Endolymphatic sac tumors in von Hippel-Lindau disease

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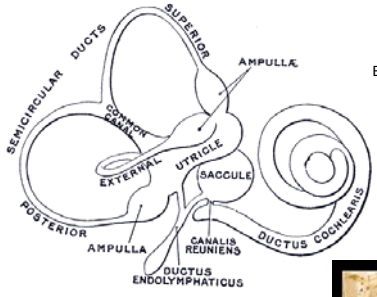
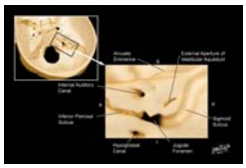
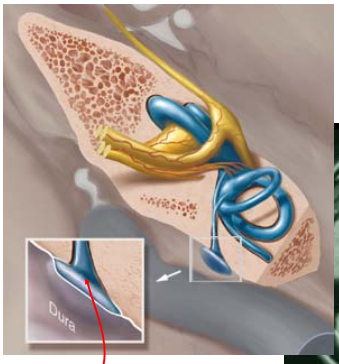
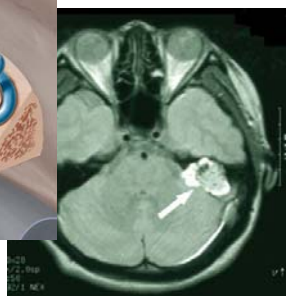


Endolymphatic duct / sac



Part of the inner ear, connecting the **cochlea** (for hearing) with the **semicircular canals** (for balance)





Tumor arises from distal endolymphatic duct and/or endolymphatic sac

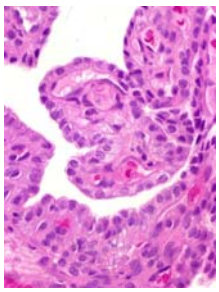
Genetics

- Rare in general population: most cases are sporadic
- Definite association with VHL
- Seen in **11-16%** of VHL patients
- Occur at younger age in VHL, can be bilateral (30%)

Fluorescent *in situ* hybridization shows loss of one copy of VHL gene in each of 2 tumor cells

Pathology

- Papillary cystadenomas = highly vascular tumor within middle ear
- Infiltrates and destroy adjacent bone and dura
- Destroys retrolabyrinthine petrous temporal bone in region of vestibular aqueduct, then spreads to supra- and infralabyrinthine and mastoidotympanic regions
- Strong vascularity, with blood supply from ascending pharyngeal and stylomastoid arteries



Differential diagnosis

V Jugular bulb anomalies (high riding bulb, dehiscent jugular bulb, and jugular bulb diverticulum), aberrant internal carotid artery (ICA), hemangioma, persistent stapedial artery

I Otitis media, otitis externa, malignant otitis externa, tuberculous otitis

T Tympanosclerosis

A Granulomatous diseases (*e.g.*, Wegener's granulomatosis)

M Osteoradionecrosis

I Retained PE tube, Foreign body

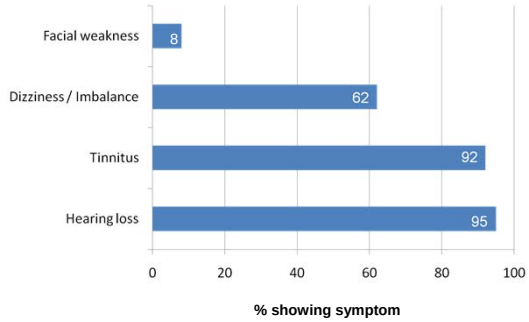
N Cholesteatoma, paraganglioma / glomus tympanicum tumor, schwannoma, adenoma, **endolymphatic sac tumor**, cholesterol granuloma, polyps, adenocarcinoma, squamous cell carcinoma, adenoid cystic carcinoma

C Cholesteatoma, encephalocele

Clinical presentation

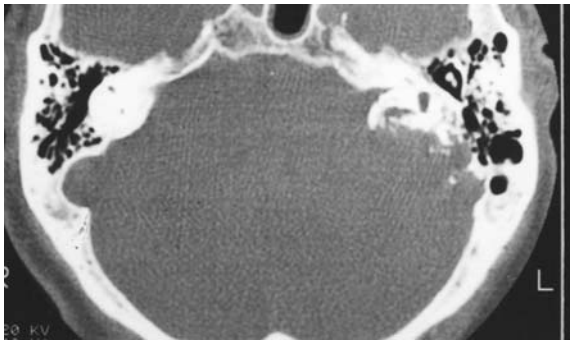
- May be found incidentally if small
- Unilateral hearing loss and vestibular dysfunction are prominent symptoms
- Tinnitus and vertigo also frequent
- Duration of hearing loss: 3-6 months' onset, but can be much longer (*i.e.* years)
- Tends to occur early: mean age of onset = 22 yrs
- Usually grow slowly and locally (no metastasis)

