Case Report

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# Alveolar soft-part sarcoma of the orbit

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## ABSTRACT

Alveolar soft-part sarcoma (ASPS) is a rare soft tissue tumour of uncertain cellular origin. It accounts for only 1% of all sarcomas, which themselves represent only a small proportion of human tumours. ASPS can arise in any soft tissue of the body, but there is an unexplained predilection for the right side. The most common site for paediatric ASPS is in the head and neck region, although involvement of the orbit is rare, with fewer than 30 reported cases. A case of a 5-year-old Kenyan boy with left-sided orbital ASPS is reported and the difficulties of diagnosing rare tumours are discussed.

**Key words:** Alveolar soft-part sarcoma, orbital, tumour

## INTRODUCTION

Alveolar soft-part sarcoma (ASPS) is an extremely rare soft-tissue tumour, with unique histological features, uncertain line of differentiation and variable clinical behaviour.[1] It represents less than 1% of all sarcomas, which themselves account for only 1% of adult and 15% of paediatric malignancies.[2,3] The tumour generally presents as a painless, slow-growing mass during adolescence or early adulthood, although childhood cases have been reported, with the head and neck being the most affected region.[4] Orbital ASPS is a very rare presentation of this tumour, with only eight cases reported in the last 25 years.[5-10] Eighteen further cases were reported prior to this.[11,12] ASPS has a female preponderance and a predilection for affecting the right side of the body; the latter being, as yet, an unexplained phenomenon.

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Since the first description of ASPS almost 60 years ago,[13] there has been much debate surrounding the cellular origin of the tumour. It has been claimed that ASPS may represent a malignant variant of granular cell myoblastoma, a non-chromaffin cell paraganglioma or a malignant angioreninoma.[1,14,15] In more recent years, the predominant theory has been that ASPS is a muscle-derived tumour, this being based on cellular morphology and molecular markers. However, it is now generally accepted that this tumour is a translocation-associated sarcoma, therefore having no normal cellular counterpart.

ASPS has an unbalanced translocation between chromosomes 17q25 and Xp11.2, resulting in the expression of a novel fusion protein.[16] This translocation joins two genes: a transcription factor, *TFE3*, on the X chromosome, and *ASPL*, a gene of unknown function on chromosome 17. The mechanism by which this fusion process causes disease remains unknown, but it has been hypothesized that loss of the upstream regulatory elements of the *TFE3* gene may play a role.[17] Translocations involving *TFE3* have been reported in some paediatric papillary renal cell adenocarcinomas.[1,18-22] Furthermore, very recent reports have shown an almost identical translocation to the one seen in ASPS in a case of adult-onset renal cell carcinoma.[23]

## CASE REPORT

A healthy 5-year-old Kenyan boy presented with a 2 year history of slowly progressive left proptosis and painless swelling of the left upper eyelid. There was no family history of neurofibromatosis.

Snellen visual acuity was 6/6 on the right, 6/60 on the left, with no relative afferent pupillary defect. There was left hypoglobus and moderate proptosis. Left hypotropia was associated with global restriction of ocular ductions and a complete absence of left

upgaze. Upper eyelid position was normal, with good levator muscle excursion. There was no intraocular abnormality. A rubbery, vermiform mass was palpable above the globe, this lying in close apposition to the upper sclera, with very large episcleral vessels visible both around and on the mass.

Computed tomography (CT) scan demonstrated a large, well-defined mass in the upper part of the left orbit, with passive remodelling of the orbital roof and downward depression of the orbital floor [Figure 1a]. The superior rectus and levator muscle complex could not be discriminated from the abnormal tissues [Figure 1b]. Imaging was suggestive of a significant vascular supply at the posterior pole of the mass [Figure 1c].

