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Case Report
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A Mimicker of Haemangioma: A Case Report of Orbital Alveolar Soft Part Sarcoma Excised by Endoscopic Transorbital Approach in A 3-Year-Old Child and A Review of Literature

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Abbreviations: ASPS: Alveolar Soft Part Sarcoma; MRI: Magnetic Resonance Imaging; CUSA: Cavitron Ultrasonic Surgical Aspirator

Introduction

Alveolar soft part sarcoma (ASPS) is a rare sarcoma arising from skeletal muscles or fascia favorably in lower extremities and trunks [1]. It was reported that the most common site was extremities while 15% was found in head and neck region in two series of pediatric and adolescent ASPS [2,3]. Pediatric ASPS may be misinterpreted as haemangioma or arteriovenous malformation by radiological investigations. Therefore, histopathological examination with immunopositivity of TFE3 is necessary for diagnosis of ASPS [4]. We present the case of a 3-year-old girl diagnosed ASPS with surgical excision done by both neurosurgeons and ophthalmologists. A comprehensive literature review was performed to investigate the clinical characteristic of pediatric orbital ASPS, treatment modalities and clinical outcomes.

Case Report

A 3-year-old girl was referred to ophthalmologists for right eye proptosis since July 2020. She had good past health with uneventful perinatal and developmental history. She did not report any visual impairment or diplopia. No suspicious skin lesion was noted.

On examination, her visual acuity of both eyes were 0.7 mm and the intraocular pressure were normal bilaterally. The exophthalmometery measurements of right and left eye were 15 and 12 mm respectively. Corneal diameters of both eyes were normal 11mm. Extraocular muscle movements were full without diplopia. Slit-lamp examination and fundal examination were unremarkable. Systemic physical examinations were also unremarkable. Magnetic resonance imaging (MRI) revealed a well circumscribed soft tissue mass at medial superior aspect of right orbit which measured 2.60 x 1.71 x 1.75 cm (Figure 1). It contained prominent internal blood vessels with no invasion to adjacent bony structures. Preliminary diagnoses were neuroblastoma or vascular tumors including haemangioma and vascular malformation. Urine vanillylmandelic acid and homovanillic acid were normal.

Right eye orbital mass biopsy was done via an endoscopic transcaruncular approach in October 2020 by ophthalmologists. The procedure was smooth, but the tumor was very vascular with torrents of bleeding after taking the biopsy which required meticulous haemostasis. The biopsy measured 4mm showed moderately cellular tumor comprising packets of epithelioid cells transversed by rich vascular septa. On immunostaining, tumor cells were stained TFE3+ and MelanA-ve. Features were compatible with TFE3-translocated neoplasm. Differential diagnosis included alveolar soft part sarcoma and perivascular epithelioid cell tumor which was not supported by the lack of melanocytic marker. On review of the imaging, the orbital mass

extended into the orbital apex and the ophthalmic artery was located just inferolateral to the tumor (Figure 2). It was anticipated that the possibility of ophthalmic artery injury was high in such a narrow space. In view of these, together with the biopsy results and high vascularity of the tumor, a decision was made for joint excision by neurosurgeons and ophthalmologists together.

