Growing mammary choristoma masquerading as a lumbosacral lipomyelomeningocele in a pubertal girl

Case report

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The authors report, to the best of their knowledge, the first case of lumbosacral choristoma of breast origin, presenting in a young girl with lumbosacral lipomyelomeningocele. Although choristomas are considered to be benign, the regrowth of this mass when the patient was 15 and 16 years of age, and its involvement in the conus medullaris and cauda equina, warranted 2 additional resections with spinal cordotomy resulting in cessation of any further growth. The authors describe the case and provide a review of pertinent literature and a discussion of the mechanisms involving the development and growth of this lesion. (DOI: 10.3171/2011.6.PEDS10480)

KEY WORDS • mammary choristoma • lumbosacral lipomyelomeningocele • conus medullaris • cauda equina

Choristomas are benign tumors/malformations composed of histologically normal tissue in an abnormal location. This is in contrast to hamartomas, which are also benign tumors but which contain an abnormal/haphazard arrangement of cells and tissue components that are normally present at a site. Spinal tethering tumors containing heterotopic components have been reported, but the histological classification is not well established. Here, we report the first case of lumbosacral spinal choristoma of breast origin. The clinical presentation, radiological features, pathological findings, and differential diagnosis are discussed, and the relevant literature is reviewed.

Case Report

History. This 11-year-old girl presented to our center with progressive foot weakness (Grade 0/5 on the right and 3+/4 on the left) and bladder incontinence. Magnetic resonance imaging revealed a caudal lipomyelomeningocele (Figs. 1A and 2A). The patient had previously undergone resection of a midline lumbar subcutaneous lipoma overseas. At our institution, she underwent a tethered cord release and partial resection of the adipose mass, which was found to extend deep into the conus medullaris. Tissue was not submitted for pathological evaluation. Her neuromuscular problems stabilized until she was 15 years of age, when she presented with further deterioration in bladder and lower-limb motor and sensory function. Surprisingly, MR imaging revealed that the lipoma had grown and contained cystic-looking cavities within the adipose tissue. Significant debulking was performed, but this was limited by the fact that the abnormal tissue seemed to heavily involve the conus and cauda equina, making a gross-total resection hazardous. Pathological examination revealed a choristoma rather than a typical spinal lipoma. The patient was followed up closely with serial MR imaging, which demonstrated rather rapid enlargement of components of the residual cystic mass (Fig. 1B) with concomitant further deterioration in lower-limb function within a year of the third surgery. The decision was made to perform a near-total resection of the tumor and a spinal cordectomy distal to the most caudal functional level, as determined by electromyographic stimulation of the cord (Fig. 2B). Postoperatively, the patient had a mild decrease in her lower-limb motor function but remained ambulatory with crutches.
Gross and Microscopic Description. The resected lipomatous tissue from the third surgery revealed several large, clear, lobulated masses. Grossly, the tissue did not resemble an epidermoid cyst. On H & E–stained tissue sections, we observed masses of benign adipose tissue containing abundant glandular elements, frequently surrounded by strands of benign smooth-muscle cells. A few fascicles of peripheral nerve were present at the periphery of the lesion and 1 fascicle exhibited traumatic changes. The gland lumina ranged from small to large cystic dilations and were lined by a single layer of cuboidal to low columnar cells lacking goblet features (Fig. 3A and B). Myoepithelial cells were present basally and confirmed with p63 antibody immunolabeling (Fig. 3E). The majority of the glands appeared to be eccrine, although focally, apical knobs are also noted, suggesting possible apocrine properties. In some areas, the glandular elements were crowded and lay back to back; however, nuclear atypia was not evident. There were also areas with features of sclerosing adenosis, a microscopic focus of epithelial hyperplasia, and only 1 gland lined by a few ciliated cells. Very rare mitoses were encountered. Strong immunolabeling with estrogen receptor (Fig. 3C) and progesterone receptor (Fig. 3D) antibodies was observed. Immunolabeling with Ki 67 revealed a small number of positively labeled nuclei scattered throughout the specimen, and 3 glandular elements contained several cells with multiple nuclei labeled (Fig. 3F). The excised tissue from the fourth surgery was about twice the volume of material as that from the third. It also consisted of benign fibroadipose tissue containing glandular elements similar to the previous material, but with less crowding. Instead, at the periphery, in addition to fascicles of peripheral nerves, bundles of atrophic and denervated skeletal muscle were found. Here, the mixture of glandular elements and fibro-
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adipose tissue extended into the skeletal muscle. Ki 67 immunolabeling of this specimen revealed a scattered population of positively labeled nuclei but no focally prominent labeling. Tissues from the third and the fourth surgeries lacked other glandular or epithelial elements, and there was no suggestion of endodermally derived tissue, central nervous tissue, bone, or cartilage.

