

Rare Adult Presentation of Embryonal Rhabdomyosarcoma: A Case Report

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ABSTRACT

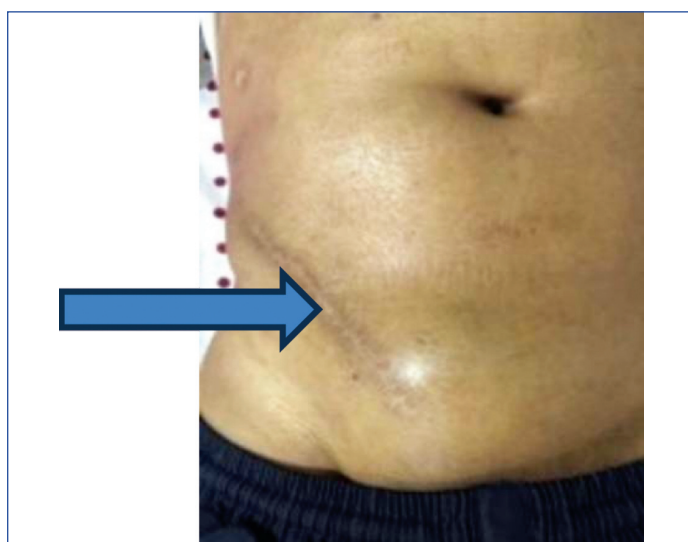
Embryonal Rhabdomyosarcoma (ERMS) is a malignant mesenchymal tumour showing skeletal muscle differentiation and represents the most common histological subtype of Rhabdomyosarcoma (RMS). It predominantly affects the paediatric population, typically arising in the head and neck, genitourinary tract, and retroperitoneum. Occurrence in adults is exceedingly rare and is associated with aggressive biological behaviour, advanced stage at presentation, limited therapeutic options, and poor prognosis. Hereby, the authors report the case of a 56-year-old male who presented with a rapidly enlarging, painful right abdominal mass of two months duration. Imaging revealed a large heterogeneous soft tissue mass with evidence of distant metastases. Histopathological examination showed pleomorphic round-to-ovoid tumour cells with rhabdomyoblastic differentiation and prominent anaplastic features. Immunohistochemistry demonstrated positivity for Myogenic Differentiation 1 (MyoD1), cytokeratin, Cytokeratin 8/18 (CK8/18) and Desmin, confirming the diagnosis of ERMS. Genomic profiling done using foundation one heme revealed amplification Mouse Double Minute 2 homolog (MDM2), Cyclin-Dependent Kinase 4 (CDK4) and Fibroblast Growth Factor Receptor Substrate 2 (FRS2). Suggesting aggressive tumour biology and potential therapeutic targets. The patient underwent surgical excision followed by multiple lines of chemotherapy and molecularly guided targeted therapy. Despite transient clinical responses, the disease showed repeated relapses with progressive metastatic spread. The patient ultimately succumbed to the illness. The present case highlights the extreme rarity of adult-onset ERMS, its aggressive clinical course, and dismal outcome despite multimodal treatment. Accurate histopathological and molecular diagnosis, along with reporting of such cases, is essential to improve understanding of this uncommon and highly aggressive malignancy.

