January 2011

Aggressive embryonal rhabdomyosarcoma presenting as schwanoma : a case report

Mohammad Sohail Awan
Agha Khan University, sohail.awan@aku.edu

Montasir Junaid
Aga Khan University

Zain A. Sobani
Aga Khan University

GLOBAL JOURNAL OF SURGERY 33 Global Journal of Surgery | Volume 2 | Issue 1 | January 2011 Aggressive embryonal…

Aggressive embryonal rhabdomyosarcoma presenting as schwanoma: A case report

Montasir Junaid, Zain A. Sobani, M. Sohail Awan*

Department of Otorhinolaryngology, Head & Neck Surgery, Aga Khan University Hospital, Karachi, Pakistan

**Author for correspondence: M. Sohail Awan, email: sohail.awan@aku.edu*
Received 13 Apr 2010; Accepted 15 May 2010; Available Online 30 Dec 2010

Abstract

Rhabdomyosarcomas (RMS) are thought to arise from pluripotent muscle cell precursors intended to form the body's future striated muscle. Due to the wide distribution of striated muscle across the body these tumors may appear anywhere and cause symptoms accordingly. Presenting symptoms are highly variable and based on the tumor size and location making clinical diagnosis difficult. The diagnosis of RMS is based on histological examination; however they express varying cytologic pictures representing the different stages of rhabdomyoblasts and their development into skeletal muscle presenting pictures from highly to poorly differentiated neoplasms. Therefore their histological diagnosis also requires the use of specialized stains and complex staining procedures which may not be available at peripheral centers. Here we would like to present a highly aggressive embryonal rhabdomyosarcoma of the maxilla, presenting as a schwanoma on initial histopathology.

Keywords: Rhabdomyosarcoma; Embryonal; Schwanoma

1. Introduction

Rhabdomyosarcomas (RMS) are thought to arise from pluripotent muscle cell precursors called rhabdomyoblasts which are intended to form the body's future striated muscle [1]. Due to the wide distribution of striated muscle across the body, these tumors may appear anywhere across the body and cause symptoms accordingly; however due to some unknown mechanism the head and neck area is the most frequently involved site. Based on their histological features they may be classified as embryonal, alveolar, botryoid or spindle cell rhabdomyosarcomas. Around 250 cases of rhabdomyosarcoma are expected in the United States each year, with the embryonal subtype accounting for about 60% of them [1]. The five year survival for localized rhabdomyosarcoma ranges between 73-80% [2-3]. Here we would like to present a highly aggressive embryonal rhabdomyosarcoma of the maxilla, presenting as a schwanoma on initial histopathology.

2. Case report

A 12-year old male presented to our emergency room with complaints of a bleeding mass on the left cheek. He gave a history of a similar non tender mass at the same location not associated with any bleeding or discharge about 2 years ago which persisted for 18 months and regressed spontaneously. The mass reappeared 2 months ago upon which he sought medical attention in his hometown. The mass was excised and histopathological analysis showed a schwanoma. The mass reappeared one week later associated with on and off bleeding and he decided to visit our emergency room.

On examination a left buccogingival lesion with an oozing point was noted, keeping the previous histopathological diagnosis in mind, recurrence or residual disease from the previously excised schwanoma was considered and he was planned for surgery.

The lesion was excised using a sublabial incision to access the anterior wall of maxilla as described by Caldwell & Luc [4] and specimen was sent for histopathological examination. His post operative recovery was uneventful, and he was discharged awaiting final histopathology report.

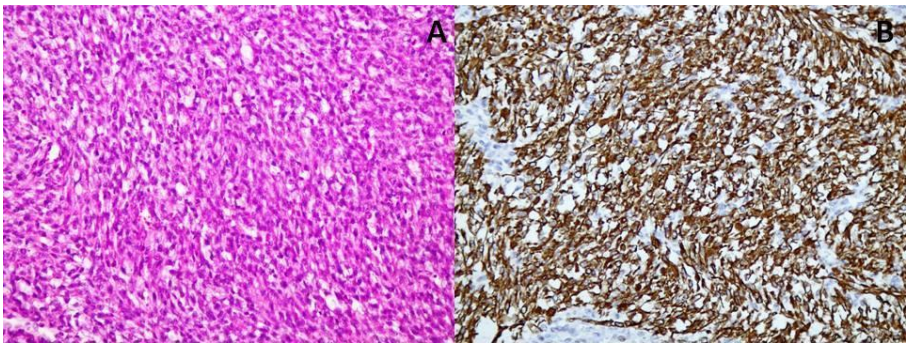

Figure 1. (A) H & E staining of the excised tissue at 20x showing embryonal rhabdomyosarcoma with sheets of moderate to poorly differentaited round cells with eosinophilic cytoplam and eccentric, small nuclei, (B) shows desmin positivity in these cells.

