Surgical and Non-Surgical Causes of Progressive Hearing Loss in Children: What can be done about it?

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Because Children Are Our Future

Cochlear Implant Team
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The Sound Wave Symposium


WELL BEHAVED
WOMEN
RARELY MAKE
HISTORY

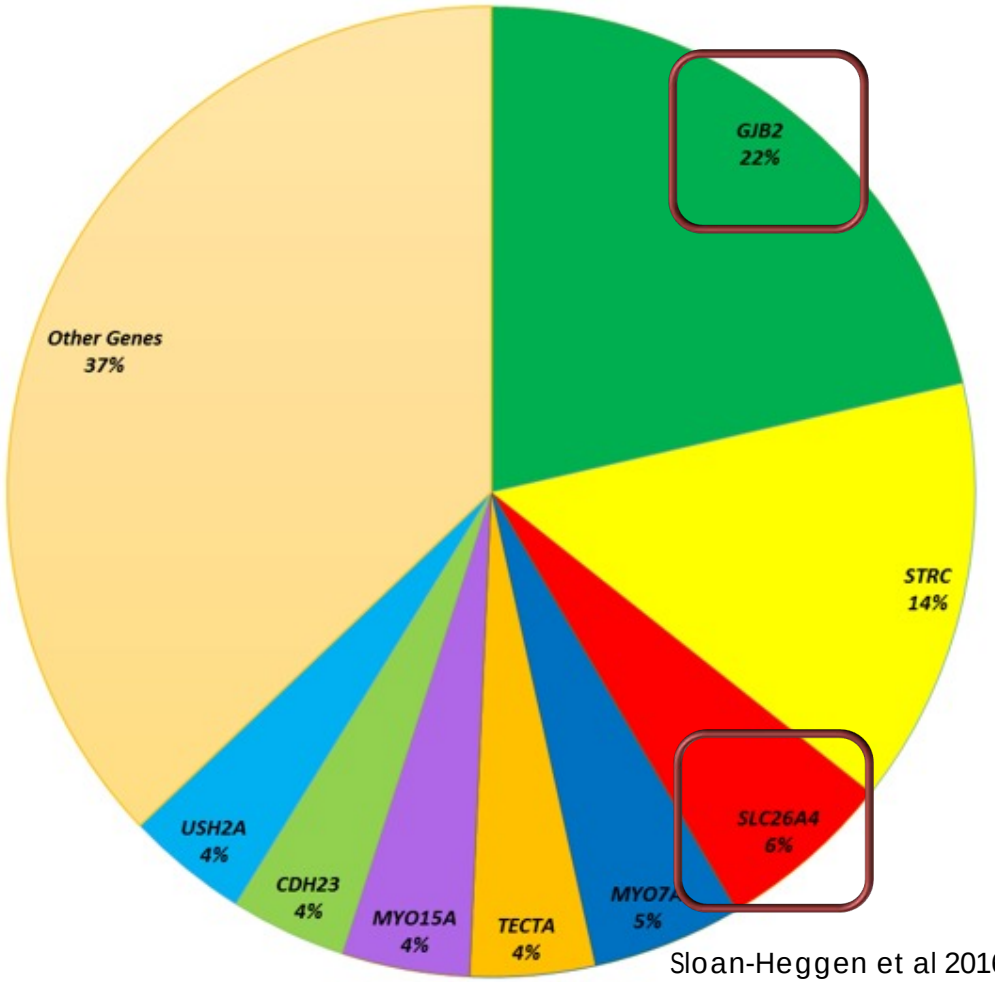
Pediatric Progressive Hearing Loss

- Non-surgical
  - Genetic
  - CMV
- Surgical
  - Tympanic membrane retraction → Cholesteatoma
  - Tympanosclerosis
  - Otosclerosis

Pediatric Progressive Hearing Loss

- Non-surgical
  - Genetic
    - Connexin 26 and Enlarged Vestibular Aqueduct
  - CMV
- Surgical
  - Tympanic membrane retraction → Cholesteatoma
  - Tympanosclerosis
  - Otosclerosis

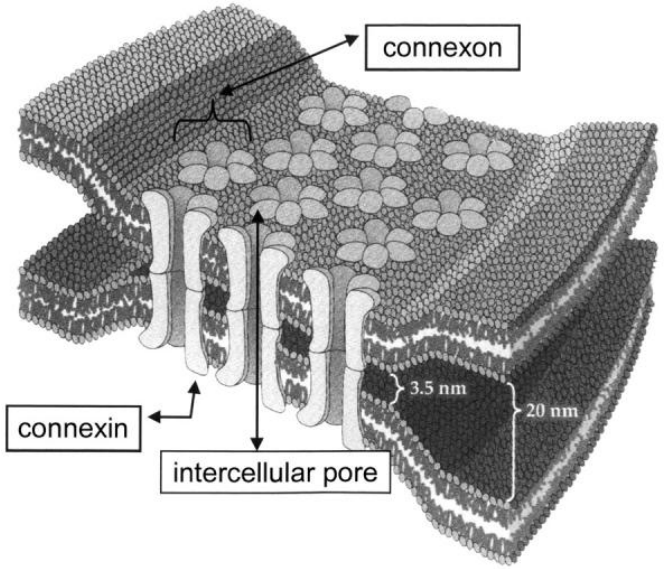
Genetic causes of SNHL

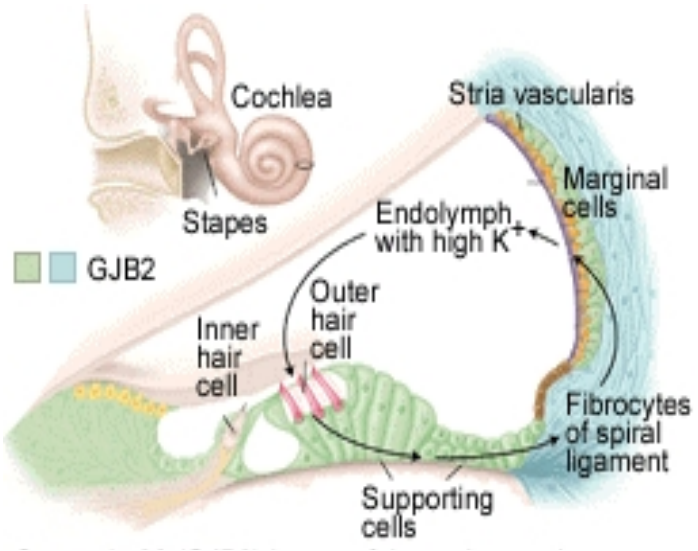


Sloan-Heggen et al 2016; Smith et al, unpublished data

GJB2 (Connexin 26)

- DFNB1
- Gene: GJB2 (gap junction beta 2)
- Connexin 26: protein





GJB2 (Connexin 26)

- Hearing loss ranges from mild to profound
- Majority congenital

- More than 50% will have HL progression
  - generally gradual
  - occasionally precipitous
  - Influenced by genetics and other factors

GJB2 – Progressive hearing loss

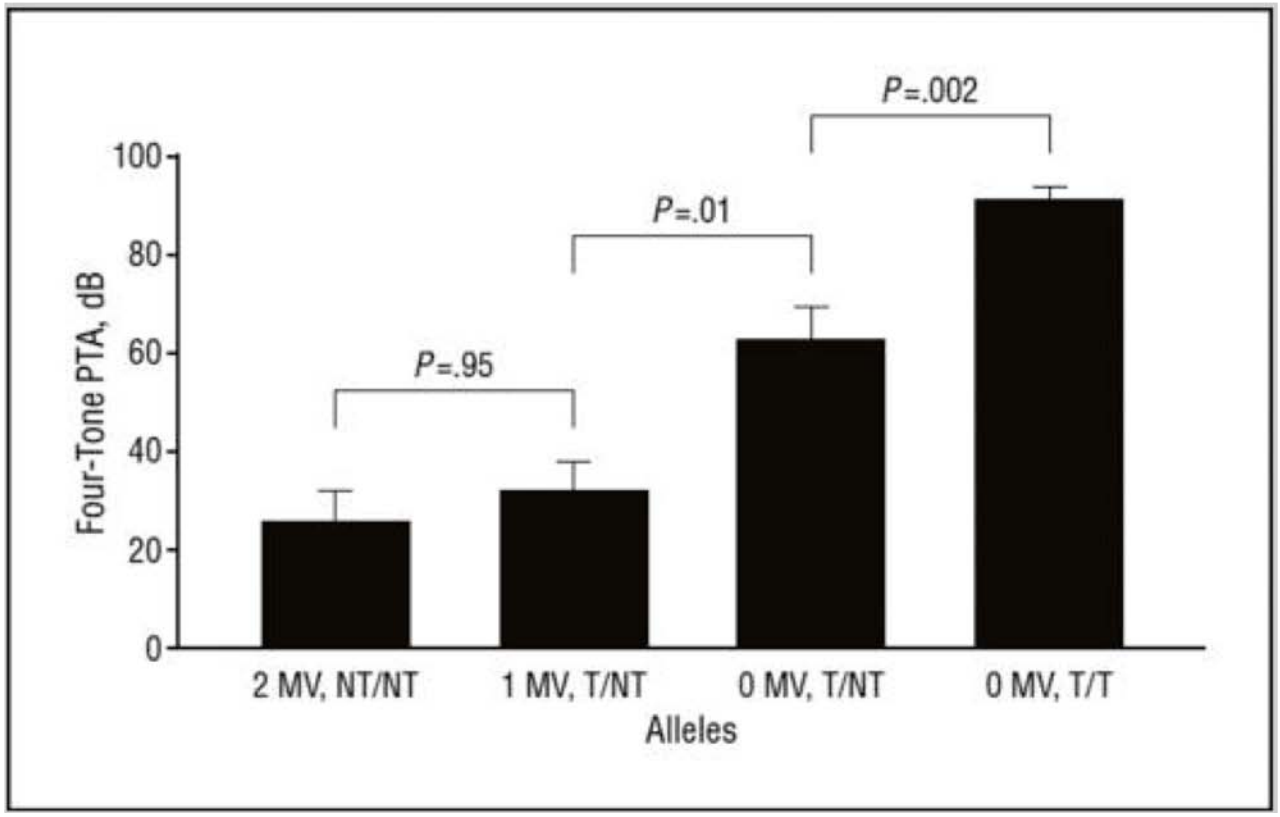
- Kenna et al, 2010
- 126 patients with SNHL due to GJB2
  - 33% had bilateral profound SNHL when initially identified
  - 67% presented with mild to severe SNHL
    - 56% have had progression of their hearing loss
    - 3 of them possibly due to other causes

Kenna et al, Arch Otolaryngol Head Neck Surg.2010

Mutation	No. (%) of 225 Alleles Identified
Truncating	
35delG	137 (53.9)
167delT	22 (8.7)
235delC	8 (3.1)
313_326del14bp	5 (2.0)
E47X	4 (1.6)
M1V	2 (0.8)
W24X	2 (0.8)
Q57X	2 (0.8)
176_191del16	2 (0.8)
299_300delAT	1 (0.4)
333_334delAA	1 (0.4)
453_460del8ins9[a]	1 (0.4)
631_632delGT	1 (0.4)
Total	**188 (74.1)**
Nontruncating	
V37I	23 (9.4)
M34T	22 (9.1)
V84L	2 (0.8)
L90P	2 (0.8)
N206S	2 (0.8)
S199F	2 (0.8)
T8M	1 (0.4)
G12V	1 (0.4)
K15T	1 (0.4)
R32C	1 (0.4)
I35S	1 (0.4)
V95M	1 (0.4)
[E114G;V27I](in *cis*)[b]	1 (0.4)
S139N	1 (0.4)
R143W	1 (0.4)
V153I	1 (0.4)
R184P	1 (0.4)

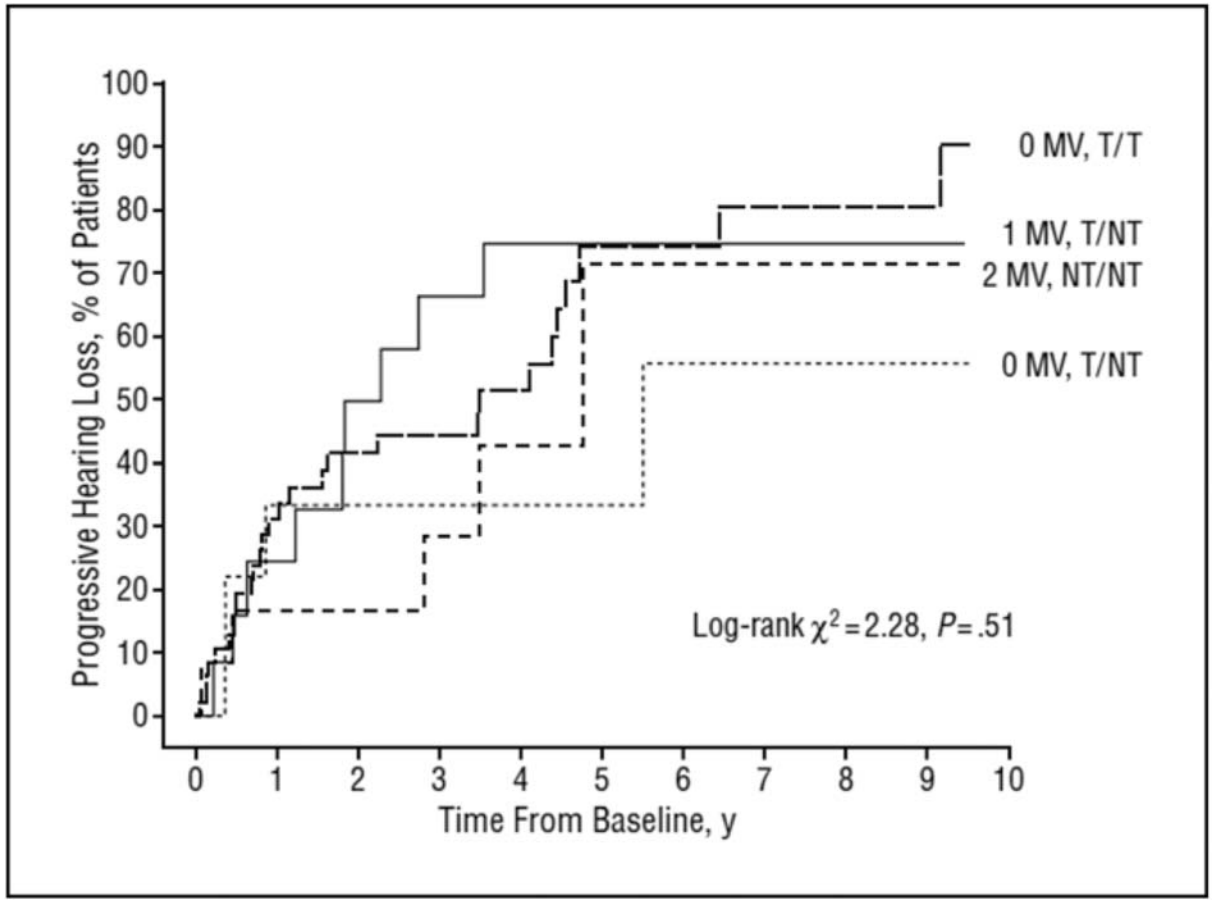
Kenna et al, Arch Otolaryngol Head Neck Surg.2010