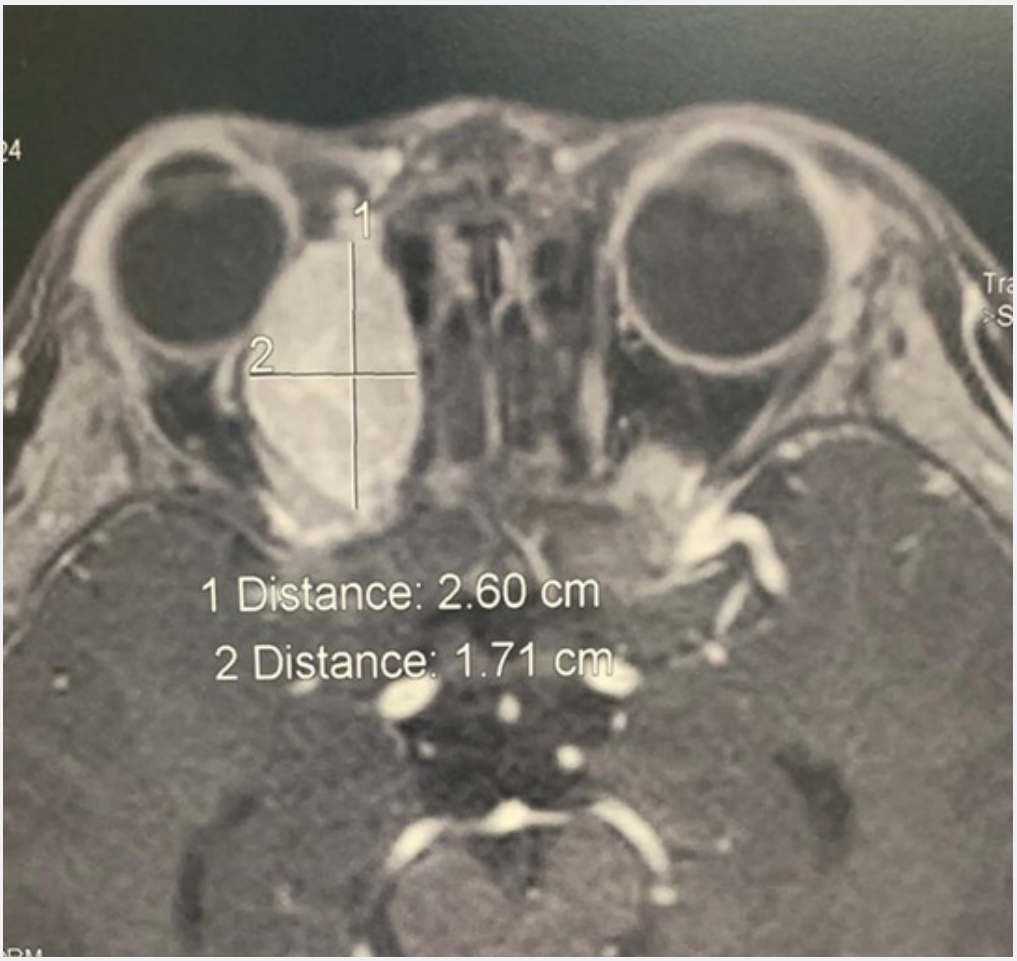

Figure 1: MRI T1 with contrast showing right orbital mass compressing onto the optic nerve.

Right subbrow approach for orbitotomy under endoscopic guidance was then performed by conjoint neurosurgeons and ophthalmologists for complete tumor excision in November 2020. Vascular tumor at superomedial side of orbit at apical region was found (Figure 3). Tumor was dissected from surrounding muscles and debulked by Cavitron Ultrasonic Surgical Aspirator (CUSA). Anterior and posterior ethmoidal arteries were cauterized and excised, and the tumor was removed enbloc phthalmic artery was found and carefully preserved during the procedure (Figure 4), there was visible pulsations of the artery under endoscopic viewing at the end of operation (Figure 5). Tumor specimen was sent for genetic testing. ASPSCR1-TFE3 chimeric transcript was detected. Overall features are suggestive of alveolar soft part sarcoma. The girl recovered very well after the operation, with no diplopia or visual compromise. Adjuvant radiotherapy was suggested however, it was declined by the parents. There was no recurrence of the tumor up until the time of writing of this report.

Literature Review

Search Strategy

A comprehensive literature search was performed to identify case reports of pediatric orbital alveolar soft part sarcoma. We searched PubMed databases until August 2021 to identify relevant articles. Appropriate key words and MeSH terms were used to identify all studies: “orbital”, “alveolar”, “soft”, “part”, and “sarcoma.”

Selection Criteria and Data Extraction

Studies eligible for inclusion were those that reported pediatric orbital alveolar soft part sarcoma in children aged 12-year -old or younger [5]. All studies selected are in English.