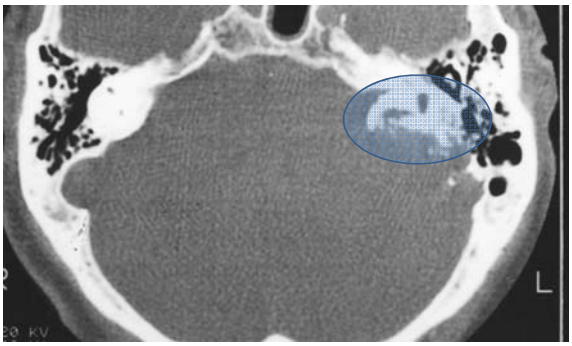
Spectrum of symptoms

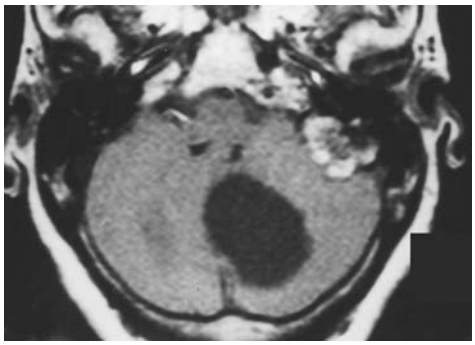


Case #1

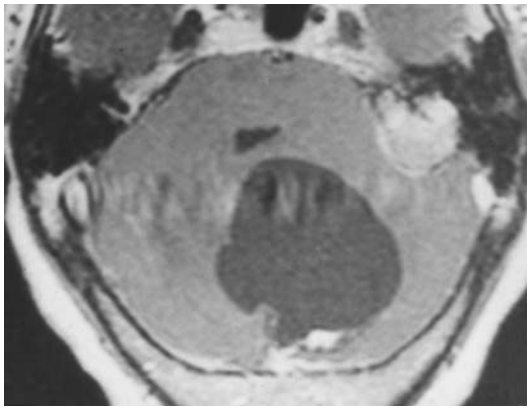
- 54 yo woman with chronic headache / nausea
- Also hearing loss x 3 years
- Exam showed ataxia and left facial weakness
- Audiogram, CT, and MRI obtained

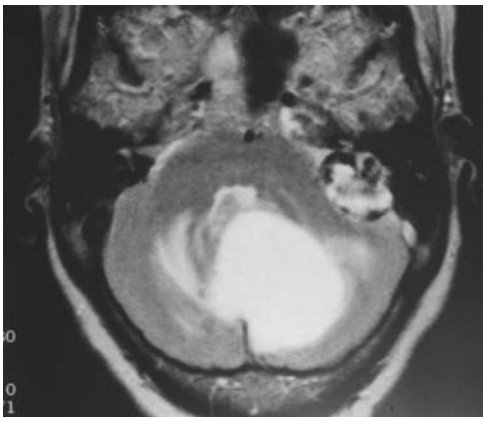




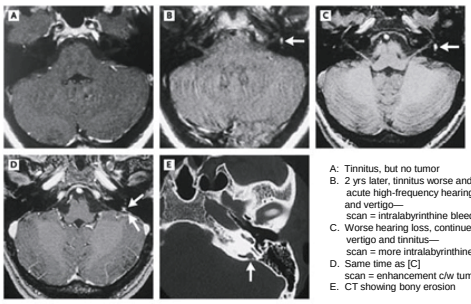


Scattered areas of increased signal intensity seen in tumors on T1-weighted images = breakdown products of subacute hemorrhage, cholesterol clefts, and proteinaceous cysts





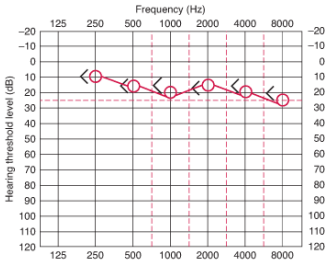
Case #2: 34 yo woman



A: Tinnitus, but no tumor
B. 2 yrs later, tinnitus worse and new acute high-frequency hearing loss and vertigo—
scan = intralabyrinthine bleed
C. Worse hearing loss, continued vertigo and tinnitus—
scan = more intralabyrinthine bleed
D. Same time as [C]
scan = enhancement c/w tumor
E. CT showing bony erosion