GJB2 – Progressive hearing loss



Kenna et al, Arch Otolaryngol Head Neck Surg.2010

GJB2 – Progressive hearing loss

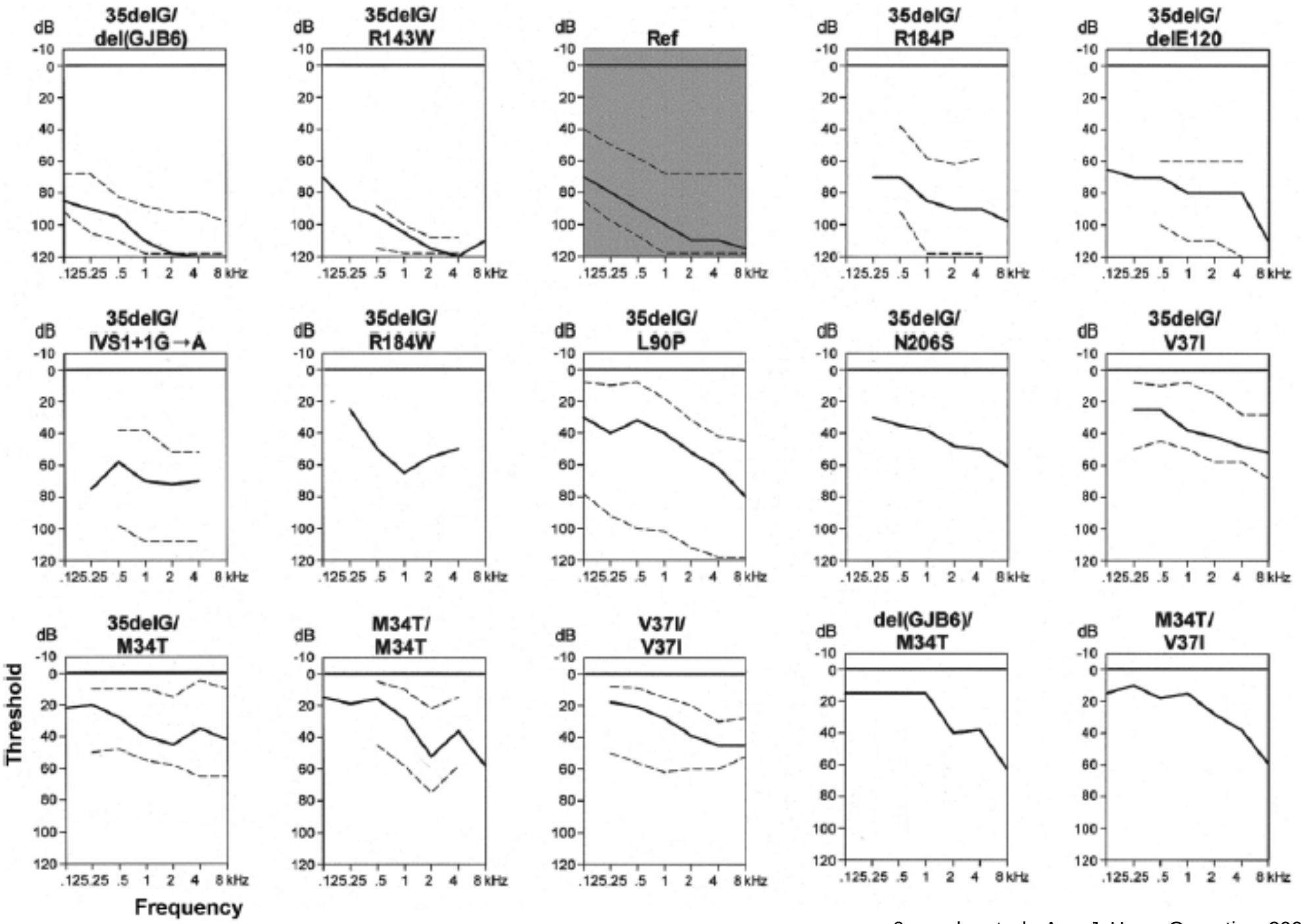


Kenna et al, Arch Otolaryngol Head Neck Surg.2010

GJB2 – Progressive hearing loss

Race/Ethnicity	Progressive HL	Non-progressive HL
White, non-Hispanic	40 (85%)	24 (65%)
White, Hispanic	3 (6%)	5 (14%)
Asian	2 (4%)	7 (19%)
African American and white	2 (4%)	0
Asian and white	1 (2%)	1 (3%)

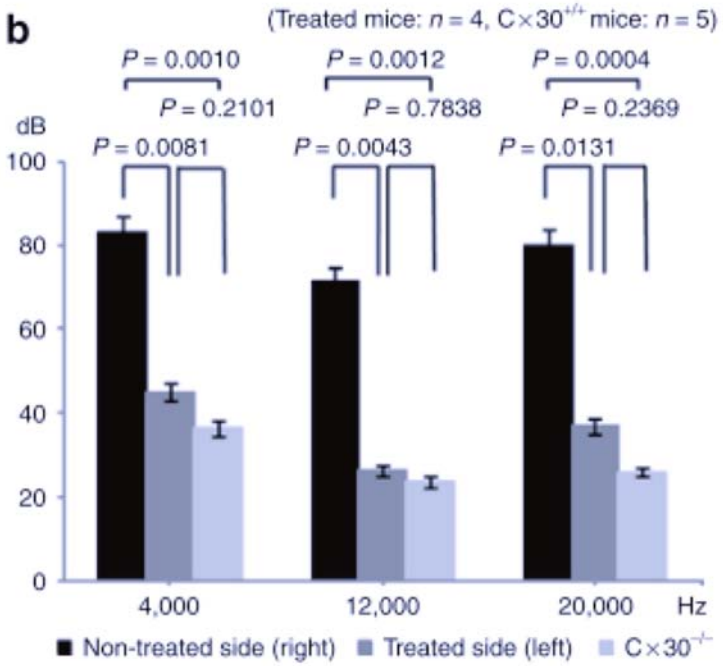
Kenna et al, Arch Otolaryngol Head Neck Surg.2010

