RADY403

Mixed Hearing Loss

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UNC SCHOOL OF MEDICINE Radiology

Focused patient history and workup

- **HPI:** One year old who presented from outside institution for failed ABR x2 in both ears. At initial presentation, no response to bone conduction. BAHA was attempted, but speech and language did not progress.
- **Family Hx:** Mother with hearing loss at age 14. Grandfather with severely impaired hearing from young age.
- **PE:** well-developed, well-nourished infant. Face symmetric. TMs in neutral position. Anterior rhinoscopy benign. OP exam wnl.
- **Diagnoses:** Bilateral severe-profound sensorineural hearing loss.

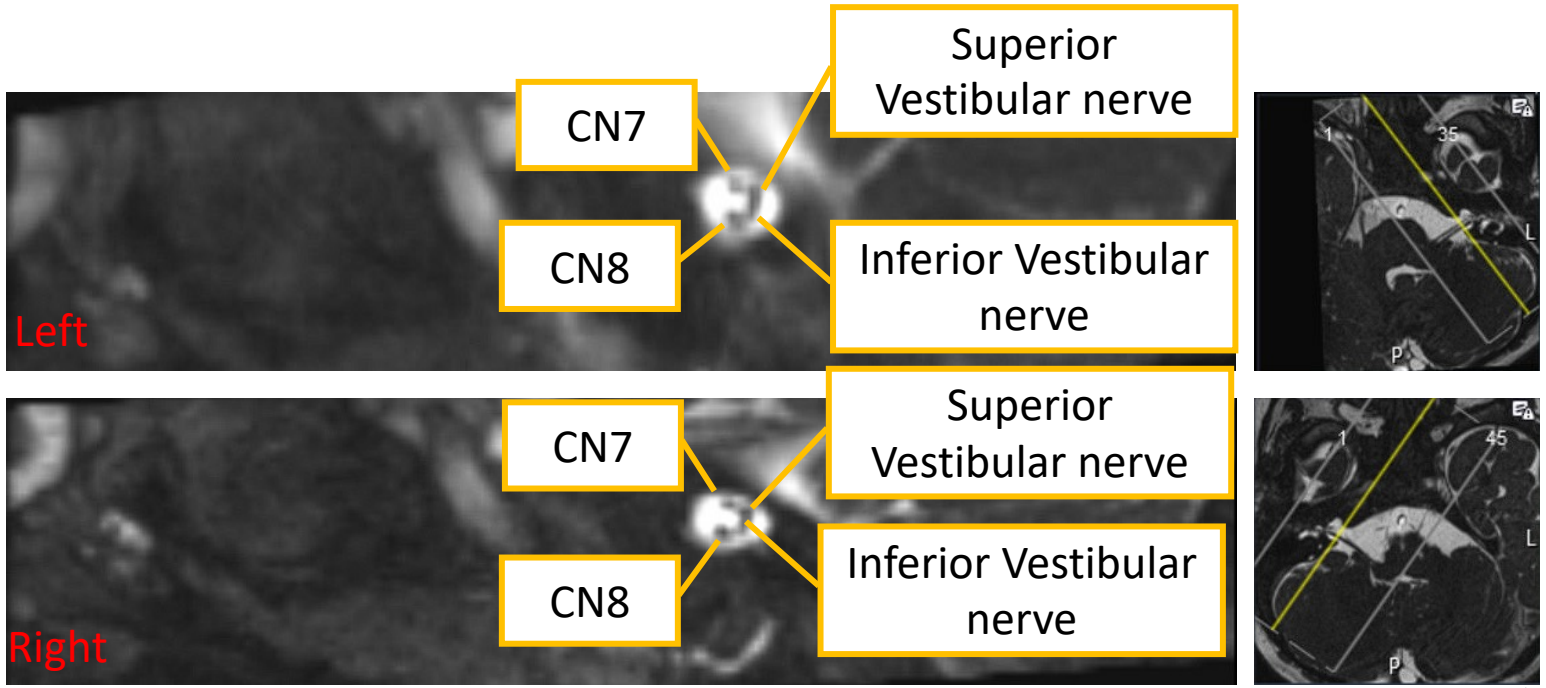
Further imaging is ordered…

List of imaging studies

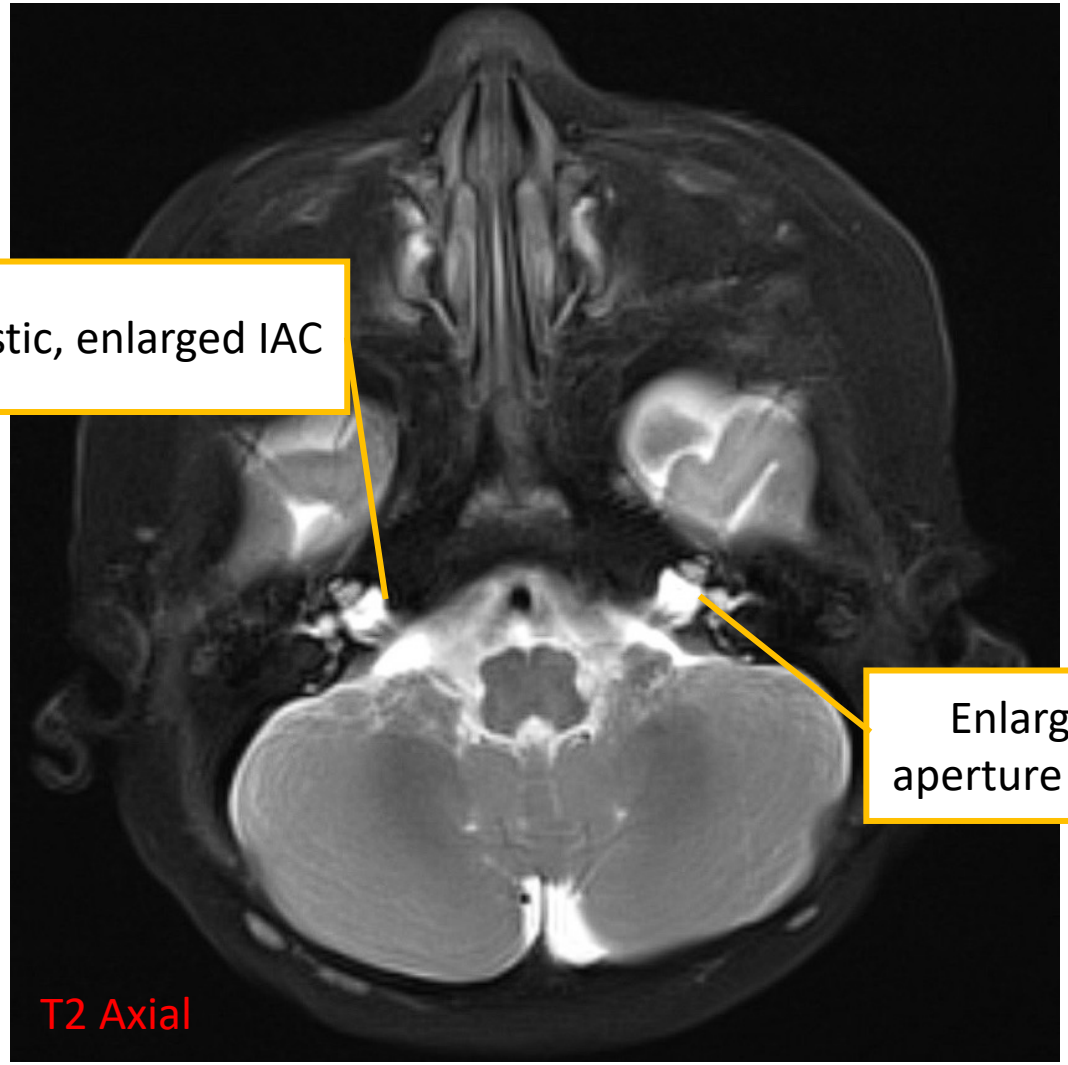
- MRI Brain
- Xray Skull (8 months after initial MRI, post-operative)
- CT Temporal Bones (1 year after Xray, post-operative)