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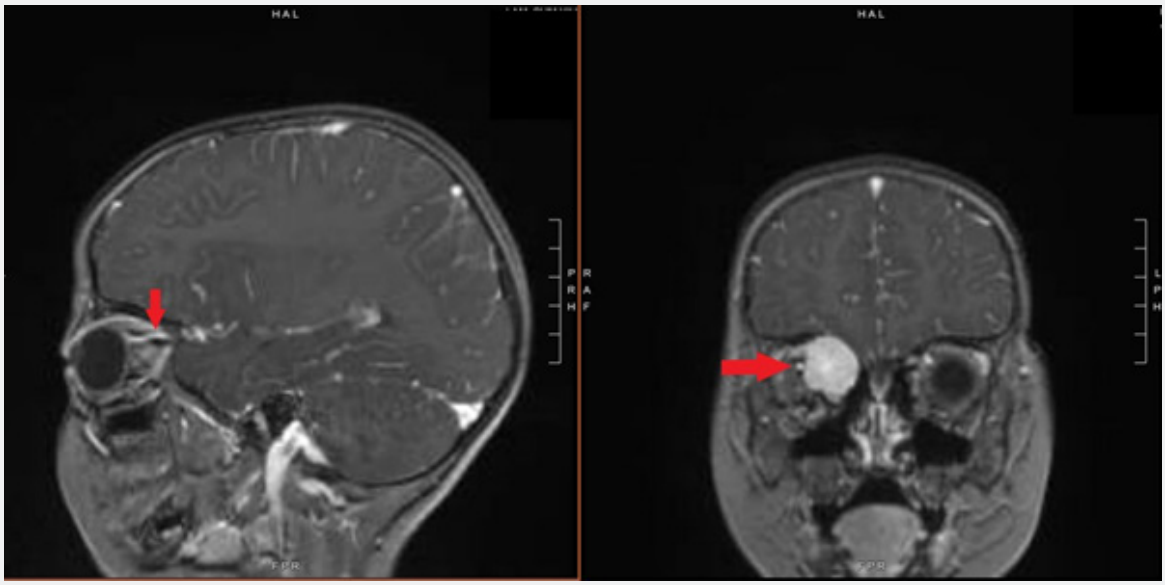

Figure 2: MR angiogram, sagittal and coronal cuts, showing relationship of ophthalmic artery (red arrow) with tumor.

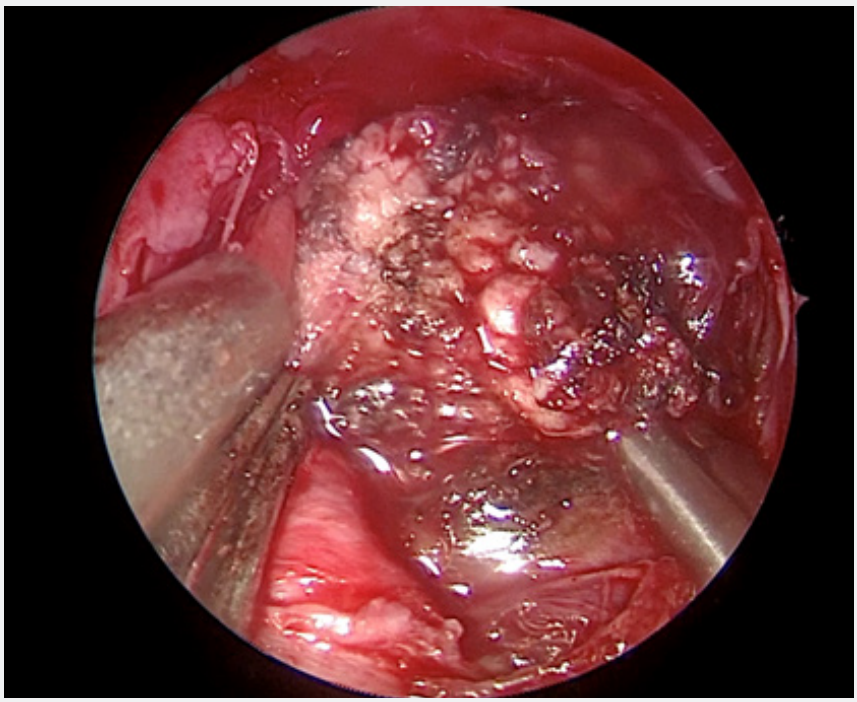

Figure 3: Intraoperative view of a fleshy and highly vascular tumor.

Study Selection

A total of 58 studies were identified through database searches and underwent title, abstract, and/or full-text review. 43 studies were excluded due to irrelevance and incomplete information. 15 studies [6-20] were included in the analysis after comprehensive review.