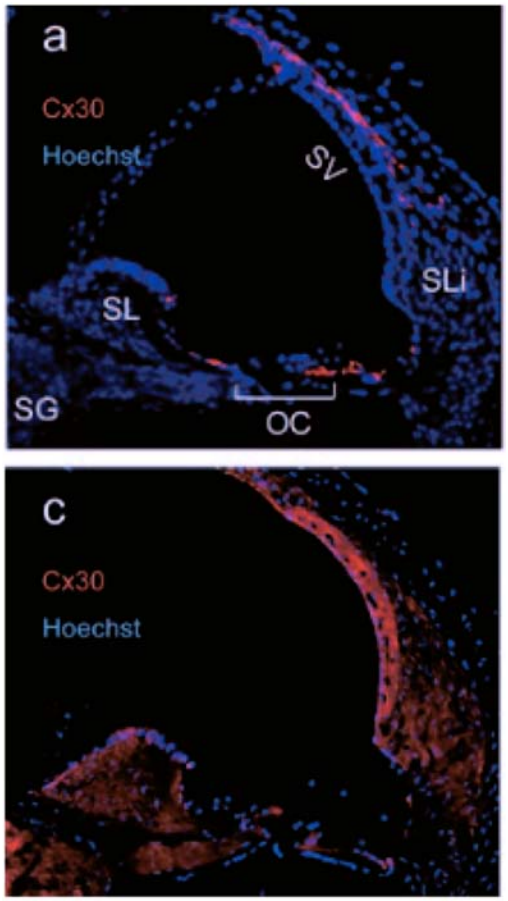


Snoeckx et al., Am. J. Hum. Genetics, 2005

GJB2 – Progressive hearing loss

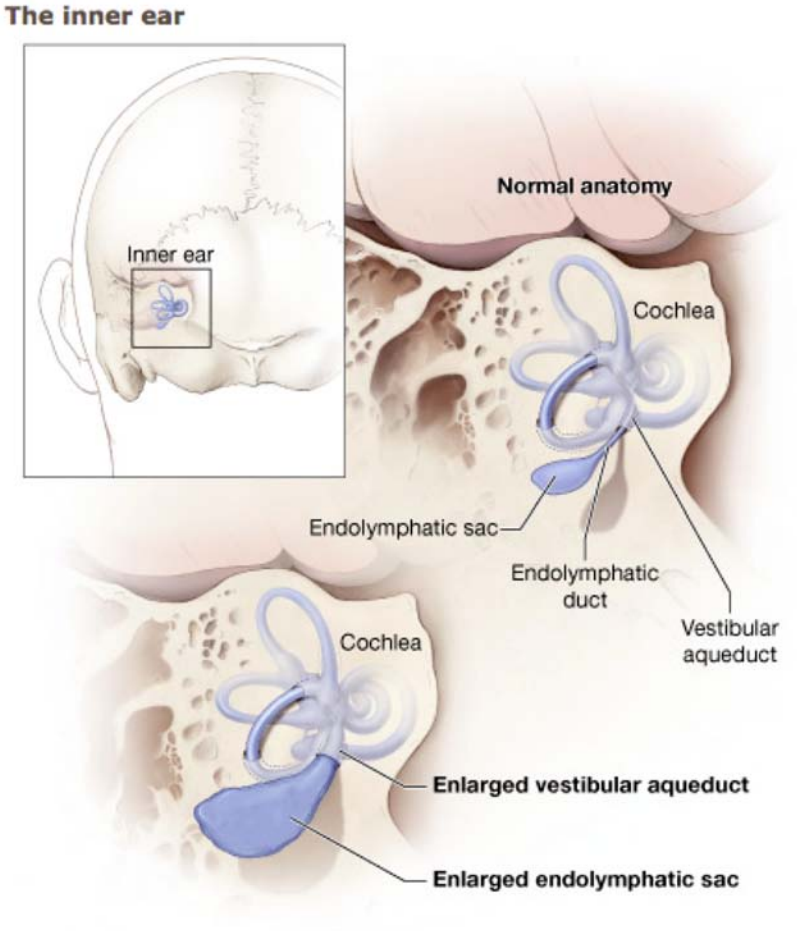
- Future





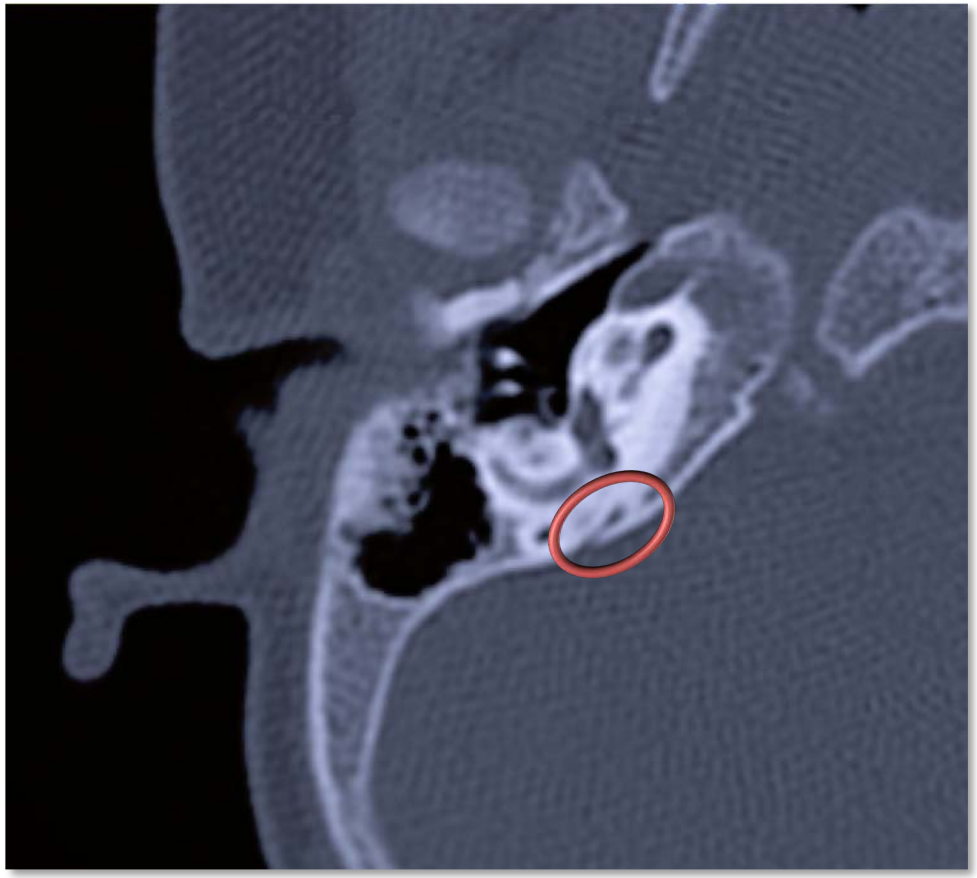
Miwa et al., Molecular Therapy, 2013

Enlarged Vestibular Aqueduct

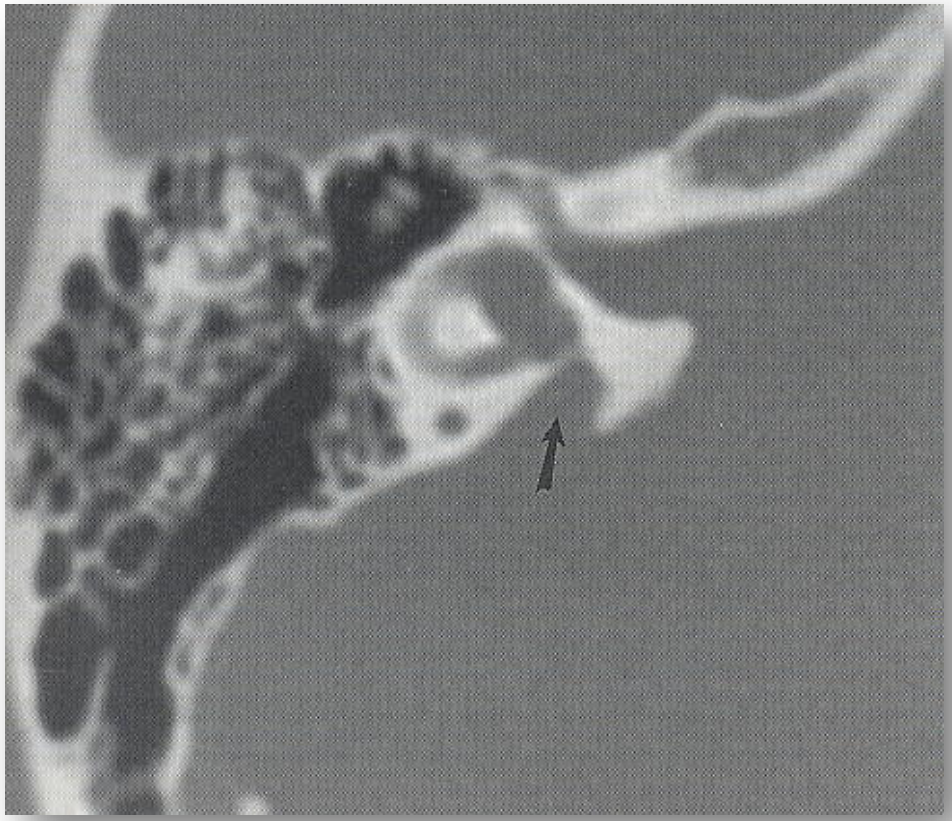


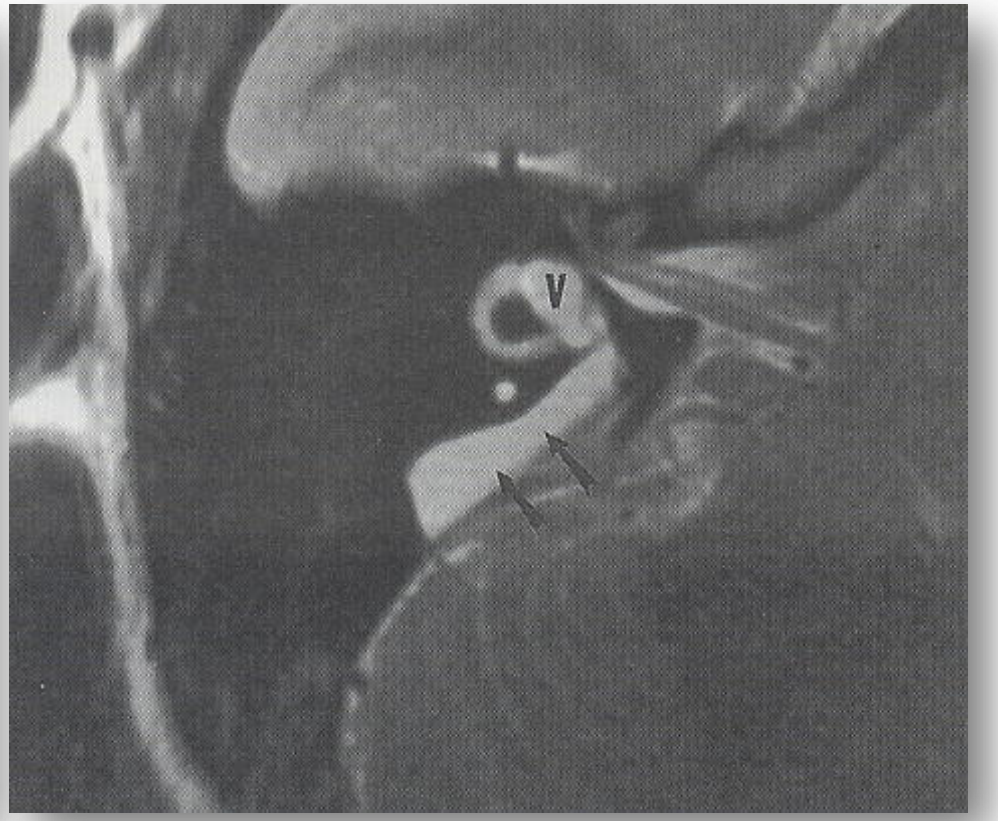
Credit: NIH Medical Arts

Enlarged Vestibular Aqueduct



Enlarged Vestibular Aqueduct





Enlarged Vestibular Aqueduct

- Most common inner ear abnormality
- Normal size 0.4 - 1.0 mm
- Abnormal >1.5 mm at its midpoint
- Bilateral (up to 94%)

Enlarged Vestibular Aqueduct

- At birth
  - Normal to profound SNHL
- Progression
  - Can deteriorate into early adulthood (25 dB per 6 years)
  - Can have sudden decrements of hearing (65%) – can be related to head trauma/pressure changes

Enlarged Vestibular Aqueduct

- Ascha et al, *JAMA* 2017

- For each millimeter increase in vestibular aqueduct size (greater than 1.5 millimeters)
  - increase of 17.5 dB in speech reception threshold
  - decrease of 21% in word recognition scores

- For every year after the initial audiogram
  - speech recognition threshold increased by 1.5 dB
  - word recognition decreased by an additional 1.7%

Enlarged Vestibular Aqueduct

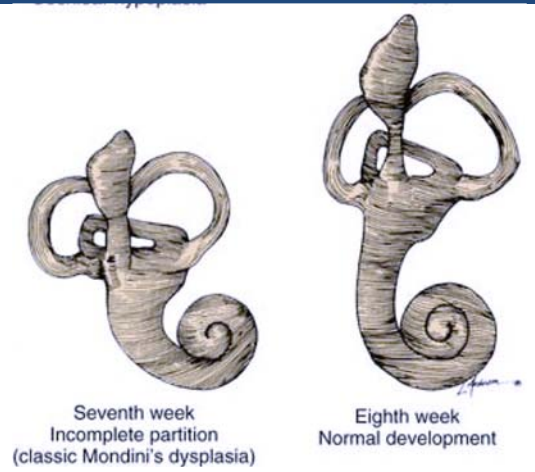
- Up to 1/3 patients with conductive component
  - Increased pressure dampening stapes
  - 3rd window effect
- Risk of CSF “gusher” with stapedectomy and cochlear implantation
  - Some have suggested increased incidence of PLF

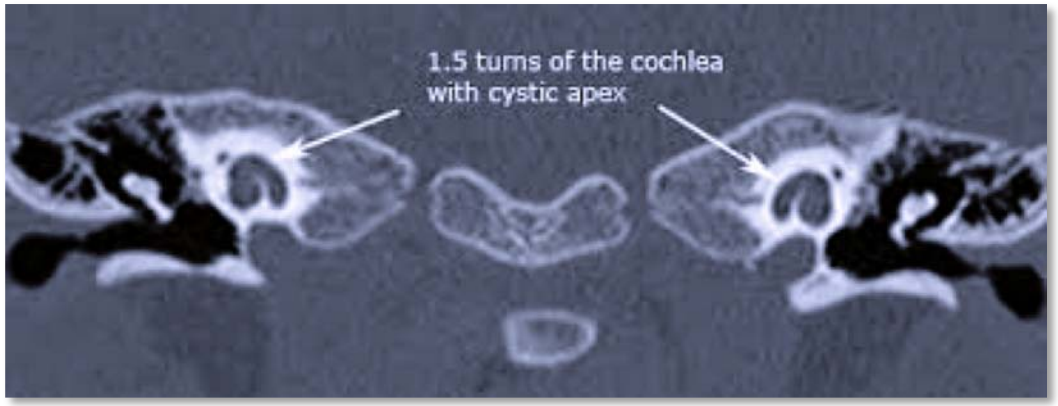
Enlarged Vestibular Aqueduct

- *Pendred syndrome* versus *Nonsyndromic enlarged vestibular aqueduct* (PDS/NSEVA)
- Phenotypic spectrum:
  - Sensorineural hearing loss (SNHL)
  - Vestibular dysfunction
  - Temporal bone abnormalities (bilateral enlarged vestibular aqueduct with or without cochlear hypoplasia)
  - PDS also includes development of euthyroid goiter in late childhood to early adulthood

Enlarged Vestibular Aqueduct

- Pendred syndrome
  - 50% with euthyroid goiter
  - EVA with/without cochlear hypoplasia (1.5 turns)
- NSEVA
  - No goiter
  - Only EVA present, no cochlear hypoplasia



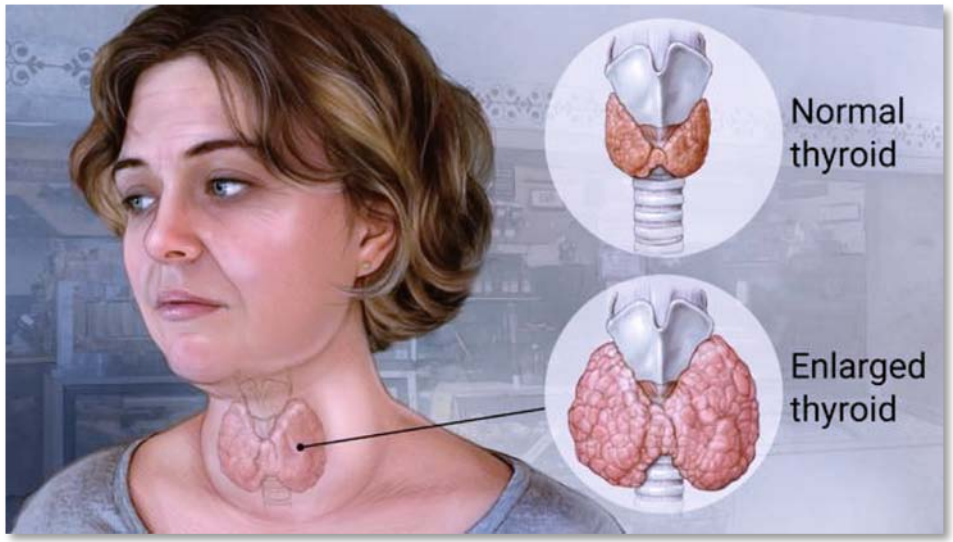


Enlarged Vestibular Aqueduct

- Autossomal recessive
  - Biallelic pathogenic variants in *SLC26A4* or
  - double heterozygosity (*SLC26A4* and either *FOXI1* or *KCNJ10*)

Enlarged Vestibular Aqueduct

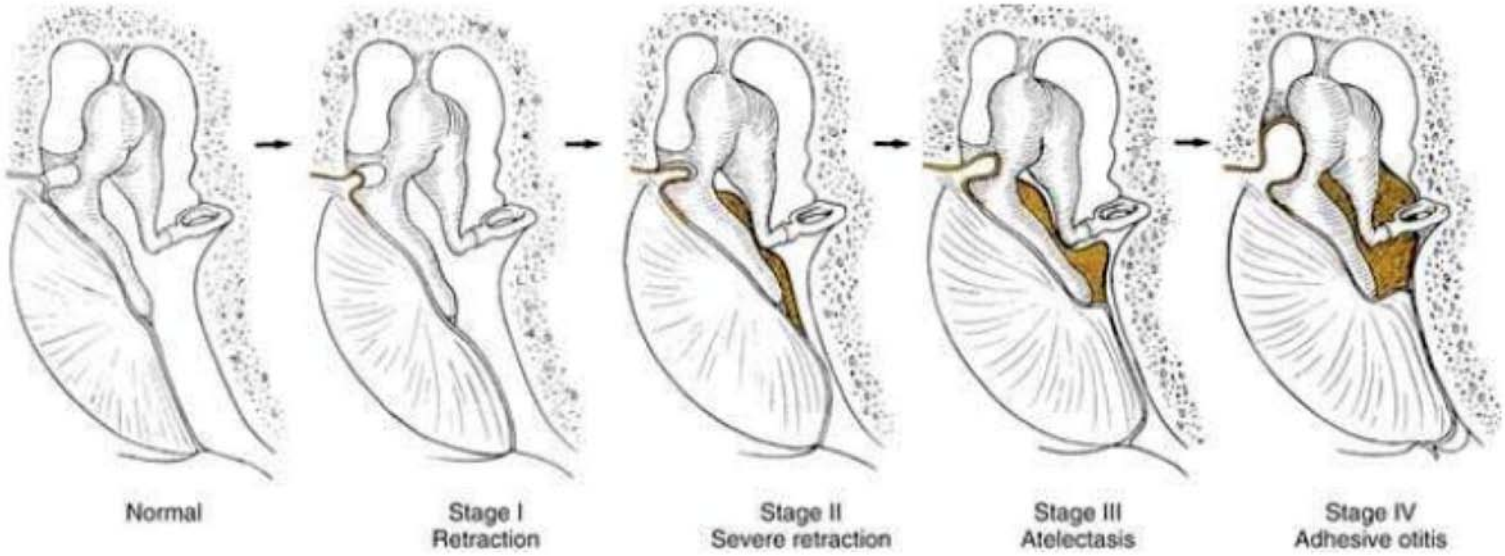
- Treatment of manifestations:
  - Hearing loss
  - Goiter/hypothyroidism



