Ventures.

References