VOLUME - 10, ISSUE - 12, DECEMBER - 2021 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Pathology

A CASE REPORT OF A YOUNG MAN WITH ABDOMINAL MASS: HUNT FOR DIAGNOSIS

Dr K Pratyusha Postgraduate student, Dept of Pathology, BLDE (Deemed to be University), Shri B M Patil medical college, hospital and research Center, Vijayapur.

Dr Satish Arakeri* Associate professor, Dept of Pathology, BLDE (Deemed to be University), Shri B M Patil medical college, hospital and research Center, Vijayapur. *Corresponding Author

Dr Surekha Arakeri Professor and HOD, Dept of Pathology, BLDE (Deemed to be University), Shri B M Patil medical college, hospital and research Center, Vijayapur.

ABSTRACT

Introduction: Extraskeletal Ewing sarcoma/PNET is a small round cell sarcoma showing gene fusions of EWSR1-FLI1. **Case Report:** A 28-year-old male patient presented with right flank pain and low grade fever since 15 days. On examination: a mass was palpable in the right hypochondrium. Provisional diagnosis of Liver abscess has been made. USG abdomen shows features cystic lesion in the liver with internal septation ?Liver abscess /Hydatid cyst. Intraoperatively, tumor was seen attached to upper pole of kidney. Since tumor was large, it was ruptured intraoperatively and debulking surgery has been done. Under microscopy, tumor was arranged in sheets with intervening stroma showing desmoplastic reaction. Perivascular pseudorosettes are seen. The diagnosis of malignant small round cell tumor has been given. On immunohistochemistry, tumor cells are positive for Vimentin, CD99, NKX2.2 , FLI1, Neurofilaments, Synaptophysin with focal immunoreactivity for EMA, Pancytokeratin. Final diagnosis was EXTRASKELETAL EWING SARCOMA/PNET.
Discussion: Extra-skeletal Ewing sarcoma/PNET is malignant soft tissue tumor seen in chest wall, thigh, paravertebal region etc. Retroperitoneum is a least common site. Most common presentation is swelling in the soft tissue with compressive symptoms. Histologically, it is composed of undifferentiated small round cells.
Conclusion: Clinical examination and radiological findings leads to ambiguous diagnosis in Ewing sarcoma/PNET. Hence proper histopathological study is essential for final diagnosis.

KEYWORDS : Ewing sarcoma, Retroperitoneum, immunohistochemistry

INTRODUCTION:

Extraskeletal Ewing sarcoma/PNET is a small round cell sarcoma showing gene fusions involving one member of FET family of genes(usually EWSR1) and a member of ETS family of genes(E26 transcription specific), most commonly EWSR1-FLI1(85-90%).

Case Report:

A 28-year-old male patient presented with right flank pain, fullness in right upper abdomen and low grade fever since 15 days. On examination: a mass was palpable in the right hypochondrium extending 8cm beyond a right costal margin occupying epigastrium and right lumbar region. Mass has irregular surface, rounded border with firm to hard consistency. Mass moves with respiration and non ballotable. Provisional diagnosis of Liver abscess has been made.

USG abdomen shows features cystic lesion in the liver with internal septation ?Liver abscess/Hydatid cyst.

CT scan shows cystic lesion in the upper pole of kidney with infiltration into the IVC , suggestive of malignant growth of kidney. Intraoperatively, tumor was seen attached to upper pole of kidney. Since tumor was large, it was ruptured intraoperatively and debulking surgery has been done.

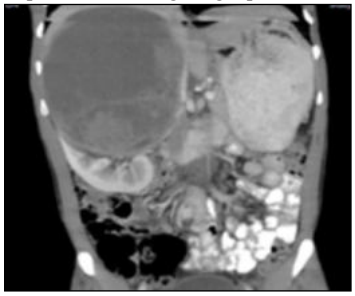

Fig no 1: CT scan showing cystic lesion in upper pole of kidney

Gross: Right nephrectomy specimen measuring 14x6x5cm, weighing 300gm. Cut section shows compressed kidney with near normal morphology. Above the kidney, there was an attached cystic mass filled with necrotic material. Cyst wall is thin. Well defined capsule was seen between cyst and kidney.