Based on our findings, we diagnosed a choristoma of the conus medullaris.

Discussion

This is the first reported case of mammary choristoma in the conus medullaris. In this case, histology and positive p63, estrogen receptor, and progesterone receptor immunolabeling clearly demonstrated that the glandular components lined by epithelial and myoepithelial cells in the spinal mass were those of breast tissue. Although there was entrainment of peripheral nerve fascicles in both specimens and the skeletal muscle in the last specimen, cytological features, low mitotic activity, and proliferative index of the lesion indicated a benign process. Both tissues revealed a moderately organized mammary tissue without any other organ or tissue elements within. At the periphery there was entrainment of lumbosacral nerve rootlets, rare sensory ganglia, and the paraspinal skeletal muscle. None of these latter elements were viewed to be part of the lesion and thus the lesion was not consistent with a teratoma. Traditionally, congenital midline lumbar spinal tethering tumors have been spinal lipomas because they are mostly composed of mature adipose tissue.8 However, several studies have reported that in addition to lipomatous tissue, a variety of heterotopic components of ectodermal, mesodermal, and/or endodermal origin, such as renal corpuscles, respiratory, or digestive epithelium-lined cysts, and even a nodule of cerebellar parenchyma, have been identified in lipomatous lesions.6,11 Histological classification of the spinal tethering tumors is not well established. Different names such as lipoma,6 hamartoma,1,8,11 choristoma,7,10 and teratoma2,4 have been applied to these lesions.

The most important differential diagnosis for spinal choristoma includes spinal hamartoma and teratoma. In general, only benign lesions composed of disordered mature tissue normally present in the affected location should be considered to be pure hamartomas. Accordingly, spinal hamartomas contain mostly mature and well-differentiated tissue from ectodermal and mesodermal layers. Similar to spinal choristomas, spinal hamartomas do not carry the potential for malignant transformation.1 Alternatively, if tissues from all 3 germ layers are identified in the lesion, a teratoma with malignant potential should be considered. Differentiating the aforementioned entities is important in the evaluation and management of the individual cases.

By definition, a choristoma is a mass of tissue histologically normal for an organ or part of the body other than the site at which it is located. Thus, in this case, we favor the designation of choristoma over hamartoma, which is defined as a mass of mature, disorganized cells and tissue components indigenous to a given site. In our patient, tissue specimens from the last 2 surgeries exhibited adipose tissue with glandular elements that, by histological and immunological features, were identified as mammary tissue. Some glands are dilated, with ductal features, and surrounded by bands of smooth-muscle cells. Even though samples of the first 2 resections were not evaluated microscopically, we suspect that they did contain choristoma. Because the tissue samples had benign histological features, we suspect that regrowth of this mass was at least in part secondary to puberty-related hormonal changes, influencing the growth of mammary glandular elements. The entrainment of lumbosacral nerve rootlets, rare ganglion cells, and surrounding skeletal muscle are likely related to both multiple surgeries at this site and growth stimulation of the lesion during puberty. The level of tissue organization in this lesion and the lack of other tissue components do not support a diagnosis of teratoma in this case. This being said, the occurrence of choristoma in the conus medullaris is extremely rare. Kurman and associates8 first described a lumbosacral choristoma of Müllerian origin. Since then, choristomas with endometrial tissue have been reported in 2 cases.7,10 The mechanism for the formation of spinal lipomatous lesions is not well understood. It has been proposed that the lesions arise from premature dysjunction of the cutaneous and neural ectodermal layers prior to neural tube closure, after which mesenchymal cells that migrate through the focal site of dysjunction to the dorsal aspect of the placode differentiate into adipose tissue, forming the lipomatous mass.3,5 However, the pathogenesis of choristoma remains elusive and the embryological origin of heterotopic tissue in the lumbosacral spinal canal has not been fully explored.

Conclusions

We report a case of spinal choristoma of breast origin that masqueraded as a lumbosacral lipomyelomeningocele. While it is generally thought that spinal choristomas have an excellent prognosis with little, if any, potential for excessive growth or malignant transformation, this particular tumor seemed to encompass the conus medullaris and cauda equina, and it grew at a noticeable rate between resections—behavior akin to a more aggressive tumor. This unusual tumor behavior eventually led to a near-total resection and a spinal cordectomy below the most caudal functional level.

Disclosure

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

Author contributions to the study and manuscript preparation include the following. Conception and design: all authors. Acquisition of data: all authors. Analysis and interpretation of data: all authors. Drafting the article: all authors. 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: Salamat. Administrative/technical/material support: all authors. Study supervision: Salamat.
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