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Pathology

MALIGNANT RHABDOID TUMOR OF MEDIASTINUM – A CASE REPORT

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ABSTRACT Malignant rhabdoid tumor (MRT) of the mediastinum is an extremely rare tumor. It follows an aggressive course. To date only 24 cases of mediastinal MRT have been reported in adults and 9 cases in paediatric age group (less than 18 years of age). (1) We report herewith a case of MRT in a 32 year old male of Indian origin who presented to emergency department with acute shortness of breath for 3 days. He had an enlarged left supraclavicular lymph node in which FNAC was performed which revealed metastasis from malignant rhabdoid tumor. Patient was further investigated and found to have a large mediastinal mass (the primary tumor).

KEYWORDS : Malignant, Rhabdoid, Mediastinum.

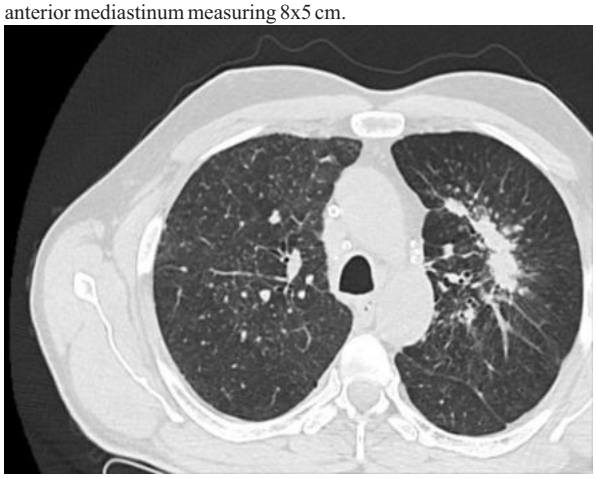
INTRODUCTION

A 32 year old male of Indian origin working as a bus cleaner presented to the emergency department with acute shortness of breath on exertion and at rest for 3 days. Patient gave history of cough which was productive with whitish expectoration which was more at night, not associated with hemoptysis. There was history of progressive ,unexplained weight loss and anorexia for 20 days. He used to chew tobacco for past 15 years. There was no history of fever or chest pain. No past history of Tuberculosis or any other severe illness.

Physical examination revealed tachycardia, tachypnea, and an enlarged left supraclavicular lymph node measuring 2x2 cm firm, nontender, mobile.

Initial chest radiograph revealed a large soft tissue opacity over the medial aspect of the upper zone of left lung.

Non contrast CT thorax showed a soft tissue density lesion in the anterior mediastinum measuring 8x5 cm.



FNAC of left supraclavicular lymph node was performed which revealed a cellular smear comprising of large non cohesive cells with abundant eosinophilic cytoplasm and large eccentric nucleus with prominent nucleoli and intracytoplasmic inclusion.

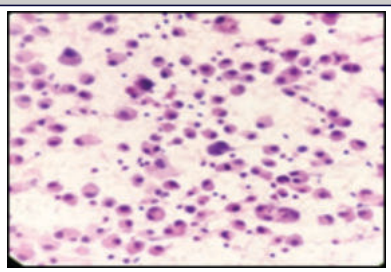


Fig 1: FNAC of supraclavicular lymph node

A provisional diagnosis of metastasis to lymph node by Malignant rhabdoid tumor was made.

Contrast enhanced CT thorax and abdomen was performed which revealed a large, well marginated mass with lobulated contour in anterior mediastinum measuring 8.5x8x8.5 cm. Mass was abutting the arch of aorta, main and left pulmonary artery and encasing left commom carotid artery and left subclavian artery. Multiple enlarged heterogeneously enhancing hilar lymph nodes were seen. Nodal mass of 3.6x3.8cm was noted in left supraclavicular region comprising the left internal jugular vein. Multiple well defined nodular lesions of varying sizes were noted in bilateral lung parenchyma in random distribution. Hypoattenuating lesion was noted in segment IVB of righ lobe of liver, heterogeneously enhancing nodular lesions were noted in bilateral adrenal glands suggestive of metastasis. Severe hydronephrosis of right kidney was seen suggestive of pelviuretric junction obstruction.