Keywords: Adult, Mesenchymal tumour, Soft-tissue sarcoma

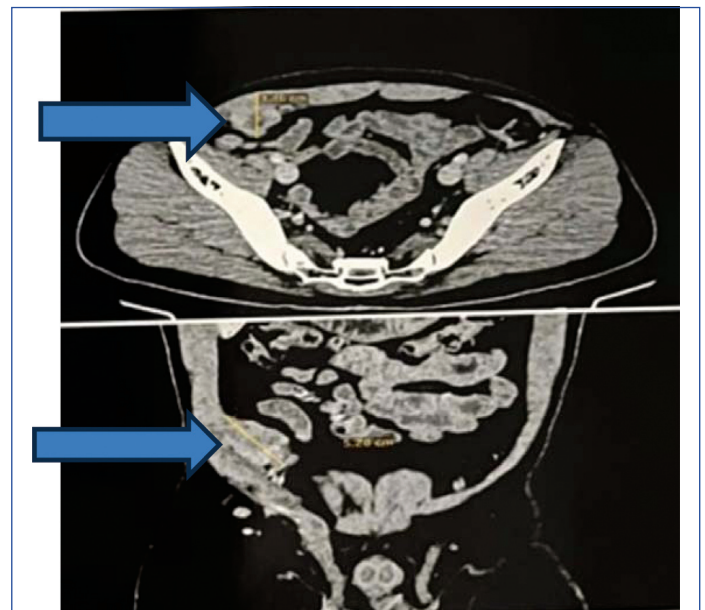
CASE REPORT

A 56-year-old male reportedly asymptomatic until one month ago when he first noticed a 1 cm lump in the right inguinal area. A history of gym-related trauma was reported 15 days prior to the appearance of the lump, following which gradual pain started for 10 days. For which no analgesics were taken for the pain as it was mild in nature. A plain Computed Tomography (CT) abdomen identified a haematoma measuring about 1x1 cm.

One month later, lump exhibited gradual progression and increase in pain over time for which CT abdomen revealed a mass measuring 35 × 34 × 52 mm in the right iliac fossa, originating from the anterior abdominal muscles [Table/Fig-1,2]. Additionally, a CT of the thorax indicated a subpleural nodule in the posterior segment of the right middle lobe. No pulmonary complaints were present at that time.



[Table/Fig-1]: Clinical image of the patient showing a large, visible swelling/mass is seen in the lower abdomen, causing fullness and outward bulging.



[Table/Fig-2]: Contrast-enhanced CT abdomen.

Oval shaped soft-tissue lesion is noted involving anterior abdominal wall muscles in right lower quadrant. The lesion measures 35 × 34 × 52 mm.

Subsequently, Fine Needle Aspiration Cytology (FNAC) was performed from the lateral aspect of the right iliac fossa. The haemorrhagic aspirate revealed cells with hyperchromatic vesicular nuclei, prominent nucleoli, a high nuclear-to-cytoplasmic (N:C) ratio, and abnormal mitotic figures, suggestive of malignancy.

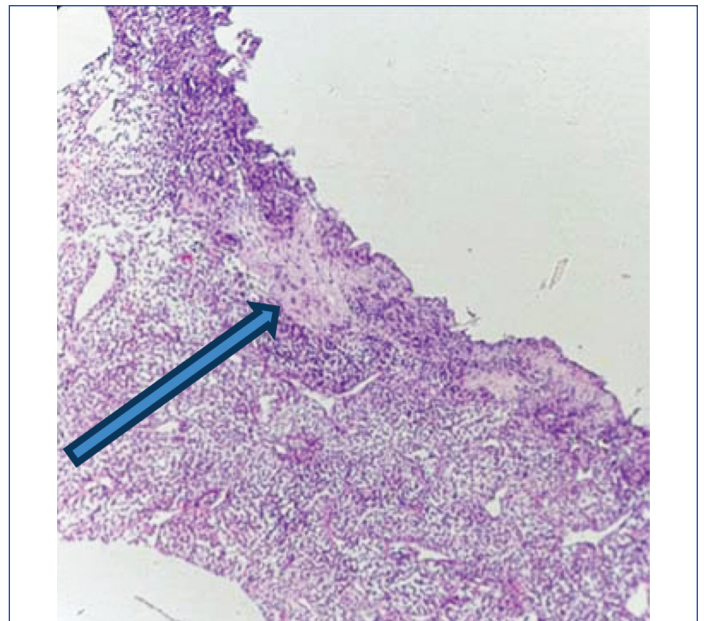
The patient then underwent a wide excision of the anterior abdominal wall (right iliac fossa), along with resection of the omentum, terminal ileum loop, parietal peritoneum, spermatic cord, and testicular vessels. The procedure was followed by mesh placement.

The histopathology report indicated a high-grade sarcoma with tumour infiltration into the adjacent soft tissue, skeletal muscle,