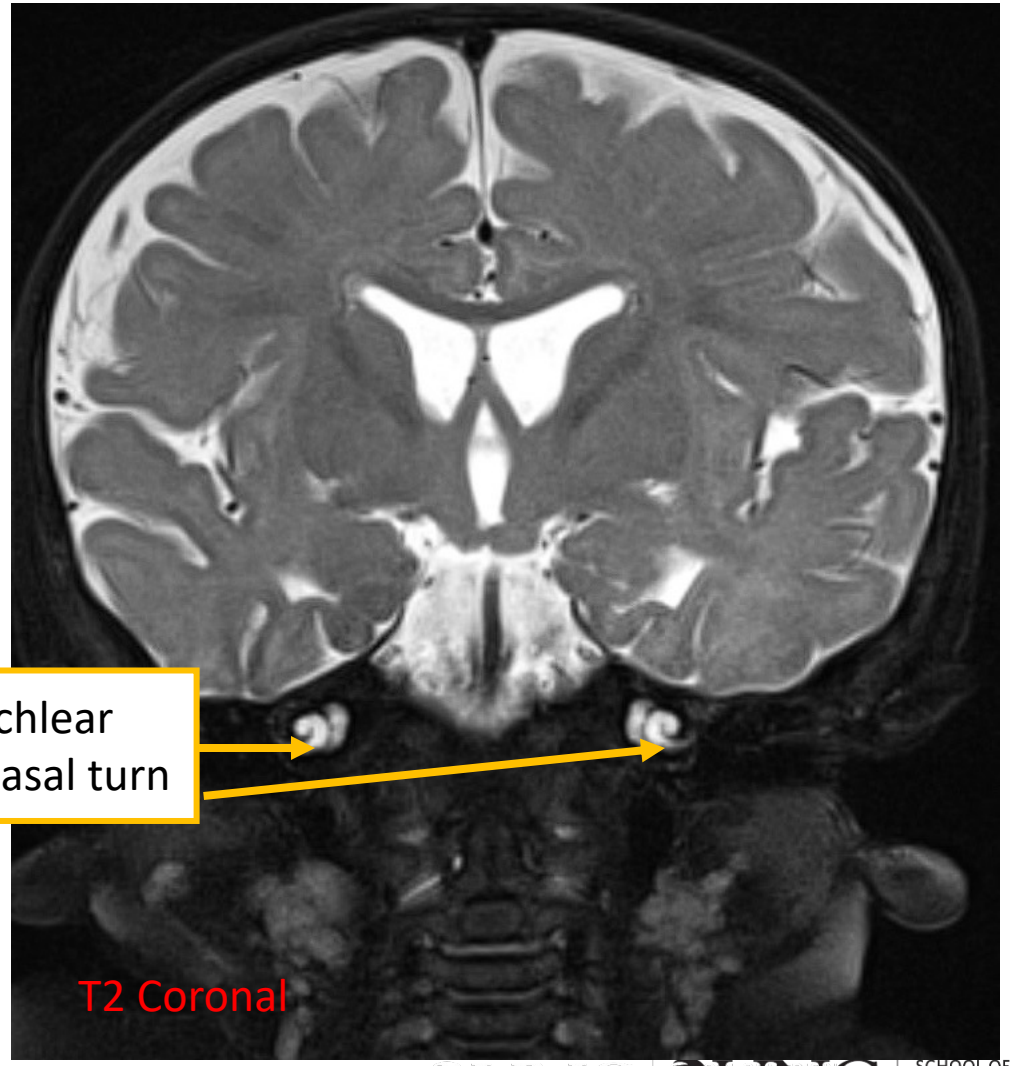
MRI Brain w/wo contrast

- "Seven-up, coke down" is a mnemonic to help remember the orientation of CN7 and CN8 in the internal auditory canal (IAC).
- Patient with appropriately present IAC structures.



