

A Case Report

Primary Atypical Lipomatous Tumor of the Orbit: A Case Report

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Abstract

Purpose: To describe a case of primary atypical orbital lipomatous tumor (ALT).

Case Report: A 35-year-old man presented with a two-month history of left eye proptosis and vertical diplopia. His visual acuity was 20/30 OD and 20/60 OS. External examination showed proptosis and downward displacement of the left eye with mild lid erythema. Extraocular movements were reduced in the left eye, with 10% and 70% motility in upgaze and abduction/adduction, respectively. Imaging showed a mass (22 × 16 × 46 mm) in the superior left orbit that infiltrated the orbital fat and the superior rectus muscle. A biopsy of the mass showed mature adipose tissue intermingled with fibrous zones of hyperchromatic stromal cells with nuclear atypia. Fluorescence *in situ* hybridization analysis demonstrated positive amplification for MDM2/CEP12. The MDM2 to CEP12 ratio was 5:7. A diagnosis of ALT was confirmed. An orbital exenteration was recommended, which the patient declined.

Conclusion: Although rare, the differential for unilateral proptosis with or without diplopia should include orbital liposarcomas including the ALT subtype. Imaging, biopsy, staining, and/or FISH analysis for proto-oncogenes can assist with diagnosis and staging, while the standard treatment is exenteration.

Keywords: Atypical Lipomatous Tumor; Orbital Liposarcoma; Primary Orbital ALT; Primary Orbital Liposarcoma; Well-differentiated Liposarcoma

J Ophthalmic Vis Res 2018; 13 (1): 78-80

INTRODUCTION

Liposarcomas are the most common soft-tissue sarcomas in adults. These malignant tumors are derived from adipocytes and can develop anywhere in the body; however, the orbit is a rare site for their presentation. According to a 2011 study, about 40 cases of orbital liposarcomas had been

reported in the literature.^[1-6] These tumors are categorized histologically into five groups (well-differentiated/atypical lipomatous tumor (ALT), dedifferentiated, pleomorphic, myxoid, round cell). ALTs in turn, are categorized into four morphologic variants: adipocytic, sclerosing, inflammatory, and spindle cell. Herein, we describe a case of an orbital ALT in a 35-year-old man who initially presented with painless left eye proptosis and diplopia.

CASE REPORT

A 35-year-old Hispanic man presented with a painless, progressive proptosis of his left eye that had developed

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How to cite this article: Dworak DP, Patel SA, Chennuri R, Falco D. Primary atypical lipomatous tumor of the orbit: A case report. *J Ophthalmic Vis Res* 2018;13:78-80.

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Received: 16-09-2015 Accepted: 15-06-2016

Access this article online

Quick Response Code:



Website:
www.jovr.org

DOI:
10.4103/jovr.jovr_208_15

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two months earlier. He complained of a vertical diplopia and a decrease in vision. He denied trauma or ocular diseases. He had no significant past medical or family history and was not taking any medications. He was a construction worker, and denied tobacco use, consuming alcohol, or illicit drug use.

On examination, his best corrected visual acuity was 20/30 OD and 20/60 OS. External examination showed significant proptosis and downward displacement of the left eye [Figure 1]. Mild lid erythema was present with no periorbital edema. There was no tenderness or a palpable mass. Hertel exophthalmometry revealed a 9-mm proptosis on the left side. Pupils were round and similar in size with a trace relative afferent pupillary defect of the left eye. Extraocular motility was complete in the right eye but diminished in the left eye, with 10% and 70% motility in upgaze and abduction/adduction, respectively. Humphrey visual fields and Ishihara color plates were noted as normal in both eyes. The slit lamp examinations of both eyes were normal. Gonioscopy was normal in both eyes without the presence of blood. The intraocular pressure measured by Goldmann tonometer was 19 mmHg in both eyes. Fundoscopic exams were not remarkable. Both optic nerves had 0.35 cup to disc ratios and were sharp without edema or pallor.

Computed tomography (CT) and magnetic resonance imaging (MRI) scans showed an infiltrating mass in the superior left orbit [Figure 2]. The mass measured 22 × 16 × 46 mm and infiltrated the intraconal and extraconal fat and the superior rectus muscle. Inferior displacement of the superior ophthalmic vein was appreciated. The optic nerves and cavernous sinuses were normal.

A biopsy of the mass was performed and showed mature adipose tissue intermingled with fibrous zones containing hyperchromatic stromal cells with nuclear atypia, many of which were multinucleated [Figure 3]. FISH analysis to assess the presence of *MDM2* gene amplification demonstrated positive amplification. The *MDM2* to *CEP12* ratio was 5:7. A diagnosis of ALT was confirmed.

The patient was sent to follow-up with oncology, which recommended an exenteration. However, the patient has denied exenteration because of personal reasons and is currently being observed.

DISCUSSION

Liposarcomas represent a diverse group of tumors arising from mesenchymal cells. Even though ALTs represent the lowest grade lesions in the liposarcoma spectrum, they have the ability to dedifferentiate, and therefore, may metastasize. Mavrogenis et al concluded that only 1 (2%) of the 67 ALTs studied showed dedifferentiation, which did not metastasize.^[7] Additionally, Zhang et al's case in 2011 was only the second reported case



Figure 1. The patient showing left eye proptosis with inferior displacement of the globe.