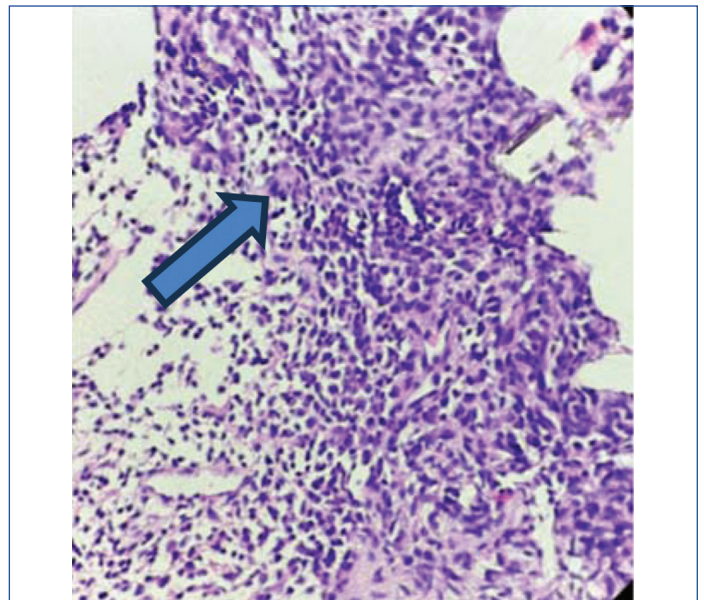
abdominal aponeurosis, outer surface of the ileum, omentum, and spermatic cord. The right iliac lymph nodes and soft tissue surgical margins were free of tumour involvement ([Table/Fig-3]: 4x magnification, [Table/Fig-4]: 20x magnification, [Table/Fig-5]: 40x magnification and [Table/Fig-6]: 100x magnification).

Immunohistochemistry of the tumour cells showed patchy expression of Desmin, MyoD1 and are focally positive for cytokeratin & CK8/18. These cells were immunonegative for Myogenin, Epithelial Membrane Antigen (EMA), CD45, CD117, Smooth Muscle Actin (SMA), CD34, S-100 protein, SOX10, H-caldesmon, CDX2 & CD38. The Integrase Interactor 1 (INI-1) & Histone H3 Lysine 27 trimethylation (H3K27me3) expression is retained.

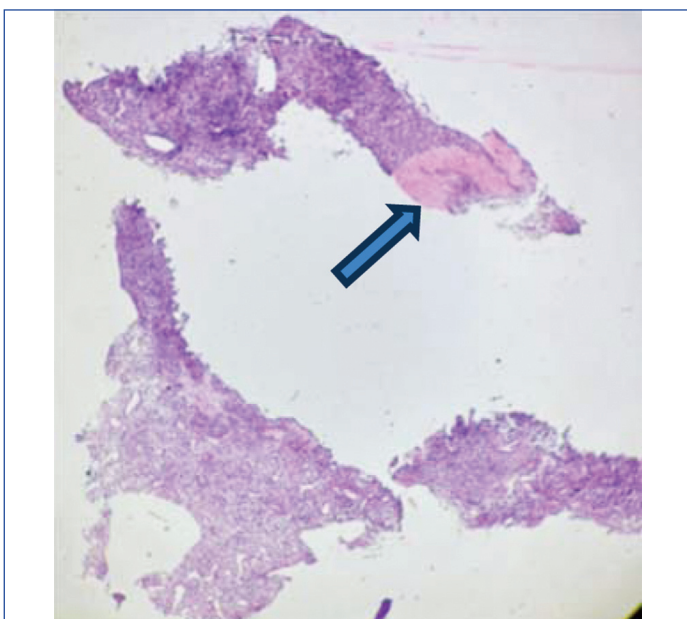
A whole-body Positron Emission Tomography (PET) scan performed 20 days postoperatively revealed scattered pulmonary nodules on both sides, with the largest measuring approximately 10.8x7.8 mm. Additionally, ill-defined and nodular areas were observed in the anterior lower abdominal wall and the right inguinal region [Table/Fig-7]. The patient was initiated on chemotherapy with Vincristine, Adriamycin, and Cyclophosphamide for two cycles, with a 20-day interval between each cycle. A follow-up CT scan of the abdomen and thorax after 1.5 months was performed to assess the response, which showed no improvement. Instead, there was an increase in the number and size of masses in both the lungs and the anterior abdominal wall [Table/Fig-8]. While the masses did not infiltrate any organs, they caused space-occupying lesions. Following this, the patient underwent two cycles of Ifosfamide and Etoposide chemotherapy, with a 20-day interval between each cycle. A subsequent PET-CT scan after two months showed regression of the masses, indicating a partial response to the chemotherapy [Table/Fig-9,10]. One cycle of Ifosfamide and Etoposide chemotherapy was administered with a 20-day gap, followed by a CT scan for evaluation. The results indicated no response to the chemotherapy, as both the size and number of masses had increased. Following this, a chemotherapy drug sensitivity test was conducted, indicating the best response to Paclitaxel. Genomic profiling done using foundation one heme revealed amplification of CDK4, MDM2, and Fibroblast Growth Factor Receptor Substrate 2 (FRS2). The patient was initiated on Lenvatinib, a multi-kinase inhibitor, for 15 days, along with weekly Paclitaxel chemotherapy for four weeks. After the 4th dose of Paclitaxel, Tablet Abemaciclib, a CDK4 and CDK6 inhibitor, was introduced. Whole body PET-CT was done for observation but no regression in mass seen. Due to the presence of large multiple masses in the abdomen, the patient developed an