MRI Brain w/wo contrast

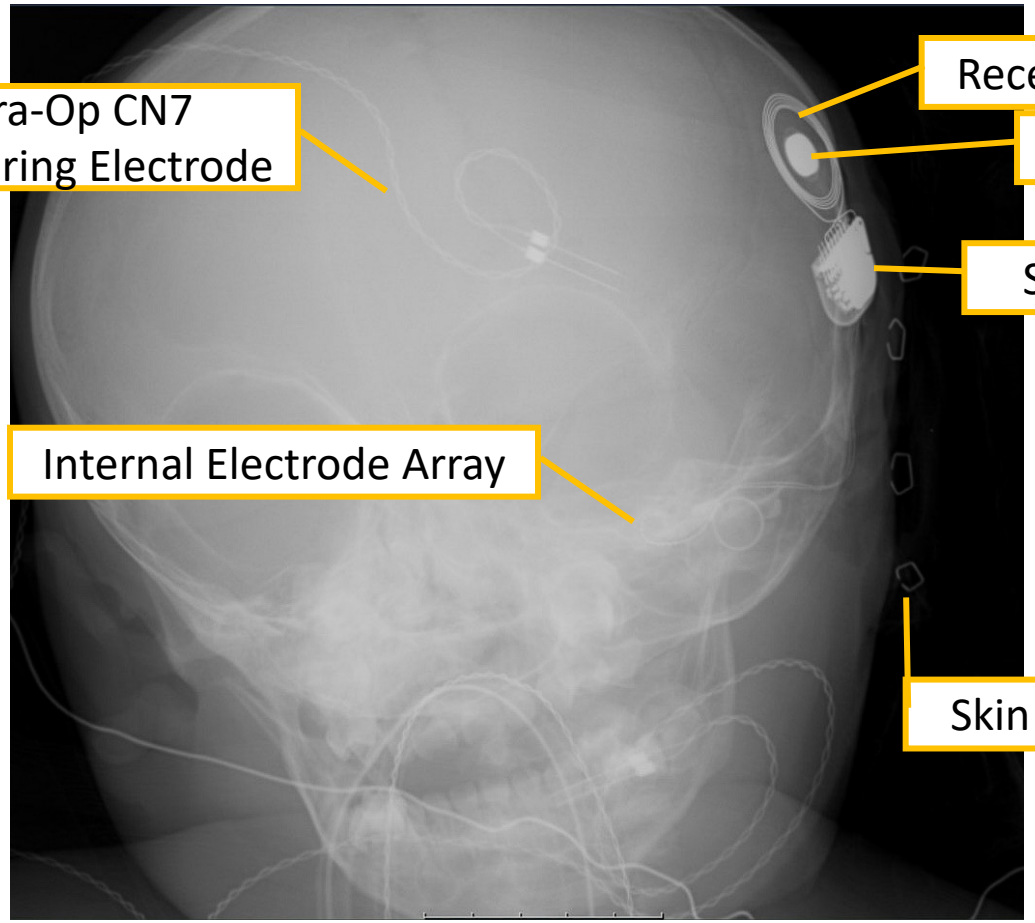


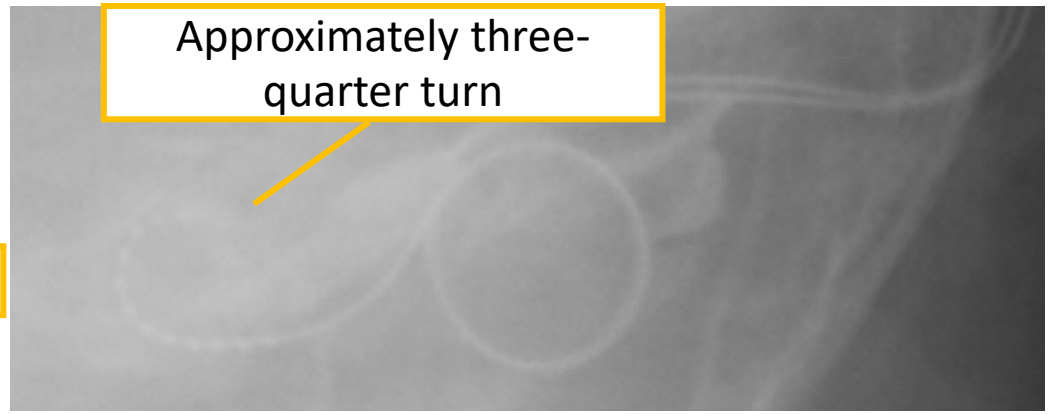


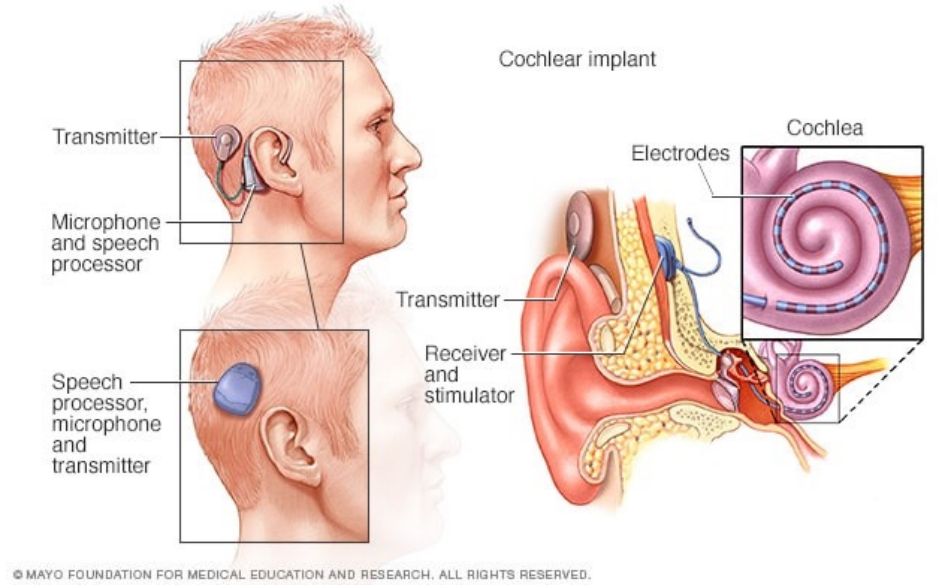
Interval History

- Patient was diagnosed with **X-linked Stapes Gusher syndrome (DFNX2)**
- Patient failed to progress developmentally over the following year despite multiple attempted treatments
  - Focused speech/developmental therapies
  - Bilateral Hearing Aids
  - Bone-anchored hearing aids
- Ultimately, the family elected for cochlear implantation 1 year later.

