A Rare Case of adult Alveolar Rhabdomyosarcoma of the Lacrimal Sac

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ABSTRACT

Rhabdomyosarcoma is a common malignancy in children but rare in adults. The most common sites of involvement in adults are the extremities, genitourinary system and head and neck region. Rhabdomyosarcoma of the lacrimal sac is exceedingly rare, with only three cases ever reported in literature. A 23-year-old man presented with complaints of a painless swelling in the medial canthus of the right eye and epiphora. He was initially treated at a local hospital for acute dacrocystitis. There was no response and swelling persisted. Further evaluation including a CT scan showed a mass arising from the lacrimal sac with extension to the orbit. A core biopsy of the mass lesion revealed a lacrimal sac tumour. Histopathology and immunohistochemistry was suggestive of an alveolar rhabdomyosarcoma. He was started on chemotherapy with the IRS-4 protocol. This case report highlights the need to keep the possibility of malignancy in a case of a medial canthus mass, which responds poorly to antibiotics.

Keywords: Adult rhabdomyosarcoma, Epiphora, Lacrimal sac

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INTRODUCTION

Rhabdomyosarcoma is the third most common extra cranial malignancy in children. However, it is rare in adults and constitutes around 5% of all soft tissue sarcomas. It is of mesenchymal origin and can arise from almost any location in the human body. The most common sites of occurrence in adults is the extremities, genitourinary system and head and neck region. Involvement of the lacrimal sac is very rare with only three cases ever reported in literature. We present a case of primary rhabdomyosarcoma of the lacrimal sac in a 23 year old male.

CASE REPORT

A 23 year old male was presented with history of epiphora from the right eye and swelling in the medial canthus of the same eye of two months duration. He was initially

treated as acute dacrocystitis with oral antibiotics and when the symptoms persisted, a biopsy of the mass was performed which revealed a small round cell tumour and was referred to our institution. Clinical examination revealed a 2 x 2 cm hard mass at the inner canthus of the right eye with excessive lacrimation (Figure1).

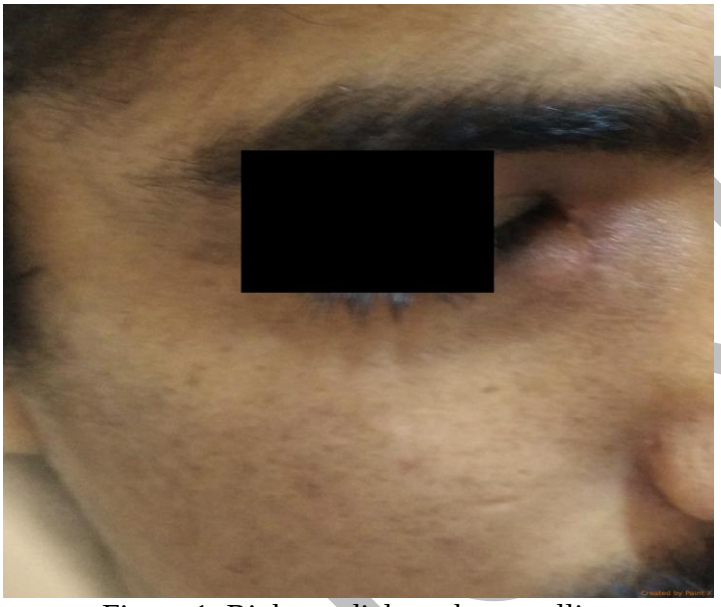

Figure 1: Right medial canthus swelling

There was no evidence of exophthalmos, conjunctival suffusion or sinus tenderness. On further evaluation, CT scan showed a 2.2 x 2.6 x 2.0 cm heterogenous soft tissue lesion in the infero-medial aspect of the right orbit, invasion of the post nasal cavity, ethmoid and frontal sinuses, loss of fat planes with the right eyeball and a 2x2 cm level 2 right cervical lymph node compressing the right internal jugular vein (Figure 2).