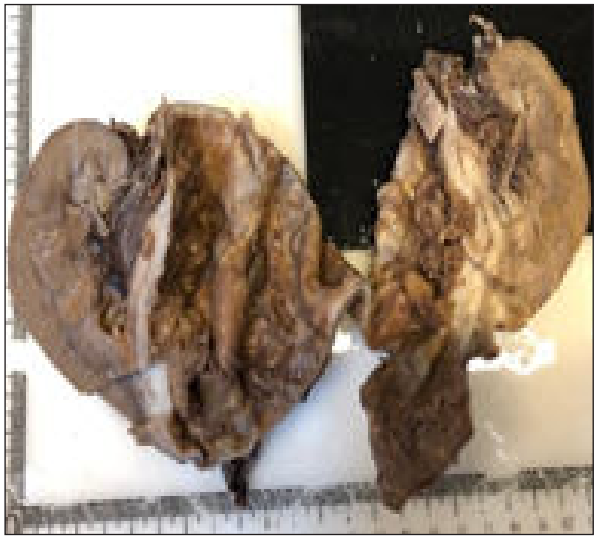

Fig no 2: Gross Image Of Right Nephrectomy Specimen

Under microscopy, cystic lesion shows tumor arranged in sheets with intervening stroma showing desmoplastic reaction. Tumor cells are small round to oval, hyperchromatic nucleus with scant amount of eosinophilic cytoplasm seen. Perivascular pseudorosettes are seen. Plenty of tumor necrosis is seen. Section from kidney shows well defined renal capsule with sclerosed glomeruli. On histopathology, the diagnosis of malignant small round cell tumor has been given.

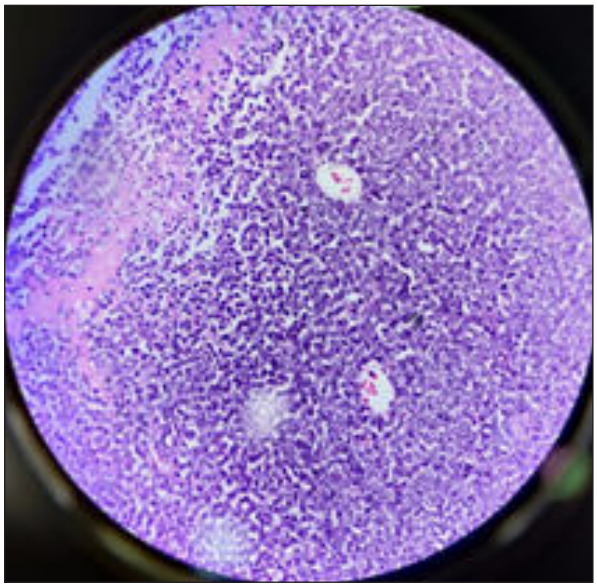

Fig no 3: Sheets of monotonous hyperchromatic cells with scant cytoplasm (H&E stain, 400x)

On immunohistochemistry, tumor cells are positive for Vimentin, CD99, NKX2.2 , FLI1, Neurofilaments, Synaptophysin with focal immunoreactivity for EMA, Pancytokeratin. Tumor cells are negative for ERG, WT1, SOX10, ALK, GFAP, TLE1, CD20, CD3, LCA. KI-67 index was 33-35%.

Final diagnosis made was EXTRASKELETAL EWING SARCOMA/PNET.

DISCUSSION:

Extraskeletal Ewing sarcoma (EES) is a rare disease which belongs to the Ewing sarcoma family of tumors. The incidence of EES is ten times lesser than the classical Ewing sarcoma of bone. It is most commonly seen in the age group of less than 5 years and more than 35 years1,2. The most common genetic profile seen is translocation t(11;22)(q24;q12) and t(21;22)(q22;q12). This translocation leads to fusion of FLI1 and EWSR1 gene3,4.

