Rhabdomyosarcoma (RMS) is a highly malignant, soft tissue neoplasm arising from undifferentiated mesenchyme.1 Its one among the most common soft tissue neoplasm occurring in the head and region with an incidence of 4.3 cases per million and around 10% of these RMS occur in the orbit.2,3 RMS is seen during the first decade of life till seventh decades. It comprises 4% of the pediatric malignancies. The incidence of RMS as reported by Turner and Richmon in his study says that the annual percentage of RMS is increasing by 1.16%, and there is no statistical change in 5 years survival rate.4 Survival rate was usually dependent on the extent and the behavior of the disease rather than primary diseases. According to recent studies, the 5 years survival rate of orbital RMS is 84.3%, which is more favorable when compared to other head and neck sarcomas.

Orbital RMS was once considered to arise from extraocular striated muscles of the orbit or the adnexal structures. However, recently its believed to arise from undifferentiated mesenchymal cells that have an innate capacity to differentiate into striated muscles.5-7 In this article, we present a rare case of 9-year-old boy who was diagnosed with histopathologically proven RMS of orbit who had underwent a salvage left orbital exenteration following a chemotherapy. The main aim of this article is also to provide an overview of RMS of orbit, clinical features, investigations required, staging and various treatment modalities.

A 9-year-old boy reported to our hospital 7 months back with a chief complaint of large soft tissues mass on left eye (Figure 1). On clinical examination, he had intraorbital soft tissue mass arising from extraconal space. Computed tomography (CT) scan was taken, which revealed a 4 cm × 3 cm well-defined, homogenously hyperdense soft tissue mass in left intraorbital, extraconal postero-lateral quadrant arising from lateral rectus muscle with abnormal anatomy of the left orbit (Figure 2). An incisional biopsy was done, which was suggestive of malignant round cell tumor. An IHC was also done, which was suggestive of RMS. The patient was classified as Group III according to International group of RMS (IRS) and was started on preoperative radiotherapy and chemotherapy of vincristine, actinomycin and cyclophosphamide (VAC) for 16 weeks.

The patient reported back presently to our hospital with a chief complaint of nasal bleed and acute febrile illness from 2 days. A complete blood cell count was done which was suggestive of increased bleeding time. CT scan was taken to know the extent of the lesion that revealed. The lesion was reduced in size but not to a considerable extent and hence a plan of salvage surgery was done.

Orbital exenteration was done under general anesthesia. The approach was transorbital and ophthalmic artery was ligated. Globe, extraocular muscles and intraconal contents of the orbit were enucleated (Figures 3-5). A split skin graft
was taken from left thigh and placed intraorbitally with a gauze pack for 5 days (Figure 6). The graft was adherent to the walls of the orbit. The histopathology examination of the excised specimen was confirmed as RMS of the left orbit. The follow-up of the patient is for 1 month and the
patient is doing fine with no further complaints. We are planning to rehabilitate the patient with orbital prosthesis in the near future.

**DISCUSSION**

Orbital RMS is often insidious in their early stage and usually mimics other space-occupying lesions of the orbit. The clinical features of RBS are proptosis that develops rapidly over weeks or displacement of the globe that is usually downward and outward because two-thirds of this neoplasm occurs near superior-nasal quadrant of the orbit. But in our case the lesion was occurring in from superior-lateral quadrant with mild orbital displacement medially and downwards. There can also be a subconjunctival or eyelid edema with chemosis. If the lesion extends to posterior orbital, it can cause edema of the optic disk, choroidal folds and some degree of ophthalmoplegia. Local infiltration of the tumor is very common. There can be an intracranial extension of the tumor by bony erosion and perforation on the medial side of the orbit or in the inferior orbital wall with the extension into the maxillary sinus. Metastatic spread is very uncommon but if the disease is not treated, there is a propensity of the disease to metastasize to lung, bone and bone marrow via vascular invasion. Metastatic orbital RMS has very unfavorable prognosis however in a joint European-North American analysis has concluded that orbital site proved to have a favorable prognosis.8-10

Often a detailed clinical history is very important if there is an orbital mass during early childhood. The ophthalmologist is the first health care personal who evaluates the patient. Normally clinical symptoms are pain and visual loss.11,12 The history is important to have a differential diagnosis of the lesion. It can be idiopathic orbital inflammation, orbital cellulitis, lymphangioma, hemangioma, sarcomas, neuroblastomas, etc., Imaging and histopathological investigations plays a key role in accurate diagnosis. Orbital RMS usually occurs in extraconal space (37-87%) but can also extend intraconally (13-47%).9,11,13,14 In our patient, it was in extraconal space on the lateral margins extending in intraconal space. The mass is usually close to extraocular muscles but initially there is no enlargement of the muscle. In later stages, if there is pseudocapsular invasion, the borders of the muscle are irregular. Ultrasound findings and usage in orbital lesions are limited since the penetration into the deeper tissues is restricted. The findings are usually to differentiate whether it’s a cyst or a tumor, and RMS appear has well circumscribed, irregular mass with low to medium echogenicity with a variable intravascular flow pattern. CT and magnetic resonance imaging (MRI) are very important for the preoperative evaluation, staging, and treatment modality of orbital RMS. CT is important to find the detection of bone involvement or erosion and to know the extent and the margins of the tumor. The lesion appears as circumscribed, a homogenous isodense mass. There can be focal hemorrhage or necrosis, which can be visible with moderate contrast enhancement. A common finding is eyelid thickening, regardless of the lid involvement, and a less common finding is a cavitated mass with ring-like enhancement.12,15