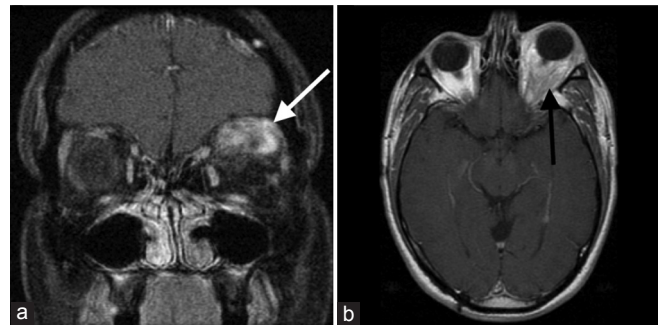


Figure 2. (a) MRI, coronal view of a heterogeneous intermediate signal (white arrow) of the left superior orbital mass, infiltrating the superior rectus muscle. (b) The transverse view of the same heterogeneous mass (black arrow) displacing the globe anteriorly (proptosis).

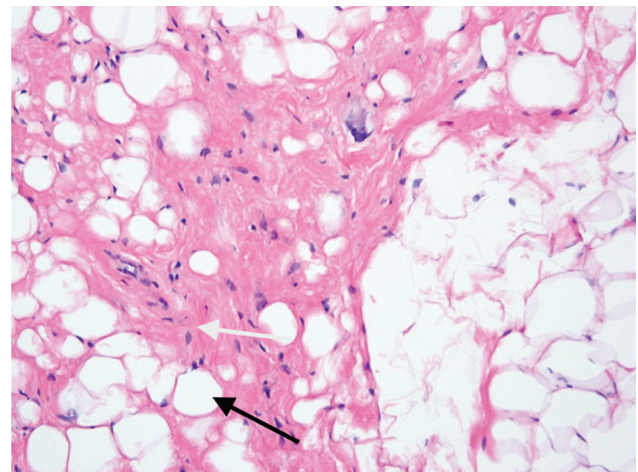


Figure 3. Mature adipocytes (black arrow) intermingled with zones of atypical stromal cells (white arrow) (200×, H & E stain).

of dedifferentiated liposarcoma of the orbit.^[1] In 2003, an atypical liposarcoma was described to consist of contained areas of dedifferentiation, but without metastasis.^[8] However, liposarcomas rarely develop rapidly, as observed in our case. For example, Stiglmayer et al's patient took one year from onset of symptoms for proptosis to become progressive enough to seek further medical care.^[8]

Liposarcomas create symptoms by having a mass effect. Orbital liposarcomas may cause proptosis, swelling, diplopia, vision loss, pain, and optic neuropathy.

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Proptosis is the most common symptom followed by diplopia occurring in over 90% and 40% of the patients, respectively.^[1] Both of these symptoms were present in our patient. The clinical differential diagnosis included vascular proliferations, inflammatory lesions, metastatic lesions, herniated orbital fat, lipomas, and liposarcomas.

No radiographic feature is pathognomonic for the diagnosis of orbital liposarcoma.^[9] Imaging with CT or MRI can show nonspecific findings of a poorly distinct mass with different densities, but some ALTs—specifically the spindle-cell variant—may show a distinct mass on imaging.^[10] Image findings can vary from well-defined homogeneous lesions of soft tissue density to heterogeneous masses containing fat.^[9] An infiltrative enhancing left supraorbital mass was demonstrated on MRI, with internal foci of fat highly suggestive of the reported well differentiated liposarcoma diagnosis.

Although ALT consists of mature adipocytic proliferation that shows significant variation in cell size with nuclear atypia and fibromyxoid stroma, these findings can also be found with other pathologies, such as herniated fat. Other histological differentials include lipoma, lipomatous angiomyolipoma, lipoblastoma, and myxoid liposarcoma. Recently, it was found that ALTs, with the exception of the spindle-cell variant, show amplification of several proto-oncogenes, such as *MDM2*, *CDK4*, and *HMG2*, and immunohistochemical techniques for these genes can be confirmatory. Jakobiec et al concluded that *MDM2* and Ki-67 markers could reliably be used for the diagnosis of ALTs.^[9] *MDM2* amplification (*MDM2/CEP12* >2) by FISH analysis is a helpful test and is associated with both ALTs and dedifferentiated liposarcomas. Furthermore, our biopsy showed no concerning signs of dedifferentiation.

Definitive treatment for liposarcomas of the orbit is exenteration given diffuse infiltration of ocular and adnexal tissue. Our patient refused exenteration for personal reasons. Radiation therapy can be used if surgery is not complete, and chemotherapy is initiated in the rare cases of metastasis. Unfortunately, these patients need to be monitored since recurrence is common. The recurrence rate of primary ALTs is greater than 10% and is even higher after initial recurrence.^[7]

Although extremely rare, the differential for unilateral proptosis with diplopia should include orbital liposarcomas. Imaging, biopsy, staining, and/or FISH analysis for proto-oncogenes can assist with diagnosing

and differentiating between the subtypes. Moreover, the standard treatment for orbital liposarcoma is exenteration.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient has given his consent for his images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial Support and Sponsorship

Nil.

Conflicts of Interest

There are no conflicts of interest.

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