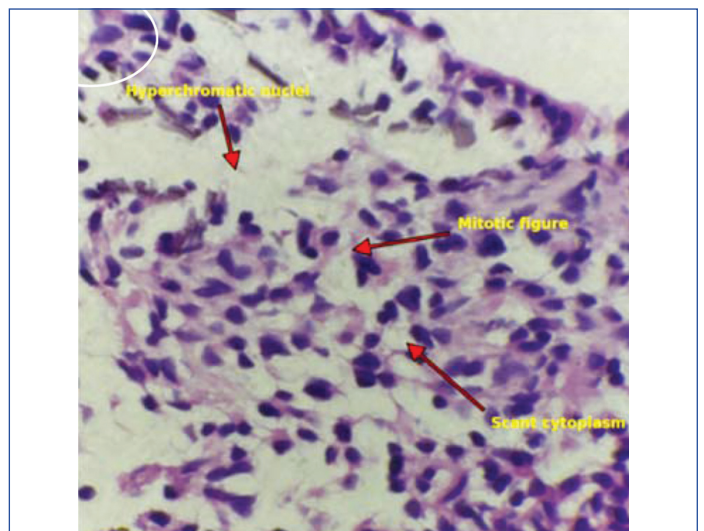
[Table/Fig-4]: This is a haematoxylin and eosin stained slide. (20x magnification) showing a densely cellular area is seen just beneath the epithelial lining, composed of poorly differentiated round to spindle-shaped cells with scant cytoplasm, along with numerous mitotic figures.



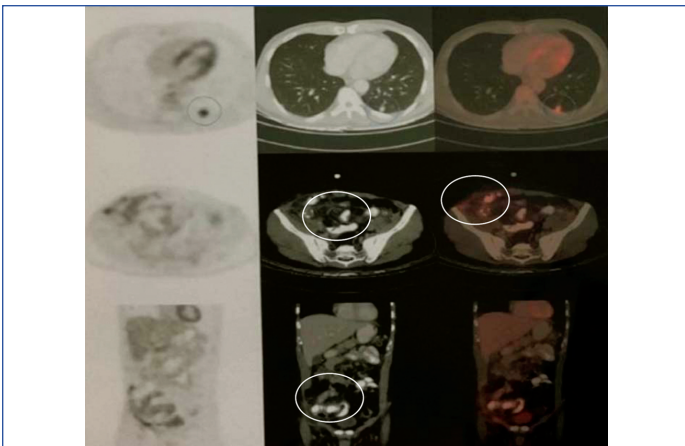
[Table/Fig-5]: This is a haematoxylin and eosin-stained slide. (40X magnification) showing the cells are round to spindle-shaped, poorly differentiated, with high nuclear-to-cytoplasmic ratio and cells with elongated tails of cytoplasm (tadpole cells).



[Table/Fig-3]: This is a haematoxylin and eosin stained slide scanner view (4x magnification) showing occasional tumour cells exhibit abundant eosinophilic cytoplasm



[Table/Fig-6]: This is a haematoxylin and eosin-stained slide (100X magnification) showing hyperchromatic nuclei, mitotic bodies and Scanty cytoplasm.



[Table/Fig-7]: This image shows multiple PET-CT fusion slices (thorax and abdomen).