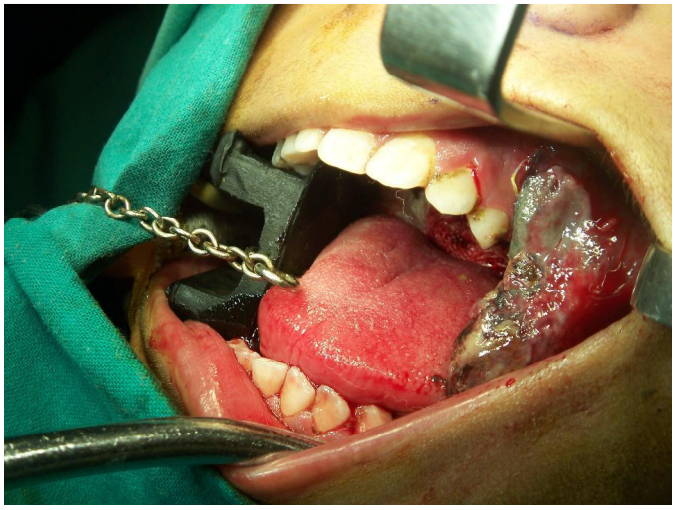

Figure 2. The lesion extending from the patients lip to the inner aspect of his buccal mucosa.

Following his discharge the final histopathology report showed embryonal rhabdomyosarcoma (Figure 1) and the patient was planned for further debridement; however he presented to the emergency room with a reappearance of the mass associated with bleeding on touch and the procedure was expedited.

The lesion was soft to touch extending from the lip to the inner aspect of the buccal mucosa (Figure 2). CT scan showed a large encapsulated lesion in the left maxillary sinus, eroding the medial and anterolateral maxillary walls and involving the buccal mucosa, measuring about 5.6x3.6x5.7 cm. Significant destruction of the left supra alveolar arch was also noted (Figure 3).

The tumor was surgically excised using a Weber Ferguson approach where the incision is made from the ipsilateral nasal vestibule laterally to the base of the nasal columella and extended transversely to the midline of the upper lip followed by a horizontal gingivo-buccal incision [4]. He is planned for further oncological management.

3. Discussion

Rhabdomyosarcoma (RMS) accounts for around 3.5% of all malignancies in children, with 5-year survival rates of 73%, according to the Intergroup Rhabdomyosarcoma Study (IRS)-IV. Presenting symptoms are highly variable and based on the tumor size and location making clinical diagnosis difficult [5].

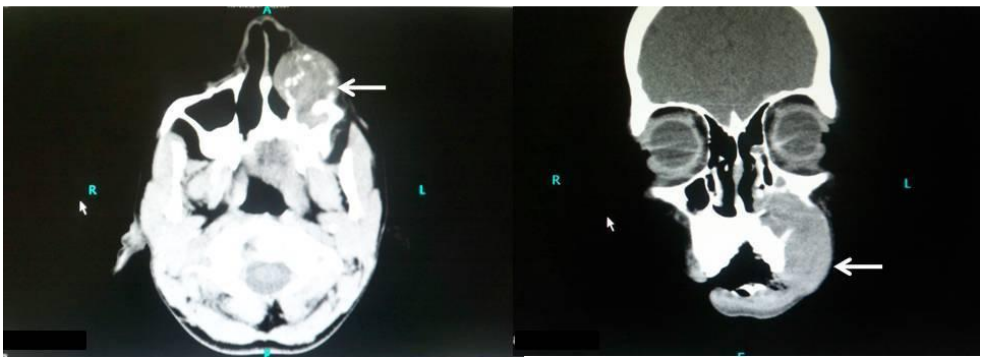

Figure 3. Axial and coronal CT scans showing the size of the lesion.

The diagnosis of RMS is based on histological examination; however they express varying cytologic pictures representing the different stages of rhabdomyoblasts and their development into skeletal muscle presenting pictures from highly to poorly differentiated neoplasms. RMS may however present without any signs of myogenic differentiation on routine staining procedures; requiring the use of specialized stains and complex staining procedures, for their histological diagnosis, which may not be available at peripheral centers [6], leading to an incorrect histological classification as may have been the case with our patient. Due to the rarity of this condition, RMS may be missed out even at major centers as this may not be considered among the differentials, in our country cases of RMS are even rarer on retrospective review of our records we found only 3 cases of embryonal RMS of the head and neck in the last five years.

