Malaysian Journal of Ophthalmology

© Malaysian Journal of Ophthalmology 2026; 8:47-53
Case report
https://doi.org/10.35119/myjo.v8i2.445

Exploring dedifferentiated orbital liposarcoma: a rare disease

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Abstract

Background: To report a rare case of dedifferentiated liposarcoma.
Case presentation: A 72-year-old man presented with worsening proptosis of the right eye for 2 months, accompanied by severe pain and redness. He had a three-year history of progressive upper eyelid swelling in the right eye. His vision in the right eye deteriorated in 1 year to no light perception. The right eye displayed severe non-axial proptosis, with inferomedial displacement and rupture of the globe caused by a 95 x 90 x 30 mm mass. Computed tomography of the orbit revealed a heterogeneous mass with calcification occupying intra- and extraconal space, with inferomedial displacement of the indented globe. Biopsy of the orbital mass in the right eye showed acute inflammation with squamous metaplasia. The patient underwent a lid-sparing modified exenteration procedure. Histopathological examination revealed dedifferentiated liposarcoma.
Conclusion: Dedifferentiated orbital liposarcoma can be debilitating, particularly when accompanied with displacement of the eyeball.

Keywords: dedifferentiated liposarcoma, proptosis, orbital mass

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Penerokaan dedifferentiated orbital liposarcoma: penyakit jarang berlaku

Abstrak

Latar belakang: Melaporkan satu kes jarang berlaku dedifferentiated liposarcoma.
Keputusan: Seorang lelaki berusia 72 tahun hadir dengan proptosis mata kanan yang semakin teruk selama dua bulan, disertai kesakitan dan kemerahan yang ketara. Pesakit mempunyai sejarah pembengkakan kelopak mata atas kanan yang semakin progresif selama tiga tahun. Penglihatan mata kanan merosot dalam tempoh satu tahun sehingga tiada persepsi cahaya. Mata kanan menunjukkan proptosis bukan aksial yang teruk, dengan bola mata teranjak dan pecah ke arah inferomedial akibat jisim berukuran 95 x 90 x 30 mm. Imbasan tomografi berkomputer orbit menunjukkan jisim heterogen dengan kalsifikasi yang melibatkan ruang intra- dan ekstrakonal, serta menyebabkan anjakan bola mata yang tertekan ke arah inferomedial. Biopsi jisim orbit pada mata kanan menunjukkan keradangan akut dengan metaplasia skuamus. Pesakit kemudiannya menjalani prosedur eksenterasi terubah suai dengan pemeliharaan kelopak mata. Pemeriksaan histopatologi seterusnya mengesahkan diagnosis dedifferentiated liposarcoma.
Kesimpulan: Dedifferentiated liposarcoma boleh menyebabkan morbiditi yang signifikan, terutamanya apabila disertai dengan anjakan bola mata.

Kata kunci: dedifferentiated liposarcoma, jisim orbital, proptosis

Introduction

Liposarcoma is a malignancy involving adipose tissue and are classified into well-differentiated, myxoid, pleomorphic, myxoid pleomorphic, and dedifferentiated.[1,2] Orbital liposarcoma is a rare entity as it predominantly manifests in the muscles of limbs and abdomen.[3] Through our literature search, we only found 6 reported cases of dedifferentiated liposarcoma; this is the first case reported in Malaysia.

Case report

A 72-year-old man presented with worsening proptosis in the right eye (OD) for 2 months, accompanied by severe pain, redness, and eye discharge. He had a 3-year history of progressive upper eyelid swelling in the OD. Vision in the OD deteriorated to no light perception in 1 year. The OD displayed severe non-axial proptosis, with a displaced and ruptured globe inferomedially, as shown in Figure 1. The mass

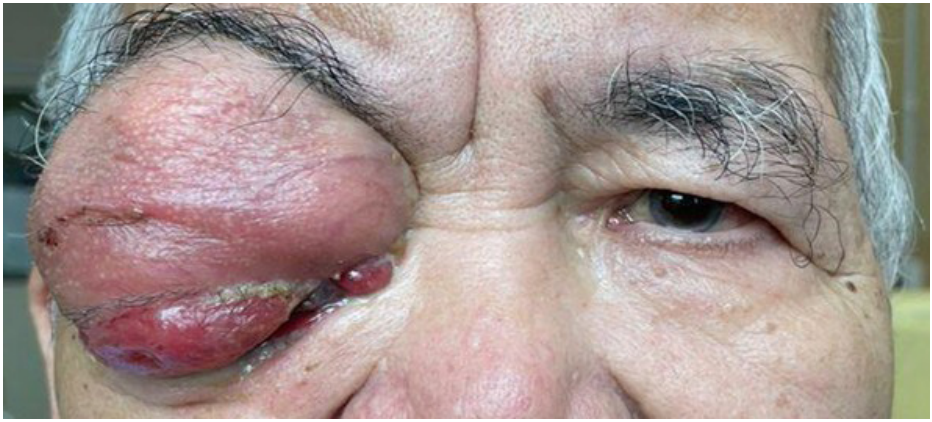

Fig. 1. Severe proptosis, redness and eye discharge.

