Case report - Unusual presentation of Embryonal Rhabdomyosarcoma

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Abstract: An eight year old girl presented to us with left superior orbital mass. On clinical and radiological investigations we made a differential diagnosis of veno lymphatic malformation or a hemangioma. The patient was treated with systemic steroids. However, she did not improve and the proptosis and swelling increased rapidly. The rapidly forced us to intervene surgically. The histopathological diagnosis came as embryonal rhabdomyosarcoma. This case highlights the different form a rhabdomyosarcoma can present with and alerts the treating ophthalmologist to have high degree of suspicion. Keyword: 1. Embryonal Rhabdomyosarcoma, 2. pediatric tumour 3. rapidly progressing, 4. orbital mass, 5. veno lymphatic malformation-Case An 8 year old girl from Kerala presented with complaints of rapidly progressive swelling in left eye for past 1 week following a fall. On presentation, she had a visual acuity of 6/6 in the right eye and 6/12 in the left eye. Clinical examination of the left orbit revealed a soft, non-tender, non-pulsatile, non-reducible mass in the superior periorbital region causing dystopia. (figure 1) Spontaneous lid opening was absent. There was no conjunctival congestion or chemosis and rest of the anterior as well as posterior segment examination was within normal limits. Both pupils were brisk. Ocular motility showed a limitation in elevation in left eye.

MRI done elsewhere previously revealed a 2.4×1.2×2.0 cm sized solid lesion in extra conal compartment of left orbit superiorly with inferior displacement of superior rectus muscle and left globe. The lesion was well defined with intensely homogenous enhancement. (figure 2&3) Haemangioma and lymphangioma were considered in the differential diagnosis. CT scan with contrast showed a poorly enhancing hypodense lesion involving superior quadrant of left orbit and blends with superior orbital tissues and the epicenter from upper eyelid originating from sub periosteal zone causing downward displacement of globe. Size of the lesion measured 3.6×2.6×2.4cms suggestive of a veno-lymphatic malformation with bleeding.

(figure 2)

(figure 3)
Based on these clinical and radiological findings we made a differential diagnosis of veno lymphatic malformation or a hemangioma. We started her on IV Dexamethasone, and continued to treat with Tab. Wysolone 5 mg od. The patient presented to us again after 4 days with a rapid increase in size of the swelling associated with severe conjunctival congestion. (figure 4) We decided to go ahead with an incision and drainage of the upper lid under G.A. Surprisingly on table the tap was dry and the mass was tense. Hence, we went ahead with an incision biopsy of the mass for Histopathology of the biopsy specimen showed structure of fibrocollagenous tissue with infiltration of elongated hyperchromatic tumour cells in sheets with myxoid stroma. Areas of alternate hypercellular and hypocellular zones with interspersed elongated cells having cytoplasmic extensions, tadpole cells, extensive areas of perivascular necrosis with mitosis was seen suggestive of rhabdomyosarcoma. (Figure 5) The ImmunoHistochemistry was strongly positive for desmin, confirming the diagnosis of embryonal rhabdomyosarcoma.

Fig 4 and Fig 5

Metastatic workup was carried out. Blood investigations and USG abdomen were within normal limits. Bone marrow and CSF analysis were also within normal limits. A medical oncologist was consulted and advised for four cycles of chemotherapy with radiotherapy after each chemotherapy cycle. She was started on chemotherapy with vincristine, Actinomycin D and cyclophosphamide. After the completion of the first cycle of chemotherapy she has been planned for local radiotherapy.

DISCUSSION: Rhabdomyosarcoma should be considered in the initial differential diagnosis in any child who presents with proptosis. The main lesions that may simulate rhabdomyosarcoma clinically are orbital cellulitis, pseudotumour, dermoid cyst, cavernous haemangioma, lymphangioma, and lymphoma. Orbital lymphangioma can occasionally be difficult to differentiate from rhabdomyosarcoma. Like the latter, it is generally diagnosed in the first decade of life, and is characterized by a rapid onset of proptosis. This usually results from haemorrhage into a preexisting lesion, often after orbital trauma or upper respiratory infection. Imaging studies frequently reveal blood levels in the larger cystic spaces that comprise the mass. Such haemorrhagic cysts would be unlikely in rhabdomyosarcoma; it usually appears as a solid mass. However, occasionally a rhabdomyosarcoma can develop cavitary changes and masquerade as a lymphangioma.

Rhabdomyosarcoma is far the most common tumours of the orbits, and embryonal rhabdomyosarcoma type is the most common among the pediatric age group. Hence any child in the first two decades of life who presents with symptoms and signs of an orbital mass should be considered to have rhabdomyosarcoma until proven otherwise. The child should have a detailed history, examination and imaging studies like CT or MRI. Prompt biopsy should be considered whenever rhabdomyosarcoma is a reasonable possibility.

References: