September 2008

Rhabdomyosarcoma embryonal type: in a three and a half year old girl

Ameer A Khowaja
Aga Khan University

Saqib Ali Gowani
Aga Khan University

Asmatullah Khan
Aga Khan University

Saulat H. Fatimi
Aga Khan University

Rhabdomyosarcoma Embryonal type: In a three and a half year old girl

Ameer A. Khowaja[1], Saqib Ali Gowani[2], Asmatullah Khan[3], Saulat H. Fatimi[4]

Department of Biological and Biomedical Sciences[1], Medical students[2,3], Cardiothoracic Section, Department of Surgery[4]
Aga Khan University, faculty of health sciences, Karachi.

Abstract

Rhadomyosarcoma is the most common tumour of the soft tissues in infants and children[1]. We report a case of a 3 ½ year old girl who presented with a swelling over the neck. Swelling was diagnosed as Rhabdomyosarcoma (embryonal type). Tumour mass was found to be unresectable initially. The patient was subjected to multiple courses of chemotherapy, which shrunk the tumour significantly. There was simultaneous development of cystic lesions in upper lobe of right lung. Excision of the remnant rhabdomyosarcoma mass and biopsy of right lung with cystectomy of right upper lobe cystic lesion was performed. The patient fully recovered and experienced an uneventful 6 months of follow-up.

Introduction

Rhabdomyosarcoma (RMS) is a malignant tumour of striated muscle origin. It is derived from primitive mesenchyme that retained capacity for skeletal muscle differentiation. Rhadomyosarcoma is the most common tumour of the soft tissues in infants and children[1,2]. It is associated with strong family history[3]. Rhabdomyosarcoma is a frequent tumour in families affected by Li-Fraumeni syndrome[4]. RMS is also associated with Neurofibromatosis, Rubinstein-Taybi syndrome, Gorlin basal cell nevus syndrome, and Beckwith-Wiedemann syndrome. Most frequent sites are head and neck with orbit. Least frequent site is thorax and extremities[3]. Currently Rhabdomyosarcoma is classified into six subtypes depending upon the histological features, that is, botryoid RMS, spindle cell RMS, embryonal RMS, alveolar RMS, undifferentiated sarcoma and RMS with rhabdoid features[5].

We report a case of 3 ½ year old girl who presented with a swelling over the neck whose diagnostic work up was suggestive of RMS (embryonal type) and was managed by our team.

Case Report

A 3 ½ year old girl was presented with swelling over the neck. Swelling increased in size within two to three weeks. Swelling was non-tender and was not associated with any other systemic ailments. Biopsy of the swelling revealed Rhabdomyosarcoma (embryonal type). CT chest showed a mass in anterior mediastinum and neck, extending to the major vessels and to the heart. Lung fields were clear and a small lymph node in cervical region was also appreciated. Bone Scan was done to exclude bony metastasis that showed no hot spots.

Since mass was unresectable at the time of diagnosis therefore the patient underwent multiple cycles of chemotherapy. The chemotherapeutic agents used were VDC (Vincristine, Doxorubicin, Cyclophosphamide), Etoposide, Ifosfamide and Mesna. During the course of chemotherapy she suffered from chemotherapy associated side effects which included alopecia, anaemia, irritability and febrile neutropenia and high grade fever. She also developed pharyngitis, otitis media and right-sided pneumonia. Blood cultures revealed growth of Staphylococcus Epidermidis. Chest X-ray showed right upper lobe haziness as a result of which pleural effusion was suspected. Ultrasound was done which was normal. On that occasion, another CT chest was done that revealed some cavitations on right side and cystic lesion in upper lobe of right lung. Febrile neutropenia in later part of chemotherapy was managed with Ceftazidime, Amikacin and Cloxacillin.

When the mass had reduced significantly and cystic lesion in upper lobe of right lung were seen it was discussed with the family and surgery was decided. With a median sternotomy approach, the excision was extended into the left side of the neck. There was no sign of any residual neck mass. Anterior mediastinum tissue was carefully cleaned off from innominate vein, the aorta and the pericardium. There was no extension into the pericardium. Phrenic nerves were identified and preserved. The mass was then carefully dissected off from the innominate vein and from the pleura. The left pleural cavity was entered and rest of the mass was excised. In the right pleural cavity the mass was adherent to right upper lobe. Cysts on the right upper lobe were identified and opened. They appeared to be chronic inflammatory type. A frozen section was sent which proved to be non-cancerous. The residual mass was then excised up to the neck along with its contagious lymph node. The mass was completely excised and sent for histopathology. The postoperative course was uneventful and satisfactory.