Study Characteristics and Outcomes

Our literature review found out a total of 29 cases of pediatric orbital alveolar soft part sarcoma. The patients' demographic characteristics, tumor location, treatment modality and outcomes

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of each case were listed in Table 1. The patients' median age at diagnosis was 6. 66% (19 cases) were female while 34% were male (10 cases). Pediatric Orbital ASPS affects primarily left eyes (72%, 21 cases) more than right eyes (28%, 8 cases). The most common presenting feature is proptosis (79%), followed by ocular motility restriction (48%) and lid swelling (41%). Treatment modalities and outcomes are summarized in (Table 1). For those have their treatment modalities mentioned, 18 patients underwent surgical excision and 8 underwent exenteration. 13 patients also received adjuvant radiotherapy. Of the 29 patients, 6 cases did not mention their clinical outcomes while 1 patient died during angiography. 16 patients had no local recurrence nor metastasis while 4 patients had local recurrence and 2 patients had metastasis. Only 1 case and our case involved neurosurgeons in surgery.

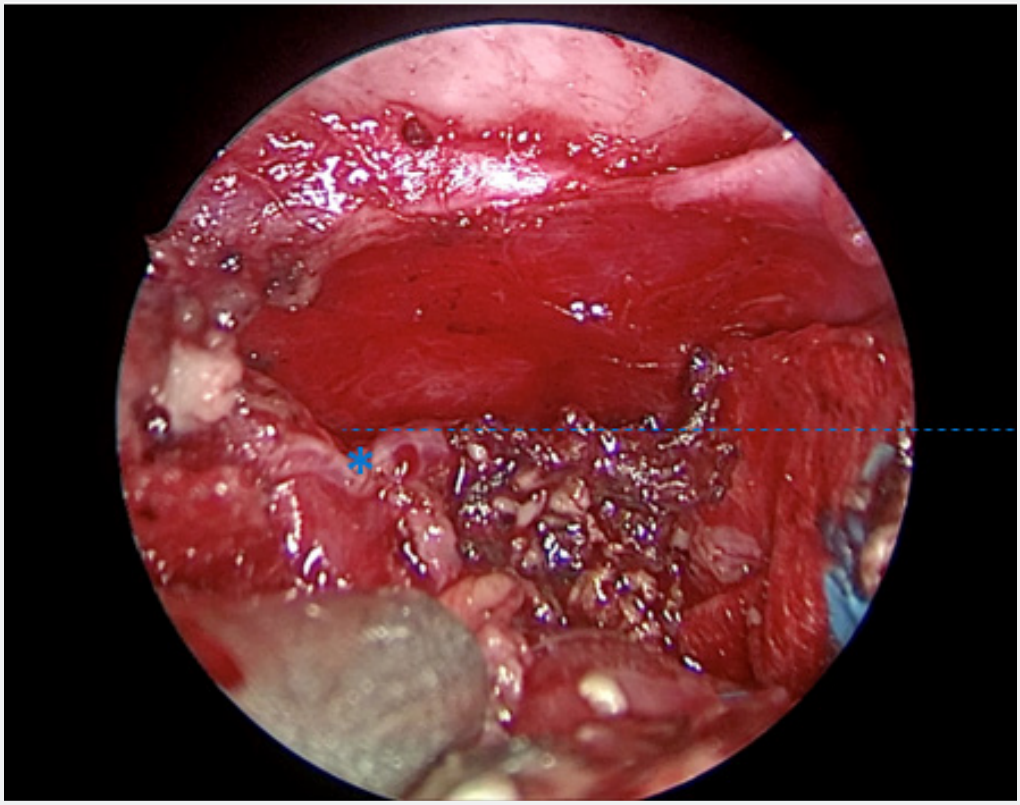

Figure 4: Endoscopic view of tumor removal at the orbital apex, with the opthalmic artery (*) well preserved.

Discussion

Alveolar soft part sarcoma is a rare neoplasm with characteristic immunohistopathological pattern infrequently reported worldwide let alone affecting orbits of pediatric group of patients. We analyzed data from 29 pediatric cases with orbital involvement of ASPS. Orbital ASPS in children showed the typical female predominance in our results [1]. Orbital ASPS in children usually present with proptosis, ocular motility limitation and lid swelling. If optic nerve is compressed, vision may be permanently affected. Interestingly, in our pooled cases, ASPS affected left eye more than right eye.

