Malignant melanoma of the rectum presenting as orbital metastasis

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An orbital mass being the presenting sign of disseminated systemic metastasis is a rare clinical picture. Here, the authors describe the case of a 52-year old Asian-Indian female who presented with unilateral proptosis and motility restriction. Imaging showed an irregular orbital mass infiltrating the right lateral rectus and with a significant intraconal component. Incisional biopsy helped to diagnose a malignant melanoma and exhaustive systemic imaging showed that the primary was found to be arising from the rectum. This represents the first reported case of malignant melanoma of the rectum metastasizing to the orbit and presenting with proptosis and reduced vision.

Key words: Anorectal melanoma, orbital melanoma, proptosis, vision loss

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Metastasis to the orbit is uncommon and accounts for only 3–7% of all orbital tumors.[1] At the time of diagnosis of orbital metastases, over 40% of patients do not have a known primary tumor, making metastatic disease an important differential diagnosis while evaluating patients with sudden onset proptosis, especially in the elderly.[2] In this communication, the authors highlight the case of a 52-year old Asian-Indian female who presented with orbitopathy and was found to harbor a metastatic melanoma, with the primary arising from the rectum. Both the secondary and the primary locations are extremely rare for melanomas. To the best of our knowledge, this is the first report in the literature of an anorectal melanoma metastasizing to the orbit.

Case Report

A 52-year-old female presented with proptosis of the right eye, which was progressively increasing over two months [Fig. 1a and b]. Best-corrected visual acuity was 20/120 OD and 20/20 OS and no afferent pupillary defect was seen. Ocular motility in the right eye was limited in all gazes. The right eye also showed lagophthalmos of 2 mm without any exposure keratopathy and fundus examination showed choroidal folds. Anterior and posterior segment examination in the left eye was

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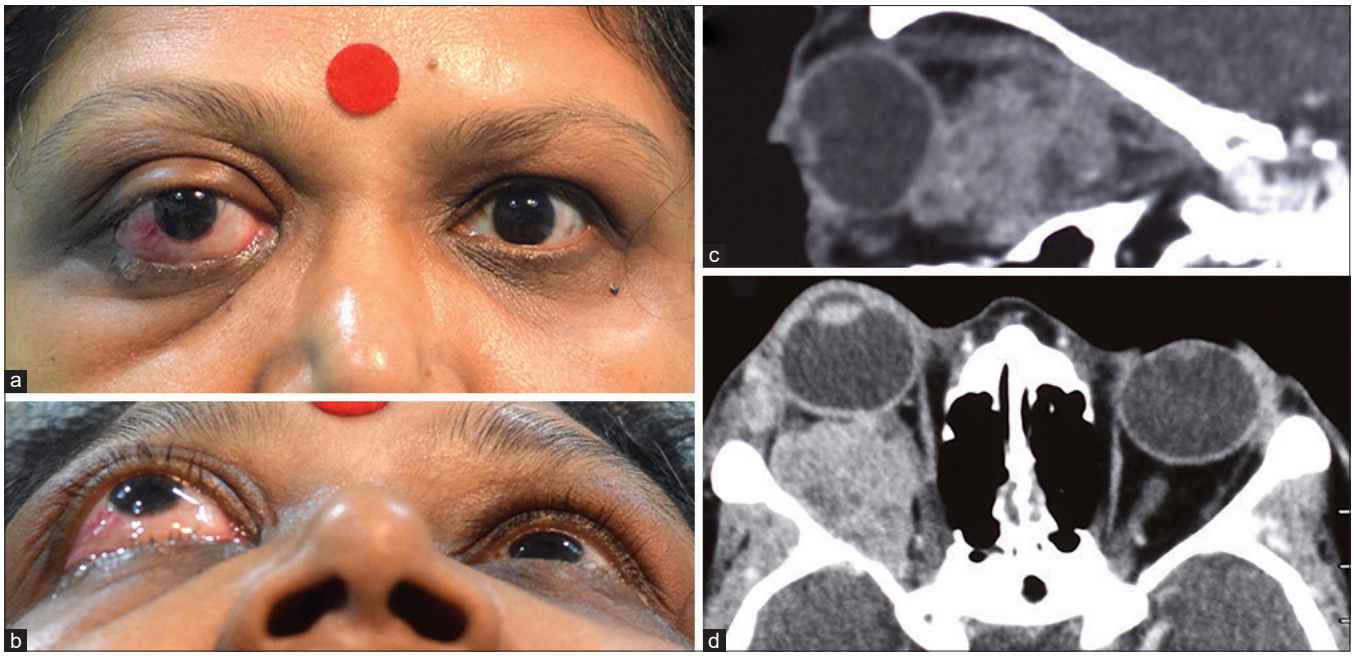

