Open Access Case Report

A Rare Case of Intradural Extramedullary Ewing's Sarcoma with Bone Marrow Infiltration

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Abstract

Ewing's Sarcoma commonly arises from long bones, ribs, pelvis and some extraskeletal regions. A Primary Intradural Extramedullary Ewing's Sarcoma is extremely rare and requires careful history taking, examination and imaging studies for proper diagnosis. Here we describe a case of a young female who presented with the symptoms of lower limb weakness and urinary incontinence. Following Magnetic Resonance Imaging of dorsolumbar region, third to fifth lumbar vertebral laminectomy and histopathology of excised mass, she was diagnosed as a case of Intradural Extramedullary Ewing's Sarcoma. On staging workup, bone marrow infiltration was revealed which is a very rare occurrence in these cases.

Keywords: Ewing's Sacrcoma, Intradural-Extramedullary Spinal Cord Neoplasms, Bone marrow, metastasis.

Case Report

A 9-year old female presented with backache, bilateral leg pains and urinary incontinence for the last 4 months. She also showed inability to walk for the last 3 weeks. There was no history of fits or visual problems. She was the first product of non-consanguineous marriage and there was no history of any sibling death or any other similar ailment. She was fully vaccinated and was developmentally normal. On examination, she was vitally stable and had a Glasgow Coma Scale (GCS) of 15/15. Upper limb examination was normal. On examination of lower limbs, there was reduced bulk, loss of power, reduced motor tone and reflexes, with bilaterally mute plantars. Sensory system was intact. She was referred for diagnostic radiological assessment. Magnetic Resonance Imaging (MRI) of dorsolumbar region showed dural ectasia at LV5-SV1 vertebral level and altered MR signal in spinal canal at lower lumbosacral region. MRI Brain with contrast was unremarkable. She underwent L3-L5 laminectomy with excision. Grossly, the specimen consisted of multiple tan brown fragments measuring 3x2x1 cm. Histopathological examination showed fragments of tumor exhibiting crushing and foci of necrosis. Tumor was composed of nests and sheets of cells showing high nuclear to cytoplasmic ratio and scant cytoplasm. Immunohistochemistry (IHC) showed tumor cells to be positive for CD99, NKX2.2; focal positive for CD56 and focal weak positive for Synaptophysin.

Markers CD20, Olig2 and GFAP were negative. Histopathological evaluation in conjunction with Immunohistochemistry revealed the lesion to be Intradural Extramedullary Ewing's Sarcoma. Liver Function Tests, Renal Function Tests, Serum Calcium and Blood Sugar were within normal limits. Blood Complete Picture showed TLC of 7.97 x 10^9/L (Neutrophils 59%, Lymphocytes 30%, Monocytes 6%, Eosinophils 5%), Hemoglobin of 11.6 g/dl and Platelet count of 375 x 10^9/L. Bone Marrow biopsy was done as a part of staging work-up. It was hypercellular showing active trilineage hemopoiesis along with focal areas of infiltration by round blue tumor cells showing high N/C ratio and condensed chromatin (figure 1). IHC showed them to be positive for NKX2.2 and CD99 (figure 2). Bone Scan and CT-Scan Chest were unremarkable. The patient is currently undergoing palliative therapy under care of Paediatric Oncology Department.

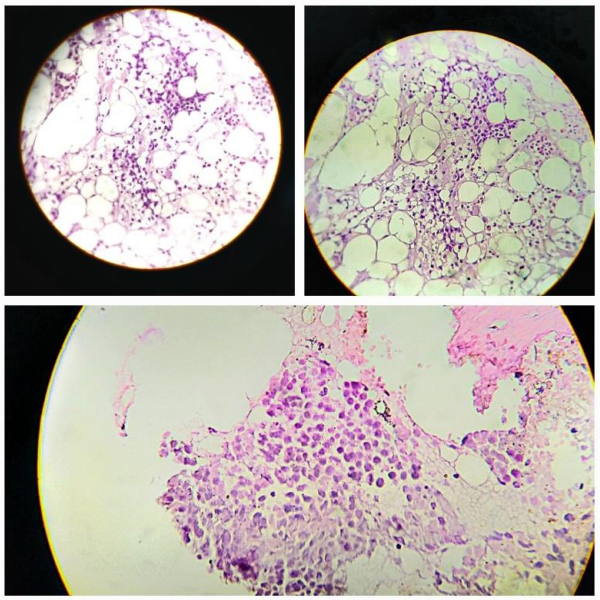

Figure 1. Bone marrow biopsy (Hematoxylin and Eosin Stain) showing focal areas of infiltration by round blue tumor cells showing high N/C ratio and condensed chromatin. (Top Half: 100X magnification; Bottom Half: 400X magnification).

