Bilateral Conjunctival Lymphangiectasia in Klippel-Trénaunay-Weber Syndrome

Klippel-Trénaunay-Weber syndrome (KTWS) is a rare congenital malformation syndrome involving blood and lymphatic vessels as well as disturbed growth of soft tissues and bone. The clinical features can be extremely variable, but most patients exhibit the triad of cutaneous capillary malformation, varicose veins, and hypertrophy of bone and soft tissue. Thromboembolic events occur at an increased rate and may be fatal. Most cases of KTWS are sporadic and no causative gene or etiology has been firmly established. Reported ophthalmological features include conjunctival, retinal, choroidal, and orbital varicosities, secondary glaucoma, iris coloboma and heterochromia, and several neuro-ophthalmic manifestations, including strabismus. To our knowledge, lymphangiectasia of the conjunctiva has not been reported in association with KTWS.

Report of a Case. A 30-year-old woman was referred for a lesion of the inferior conjunctiva of the right eye that had been present and stable for more than 10 years. Her medical history was significant for KTWS, including the following features: multiple congenital cutaneous capillary malformations on virtually all body surfaces (Figure 1A), right leg hypertrophy (Figure 1B), left leg venous angiomata, communicating varices, an incomplete deep venous system, and atrophy of the metatarsal and long bones. Her parents and 2 sisters were not affected.

Ophthalmic examination revealed best-corrected visual acuity of 20/25 OD and 20/70 OS as well as a left dissociated vertical deviation. Intraocular pressure was 15 mm Hg OD and 14 mm Hg OS. An extensive, cystic-appearing, translucent lesion consistent with lymphangiectasia was seen over the inferior 270° of the conjunctiva of the right eye (Figure 1C).
staining of the conjunctiva with the lymphatic marker D2-40 shows positive staining of the dilated channel in the substantia propria (original magnification ×250). The localized form is often seen secondary to conjunctival pathological findings such as pinguecula or scar. The diffuse form is less common and of-
and D). A similar lesion was seen on the inferotemporal conjunctiva of the left eye. Funduscopic examination disclosed Bergmeister papilla and retinal vascular tortuosity bilaterally. The only facial cutaneous capillary malformations were on the lips and periorally. Lymphatic malformations had not been identified elsewhere in the body.

Following incisional biopsy of the lymphangiectasia in the right eye, the entire conjunctival surface flattened, presumably because the lymphatic channels were interconnected. Histopathological examination showed conjunctiva with focal epithelial thinning and numerous endothelium-lined, dilated channels filled with serous material in the substantia propia (Figure 2A). Capillaries were seen intermixed with the dilated lymphatic channels. Immunohistochemical examination revealed positive staining for CD34 in the endothelial cells of the blood vessels and negative staining for CD34 in the endothelial lining of the dilated, blood-free channels, consistent with lymphatic channels. Immunofluorescence with the lymphatic marker D2-40 was positive in the dilated channels (Figure 2B).

Six weeks postoperatively, the lesion again was elevated in all areas except for the site from which the biopsy specimen was taken.

Comment. Lymphangiectasia of the conjunctiva is a localized or diffuse enlargement of the lymphatics that appears as chemosis or a freely movable cyst or series of cysts (“string of pearls”). The localized form is often seen secondary to conjunctival pathological findings such as pinguecula or scar. The diffuse form is less common and often there is no attributable cause, but it has been reported in a patient with Turner syndrome and hereditary lymphedema (Milroy disease).8 When a connection exists between a blood vessel and the dilated lymphatics, intermittent filling with blood occurs and the lesion is known as lymphangiectasia hemorrhagica conjunctivae.

Lymphatic abnormalities are among the congenital vascular malformations seen in KTWS and are predominantly located in the extremities and adjacent parts of the trunk (shoulder, pelvis). Other reported sites include the genitourinary system, lower gastrointestinal tract, liver, lung, and spine, with facial involvement being exceedingly rare.9 The bilateral conjunctival lymphangiectasia in this patient represents another manifestation of this poorly understood syndrome. Although lymphangiectasia is benign in nature, excision for cosmesis or diagnosis may be required.

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Soft-Tissue Perineurioma of the Bulbar Conjunctiva

Perineuromas are rare peripheral nerve sheath tumors composed exclusively of neoplastic perineurial cells and showing distinctive morphologic, ultrastructural, and immunophenotypic features that distinguish them from other nerve sheath tumors. Perineuromas can be broadly divided into 2 histological categories: an intraneu-