The aim of treatment in RMS is local control with prevention of systemic metastasis by aggressive surgery with associated chemo and radiotherapy [7]; however the treatment for a schwanomma in a similar setting may not be as aggressive due to the lower chances of residual and recurring disease indicating that differentiation between the two prior to excision is of vital importance. Studies have reported an increased risk of embryonal RMS with high birth weight and increased gestational size suggesting that accelerated in utero growth may be associated with an increased risk for developing embryonal RMS; however the study failed to find such a correlation with other subtypes of RMS [8].

Although the prognosis of Embryonal RMS has been shown to be better than other forms of RMS it shows a frequent loss of the 11p15.5 allele, which has been suggested to possess tumor suppressive activity [9]. Another study comparing the chromosomal imbalances between different subtypes of RMS found frequent gains on 7p, 9q, 2p, 18q, 1p and 8q; losses on 11p in cases of embryonal RMS [10].

Autocrine signalling via vascular endothelial growth factor (VEGF) has also been implicated in the growth of the tumor, with increases in cell proliferation demonstrated on administration of exogenous VEGF. All-trans-retinoic acid has been shown to decrease VEGF secretion, leading to a decrease in cell proliferation in RMS [11]. Another study found that genistein arrested the cell-cycle at different checkpoints in RMS cells [12].

4. Conclusions

RMS can be very aggressive and extensive surgical debridement with chemo and radio therapy is recommended with close follow up over an extended time period as recurring diseases have been reported 20 years after initial multi modality treatment [13].

The authors recommend that even though a rarity in our population, RMS be kept in mind as a differential for pediatric soft tissue malignancies.

References

1. Drake, A.L., SC, Pathology – Rhabdomyosarcoma, eMedicine, (2009).
2. Crist, W.M., et al., Intergroup rhabdomyosarcoma study-IV: results for patients with nonmetastatic disease, J Clin Oncol, 19 (2001) 3091-3102.
3. Punyko, J.A., et al., Long-term survival probabilities for childhood rhabdomyosarcoma. A population-based evaluation, Cancer, 103 (2005) 1475-1483.
4. Paul W. Flint, B.H.H., Valerie J. Lund, John K. Niparko, Mark A. Richardson, K. Thomas Robbins, and J. Regan Thomas, Cummings Otolaryngology - Head and Neck Surgery, 5th Edition, Mosby (2010).
5. Parham, D.M. and D.A. Ellison, Rhabdomyosarcomas in adults and children: an update, Arch Pathol Lab Med, 130 (2006) 1454-1465.
6. Kodet, R., et al., Is fetal cellular rhabdomyoma an entity or a differentiated rhabdomyosarcoma? A study of patients with rhabdomyoma of the tongue and sarcoma of the tongue enrolled in the intergroup rhabdomyosarcoma studies I, II, and III, Cancer, 67 (1991) 2907-2913.
7. Abbas, A. ,S. Awan, Rhabdomyosarcoma of the middle ear and mastoid: a case report and review of the literature, Ear Nose Throat J, 84 (2005) 780, 782, 784.
8. Ognjanovic, S., et al., Birth characteristics and the risk of childhood rhabdomyosarcoma based on histological subtype, Br J Cancer, 102 (2010) 227-231.
9. Schaaf, G.J., et al., Full transcriptome analysis of rhabdomyosarcoma, normal, and fetal skeletal muscle: statistical comparison of multiple SAGE libraries, FASEB J, 19 (2005) 404-406.
10. Li QX, L.C., Chun CP, Qi Y, Chang B, Li XX, Chen YZ, Nong WX, Li HA, Li F., Chromosomal imbalances revealed in primary rhabdomyo-sarcomas by comparative genomic hybridization, Chin Med J (Eng), 122 (2009) 1277-1282.
11. Gee MF, T.R., Eichler-Jonsson C, Das B, Baruchel S, Malkin D., Vascular endothelial growth factor acts in an autocrine manner in rhabdomyosarcoma cell lines and can be inhibited with all-trans-retinoic acid, Oncogene, 24 (2005) 8025-8037.
12. Regenbrecht, C.R., et al., The molecular basis of genistein-induced mitotic arrest and exit of self-renewal in embryonal carcinoma and primary cancer cell lines, BMC Med Genomics, 1 (2008) 49.
13. Wight, R.G., et al., Rhabdomyosarcoma of the nasopharynx a case with recurrence of tumour after 20 years, J Laryngol Otol, 102 (1988) 1182-1184.