Case Report

Primary extragonadal yolk sac tumor of the orbit: A rare case and review of literature

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ABSTRACT

Germ cell tumors usually arise from primordial cells in gonads, mainly testes, and ovaries. Extragonadal involvement of orbit is very unusual. We report one case of primary yolk sac tumor of orbit in a 3-year-old male child presented as a left lateral orbital mass. Its clinical presentation may mimic many common pediatric orbital tumors causing misdiagnosis which affects ocular morbidity and mortality. Awareness about this rare entity to clinicians is helpful in the early diagnosis and proper management of young kids.

Key words: Extragonadal germ cell tumor, Pediatric orbital tumor, Yolk sac tumor of orbit

Extragonadal germ cell tumor (GCT) of the head and neck is an extremely rare condition that comprises only 5% of all benign and malignant GCTs. Orbit is an uncommon site for the occurrence of extragonadal GCT and usually, these are orbital teratomas. Extragonadal GCTs can be highly aggressive and yolk sac tumor (YST) or endodermal sinus tumor is a malignant variant of extragonadal GCT. Pure YSTs are rare (20% of all cases) [1].

We describe the case of primary orbital YST in a 3-year-old male child who presented with a history of left lateral orbital mass. To the best of our knowledge, very few case reports or case series are reported in the English literature to date.

CASE REPORT

A 3-year-old male child presented with a history of the left lateral orbital mass which is gradually progressive causing forward protrusion of the left eye for 6 months. There was no history of fever, trauma, or any significant medical and sibling history.

On clinical examination, there was proptosis of the left eye with upper lid and lower lid swelling extending over the bulbar conjunctiva. The anterior and posterior segment examination of both eyes was unremarkable. Computed tomography of the orbit and brain showed a lobulated well-defined soft-tissue attenuation mass lesion measuring 30×30×25 mm as seen in the left orbit lateral preseptal compartment, the lesion was in contact with the left eyeball and seen markedly compressing and displacing it medially. Posteriorly, the lesion was extending into the lateral and superior extraconal post-septal compartment without any recti muscle involvement. No calcification, skull wall defect, or intracranial communication was seen. Clinical examination revealed no abnormalities of the abdomen and genitals.

His routine hematology investigations were within normal limits. Informed consent for biopsy was taken and the tissue was sent for a histopathological examination.

Grossly, multiple gray-brown tissue, the largest measuring 1.8×1.5 cm was seen. On cutting, a fleshy mucoid-like area was seen. The entire tissue was submitted for microscopy. Microscopic examination shows large cystic spaces lined by cuboidal flattened tumor cells having eosinophilic cytoplasm surrounded by myxoid stroma comprising spindle to stellate cells. At places, pleomorphic vesicular tumor cells having prominent nucleoli arranged in a papillary and glandular infiltrative patterns in muscles were seen (Fig. 1a-d). Overall, findings were suggestive of extragonadal GCT and YST of the orbit. The patient was advised for immunohistochemistry (IHC) and serum alpha-fetoprotein (AFP) levels for confirmation. Subsequently, it was confirmed by IHC and his serum AFP was increased to 2374 ng/mL (reference range/5.8 ng/mL).

DISCUSSION

The most common sites for extragonadal GCT in young children include midline structure, Sacrococcyx, and retroperitoneum.

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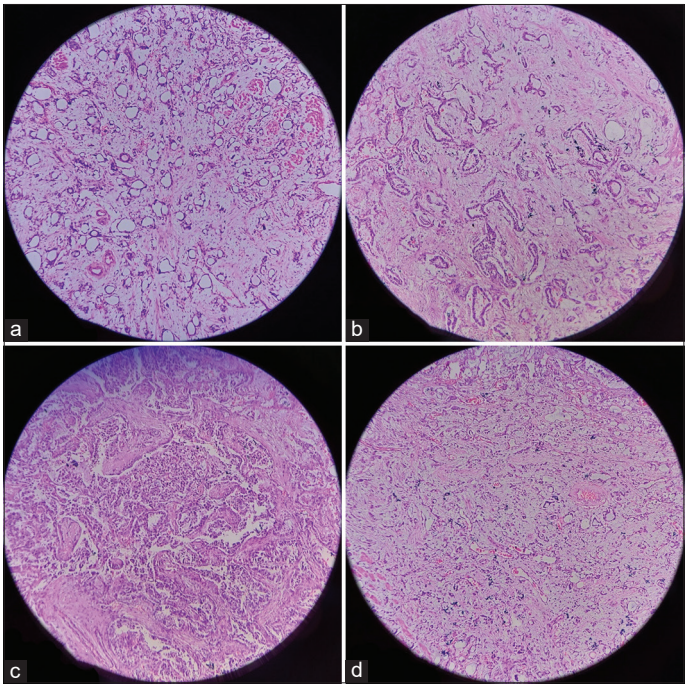

