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Case Report

Primary Malignant Melanoma of the Lacrimal Sac: A Case Report

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A B S T R A C T

Background: Primary malignant melanoma of the lacrimal sac is a sporadic and aggressive tumor, accounting for less than 1% of all lacrimal sac neoplasms. Because its symptoms-such as persistent epiphora, medial canthal swelling or bloody discharge-closely resemble chronic dacryocystitis, diagnosis is often delayed until the disease has reached an advanced stage. Radiological imaging, particularly MRI and CT, plays a pivotal role in early detection, while definitive diagnosis relies on histopathology and immunohistochemistry. Recent advances in immunotherapy have expanded therapeutic options for mucosal melanomas in anatomically complex sites such as the lacrimal drainage system.

Case presentation: A 60-year-old man presented with progressive tearing and swelling at the left medial canthus, accompanied by intermittent blood-stained discharge for three months. Physical examination revealed a firm, non-mobile mass over the lacrimal sac without cutaneous ulceration. CT and MRI demonstrated a well-defined lesion within the left nasolacrimal canal, showing T1 hyperintensity and T2 hypointensity suggestive of a pigmented neoplasm. Histopathological examination confirmed malignant melanoma with immunopositivity for HMB-45, Melan-A and S-100. PET-CT revealed distant metastases at diagnosis. The patient received systemic immunotherapy with pembrolizumab (200 mg every three weeks), achieving partial remission before progression at 18 months. He was subsequently placed on palliative care.

Discussion: This case underscores the diagnostic challenges of lacrimal sac melanoma, given its rarity and nonspecific presentation. Multimodal imaging and early biopsy are crucial to distinguish it from chronic inflammatory disease. Immunotherapy, particularly PD-1 blockade, represents a promising alternative to extensive surgery in metastatic or unresectable cases.

Conclusion: Prompt recognition and multidisciplinary management are essential for improving outcomes in this aggressive malignancy. Lifelong surveillance remains mandatory due to the high risk of recurrence and metastasis.

Keywords: Lacrimal sac, Malignant melanoma; Mucosal melanoma; Immunotherapy; Pembrolizumab; Case report

Introduction

Primary malignant melanoma originating from the lacrimal sac is exceedingly rare, with only a few dozen cases described in the medical literature[1,2]. Because its initial manifestations-such as persistent epiphora or medial canthal swelling-are nonspecific and often resemble chronic dacryocystitis, diagnosis is frequently delayed[2,3]. Advanced imaging modalities, including CT and MRI, are invaluable for evaluating the lesion's extent and guiding management; typical features include T1 hyperintensity, T2 hypointensity and variable post-contrast enhancement, although signal characteristics may vary[4,5]. The definitive diagnosis depends on histopathological analysis supported by immunohistochemical markers such as HMB-45, Melan-A and S-100[1,4]. Given its aggressive nature and high recurrence potential, early identification and multidisciplinary treatment are critical[2].

Case Presentation

A 60-year-old male presented to the ENT Department with a three-month history of progressive tearing and swelling in the left medial canthal region, accompanied by nasal obstruction. The patient reported intermittent discomfort and occasional blood-stained discharge from the left punctum. He had no history of trauma, previous sinonasal surgery or cutaneous melanoma.

On clinical examination, a firm, non-mobile mass measuring approximately 20 × 15 mm was palpable over the left lacrimal sac, causing mild lower eyelid displacement. The overlying skin appeared slightly discoloured but intact. Gentle pressure on the sac produced no discharge. Ocular motility and visual acuity were preserved **(Figure 1)**.

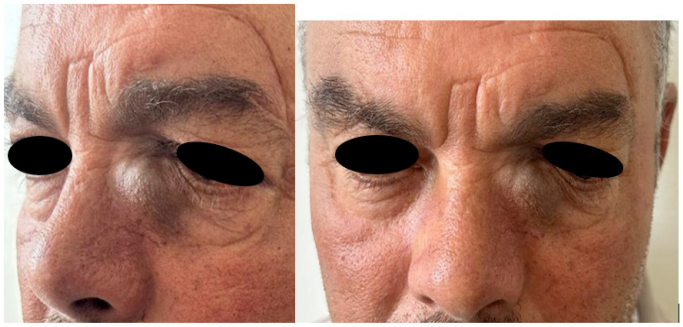

Figure 1: Nasal endoscopy revealed a pigmented mass in the region of the left middle meatus. CT dacryocystography demonstrated a well-circumscribed soft-tissue lesion occupying the left nasolacrimal canal and extending into the inferior meatus, measuring around 21 × 18 × 25 mm.

(Figure 2) the lesion appeared spontaneously hyperdense, with smooth bony remodelling but no destruction.

