Clinical Case Report

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Case report: an intriguing sign in a patient with an inferior rectus muscle granular cell tumor

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Abstract

Rationale: Occurrence of granulosa cell tumors (GCTs) in the eye are rare and may be diagnosed by imaging examination and immune-histochemical studies. Two common signs of a rectus muscle tumor are the proptosis of the eye ball and the complaint of bi-ocular diploma.

Patient concerns: A 45-year-old man visited our ophthalmology department with an about a 3-year history of vertical diplopia. His visual acuity when looking forward was normal but was severely low when gazing upward.

Diagnosis: Histopathological analysis demonstrated that the encapsulated tumor contained large nested or cord-like cells with small nuclei and abundant eosinophilic cytoplasmic particles. Immunohistochemistry showed that tumor cells strongly expressed CD68, S100 and vimentin, were weakly positive for Ki67, and negative for CK. The tumor was diagnosed as a GCT.

Interventions: The tumor was surgically removed via a transconjunctival approach along inferior rectus muscle.

Outcomes: The severe loss of acuity when gazing upward was ameliorated after surgery, but global mobility did not improve. Long-term follow-up was still needed.

Lessons: Ophthalmologists should be aware that when a patient's visual acuity is normal when looking forward but severely low when gazing upward, it may be a sign of a GCT of the inferior rectus muscle.

Abbreviations: CT = computed tomography, EOMs = extraocular muscles, GCTs = granular cell tumors, MRI = magnetic resonance images.

Keywords: EOMs, GCTs

1. Introduction

Orbital rectus muscle tumors, such as orbital rhabdomyosarcoma, intermuscular hemangiomas, and primary orbital lymphoma, are relatively rare. Neoplastic involvement of extraocular muscles may be the result of a primary muscle tumor, local infiltration from an orbital tumor or metastasis. Metastases to the extraocular muscles are uncommon, and are most frequently associated with breast cancer and melanoma.[1] It was believed that granular cell tumors (GCTs) originate from rhabdomyosarcoma cells, but it has been recently confirmed that they are derived from Schwann cells and neurogenic tumor cells.[2,3] Head, neck, underarms, breast, and waist are the most common sites for GCTs. Extraocular muscles were involved in 67.5% of the GCTs, with the most common infiltration, 40.7%, of the inferior rectus muscle.[4]

Rectus muscle tumors can directly restrict globe mobility, and the patients often come to the ophthalmologist with the complaint of biocular diploma. Another sign for a rectus muscle tumor is proptosis of the eye ball due to the mass effect of the tumor.

In the current case, we report an uncommon sign in a patient clinically diagnosed with a GCT of the inferior rectus muscle. His visual activity when looking forward was normal but was severely low when gazing upward. This is the 1st report for such a sign that ophthalmologists should be aware of.

2. Case

A 45-year-old man visited our ophthalmology department with an about a 3-year history of vertical diplopia. Intriguingly, his best corrected visual acuity was 20/20 in both eyes when looking forward that decreased to hand motion /10 CM when gazing upward. The patient denied any orbital trauma or other ocular diseases, and he had no significant past medical or family history.

External examination showed slight proptosis and upward displacement of the right eye (Fig. 1). There was no palpable mass in periorbital area. Hertel exophthalmometry revealed a 3 mm proptosis on the right side. Globe mobility was complete in the left eye, but infraduction and supraduction was comprimised in