Intra-operative Xray Skull







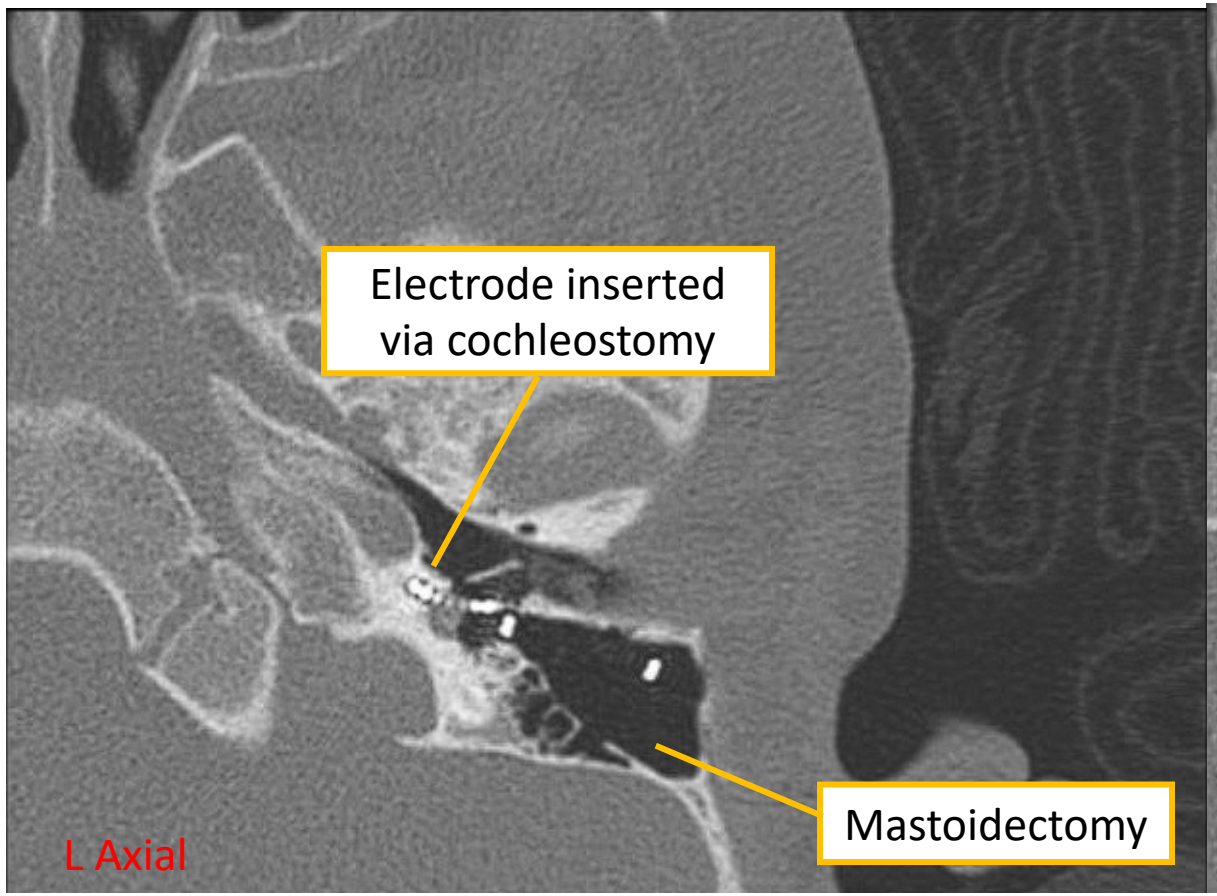
Citation: Mayo Clinic

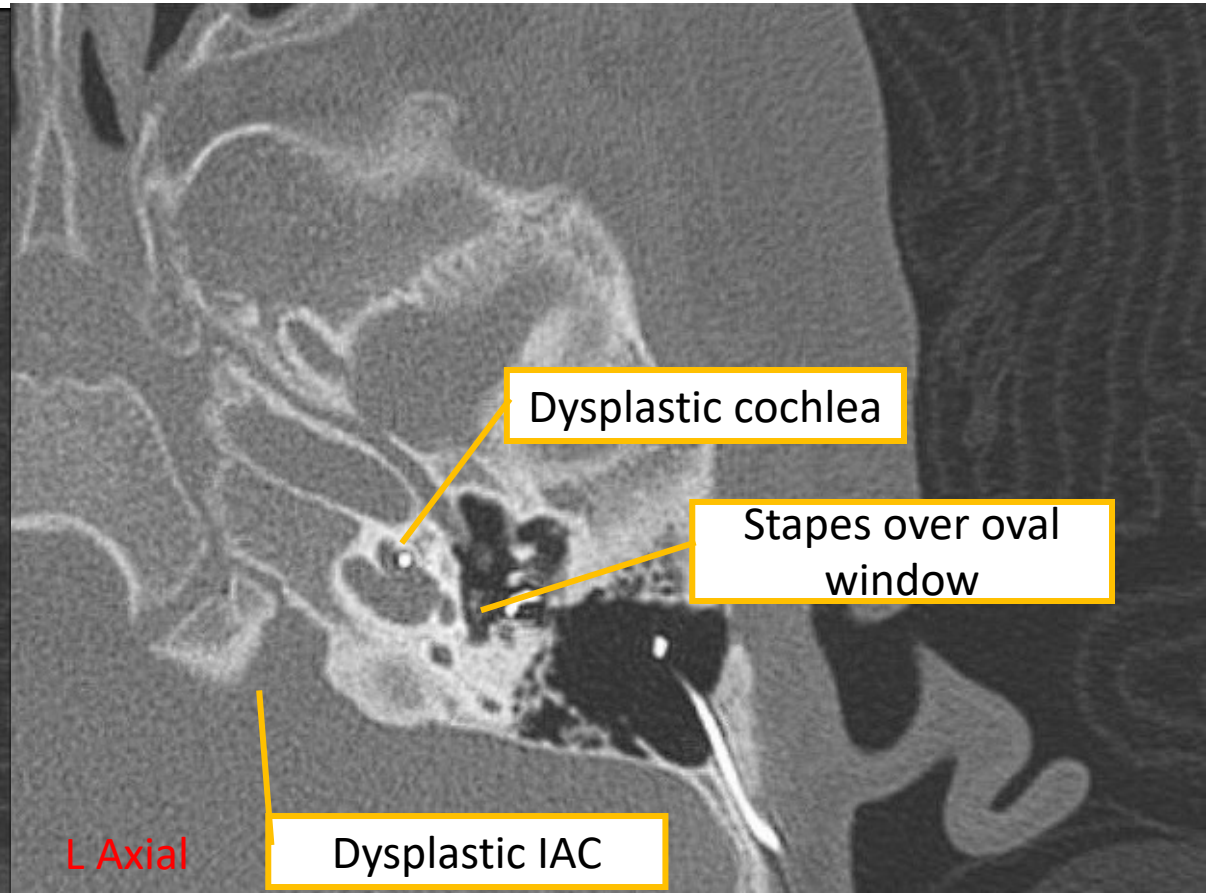
Interval History

- Patient continued on multidisciplinary treatment for approximately 8 months, but continued to have poor speech outcomes
- Due to the patient's age and language development, determination of device failure was difficult