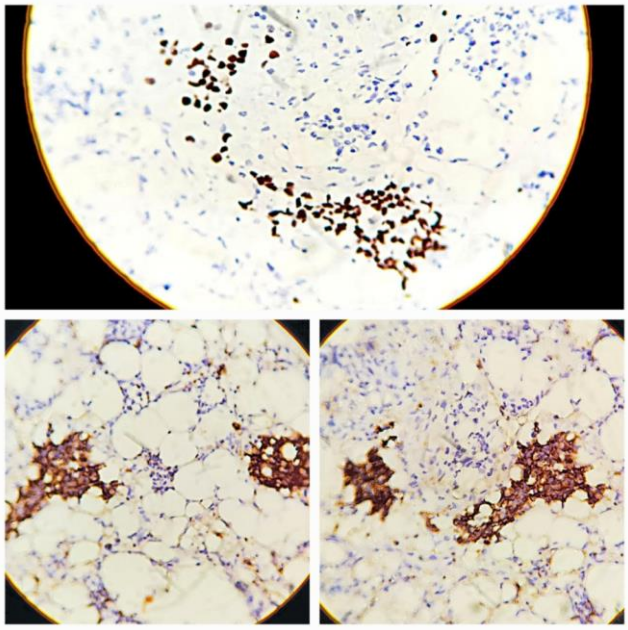

Figure 2. Top Half: Bone Marrow Biopsy NKX2.2 Immunohistochemical stain 400X; Nuclear NKX2.2 positivity seen confirming infiltration by tumor cells.(Bottom Half: Bone Marrow Biopsy CD99 Immunohistochemical stain 400X. Diffuse, strong membrane positivity for CD 99 can be seen in the tumor cells.)

Discussion

Ewing's sarcoma (ES) is an aggressive tumor, primarily involving bone, often with soft tissue extension. It differs from other sarcomas due to its aggressiveness and distinct molecular pattern.[1] Extraosseous ES is uncommon accounting for 16% of all the cases. It is most likely to originate from soft tissue of thorax and abdomen (43%), extremities (26%) and pelvis (14%). The paravertebral space constitutes about 7% of all extraosseous ES tumors, which almost always arise from extradural paraspinal soft tissue. The primary intradural extramedullary Ewing's Sarcoma is extremely rare. Only a limited number of cases have been reported in the literature.[2]

Here we have presented a rare case of primary intradural extramedullary ES with secondary metastasis in the bone marrow. From the previous knowledge, around quarter of patients with ES have metastasis at the time of diagnosis with 50% metastasizing in the lungs and remaining in the bone and bone marrow[3]

In our case there was a primary intradural extramedullary Ewing's Sarcoma with metastasis in the bone marrow. Although in the previous case reports, the metastasis have been reported in the brain [2,4] but here the MRI brain showed no abnormality. In another report metastasis in lungs has also been reported. [5] Similarly, skip metastasis is also common in these cases.[5] Therefore MRI brain and spine with whole body PET scan is mandatory.[6] The presence or absence of identifiable metastasis is the principal prognostic factor, with five year survival rate being doubled if no metastasis is present at the time of diagnosis.[7] In our case, bone marrow was infiltrated and according to current literature as of date, no case report has been presented of this rare occurrence.

