ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/25mar1530

Metastatic Alveolar Rhabdomyosarcoma with Occult Primary Tumor in an Adult: A Case Revealed by Trephine Biopsy

Dr. Thounaojam Joytamala Devi[1]; Dr. Rajeshwari.S. Handigund[2*]; Dr. Rohan Bhise[3]

[1]Junior Resident in Pathology; [2]Consultant Pathologist; [3]Professor and HOD Medical Oncology
[1,2,3]KLE'S Dr Prabhakar Kore Hospital and Medical Research and Jawaharlal Nehru Medical College, Belagavi, Karnataka

Corresponding Author: Dr. Rajeshwari. S. Handigund[2*]

Publication Date: 2025/04/03

Abstract:

- ***Introduction:***
Rhabdomyosarcoma (RMS) is a malignant tumor, commonly seen in children and adolescents but rare in adults. In children, RMS present in the head and neck, genitourinary tract, or extremities, while in adults, it mostly occurs in the extremities, rarely in head and neck region.

- ***Case Details:***
A 60-year-old man, with Type2 diabetes mellitus and Alveolar rhabdomyosarcoma (ARMS) with unknown primary under chemotherapy since1 year, came with the complaints of fever, generalised weakness since 1 month. Complete blood count showed Pancytopenia with Haemoglobin - 6.9 g/dl, Total leucocyte count - 2.4x10^6/µL, Platelet count - 53x10^3/µL. PET-SCAN revealed increase FDG uptake in axial and appendicular skeletal marrow. Bone marrow study showed suppression of trilineage haematopoiesis with 80% atypical elongated cells with hyperchromatic nuclei and abundant eosinophilic cytoplasm, favouring rhabdomyosarcoma cells with focal areas of necrosis. Patient continued chemotherapy with blood transfusions and on regular follow-up.

- ***Conclusion:***
ARMS is characterized by aggressiveness and metastatic tendencies. It spreads to lungs, bone marrow, liver and brain. This case highlights a rare case of ARMS with occult primary metastasis in the lymph node and bone marrow, making hematopathological analysis essential for diagnosis and understanding disease progression.

Keywords: *Bone Marrow Metastasis, Rhabdomyosarcoma, Alveolar Rhabdomyosarcoma, Haematopathology.*

How to Cite: Dr. Thounaojam Joytamala Devi; Dr. Rajeshwari.S. Handigund; Dr. Rohan Bhise; Dr. Rajeshwari. S. Handigund. (2025). Metastatic Alveolar Rhabdomyosarcoma with Occult Primary Tumor in an Adult: A Case Revealed by Trephine Biopsy. *International Journal of Innovative Science and Research Technology*, 10(3), 1979-1982. https://doi.org/10.38124/ijisrt/25mar1530.

I. INTRODUCTION

In children, RMS typically presents in the head and neck, genitourinary tract, or extremities, while in adults, it most commonly occurs in the extremities and rarely in the head and neck region. It is commonly seen in children and adolescents but is rare in adults, comprising less than 1% of solid tumors. RMS is a primitive mesenchymal malignant tumor. It is commonly seen in children and adolescents but rare in adults, comprising less than 1% of solid tumors. In children, RMS typically presents in the head and neck, genitourinary tract, or extremities, while in adults, it most commonly occurs in the extremities, rarely in head and neck region. (2,3,4) RMS are of four types: embryonal (most common), alveolar, pleomorphic, and spindle cell/sclerosing. The Alveolar rhabdomyosarcoma (ARMS) being more common in the adult population, are characterized by a specific reciprocal chromosomal translocation involving

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/25mar1530

PAX3-FKHR or PAX7- FKHR fusion proteins(5). RMS with bone marrow metastasis accounts for approximately 6%-16% of all RMS cases and very rare in lymph nodes. (6)