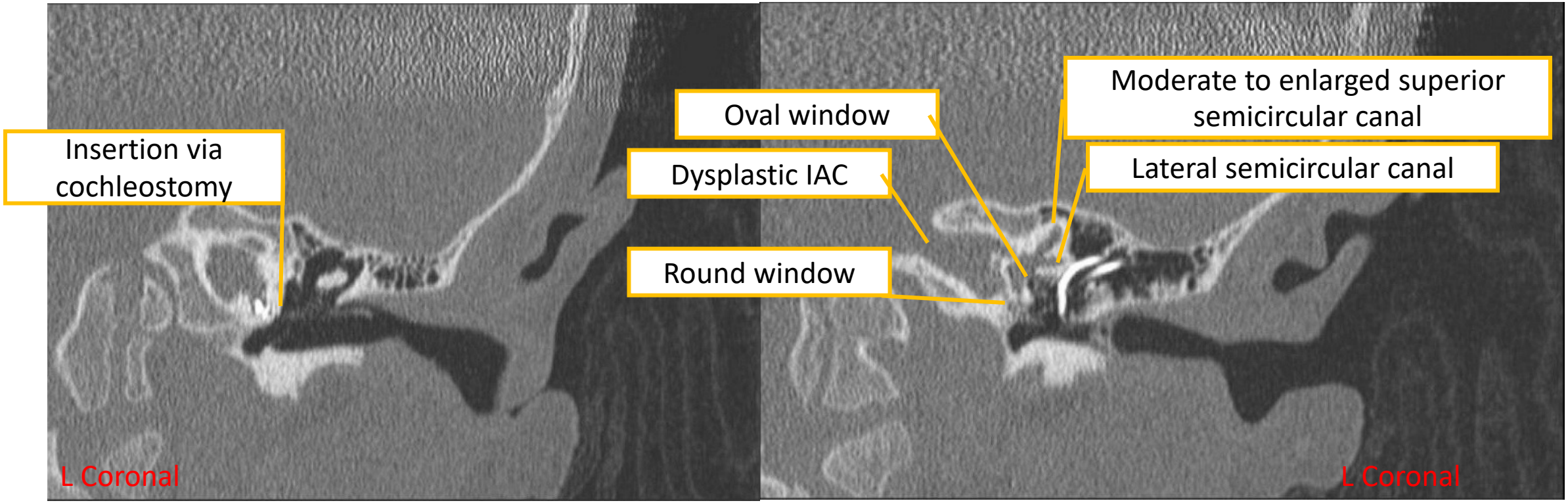
CT TEMPORAL BONE WAS ORDERED TO ASSESS POSITIONING OF THE IMPLANT

CT Temporal Bones





CT Temporal Bones



Patient Treatment Outcome

- Audiological outcomes were moderate to poor
- Patient ultimately underwent cochlear implant revision about one year later to advance his electrode (re: three-quarter turn) and reposition the internal processor
- After revision, patient recovered well and improved speech recognition performance
- Ultimately, the implant became lateralized again 9 years later, and the patient underwent repeat revision

Clinical Characteristics of X-Linked Stapes Gusher Syndrome

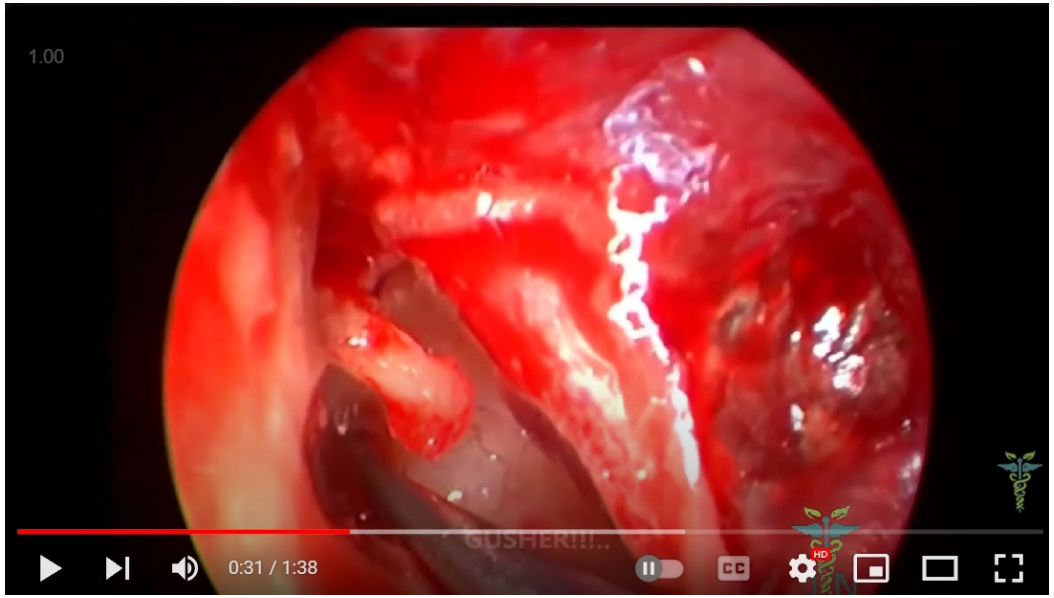
- Often mixed sensorineural/conductive hearing loss due to POU3F4 mutation.[2,3]
- Sometimes conductive-predominant phenotypes can prompt exploration of the middle ear (stapes) causing the infamous "stapes gusher."[3]
- The condition is due to sudden, rapid flow of perilymph due to abnormal connection between the subarachnoid and perilymphatic spaces, often in cases of congenital malformation of the cochlear aqueduct/IAC.[4]
- Stapes gusher can occur in a **wide** variety of clinical contexts, which is why more specific names have been suggested: Incomplete Partition type 3, or DFNX2.[5,6,7]
- Accounts for ~50% of all familial non-syndromic deafness.[8]