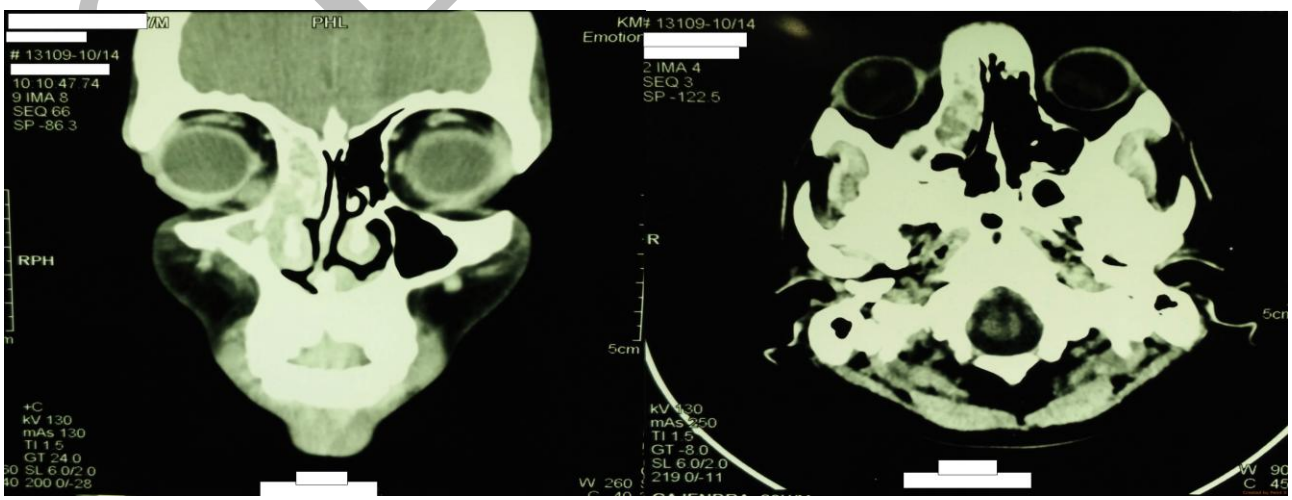

Figure 2: Contrast enhanced CT scan of PNS, Coronal and Axial views

Bone scan was normal except for an increased uptake in the medial aspect of the right orbit. Bone marrow aspiration and biopsy showed no involvement of the tumour. CSF analysis was also negative for abnormal cells. Histopathological review showed a small round cell tumour arising from the lacrimal sac. Immunohistochemistry revealed tumour cells positive for myogenin and desmin and negative for LCA, CK, synaptophysin, CD99 and chromogranin suggestive of alveolar rhabdomyosarcoma (Figure 3).

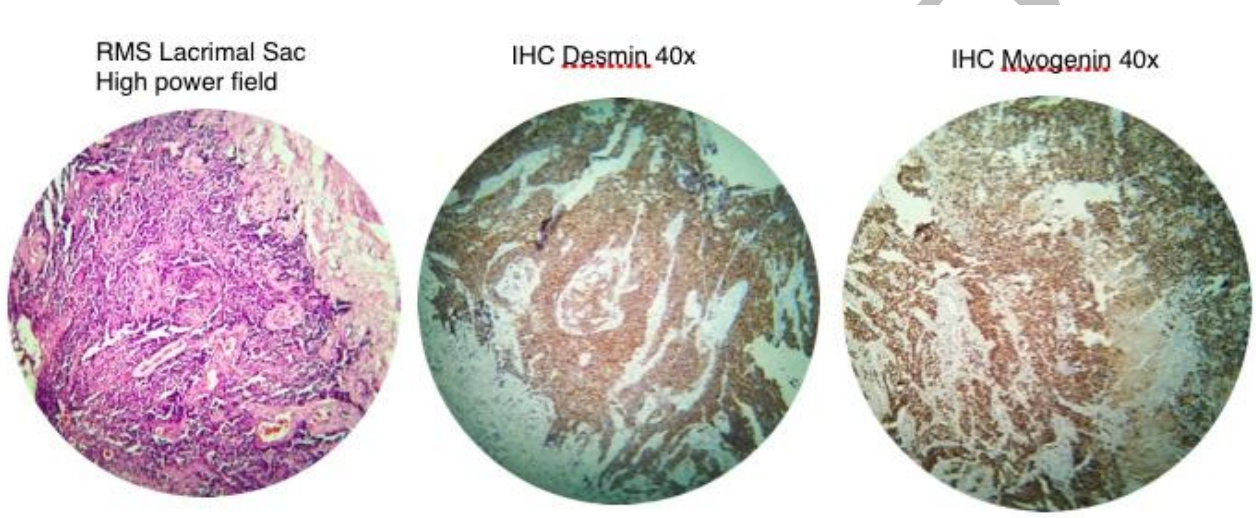

Figure 3: Histopathology high power field, Desmin 40x, Myogenin 40x

He was started on the IRS-4 protocol with Vincristine, Actinomycin-D and Cyclophosphamide with growth factor support.

DISCUSSION

Rhabdomyosarcoma is a malignant tumour of the mesenchymal tissue which exhibits skeletal muscle cells in varying degrees of differentiation.[1] It was first described by Weber in 1854.[2] It is the third most common extra cranial malignant tumor in children after neuroblastoma and Wilms tumour but is quite rare in adults. It constitutes only 5% of all soft tissue sarcomas in adults but around 60% in children under 15 years of age.[3] This tumor most commonly occurs in the head and neck, genitourinary tract, and extremities of children. In adults, the extremities, trunk and the genitourinary tract are the most common sites.[4] There are four histologic types, namely, embryonal, alveolar, pleomorphic and undifferentiated. According to a comparative analysis done in the SEER program, tumors in adults were more likely to be at an unfavorable site and to have histologies that are unusual during

childhood, particularly the pleomorphic subtype and not otherwise specified. [5] Adults had significantly worse outcome than children with similar tumors.