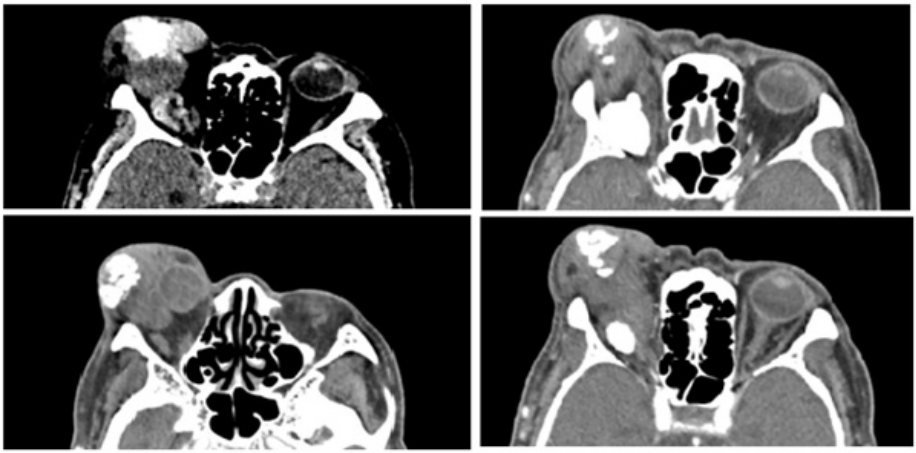

Fig. 2. Computed tomography shows a lobulated mass with dystrophic calcification at the lateral periorbital with extension to the intra- and extraconal spaces. The periorbital mass measured 4.3 x 5.0 x 4.4 cm; the intraorbital extension measured 4.0 x 2.4 x 2.1 cm. The intraorbital component is seen abutting the optic nerve.

measured 95 mm horizontally x 90 mm vertically x 30 mm in width, protruding anteriorly and causing superior displacement of the eyebrow. This mass pushed the right upper tarsus downward, leading to severe exposure and a thick layer of keratinization of the cornea. The surface of the mass appeared lobulated and exhibited a firm consistency. Visual acuity in the left eye was 6/9, with normal anterior and posterior segment ocular examination.

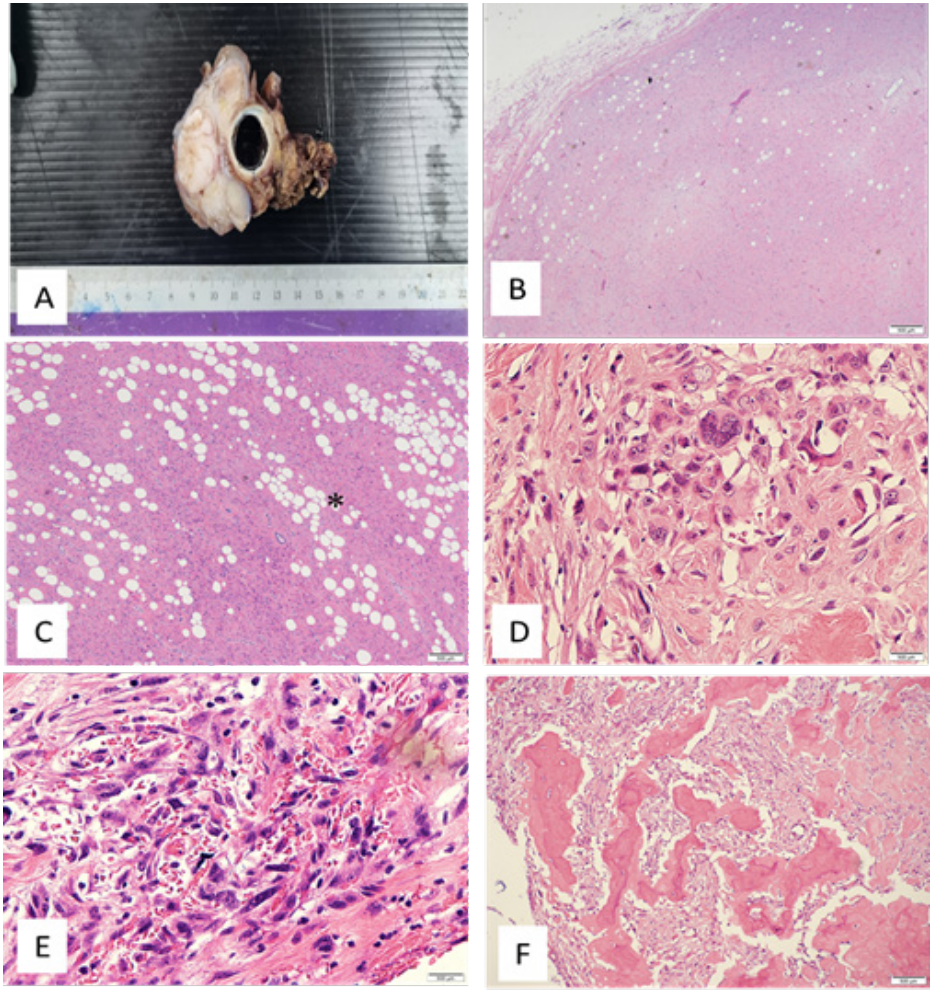

Fig. 3. Dedifferentiated liposarcoma. *(A)* Orbital tumour showing large firm mass with lobulated surface *(B, C)* Low-grade component (H&E x20, H&E x40). *(D)* Tumour cells showing pleomorphic bizzare nuclei; some show vacuolated cytoplasm (H&E x400). *(E)* Dedifferentiated component showing tumour-forming vascular channels forming angiosarcomatous elements (H&E x400) and *(F)* bone formation with lace appearance rimmed by pleomorphic bizarre tumour cells (H&E x200).