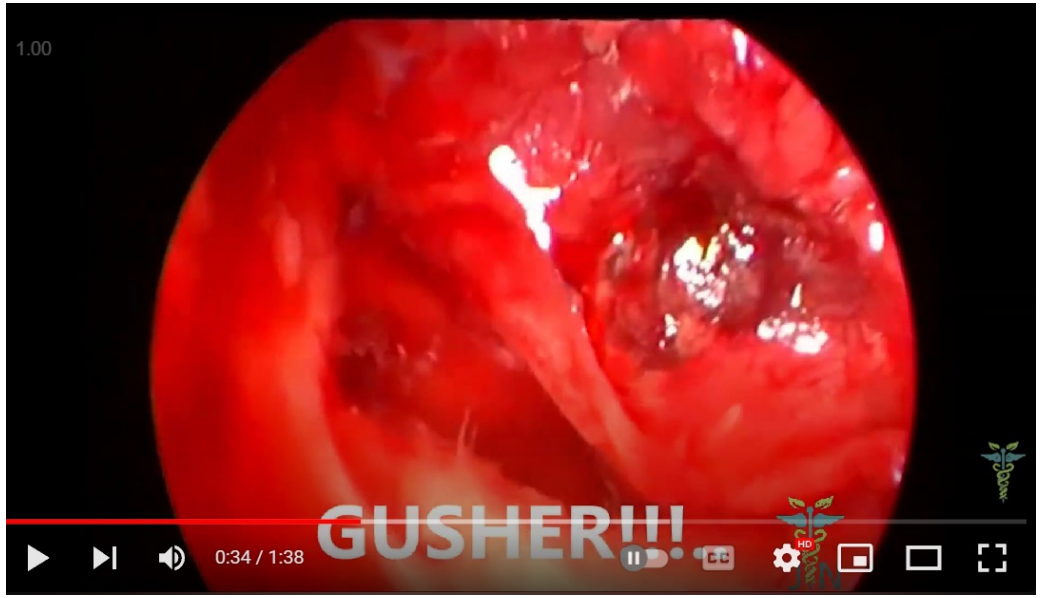
Clinical: What to call X-Linked Stapes Gusher Syndrome

- A syndrome of many names:[6,7]
  - Preferred Radiologic Terminology: Incomplete partition type 3
  - X-linked deafness type 2 (DFNX2)
  - Deafness type 3 (DFN3)
  - Conductive deafness with stapes fixation
  - Nance Deafness
  - X-linked mixed conductive and sensorineural hearing loss
  - X-linked progressive mixed deafness with perilymphatic gusher during stapes surgery
  - X-linked stapes gusher

Intra-operative stapes gusher

- https://www.youtube.com/watch?v=jbn3n9jXRFw – Dr. Joseph Nadakkavukaran
- 0:29





Radiology: ACR Appropriateness Criteria

Variant 4: **Mixed conductive and sensorineural hearing loss. Initial imaging.**

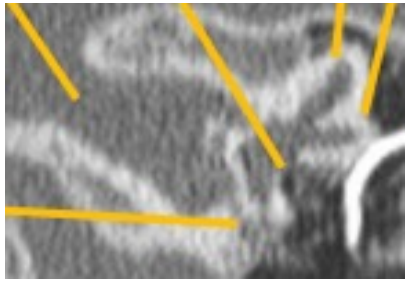
Procedure	Appropriateness Category	Relative Radiation Level
CT temporal bone without IV contrast	Usually Appropriate	☢☢☢
MRI head and internal auditory canal without and with IV contrast	Usually Appropriate	O
MRI head and internal auditory canal without IV contrast	Usually Appropriate	O
CT head with IV contrast	Usually Not Appropriate	☢☢☢
CT head without and with IV contrast	Usually Not Appropriate	☢☢☢
CT head without IV contrast	Usually Not Appropriate	☢☢☢
CT temporal bone with IV contrast	Usually Not Appropriate	☢☢☢
CT temporal bone without and with IV contrast	Usually Not Appropriate	☢☢☢
CTA head with IV contrast	Usually Not Appropriate	☢☢☢
MR venography head with IV contrast	Usually Not Appropriate	O
MR venography head without IV contrast	Usually Not Appropriate	O
MRA head without and with IV contrast	Usually Not Appropriate	O
MRA head without IV contrast	Usually Not Appropriate	O

Red text indicates characteristic findings of the disease present in this case.

Radiology: Incomplete partition type 3

- The radiographic evidence of DFNX2 primarily involve the cochlea.[9]
  - Present interscalar sella, leading to "corkscrew" appearance
  - Absent or dysplastic modiolus
  - Absent or dysplastic lamina cribrosa at the basal turn of the cochlea

- Some common findings outside the cochlea include:[8,9]
  - Thin otic capsule
  - Bilateral widening of the IAC
  - Enlarged SSSC
  - Dysplastic oval and/or round windows
  - Dilated labyrinthine segment of facial nerve canal (mentioned in Op note)



UNC Top Three Teaching Points

- Familial deafness requires a thorough radiological work-up prior to procedures.
- DFNX2 is characterized radiographically by dysplastic, enlarged, “corkscrew” cochlea with abnormal IACs.
- DFNX2 should be suspected clinically in the setting of profound, bilateral sensorineural +/- conductive hearing loss with positive family history.

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