CT guided biopsy of anterior mediastinal mass was performed. Tissue was processed and stained with hematoxylin and eosin.

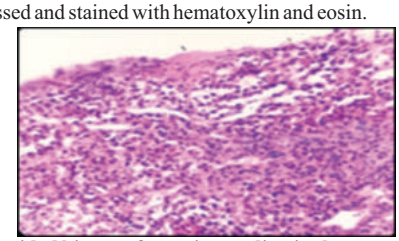


Fig 2: CT guided biopsy of anterior mediastinal mass.

Section revealed presence of round to oval hyperchromatic pleomorphic cells having eccentrically placed nucleus with prominent nucleolus. Cells show abundant eosinophilic cytoplasm with eosinophilic hyaline cytoplasmic inclusion confirming the provisional diagnosis of malignant rhabdoid tumor with metastasis to left supraclavicular lymph node.

Patient was started on cytoreductive chemotherapy with vincristine and cyclophosphamide after explaining risks involved. However his condition deteriorated thereafter, he developed type 2 respiratory failure and was put on mechanical ventilation. Patient succumbed within 7 days of diagnosis.

DISCUSSION

Rhabdoid tumors are malignant neoplasms of low prevelance, aggressive behaviour and high mortality.(1) To date only 33 cases of the mediastinal Malignant rhabdoid tumor have been reported in adults and 9 cases in the peadiatric age group.(2) The Rhabdoid tumor was originally described by Beckwith and Palmer in 1978.(3) It commonly arises in kidneys of young children before age of 1 year.(4)

Other rare extrarenal sites described are head and neck region, thorax, mediastinum, liver, uterus, adrenal gland, shine, genitourinary tract, retroperitoneum, trunk and extremities.(2) Although predominantly seen in paediatric population, we describe a case of malignant rhabdoid tumor in the anterior mediastinum in a young adult male .

Malignant rhabdoid tumors are known to show a bimodal age distribution, 1st peak in childhood and 2nd peak between 4th to 5th decade.(2) Mediastinal Malignant rhabdoid tumors usually present with chest pain, dyspnea, cough and respiratory distress which are nonspecific and are due to mass effect of the tumor.(5) some patients do have constitutional symptoms such as fever, loss of weight, loss of appetite,joint pain and generalized weakness.(5) Our patient had unexplained weight loss and anorexia.

Contrast enhanced CT thorax and abdomen revealed that our patient had both compressive and infiltrative features encasing the major blood vessels in the mediastinum along with metastatic lesions in left supraclavicular lymphnodes, bilateral lungs, liver, and both adrenal glands.

Radiologically the differential diagnosis for a solid anterior mediastinal mass is lymphoma, germ cell tumor, invasive thymoma. It may be very difficult to differentiate one from the other radiologically. (2)

Definitive diagnosis can be made on histopathological examination. Our patient had a preliminary diagnosis of metastasis from malignant rhabdoid tumor on fine needle aspiration of left supraclavicular lymph node.

The preliminary diagnosis was confirmed on CT guided biopsy of the mass in the anterior mediastinum. Immunohistochemistry performed on the cells revealed strong and diffuse staining for CD 34 and typically the tumor show lack of INI-1.

Molecular analysis reveals mutation or alteration in the SMARCB1/ INI gene which results in loss of INI-1 expression.(6)

Due to its rarity ,no standard or consistently effective chemotherapy or radiotherapy regimen for mediastinal Malignant rhabdoid tumor has been established.(2)

These patients usually present at an advanced stage and is rendered unresectable with very poor prognosis.

CONCLUSION

Mediastinal Malignant rhabdoid tumors are rare tumors with an extremely aggressive clinical course. This diagnosis should be kept mind in the differential diagnosis of any aggressive anterior mediastinal mass. A biopsy is required to establish the final diagnosis.

Hence, Prompt evaluation and early diagnosis with a core biopsy should be done to initiate early treatment and maximize survival. Further research needs to be done to identify targeted therapy and improve the efficacy of treatment modalities.

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