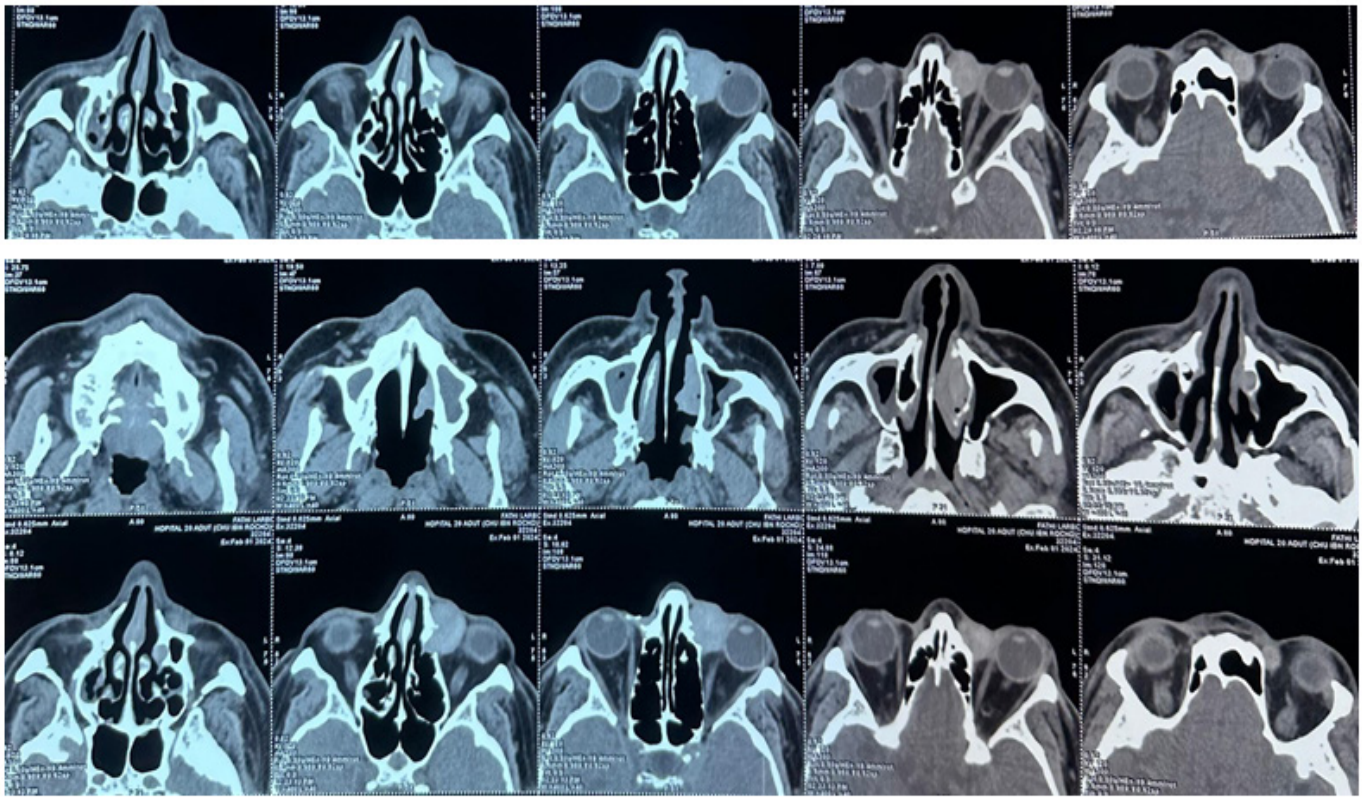

Figure 2: Lesion occupying the left nasolacrimal canal and extending to the inferior meatus.

MRI revealed an ovoid lesion at the left medial canthus that was hyperintense on T1-weighted images, hypointense on T2 and showed heterogeneous enhancement after contrast administration **(Figure 3)**. These features favoured a neoplastic process over inflammatory dacryocystitis[3-5].

Histopathological examination of a biopsy specimen revealed sheets of epithelioid and spindle cells containing melanin pigment. Immunohistochemistry confirmed melanocytic differentiation, positive for HMB-45, Melan-A and S-100[1,4].

Staging PET-CT demonstrated distant metastases. The patient received immunotherapy with pembrolizumab. Follow-up CT revealed new metastatic lesions **(Figure 4)** and the patient was transitioned to palliative care approximately 18 months after presentation. He was subsequently lost to follow-up.

