



Orbital-Ethmoidal Rhabdomyosarcoma in an Adult Patient: A Case Report

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Abstract

Rhabdomyosarcomas (RMS) is one of the most common sarcomas in children but rare in adults. It can occur in any anatomic location but most often presents in the head and neck region. Herewith, we report a rare case of primary orbital-ethmoidal rhabdomyosarcoma in a 29-year-old female. The patient underwent induction chemotherapy and achieved surprising results.

Subject Areas

Oncology

Keywords

Case Report, Rhabdomyosarcomas (RMS)

1. Introduction

Rhabdomyosarcomas (RMS), a malignant tumor of skeletal muscle origin, is one of the most common sarcomas in children. Although rhabdomyosarcoma (RMS) is rare in adults, accounting for 2 to 5 percent of adult sarcomas, approximately 40 percent of RMS cases arise in adults. It most often presents in the head and neck, approximately 35% - 40% [1]. Experience in treatment of adults with RMS is limited. Some data suggest that compared with children, adults have an inferior outcome [2]. We report a case of 29-year-old woman who was diagnosed with orbital-ethmoidal rhabdomyosarcoma, and had complete response with chemotherapy.

2. Case Presentation

A 29-year-old woman presented with a gradual protrusion of the left eye of a six-month duration. It was associated with pain, redness and eye discharge. She had no history of trauma, fever or any systemic problems. There was a proptosis

on the left-side eye. Palpation revealed a 5 cm firm mass in the superior of the orbit. The right eye examination was normal. The regional lymph nodes were not palpable (**Figure 1**).

The MRI of the head and neck showed a mass centered in the left nasoethmoidal region. It invaded the left side of frontal sinus, left orbit. It also extended to the meninges (**Figure 2**).

A biopsy was performed. Immunohistochemistry tests were positive for Desmin, myosin and negative for S-100, Melan-A, Synaptophysin and Chromogranin (**Figure 3**, **Figure 4**). Finally, the tumor was diagnosed as alveolar RMS.

She was treated with a combination of chemotherapy-VAC regimen, including Vincristine 1.5 mg/m²; Dactinomycine 0.045 mg/kg and Cyclophosphamide 1200 mg/m². The patient responded well to this regimen after 4 cycles and cancer reached complete remission (**Figure 5**, **Figure 6**).

3. Discussion

Rhabdomyosarcoma (RMS) is the most common soft tissue tumor of childhood and is accounting for one-half of all soft tissue sarcomas after osteosarcoma. Approximately two-thirds of cases are diagnosed in children younger than six



Figure 1. Left-side eye: proptosis, pain, redness and eye discharge.

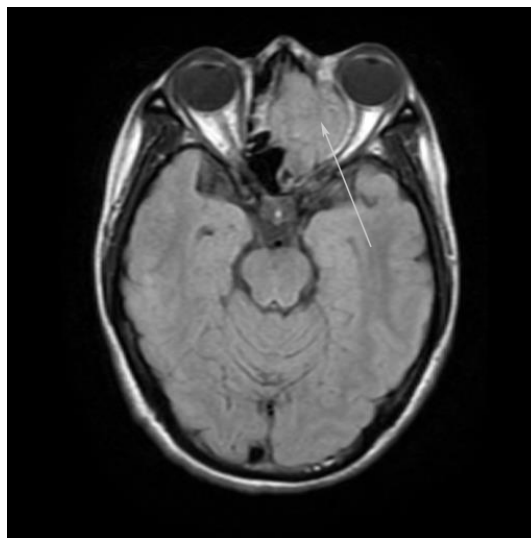


Figure 2. Mass centered in the left nasoethmoidal region (the white arrow).

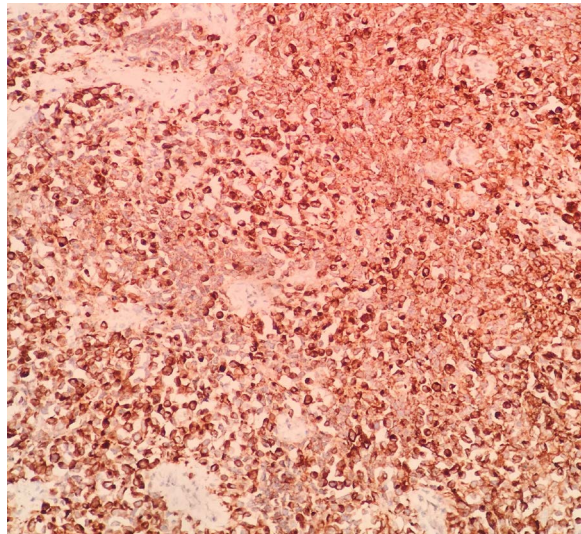


Figure 3. Desmin-positive staining.