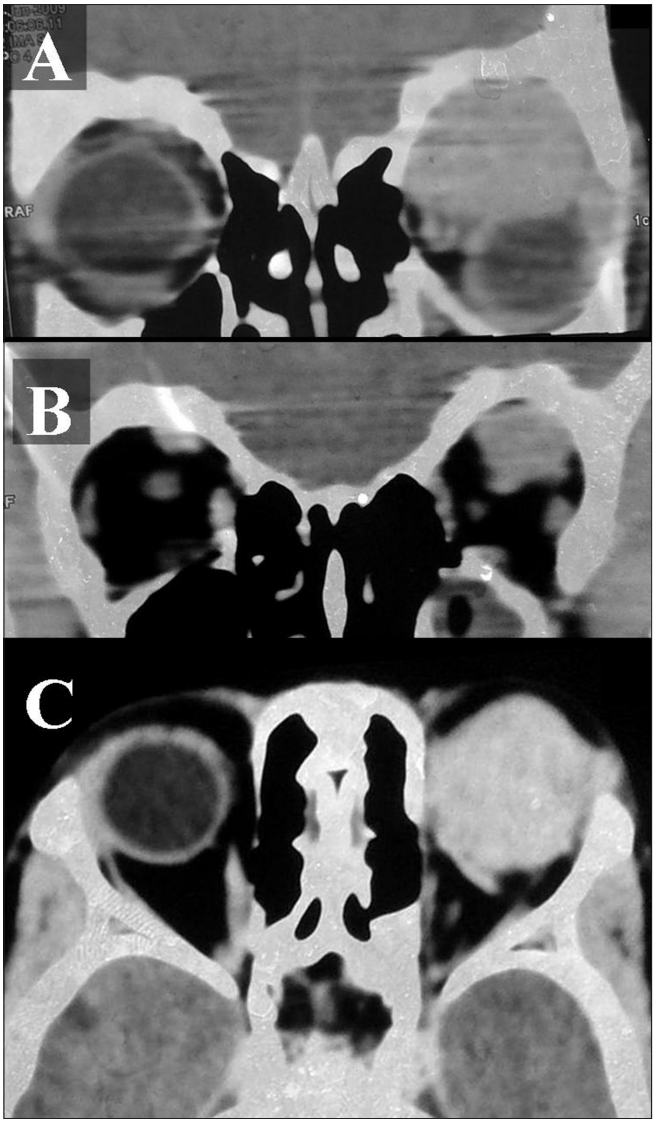



**Figure 1: CT imaging of tumour. (a) Coronal section showing hypoglobus and passive remodelling of the orbital roof and floor; (b) coronal section demonstrating absence of discernable superior rectus/levator muscle complex; (c) axial section showing significant vascular supply to tumour at posterior pole**

The tumour was resected through a trans-conjunctival superior orbitotomy, during which it was found to have replaced the left superior rectus muscle throughout its length. Large feeder vessels were evident both around and within the large, lobulated, grey-white tumour [Figure 2].

Histology showed a partially encapsulated tumour, comprising cohesive sheets of cells with mildly pleomorphic nuclei. There were small, but prominent nucleoli and copious pale cytoplasm which was eosinophilic and somewhat flocculent. Although partial thick, fibrous septa were present, an alveolar pattern was not prominent. Immunohistochemistry was negative for S-100 and smooth muscle actin, but diastase periodic acid - Schiff (PAS) staining highlighted intracellular granules which showed focally elongated crystalline structure. These histological findings are consistent with the diagnosis of ASPS.

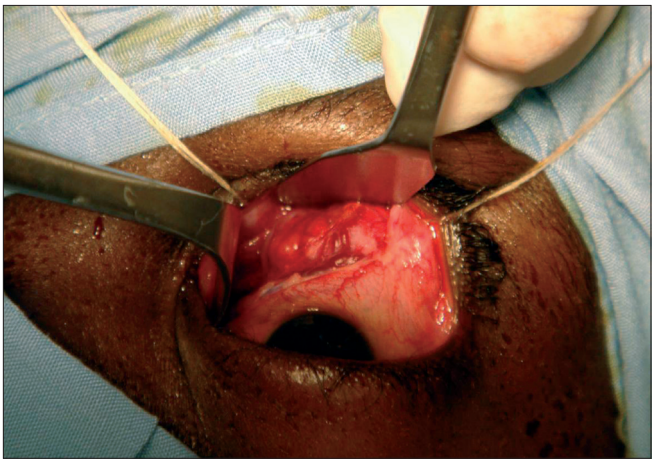


**Figure 2: Intraoperative view of tumour, demonstrating massive vascular supply**



**Figure 3: Cosmetic result, 1 month postoperatively**

One month postoperatively, the child was well and both he and his mother were pleased with the cosmetic result [Figure 3]. Corrective strabismus surgery was planned for a future date. The child was referred to the oncology specialists for further advice on management of ASPS.

## DISCUSSION

ASPS of the orbit is an extremely rare malignancy, accounting for only a small proportion of orbital tumours. The clinical presentation of the case was suggestive of an arteriovenous malformation or a vascular tumour such as haemangioma, but at surgery, the lobulated, grey-white tumour was not consistent with this clinical diagnosis. The tumour appeared more like a granular cell tumour, although the massive vascular supply would be unusual with this type of malignancy. Histology showed that the tumour was most likely to be ASPS, despite the lack of classical alveolar morphology. This case demonstrates that clinical diagnosis of orbital tumours can be problematic even when good imaging is available, and that the histological diagnosis may also be difficult to reach when the classic histological pattern is absent. In such cases, greater reliance has to be placed on immunohistochemistry and the identification of tumour markers.

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