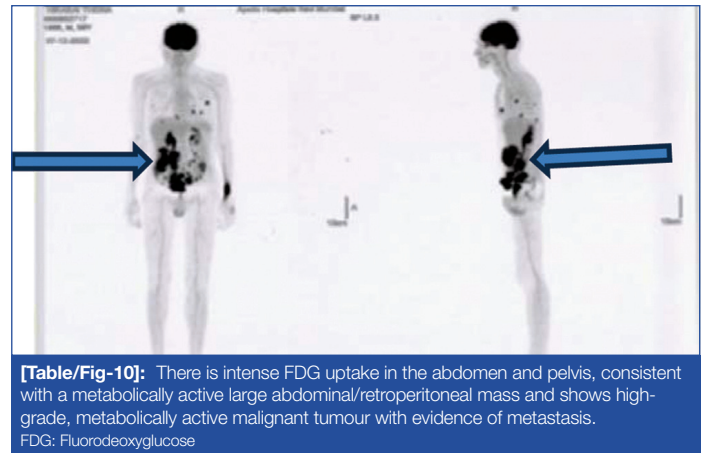
Thoracic section (top row):

- A hypermetabolic focus is seen in the right lung base, suggesting pulmonary metastasis.
- The fusion image confirms FDG uptake in this lesion.

Abdominal/pelvic section (middle row):

- There is a large FDG-avid soft tissue mass in the pelvis/retroperitoneum, consistent with the primary lesion.
- Adjacent structures appear displaced, and uptake is heterogeneous (possible necrotic areas within).

FDG: Fluorodeoxyglucose



[Table/Fig-10]: There is intense FDG uptake in the abdomen and pelvis, consistent with a metabolically active large abdominal/retroperitoneal mass and shows high-grade, metabolically active malignant tumour with evidence of metastasis.
FDG: Fluorodeoxyglucose

intestinal obstruction and underwent surgery for a jejunotransverse anastomosis. The patient experienced symptom relief following the procedure. However, one-month post-surgery, the patient went into septic shock and was unable to recover.

DISCUSSION

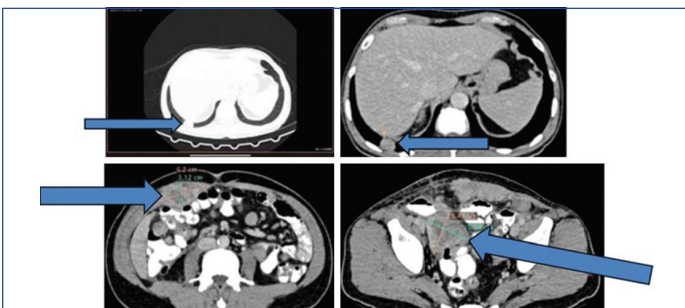
RMS is predominantly a paediatric malignancy, accounting for approximately 3-4% of childhood cancers, whereas its occurrence in adults is rare and biologically distinct [1]. Adult RMS constitutes less than 4% of all soft-tissue sarcomas and is frequently associated with advanced-stage disease, larger tumour size, unfavourable anatomical locations, and early metastatic spread, resulting in inferior outcomes compared to paediatric populations despite similar treatment approaches [2]. These features were evident in the present case, which involved an adult patient with abdominal wall primary and pulmonary metastases at diagnosis.

The diagnosis of ERMS in adults is particularly challenging because of its rarity and significant histomorphological overlap with other high-grade sarcomas. Differential diagnoses include pleomorphic RMS, alveolar RMS, undifferentiated pleomorphic sarcoma, leiomyosarcoma, synovial sarcoma, malignant peripheral nerve sheath tumour, and poorly differentiated carcinoma, especially in cases showing cytokeratin positivity [3]. In tumours arising from the abdominal wall, desmoid-type fibromatosis and gastrointestinal stromal tumours must also be considered [4]. Therefore, accurate diagnosis requires a multimodal approach that integrates imaging, histopathology, immunohistochemistry, and molecular studies.

Cross-sectional imaging with CT and PET-CT plays a crucial role in defining tumour extent, identifying metastatic disease, and monitoring therapeutic response [5].

Histopathologically, ERMS is characterised by primitive round-to-ovoid cells with rhabdomyoblastic differentiation; the presence of marked nuclear pleomorphism and anaplastic features, as seen in this case, has been reported more frequently in adult patients and may indicate aggressive tumour biology [6]. Immunohistochemistry is indispensable for confirmation, with desmin indicating myogenic differentiation and nuclear expression of MyoD1 and myogenin being highly specific for RMS [7]. Molecular testing is essential to exclude alveolar RMS by assessing PAX-FOXO1 fusion status, as this has prognostic implications. In the present case, comprehensive genomic profiling revealed CDK4 and MDM2 amplification, supporting aggressive behaviour and offering potential therapeutic targets [8].

