Case Report

# Cauda equina syndrome due to primary extra-skeletal Ewing's sarcoma: a diagnostic challenge

**Habib R[a], Alam D[b], Munshi MA[c]**


## ABSTRACT

*Ewing's sarcoma causing cauda equina syndrome is a rare entity. We report case history of a 20-year-old man who presented with features of cauda equine syndrome preceded by 4-month history of pain in the buttock and tip of the coccyx. Magnetic resonance imaging revealed a lesion within the spinal canal at S1–5 level and a large lobulated heterogeneously enhancing mass lesion in pelvic cavity in sacral region. Excisional biopsy revealed Ewing's sarcoma by characteristic features on histologic analysis and immunohistochemistry. Patient was referred to oncologist for combined chemotherapy with radiotherapy.*


***Key words:*** *Ewing's sarcoma, cauda equina syndrome.*

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## INTRODUCTION

Ewing's sarcoma (ES) is a poorly differentiated, highly malignant, round cell tumour without cellular or structural differentiation.[1] First described by James Ewing in 1921[2], ES of bone is the most common malignant tumor in the first decade of life and the second after osteosarcoma in the second decade.[3] It can affect any part of the skeleton but the most affected parts are the ilium and the diaphyses of the femur and tibia.[4] Extra-skeletal ES was first described by Tefft et al. in 1969.[5] It can arise in various locations like the chest wall, paravertebral muscles, extremities, pelvis and retroperitoneal space.[6] Patients may have a neurological deficit when the tumor extends into the spinal canal, causing compression of the spinal cord or primarily arises from epidural space. We report a case of 20-year-old patient presenting with cauda equina syndrome who was finally diagnosed with Extra- skeletal Ewings Sarcoma.

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## CASE REPORT

A 20-year-old previously healthy young man presented with the complaints of bowel and bladder incontinence with perianal anesthesia for one month. This was preceded by history of progressively increasing pain in the buttock and tip of the coccyx with associated paresthesia in back of thigh and leg muscles for four months. He denied any history of back pain, injury, fever, weight loss, trauma or previous history of tuberculosis. He was treated as a case of coccydynia for initial three months without any significant improvement. His symptoms evolved with marked accentuation of the pain which made it difficult for him to maintain sitting posture and hampered his activity of daily living. Clinical examination revealed tenderness at the sacral spine with mild weakness of ankle planter flexion in right side. Deep tendon reflexes were normal with flexor planter responses. Sensory system examination also revealed no abnormality except diminished sensation in peri-anal area with reduced rectal tone.

His initial laboratory workup showed normal findings. Radiographs of the spine revealed no evidence of bony destruction. But abdominal ultrasonogram showed features of chronic cystitis with post voidal residual volume of 284 cc. MRI of lumbosacral spine revealed large lobulated mass lesion in pelvic cavity in sacral region heterogeneously enhanced after injection of gadolinium, infiltration of sacral vertebral bodies and spinal epidural component causing significant canal compromise (Figure 1).

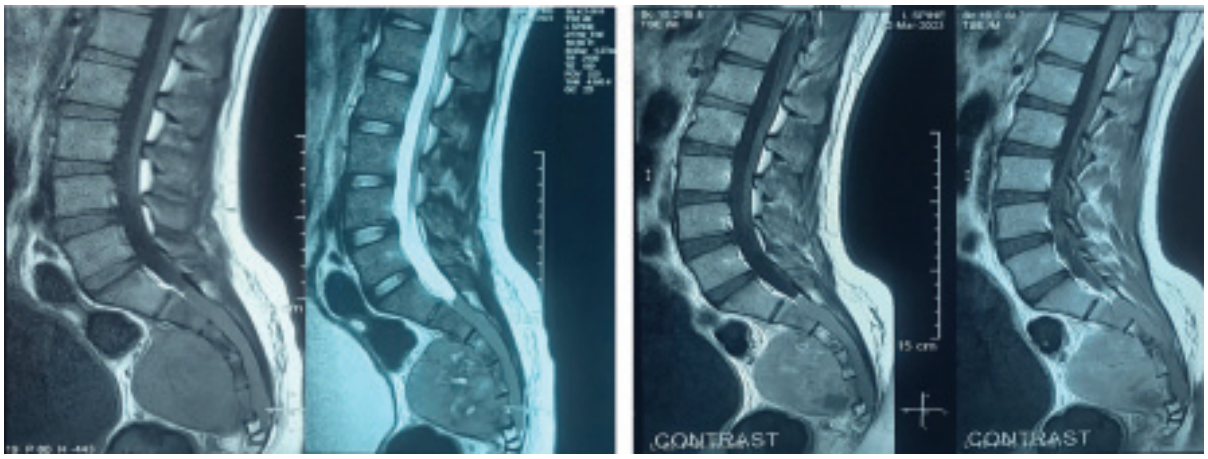


**Figure 1.** Gadolinium enhanced MRI of lumbosacral spine in sagittal view showed large lobulated mass lesion in pelvic cavity,infiltration of sacral vertebral bodies and spinal epidural component

The patient was catheterized and inj. dexamethasone 10 mg I/V 8 hourly was started. An exploratory laparotomy was done. The growth measuring about (5 cm X 6 cm) over L5-S1 region deeply adherent with sacral promontory, posterior wall of rectum and between two layers of Waldeyer's fasia, was removed as much as possible and the specimen was sent for histopathology and immunohistochemistry. Histopathological examination of the specimen showed malignant neoplasm composed of nests, clusters and occasional rosette like aggregates of small round cells with round to oval hyper chromatic nuclei surrounded by prominent desmoplastic stroma, giving the impression of malignant small round cell tumor suggestive of desmoplastic small round cell tumor (Figure 2). Immunohistochemistry was positive for Vimentin, CD99, FL1 and moderately positive for Synaptophysin but negative for S-100, Desmin, CD45, WT1. Based on these immunohistochemistry reports, a diagnosis of Ewing's Sarcoma was confirmed. The neurological outcome of surgery was good with mild residual deficits. He was referred for adjuvant systemic chemotherapy and local radiotherapy.