Figure 1: A 52-year-old female presenting with gross proptosis of the right eye (a), more evident in the worm's eye view (b). CT scans show the intraconal extent of the mass (c). Axial slice showing the diffuse irregular iso-to-hyper dense mass infiltrating the lateral rectus and displacing the optic nerve medially (d)

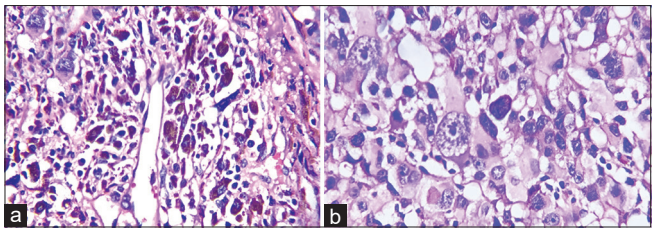

Figure 2: Histopathological examination showed a high grade, pigment-laden, malignant tumor cells with epithelioid morphology (a) (Hematoxylin and Eosin 40×). Bleached sections show prominent nucleoli with frequent mitotic figures within the cells. They also have abundant eosinophilic to clear cytoplasm with residual intracytoplasmic brown pigment (b) (Hematoxylin and Eosin 40×)

unremarkable with intraocular pressures in both eyes being normal. No regional lymphadenopathy was noted.

Computed tomography (CT) scans of the orbit showed an irregular mass along the lateral wall of the right orbit extending into the intraconal and extraconal spaces; displacing the optic nerve medially. Posteriorly, the mass extended up to the orbital apex [Fig. 1c and d]. The mass appeared hyperdense with a variegated appearance and focal contrast enhancement. The differential diagnoses considered were non-specific orbital inflammation, lymphoproliferative mass, and orbital metastasis. An incisional biopsy was performed and histopathological examination showed a high grade, pigment-laden, malignant tumor with epithelioid morphology [Fig. 2a]. The tumor cells showed prominent nucleoli with abundant eosinophilic to clear cytoplasm with intracytoplasmic brown pigment [Fig. 2b]. The cells stained positive for S-100 and had a high Ki-67 index of 18–20; suggestive of a high-grade malignant melanoma. Since the intraocular examination was normal, the working diagnosis, now was a primary orbital melanoma.

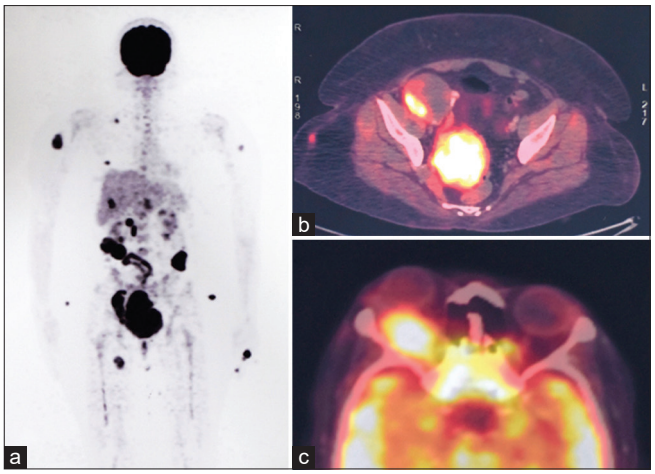

Figure 3: Positron emission tomography scans show a heterogeneously enhancing soft tissue lesion in the right orbit (a). Multiple metabolically active lesions demonstrating high FDG uptake are seen in the right supraclavicular fossa, both arms, right breast, liver, and scattered within the abdomen and pelvis. The largest metabolically active lesion is seen in the rectum (b). The residual orbital mass is also seen as a large metabolically active lesion (c)