II. CASE DETAILS

A 60-year-old man, with Type2 diabetes mellitus presented with left cervical lymph nodes swelling for 4/5months. Histopathological examination of the biopsy specimen from the left level II cervical lymph node showed metastatic malignant round tumor cells having high Nuclear : Cytoplasmic ratio, hyperchromasia and marked pleomorphism that are arranged in nests separated by thin fibrous septate. Brisk mitosis and prominent apoptosis are also seen. An Immunohistochemistry (IHC) panel showed diffusely and strongly positive for Desmin and MyoD1 with focal positivity for Synaptophysin. This immunophenotype supports the diagnosis of Rhabdomyosarcoma. Genetic analysis of the biopsy by RT-PCR detected the PAX3-FKHR gene translocation. Whole body FDG PET showed no suspicious uptake and or lesions. Altogether confirmed the diagnosis of an ARMS confined to cervical lymph nodes with no identifiable primary.

Patient was started neoadjuvant chemotherapy with vincristine, actinomycin- D and cyclophosphamide (VAC) followed by radiotherapy and adjuvant chemotherapy (VAC) for total 24-25 weeks cycle and on regular follow-up since 1 year. Now he came with the complaints of on and off fever, generalised weakness and myalgia since 1 month. On further workup, Complete blood count showed Pancytopenia with Haemoglobin - 6.9 g/dl, Total leucocyte count - $2.4x10^6/\mu L$ and Platelet count - $53x10^3/\mu L$. FDG PET scan revealed increased uptake in axial and appendicular skeletal marrow maximum in right iliac bone.

Bone marrow study showed suppression of trilineage haematopoiesis with presence of 80% atypical elongated cells arranged singly and in clusters floating in the intertrabecular region admist necrosis. These cells are large round to oval with abundant cytoplasm with cytoplasmic vacuolation and having irregular, hyperchromatic nuclei with conspicuous nucleoli favouring rhabdomyosarcoma cells. IHC on trephine biopsy showed tumor cells are strongly and diffusely positive for MyoD1, but negative for Desmin and confirmed marrow infiltration by ARMS Patient continued with chemotherapy and blood transfusions but still in a poor state of health.

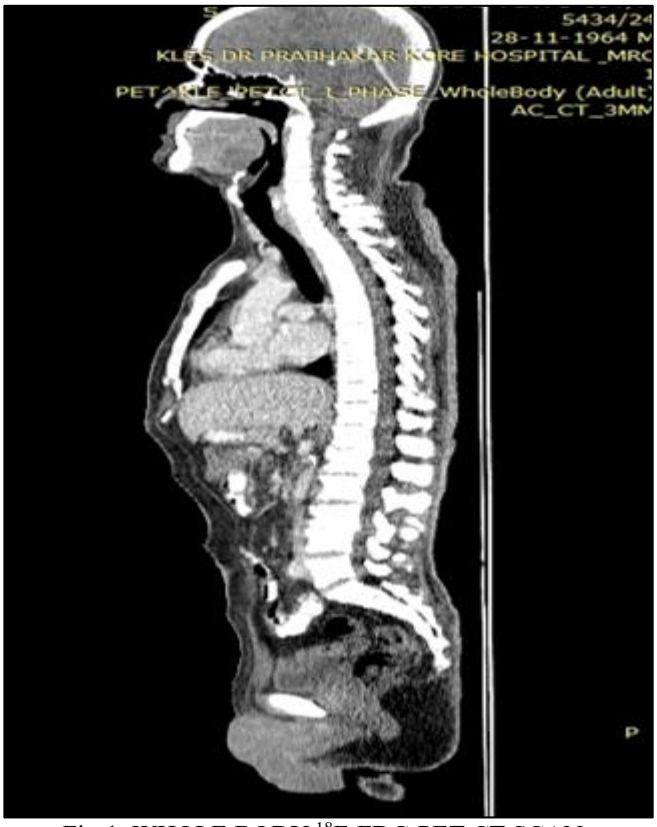

Fig 1: WHOLE BODY ^{18}F-FDG PET-CT SCAN- Heterogeneously Increased FDG Uptake in Axial and Appendicular Skeleton, Max in Right Iliac Bone with Osteophytic End Plate Degeneration Changes in Lumbar Vertebrae