Comparative analyses from published series demonstrate that adult ERMS differs significantly from paediatric disease. Studies by Little DJ et al., and Sultan I et al., have shown that adults present more frequently with advanced and metastatic disease and experience significantly worse survival outcomes [2,9]. Similar to previously reported adult ERMS cases involving deep abdominal or retroperitoneal sites, this patient presented with an unfavourable primary location and early lung metastases, both established adverse prognostic factors. While some adult ERMS cases have

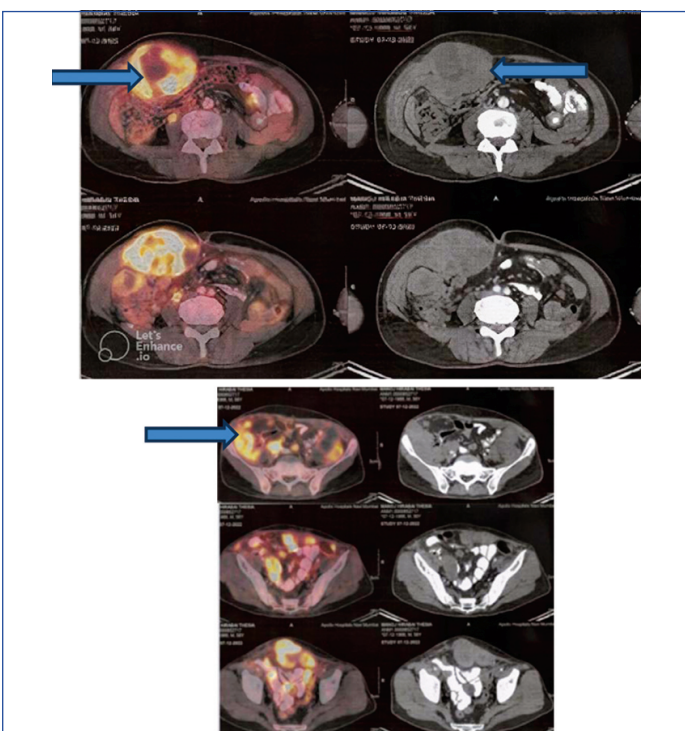


[Table/Fig-8]: Contrast-Enhanced CT Abdomen. Thoracic section (top row):

- Largest nodule is about 22x14mm at posterior basal segment of right lower lobe.

Abdominal/pelvic section (middle row):

- Multiple enhancing soft tissue lesion within peritoneal cavity predominantly in right lower quadrant and paracolic gutter largest of size measures 84x74x40mm.
- Enhancing nodule encasing the bowel loops RIF region without causing dilatation of proximal bowel loops. Multiple enhancing peritoneal nodules are seen along the serosal surface of small and large bowel loops



[Table/Fig-9]: PET images (left-side), the lesion demonstrates intense FDG uptake, indicating high metabolic activity, consistent with a malignant tumour and The CT images (right-side) show the same lesion with areas of soft-tissue density and possibly necrosis/heterogeneous enhancement.

FDG: Fluorodeoxyglucose

shown partial or durable responses to multimodal therapy, the present case exhibited only transient responses to successive chemotherapy regimens and targeted therapy, followed by rapid disease progression [10]. A contrasting feature of this case is the use of contemporary genomic profiling, which is infrequently described in earlier reports; however, despite molecularly guided therapy, clinical benefit remained limited.

Overall, the current case closely reflects patterns reported in the literature-adult age, abdominal primary site, metastatic presentation, aggressive clinical course, and poor outcome-while also highlighting the diagnostic complexity and evolving role of molecular characterisation. It underscores the persistent therapeutic challenges in adult ERMS and the need for continued reporting and collaborative research to improve understanding and outcomes in this rare and highly aggressive malignancy [11].

CONCLUSION(S)

The present case highlights the rare occurrence of ERMS in adults and reinforces its aggressive clinical behaviour and poor prognosis, particularly when presenting with metastatic disease. Adult-onset ERMS often manifests at advanced stages and demonstrates limited and short-lived responses to conventional chemotherapy, despite aggressive multimodal treatment. Early and accurate diagnosis using detailed histopathological, immunohistochemical, and molecular evaluation is essential to establish the diagnosis and guide therapy. Comprehensive genomic profiling may help identify actionable targets and should be considered early in the disease course. Given the lack of standardised treatment protocols and inferior outcomes in adults, reporting such rare cases is crucial to improve understanding of disease biology and to inform future therapeutic strategies.

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