The diagnostic immunohistochemical marker for Ewing's sarcoma is CD 99.[6] The results of our histochemical markers are comparable to other studies. However, due to limitation of resources, cytogenetics was not performed at the time of diagnosis. The specific translocation occurring in Ewing's sarcoma is t(11;22)(q24;q12), resulting into production of EWSR1-FLI1 fusion transcript and protein. This chimeric protein has been reported to act as an oncogenic transcription factor.[6]

The symptoms of intradural extramedullary ES are almost similar in all cases; of spinal cord compression.[8,9] The most common presenting complaint is pain accompanied with motor disturbance of upper and lower limbs. Bladder and rectal disturbance is also present in one quarter of patients.[4] Our patient also presented with similar findings that is backache, bilateral leg pains and urinary incontinence with preservation of sensory system.

The treatment of these cases lies in the complete resection of tumor with excellent micro surgical techniques followed by chemotherapy and radiotherapy. The choice of radiotherapy varies from case to case. Due to scarcity of the cases, proper treatment guidelines have not been established.[6] Furthermore, it is a highly aggressive tumor with very poor prognosis and very high mortality.[10]

Conclusion

In summary, we report a rare case of primary intradural extramedullary Ewing's Sarcoma with secondary bone marrow metastasis, an unusual presentation in the existing literature. Despite the absence of brain metastasis on MRI, bone marrow infiltration was observed, underscoring the importance of comprehensive staging.

References

1. Cuello CM, De Jesus O, de Jesús Espinosa A, Fernández-de Thomas RJ, Murray G, Pastrana EA, et al. Prognosis and outcome of cervical primary extraosseous intradural extramedullary Ewing sarcoma: a systematic review. *Cureus*. 2022;14(7):1-8.
2. Müngen E, Kurucu N, Kutluk T, Oğuz KK, Söylemezoğlu F, Yalçın B. Primary spinal multifocal intradural-extramedullary Ewing sarcoma in children: presentation of a case and review of the literature. *Turk J Pediatr*. 2021;63(6):1084-90. doi:10.24953/turkjped.2021.06.018
3. Bazzocchi A, Bacci A, Serchi E, Salerno A, Salizzoni E, Leonardi M. Intradural extramedullary Ewing's sarcoma recurrence with acute clinical presentation and literature review. *Neuroradiol J*. 2013;26(4):476-81. doi:10.1177/197140091302600413
4. Izubuchi Y, Nakajima H, Honjoh K, Imamura Y, Nojima T, Matsumine A. Primary intradural extramedullary Ewing sarcoma: a case report and literature review. *Oncol Lett*. 2020;20(3):2347-55. doi:10.3892/ol.2020.11786
5. Haresh KP, Chinikkatti SK, Prabhakar R, Rishi A, Rath GK, Sharma DN, et al. A rare case of intradural extramedullary Ewing's sarcoma with skip metastasis in the spine. *Spinal Cord*. 2008;46(8):582-4. doi:10.1038/sc.2008.8
6. Takami H, Kumar R, Brown DA, Krauss WE. Histologic features and prognosis of spinal intradural extramedullary Ewing sarcoma: case report, literature review, and analysis of prognosis. *World Neurosurg*. 2018;115:448-52. doi:10.1016/j.wneu.2018.04.015
7. Arena JD, Sinha S, Wathen C, Ghenbot Y, Zhang PJ, Welch WC. Primary intradural extraosseous Ewing's sarcoma of the L3 nerve root: illustrative case. *J Neurosurg Case Lessons*. 2024;7(3). doi:10.3171/CASE23459
8. Scantland JT, Gondim MJ, Koivuniemi AS, Fulkerson DH, Shih CS. Primary spinal intradural extraosseous Ewing sarcoma in a pediatric patient: case report and review of the literature. *Pediatr Neurosurg*. 2018;53(4):222-8. doi:10.1159/000488767
9. Kartal A, Akatlı A. Primary intradural extraosseous Ewing's sarcoma in a young child. *Childs Nerv Syst*. 2016;32:409-10. doi:10.1007/s00381-016-3020-7
10. Pu F, Liu J, Zhang Z, Guo T, Shao Z. Primary intradural extramedullary extraosseous Ewing's sarcoma/peripheral primitive neuroectodermal tumor (PIEES/PNET) of the thoracolumbar spine: a case report and literature review. *Open Med (Wars)*. 2021;16(1):1591-6. doi:10.1515/med-2021-0377