EES is a fast growing tumor with high tumor burden. It usually presents with pain and swelling on involved site. It is most commonly seen in thigh, arm and gluteal region. The most common sites for metastasis are lungs and bone marrow5,6.

The imaging modality which aids in diagnosis is ultrasound examination, computed tomography and MR imaging. Ultrasound imaging shows heterogenous mass with low echogenicity and increase vascularity within the tumor. On CT imaging, EES shows well defined mass with intensity similar to muscle bundles. On MR imaging, it shows low to intermediate signals on T1 weighted image7,8.

The modality of treatment is surgical debulking of tumor followed by chemotherapy and radiotherapy which will be decided on case basis.

Grossly the tumor is greyish color with areas of hemorrhage, necrosis and cystic degeneration. Under microscope, tumor cells are monomorphic, small, round cells with large nucleus, inconspicuous nucleoli and scant cytoplasm. On immunohistochemistry, tumor cells of EES are positive for CD99, S-100, Synaptophysin and FLI1. 9,10.

CONCLUSION

Clinical examination and radiological findings leads to ambiguous diagnosis in Ewing's sarcoma/PNET. Hence proper histopathological study with immunohistochemistry is essential for final diagnosis.

REFERENCES

1. Abboud A, Masrouha K, Saliba M, Haidar R, Saab R, Khoury N et al. Extraskeletal Ewing sarcoma: Diagnosis, management and prognosis (Review). Oncology letters 2021;21(5):1-6.
2. Srivastava S, Arora J, Parakh A, Goel RK. Primary extraskeletal Ewing's sarcoma/primitive neuroectodermal tumor of breast. Indian J Radiol Imaging. 2016 Apr-Jun; 26(2): 226–230.
3. Javery O, Krajewski K, O'Regan K, Kis B, Giardino A, Jagannathan J. A to Z of Extraskeletal Ewing Sarcoma Family of Tumors in Adults: Imaging Features of Primary Disease, Metastatic Patterns, and Treatment Responses. American Journal of Roentgenology 2011;197: 1015-1022.
4. Tural D, Mandel NM, Dervisoglu S, Dincbas FO, Koca S, Colpan Oksuz D et al. Extraskeletal Ewing's Sarcoma Family of Tumors in Adults: Prognostic Factors and Clinical Outcome. Japanese Journal of Clinical Oncology 2012; 42(5): 420–426.
5. Kennedy, John G, Stephen E , Robert C, David F, Brian H ET AL. Extraskeletal Ewing's Sarcoma - Spine2000; 25 (15) : 1996-1999.
6. F O'Keeffe, Lorigan JG, Wallace S. Radiological features of extraskeletal Ewing sarcoma. Br J Radiology 1990 ;63(750):456-60.
7. Murphey MD , Senchak LT, Mambalam PK, Logie CL, Klassen-Fischer MK, Kransdorf MJ. From the Radiologic Pathology Archives: Ewing Sarcoma Family of Tumors: Radiologic-Pathologic Correlation. RadioGraphics 2013; 33 (3): 1-5.
8. O'Keeffe F, Lorigan JG, Wallace S. Radiological features of extraskeletal Ewing sarcoma.The British Journal of Radiology 2014; 63(750): 70-76.
9. Chun-Fang Xi, Meng-Zhong L, Mian Xi. Extraskeletal Ewing's sarcoma: a report of 18 cases and literature review. Chin J Cancer 2010;29(4):420-4.
10. Murthy SS, Challa S, Raju KVVN, Rajappa SJ, D Fonseca, Gundimeda SD et al. Ewing Sarcoma With Emphasis on Extra-skeletal Ewing Sarcoma: A Decade's Experience From a Single Centre in India. Clinical pathology 2020;13:1-6.