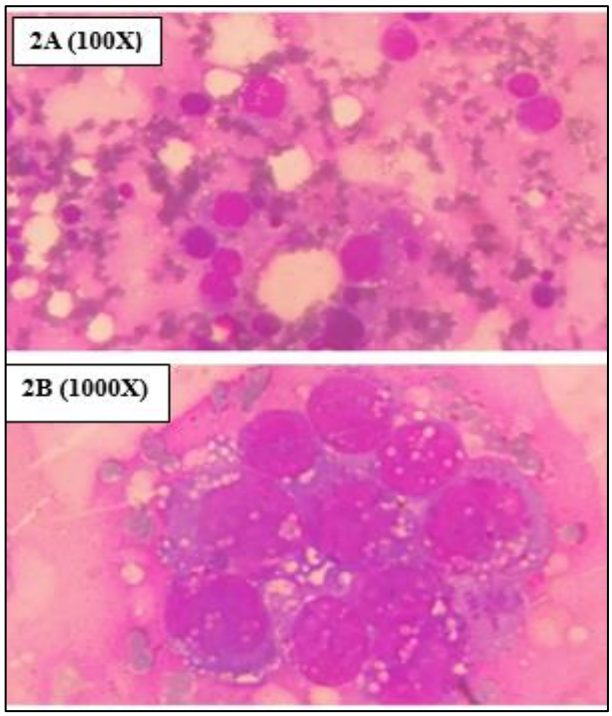

Fig 2A, B: Wright Giemsa-Stained Aspirate Smear Showing Atypical Large Cells with abundant Cytoplasm with Cytoplasmic Vacuolation and having Irregular, Hyperchromatic Nuclei with Conspicuous Nucleoli Cells Arranged Singly and in Clusters

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/25mar1530

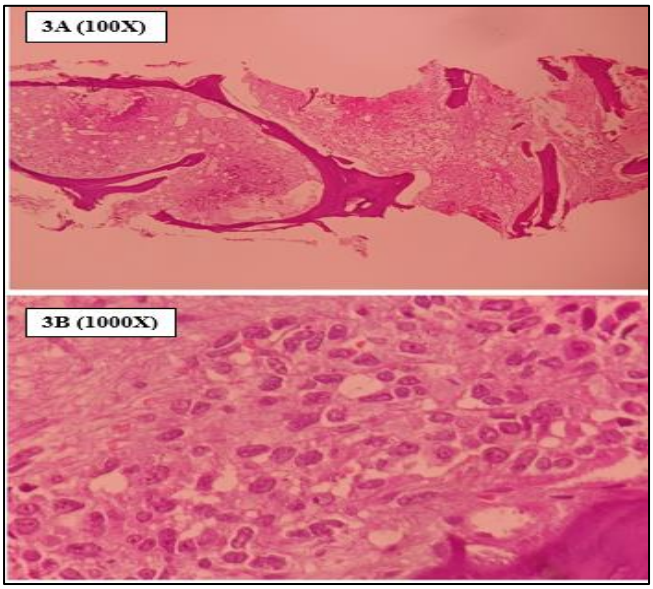

Fig 3A, B: Trephine Biopsy Showing Intertrabeculae Region with Marked Suppression of Trilineage Hematopoiesis by Atypical Round or Polygonal, Strap-Shaped with Eosinophilic Cytoplasm having Large Round to Ovoid Nucleus, Often Tilted to One Side, with Vesicular Chromatin and Conspicuous Nucleoli

III. DISCUSSION

The presence of metastases is one of the most adverse prognostic factors in RMS, and the bone marrow is a frequent site of tumor infiltration, but is rare in cervical lymph nodes. The VAC regimen is recommended for individuals with high-risk rhabdomyosarcoma (7). The patient's prognosis at the time of diagnosis was unfavourable and may not have been altered with the initiation of treatment at an earlier stage.