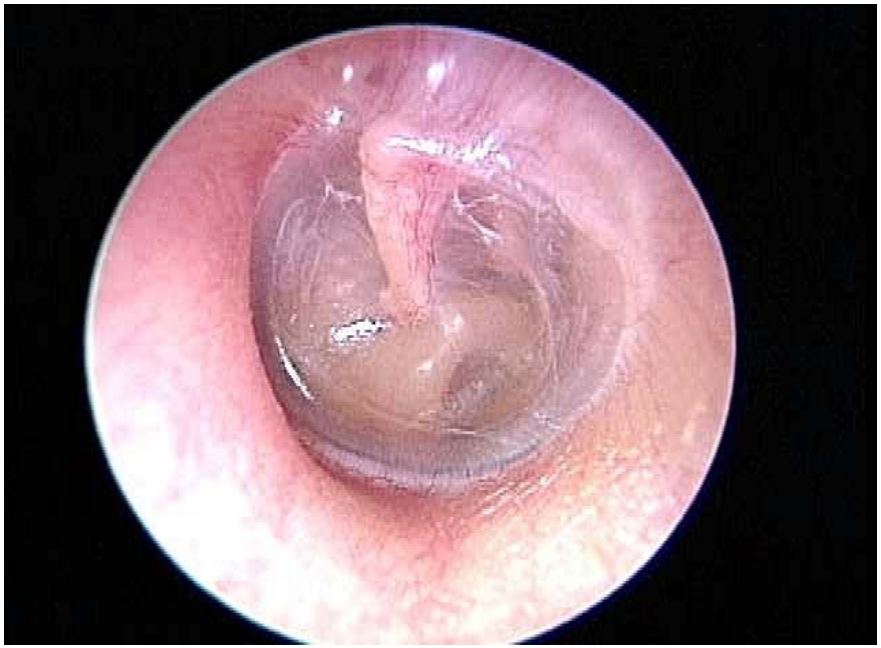
Enlarged Vestibular Aqueduct

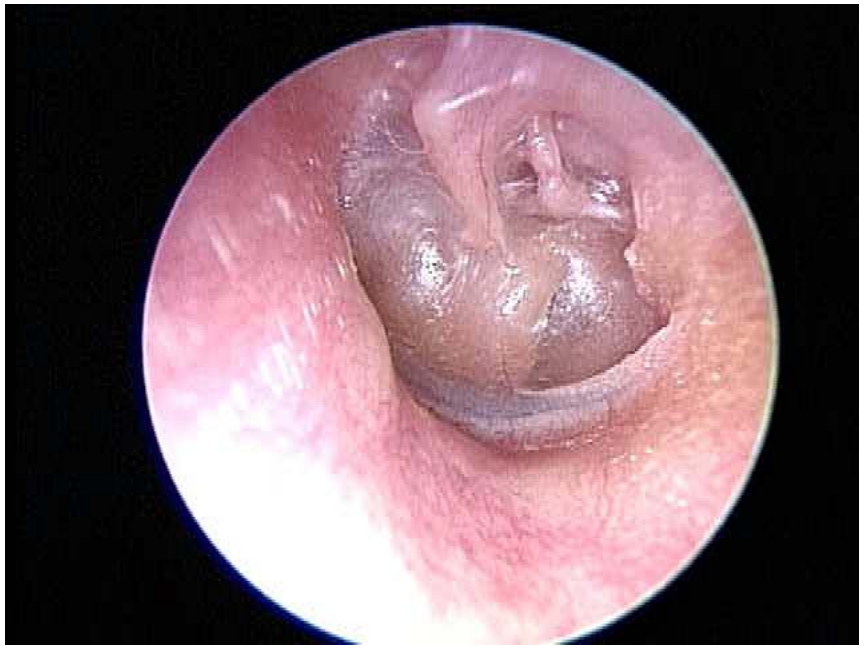
- Surveillance:
  - Audio every 3-6 months initially, then 6-12 months
  - Baseline ultrasound examination of the thyroid
    - Monitor volumetric changes (clinical and/or US)
- Agents/circumstances to avoid:
  - Dramatic increases in intracranial pressure can be associated with a sudden drop in hearing
  - ➢ Avoiding weightlifting and/or contact sports should be discussed with a physician

TM retraction / Cholesteatoma

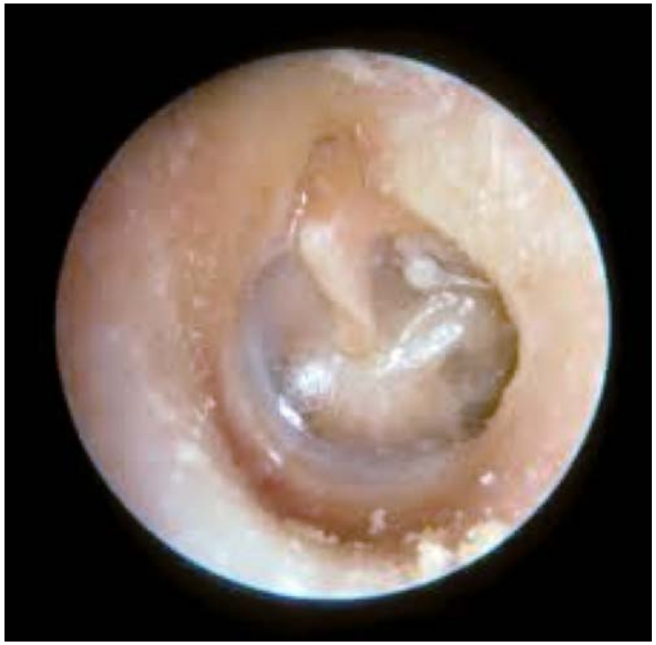


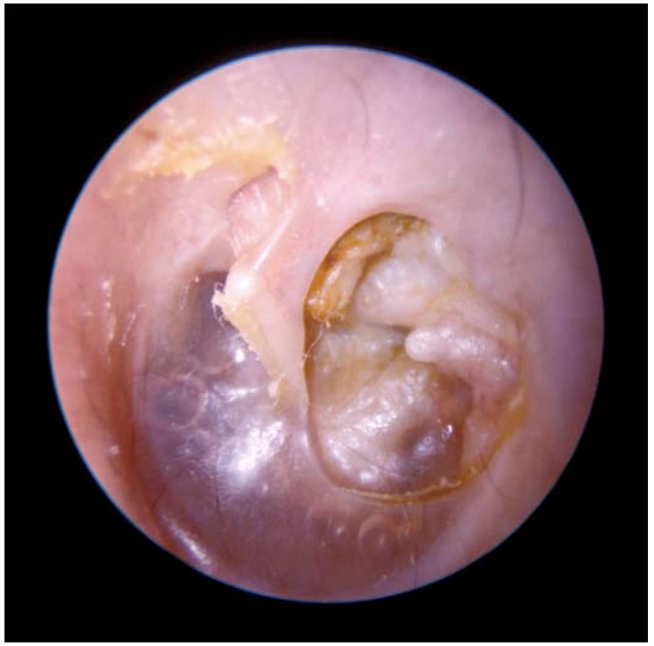
Cholesteatoma - progression





Cholesteatoma - progression

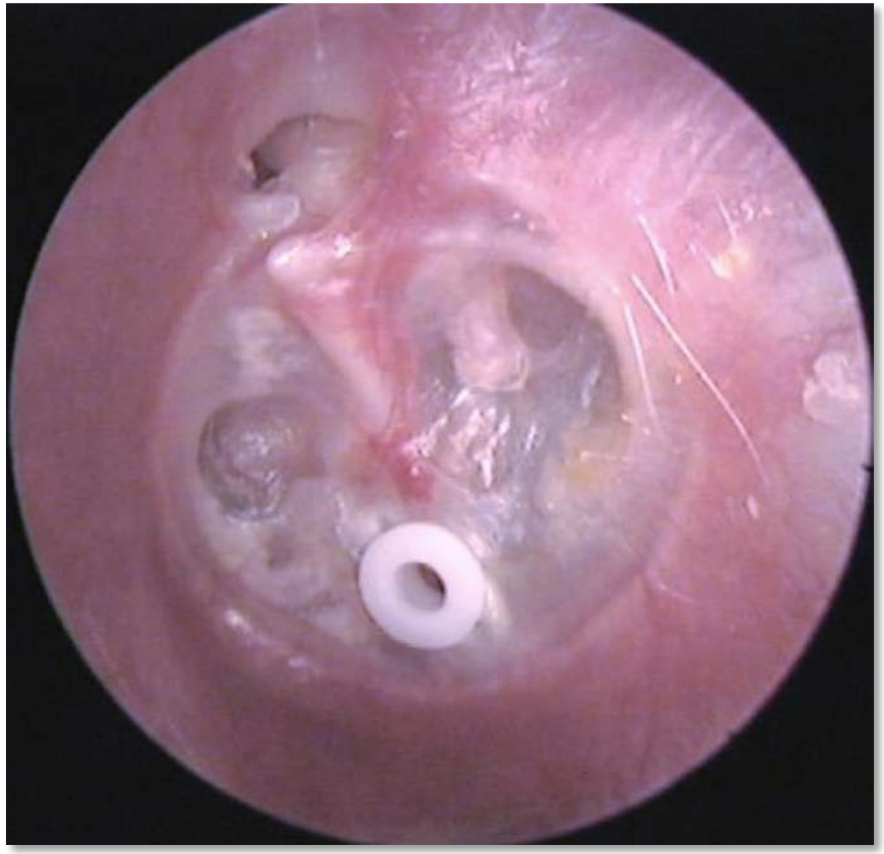




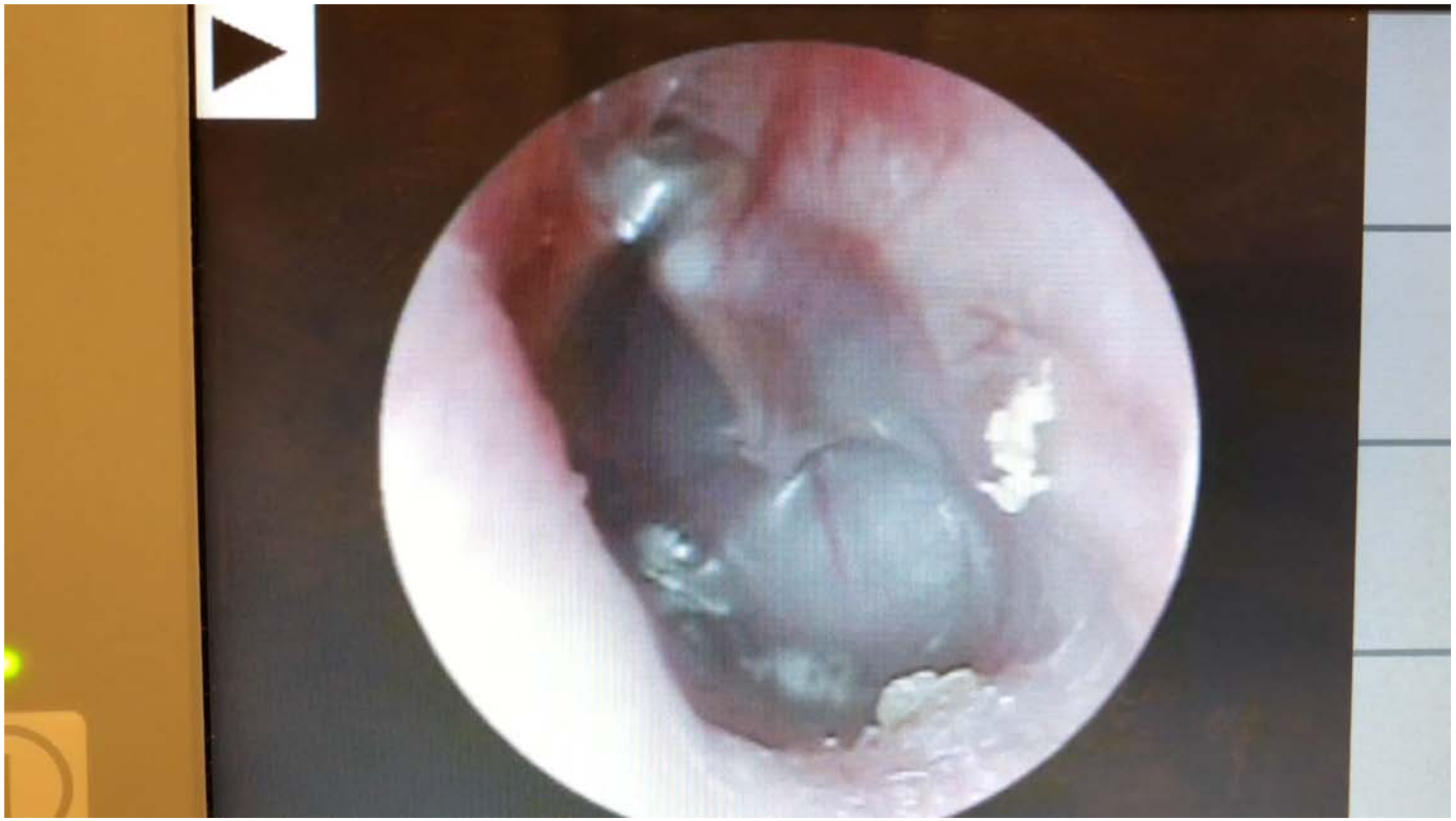
Retracted Tympanic Membrane

- What can be done?