Theoretically, rhabdomyosarcoma can develop in any location due to its myogenic nature. The orbit is a common location for rhabdomyosarcoma in children and not unheard of in adults as well. However, involvement of the lacrimal gland with rhabdomyosarcoma in adults is exceedingly rare with only three cases reported in literature.[6,7,8]. Of these, two were of alveolar histology[6,7] and one was of embryonal histology[8]. As the initial histological diagnosis was that of a small round cell tumour, immunohistochemistry was done which showed positivity for design and myogenin which supported the diagnosis. Over 99 percent of RMS stain for polyclonal desmin, while muscle-specific actin, myogenin, and myoglobin positivity is found in approximately 95, 95, and 78 percent of tumors, respectively. Myogenin is an extremely reliable and specific marker for rhabdomyoblastic differentiation.[9] Myogenin has been associated with a poor prognosis independent of histologic subtype, tumor site, cytogenetics, and stage.[10]

Multivariate analysis in the SEER program showed that age, histologic subtype, primary site location, stage and local control with surgery and/or radiation were significant predictors of survival.[5] Although almost 30% of adults present with distant spread, our patient had no evidence of distant metastasis. However, the disease was locally advanced with regional nodal involvement.

The management of rhabdomyosarcoma involves multimodal therapy with surgery, radiotherapy and chemotherapy. As rhabdomyosarcoma is a high-grade tumour, the standard of care is neoadjuvant chemotherapy with or without radiotherapy followed by function sparing complete excision. Since it is primarily a disease of childhood, most studies were done in the paediatric age group and there is a paucity of data for treatment of rhabdomyosarcoma in adults. Hence, at our centre, all cases of adult RMS is subjected to the same chemotherapy protocol as of the pediatric age group. Hence, this patient was started on the International Rhabdomyosarcoma Study Group-4 (D9602) protocol.[11]

CONCLUSION

Although rhabdomyosarcoma of the lacrimal sac is an extremely rare entity, we conclude that the possibility of malignancy should be kept in mind in patients who present with a medial canthus swelling and epiphora who do not respond to antibiotics. Imaging and histopathology including

immunohistochemistry form an essential part of diagnosis and evaluation in such patients.

REFERENCES

[1] Durães G V, Jham B C, Mesquita A T M, Santos C R R, Miranda J L: Oral embryonal rhabdomyosarcoma in a child: A case report with immunohistochemical analysis. Oral oncology extra vol. 42 issue 3.p105 –108, March 2006.

[2] Pastore G,Peris–Bonet , Caril M , et al .Childhood soft tissues sarcomas incidence and survival in European children (1978 - 1997) : report from the automated children cancer information system project .Eur. j cancer 42 : 2136 , 2006 .

[3] Stuart A, Radhakrishnan J, Rhabdomyosarcoma, Indian J Pediatr 71 pp. 331–337 (2004)

[4] Tsuchiya A, Furukawa H, Endo Y, et al. Rhabdomyosarcoma presenting as an anterior cervical mass in an adult: report of a case. Surg Today 2000; 30: 1107-9

[5] Sultan I, Qaddoumi I, Yaser S, Galindo C R, Ferrari A. Comparing Adult and Pediatric Rhabdomyosarcoma in theSurveillance, Epidemiology and End Results Program, 1973 to 2005: An Analysis of 2,600 Patients J Clin Oncol Vol.27:Jul.10.2009: 3391-3397

[6] Neffendorf J E, Bagdonaite L, Mudhar H S, Pearson A R. Adult alveolar rhabdomyosarcoma of the lacrimal sac. Orbit. 2014 Dec;33(6):468-70. doi: 10.3109/01676830.2014.950295. Epub 2014 Sep 10.

[7] Woo K I, Kim Y D. Rhabdomyosarcoma of The Lacrimal Sac in 32-year-old Man. *J Korean Ophthalmol Soc*. 1996 Apr;37(4):702-707

[8] Lim J W, Lee S J. Primary Rhabdomyosarcoma of the Lacrimal Sac in a 68-year-old Woman.*J Korean Ophthalmol Soc*. 2006 Sep;47(9):1486-1490.

[9] Kumar S, Perlman E, Harris C A, M Raffeld, M Tsokos. Myogenin is a Specific Marker for Rhabdomyosarcoma: An Immunohistochemical Study in Paraffin-Embedded Tissues. Mod Pathol 2000;13(9):988–993

[10] Heerema-McKenney A, Wijnaendts LC, Pulliam JF, et al. Diffuse myogenin expression by immunohistochemistry is an independent marker of poor survival in pediatric rhabdomyosarcoma: a tissue microarray study of 71 primary tumors including correlation with molecular phenotype. Am J Surg Pathol 2008; 32:1513.

[11] Raney, R B, Walterhouse D O, Meza J L, Andrassy R J, et.al. Results of the Intergroup Rhabdomyosarcoma Study GroupD9602 Protocol, Using Vincristine and Dactinomycin Withor Without Cyclophosphamide and Radiation Therapy, forNewly Diagnosed Patients With Low-Risk

Embryonal Rhabdomyosarcoma: A Report From the Soft Tissue Sarcoma Committee of the Children's Oncology Group. J Clin Oncol Vol.29:No.10. Apr.1 2011 :1312-1318.