Degree relatives should be encouraged to undergo detailed systemic evaluation to exclude gastrointestinal and genitourinary malignancy according to published protocols. Because systemic malignancy may occur many years after the onset of sebaceous adenoma, continued long-term surveillance is necessary.

The principal genetic aberrations in Muir-Torre syndrome are mutations in the mismatch repair genes, hMLH1 and hMSH2, leading to microsatellite instability. Microsatellite testing requires the services of a molecular diagnostic laboratory, but tumors that exhibit microsatellite instability can be detected with immunohistochemical staining. The absence of hMLH1 and hMSH2 nuclear expression identifies tumors with mismatch repair deficiency. Application of this technique to cutaneous sebaceous neoplasms has proved to be a reliable screening method, with high predictive value for the diagnosis of DNA mismatch repair-deficient Muir-Torre syndrome. In this study, the sebaceous adenoma did not express MLH1 protein but expressed MLH1 protein; the same pattern was seen in the rectal adenocarcinoma. The overall findings indicate mutations in the hMSH2 gene and, coupled with the clinical findings, are highly suggestive of Muir-Torre syndrome.

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**Desmoplastic Small Round Cell Tumor:**

Desmoplastic small round cell tumor (DSRCT) is an uncommon yet potentially lethal tumor typically affecting young adult males. This extremely aggressive tumor was first described by Gerald and Rosai in 1989, and since then, more than 250 cases have been reported, with recent cytogenetic work revealing a unique translocation involving the Ewing sarcoma breakpoint region (EWSR1) at t(11;22)(p13;q12). In the largest case report to date, 94% of 109 cases were located in the abdominal cavity. Other serosal surfaces may be involved, including the testis, ovary, pleura, and parotid gland. Manifestations in the head are extremely uncommon. Desmoplastic small round cell tumor has been reported once in the ethmoidal sinuses and once in the postero-crinal fossa. The current report describes the first patient, to our knowledge, with DSRCT producing visual disturbances as an initial complaint.

**Report of a Case.** A 32-year-old man was in his usual state of health when he saw his physician and complained of blurry vision. The patient initially attributed his blurry vision to the recent administration of fluoxetine hydrochloride. Fluoxetine administration was discontinued, but the patient continued to experience blurry vision. In addition, the patient developed pain in his left eye and reduced vision with right gaze. He was evaluated by a general ophthalmologist who diagnosed an orbital abnormality in his left eye. Computed tomographic scans and magnetic resonance images of the patient’s orbits revealed a lobular heterogeneous soft tissue mass within the left orbit measuring 2.5 x 2.5 x 1.8 cm (Figure 1), producing proptosis of the left globe, and flattening and decreases...
ing the anteroposterior diameter of that eye. Imaging also revealed displacement of the left optic nerve medially and slightly superiorly, with no extension of the tumor mass through the optic foramen. Laboratory studies revealed no hematologic abnormalities. Imaging results of the chest, abdomen, and pelvis were normal. The patient underwent orbital excisional biopsy, and excised soft pink tissue was submitted for histopathologic examination.

Hematoxylin-eosin sections of tumor tissue revealed well-defined nests and irregular sheets of small round cells infiltrating a desmoplastic stroma. The tumor showed irregular nuclear contours, indistinct cytoplasmic boundaries, and areas of central necrosis (Figure 2A and B). In some areas, rosette-like structures and cords of single cells were occasionally visible. Immunohistochemical staining revealed positive reactivity for cytokeratin and desmin with a perinuclear dot-like pattern (Figure 2C). The tumor also tested positive for neuron-specific enolase and weakly positive for CD99. Immunohistochemical stains tested negative for OC-125, smooth muscle actin, synaptophysin, S100, HMB45, CD3, CD10, and CD20. Cytogenetic analysis using fluorescence in situ hybridization revealed an abnormal signal pattern in 69% of cells, indicating a translocation of the EWS gene (an abnormal signal pattern is characterized by fusion [orange + green = yellow] of the orange and green fluorescence signals within a cell, indicating a break-apart rearrangement of 1 copy of the EWSR1q region) (Figure 2D).

The patient underwent adjuvant radiation therapy for the treatment of his tumor. He received 45 Gy to the orbit and 55.8 Gy to the tumor bed, using intensity-modulated radiation therapy to limit the retinal dose to 45 Gy and the corneal dose.

Figure 1. Coronal T1-weighted magnetic resonance image demonstrating a lobular heterogeneous mass with intraconal localization.

