Synovial sarcoma of the cauda equina

Case report

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Primary synovial sarcoma originating from the cauda equina is extremely rare. Only one case, involving an 11-year-old girl, has been reported. The authors describe the case of a 23-year-old woman with a primary synovial sarcoma of the cauda equina.

The patient visited a local hospital and described a 2-month history of low-back pain. She was referred to the authors’ hospital for further evaluation. On physical examination, she had a straight-leg raising result of 70° bilaterally. Motor examination revealed Grade 4/5 strength in the bilateral extensor hallux longus muscles. There was normal sensation to light touch and vibration in the lower extremities. Sagittal Gd-enhanced T1-weighted MR imaging demonstrated an intradural, extramedullary, and uniformly enhancing mass that extended from L-3 to L-4. The mass was totally resected and adjuvant local radiation therapy was administered. Reverse transcriptase polymerase chain reaction (RT-PCR) of a paraffin-embedded tissue sample revealed SYT-SSX fusion transcripts, and the diagnosis of synovial sarcoma was confirmed. Five and a half years after surgery, the patient is free of local recurrence and metastatic disease. The RT-PCR detection of SYT-SSX fusion transcripts played a key role in establishing the diagnosis of synovial sarcoma of the cauda equina. Complete resection of the mass with adjuvant local radiation therapy proved to be effective. (DOI: 10.3171/2011.10.SPINE11359)

Key words • synovial sarcoma • cauda equina • SYT-SSX gene • lumbar

SYNOVIAL sarcoma is a malignant tumor of mesenchymal origin that represents 5.8%–10% of all soft-tissue sarcomas.1,14 It usually originates in the extremities, especially around the knee joint.1,14,22 Less commonly, however, it has been reported to occur in nearly all sites including the heart,8 colon,12 liver,17 and nerves.23 In this report we describe an extremely rare case of synovial sarcoma of the cauda equina in an adult.

Case Report

Presentation and Examination. This 23-year-old woman presented to her a local hospital with a 2-month history of low-back pain. She was referred to our hospital for further evaluation. On physical examination, we documented a straight leg–raising result of 70° bilaterally. Motor examination revealed Grade 4/5 strength in the bilateral extensor hallux longus muscles. There was normal sensation to light touch and vibration in the lower extremities. The deep tendon reflexes were normal. The patient had no bowel or bladder dysfunction. Plain radiographs of the lumbar spine demonstrated no abnormality. Sagittal Gd-enhanced T1-weighted MR imaging demonstrated an intradural, extramedullary, and uniformly enhancing mass that extended from L-3 to L-4 (Fig. 1).

Operation and Adjunct Treatment. We performed an L2–4 left hemilaminectomy, and the dura mater was opened from the L-2 to L-4 level. We found an intradural circumscribed mass (4.5 × 1.5 × 1.5 cm) that involved the nerve root of the cauda equina within the intradural space. The tumor including the nerve root was completely excised. Adjuvant local radiation therapy (50 Gy) was administered. Three months after surgery, the patient’s motor weakness had resolved completely.

The patient undergoes follow-up examination at 6-month intervals. The whole spine, brain, and lungs are closely evaluated. Five and a half years after surgery, the patient is free of local recurrence and metastasis.

Abbreviations used in this paper: MPNST = malignant peripheral nerve sheath tumor; RT-PCR = reverse transcriptase polymerase chain reaction.
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Pathological Examination. This tumor was composed of a proliferation of spindle-shaped cells in a fascicular fashion with focally hyalinized stroma (Fig. 2A). Pseudo-rosette and pseudopapillary structures were focally seen (Fig. 2B and C); however, no apparent glandular epithelial differentiation with a lining of epithelial cells was observed. Also notable was the absence of tumor necrosis or heterotopic elements such as muscle, cartilage, and bone. Based on these histological findings and the site at which the tumor developed, an MPNST and monophasic fibrous synovial sarcoma were considered in the diagnosis. Immunohistochemical evaluation revealed diffuse vimentin expression and no distinct expression of other markers including S100 protein, keratin (AE1/AE3, CAM5.2), and epithelial membrane antigen (Fig. 2D).

