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Co-occurrence of subcutaneous myxopapillary ependymoma, dermal sinus tract, and filum terminale lipoma: a review of the pathobiology of caudal spinal cord development and spinal cord tethering. Illustrative case

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BACKGROUND Myxopapillary ependymoma (MPE) is typically benign and found in the conus medullaris and/or filum terminale, although rare cases of subcutaneous and extra-axial MPE have been reported. The co-occurrence of MPE, tethered cord syndrome (TCS) with lipoma of the filum terminale, and a dermal sinus tract is extremely rare, with only 6 reported cases in the literature. Here, the authors present the first case, to their knowledge, of an extra-axial, subcutaneous MPE co-presenting with TCS, lipoma of the filum terminale, and a dermal sinus tract and discuss the underlying pathobiology.

OBSERVATIONS A 14-month-old male who presented for evaluation of a dermal sinus tract underwent magnetic resonance imaging, which revealed a tethered cord with associated lipoma. At 14 months, the patient underwent spinal cord detethering with resection of his sacral dimple and sinus tract. Histopathological evaluation revealed an incidentally found MPE within the dermal sinus tract.

LESSONS The authors review the underlying biology of MPEs, tethered cord syndrome, and dermal sinus tracts, and explore possible points of convergence within the developmental pathways that may result in this unique concomitant presentation. Additionally, they suggest that extra-axial MPE may be underappreciated and underdiagnosed; this case suggests that extra-axial MPE may be only effectively diagnosed with histological studies.

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KEYWORDS myxopapillary ependymoma; tethered cord syndrome; spinal cord tethering; pediatrics; dermal sinus tract

Myxopapillary ependymoma (MPE) is a molecularly distinct subtype of ependymoma with a predilection for the conus medullaris and/or filum terminale (FT). Although ependymomas characteristically originate from the ependymal surface of the ventricular system or spinal central canal, rare cases of subcutaneous MPE and extra-axial MPE have been previously reported.[1]

Tethered cord syndrome (TCS) is a neurological disorder most often characterized by a low-lying conus medullaris and adhesions that limit the movement of the cord within the thecal sac and spinal canal. The presence of a dermal sinus tract and/or a thickened, fat-infiltrated FT are commonly associated with TCS.[2]

The co-occurrence of MPE, TCS with lipoma of the FT, and a dermal sinus tract is extremely rare, with only 6 reported cases in the literature. Here, we present the first case, to our knowledge, of an extra-axial, subcutaneous MPE co-presenting with TCS, lipoma of the FT, and a dermal sinus tract and discuss the underlying pathobiology.

Illustrative Case

A sacral dimple was noted in a 4-month-old male born via vaginal delivery at 39 weeks' gestation. Magnetic resonance imaging (MRI) was performed to evaluate for possible spinal cord tethering

ABBREVIATIONS FT = filum terminale; MPE = myxopapillary ependymoma; MRI = magnetic resonance imaging; TCS = tethered cord syndrome.
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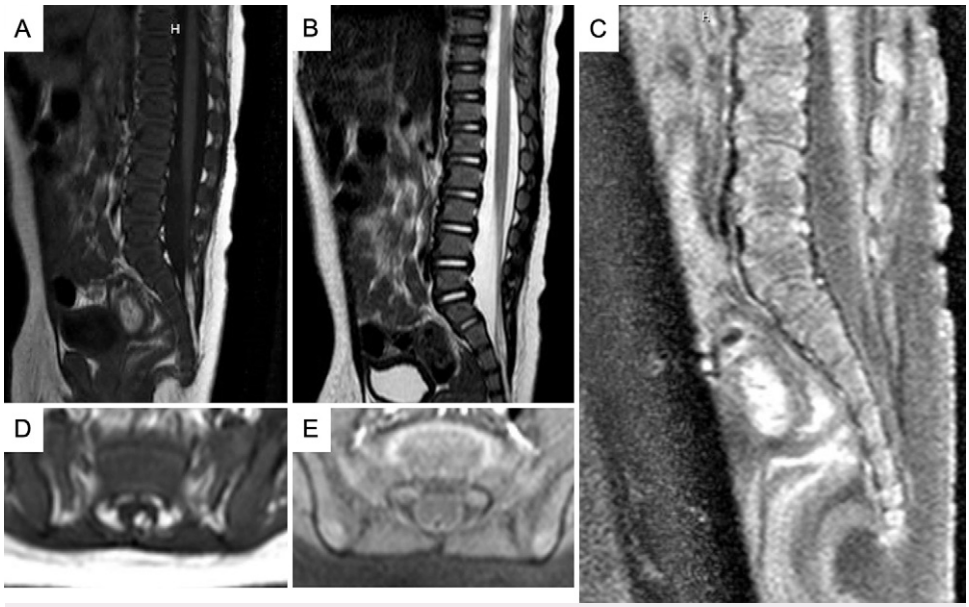

