Endolymphatic Sac Tumors and von Hippel-Lindau Disease

Samuel Gubbels, M.D.
Associate Professor, Neurotology
Director, UCHealth Hearing and Balance Clinics
Department of Otolaryngology
University of Colorado
Denver, CO, USA

Disclosures

• Scientific advisory board
  – Roche Pharmaceuticals, Applied Genetic Technologies Corporation
• Investigator
  – Multi-institutional 5 year study of clinical outcomes of the Cochlear Nucleus Hybrid L24 device
• Grant funding – NIH/NIDCD
• Grant funding – Med El Corporation

Outline

• Anatomy of ELS
• Definition and epidemiology
• Symptoms
• Evaluation and treatment
• Adjunctive measures
• Questions

Where is the Endolymphatic Sac (ELS)?

• Along the dural lining of the brain
• Back portion of the skull – posterior fossa
• <1 cm triangular shape
• Flat
• Fibrous in quality

https://www.semanticscholar.org/paper/Tumors-of-the-endolymphatic-sac-in-von-disease.-Lonser-Kim/978cfd5f3a0d7bee1649eeab6f5c6b70d3e6bb7/figure/0
What does the ELS do?

- Part of the inner ear membranous system
- Endolymph homeostasis
- Removes waste
- Endocrine/paracrine
- Meniere’s disease
  - Vertigo attacks
  - Hearing loss, ringing, ear fullness

What is an Endolympathic Sac Tumor?

- Rare tumor
- Slow growing
- Locally erosive
- Low grade
- Vascular – red/brown
- Type of adenocarcinoma
- Metastases are rare

History of ELST

- Heffner (1989) – 20 cases
  - “probable” endolymphatic sac origin
- Li (1993) – Reclassified as “ELST”
- Magerian (1995) – found 8 cases of ELST in cadaveric skull base collection
- Manski (1997) – part of vHL
- World Health Organization (2012)
  - Endorsed “Endolympathic Sac Tumor” officially

Epidemiology

- 300 reported cases
- No reported metastases
  - 1 spinal “drop” metastasis
- No gender predilection
- 15-77 years old
- 40 yo average
- vHL
  - pVHL tumor suppressor
  - Female 2:1 ELST
  - 12-50 years old
  - 30 year old average
  - 6-15% of vHL patients
    - 30% both sides with ELST
    - 32% with ELST at initial presentation of vHL
What Symptoms do ELST Cause?

- Hearing loss (86-100%)
  - Progressive > sudden
- Ringing in the ear/tinnitus – (71-89%)
- Vertigo/dizziness (47-70%)
- Ear fullness (37%)

How is an ELST Diagnosed?

- Audiogram
  - Inner ear pattern
    - Sensorineural
  - Asymmetric hearing
  - Triggers imaging workup

- Imaging
  - Computed Tomography (CT)
How is an ELST Diagnosed?

- Audiogram
  - Inner ear pattern
    - Sensorineural
- Sensorineural asymmetry
  - Triggers imaging workup
- Imaging
  - Computed Tomography (CT)
  - Magnetic Resonance Imaging (MRI)
  - PET/CT

Screening for ELST in vHL?

<table>
<thead>
<tr>
<th>Table 1</th>
</tr>
</thead>
<tbody>
<tr>
<td>Grade</td>
</tr>
<tr>
<td>I</td>
</tr>
<tr>
<td>II</td>
</tr>
<tr>
<td>III</td>
</tr>
<tr>
<td>IV</td>
</tr>
</tbody>
</table>

Staging of ELST


Papadakis et al. (2016) Clin Nuc Med

Varshney et al. (2017) J. Kidney Cancer and vHL

How are ELST’s Treated?

• Surgery as mainstay
• Complete excision paramount
• Hearing preservation possible with small tumors
• Embolization possibly
• Variety of surgical approaches
  – Incisions behind ear in general

Surgical Details

• Inpatient procedure in general
• Length of stay – 2-3 days
• Risks
  – Hearing loss, dizziness
  – Facial nerve
  – CSF leakage
  – Recurrence
  – Bleeding
  – Infection

Outcomes after Surgery

• Small studies
• Control rates – 75-92% at 1-2 years
• Hearing loss common
• Cranial nerve problems
• Radiotherapy or addl surgery can be required
  – Incomplete excision, recurrence

Additional Treatments

• Radiation therapy – multiple types
  – Poor surgical candidates
  – Recurrences - multifocal
  – Recurrence – surgical morbidity unacceptable
• Reports
  – Primary
  – Adjunctive - incomplete resection
• Control rates vary
  – Possibly better with radiosurgical techniques
Rehabilitation

• Dizziness – Vestibular therapy
• Facial nerve problems
  – Variety of procedures to help
  – Rehabilitation can be beneficial depending
• Hearing loss
  – Generally a complete loss
  – Hearing aids not helpful
  – Route to other side
    • CROS or Bone anchored hearing aid

Hearing Rehabilitation

• Cochlear implantation sometimes possible
• Hearing on other side?
• Status of cochlea, auditory nerve
• Multiple benefits
  – Hearing in noise
  – Localization

Conclusions

• Screening and early detection paramount
• Hearing loss most common symptom
• Small tumor with better outcomes
• Surgery as mainstay of treatment
  – Complete resection very important
• Other treatments may be necessary
• Multiple hearing rehabilitation options