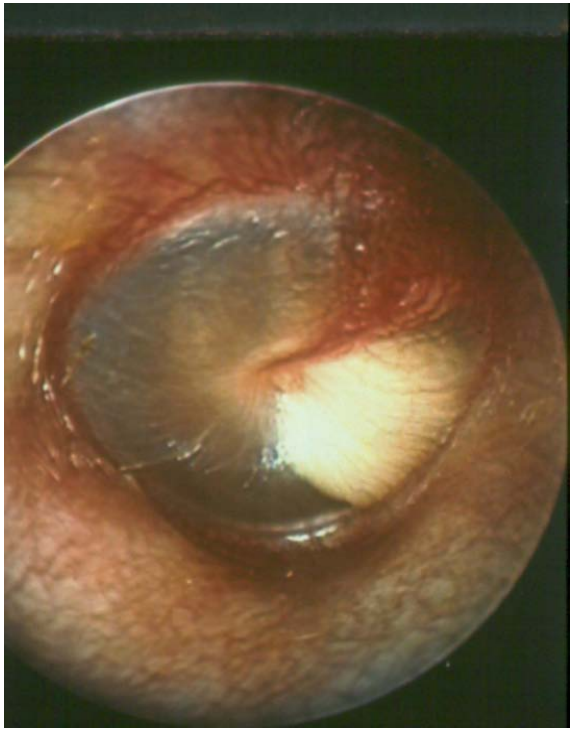
Retracted Tympanic Membrane

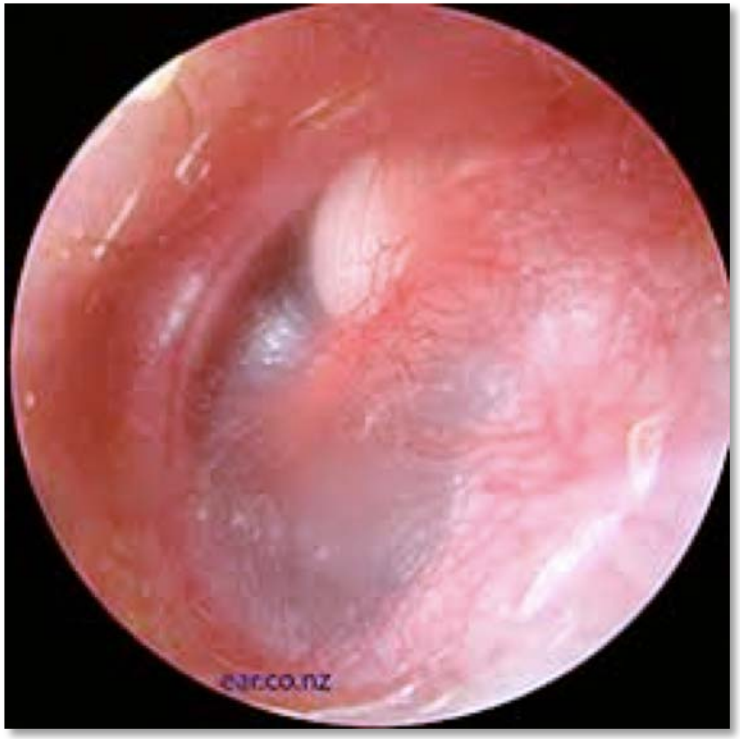


Retracted Tympanic Membrane

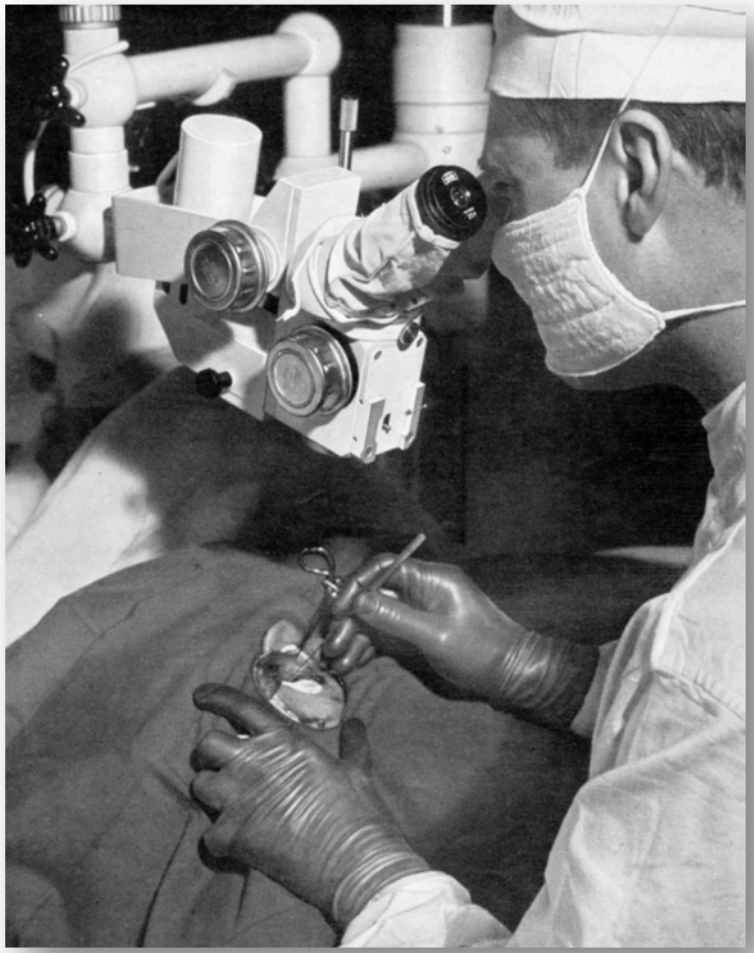


Cholesteatoma





Otologic surgery



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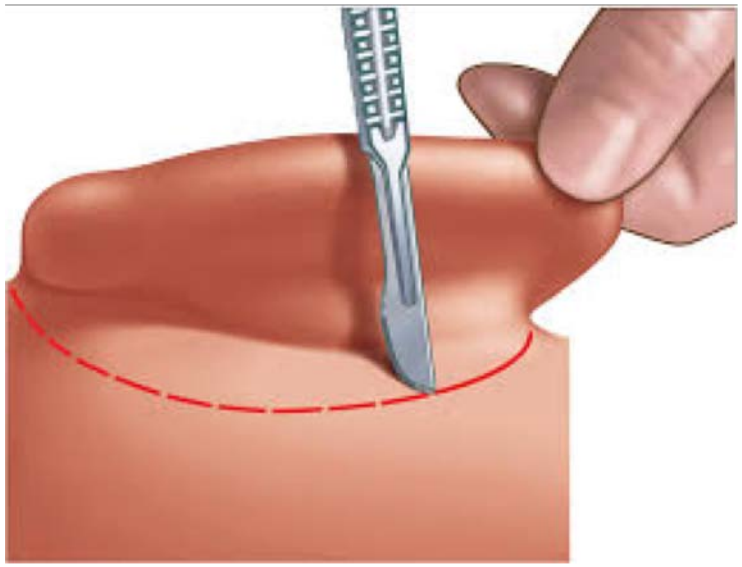
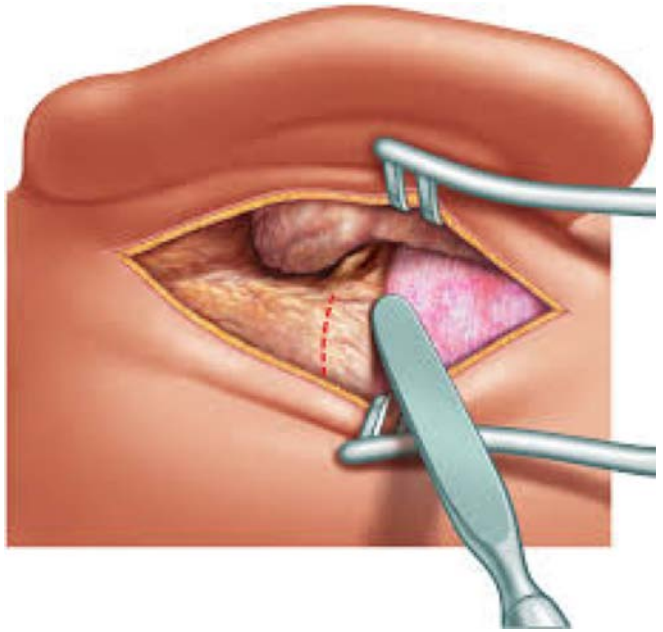
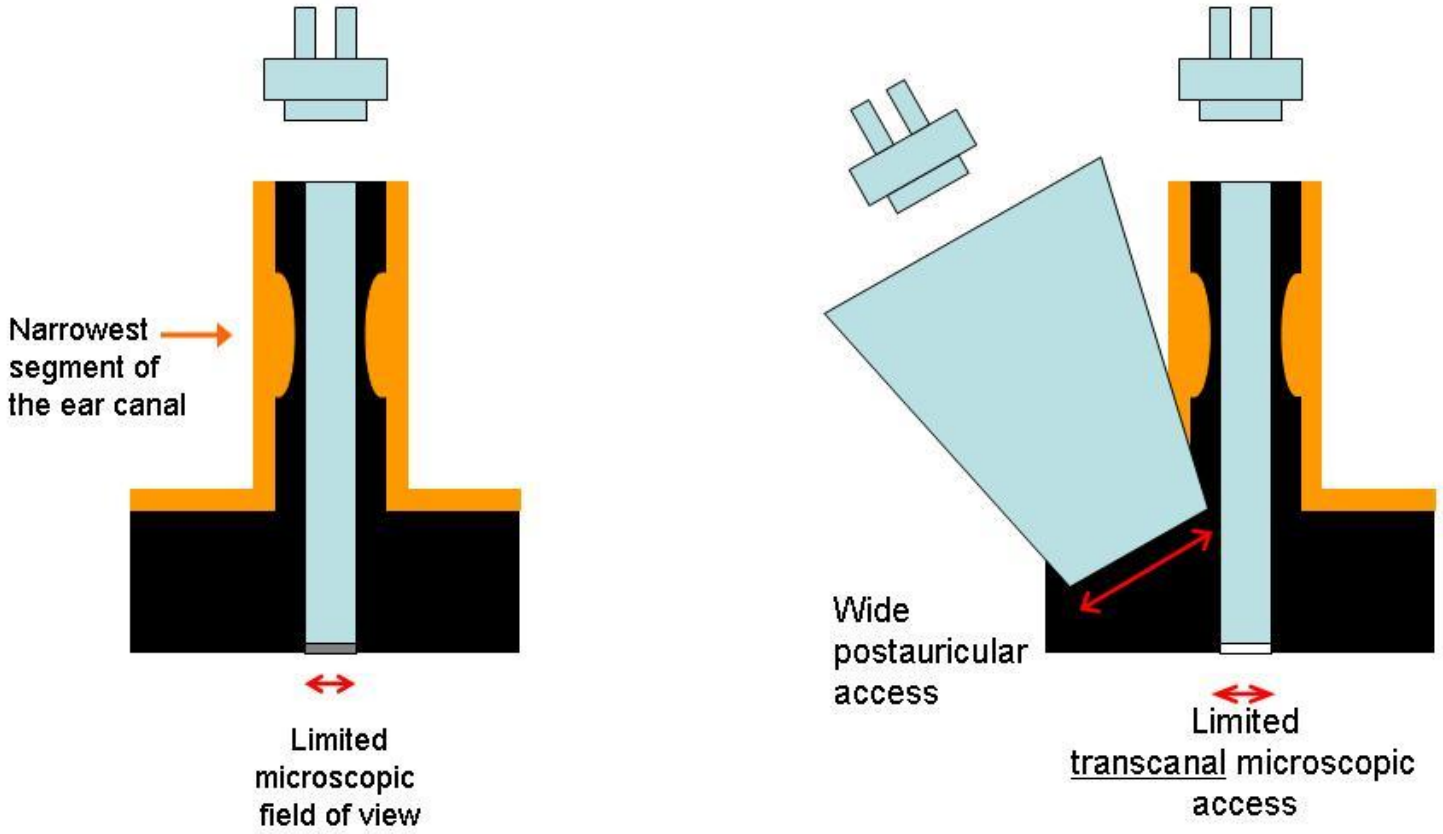
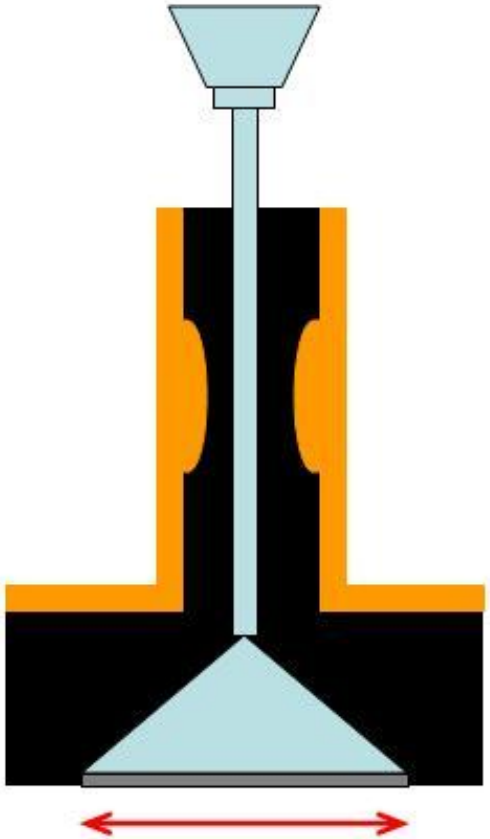


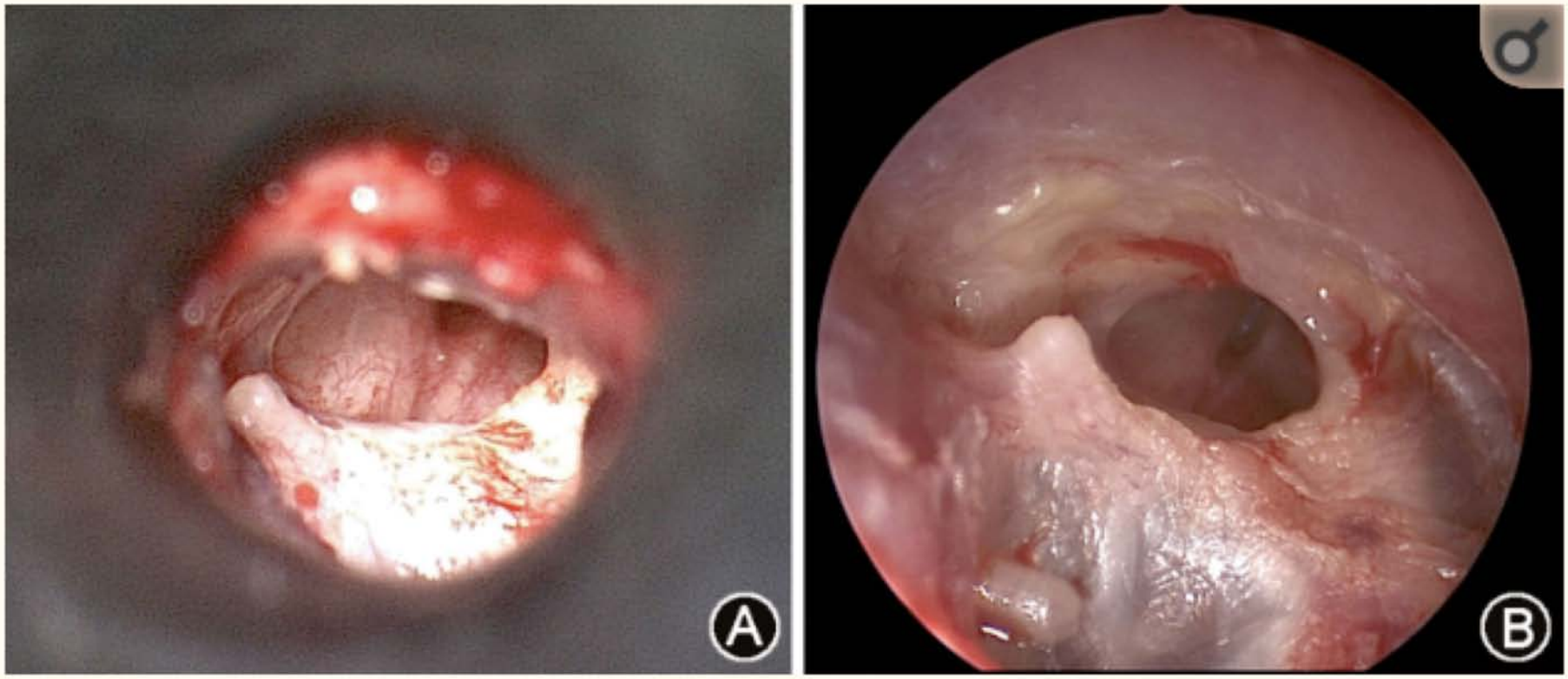



Narrowest segment of the ear canal
Limited microscopic field of view
Wide postauricular access
Limited transcanal microscopic access