Figure 1: (a) (Hematoxylin and eosin stain, H&E [×100]) Tumor cells arranged in microcystic, reticular, and glandular pattern in loose myxoid stroma infiltrate in muscle; Fig 1(b) (Hematoxylin and eosin stain, H&E [×400]) – Pleomorphic, hyperchromatic tumor cells arranged in glandular, and papillary pattern in hyalinized stroma. Fig 1(c) (Hematoxylin and eosin stain, H&E [×400]) – Pleomorphic, hyperchromatic tumor cells arranged in glandular, and papillary pattern in hyalinized stroma Figure 1 (d) (Hematoxylin and eosin stain, H&E [×100]) Tumor cells arranged in microcystic, reticular, and glandular pattern in loose myxoid stroma infiltrate in muscle.

Head and neck comprise only 5% of all cases. Other reported rare sites include the jaw, cheek, upper lip, the floor of the mouth, and nasopharynx [1,2]. Malignant GCT accounts for about 3% of all malignancies in children [1,2]. Presenting symptoms may be non-specific and include proptosis, strabismus, and ophthalmoplegia.

Katz *et al*. reported the first case of YST in orbit in 1982 [3]. Devaney and Ferlito found 27 cases of YST of the head and neck region and in one-third of the cases; YST was associated with teratoma and the rest with malignant GCT or non-germ cell malignancies [4]. In the present case, no other tumor component was identified. Kamal *et al*. reviewed 16 patients with primary orbital YST, the most common presentation of orbital YST ranges from 3 months to 14 years with an average being 32 months (2.6 years) [4-6]. With slight female preponderance, the male: female ratio is 1:1.5 [5]. The mean duration of clinical presentation is 2 weeks and suggests that these tumors increase rapidly in size with the invasion of surrounding structures [5-7]. Kamal *et al*., Mishra *et al*., and Mogaddam *et al*. reported primary YST of orbit in a 2-year-old child, an 11-month-old infant, and a 1-year-old child, respectively [1,5,8]. Kumari *et al*. presented primary orbital YST as a fungating mass [9]. Ahmad *et al*. reported a case of in a 14-year-old girl, primary orbital YST with the invasion of periorbital and intracranial spaces [6].

Clinical differential diagnosis includes rhabdomyosarcoma, optic nerve glioma, Langerhans cell histiocytosis, myeloid sarcoma, metastatic disease from neuroblastoma, advanced retinoblastoma, and pseudotumor [5,7,8]. Initial histopathological misdiagnoses have been made in 5/14 (35%) reported cases. Due to the lack of awareness about this entity, many orbital YSTs are often misdiagnosed [5]. Orbital YST may have a better prognosis since these present at a younger age and may cause noticeable proptosis, or other symptoms/signs; therefore, early detection and proper management can be instituted [10].

AFP is a glycoprotein produced by the yolk sac and liver during fetal development [8]. Serum AFP level was raised in our case. Serum AFP levels are also useful to monitor the response to chemotherapy and identify early tumor recurrence.

Due to the rarity of the orbital YST, the clinical diagnosis is difficult and diagnosis solely rests on detailed histopathological examination. For this reason, optimum management guideline is not defined. Multidisciplinary discussion and multimodal therapy includes surgery, chemotherapy, and radiotherapy that may be needed to achieve optimum long-term disease-free survival.

CONCLUSION

The most common histological subtype of extragonadal GCT is teratoma, whereas YST as observed in our case is extremely rare. In our case, a 3-year-old male child having no other benign or malignant germ cell component was identified and diagnosed as a primary YST of the orbit. A high index of clinical suspicion according to age and presentation followed by biopsy, proper histopathology, IHC, and serum AFP level correlation clinch early diagnosis and proper management. Any delay may adversely impact the outcome of vision and survival.

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