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

To The Editor: Our article1 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 review5 and a case report.6

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,2 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).5 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 substantiated1), 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.

**Disclosure**

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

**References**

Medulloblastoma and the inferior medullary velum

To The Editor: Tubbs et al. have published an interesting anatomical study in your journal recently about the microscopic and macroscopic morphology of the inferior medullary velum (Tubbs RS, Bosmia AN, Loukas M, et al: The inferior medullary velum: anatomical study and neurosurgical relevance. Laboratory investigation. J Neurosurg 118:315–318, February 2013).8

Countering what was almost a mainstream notion in neuroscience,4,5 Tubbs et al. based on their findings, argue against hypothesizing that the inferior medullary velum is the origin of medulloblastoma. Because we agree with Tubbs et al. in challenging this popular concept regarding the relationship between inferior medullary velum and medulloblastomas, we would like to provide more details about the embryological development of the inferior medullary velum to show that if we couple the embryological development with well-known radioanatomical features of medulloblastomas, we can rationalize further that these lesions are unlikely to arise from the inferior medullary velum. Instead we can establish a new interpretation for this relationship and challenge further that these lesions are unlikely to arise from the inferior medullary velum. Instead we can establish a new interpretation for this relationship and challenge further that these lesions are unlikely to arise from the inferior medullary velum.

As an anatomical structure that belongs to the roof of the fourth ventricle,2,3,6,10 the inferior medullary velum has a distinctive embryological development from the cerebellum and the floor of the fourth ventricle.1 Even though the inferior medullary velum is derived from the rhomboid lip, which is one of the germinal zones of the developing brain, the rhomboid lip itself is differentiated from the early stage of development into primary and secondary lips. Primary lip gives origin to the cerebellar plate, and secondary lip gives origin to the roof plate. A detailed description of the rhomboid lip was part of the remarkable work of Joseph A. Blake and was published in a comparative neurology journal in 1900.

In his work, and by studying the embryological development of the hindbrain of several species, Blake illustrated the features of primary and secondary rhomboid lips and their development in the hindbrain. According to his work, the lips coincide at the level of the lateral recess and enter the acoustical ganglia together, are continuous with the auditory tract region, and are fused at the level of the calamus. Fusion of the secondary lips creates the obex. The line of fusion between the secondary lip and medulla is called the ligula. The secondary lip anatomy varies greatly among individuals. Secondary lip lies between the ligula and ala cinerea (vagal trigone). The lip refolds prior to reaching the lateral recess. At the pontine level the secondary lip might appear to be contributing to the enlargement of the middle cerebellar peduncle.1

Regarding the relationship between the inferior medullary velum and medulloblastoma, the following points argue against considering the inferior medullary velum as the origin of these tumors.

1) The radiological characteristics of medulloblastoma and their pattern of extension and infiltration makes considering this lesion as originating from the region dorsal to the inferior medullary velum more logically consistent. In other words, it is well known that medulloblastomas infiltrate the floor of the fourth ventricle late in their course and rarely go beyond the foramen of Luschka, whereas the ependymomas infiltrate the floor early in their course even though medulloblastoma is more aggressive pathologically than ependymoma.3,5,11

2) Based on the above-mentioned type of extension and given the fact that the primary lip undergoes more complicated embryological development, medulloblastoma most likely develops from undifferentiated cells located in the primary lip.

If we consider that the origin of medulloblastoma is the primary lip, and ependymoma arises from the secondary lip, it will be easier to explain the pattern of extension of both lesions. Also, it seems that the inferior medullary velum plays the role of a natural anatomical barrier that prevents the medulloblastomas from extending toward the floor, at least early in the course of these tumors. In the same way, the inferior medullary velum prevents the extension of ependymoma toward the vermis of the cerebellum.

The anatomy and the embryology of inferior medullary velum described here and in the paper by Tubbs et al., as well as the radioanatomical patterns of extension that tumors of the fourth ventricle usually follow, could be used to refine our neurosurgical techniques to deal with tumors of the fourth ventricle. In this regard, the inferior medullary velum could be used as a natural dissection plane when dealing with tumors of the fourth ventricle; that is, to circumscribe and determine the possible margins of these tumors rather than cutting it from the beginning.

Finally, we would like to emphasize that we are pleased that Tubbs et al. drew attention to some aspects of the embryological development of this hidden anatomical structure, because we believe that coupling the embryological development with the anatomical features has a significant potential role in explaining at least some
aspects of the development and the behavior of CNS tumors.

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Disclosure
The authors report no conflict of interest.

References


RESPONSE: We appreciate the comments of Drs. Salma, Lin, and Fassett and agree with their conclusions. Based on our anatomical study and the embryological study of Blake,1 it appears that the inferior medullary velum is not a likely source of medulloblastoma.

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Please include this information when citing this paper: published online May 24, 2013; DOI: 10.3171/2013.2.JNS13237. ©AANS, 2013