After surgery, hearing unchanged and tinnitus/vertigo better

Normal audiogram



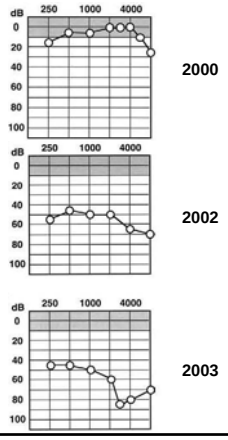
Hearing loss

- 60% of VHL patients with hearing loss show no tumor on scan, indicating:
  - microscopic tumor
  - other cause of hearing loss
- Often caused by bleeding inside tumor, rather than direct invasion of inner ear structures by tumor
- Do not confuse with Menière's disease
  - Hearing loss, tinnitus, vertigo due to excess endolymph
  - Tumor can cause this by producing fluid, blocking its reabsorption, or bleeding leading to inflammation
- Once lost, hearing usually does not return…due to cochlear disruption or eighth nerve damage
- → Intervene early!

Hearing loss

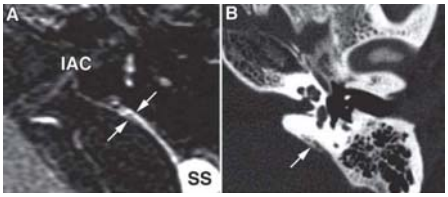
- Occurs variably:
  - Suddenly 43%
  - Stepwise over 3-6 m 43%
  - Gradually (over > 6 m) 14%

Deterioration of hearing over time (case 2)



Best imaging

- CT (bone windows) with thin cuts through skull base
- MRI = fluid-attenuated inversion-recovery [FLAIR], spoiled-gradient [SPGR], standard T1 and T2…. with thin cuts
- Tumor can be VERY subtle



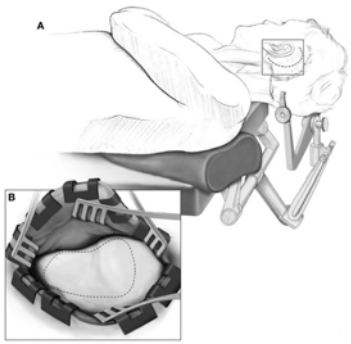
Treatment

- First-line treatment: **SURGERY**
- Radiosurgery can be considered but experience is almost non-existent
  - reported in several cases of recurrence, but follow-up inadequate
- Must remove dura, endolymphatic sac, and involved portion of endolymphatic duct
- Visible tumor with intact hearing: operate to preserve hearing
- Visible tumor and pt deaf: operate to help other symptoms *or* to protect brainstem

Surgical approaches

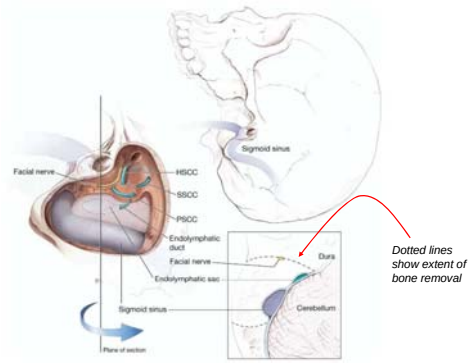
- Choices
  - Retrolabyrinthine posterior petrosectomy [BEST]
  - Transmastoid approach [does not expose EL sac and duct completely unless mastoid air cells are very generous]
  - Retrosigmoid approach [hard to show entire posterior semicircular canal and EL duct safely, and has higher risk for CSF leak]
- If tumor is large and hearing is present
  - Combined RPP + retrosigmoid
- If tumor is large, but hearing absent
  - Combined retrosigmoid + translabyrinthine