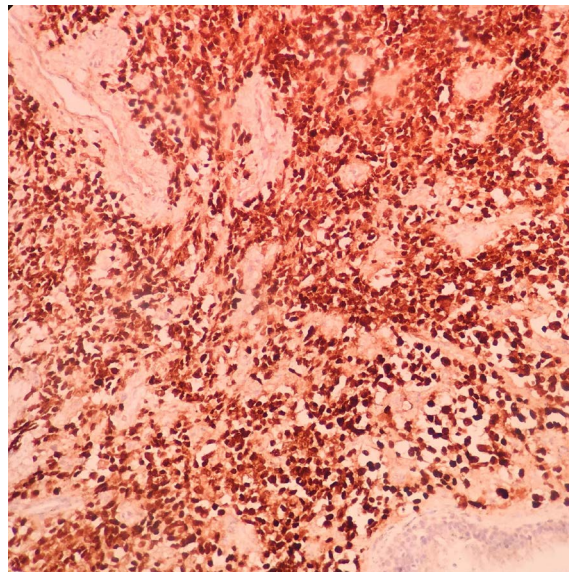


Figure 4. Myogenin-positive staining.

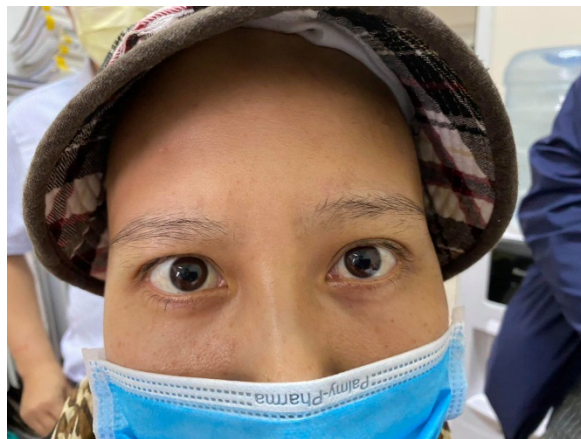


Figure 5. Her eye was treated with chemotherapy reached complete remission.

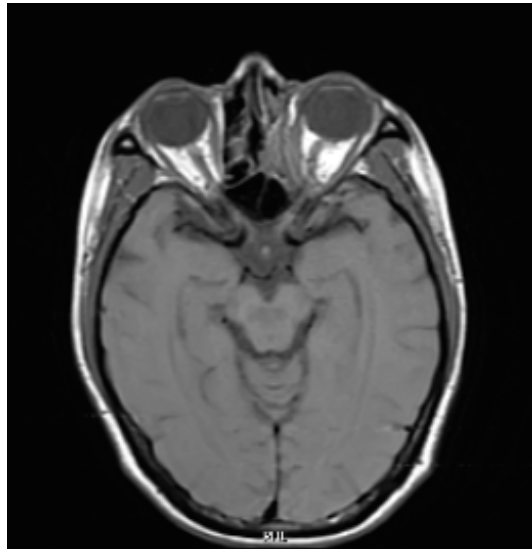


Figure 6. MRI image showed complete remission.

years of age, and there is a slight male predominance (male to female ratio between 1.3 and 1.5) [3]. Most cases of RMS are considered sporadic in origin, but there have been associations of RMS with several genetic syndromes (7 to 8 percent of the case). These syndromes associated with RMS include neurofibromatosis, Li-Fraumeni syndrome, Beckwith-Wiedemann syndrome, DICER1 syndrome, and Costello syndrome [4]-[10].

RMS has four different histopathological types: Embryonal, alveolar, pleomorphic and undifferentiated. The embryonal type is the most common, represents 70 percent of all RMS cases with an intermediate prognosis. Alveolar RMS is accounting for 21 percent of all cases with a relatively poorer prognosis. Alveolar RMS also presents with chromosomal translocations, including $t(2;13)(q35;q14)$ fused the PAX3 gene with FOXO1 gene and $t(1;13)(p36;q14)$ fused the PAX7 gene with FOXO1 [11] [12].

Patients with RMS can present with an asymptomatic mass or with signs and symptoms that are associated with the primary tumor site and the presence or absence of distant metastases. RMS can affect any body part. The most common primary site is head and neck (approximately 35 to 40 percent), the second common site are the genitourinary tract and the extremities (25 percent and 20 percent). Head and neck RMS include parameningeal site (almost half of all head and neck case); the orbit (25 percent of all head and neck case) and other locations (25 percent including the scalp, parotid gland, oral cavity, pharynx, thyroid, and parathyroid glands, and neck) [13]. Parameningeal lesions can cause nasal, aural, or sinus obstruction with or without a mucopurulent or sanguineous discharge while orbital tumors cause proptosis and ophthalmoplegia. RMS arising from sites other than these typically present as a localized, painless enlarging mass.