Figure 2. Histopathologic section of the surgical specimen. A, Low-power photomicrograph. Irregular sheets of small, round, infiltrating cells with abundant desmoplastic stroma (hematoxylin-eosin ×25). B, High-power photograph demonstrating malignant cells with irregular nuclear contours and high nuclear-to-cytoplasmic ratio. Mitotic figures and apoptotic cells are also found within the section (hematoxylin-eosin ×100). C, Tumor cells demonstrated strong perinuclear desmin immunoreactivity. D, Cytogenetic analysis using fluorescence in situ hybridization. An abnormal signal pattern (yellow fluorescence) was detected in 69% of cells, indicating a translocation in the EWS gene.
to less than 40 Gy. The patient remains in complete remission with more than 11 months’ follow-up.

Comment. Small round cell tumors manifesting in the orbit should raise the suspicion for a wide variety of tumors. The differential diagnosis includes lymphoblastic lymphoma, embryonal rhabdomyosarcoma, neuroblastoma, primitive neuroectodermal tumor, Ewing sarcoma, melanoma, and ependymoma, as well as DSRCT.

Several features are consistent with the diagnosis of DSRCT in this patient. First, the majority of patients with this tumor are between the ages of 15 and 35 years with a 4:1 male-to-female preponderance. Second, immunohistochemical examination revealed positive cytokeratin and desmin staining in a characteristic perinuclear dot-like pattern, which is pathognomonic for DSRCT. In addition, although the histogenesis of DSRCT remains unknown, positive CD99 and neuron-specific enolase staining have been observed in these tumors, supporting an origin from a progenitor cell with potential for multiphenotypic differentiation. Finally, the fluorescence in situ study demonstrates an EWS gene rearrangement that is characteristic of this tumor. Primitive neuroectodermal tumor is excluded on the basis of strong positive cytokeratin and desmin staining.

The unusual feature of this clinical manifestation is the location of the tumor, which typically appears in the abdomen or involves other serosal surfaces. Only 4 cases of nonserosal DSRCT have been reported. Two cases involved soft tissues and bone. Adsay et al. described a hypophenar mass in the right hand of a 34-year-old man, and Mihok and Cha reported a neck mass in a 16-year-old boy. Two other cases manifested in the head. Tison et al. described an intracranial lesion located in the posterior fossa in a 24-year-old man. Finke et al. reported an ethmoidal sinus lesion in a 21-year-old woman.

To our knowledge, this is the first case of DSRCT in the orbit, causing visual disturbances and eventually pain in the eye with proptosis and loss of vision. Resection followed by intensity-modulated radiation therapy has resulted in disease control.

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Optical Coherence Tomography of Optic Disc Swelling in Acute Primary Angle-Closure Glaucoma

Acute primary angle-closure glaucoma (APACG) is an ophthalmic emergency with poor outcome if initial recognition and treatment are delayed. Posterior segment signs are often difficult to detect owing to corneal edema, anterior chamber inflammation, cataract, and the use of miotics. Optical coherence tomography (OCT) is a noncontact, noninvasive imaging technology capable of obtaining high-resolution tomographic images of posterior segment ocular structures. We report a case of optic disc swelling and branch retinal vein occlusion after APACG with imaging performed with OCT and fluorescein angiography. We discuss the disc appearance on OCT imaging and a finding of a partial thrombus in the venous circulation on fluorescein angiography.

Report of a Case. A 64-year-old Chinese woman had pain, blurring of vision, and redness in the left eye for 5 days. She had no previous episodes of eye pain. Her visual acuity was 20/30 OD and hand motion OS with a left relative afferent pupillary defect and mid-dilated pupil. The left eye had conjunctival injection, corneal edema, and a shallow anterior chamber. The intraocular pressure (IOP) was 14 mm Hg OD and 54 mm Hg OS. Gonioscopy confirmed angle closure. The IOP was lowered with intravenous acetazolamide, topical timolol maleate, pilocarpine hydrochloride, and brimonidine tartrate. Topical steroids were administered. Six hours after initial examination, her IOP was 12 mm Hg.

The left cornea had cleared 12 hours after initial examination to allow a view of the left disc. This was swollen and hyperemic with peripapillary flame hemorrhages. The fundus was otherwise unremarkable. The IOP was 10 mm Hg OS. Bilateral laser peripheral iridotomy was performed. The OCT (OCT 3; Zeiss-Humphrey Systems, Dublin, Calif) of the left disc showed swelling with adjacent thickening of the retina in the papilla-macula region (Figure 1). The right disc showed no swelling.

One week later, the left disc was still swollen and hyperemic, with fluorescein angiography showing no central retinal vein occlusion. There was, however, a partial thrombus of the left superotemporal branch vein (Figure 2). One month later, the left disc was pale and cupped (0.7). The visual acuity was counting fingers OS.