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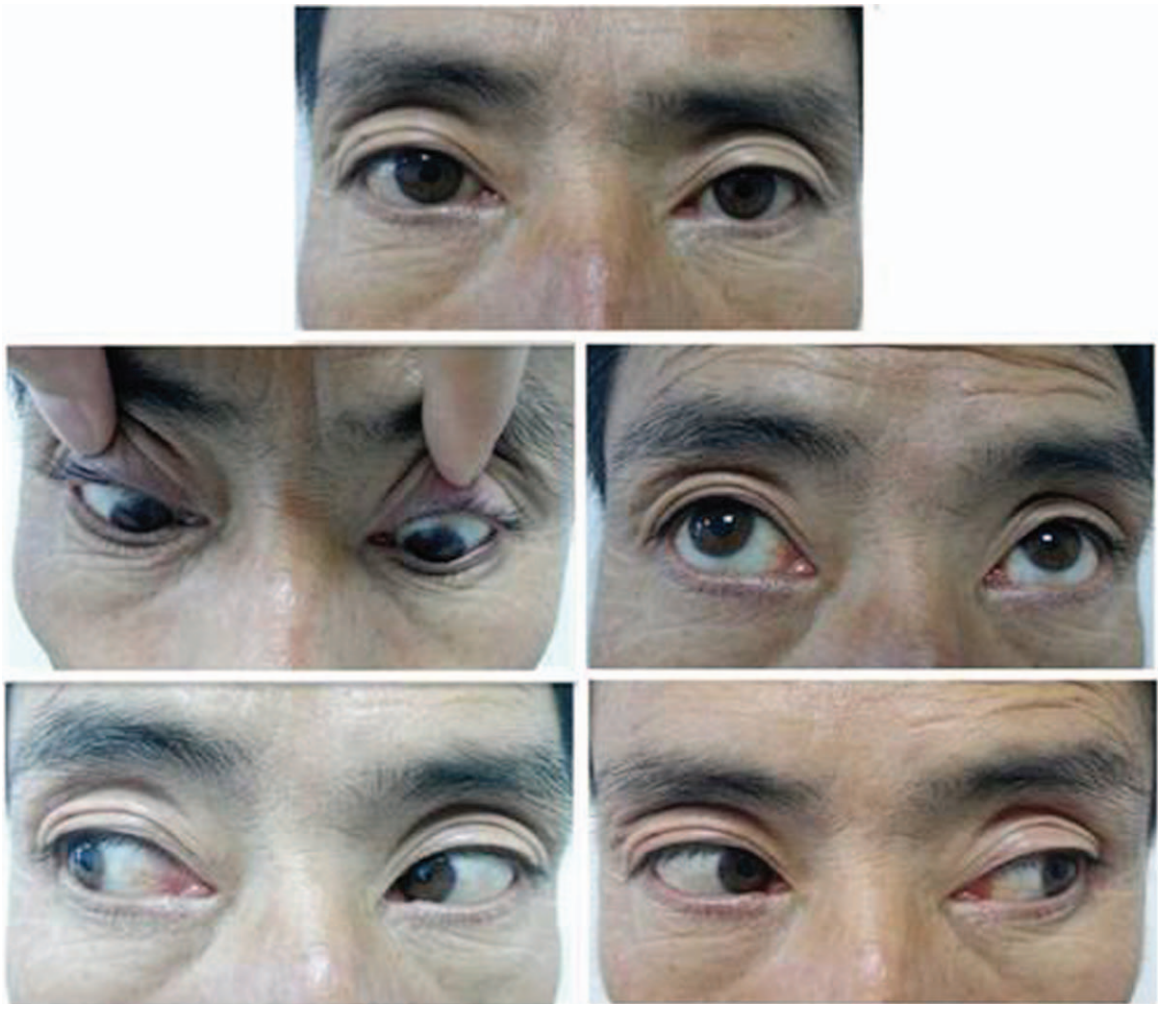

Figure 1. Photographs of patient presenting with a slight proptosis and upward displacement of the right eye.

the right eye. Slit lamp, fundoscopic, Humphrey visual fields and visual evoked potential examinations of both eyes were unremarkable. The intraocular pressure measured by Goldmann tonometer was 15 mm Hg in both eyes.

Orbital computed tomography (CT) scanning showed enlargement of the right inferior rectus muscle infiltrated by a muscle-density tumor which was about $11 \times 17 \times 7\,mm^3$ that displaced the globe upward. An axial magnetic resonance images (MRI) view of the orbit revealed a nodule-like tumor derived from the inferior rectus muscle, which was slightly hypo-intense to the extraocular muscles on T1W with a very low central intensity on T2W and a higher intensity along the periphery. Sagittal MRI images showed that the tumor was very close to the optic nerve with almost no gap in between. Enhanced MRI after intravenous contrast administration showed that the tumor was significantly enhanced in the periphery but only slight enhanced in the center (Fig. 2).

The tumor was surgically removed via a transconjunctival approach along inferior rectus muscle. Intraoperatively, the tumor was completely separated from muscle tissue since it was pseudoencapsular. The gross specimen exhibited a firm fibrotic nodule-like and pale-colored mass (Fig. 3). There was no significant improvement in globe mobility after surgery; however, a longer term follow-up is required.