Table 1: A Systematic Review of Cases of Rhabdomyosarcoma with Symptoms and Metastatic Site

STUDY	AGE (Years) AND GENDER	PRESENTING SYMPTOMS	TYPE OF RHABDOMYSOSARCOMA	SITE OF METASTASIS
Panagiotis et al (8)	61/F	Shortness of breath and fatigue	Alveolar RMS	Bone marrow
Manasa et al (9)	33/M	Mass in left arm	Alveolar RMS	Pulmonary
Justin J et al (10)	67/F	Epistaxis with bony pain	Alveolar RMS	Bone marrow
Present study	60/M	Fever, generalised weakness and bony pain	Alveolar RMS	Cervical lymph nodes and Bone marrow

IV. CONCLUSION

ARMS is extremely aggressive, with a 5-year survival rate of approximately 27%, which is lower than the survival rates in paediatric populations (10).

The clinical characteristics of this case highlight that metastatic ARMS with unknown primary in adults and the elderly necessitates further investigation, making hemato pathological analysis essential for diagnosis, understanding disease progression, and guiding treatment decisions to resolve these unresolved issues.

REFERENCES

[1]. Meza JL, Anderson J, Pappo AS, Meyer WH. Analysis of prognostic factors in patients with nonmetastatic rhabdomyosarcoma treated on intergroup rhabdomyosarcoma studies III and IV: The Children's Oncology Group. J Clin Oncol. 2006;24(24):3844–51

[2]. Meyer WH, Spunt SL. Soft tissue sarcomas of childhood. Cancer Treat Rev. 2004; 30(3): 269–80.

[3]. Yassir H, Ngham H, Lyoubi M, Abada RA, Oukessou Y, Roubal M, et al. Alveolar rhabdomyosarcoma of the mandible in an adult invading the parotid gland: a rare case report. Int J Surg Case Rep. 2020; 77: 848–52.

[4]. Stepan K, Konuthula N, Khan M, Parasher A, Del Signore A, Govindaraj S, et al. Outcomes in adult sinonasal rhabdomyosarcoma. Otolaryngol Head Neck Surg. 2017; 157(1): 135–41.

[5]. Sorensen PH, Lynch JC, Qualman SJ, Tirabosco R, Lim JF, Maurer HM, et al. PAX3-FKHR and PAX7-FKHR gene fusions are prognostic indicators in alveolar rhabdomyosarcoma: a report from The Children's Oncology Group. J Clin Oncol. 2002;20(11):2672–9

[6]. Weiss AR, Lyden ER, Anderson JR, et al.: Histologic and clinical characteristics can guide staging evaluations for children and adolescents with rhabdomyosarcoma: a report from the Children's Oncology Group Soft Tissue Sarcoma Committee. J Clin Oncol. 2013, 31:3226-32. 10.1200/JCO.2012.44.6476)

[7]. Raney RB, Maurer HM, Anderson JR, Andrassy RJ, Donaldson SS, Qualman SJ, et al. The Intergroup Rhabdomyosarcoma Study Group (IRSG): major lessons from the IRS-I through IRS-IV Studies as

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/25mar1530

background for the current IRS-V treatment protocols. Sarcoma. 2001; 5(1): 9–15.

[8]. Manasa Dondapati, Vincent J, Ahmad S, Stern AS, Lieber JJ. Rare Adult Subtype of Rhabdomyosarcoma, a Common Childhood Soft Tissue Carcinoma. Journal of Investigative Medicine High Impact Case Reports. 2021 Jan 1;9:232470962110422-232470962110422.

[9]. Karagiannis P, Guth N, Thoennissen GB, Bern C, Sperveslage J, Ilske Oschlies, et al. Alveolar rhabdomyosarcoma confined to the bone marrow with no identifiable primary tumour using FDG-PET/CT. Clinical Sarcoma Research. 2015 Nov 18;5(1).

[10]. Cheng JJ, Mott RT, Savage PD, Paluri RK. Metastatic Alveolar Rhabdomyosarcoma with Extensive Bone Marrow Replacement in an Older Adult. Case Reports in Oncology. 2021 Oct 21;14(3):1505–10.