

Endolymphatic Sac Tumor

by Lester D. R. Thompson, MD

Endolymphatic sac tumor (ELST) is a papillary epithelial neoplasm arising within the endolymphatic sac/duct that shows a high association with von Hippel-Lindau disease (VHL). There is usually a VHL tumor suppressor gene germline mutation with an autosomal dominant inheritance pattern. Approximately 1 in 35,000 to 40,000 people have VHL, of which approximately 10 to 15% have endolymphatic sac tumors. There is a wide age range at presentation, although most patients are between 30 and 40 years; there is no gender predilection.

As the name implies, the endolymphatic duct/sac system within the posterior petrous bone is affected by the tumor. Patients present with progressive, ipsilateral hearing loss (more often sensorineural than conductive), along with tinnitus, vertigo, ataxia, and vestibular dysfunction. Importantly, patients may show signs of VHL at other anatomic sites (such as kidney, pancreas, and cerebellum).

Wide excision with careful attention to hearing preservation is attempted, although tumor size and extent may limit the ability to achieve complete removal. All patients with VHL should be radiographically screened for ELSTs. If bilateral tumors are present, they are almost always associated with VHL. The tumors will frequently expand into the posterior cranial fossa, ranging up to 10 cm in greatest dimension.

Histologically, the tumors are unencapsulated, destructive growths that result in bone invasion and remodeling. The tumor is arranged in simple, coarse, broad papillary projections within cystic spaces (figure 1). Fibrovascular cores are seen within the papillary structures. The cystic spaces may contain serum, secretions, or erythrocytes. The acinar-follicular spaces may be filled with inspissated material that mimics thyroid gland colloid. A single layer of low cuboidal to columnar epithelial cells lines the papillary projections. The cytoplasm is clear to slightly eosinophilic with indistinct cell borders. The nuclei are small, round, and hyperchromatic (figure 2). Pleomorphism, mitosis, and necrosis are nearly always absent.

Metastatic adenocarcinoma, metastatic clear-cell renal cell carcinoma, and metastatic thyroid papillary carcinoma need to be eliminated by clinical, imaging, histologic, and immunohistochemical evaluation. Specifically, a renal cell carcinoma may be challenging, as renal cell carcinoma is a component of VHL. A choroid plexus papilloma is usually midline without temporal bone destruction.

Figure 1. Low-power magnification shows many short, simple papillary structures lined by a single layer of cells. Note the secretions between the papillae.

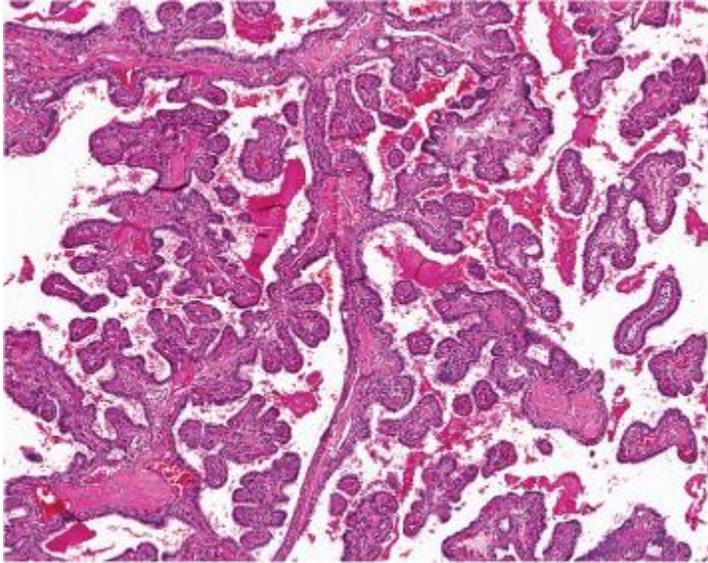
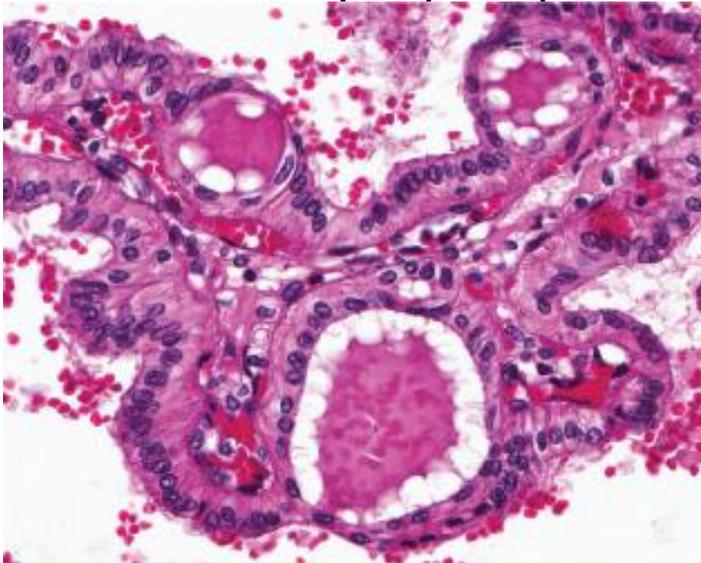


Figure 2. High-power magnification shows cuboidal to columnar cells arranged in papillary and follicular-like structures. The nuclei are round to oval with small grooves. Note the inspissated material in the lumen. Erythrocytes are present in the cysts.



Suggested reading

1. Bisceglia M, D'Angelo VA, Wenig BM. Endolymphatic sac papillary tumor (Heffner tumor). *Adv Anat Pathol* 2006; 13 (3): 131-8.
2. el-Naggar AK, Pflatz M, Ordoñez NG, Batsakis JG. Tumors of the middle ear and endolymphatic sac. *Pathol Annu* 1994; 29 (Pt 2): 199-231.
3. Heffner DK. Low-grade adenocarcinoma of probable endolymphatic sac origin. A clinicopathologic study of 20 cases. *Cancer* 1989; 64 (11): 2292-302.
4. Michaels L. Origin of endolymphatic sac tumor. *Head Neck Pathol* 2007; 1 (2): 104-11.

*From the Department of Pathology, Woodland Hills Medical Center, Southern California
Permanente Medical Group, Woodland Hills, Calif.*

Ear Nose Throat J. 2013 April-May;92(4-5):184-188.