Computed tomography of the orbit revealed a heterogenous mass with dystrophic calcification occupying intra- and extraconal space with displacement of the indented globe inferomedially. The apical calcification was in continuation of the roof of the orbit with no intracranial extension, as seen in Figure 2. Incisional biopsy of the orbital mass in the OD showed acute inflammation with squamous metaplasia. The patient was counselled for exenteration in view of malignancy in the blind eye with ruptured globe. Consequently, the patient underwent a lid-sparing modified exenteration surgery under general anaesthesia. A skin incision was made 2 mm from the lid margin, extending to the subcutaneous tissue and carried to the level of the orbital rim. The periosteum was incised at the marcus marginalis. The orbital contents and periosteum were removed manually. The brow was anchored to the periosteum with 5-0 Prolene sutures. Reconstruction of the lateral and inferior socket was performed using 7-0 Vicryl sutures.

Histopathological examination (Fig. 3) revealed dedifferentiated liposarcoma. Histology showed a heterogenous pattern with areas of low- and high-grade transformation consisting of lipomatous area with lipoblasts, prominent osteod formation, pleomorphic spindle cells with bizarre nuclei and multinucleation, and tumour forming vascular-like channels with areas of destructive invasion and myxoid areas.

Immunohistochemical studies showed strong positivity for MDM2, p16, and S100, and patchy immunopositivity for CD34. Fluorescence in situ hybridization (FISH) analysis for MDM2 and CDK 4 gene amplification was positive, CDK4/CEP12 ratio > 2.0. Following surgery, the patient was referred to oncology for further management. The patient subsequently defaulted after the 1-month postoperative follow-up.

Discussion

Liposarcoma is a one of the most commonly diagnosed soft tissue sarcomas, comprising up to 12.8% of all soft tissue malignancies.[4] Liposarcoma in the head and neck region constitute a minority, representing less than 5% of all liposarcomas.[5] Liposarcomas occurring in the orbit are exceptionally uncommon, with approximately 40 documented cases in English literature, primarily in the form of case reports or small series.[5] In cases of orbital liposarcoma, presentation typically mirrors that of other slowly growing tumours in the orbit. Common signs include proptosis, eye displacement—with and without diplopia—as well as optic nerve compression resulting in pain and vision impairment, particularly if the tumour resides at the orbital apex.[6]

Based on Enzinger and Weiss, it has been categorized into five subtypes: well-differentiated, myxoid, round-cell, dedifferentiated, and pleomorphic.[7] Myxoid and well-differentiated subtypes are more common, as indicated by previous studies.[8] Well-differentiated liposarcomas typically exhibit increased mature adipocytes and

various lipoblasts in their pathology. Conversely, dedifferentiated liposarcomas present distinct histological features, characterised by fewer mature adipocytes and a prevalence of highly dysmorphic cells.[3]

Gene amplication of MDM2 and CDK4 proteins has been observed in well- differentiated and dedifferentiated liposarcomas.[9] According to Aleixo *et al.*, immunohistochemistry can be utilized to detect the overexpression of MDM2/CDK4 proteins, aiding in the diagnosis of well-differentiated liposarcomas and dedifferentiated liposarcomas. Similarly in this case, the exenterated eye was sent for FISH analysis for MDM2 & CDK 4 gene amplification and was found to be positive for both.

Diagnosing orbital liposarcoma clinically can be challenging due to the absence of specific diagnostic symptoms. When a tumour is suspected, imaging such orbital computerised tomography and magnetic resonance imaging are crucial for detecting any invasion into neighbouring structures. Histopathology remains the gold standard for diagnosis. Complete surgical resection is recommended once a confirmed diagnosis is obtained.[1] The efficacy of radiotherapy and chemotherapy in managing primary orbital liposarcoma remains uncertain, although radiotherapy has been reported for dedifferentiated liposarcoma.[3] Especially in this case, where we were unable to get clear margins, radiotherapy could have been considered an adjunct.

Conclusion

Dedifferentiated liposarcoma can be debilitating, particularly when accompanied with displacement of the eyeball. In such cases, the importance of a swift and precise management strategy plays a critical role in addressing the condition effectively.

Declarations

Informed consent for publication
The patient provided written informed consent for the inclusion of the clinical data and images presented in this report.

Competing interests
None to declare.

Funding
None to declare.

Acknowledgement
None to declare.

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