A positron emission tomography (PET) CT scan showed multiple metabolically active lesions in the abdomen, right orbit, right supraclavicular fossa, and right inguinal lymph node [Fig. 3]. The largest lesion was in the right pelvis arising in the recto-sigmoid junction and extending into the anal canal, measuring approximately 8.8 cm × 7.0 cm. Multiple clinical, laboratory, and imaging examinations showed no other suspicious lesions in the skin, eye, or leptomeninges. A colonoscopy and subsequent biopsy confirmed the diagnosis of malignant melanoma arising in the rectum. A detailed

history was elicited and the patient recounted having episodes of vague lower abdominal pain, altered defecation patterns, and at least two episodes of rectal bleeding—six months prior to the onset of proptosis; suggesting that the rectal mass was the primary tumor. BRAF V600E mutation analysis was performed and primary and the metastatic tumors tested negative. The patient was offered palliative chemotherapy and succumbed to the disease, three months after the initial diagnosis.

Discussion

Orbital lesions most commonly present with ocular motility restriction, proptosis, ptosis, vision loss, palpable mass.[3] Carcinomas account for 91% of orbital metastatic disease, with breast, lung, and prostate being the most common primary sites.[4] Melanomas, however, metastasize to the orbit as a late event.[5] There have been many reports that have documented melanomas metastatic to the orbit: however, no such case with a primary lesion in the rectum has been documented.[5-8]

Melanoma in the orbit can either be primary or secondary. Primary orbital melanoma (POM) is extremely rare, with only about 50 cases reported to date. It is postulated that POMs arise from melanocytic cells of the leptomeninges or ciliary nerves, or from ectopic intraorbital nests of melanocytes. They may also occur de novo, but usually in association with pigmentary changes within periocular tissues; like nevus of Ota, blue cellular nevus, or oculodermal melanosis.[9] Secondary orbital melanoma can be seen in cases of extrascleral extension of an intraocular melanoma; as orbital extension of conjunctival and eyelid melanoma; as local recurrence after surgical treatment of ocular melanomas; and finally as orbital metastases from a distant cutaneous melanoma.[7] Metastases from nonocular and noncutaneous locations are very uncommon with very few reported cases. Furthermore, it is necessary to distinguish POM from metastasis since the treatment for POM is usually orbital exenteration, whereas it only works as a palliative measure for orbital metastasis.[10]

Malignant melanoma of the rectum, itself, is an extremely rare malignancy that typically presents in the fifth or sixth decade of life with nonspecific complaints such as rectal bleeding or anal pain; and accounts for less than 1% of all anorectal malignancies.[11] In the present case, the dilemma was to decide if it was a primary orbital melanoma with disseminated metastases or a primary anorectal melanoma with metastasis to the orbit among other sites. Charting of the timeline of the patient's symptoms indicated that gastro-intestinal disturbances preceded the onset of proptosis, diplopia, and reduced vision suggesting that the anorectal mass was the primary tumor.

Conclusion

A large proportion of orbital metastases are diagnosed where the patient does not have a previously diagnosed malignancy, highlighting the need for exhaustive clinical examination of all systems, and meticulous history taking. Upon diagnosis of a melanocytic orbital mass, a thorough systemic screen in the form of PET-CT can help in localizing the primary site. Rarely, an orbital mass may be the presenting sign of a rectal melanoma with disseminated metastases.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their 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.

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Conflicts of interest

There are no conflicts of interest.

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