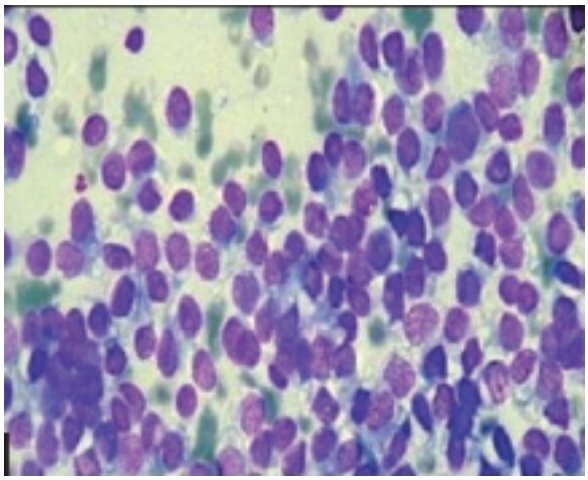

**Figure 2**. Histopathology of the specimen showed malignant small round cell tumor

## DISCUSSION

Cauda equina syndrome is a surgical emergency that requires early diagnosis and prompt intervention. It is a challenging diagnosis especially in young and a delay in the diagnosis can result in complications that severely affect the patient's quality of life. The cause of cauda equina syndrome with low back pain in children and young include tuberculosis, pyogenic osteomyelitis, lymphoma, chordoma, osteogenic sarcoma, and ES and other intradural extramedullary tumors of the spinal cord and cauda equina.[7]

Extra skeletal ES (EES) has no specific clinical manifestation. The most frequently presenting symptom is a rapidly growing mass with local pain.[8] However, the type of accompanying symptoms depends largely on the sarcoma's site of origin. EES primarily arising in the epidural space is a rare entity and very few cases of cauda equina syndrome due to primary epidural ES mostly involving the lumbosacral region have been reported.[9]

To accurately diagnose extra-skeletal epidural ES, it is important to rule out several other potential etiologies

of an epidural mass. MRI represents the gold standard for early diagnosis, assessment of the extent of the tumor in the soft tissues and determination of the therapeutic strategy.[10]

Although clinical and radiological findings are essential in the early diagnosis of primary ES definitive diagnosis is based on histology, immunophenotyping and the demonstration of one of the specific translocations. The classical histopathologic findings are sheets, lobules and occasional rosettes of round cells with irregular nuclei and sparse cytoplasm.[11] The small, blue, round cell tumors are the histological features akin to ES are hard to differentiate from neuroblastoma, malignant lymphoma and rhabdomyosarcoma.[12] Thus, ES is able to be distinguished by using the cell surface marker, CD99 (encoded by MIC2 gene) or Friend leukemia integration 1 transcription factor (FLI1) with immunohistochemistry. However, the sensitive marker also shows a positive result in other types of malignancies, such as neuroectodermal tumors.[13] These diagnoses were ruled out by the negativity of the respective markers (CD45; demsin and EMA), as well as neuroblastoma (negative synaptophysin). Cytogenetic analysis, by fluorescence in situ hybridization (FISH) technique, allows to confirm the diagnosis. This technique allows to find the reciprocal translocation t(11;22) (q24;q12) involving chromosome 22 located on EWS-FLI 1 in more or less 90% of cases.[4] This technique is not available in our institute.

The National Comprehensive Cancer Network (NCCN) has generated guidelines for the treatment of bone cancers including ES and the authors suggest that any member of the Ewing tumor family can be treated according to the same algorithm.[14] The treatment recommended is local treatment (surgery and/or radiotherapy) plus chemotherapy. However, many authors have suggested that surgery may have a more important role in EES than in skeletal ES and that complete resection predicts a favourable survival.[15]EES is quite radiosensitive, but improvements in surgical technique and the risks associated with radiation (secondary malignancies) have reduced the reliance upon radiation.[16]. Some researchers have underlined the important role of preoperative radiotherapy (RT) for successful local treatment in spinal Ewing tumours especially when an intralesional resection is possible.[17]

Chemotherapy plays a pivotal role in the treatment of ESs. Recent studies have shown that neoadjuvant and adjuvant chemotherapies produce comparable results in patients with localized disease.[18] Chemotherapy can be provided after surgery to improve overall survival rates and reduce the likelihood of tumour recurrence. [19]

The prognosis for extraosseous ES appears more favourable than that for ES in bone.[20] Patients with localized disease have estimated 5-year overall survival rates of about 70 % due to considerable progress in both local and systematic therapy during the past four decades. [21] Patients with metastatic or recurrent disease have a worse outcome; 5-year overall survival remains about 25 %.[22]

Because of the distressing nature of the cauda equina syndrome, a rare case of extra-skeletal ES came to be diagnosed. A multi-specialty approach is paramount in such a diagnostically challenging case, including radiological expertise in the identification of lesion and urgent surgical management in the form of decompression and tumor excision, followed by immune-histopathological correlation. Co-management with an oncological team in form of chemo- and radiotherapy is essential for adequate treatment.

**Authors' contribution:** RH was involved in the diagnosis, patient management, manuscript writing and literature review. All authors were involved in evaluation and management of the case.

**Consent:** Informed consent was taken from the patient and his attendant for publication of this case report and accompanying images.

**Conflicts of interest:** Nothing to declare.

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