Subsequent histopathological analysis demonstrated that the encapsulated tumor contained large nested or cord-like cells with small nuclei and abundant eosinophilic cytoplasmic particles. Immunohistochemistry showed that tumor cells strongly expressed CD68, S100 and vimentin, were weakly positive for Ki67, and negative for CK (Fig. 4) and was diagnosed as a GCT.

3. Discussion

The most common complaints of patients with orbital GCT are diplopia, proptosis, and mobility restriction. The GCTs appear as diffuse or oval masses isodense relative to brain tissue in computed tomography (CT) scans. The tumors are isointense in T1-weighted MRI and isointense or hypointense in T2-weighted MRI relative to the extraocular muscles (EOMs), and can be slightly enhanced with contrast administration. Intriguingly, in the current case, the vision of the patient was sharply lower in his right eye when gazing upward which is very rare. Upon further examination with enhanced MRI sagittal imaging, we found a tumor that was very closed to the optic nerve. We speculate that when the eyeball turned upwards, the enlarged tumor contacted the optic nerve leading to a temporal decrease of vision.

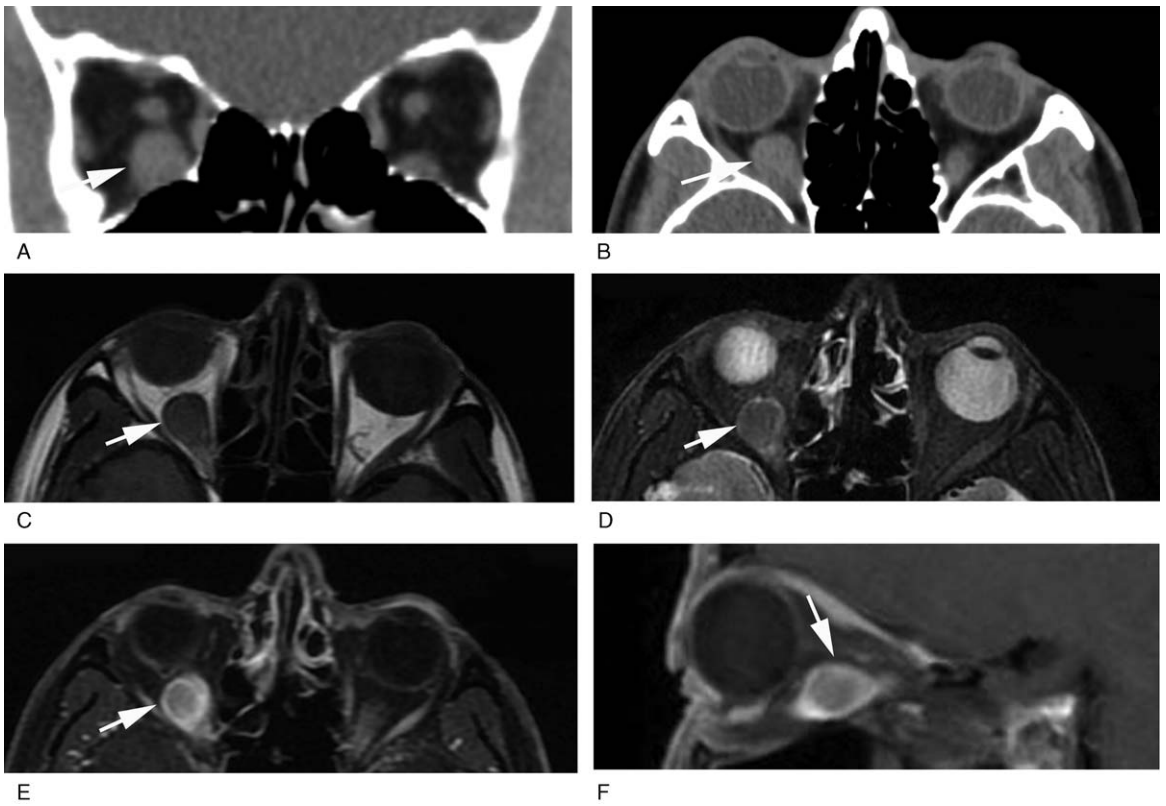

Figure 2. (A, B) Orbital computed tomography(CT) scanning showed enlargement of the right inferior rectus muscle infiltrated by a muscle-density tumor which was about 11 × 17 × 7 mm3. (C, D) An axial magnetic resonance images (MRI) view of the orbit revealed a nodule-like tumor derived from the inferior rectus muscle, which was slightly hypo-intense to the extraocular muscles on T1W with a very low central intensity on T2W and a higher intensity along the periphery. (E, F) Enhanced MRI after intravenous contrast administration showed that the tumor was significantly enhanced in the periphery but only slight enhanced in the center. The tumor was very closed to the optic nerve (arrows) and extended into the orbital apex. The mass was with significantly enhanced peripherally and slightly enhanced in the center.