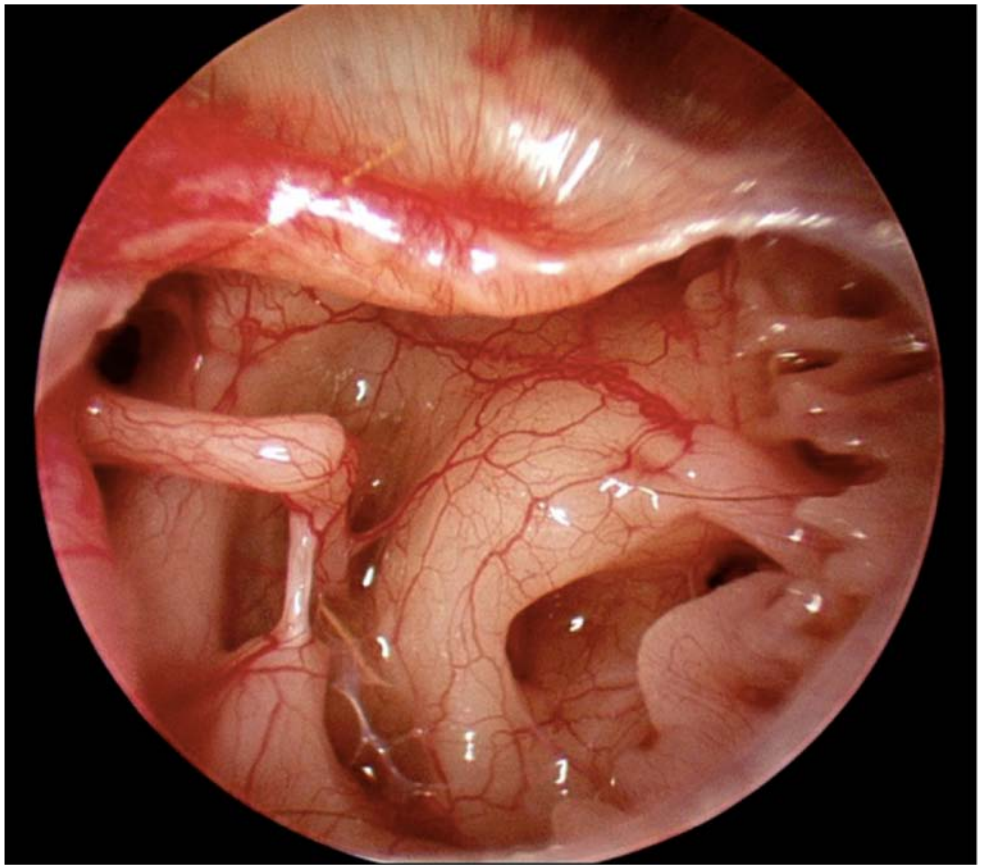


Wide endoscopic
field of view

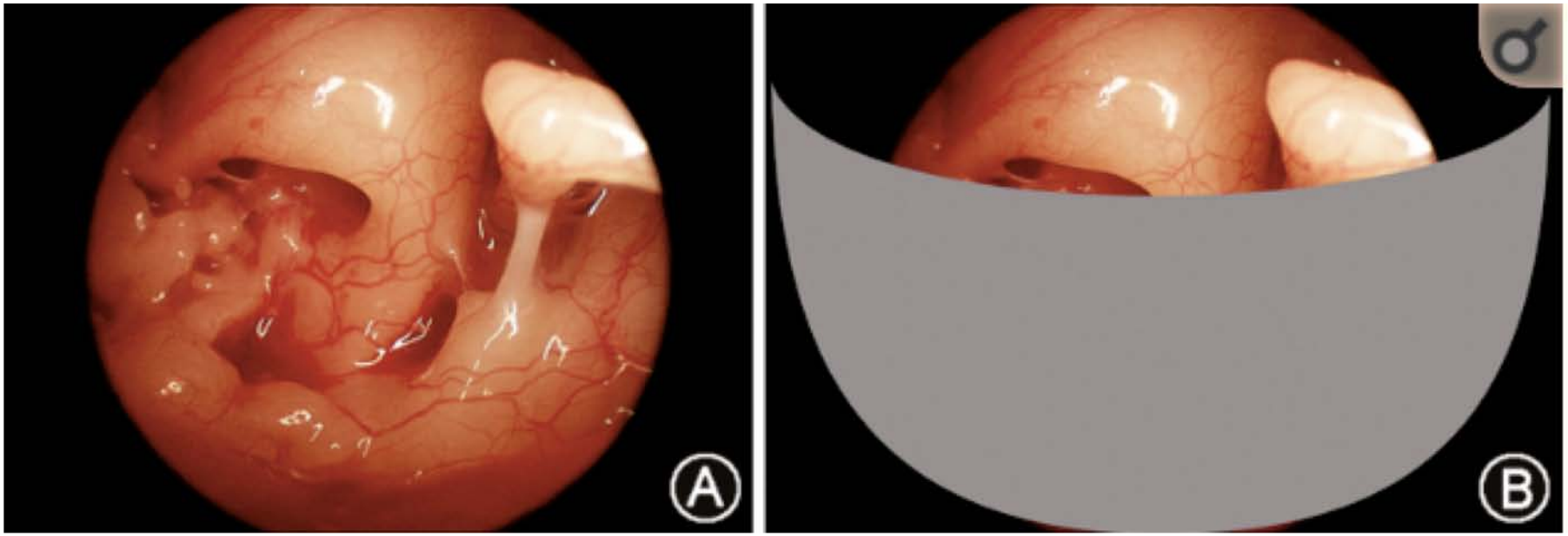
Microscope versus Endoscope



Endoscopic view

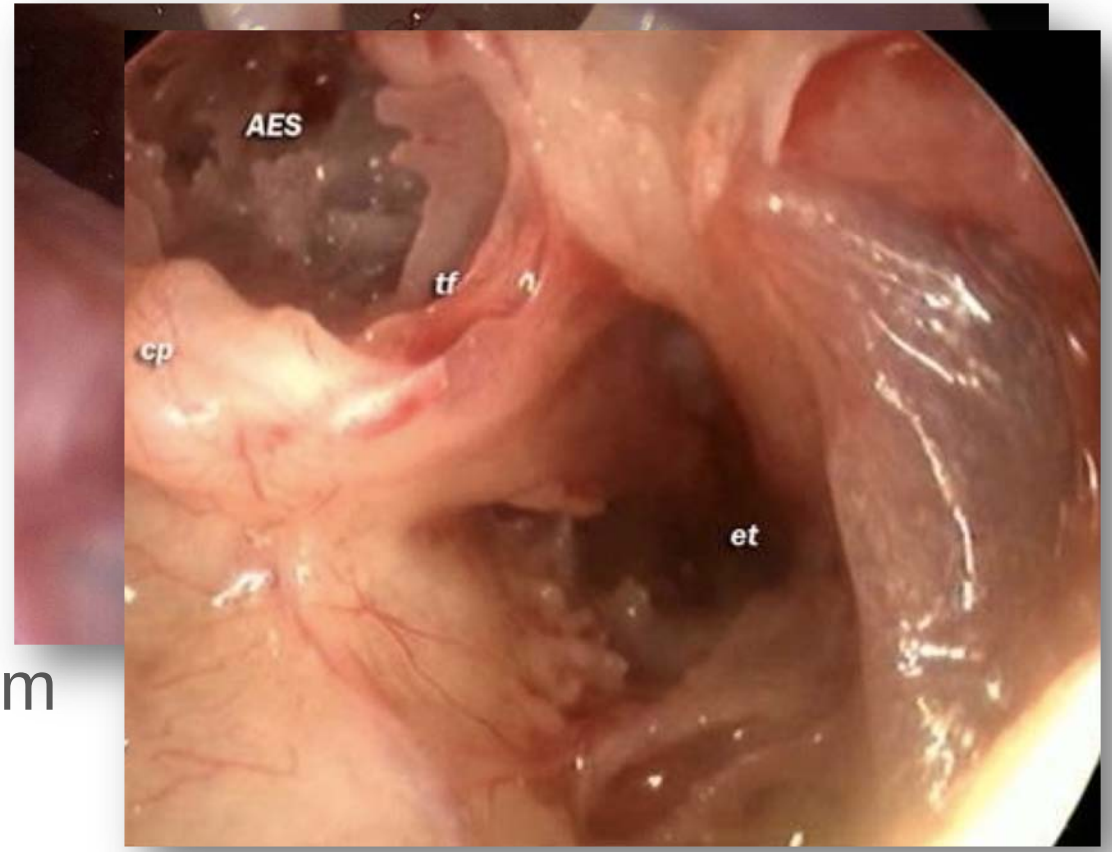


Hard-to-reach areas



Hard-to-reach areas

- Sinus tympani
- Facial recess
- Hypotympanum
- Attic
- Anterior epitympanum