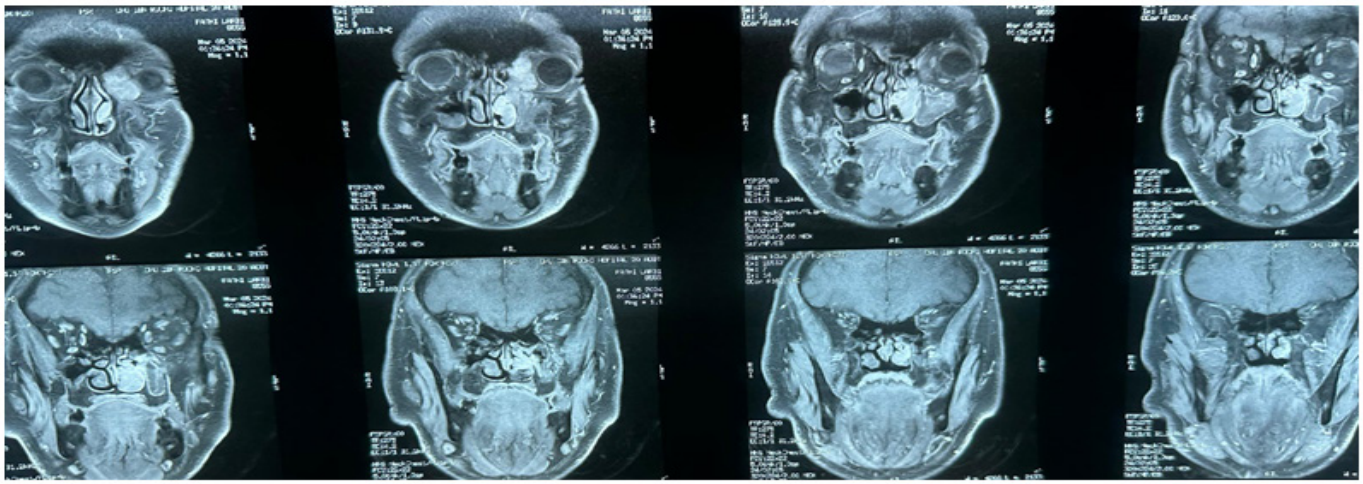

Figure 3: Lesion at the left medial canthus that was hyperintense on T1-weighted images.

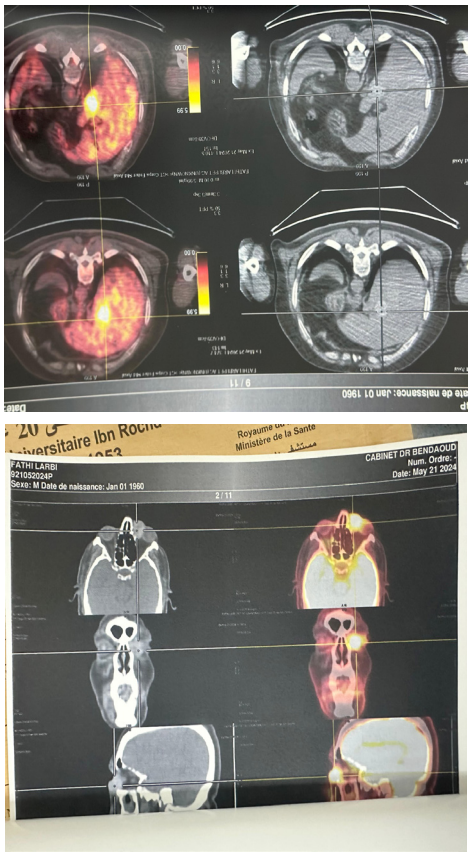

Figure 4: Metastatic lesions.

Discussion

Primary malignant melanoma of the lacrimal sac accounts for less than 1% of tumours in this area and poses a significant diagnostic challenge[2]. It generally affects individuals in their sixties or seventies, with a slight female predominance[1,2]. Due to the similarity of symptoms to benign conditions, numerous patients receive inappropriate treatment for suspected chronic dacryocystitis prior to the accurate diagnosis being made[2,3].

The histogenesis of melanoma in the lacrimal sac is still not fully understood. While melanocytes are typically absent from the lacrimal drainage system, their presence may be attributed to abnormal migration of neural crest-derived cells during embryonic development or secondary migration from the conjunctiva[6,7]. This phenomenon accounts for the emergence of primary melanocytic neoplasms in this otherwise atypical location.

Radiological imaging is crucial in distinguishing melanoma from other lesions. CT scans frequently display a well-defined soft-tissue mass within the lacrimal sac or nasolacrimal duct, usually accompanied by smooth bony expansion rather than erosion[3,5,7]. MRI results can vary based on melanin levels and the presence of haemorrhage, but lesions often exhibit T1 hyperintensity and T2 hypo intensity[4,5]. Although these characteristics are not definitive, they may indicate a pigmented neoplasm and lead to an early biopsy.

Histopathological examination is vital for diagnosis confirmation. The tumour is characterized by nests or sheets of epithelioid or spindle-shaped cells that contain coarse melanin granules. Immunohistochemical positivity for HMB-45, Melan-A and S-100 reinforces the diagnosis and aids in ruling out poorly differentiated carcinomas or sarcomas[1,4].

Due to the rarity of cases, there is no established treatment protocol. Traditionally, wide surgical excision-often involving dacryocystectomy and partial medial maxillectomy-has been the main treatment strategy, with adjuvant radiotherapy for close or positive margins[2,3,7,8]. Nevertheless, recurrence rates surpass 50% within two years and the prognosis remains uncertain[2]. Recent advancements in treatment options are being explored[9,10].

Conclusions

Primary malignant melanoma of the lacrimal sac is an uncommon and highly aggressive neoplasm that often presents with nonspecific symptoms, leading to delayed diagnosis. Comprehensive imaging, prompt histopathologic evaluation and coordinated multidisciplinary care are vital for optimizing outcomes. Although surgical excision remains the traditional mainstay of therapy, immunotherapy represents a promising organ-preserving alternative, particularly in unresectable or metastatic cases. The favourable outcome observed in this case supports the expanding role of immune checkpoint inhibitors in managing rare mucosal melanomas. Lifelong follow-up remains crucial to monitor for recurrence or distant metastasis.

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