FIG. 1. Preoperative imaging with an incidentally found myxopapillary ependymoma within a resected dermal sinus tract: sagittal T1-weighted (**A**), sagittal T2-weighted (**B**), sagittal T1-weighted fat saturated postcontrast (**C**), axial T1-weighted (**D**), and axial T1-weighted fat saturated (**E**) MRI.

and revealed a low-lying conus with a T1 hyperintense, thickened, fatty filum measuring 4 mm in diameter and extending caudally from S1. The signal was suppressed with fat-saturation techniques and did not enhance (Fig. 1). A sacral defect and fibrous tract were also noted.

The patient presented at 11 months for evaluation and routine evaluation after moving to the region. On examination, he had no neurological deficits. The sacral dimple was located just superior to the gluteal cleft without redness or drainage. After discussing treatment options, the parents elected for the patient to undergo spinal cord detethering. At 14 months, the patient underwent excision of his dermal sinus tract and, secondarily, laminectomy for spinal cord detethering. Intraoperatively, an elliptical incision was made around the sacral dimple/dermal sinus tract, and the tract was dissected down to the level of the sacrum, where it terminated without an apparent bony penetration or connection to the thecal sac. The dermal sinus tract was then resected and sent for histopathological examination. A partial inferior L5 laminectomy was then performed, and a 1-cm midline dural incision was made. With the operative microscope, a single adherent sacral nerve root, which activated the sphincter on stimulation, was dissected off the filum. No intradural abnormalities were noted with microsurgical dissection. The filum was then tested with stimulation, and eventually cauterized and transected, with recoil of the rostral end. Routine closure was performed. His operative and postoperative course were without complications, and he remained neurologically intact on discharge from the hospital and at last follow-up in our clinic 15 months postoperatively (his family subsequently moved out of the region, but radiological follow-up continued as described below).

Histopathological evaluation of the resected sacral dimple and sinus tract showed evidence of a papillary and myxoid lesion located in the deep dermis and abutting the subcutis with occasional palisading of the papillae. Immunohistochemical analysis revealed strong positivity for glial fibrillary acid protein (GFAP) and vimentin, with weak cytokeratin staining of the surface cells. These findings were consistent with microextramedullary MPE.

Due to the pathological diagnosis of extramedullary MPE, the patient underwent complete neuroaxis imaging without evidence of additional lesions. Lumbar spine MRI at 6 months post-detethering demonstrated postsurgical changes with fatty remnant along the anterior surface of the cord and a small residual fatty nodule at S2 (Fig. 2A and B), which remained stable on subsequent imaging (Fig. 2C). The patient was monitored with serial imaging for 5 years without recurrence, with the patient subsequently being lost to follow-up.

Discussion

Observations

This case demonstrates several unique features that provide insight into the underlying pathobiology of MPE, TCS, and dermal sinus tracts. The cooccurrence of MPE and TCS due to fatty filum is extremely rare. To date, there have been only 3 adult cases (28, 40, and 67 years old)[3–5] and 3 pediatric cases (9, 11, and 16 months old)[6–8] of TCS, FT lipoma, and concomitant intra-axial MPE reported. The natural history of these co-occurring lesions seems relatively indolent, with these patients generally living without complications after treatment.

Dermal sinus tracts arise from nondisjunction of the cutaneous ectoderm and neuroectoderm during spinal cord development, making dermal sinus tracts a relatively common manifestation in TCS.[9] It is believed that MPEs originate from embryonic ependymal cell rests, a glial population that is normally present in the FT. These ependymal cell rests are well circumscribed and without neoplastic characteristics.[10] Based on our literature review, there have been 43 reports of subcutaneous, extra-axial MPE since its first description in 1902.[1,11–15] Subcutaneous MPEs are hypothesized to arise