RMS should undergo diagnostic biopsy and staging evaluation before starting therapy. There are two classifications for RMS into risk-based therapy groups:

the clinical group (CG) and the tumor, node, metastasis (TNM) system. In 1972, the Intergroup Rhabdomyosarcoma Study Group (IRSG) developed the CG system. This system recognizes four disease categories based on the postoperative extent of disease with different prognoses (**Table 1**) [14].

TNM system has four disease stages (1 - 4) based upon the site and size of the primary lesion, regional nodal involvement, and metastatic disease (**Table 2**) [15].

Table 1. Clinical grouping of rhabdomyosarcoma by the intergroup rhabdomyosarcoma study group (IRSG).

Clinical group	Extent of disease/surgical result
I	A Localized tumor, confined to site of origin, completely resected
	B Localized tumor, infiltrating beyond site of origin, completely resected
II	A Localized tumor, gross total resection, but with microscopic residual disease
	B Locally extensive tumor (spread to regional lymph nodes), completely resected
III	C Locally extensive tumor (spread to regional lymph nodes), gross total resection, but microscopic residual disease
	A Localized or locally extensive tumor, gross residual disease after biopsy only
IV	B Localized or locally extensive tumor, gross residual disease after major resection (≥ 50 percent debulking)
	Any size primary tumor, with or without regional lymph node involvement, with distant metastases, irrespective of surgical approach to primary tumor

Table 2. TNM staging system for rhabdomyosarcoma.

Stage	Sites	Tumor stage invasiveness	T stage size	N	M
1	Orbit	T_1 or T_2	a or b	Any N	M_0
	Head and neck				
	Genitourinary Biliary tract				
2	Bladder/prostate	T_1 or T_2	a	N_0 or N_x	M_0
	Extremity				
	Cranial parameningeal Other				
3	Bladder/prostate	T_1 or T_2	a	N1	M_0
	Extremity				
	Cranial parameningeal Other				
4	All	T_1 or T_2	a or b	N_0 or N_1	M_1
T: Tumor stage		N: Regional nodes		M: Metastases	
T_1 : Confined to anatomic site of origin		N_0 : Not clinically involved		M_0 : No distant metastases	
T_2 : Extension		N_1 : Clinically involved		M_1 : Distant metastases present	
a: ≤ 5 cm in diameter		N_x : Clinical status unknown			
b: > 5 cm in diameter					

Currently, treatment for RMS is multimodality therapy, includes surgery, chemotherapy, and radiation therapy. The IRSG performed large collaborative randomized trials for the treatment of rhabdomyosarcoma. Since its inception, there have been five major trials, listed as the study I through V. As a result of these trials, the overall five-year survival rate of rhabdomyosarcoma at all sites has improved from 55 percent in IRS-I to 63 percent in IRS-II to approximately 71 percent in the IRS-III and IRS-IV protocols [16] [17] [18]. In head and neck tumors, the role of surgery is limited to initial diagnostic biopsy because of proximity to vital structures and cosmetic concerns. For COG treatment protocols, the three-drug combination of vincristine, actinomycin D, and cyclophosphamide (VAC) has been the gold-standard regimen of RMS. Different agents (Etoposide, Ifosfamide, Irinotecan, Topotecan, and Doxorubicin) or intensifying cyclophosphamide have attempted but they have not significantly improved clinical outcomes [19]. Radiation therapy (RT) is an important component of multimodality therapy for pediatric RMS. Induction chemotherapy, followed by concurrent chemoradiation, is the current standard of care for patients with the unresected disease and for patients with alveolar histology. RT is generally given after four cycles of chemotherapy.

In adults, there is a lack of standardized treatment. Generally, adults with RMS should be treated with the same treatment protocols as children although data are lacking with regards to some specifics, such as a way to integrate surgery, radiation, and chemotherapy in these patients or the total radiation dose.

Our patient at the time was diagnosed with RMS CG III. Therefore, she was treated with a VAC regimen. She had a clinical complete response to induction chemotherapy. She is intended to continue concurrent chemoradiation.

4. Conclusion

RMS is a rare head and neck tumor that occurs in the adult population and has a poor prognosis despite aggressive therapy. We believe that with multimodality therapeutic protocols and the best supportive care for complications may improve the prognosis of patients with this disease.

Acknowledgements

We have the patient's consent to report her clinical case.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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