Klippel-Trénaunay Syndrome and Rhabdomyosarcoma in a 3-Year-Old

Klippel-Trénaunay syndrome (KTS) is a congenital vascular anomaly with soft tissue and skeletal hypertrophy. It has been associated with capillary, venous, lymphatic, and soft tissue malformations, but not with malignancies. Orbital rhabdomyosarcoma typically demonstrates a rapid-onset orbital process that can be confused with trauma or benign tumors. This case report underscores that rhabdomyosarcoma can mimic a benign, lymphatic malformation, particularly in the setting of expected vascular lesions. A comprehensive MEDLINE search failed to identify a previous case of concurrent KTS and orbital rhabdomyosarcoma. Current chemotherapeutic regimens are reviewed.

Report of a Case. A 3-year-old boy had an inferonasal conjunctival mass of 10 days' duration and a recent upper respiratory tract infection (URTI), including nasal congestion, rhinorrhea, and epiphora. Examination revealed a right, vascular conjunctival mass (Figure 1) and a grade 1 port-wine stain (diffuse capillary and venular malformation) of the left upper extremity (Figure 2A), with soft tissue hypertrophy of the left thenar eminence (Figure 2B). Computed tomography demonstrated an orbital mass with faint areas of internal septation (Figure 3).

The patient was referred to the multidisciplinary Hemangioma and Vascular Malformation Clinic at the

![Figure 1](http://archopht.jamanetwork.com/index.aspx?cmd=pdfaccess&journal=ophth/9906) Clinical features of the right inferonasal conjunctival mass.

![Figure 2](http://archopht.jamanetwork.com/index.aspx?cmd=pdfaccess&journal=ophth/9906) Clinical features of Klippel-Trénaunay syndrome. A, Reticulated pink mottling of the left chest and arm, consistent with a diffuse capillary malformation. B, Hypertrophied, hypervascular left thenar eminence in this right-handed boy.
Massachusetts General Hospital (Boston). Physical examination was consistent with KTS. Given the sudden onset of symptoms, recent URTI, radiologic interpretation, and the known association of vascular malformations with KTS, a presumptive diagnosis of orbital lymphangioma was made.

After a 1-week trial of prednisone, the mass had doubled in size. Subtotal resection of a gelatinous, poorly delineated mass was performed. Histopathologic examination demonstrated small, round, blue cells with scant cytoplasm most suggestive of rhabdomyosarcoma or metastatic neuroblastoma in this age group. Positive immunoperoxidase stains for muscle actin, vimentin, and desmin suggested either smooth or striated muscle. Stains for myogenin and myo-D1 were also positive, indicating striated muscle. Neurofilament staining was negative for tumors of neuronal origin.2 Despite negative myoglobin staining, a histopathologic diagnosis of embryonal rhabdomyosarcoma was made (Figure 4).

Radiographic staging evaluation revealed no evidence of metastatic disease. The child was treated with a 48-week course of chemotherapy consisting of vincristine and actinomycin D. Local control was established with conformal radiation to the involved portion of the orbit. The patient received a total of 4500 rad (45 Gy) at 180 rad (18 Gy)/fraction per day during 32 elapsed days. Nine months after commencing therapy, there was neither clinical nor radiographic evidence of residual disease. The child was thriving.

Comment. Rhabdomyosarcoma accounts for approximately 4% of all pediatric malignancies.3 Ten percent originate in the orbit,4 representing 4% of all biopsied pediatric orbital masses.5 An extensive description of clinical presentations in ophthalmic cases was recently published by Shields and associates.6 Both orbital rhabdomyosarcoma and orbital lymphatic malformations commonly exhibit proptosis. Furthermore, Fetkenhour and associates7 recently reported a case of orbital rhabdomyosarcoma with radiographic features, including internal cavitation, which strongly suggested a lymphatic malformation. In the patient we described, the presence of KTS provided a unique clinical context for similar diagnostic confusion.

Figure 3. Computed tomographic coronal (A) and axial (B) views demonstrating a low-density intraconal mass with rim enhancement and faint areas of internal septation in the right orbit.

Figure 4. Histopathologic analysis. A, Light microscopy reveals small, blue, round cells with scant cytoplasm, characteristic of embryonal rhabdomyosarcoma (hematoxylin-eosin). B, The deep purple color of the Myo-D1 stain is specific for nuclear proteins expressed in striated muscle. Additional immunoperoxidase stains were positive for vimentin, desmin, myogenin, and muscle actin (not shown) (original magnification ×40).


Secondary Chronic Open-Angle Glaucoma After Intravitreal Triamcinolone Acetonide

Intravitreal injections of triamcinolone acetonide have increasingly been used for treatment of various intraocular neovascular, proliferative, or edematous diseases, such as diffuse diabetic macular edema, proliferative diabetic retinopathy, proliferative vitreoretinopathy, chronic uveitis, and persistent pseudophakic cystoid macular edema. In view of the widening spectrum of therapeutic indications of intravitreal triamcinolone acetonide, we report the clinical course of a patient who repeatedly received intravitreal injections of triamcinolone acetonide 14 months apart, who showed intravitreal triamcinolone acetonide crystals still present 9 months after the second injection, and who developed secondary open-angle glaucoma uncontrollable by topical antiglaucomatous medication.

Report of a Case. A 79-year-old woman sought treatment for progressive exudative age-related macular degeneration with subfoveal occult neovascularization in her left eye. Snellen chart visual acuity decreased from 0.80 to 0.50 OS with the accompanying complaint of marked metamorphopsia. Intraocular pressure measured 16 mm Hg, and the appearance of the optic nerve head was normal. The right eye demonstrated a large subfoveal disciform scar due to exudative macular degeneration covering the whole macular region be-