MRI is important for the soft tissue contrast, for perineural invasion and to detect any intracranial spread. The lesion appears isointense with extraocular muscles and hypointense with respect to orbital fat on T1-weighted images and hyperintense with T2-weighted images. Both of these imaging modalities can be used pre- and post-operative for the follow-up. Pre- and post-contrast comparison is useful to best detect intracranial and adjacent paranasal sinus invasion. Bone scintigraphy is particular used to find for occult metastasis. Positron emission tomography (PET-CT) is very innovate imaging that is better detecting bone and lymph node metastasis. Its sensitivity is 90%, and specificity is 100%. In our case, we have perfomed CT scan to know the extent of the disease before giving chemotherapy and after giving chemotherapy.12,16,17

Orbital RMS can be confirmed by histopathological diagnosis. The biopsy can be incisional or excisional biopsy based on the clinical findings. Fine-needle aspiration cytology has very limited role. They histopathological types of orbital RMS are embryonal, alveolar and pleomorphic. The embryonal variety is most common with good prognosis when compared to alveolar variety. Our patient had embryonal type of variety. There is also a tumor relation of the orbit with respect to histopathological type. Embryonal variety is more common in superior nasal quadrant whereas alveolar type in inferior orbit. The embryonal variety comprises of 50-70% of orbital RMS.17-19 They are frequently seen with bipolar cells with tapered cytoplasmic processes and the cells seen cells are “tadpole-like” with long cytoplasmic extensions. These cells are usually arranged in an interlacing fascicular pattern. The alveolar variety comprises of 20-30% of orbital RMS and are characterized by ill-defined aggregates of poorly differentiated malignant cells that are loosely arranged and separated into irregular ovoid spaces by thin fibrovascular septa in an alveolar pattern which is absent in the “solid” form. Immunohistochemical studies can also be done with markers like antibodies against desmin (90%), muscle specific actin, myo D1 (71-91%) and myoglobin.
In our case, incisional biopsy was done before starting preoperative radiotherapy and chemotherapy which was confirmed after salvage surgery.20

Management of orbital RMS was introduced by the North American Intergroup RMS Study Group and European cooperative groups in early 1970. Prior to that, overall survival (OS) was about 25-30 with orbital exenteration being the prime modality of the treatment but later with adjuvant chemotherapy and radiotherapy the OS improved to 90%. Following biopsy, staging for orbital RMS is internationally uniformly done according to the IRS postsurgical staging system.18

Group I: Localized disease, completely resected (excisional biopsy).
Group II: Microscopic disease remaining after biopsy.
Group III: Gross residual disease remaining after biopsy.
Group IV: Distant metastasis present at onset.

Based on this staging the treatment modality is decided. Current management includes surgery, irradiation and chemotherapy depending on the stage. Group I patients are treated with chemotherapy only: Vincristine and actinomycin. Group II patients are treated with a combination of chemotherapy VAC and radiotherapy of 36 Gy. Group III patients are treated with a combination of chemotherapy (VAC) and radiotherapy of 45 Gy. Group IV are treated with a combination of intensive chemotherapy and radiotherapy. Our patient was in Group III who had undergone chemotherapy and salvage surgery for orbital exenteration.

Interstitial brachytherapy is being used for local treatment since 1991. The main aim of this protocol is to maximize local treatment and to avoid external beam radiotherapy (EBRT). Ablative surgery is performed as conservatively as possible with the effort not to sacrifice important tissue at the expense of possible microscopic remnants. Evaluation for the metastatic disease can be done with chest-CT, TC bone scan, and bone marrow punctures and trephines. Recurrence of the orbital tumors can occur in 17% of the cases with a mean time of 18 months with 92% of local recurrence and 8% distant sites. There is no clear protocol for the management of the recurrent lesions. Possible options include salvage surgery, radiotherapy (additional EBRT).16-19,21

Follow-up of the patients are very important with generally 3 months interval with a good ocular examination. The follow-up of our patient is for 1 month, and the patient is doing well.

CONCLUSION

Orbital RMS is one of the few fulminating and life-threatening diseases that present first to the ocular surgeon/ head and neck surgeon and hence prompt and accurate diagnosis and treatment is very important to limit the extent of the disease. Hence, a thorough knowledge of the clinical, radiographic and histopathological, features as well as the more recent advances in the management of this neoplasm is very essential.

REFERENCES


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