Retrolabyrinthine posterior petrosectomy



Lonser and Kim, Operative Tech Otolaryngol 18:65, 2007

Retrolabyrinthine posterior petrosectomy



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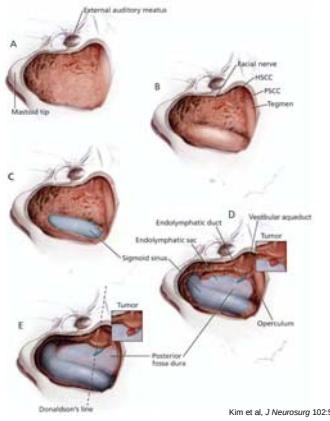
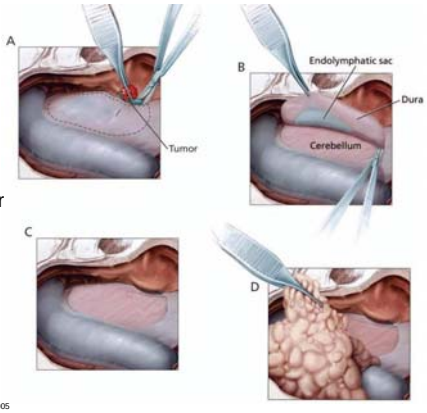
Stages of the operation



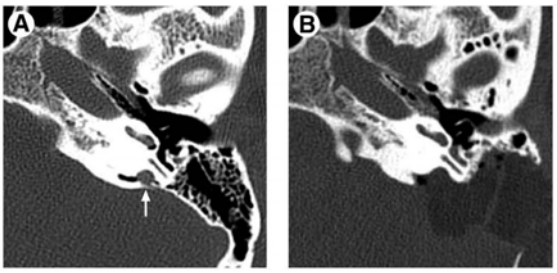
Kim et al, *J Neurosurg* 102:503, 2005

The operation proceeds further



Kim et al, *J Neurosurg* 102:503, 2005

Before and after….



SURGICAL RESULTS

Summary of characteristics in five patients who underwent resection of ELSTs

Case No.	Age (yrs), Sex	Tumor Side	Tumor Diameter (mm)*	Preop Symptoms†	Postop Results	Follow Up (mos)
1	42, M	lt	5	hearing loss (SRT 60 dB), vertigo, & tinnitus	stable hearing, resolution of vertigo, & tinnitus	18
2	40, M	lt	19	hearing loss (SRT 35 dB), vertigo, tinnitus, & aural fullness	stable hearing, resolution of vertigo, tinnitus, & aural fullness	18
3	27, F	lt	11	temporary mild hearing loss (SRT 25 dB) & vertigo	stable hearing & resolution of vertigo	12
4	34, F	lt	3	hearing loss (SRT 40 dB), vertigo, tinnitus, & aural fullness	stable hearing, resolution of vertigo, tinnitus, & aural fullness	12
5	31, M	lt	7	vertigo	stable hearing & resolution of vertigo	6

* Based on the largest diameter measured on MR images at the time of surgery.
† Audiogram speech reception threshold (SRT) hearing loss in decibels immediately before resection.

All resolved vertigo
All resolved tinnitus
All resolved stable hearing
All had SMALL tumors

Kim et al, *J Neurosurg* 102:503, 2005

Surgical results: NIH series
(n = 31 with 33 ELSTs)

Tumor seen on CT or MRI	88%
Prior intralabyrinthine bleed	12%
Approach	
Retrolabyrinthine petrosectomy	73%
Transotic	12%
Translabyrinthine	6%
Presigmoid / retrosigmoid	9%
Resection	
Complete	91%
Delayed recurrence: one patient at 46 m	

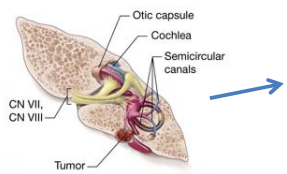
Kim et al: Laryngoscope 123:477 (2012

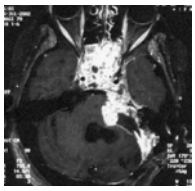
Surgical results: NIH series
(n = 31 with 33 ELSTs)

Resolution of symptoms	
Hearing stable or improved [improvement in 3/23 = 13%]	97%
Vertigo resolved	86%
Vertigo improved	14%
Tinnitus resolved or improved	96%
Complications	
CSF leak	6%
Transient cranial nerve paresis	3%

Kim et al: Laryngoscope 123:477 (2012)

Sometimes it can get bad…..





Endolymphatic sac tumor metastatic to the spine

Case report

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Endolymphatic sac tumors (ELSTs) are aggressive papillary lesions of the temporal bone. Although histologically benign, they may exhibit invasive growth and destruction of the skull base. Patients generally present with symptoms referable to the lesion's location within the middle or posterior cranial fossa. Although well characterized as a distinct entity, ELSTs involved in metastatic dissemination have never been reported. In the present report the authors describe a case of ELST metastatic to the spine treated with resection.

Key Words • endolymphatic tumor • metastasis • lumbar spine • resection

Thanks for listening! -- Hearing is important!