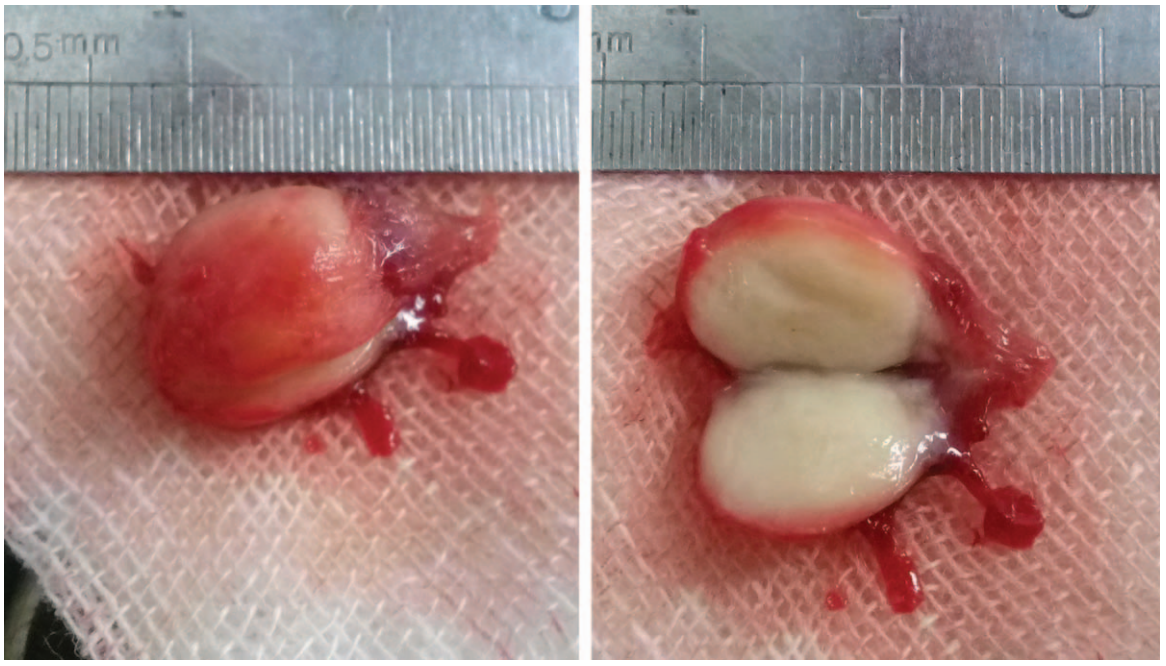

Figure 3. Photographs of the gross specimen exhibiting a firm fibrotic nodule and pale-colored mass.