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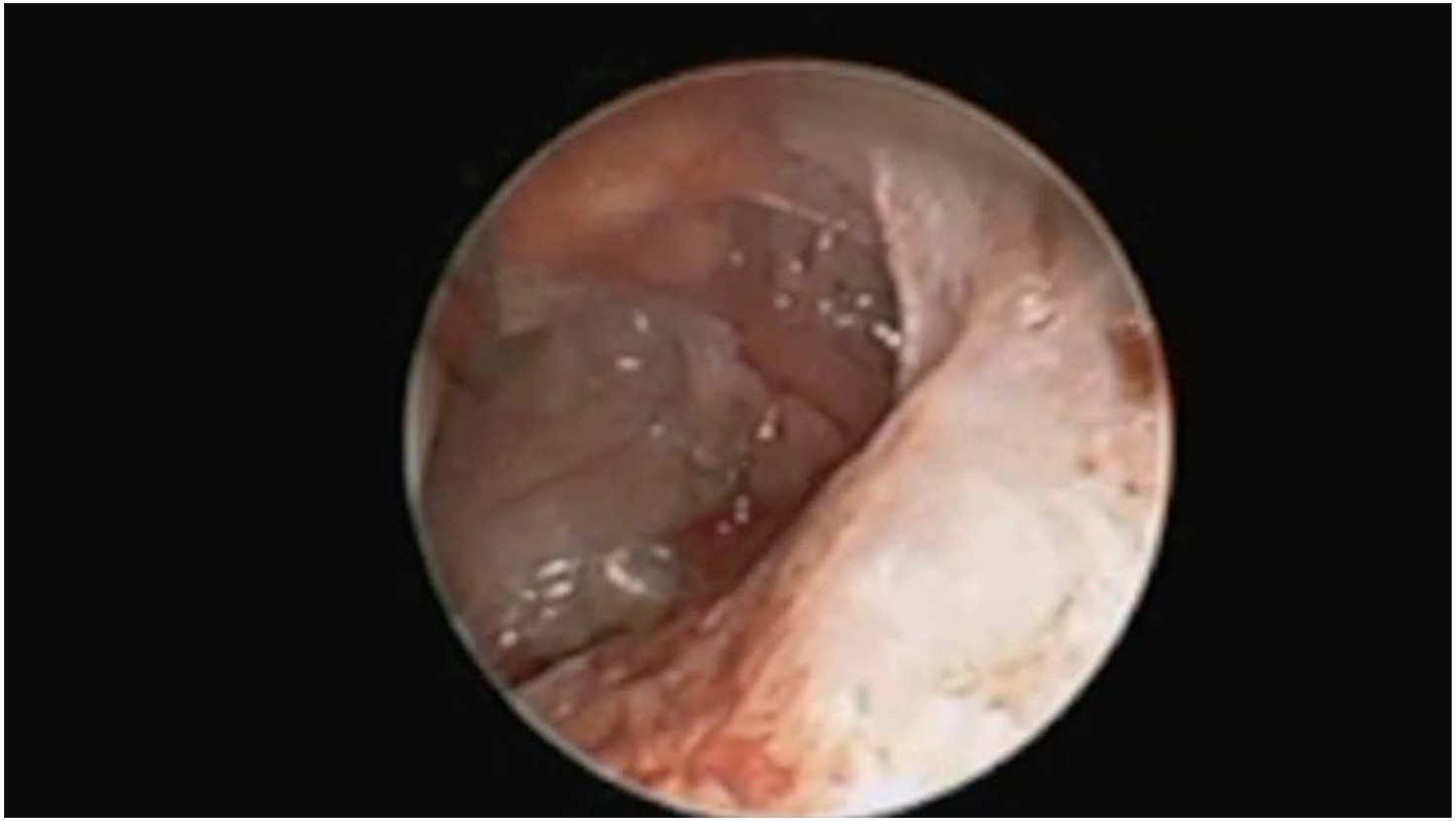
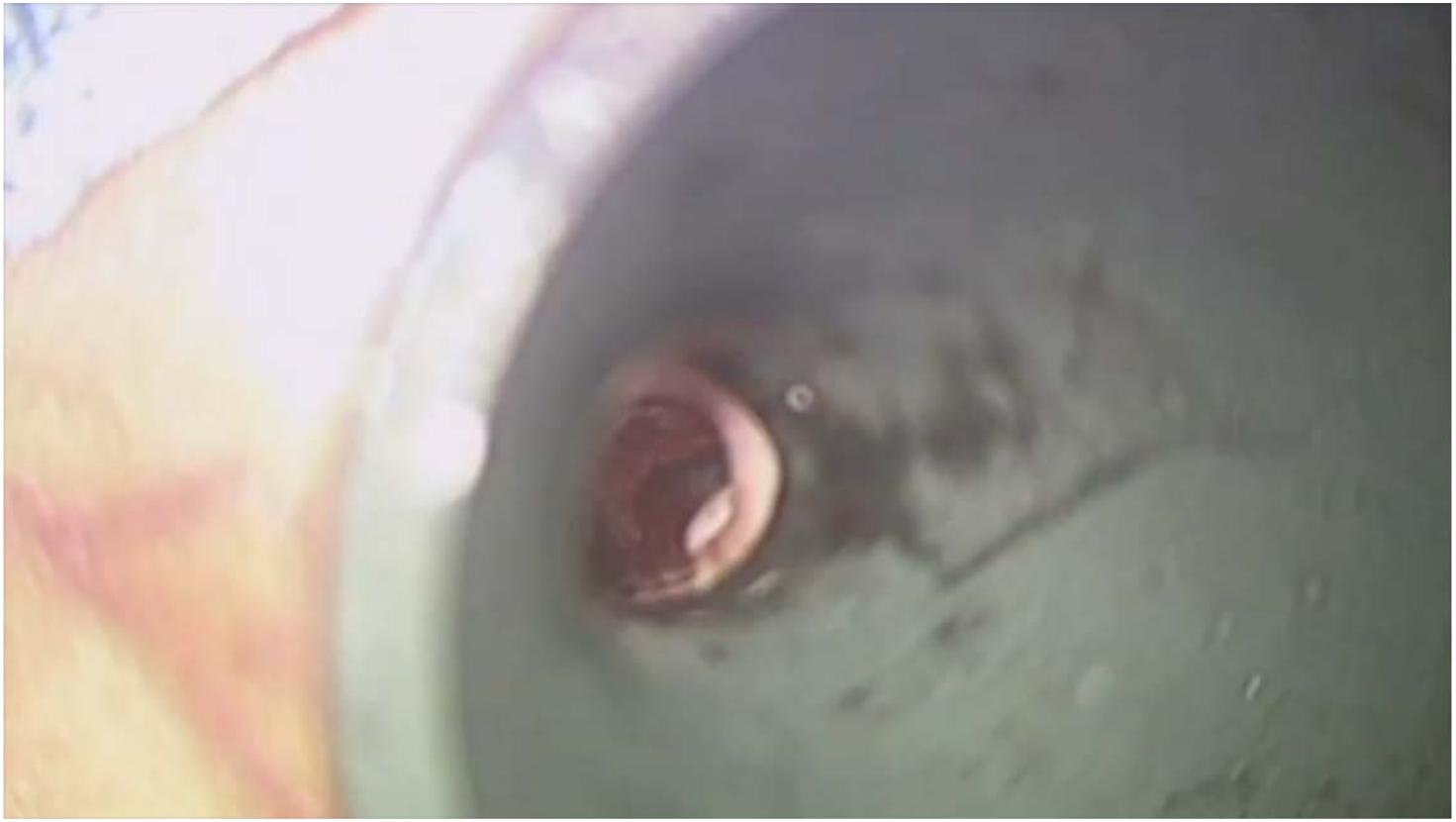
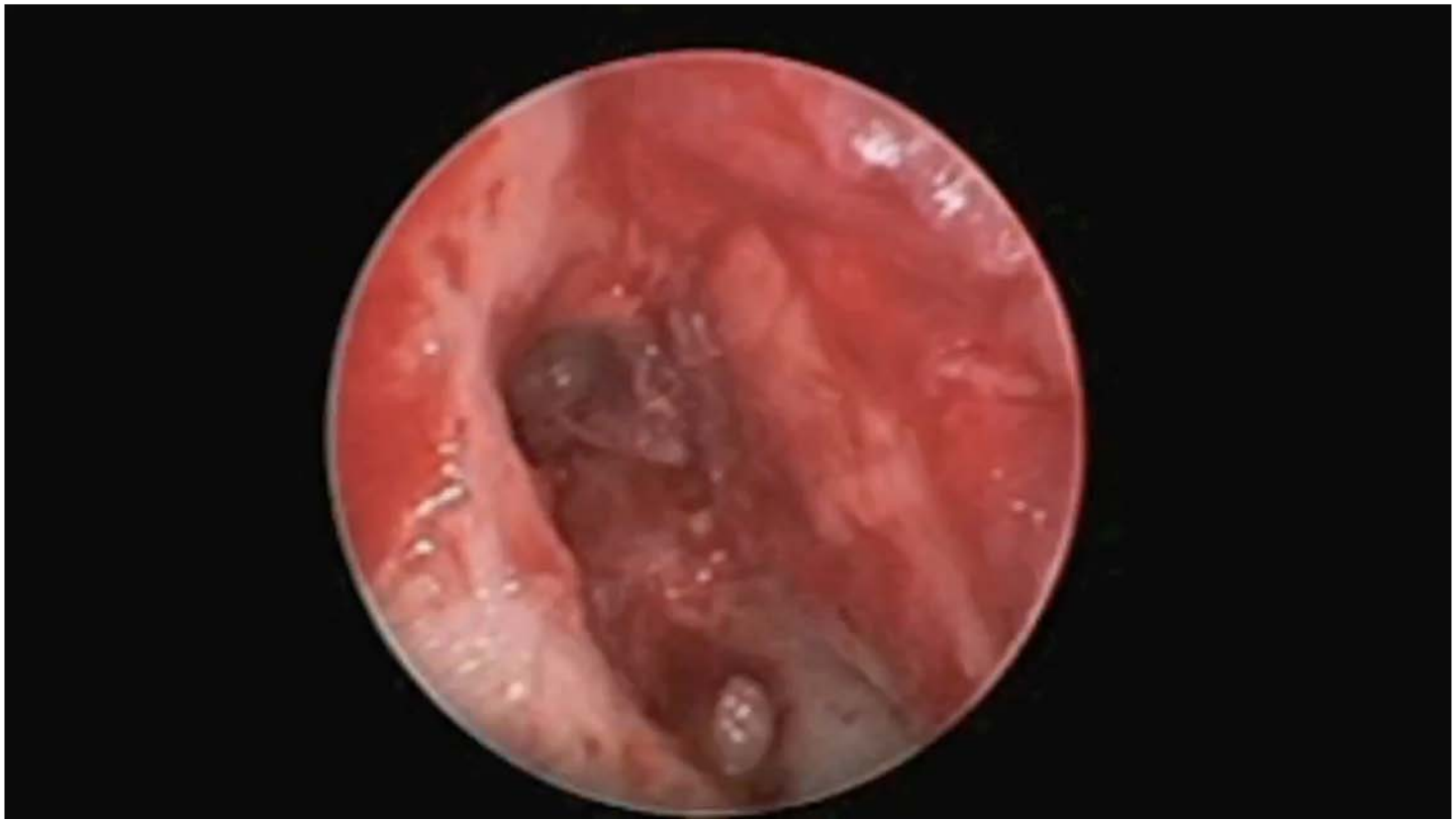


Cholesteatoma



Cholesteatoma

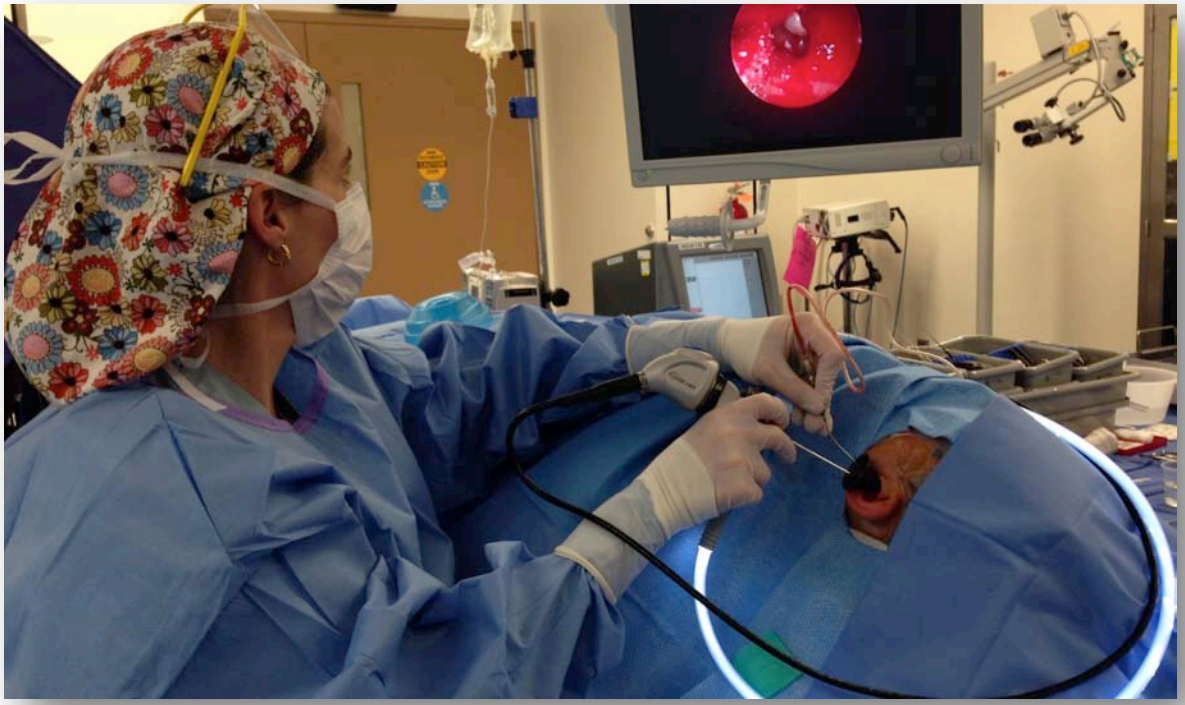


Cholesteatoma recurrence

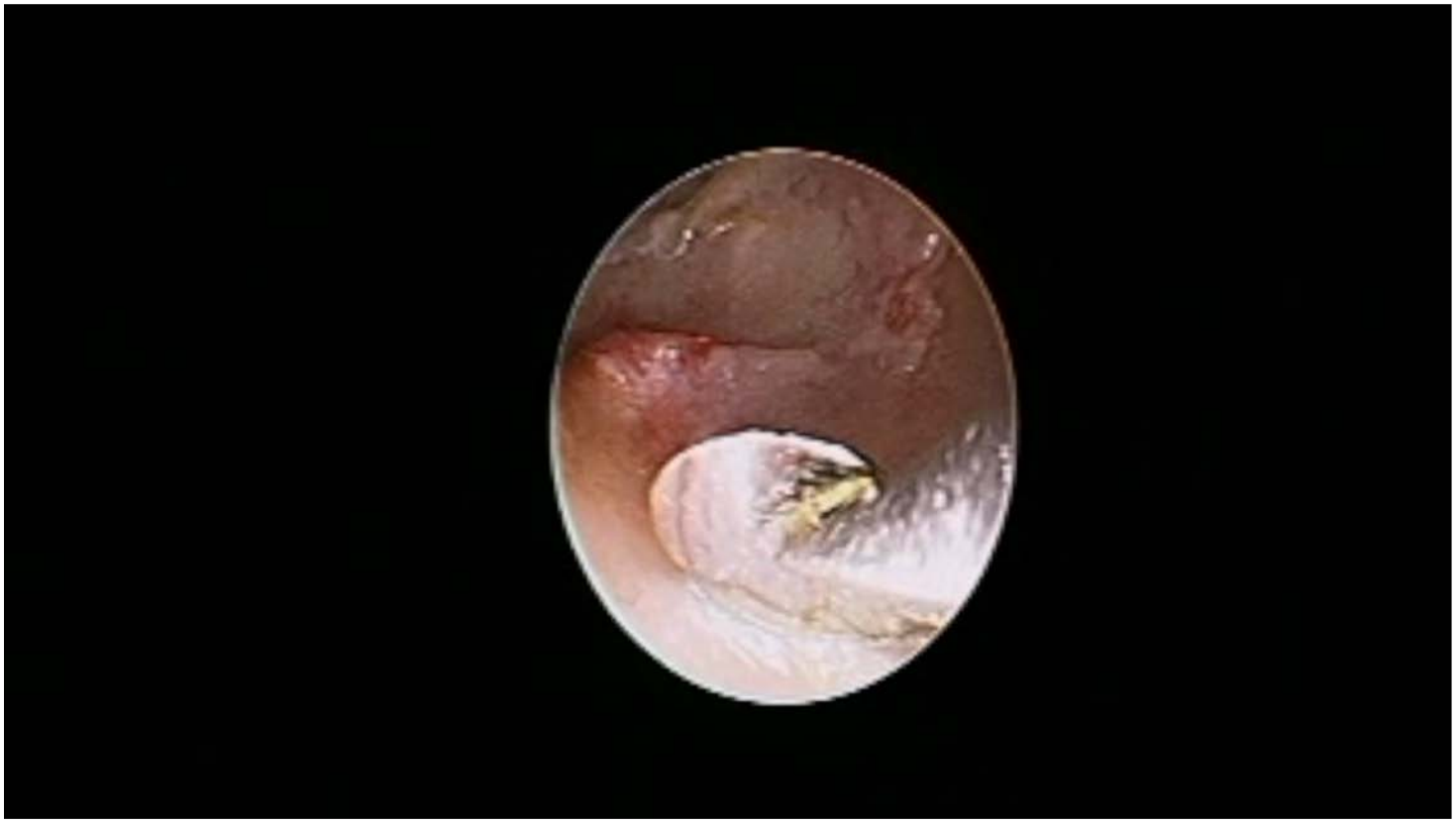
- Recurrence rate
  - Endoscopic = 8.6%
  - Canal-wall-up mastoidectomy = up to 25%