Antibiotic prophylaxis was continued. Patient was discharged in a good condition and followed up in the outpatient clinic. She made satisfactory progress.

Discussion

Though there are many reports and case series in literature available on rhabdomyosarcoma virtually of every organ of body, but to the best of our knowledge embryonal rhabdomyosarcoma of thorax with such atypical presentation as our patient had has not been reported yet. One has to have very high index of suspicion about rhadomyosarcoma. Presentation as our patient had with painless, non-tender swelling has not been documented in literature so far. Nevertheless rhabdomyosarcoma can present with myriad of clinical presentations primarily depending upon the region involved as evident by Japanese paediatric tumour registry analysis.[6] CT scan delineates much better surgical anatomy and response to chemotherapy on repeat scans. Therefore we resorted to CT scan as initial and only radiological diagnostic modality but Alemberger et al. have reported a case of thoracic rhabdomyosarcoma using ultrasonography as initial radiological diagnostic modality.[7] This may improve the cost-effectiveness but efficacy of repeat ultrasonography for following up the mass and/or postsurgical evaluation needs to be explored. Histopathologic studies remain the cornerstone for definitive diagnosis of embryonal rhabdomyoarcoma. Cohen et al. have demonstrated the role of pleural fluid cytology in diagnosing the rhabdomyosarcoma with its specific subtype.[8] Biopsy diagnosis is usually established per-operatively as frozen section specimen because of varied tumour location and accessibility. Though histological classification of rhabdomyosarcoma has academic importance it does not alter the management of patient much. Chemotherapy, radiation therapy, and surgical intervention have markedly improved the survival of patients treated for rhabdomyosarcoma. But the decision of chemotherapy before surgical resection depends upon size and location of tumour since chemotherapy have deleterious effects on the lungs as reported by Kaplan et al.[9] Our patient underwent chemotherapy because of unresectability of tumour at the time of presentation. Shaving of major thoracic blood vessels encased by tumour presented a major surgical challenge that was taken care of and entire tumour was resected without damaging any major blood vessel. We presented a case of a child without any potential complication of RMS but involving least frequent site. All possible complications should be kept in mind while managing such patients who are in moderate to high risk group.

Conclusion

Rhabdomyosarcoma (RMS) is a malignant tumour of striated muscle origin associated with strong family history. Most frequently sites are head and neck with orbit, thorax and extremities being very less frequently involved. Patients present with complaint of an expanding mass and symptoms depend upon the tumour's location. Removal of tumours surgically should be done whenever possible.

References

1. Ries LAG, smith MA, Gurney JG, et al., eds. Cancer incidence and survival among children and adolescents: united stated SEER program 1975-1995. National cancer institute, SEER program, NIH publication no. 99-4649, Bethesda, 1999
2. Parkin DM, Stiller GA, Draper GJ, Bicber CA. The international incidence of childhood cancer. Int J cancer. 1988; 42:511-20
3. Green DM. Diagnosis and management solid tumors in infants and children. Boston: Martinus Nijhoff, 1985
4. Li FP, Fraumeni JF Jr. Mulvihill JJ, et al, A cancer family syndrome in twenty four kindreds. Cancer res 1988; 48:5358-62.
5. Newton WA Jr., Gehan EA, Webber BL, et al., Classification of Rhabdomyosarcomas and related sarcomas. Pathologic aspects and proposal for new classification-an international group rhabdomyosarcoma study. Cancer 1995;76:1073
6. Nakada K., The clinical features and prognosis of rhabdomyosarcoma: follow-up studies on pediatric tumors from the Japanese Pediatric Tumor Registry 1971-1980. Part II. Committee of Malignant Tumors, Japanese Society of Pediatric Surgeons. Jpn J Surg. 1990; 20:503-9.
7. Almberger M., Iannicelli E., Matrunola M., Schiavetti A., Capocaccia P., Integrated diagnostic imaging of primary thoracic rhabdomyosarcoma. Eur Radiol. 2001; 11:506-8.
8. Cohen I, Loberant N, King E, Herskovits M, Sweed Y et al., Rhabdomyosarcoma in a child with massive pleural effusion: cytological diagnosis from pleural fluid: Diagn Cytopathol. 1999; 21:125-8.
9. Kaplan E, Sklar C, Wilmott R, Michaels S, Ghavimi F, Pulmonary function in children treated for rhabdomyosarcoma. Med Pediatr Oncol. 1996; 27:79-84.