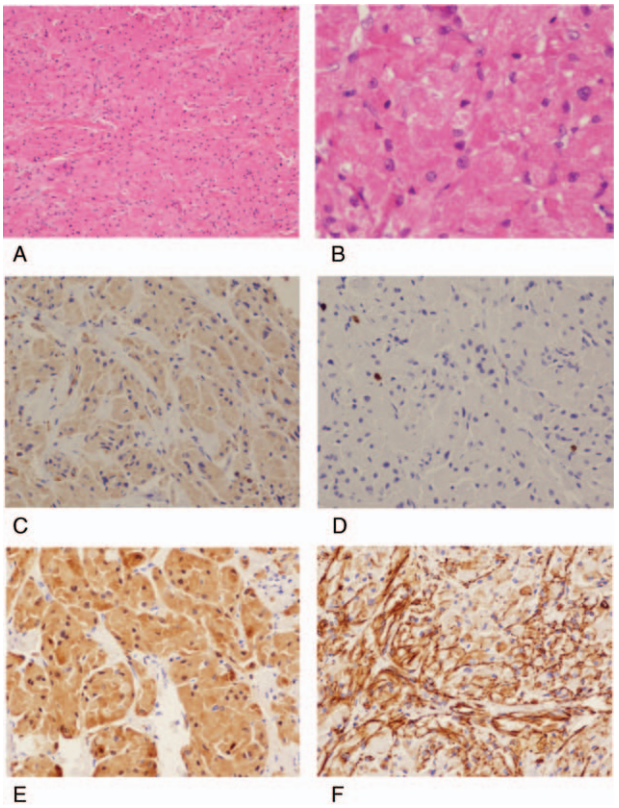

Figure 4. (A, B) Histopathological staining showing that the tumor cells are large, the nuclei are small, and a large number of eosinophilic particles can be seen in the cytoplasm. Immunohistochemistry showing: (C) positive staining for CD68, (D) staining for Ki67 was less 1%, (E) positive staining for S100 (F) positive staining for vimentin and negative staining for CK and SMA.

Immunohistochemical studies of the patient was consistent with previous reports[4] that showed 100%, 90%, and 66.7% of cases were positive for vimentin, S-100, and CD68, respectively. Therefore, the final diagnosis was a GCT.

4. Differential diagnosis of rectus muscle diseases

4.1. Graves disease

Graves disease is the most common type of hyperthyroidism. The clinical manifestations may include exophthalmos and extraocular muscles involvement, but the functional diagnosis is mainly based on the increase of thyroid hormone secretion. Orbital CT scans show a thickening of the muscle abdomen without tendon involvement.[5]

4.2. Rhabdomyosarcoma

Rhabdomyosarcoma is the most common malignancy of soft tissue in children.[6] Since it is highly malignant, the rapid progression of ocular symptoms such as proptosis, eyeball displacement, conjunctival congestion and orbital mass are often major signs. Orbital CT scanning shows enlargement of the abdomen muscle and a local lesion with relative sparing of the tendons.[7] The tumor can be enhanced in MRI images with intravenous contrast administration.

4.3. Hypertrophic myositis

Hypertrophic extraocular myositis is a type of localized nonspecific fibroid pseudo-tumor. The common clinical manifestations of hypertrophic extraocular myositis are acute paroxysmal eye pain, diplopia, ptosis, mild edema of the eyelids, slight protrusion of the eyeball, congestive edema of the bulbar conjunctiva, and limited rotation. Orbital CT scans often show a blurred contour of extraocular muscles (EOMs) and surrounding immunoreactive lesion. The main manifestations are the enlargement of EOMs including the muscle abdomen and tendons. Eyelid ultrasound can also be used for assisted examination, where CDFI shows that the blood flow signal within the lesion is not abundant.[8]

4.4. Intermuscular hemangiomas

The structures of intramuscular hemangiomas in EOMs resemble cavernous hemangiomas without a capsule, and consist of abundant thin-walled sinusoids. Intermuscular hemangiomas distribute between muscle fibers, which results in thickening of the muscles and decreased globe mobility. The MRI examination often shows a hyperintense high edge, and the lesion is with clear, enhanced boundaries after enhancement.[9]

4.5. Lymphoma

The incidence of orbital lymphoma is low, accounting from 6% to 8% of orbital tumors, yet there are some cases involving EOMs. Orbital lymphoma involving EOMs is more common unilaterally and may infiltrate multiple EOMs.[10] Pathologically, lymphomas are composed of undifferentiated cells and lymphoid cells, that lack a fibrous matrix. Orbital lymphomas often infiltrate the eyelids and sclera.

5. Treatment and prognosis

Orbital GCTs are usually benign and most can be completely removed or partially resected with surgical treatment. The chance of postoperative recurrence is low and some spontaneously regress after partial resection.[4,11] However, in patients with preoperative diplopia, diplopia may persiste after surgery. Cases of relapsed and systemic metastases of malignant GCTs are rare.

In conclusion, ophthalmologists should be aware that when a patient's visual acuity is normal when looking forward but severely low when gazing upward, it may be a sign of a GCT of the inferior rectus muscle.

Author contributions

LGY designed the study and drafted the manuscript. LC, PH, and BF collected and interpreted the data. LGY performed the operation. All authors read and approved the final manuscript.
Supervision: Pei He, Bin Fan, Guang-Yu Li.
Writing – original draft: Lin Che.
Writing – review & editing: Lin Che.
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