Badr-el-Dine M, et al. Otol Neurotol. 2002

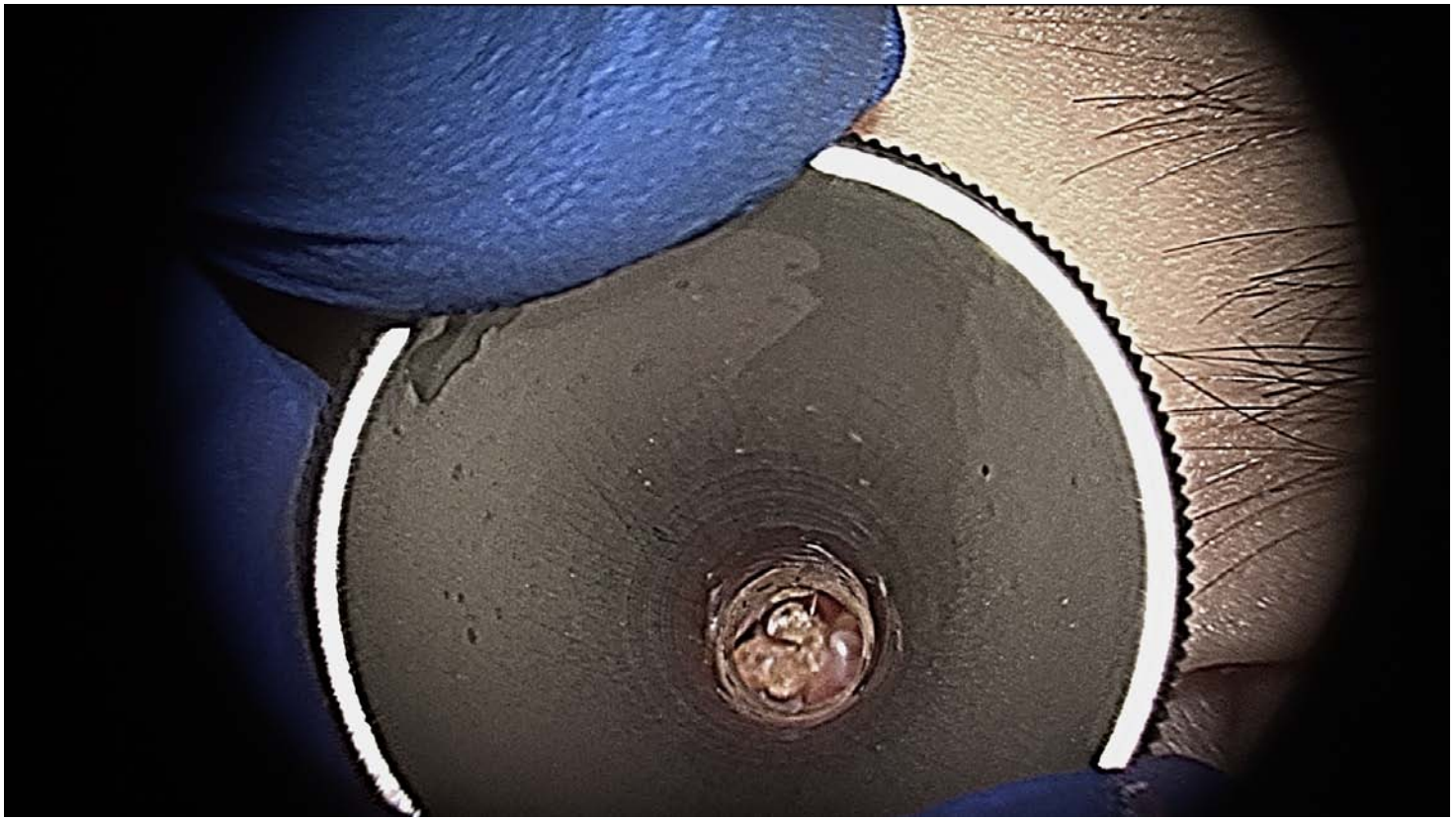
Set up



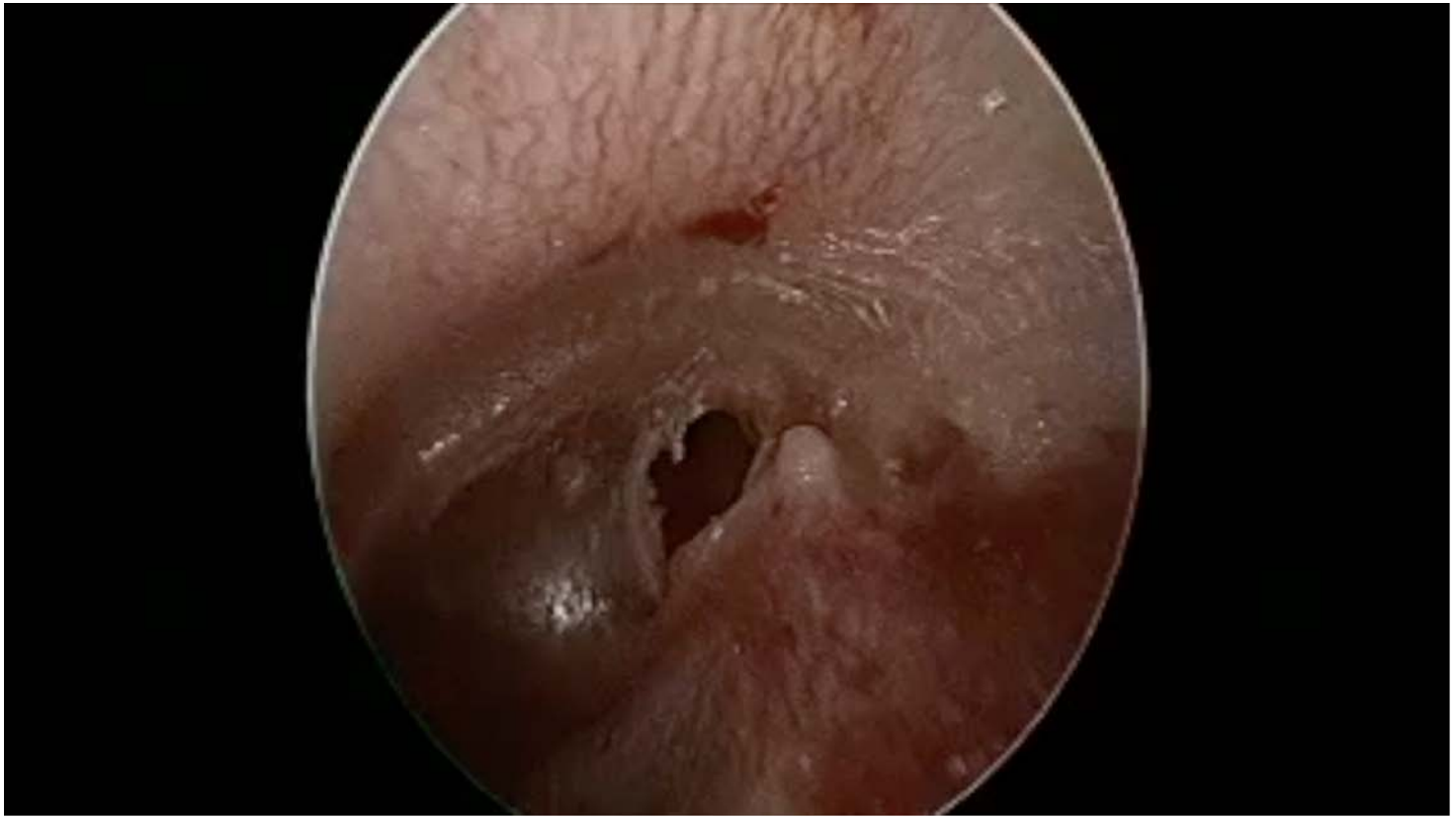
Middle ear exploration



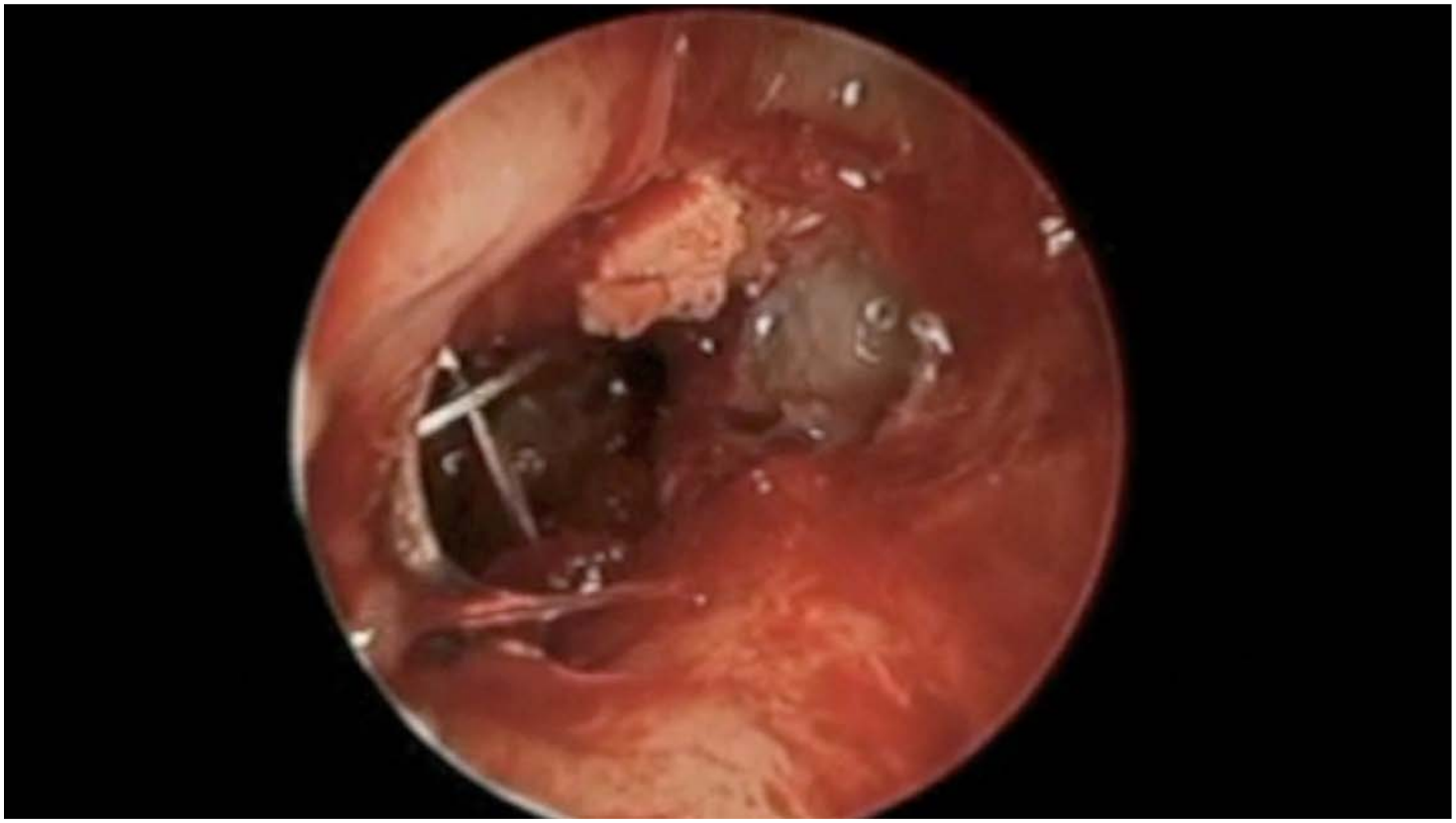
Anterior retraction/cholesteatoma



Anterior retraction/cholesteatoma



Ossicular Reconstruction



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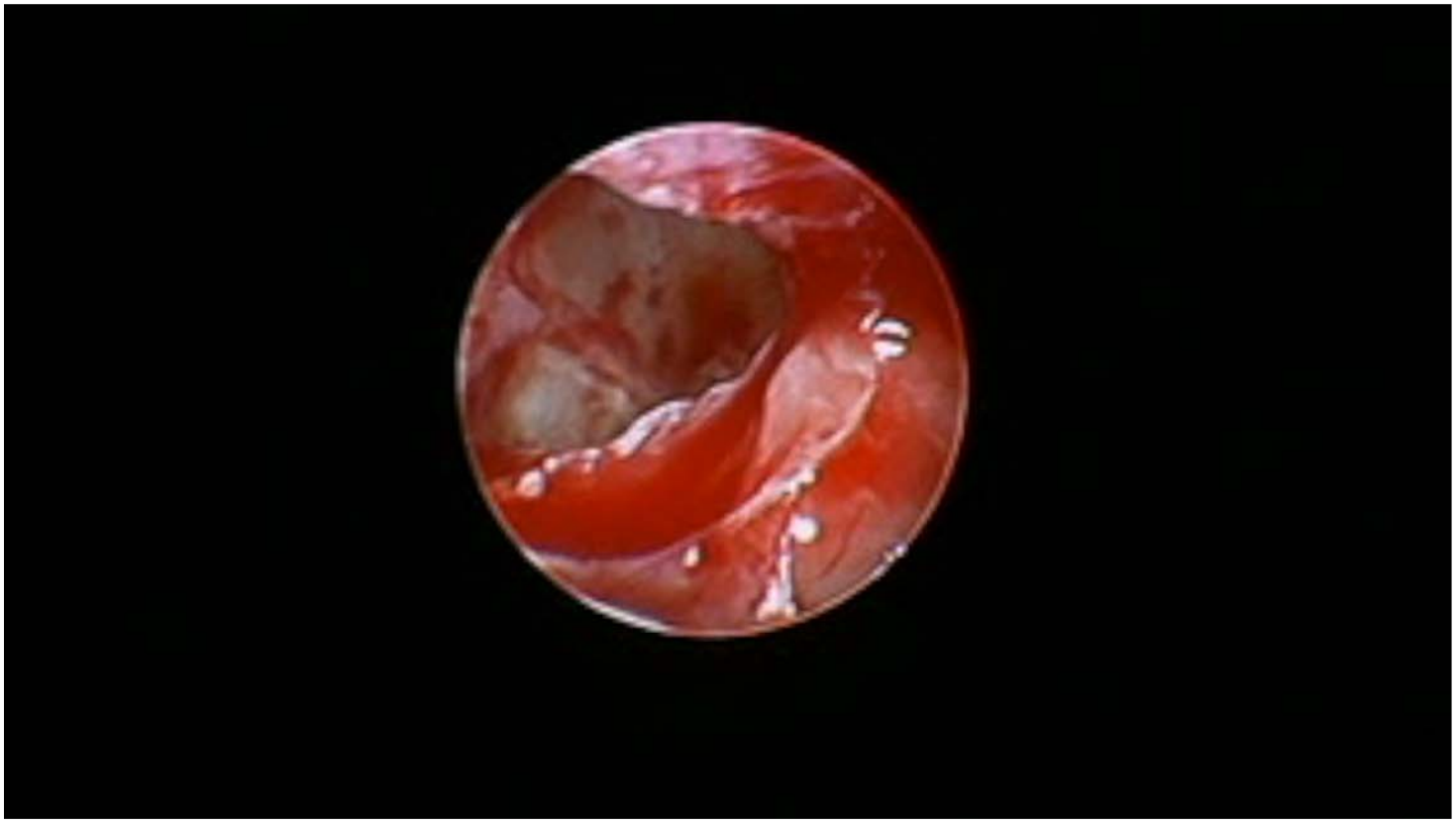

SENTAC

Houston, November 30 – December 2, 2018

San Diego, 2019

Thank you!

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Because Children Are Our Future

Cochlear Implant Team

Rady Children's Hospital · San Diego