ASPS showed high vascularity features in CT imaging and frequently mistaken as vasogenic neoplasm such as haemangioma or lymphangioma before histopathological report available. However, radiological imaging would help identifying the extent of involvement and surgical planning. One patient died during angiography because of pulmonary metastasis. Immunohistological characteristic of ASPS includes organoid or nested architecture with thin, richly vascularized fibrous septae stained with TFE-3 marker positivity [21]. Currently, it is believed that tumorigenesis of ASPS involves the translocation t(X;17) p (11.2;q25). ASPSCR1-TFE3 transcription factor is also an important aid for the diagnosis of ASPS [4].

In view of its rarity, there is no evidence on the optimal management available yet. The mainstay of treatment in our literature review was surgical excision with or without adjuvant radiotherapy so as to preserve patient's vision. The surgical approach was mainly conventional orbitotomies, and only 2 required a transcranial approach for tumor removal.

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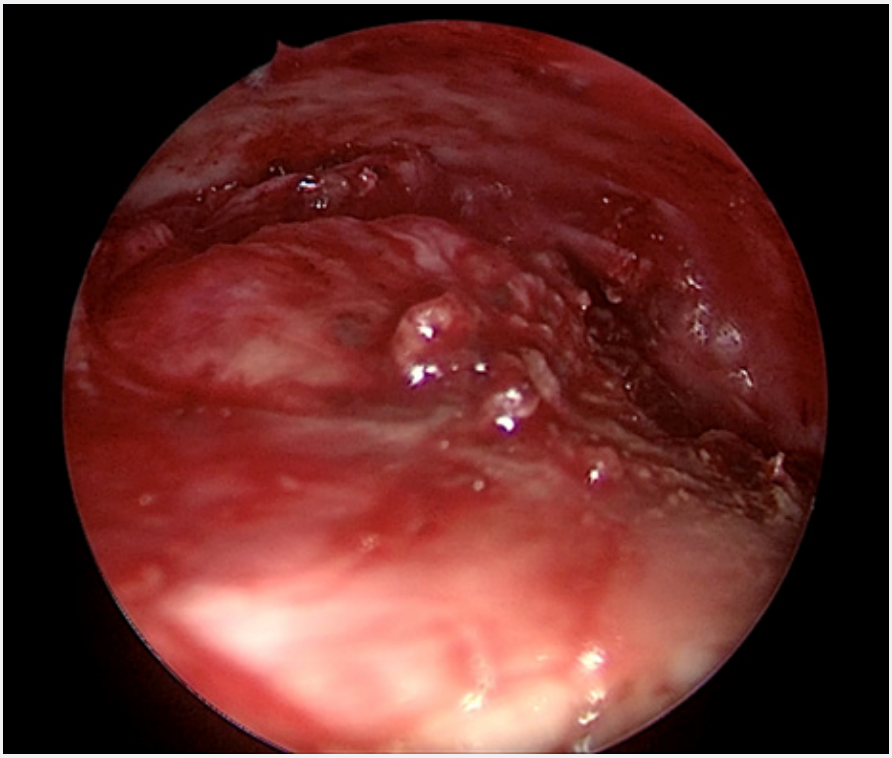

Figure 5: Complete removal of tumor via the endoscopic transorbital approach.

To our knowledge, this is the first reported case to adopt a joint excision by neurosurgeons and ophthalmologists via an endoscopic transorbital approach for tumors near the orbital apex. The endoscope enables us to have a better view of the vital structures during dissection of the tumor in the orbital apex. A good teamwork is required between the 2 surgeons operating on the patient in order to perform bimanual dissection techniques in this vascular tumor.

As these tumors are poorly circumscribed, post-operative radiotherapy is suggested in some practice to reduce the chance of local recurrence [22]. However, More studies evidence is required are needed to proof its effectiveness to reduce the chance of local recurrence provide evidence on the effect of adjuvant local radiotherapy. Larger tumor may need be excised by a modified or total exenteration to ensure complete tumor removal. Chemotherapy has limited use in treating metastatic ASPS [23,24]. The prognosis of ASPS with complete surgical excision is generally good with no local recurrence and metastasis.

Conclusion

Pediatric orbital alveolar soft part sarcoma is exceedingly rare. The most common presenting clinical feature was proptosis. Magnetic resonance imaging or computed tomography are important for tumor location and surgical planning. Endoscopic transorbital approach is a safe and novel technique in treating these patients. Complete surgical excision combined with adjuvant radiotherapy may be enough to prevent recurrence at the same time preserving vision of patients. Multidisciplinary team including ophthalmologists, neurosurgeons and radiologists is important for the holistic care for the patient.

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