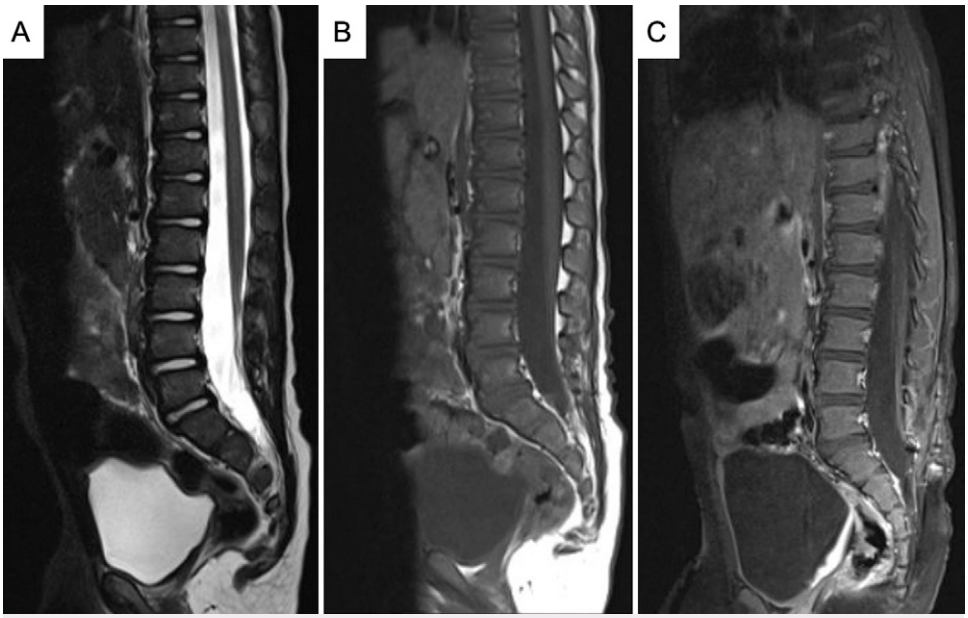

FIG. 2. Lumbar spine imaging performed after spinal cord detethering: 6-month postoperative sagittal T2-weighted (**A**), 6-month postoperative sagittal T1-weighted (**B**), and 15-month postoperative sagittal T1-weighted fat saturated (**C**) MRI.

from heterotopic ependymal cell rests located in the dermis and subcutis.[13,16–18] The caudal segment of the spinal cord, including the distal lumbar cord, conus medullaris, and FT, arises via the processes of canalization and retrogressive differentiation, both of which are less-organized processes than primary neurulation.[19] Disorganization during these processes may account for the heterotopic ependymal rests found in the FT and conus that can lead to MPE. While ependymal cell rests are relatively indolent,[10] subcutaneous MPEs arising from ependymal cell rests have been documented to recur and metastasize, with one case series reporting a metastasis rate of 17%.[13] As a result, treatment and prognosis depend on careful histological examination to distinguish between these histogenetically similar ependymal cell rests and pathological MPEs.

Lessons

Extra-axial MPE may be underappreciated and underdiagnosed. As in the case of our patient, on radiographic imaging it may be challenging to distinguish an extra-axial MPE from a dermal sinus tract. In our patient, the sacral lesion was only confirmed as an MPE upon biopsy, suggesting that extra-axial MPE may be only effectively diagnosed with histological studies. Although clearly rare, the incidence of extra-axial, subcutaneous MPE, dermal sinus tract, and FT lipoma with TCS may be underappreciated since histological analysis is not routinely performed in such cases.

While this case offers a unique clinical presentation of MPE, its limitations should be considered. The patient's follow-up spanned several years, but the extended natural history of this rare cooccurrence of lesions remains unclear. Nevertheless, this report serves as a lesson and reference as clinicians evaluate similar, concomitant presentations in future cases.

Although our patient's extra-axial, subcutaneous MPE, dermal sinus tract, FT lipoma, and TCS are seemingly unrelated, their etiologies suggest a convergence during neural tube development, with all three stemming from disruption of normal processes during caudal spinal cord development. This report and review of the underlying biology of these concomitant pathologies offers clinicians and researchers unique evidence regarding the etiology and effective diagnosis of similar, future cases.

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Author Contributions

Conception and design: all authors. Acquisition of data: Johnson, Xu, Mian. Analysis and interpretation of data: all authors. Drafting of the article: Johnson, Xu, Limbrick. Critically revising the article: all authors. Reviewed submitted version of manuscript: all authors. Approved the final version of the manuscript on behalf of all authors: Johnson. Administrative/technical/material support: Mian, Limbrick. Study supervision: Mian, Limbrick. Reviewed imaging: Mian.

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