Extraction of RNA and RT-PCR Analysis. Extraction of RNA from paraffin-embedded tissue was performed as previously described.20 We performed PCR using the following primer set—SYT-common: 5ʹ-CCA GCAGAGCCCTATGGATA-3ʹ, SSX-common: 5ʹ-TTT GTGGGCCAGATGCTTC-3ʹ (previously described to specifically amplify both of the junctional regions of the SYT-SSX1 and SYT-SSX2 fusion gene transcripts).10 After PCR, an aliquot of the PCR product was electrophoresed on a 2% agarose gel and stained with ethidium bromide. As positive controls for the integrity of mRNA in this sample, PCR for ubiquitously expressed porphobilinogen deaminase (PBGD) gene transcript was performed with the following primers—PBGD-F: 5ʹ-TGT CTGGTACGGCAATGCGGCTGCAA-3ʹ, PBGD-R: 5ʹ-TCAATGTGGCCACCACA-CCTGCCGTCT-3ʹ. These primers amplify a 98-bp fragment of SYT-SSX and a 127-bp fragment of PBGD mRNA, respectively. Reaction mixtures of reagents with SYT-SSX1 expression vector and devoid of template cDNA were included in each PCR procedure as a positive control and a negative control, respectively. A PCR product of the SYT-SSX fusion transcript was obtained from this sample (Fig. 3 upper). A subsequent direct sequencing revealed that this transcript contained base sequences identical to the SYT-SSX1 fusion transcript (Fig. 3 lower).

These findings provided a diagnosis of monophasic fibrous synovial sarcoma.

Discussion

To our knowledge, this is the first report of a primary synovial sarcoma of the cauda equina in an adult. Previously only one case involving an 11-year-old girl has been reported.5 Most synovial sarcomas arise primarily in the articular regions of the extremities, especially around the knee joint.1,14,22 It has been reported that synovial sarcoma is a malignant mesenchymal tumor of unknown histogenesis and that it develops in any soft tissue. Regarding this point, a recent study has demonstrated that a synovial sarcoma is a stem cell malignancy, and thus, this tumor can arise in any tissue including the cauda equina.11 Histologically, 2 major types of synovial sarcoma have been described.22 One is a biphasic type consisting of an apparent glandular structure composed of epithelial cells in the background of a proliferation of spindle-shaped cells. The other is a monophasic fibrous type composed totally of spindle-shaped cells. An additional minor subtype of synovial sarcoma has been described as a poorly differentiated type that typically contains primitive round tumor cells.

In our case, the tumor was composed of a proliferation of spindle-shaped cells. Pseudopapillary and pseudorosette structures were focally seen without an apparent glandular structure. Based on the histological findings and the site at which the tumor developed, an MPNST and monophasic fibrous synovial sarcoma were considered in the diagnosis. Immunohistochemical evaluation revealed diffuse vimentin expression and no distinct expression of other markers including S100 protein, keratin (AE1/AE3, CAM5.2), and epithelial membrane antigen (Fig. 2D).

Fig. 1. Preoperative sagittal Gd-enhanced T1-weighted MR image demonstrating an intradural, extramedullary, and uniformly enhancing mass extending from L-3 to L-4.

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Recently a specific chromosomal translocation between chromosome 18 and chromosome X was identified in synovial sarcoma.9 The SYT-SSX fusion gene is a chimeric gene resulting from this translocation. This fusion gene has been detected in more than 90% of synovial sarcomas. The RT-PCR detection of transcripts of the SYT-SSX fusion is the most useful and reliable tool for diagnosing a synovial sarcoma. In the present study, this fusion transcript was detected using the formalin-fixed paraffin-embedded tissue sample. This fusion gene was the decisive key in establishing our final diagnosis of synovial sarcoma.

Synovial sarcoma has been considered to have a poor prognosis and typically recurs within 2 years.22 Five-year survival rates reportedly range from 60% to 71%,4,6,15,19 and 10-year survival rates from 34% to 60%.2,6,15 The lungs, lymph nodes, and bone marrow are the most common sites of metastatic lesions. Prognostic factors associated with poor patient outcome include high-stage disease, tumor size (> 5 cm), presence of necrosis, tumor location, and extent of resection.15,16,18 For an adjuvant postoperative therapy, we chose radiotherapy because the tumor was less than 5 cm in size without necrosis and because the age of the patient was less than 23 years. Chemotherapy was not administered because its effectiveness is controversial.2,3,7 In the present case, margin resection and radiotherapy yielded more than 5 years of disease-free survival. Physicians should consider the possibility of malignancy of cauda equina tumors.

Conclusions

The RT-PCR detection of transcripts of the SYT-SSX fusion was the most useful and reliable tool for diagnosis of synovial sarcoma of the cauda equina. Complete resection of the mass and adjuvant local radiotherapy proved to be effective.

Disclosure

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![Fig. 2. Photomicrographs. A: Spindle-shaped cells proliferating in a fascicular fashion with stromal hyalinization. H & E, original magnification × 100. B: Pseudorosette formations are focally seen. H & E, original magnification × 100. C: Pseudopapillary structures are frequently seen. H & E, original magnification × 40. D: Distinct expression of S100 protein as seen in MPNSTs is not demonstrated. Original magnification × 100.](image)

![Fig. 3. Upper: A PCR product for SYT-SSX chimeric fusion transcript was detected. contl. = control. Lower: Direct sequencing confirmed that this PCR product contained a sequence identical to that of SYT-SSX1. SYT-SSX1 and SYT-SSX2 sequences are of this amplified region. Characters shown in red indicate different bases between sequences.](image)
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